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Journal of Bioanthropology is a multi and interdisciplinary scientific journal that focuses on scientific research in the field of biological anthropology, bioarchaeology, biomechanics, biomedicine, ergonomics, forensics, genetics, human evolution, molecular anthropology, public health, and related subjects. Official language of the Journal is English.
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Submitted manuscripts will generally be reviewed by two to three experts who will be asked to evaluate whether the manuscript is scientifically sound and coherent, whether it duplicates already published work, and whether or not the manuscript is sufficiently clear for publication. Reviewers will also be asked to indicate how interesting and significant the research is. The Editors will reach a decision based on these reports and, where necessary, they will consult with members of the Editorial Board.

For issue vol.6, no. 1 in 2026. we were privileged for the opportunity to announce excellent scientific lectures to be presented at the ISABS Conference on Forensic and Anthropological Genetics and Mayo Clinic Lectures in Individualized Medicine.

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Journal of Bioanthropology

PROGRAM AND ABSTRACTS

ISABS and Mayo Clinic

14th Conference: Advances in Application of Artificial Intelligence in Precision Medicine

June 15-19, 2026, Dubrovnik, Croatia

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ISABS International Affiliate Organization of AAFS AMERICAN ACADEMY OF FORENSIC SCIENCES

ISABS

PROGRAM AND ABSTRACTS

THE FOURTEENTH ISABS AND MAYO CLINIC CONFERENCE: ADVANCES IN APPLICATION OF ARTIFICIAL INTELLIGENCE IN PRECISION MEDICINE

JUNE 15-19, 2026
Hotel Dubrovnik Palace
Dubrovnik
CROATIA

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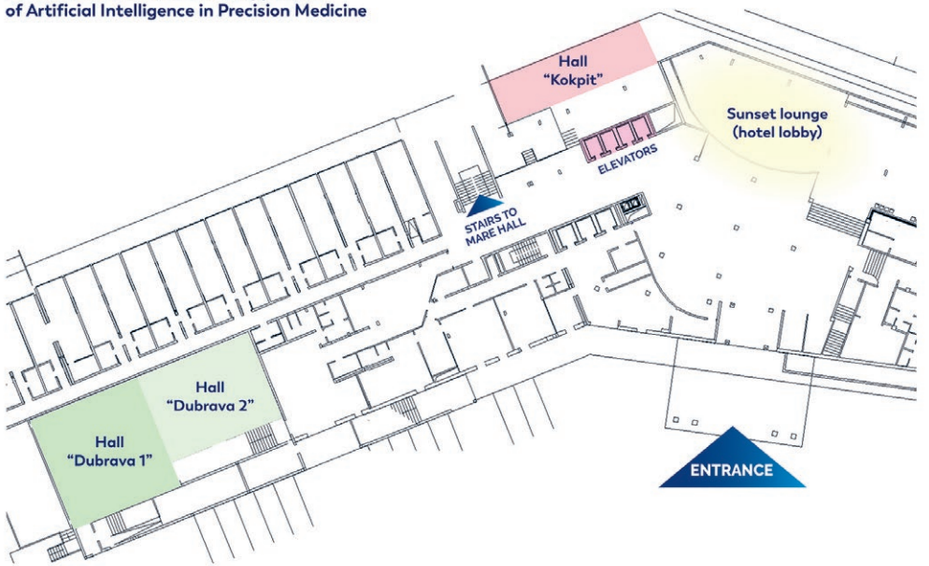
iSABS and MAYO CLINIC

14th Conference: Advances in Application of Artificial Intelligence in Precision Medicine

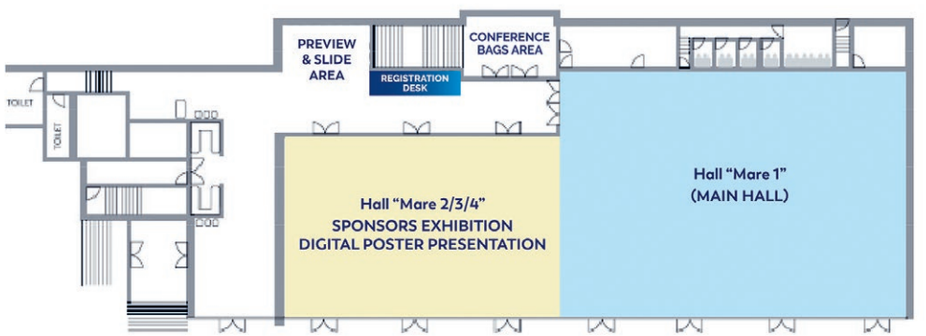
9th Floor



iSABS and MAYO CLINIC

14th Conference: Advances in Application of Artificial Intelligence in Precision Medicine

10th Floor



Hall	Sunday, June 14th	Monday, June 15th	Tuesday, June 16th	Wednesday, June 17th	Thursday, June 18th	Friday, June 19th	Saturday, June 20th
MARE 1		08:30–11:30 OPENING SCIENTIFIC BLOCK 13:00–14:00 NOBEL LAUREATE LECTURE 14:00–16:45 AFTERNOON SCIENTIFIC SESSION 16:45–18:30 PANEL DISCUSSION: Sudden Cardiac Death	08:30–11:30 MORNING SCIENTIFIC SESSION 13:00–14:00 ISABS SPECIAL LECTURE 14:00–15:50 AFTERNOON/EVENING SCIENTIFIC SESSION 16:00–17:00 ROUND-TABLE DISCUSSION: Too Much Too Soon?	08:30–10:45 MORNING SCIENTIFIC SESSION 13:00–14:00 NOBEL LAUREATE LECTURE 14:00–15:40 AFTERNOON SCIENTIFIC SESSION	08:30–10:35 MORNING SCIENTIFIC SESSION 13:00–14:00 NOBEL LAUREATE LECTURE 14:00–15:40 AFTERNOON SCIENTIFIC SESSION 19:30–21:30 ISABS SPECIAL EVENT: Garry Kasparov: The Chessboard of Artificial Intelligence and Human Creativity		
MARE 2/3/4		08:30–17:00 Sponsors Exhibition / Digital Poster Presentation	08:30–17:00 Sponsors Exhibition / Digital Poster Presentation	08:30–17:00 Sponsors Exhibition / Digital Poster Presentation	08:30–17:00 Sponsors Exhibition / Digital Poster Presentation 16:30–18:30 POSTER SESSION		
DUBRAVA 1		11:55–16:05 SATELLITE EVENT: 10th Croatian Human Genetics Conference & 3rd Croatian Personalized and Precision Medicine Conference				09:00–11:05 MORNING SCIENTIFIC SESSION: Twenty-Five Years of Inflammaging 11:05–12:00 GENERAL DISCUSSION CONFERENCE WRAP-UP	9:00–16:35 SATELLITE EVENT: Inflammaging in Clinical Practice
DUBRAVA 2		08:30–18:00 PROJECT CONFERENCE: Technology Transfer in Precision Medicine and Sports Injury Prevention (by invitation; project partners only)	11:00–13:00 JOINT EVENT by ISABS, Ministry of the Interior, American Academy of Forensic Sciences (AAFS), and University of Split, Faculty of Forensic Sciences				
KOKPIT		11:15–11:45 CSHG/CSPPM Managing Boards Meeting (by invitation) 14:00–18:00 SATTELITE EVENT: European Olympic Committees Medical Commission (by invitation) 18:00–19:00 CSHG Youth Section Meeting (by invitation)			11:00–13:00 SATELLITE EVENT: 2nd International Regenerative Medicine Experts Society (IARMES) Conference (by invitation)		
ROOFTOP		19:00–20:00 WELCOME RECEPTION					
SWIMMING POOL AND BEACH AREA		19:00–00:00 GALA DINNER, MOSES SCHANFIELD YOUNG INVESTIGATOR AWARD (MSYIA) AND FUTURE SCIENTIST AWARD (FSA) CEREMONY					
RECTOR'S PALACE					16:30–18:00 NOBEL SPIRIT: A nationally televised discussion with Nobel Laureates participating in the 14th Conference (by invitation)		

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WELCOME TO THE FOURTEENTH ISABS CONFERENCE ON APPLIED GENETICS AND MAYO CLINIC LECTURES IN TRANSLATIONAL MEDICINE!

Dear Colleague,

We invite you to join us at the 14th ISABS and Mayo Clinic Conference: Advances in Application of Artificial Intelligence in Precision Medicine, ***Dubrovnik, Croatia, June 15-19, 2026****. The conference is the next in a series of biennial events organized by the International Society for Applied Biological Sciences (ISABS), a society dedicated to promoting applied molecular biology (www.isabs.hr).*

Since initiating the series in 1997, we have strived to focus and broaden the scope of the conferences. The focus has been on applying cutting-edge analytical methodology in forensic science. In 2003, programs in forensic medicine and in cellular and molecular medicine ran in parallel with the introductory and closing sessions held jointly. The Cellular and Molecular Medicine program was co-organized with the Mayo Clinic, Rochester, Minnesota, USA. Since 2007, we have broadened the area of interest by the introduction of molecular anthropology that, in large part, shares the methodology with forensic genetics. In 2009, we introduced selected topics from individualized medicine, another applied discipline based on the advances in mapping the human genome. In 2011, we included the most recent and most interesting topics in molecular medicine. In 2013, we included genetics applied to the crossing of forensic science, anthropology, and translational medicine. Mayo Clinic again, same as in the past ten years, joined our efforts. It provided a critical link to the cutting-edge clinical applications of genetics. The overall effort culminated in the incorporation of individualized medicine as the third cornerstone, together with forensic and anthropological genetics. We feel this integration of the three areas, united by technology and applicative intent, provides an unprecedented opportunity for further progress in every respect. In the clinical part of the conference, topics covered the latest achievements in regenerative medicine, gene and cell therapy, individualized medicine, new molecular procedures, and methodology for early detection of cancer, the clinical importance of circulating tumor cells, and immune therapy in cancer treatments. In 2015, we introduced up-to-date results in genomics of individualized medicine, a program in anthropological genetics concerning ancient and modern human genome history, and human genetic history of the continents, and a forensic genetics program with special emphasis on new knowledge in Next Generation Sequencing (NGS) in forensics, DNA investigative intelligence, and advancements in forensic DNA routine. At the 10th ISABS conference's jubilee in Split in 2017, the Nobel Spirit session took place for the first time. This jubilee conference marked a breakthrough towards regenerative medicine and microbiome analysis in forensics, anthropology and medicine, and in 2019, we continued with the same "Nobel Spirit" achievements. The program in 2024 covered Whole Genome Sequencing for Implementation of Precision Medicine, Gene and Cellular Therapy, Immunotherapy, Pharmacogenomics, Tissue Engineering, Artificial Intelligence in Clinical Medicine, Dendritic Cell Vaccination, Forensic Genetic Genealogy, etc. "Nobel Spirit" provided a forum for the four Nobel laureates to stimulate public discussion on the role of science in solving global health issues, acute regional problems such as brain drain, demographic decline, and cultural and social change.

Together with several conference regulars, this year 2026, will bring many new and exciting names, including Nobel Prize laureates Gregg Semenza and Thomas Südhof. We are pleased that it will be more interesting than ever, and it will include the following topics: AI for Clinical Decision Support, AI-Powered Multi-Omics Integration for Precision Medicine – AI in Precision Oncology, AI-Driven Innovation in Tissue Engineering and Regenerative Therapies, and The Future of AI in Precision Medicine: Ethical, Legal, and Social Implications. Satellite Symposium, Moses Schanfield Memorial Symposium on Artificial Intelligence in Forensic and Anthropological Genetics includes topics: AI in Forensic Reconstruction, AI-Driven Forensic Genomics, AI for Ancestry Inference, and Justice by Design: Ethics of AI in Forensics.

This year, the fifth "Nobel Spirit" will provide a forum for the Nobel laureates to stimulate public discussion on advances in the application of artificial intelligence in precision medicine.

As before, the conference is structured to allow close interaction of the international faculty and attendees. We continue to have traditional Young Investigator Awards and High School Student Future Scientist Awards. Together with formal presentations, there will be meet-the-professor sessions, a gala dinner, and other social occasions meant to enhance opportunities for scientific intercourse, but also to introduce the participants to the town of Dubrovnik, one of the most prominent tourist destinations in the Mediterranean Sea; it carries the appellation of the pearl of the Adriatic. With its remarkable history, Dubrovnik is a city that leaves nobody unmoved, so delighted by its beauty, George Bernard Shaw said, "Those who seek paradise on Earth should come to Dubrovnik". Discover and enjoy the beauties of Croatia, the mild climate, the crystal clear and warm sea, beautiful beaches, virgin nature, and the rich history and cultural heritage. And, above all, enjoy Croatia's warm and friendly people.

We look forward to seeing you in Dubrovnik!

Dragan Primorac
Stanimir Vuk-Pavlović
Program/Conference Directors

Welcome

ABOUT DUBROVNIK

The over a thousand-year-old history of Dubrovnik is visible in every part of this city. The city is a living museum and a live stage, and has an ideal connection between its historical past and the present day. It is surrounded by medieval walls that are 1940 meters long and are preserved in their original form. They are open to visitors and are the city's greatest attraction. Since 1979, the town has been under UNESCO protection.

Dubrovnik is mainly a cultural destination, which, aside from monuments of interest, offers a series of cultural events and festivals. Dubrovnik is a destination where you can enjoy a rest, and it has extremely good air connections with all the larger European cities. Dubrovnik is a city that charms, a city that you fall in love with and always return to like new, to discover more unique experiences.

Church of St. Blaise
Constructed in 1715 in the flamboyant Venetian Baroque style, the Church of St. Blaise was designed by the Venetian master Marino Gropelli, commissioned by the Dubrovnik Senate to replace the old 14th-century Romanesque church.

Originally damaged by an earthquake, the church was then destroyed in a devastating fire in 1706. Everything perished in the flames except for the silver statue of St. Blaise, which was miraculously preserved. After years of exile in the Church of St. Nicholas at Prijeko, the statue was returned to its original place in 1715. The citizens of Dubrovnik added the following inscription: all other statues made of gold, silver, and bronze melted in the fire, while the saint's statue was miraculously undamaged.

This statue is among the most important in Dubrovnik, and the model of the city that the saint holds in his hand reveals the city's architecture of the time. St. Blaise has been honored as the patron saint of Dubrovnik since the 10th century.

Franciscan Monastery
On the western side of the city, protected by the city walls and the unconquerable Minčeta Fort, lies the Franciscan Monastery Male braće ("Little Brothers"). Together with the Dominican Monastery, it represents a cultural, artistic, and historical legacy of the Dubrovnik Republic.

The original Franciscan monastery was located outside the city walls in the suburb of Pile. However, due to the threat of war in the early 14th century, the Franciscans were forced to move inside the walls, demolish the old monastery, and build a new one within the protected city. Construction began in 1317 and continued for many years.

City walls
The most recognizable landmark that defines the physiognomy of historic Dubrovnik and gives the city its distinctive, world-renowned reputation is its remarkably preserved city walls, stretching a total length of 1,940 meters.

This imposing fortress system – one of the most beautiful and solid in the Mediterranean – comprises forts, bastions, casemates, towers, and freestanding fortresses. Built in times when the city and the Republic faced constant threats, the walls have survived not only thanks to the skill of their builders and the care of citizens who maintained and rebuilt them as needed, but also because of Dubrovnik's masterful diplomacy, which often succeeded in averting enemy aggression.

The Old Town is completely encircled by these fortifications, including the Old City Port. The history of Dubrovnik's defences dates back to the early Middle Ages.

Stradun
The famed Stradun, officially known as Placa, is Dubrovnik's favorite promenade – for locals, especially the young, and for visitors from around the world. A walk along Stradun is an essential experience, offering an unforgettable taste of the city's atmosphere.

Rebuilt after the great earthquake of 1667 as part of the accelerated reconstruction program, Stradun was given a wide and harmonious appearance, dignified yet beautiful in the simplicity of its stone architecture. Before the earthquake, it had been lined with elegant, luxurious palaces. After the disaster, however, the city's priority was survival and defense, and reconstruction focused on those needs.

All the houses along Stradun were built according to a plan approved by the Republic Senate. Uniform in height, with nearly identical facades and similar layouts, each house was required to include several shops on the ground floor – an enduring sign of the Republic's trading spirit.

14TH ISABS AND MAYO CLINIC CONFERENCE JUNE 15-19, 2026, DUBROVNIK, CROATIA

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URL: https://isabs.hr

Mayo Clinic

URL: https://www.mayoclinic.org

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International Center for Applied Biological Research

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ISABS is an International Affiliate Organization of the American Academy of Forensic Sciences (AAFS)

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Damir Marjanović (Institute for Anthropological Research, Zagreb, Croatia and International Burch University, Sarajevo, Bosnia and Herzegovina)
Henry Lee (†) (Henry C. Lee College of Criminal Justice and Forensic Sciences, University of New Haven and Henry C. Lee Institute of Forensic Science, West Haven, CT, USA)
Petar Projić (International Center for Applied Biological Sciences, Zagreb, Croatia)

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Johannes Brachman (Sana Kliniken, Coburg, Germany)

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Đurđica Kovačić (Medical School, University of Zagreb, Zagreb, Croatia)
Jelena Kovačić (Medical School, University of Zagreb, Zagreb, Croatia)
Jana Mešić (St. Catherine Specialty Hospital, Zagreb, Croatia)
Nika Miličević (Faculty of Science, University of Zagreb, Zagreb, Croatia)
Zvonimir Mlinarić (Faculty of Pharmacy and Biochemistry, University of Zagreb, Zagreb, Croatia)
Tonia Škoprc (Medical School, University of Split, Split, Croatia)

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Frederick Bieber (Harvard Medical School and Brigham and Women's Hospital, Boston, MA, USA)
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Mitchell Holland (Pennsylvania State University, State College, PA, USA)
Nenad Hrvatin (Clinical Hospital Centre Rijeka, Rijeka, Croatia)
Ante Ivkošić (Clinical Hospital "Sveti Duh; Zagreb, Croatia)
Natalija Novokmet (Institute for Anthropological Research, Zagreb, Croatia)
Petar Projić (International Center for Applied Biological Research, Zagreb, Croatia)
Vedrana Škaro (International Center for Applied Biological Research, Zagreb, Croatia)
Inga Urlić (Faculty of Science, University of Zagreb, Zagreb, Croatia)

AWARDS

Honorary Membership Award

Honorary membership award has been awarded to **Dr. Henry C. Lee in recognition of his outstanding work in the profession of the forensic sciences and continuous support to ISABS.** Dr. Lee was the director of Forensic Research and Training Center at the Henry C. Lee Institute of Forensic Science and Distinguished Chair Professor in Forensic Science at the University of New Haven. Lee was the Chief Emeritus for the Connecticut State Police during 2000 to 2010 and was the Commissioner of Public Safety for the State of Connecticut during 1998 to 2000 and has served as that state's Chief Criminalist and Director of State Police Forensic laboratory from 1978 to 2000.
Lee lectured widely, written hundreds of articles published in professional journals, and authored or co-authored more than 40 books on forensic science, crime scene investigation and crime scene reconstruction. He acted as an advisor or consultant to many law enforcement agencies. He hosted a show on the truTV network, formerly Court TV, titled Trace Evidence: The Case Files of Dr. Henry Lee, which highlighted his work on well-known cases. Lee has also appeared widely on television. He was a guest on the Taiwanese talk show KangXi Lai Le.

Moses Schanfield Young Investigator Award (MSYIA)

Knowing the importance of including young researchers as active participants of the conference, **ISABS encourages PhD and graduate students working toward a degree in the fields related to the conference program themes as well as postdoctoral researchers, to join the conference and apply for the Moses Scahnfield Young Investigator Award (MSYIA).** Therefore, for each ISABS conference, MSYIA are presented for outstanding research presented by investigators who are less than thirty-five years old. The scientific Advisory Committee of the conference selects finalists from nominees who submit abstracts. Besides the attractive prize, each award recipient is given the opportunity for a podium presentation. The author of each selected abstract receives the MSYIA Certificate as well.

The ISABS Future Scientist Award

A joint project of the International Society of Applied Biological Sciences (ISABS), the Ministry of Science and Education of the Republic of Croatia and the Croatian Education and Teacher Training Agency

The Scientific Committee of the International Society for Applied Biological Sciences (ISABS) in collaboration with the Croatian Education and Teacher Training Agency and the Croatian Ministry of Science and Education awards the best Croatian high school students' essays with The ISABS Future Scientist Award. The essays must be in the fields of human biology, genetics and chemistry, and infectious diseases (such as COVID-19). ISABS will also reward all teachers-mentors of the winning students.

Recipient of the 2026 ISABS Brave Heart Award

Ella Jovanović, Bosnia and Herzegovina, in recognition of exceptional courage and resilience

Recipients of the 2026 Moses Schanfield Young Investigator Award

Petar Brlek and Luka Bulić, Croatia (AI in Precision Oncology)
Petar Trović, Croatia (AI in Precision Medicine: Ethical, Legal, and Social Implications)

Recipients of the 2026 High School Student Future Scientist Award

Petra Cvitković and Klara Tabak, III. Gymnasium, Split, Croatia
Martina Kokić, Geodetic School Zagreb, Zagreb, Croatia
Irina Prša Krunić, X. Gymnasium Ivan Supek, Zagreb, Croatia
Ana Karla Vodanović, XV. Gymnasium, Zagreb, Croatia
Elena Weiser, I. Gymnasium, Varaždin, Croatia

Recipients of the 2024 Moses Schanfield Young Investigator Award

Floor Claessens, Denmark (Forensic Genetics)
Ivana Domljanović, Switzerland (Molecular Diagnostics)
Martina Glavan, USA (Personalized Medicine)
Vilim Molnar, Croatia (Personalized Medicine)
Illona Uzieliene, Lithuania (Molecular Diagnostics)

Recipients of the 2024 High School Student Future Scientist Award

Dorotea Alaburić, II. Gymnasium, Zagreb, Croatia
Klara Gnjidić, XV. Gymnasium, Zagreb, Croatia
Tončica Grubišić & **Vita Medić**, III. Gymnasium, Split, Croatia
Sara Polašek, School of Natural Sciences Vladimir Prelog, Zagreb, Croatia
Domagoj Praljak, XV. Gymnasium, Zagreb, Croatia
Jan Vlahinić, Andrija Mohorovičić Gymnasium, Rijeka, Croatia

Recipients of the 2022 Young Investigator Award

Elena Essel, Germany (Anthropological Genetics)
Karlo Miškec, Croatia (Epigenetics)
Amira Nabil, Egypt (Molecular Diagnostics)
Vid Matišić, Croatia (Pharmacogenomics)
Rachelle Turiello, USA (Forensic Genetics)

Recipients of the 2022 High School Student Future Scientist Award

Filip Bulat, VII. Gymnasium, Zagreb, Croatia
Ante Čolak, II. Gymnasium, Zagreb, Croatia
Mei Đulabić Chalfe, V. Gymnasium, Zagreb, Croatia
Robertina Filković, V. Gymnasium, Zagreb, Croatia
Lucija Glavičić Marović, XV. Gymnasium, Zagreb, Croatia

Đurđica Kovačić, III. Gymnasium, Split, Croatia
Nika Adriana Marijanović, Classical Gymnasium, Zagreb, Croatia
Frane Marušić, Jure Kaštelan High School, Omiš, Croatia
Nika Miličević, V. Gymnasium, Zagreb, Croatia
Goran Narančić, XV. Gymnasium, Zagreb, Croatia
Rea Pešušić, Vladimir Prelog Science School, Zagreb, Croatia

Recipients of the 2019 Young Investigator Awards

Viktoria Dotz, The Netherlands (Personalized Medicine)
Benjamin Planterose Jiménez, The Netherlands (Forensic Genetics)
Elena Zavala, Germany (Anthropological Genetics)

Recipients of the 2019 High School Student Future Scientist Award

Božo Bradarić Lisić, Vladimir Prelog Science School, Zagreb, Croatia
Matea Bürger, VII. Gymnasium, Zagreb, Croatia
Sara Caktaš, XV. Gymnasium, Zagreb, Croatia
Angela Kalpić, Medical School Split, Split, Croatia
Rea Pešušić, Vladimir Prelog Science School, Zagreb, Croatia
Petar Škrobo, M.A. Reljković Gymnasium, Vinkovci, Croatia

Recipients of the 2017 Young Investigator Awards

Sabriya Syed, USA (Personalised Medicine)
Goran Josipović and **Vladimir Zanki**, Croatia (Personalised Medicine)
Atina Vidaki, The Netherlands (Forensic Genetics)
Mateja Hajdinjak, Germany (Anthropological Genetics)

Recipients of the 2017 Future Scientist Award

Filip Bognar, XV. Gymnasium, Zagreb,Croatia
Lovro Jančić, Karlovac Gymnasium, Karlovac, Croatia
Rej Kovačević, VII. Gymnasium, Zagreb, Croatia
Lara Primorac, XV. Gymnasium, Zagreb, Croatia
Magda Topić, XV. Gymnasium, Zagreb, Croatia
Borna Branimir Vuković, V. Gymnasium, Zagreb, Croatia

Recipients of the 2015 Young Investigator Awards

Dora Polšek, Croatia (Individualized Medicine)
Barbara Zajac, Germany (Forensic Genetics)
Niraj Rai, India (Anthropological Genetics)

Recipients of the 2013 Young Investigator Awards

Matko Čančer, Sweden (Gene therapy)
Dora Markulin and **Branka Gršković**, Croatia (Genome-based applications in forensic science)
Slavé Petrovski, USA (Personalized genomics)
Antoinette Westen, The Netherlands (Genome-based applications in forensic science)

Recipients of the 2011 Young Investigator Awards

Rebecca S Just, USA (Genome-based applications in forensic science)
Mark Barash, Australia (Forensic DNA phenotyping)
Renato Polimanti, Italy (Molecular anthropology)
Martina Smolić, Croatia (Molecular therapy)

Recipients of the 2009 Young Investigator Awards

Chiara Barbieri, Germany (Molecular Anthropology)
Fernanda Gonçalves, Brasil (Individualised Medicine)
Pavlo Tatarskyy, Ukraine (Individualised Medicine)
Antoinette Westen, Netherlands (Forensic Genetics)

Recipients of the 2007 Young Investigator Award

Grzegorz Kaczmarczyk, Poland (Forensic Genetics)
Agnieszka Krzyżańska, Poland (Forensic Genetics)
Kaye Ballantyne, Australia (Molecular Anthropology)
Tomislav Domazet-Lošo, Croatia (Molecular Anthropology)
Coralie Frassati, Switzerland (Molecular Anthropology)
Taeko Kashima, Japan (Molecular Anthropology)

Recipients of the 2005 Young Investigator Award

Caroline Round, United Kingdom (Forensic Genetics)
Tracy Johnson, USA (Forensic Genetics)
Vedrana Montana, USA (Molecular and Cellular Medicine)
Mirela Baus Lončar, Germany (Molecular and Cellular Medicine)

Recipients of the 2003 Young Investigator Award

Robert J. Shelton, CO, USA (Forensic Genetics)
Chiara Magri, Italy (Molecular and Cellular Medicine)

Recipients of the 2001 Young Investigator Award

Forensic IdentityTesting: Frontiers in Molecular and Cellular Medicine:
Lucia Cifuentes Ovalle, Chile
Rima Dada, India
Katja Drobnič, Slovenia
Anna Gareeva, Russia
Nguyen Hoai Giang, Vietnam
Tomasz Kupiec, Polan

SCIENTIFIC PROGRAM INFORMATION

Visa and invitation letter

Please make sure you have the necessary valid documents to enter Croatia. It is the sole responsibility of the attendee to take care of their visa requirements. For more information, please check .
An official **invitation letter** can be sent upon request to isabs@etours.hr only for participants who have registered, paid the registration fee, and submitted an abstract that was not rejected. It does not cover any fees, nor is a guarantee to obtain the visa.
Participants should make their own arrangements with respect to **health and travel insurance**.

Badges

Participants, accompanying persons, exhibitors, and press will be given badges at registration. They will be required for admission to all conference facilities and scientific and social events during the conference. Security guards will check the badges at the conference venue. Any individual not wearing an official meeting badge will be directed to the registration desk to register or, if already registered, to purchase a replacement badge. The handling fee for replacement badges is €10.

Certificate of Attendance

Certificates of attendance will be issued at the registration desk.

Credits

The 14th ISABS and Mayo Clinic Conference has been approved for **20** (participants) or **30** (lecturers) points by the Croatian Medical Chamber. Credits are intended for medical doctors and members of the Croatian Medical Chamber to extend their medical doctor's license. If you are a Croatian Medical Chamber member and if you qualify for the credits approved for participation in the 14th ISABS and Mayo Clinic Conference, please enter your personal identification number during online registration.

Registration Desk Hours

Sunday, June 14th, 2026	10:00 AM – 8:00 PM
Monday, June 15th, 2026	10:00 AM – 8:00 PM
Tuesday, June 16th, 2026	7:30 AM – 5:00 PM
Wednesday, June 17th, 2026	7:30 AM – 5:00 PM
Thursday, June 18th, 2026	7:30 AM – 4:00 PM
Friday, June 19th, 2026	7:30 AM – 4:00 PM

Sponsor Exhibition

Setup: June 14th, 2026
Dismantling: June 19th, 2026 by 5:00 PM

Exhibit Hall Hours

Monday, June 15th, 2026	8:30 AM – 5:00 PM
Tuesday, June 16th, 2026	8:30 AM – 5:00 PM
Wednesday, June 17th, 2026	8:30 AM – 5:00 PM
Thursday, June 18th, 2026	8:30 AM – 3:00 PM
Friday, June 19th, 2026	8:30 AM – 3:00 PM

Program Changes

Organizers assume no liability for any changes in the program due to external or unforeseen circumstances. For any program changes, please check the updated Conference Program or ask at the Registration desk.

Language

The official language of the conference is English (no simultaneous translation)

Slide and PowerPoint Preview Area

A slide and PowerPoint preview area will be available to all presenters.

Info Center

The Info Center will be available at the registration desk. Service Centre provides photocopying, typing, and computer printouts at cost.

Smoking Policy

The 14th ISABS Conference is officially declared as a "Non-Smoking-Conference".

Special requirements

Registrants with special requirements for physical communication and dietary requirements should contact technical organizer in advance: isabs@etours.hr

Staff

If you should have any questions, the conference staff will be pleased to help you. It will be easy to recognize them by the special name badge they will be wearing.

e-Poster Information

E-poster presentations will be displayed at the conference in the e-poster area and in the ISABS 2026 mobile app.
Authors will have the opportunity to present their work to delegates during the **POSTER SESSION on Thursday, June 18th, 16:30–18:30**.
Scheduled presentation slots will be available on the mobile app from Sunday, June 14th, 2026.

Please note that the program is subject to change, so please check the updated Conference Program on the mobile app or ask at the Registration desk.

ONSITE ATTENDANCE

As the presenting author, your presence is requested onsite in Dubrovnik, and therefore, you must register for the Conference. Should you encounter any issues, please contact us at isabs@etours.hr

Please note that the program is subject to change, so please check the updated Conference Program or ask at the Registration desk.

ISABS is not responsible for material left after the Conference is over.

GENERAL INFORMATION

GSM OPERATORS

Currently, there are five GSM operators offering the GSM service in Croatia.
T-Mobile operating under +385 98 xxxxxx and +385 99 xxxxxx.
A1 operating under +385 91 xxxxxx.
TELE2 operating under +385 95 xxxxxx.
Tomato operating under +385 92 xxxxxx.
Bonbon operating under +385 97 xxxxxx.
Please contact your local GSM operator to check the availability and the costs of roaming services.

USEFUL TELEPHONE NUMBERS

Country code: (+/00) 385
Area code (Dubrovnik): (0)20
Emergency: 112
Police: 192
Fire-fighting center: 193
Emergency: 194
Time check: 18095
Information local calls: 18981
General information service: 18981
Information national and international calls: 11802
Wake-up calls: 18100
Roadside vehicle assistance: 1987
Weather forecast: 18166

OPENING HOURS

Banks and post offices are normally open from 7:00 or 7:30 a.m. to 7:00 or 8:00 p.m.
Non-Governmental offices work from 8:30 a.m. to 5:00 p.m., Monday to Friday.
Most grocery stores and department stores are open non-stop, from 6:00 or 7:30 a.m. to 7.30 or 8:00 p.m.

CURRENCY & EXCHANGE

The basic Croatian currency unit is the euro (EUR). Foreign currency can be exchanged for local money at banks, post offices, and exchange offices, according to the valid rates of exchange. Visit the exchange office on the Web for valid exchange rates.
Cash Machines: ATMs accepting all major bank cards and credit cards are located at numerous sites in Dubrovnik

CREDIT CARDS

All major credit cards are normally accepted throughout Croatia, as advertised at points of sale, such as Visa, Mastercard, Maestro, Diners Club

TAX-FREE

Foreigners can claim a sales tax refund within one year for purchased goods. Don't forget to ask the salesman to fill out the tax refund form when purchasing goods.

MISCELLANEOUS

Electricity Supply: 220-240 V, 50 Hz.

Water: Tap water is drinkable in all parts of Croatia.

Travel and Health Insurance: Participants are advised to make their own arrangements pertinent to health and travel. By registering for the 14th ISABS Conference on Applied Genetics and Mayo Clinic Lectures in Translational Medicine, participants agree that neither the organizers and its agents nor the sponsors and exhibitors nor the Hotel Dubrovnik Palace assume any liability whatsoever.

Taxi: Numerous taxi stands are located throughout Dubrovnik city center and in front of hotels. Hotel staff will be glad to help you.

INVITED SPEAKERS

Nobel Laureate Lectures:

Aaron Ciechanover (Nobel Prize in Chemistry 2004; Technion – Israel Institute of Technology, Haifa, Israel): TBA

Gregg Semenza (Nobel Prize in Physiology and Medicine 2019; John's Hopkins School of Medicine, Baltimore, MD, USA): (My) Life with Oxygen

Thomas Südhof (Nobel Prize in Physiology and Medicine 2013; University of Stanford, Palo Alto, CA, USA): My Path in Science Before and After the Prize.

ISABS Special Lecture:

Henry Erlich (Benioff UCSF Children's Hospital Oakland Research Institute, Oakland, CA, USA): From the Double Helix to the Genomics Information Explosion

ISABS Special Event:

Garry Kasparov (Office of Garry Kasparov, New York, NY, USA): The Chessboard of Artificial Intelligence and Human Creativity

14th ISABS and Mayo Clinic Conference: Advances in Application of Artificial Intelligence in Precision Medicine:

Itzhak Attia (Mayo Clinic, Rochester, MN, USA): From Screening to Action: AI for Detecting and Managing Asymptomatic Cardiac Disease

Veronique Belzil (Vanderbilt University Medical Center, Nashville, TN, USA): Revealing ALS Subtypes for Earlier Diagnosis Using Machine Learning

Richard Braatz (MIT, Cambridge, MA, USA): Artificial Intelligence and Machine Learning in Biomanufacturing

Gian Marco Conte (Mayo Clinic, Rochester, MN, USA): From Code to Clinic: Challenges and Opportunities in Translating AI into Radiology Practice

Antonio Esposito (San Raffaele Hospital and Vita-Salute San Raffaele University, Milan, Italy): Imaging-Driven AI Models for Personalized Cardiovascular Medicine

Claudio Franceschi (Alma Mater Studiorum, University of Bologna, Bologna, Italy): Twenty-Five Years of Inflammaging: It Is Time for Clinical Application

Alex Gotler (Teva Pharmaceuticals, Tel Aviv, Israel): Realizing the Promise of Digital Biomarkers: Challenges and Pharma's Role in Development

Steven Hart (Mayo Clinic, Rochester, MN, USA): AI in Pathology: Use Cases For and Against – A Framework for Preserving Brain Capital in the Diagnostic Apex

Vitaly Herasevich (Mayo Clinic, Rochester, MN, USA): From Paper Charts to Hospital-Wide Deterioration Detection: AMP, CEDAR, and Sepsis Surveillance

Yufei Huang (UPMC Hillman Cancer Center, University of Pittsburgh, Pittsburgh, PA, USA): Spatially Intelligent Oncology: Integrating Multimodal AI to Decode and Treat Tumors

David Jones (Mayo Clinic, Rochester, MN, USA): Treating the Complex Brain: How AI Enhances Expertise in Dementia Care

Manolis Kellis (Massachusetts Institute of Technology, The Broad Institute, Cambridge, MA, USA): AI Foundation Models for Genomic Medicine, Drug Development, and Personalized Therapeutics

Shelby Kutty (BayCare Health System, Clearwater, FL, USA): AI and Echocardiography for Cardiovascular Procedure Planning

Edward Laskowski (Mayo Clinic, Rochester, MN, USA): Too Much Too Soon? The Effect of AI on Critical Thinking and the Physician-Patient Relationship

Gordan Lauc (University of Zagreb, Faculty of Pharmacy and Biochemistry, Zagreb, Croatia; Genos Ltd., Zagreb, Croatia): immunoglobulin Glycans as Biomarkers and Functional Effectors of Inflammaging

Andrea Britta Maier (National University of Singapore, Singapore): Precision Geromedicine: Changing the Health Narrative

Shounak Majumder (Mayo Clinic, Rochester, MN, USA): AI Tools for the Diagnosis and Management of Pancreatic Diseases

Michal Melamed (Teva Pharmaceuticals, Tel Aviv, Israel): From Images to Insights: Using AI to Identify the Right Cells

Samia Mora (Brigham and Women's Hospital and Harvard Medical School, Boston, MA, USA): Reducing Inflammation to Prevent Cardiovascular Disease

Eskeatnaf Mulugeta (Erasmus University Medical Center Rotterdam, Rotterdam, Netherlands): AI Meets the Non-Coding Genome: Decoding Biological Function and Disease

Dennis Murphree (Mayo Clinic, Rochester, MN, USA): AI Advances in Dermatology

Falk Nimmerjahn (Friedrich-Alexander-Universität Erlangen-Nürnberg, Erlangen, Germany): The Sugar Switch: Pro- and Anti-Inflammatory Activities of IgG Regulated by Glycosylation

Dragan Primorac (St. Catherine Specialty Hospital, Zagreb, Croatia; Medical Schools, Universities of Split, Osijek and Rijeka, Croatia; University of Pittsburgh School of Medicine, Pittsburgh, PA; Eberly College of Science, The Pennsylvania State University, University Park, State College, PA, USA; The Henry C. Lee College of Criminal Justice and Forensic Sciences, University of New Haven, West Haven, CT, USA): Artificial Intelligence–Driven Multi Omics Comparison of Micro Fragmented Adipose Tissue (MFAT) and Hyaluronic Acid (HA) in Knee Osteoarthritis

Nataša Pržulj (Mohamed bin Zayed University of Artificial Intelligence, Abu Dhabi, UAE): AI for Mining Multi-Omics Data in Precision Medicine and Pharmacology

Juan Retamero (Tempus AI, Chicago, IL, USA): Reimagining Precision Oncology Through AI-Native Workflows

Santiago Romero-Brufau (Mayo Clinic, Rochester, MN, USA): Generative AI for Identifying Surgical Candidacy in Chronic Sinusitis

Parth S. Shah (Dartmouth Health, Lebanon, NH, USA): Enterprise Genomic Healthcare in the AI Era

Heath Skinner (UPMC Hillman Cancer Center, University of Pittsburgh, Pittsburgh, PA, USA): AI-Assisted Biomarker Development in Head and Neck Cancer

Natalia Trayanova (John's Hopkins University, Baltimore, MA, USA): Multimodal AI to Predict Risk of Sudden Cardiac Death

Carmen Terzic (Mayo Clinic, Rochester, MN, USA): AI-Enhanced Spine X-Ray Analysis for Predicting Metastases

Louis Vaickus (Dartmouth Health, Lebanon, NH, USA): Applications of AI in Pathology

Samuel Volchenboum (University of Chicago, Chicago, IL, USA): AI Won't Cure Childhood Cancer, But It Will Help the People Who Do

Mark Waddle (Mayo Clinic, Rochester, MN, USA): Real World Applications of AI in Radiation Oncology: Personalizing Treatment and Improving Efficiency

Moses Samuel Schanfield Memorial Session on Forensic Genetics

Bruce Budowle (University of Helsinki, Helsinki, Finland): Artificial Intelligence Enables Scale, Consistency, and Rigor in Forensic Identity Inference

John A. (Tony) Capra (University of California, San Francisco, CA, USA): AI as a Time Machine: Inferring Ancient Molecular Phenotypes

Mateja Hajdinjak (Max Planck Institute for Evolutionary Anthropology, Leipzig, Germany): Neandertals in Focus: What New Genomes Tell Us About Their Biology and Interactions With Humans

Mitchell Holland (Pennsylvania State University, State College, PA, USA): Probabilistic Intelligence for Mitochondrial DNA Mixture Deconvolution

Manfred Kayser (Erasmus University Medical Center Rotterdam, Rotterdam, Netherlands): AI-Driven Insights into the Genetic Basis of Human Facial Variation

Johannes Krause (Max Planck Institute for Evolutionary Anthropology, Leipzig, Germany): Genetic History of the Western Balkans and the Impact of Slavic Migrations

Ewelina Pośpiech (Pomeranian Medical University in Szczecin, Szczecin, Poland): Bridging Genomic and Epigenomic Data via Machine Learning for Forensic DNA Phenotyping

Antti Sajantila (University of Helsinki, Helsinki, Finland): Predicting Geolocation from Viral DNA Using AI

Joint Event by ISABS, Minstry of the Interior, American Academy of Forensic Sciences (AAFS), and University of Split, Faculty of Forensic Sciences

Crime Scene Investigation Training Course: "The 1996 Crash of U.S. Commerce Secretary Ron Brown's Aircraft in Croatia: Forensic Investigation, Identification, and Legacy":

Šimun Anđelinović (University Hospital of Split, Split, Croatia): Revisiting the CT-43 Crash Near Dubrovnik After Three Decades: Forensic Inquiry, Diplomatic Legacy, and Croatian–American Partnership

Ira Titunik (N/A, Sarasota, FL, USA): The Ron Brown Forensic Controversy

SCIENTIFIC PROGRAM

Please note that the program and speakers are subject to change.

Sunday, June 14th

10th Croatian Human Genetics Conference & 3rd Croatian Personalized and Precision Medicine Conference (Hall "Dubrava 1", 9th Floor)

11:15 **CSHG/CSPPM Managing Boards Meeting** (by invitation; members only) (Hall "Kokpit", 9th floor)

11:55 ***OPENING REMARKS*** (Dragan Primorac, chair)

12:00 **Scientific Session 1 – Dragan Primorac and Bernarda Lozić**, chairs

12:00 **Ivana Babić Božović** (Clinical Institute of Genomic Medicine, University Medical Centre Ljubljana, Ljubljana, Slovenia; Clinical Hospital Centre Rijeka, Rijeka, Croatia): *Genetic Diagnosis of Rare Neurological Disorders: Current Trends and Future Directions*

12:20 **Luka Bulić** (St. Catherine Specialty Hospital, Zagreb, Croatia): The Hidden Language of Cancer: *Predicting Clinical Outcomes Through Genomic Signals*

12:40 **Ljubica Odak** (Children's Hospital Zagreb, Zagreb, Croatia): *Clinical features, genetic landscape and management of osteogenesis imperfecta patients in Croatia*

13:00 **Ana-Maria Meašić** (Children's Hospital Zagreb, Zagreb, Croatia; Scientific Centre of Excellence for Reproductive and Regenerative Medicine (CERRM), University of Zagreb School of Medicine, Zagreb, Croatia; European Reference Network on congenital malformations and rare intellectual disability ERN-ITHACA, Zagreb, Croatia): *Genetic Causes of Lissencephaly: Insights from Chromosomal Microarray and Clinical/Whole-Exome Sequencing*

13:20 **Darko Antičević** (St. Catherine Specialty Hospital, Zagreb and School of Medicine, Josip Juraj Strossmayer University of Osijek, Osijek, Croatia): *Dysplasia epiphysealis hemimelica: Personalized and multiple treatment modalities for a single rare disease*

13:40 **Intermission**

14:00 **SATELLITE EVENT**: European Olympic Committees Medical Commission (by invitation only) (Hall "Kokpit", 9th floor)

14:00 **Scientific Session 2 – Dragan Primorac and Martina Smolić**, chairs (Hall "Dubrava 1", 9th floor)

14:00 **Kristian Hodak** (Clinical Hospital Centre Rijeka, Rijeka, Croatia): *Implementation of Next-Generation Sequencing in Routine Cardiology Practice: A Retrospective Study from Clinical Hospital Centre Rijeka*

14:20 **Tea Mladenić** (University of Rijeka, Faculty of Medicine, Rijeka, Croatia): *Artificial Intelligence in Cytogenetics: Progress in Automated Karyotype Analysis*

14:40 **Nenad Kunac** (University Hospital Center Split, Split, Croatia): *From Sequencing to Therapy: Implementing Comprehensive Genomic Profiling in Routine Clinical Practice*

15:00 **Damir Primorac** (Faculty of Forensic Sciences, University of Split, Split, Croatia): Legal Determinants in Medical Genetics

15:20 **Šimun Anđelinović** (University Hospital Center Split, Split, Croatia): *Croatia's Pioneering Role in First Systematic DNA-Based Identification of Mass Grave Victims*

15:40 **Damir Marjanović** (Institute for Anthropological Research, Zagreb, Croatia): Y Chromosome Story and Croatian Heritage: *Modern Croatian Genetic Pool Vs Ancient Genetic Data*

16:00 **Adjourn** (**Dragan Primorac**, chair)

18:00 **CSHG Youth Section Meeting** (by invitation; members only) (Hall "Kokpit", 9th floor)

08:30-18:00 **PROJECT CONFERENCE**: *Technology Transfer in Precision Medicine and Sports Injury Prevention* (by invitation; project partners only)

The project „*Biochemical and Genetic Pathways to Anterior Cruciate Ligament Injuries: A New Framework for Injury Prevention*" is funded through the "Targeted Scientific Research" call issued by the Ministry of Science, Education, and Youth of the Republic of Croatia under the National Recovery and Resilience Plan (NRRP) (Hall "Dubrava 2", 9th floor)

Monday, June 15th

AI Foundations in Clinical Reasoning & Patient Care

08:30 **Opening Scientific Block & Ceremony – Dragan Primorac and Stanimir Vuk-Pavlović**, Chairs
(Hall "Mare 1", 10th floor)

08:30 **WELCOME AND INTRODUCTORY REMARKS – Stanimir Vuk-Pavlović**

08:35 **Manolis Kellis** (Massachusetts Institute of Technology, The Broad Institute, Cambridge, MA, USA): *AI Foundation Models for Genomic Medicine, Drug Development, Protein Structure-to-Function, and Personalized Therapeutics*

09:05 **Intermission**

09:15 **OPENING CEREMONY & WELCOME ADDRESSES**

10:30 **John A. (Tony) Capra** (University of California, San Francisco, CA, USA): *AI as a Time Machine: Inferring Ancient Molecular Phenotypes*

11:00 **Vitaly Herasevich** (Mayo Clinic, Rochester, MN, USA): *From Paper Charts to Hospital-Wide Deterioration Detection: AMP, CEDAR, and Sepsis Surveillance*

11:30 **Informal networking / Lunch**

13:00 **NOBEL LAUREATE LECTURE: Gregg Semenza** (John's Hopkins School of Medicine, Baltimore, MD, USA): *(My) Life with Oxygen – Introduction*: Dragan Primorac (Hall "Mare 1", 10th floor)

14:00 **Afternoon Scientific Session – Gian Marco Conte**, Chair
(Hall "Mare 1", 10th floor)

14:00 **Edward Laskowski** (Mayo Clinic, Rochester, MN, USA): *Too Much Too Soon? The Impact of AI on Clinical Reasoning and the Physician-Patient Relationship*

14:25 **Gian Marco Conte** (Mayo Clinic, Rochester, MN, USA): From Code to Clinic: *Challenges and Opportunities in Translating AI into Radiology Practice*

14:50 **Petar Todorović** (School of Medicine, University of Split, Split, Croatia): *From Speed to Trust: The Governance of Artificial Intelligence-Assisted Knowledge Production in Precision Medicine* (Moses Schanfield Young Investigator Awardee)

15:00 **David Jones** (Mayo Clinic, Rochester, MN, USA): *Treating the Complex Brain: How AI Enhances Expertise in Dementia Care*

15:25 **Marijo Bekić** (General Hospital Dubrovnik, Dubrovnik, Croatia): *Preparing for Precision Medicine AI Deployment: A Fundamental Rights Impact Assessment Framework from a Croatian General Hospital* (Selected Oral Presentation)

15:35 **Samuel Volchenboum** (University of Chicago, Chicago, IL, USA): *AI Won't Cure Childhood Cancer, But It Will Help the People Who Do*

16:00 **Petar Brlek & Luka Bulić** (St. Catherine Specialty Hospital, Zagreb, Croatia): *Decoding Tumor Origin Using Artificial Intelligence: Transforming Diagnostic Strategies in Precision Oncology* (Moses Schanfield Young Investigator Awardee)

16:10 **Louis Vaickus** (Dartmouth Hitchcock Medical Center, Lebanon, NH, USA): *Applications of AI in Pathology*

16:35 **Noël Malod-Dognin** (Mohamed bin Zayed University of Artificial Intelligence, Abu Dhabi, UAE): *Exploiting the Linearity of Joint Embedding Spaces to Mine Cancer Precision Medicine Knowledge* (Selected Oral Presentation)

16:45 **Panel Discussion: Sudden Cardiac Death, Moderators: Dragan Primorac, Đorđe Alempijević, Stefano D'Errico, Johannes Brachmann**
(Hall "Mare 1", 10th floor)

16:45 **Opening and framing**

16:50 **Johannes Brachmann** (Sana Medical School, Coburg, Germany): *Sudden Cardiac Death: Clinical Aspects and Challenges*

17:05 **Adna Ašić** (Verlab Research Institute, Sarajevo, Bosnia and Herzegovina): *Towards Clinical Integration of CardioPharmaGENET Approach to Leveraging Cardiovascular Pharmacogenomics to Improve Risk Prediction, Prevention and Therapeutic Decision-Making in Sudden Cardiac Death*

17:20 **Djordje Alempijević** (University of Belgrade, Belgrade, Serbia): *Medico-Legal Death Investigation of Sudden and Unexpected Death: Focus on Sudden Cardiac Death*

17:35 **Stefano D'Errico** (University of Trieste, Trieste, Italy): *From the Friuli Venezia Giulia SCD Registry to a Balkan SCD Registry: Lessons Learned and the Way Forward*

17:50 **Dragan Primorac, Petar Brlek, Luka Bulić** (St. Catherine Specialty Hospital, Zagreb, Croatia): *Genetic Testing after Sudden Cardiac Death: From Casework to Family Prevention*

18:05 **Davide Radaelli** (University of Trieste, Trieste, Italy): *Metabolomic Profiling in Sudden Cardiac Death: Identification of Potential Biomarkers for Ischaemic Heart Disease*

18:20 **SCD Registries, Screening of High-Risk Groups and Personalised Medicine:** *Towards Regional Prevention Strategies*

19:00 **WELCOME RECEPTION**
(Rooftop)

Tuesday, June 16th

Imaging, Pattern Recognition & Phenotyping (Clinical & Forensic):
Carmen Terzic, Chair
(Hall "Mare 1", 10th floor)

08:30 **Carmen Terzic** (Mayo Clinic, Rochester, MN, USA): *Cutting-Edge Tools for Critical Diagnoses: Deep Learning and Large Language Models in Spinal Metastasis Detection*

08:55 **Antonio Esposito** (San Raffaele Hospital and Vita-Salute San Raffaele University, Milan, Italy): *Imaging-Driven AI Models: Personalizing Cardiovascular Medicine*

09:20 **Luka Bulić** (St. Catherine Specialty Hospital, Zagreb, Croatia): *Decoding Breast Cancer through Gene Expression: Preliminary Results from the Cancer Transcriptomics Intelligence Framework (CTIF)* (Selected Oral Presentation)

09:30 **Shelby Kutty** (Analytic Intelligence, Baltimore, MD, USA): *Artificial Intelligence and Echocardiography for Cardiovascular Procedure Planning: From Automated Quantification to Patient-Specific Digital Twins*

09:55 **Stephen Hart** (Mayo Clinic, Rochester, MN, USA): AI in Pathology: *Use Cases For and Against–A Framework for Preserving Brain Capital in the Diagnostic Apex*

10:20 **Željana Bašić** (Faculty of Forensic Sciences, University of Split, Split, Croatia): *Artificial Intelligence in Skeletal Biological Profiling: Toward an Interpretable and Automated Forensic Anthropology* (Selected Oral Presentation)

10:30 **Michal Melamed** (Teva Pharmaceuticals, Tel Aviv, Israel): *From Images to Insights: Using AI to Identify the Right Cells*

10:55 **Heath Skinner** (UPMC Hillman Cancer Center, University of Pittsburgh, Pittsburgh,PA, USA): *AI-Assisted Biomarker Development in Head and Neck Cancer*

11:20 **Hafiz Mohammad Hasan Babu** (Vanderbilt University Medical Center, Nashville, TN, USA): *Transforming Cancer Diagnosis: Quantum AI for Global Leukemia Screening* (Selected Oral Presentation)

11:30 **Informal networking / Lunch break**

11:00 **Joint Event by ISABS, Ministry of the Interior, American Academy of Forensic Sciences (AAFS), and University of Split, Faculty of Forensic Sciences**
Crime Scene Investigation Training Course: "*The 1996 Crash of U.S. Commerce Secretary Ron Brown's Aircraft in Croatia: Forensic Investigation, Identification, and Legacy*"
Ira Titunik (N/A, Sarasota, FL, USA) *The Ron Brown Forensic Controversy*

Šimun Anđelinović (University Hospital Center Split, Split, Croatia): *Revisiting the CT-43 Crash Near Dubrovnik After Three Decades: Forensic Inquiry, Diplomatic Legacy, and Croatian–American Partnership*
(Hall "Dubrava 2", 9th floor)

13:00 **ISABS SPECIAL LECTURE: Henry Erlich** (Benioff UCSF Children's Hospital Oakland Research Institute, Oakland, CA, USA): *From the Double Helix to the Genomics Information Explosion – Introduction*: **Stanimir Vuk-Pavlović** (Hall "Mare 1", 10th floor)

14:00 **Afternoon/Evening Scientific Session – Manfred Kayser**, Chair
(Hall "Mare 1", 10th floor)

14:00 **Yufei Huang** (UPMC Hillman Cancer Center, University of Pittsburgh, Pittsburgh, PA, USA): *Spatially Intelligent Oncology: Integrating Multimodal AI to Decode and Treat Tumors*

14:25 **Ewelina Pośpiech** (Pomeranian Medical University in Szczecin, Szczecin,Poland): *Bridging Genomic and Epigenomic Data via Machine Learning for Forensic DNA Phenotyping*

14:50 **Muhammed Erkan Karabekmez** (Istanbul Medeniyet University, Istanbul, Turkey): *Integration of Dietary and Multi-omics Data Reveals Microbiome Metabolism Interactions in Ankylosing Spondylitis* (Selected Oral Presentation)

15:00 **Manfred Kayser** (Erasmus University Medical Center Rotterdam, Rotterdam, Netherlands): *AI-Driven Insights into the Genetic Basis of Human Facial Variation*

15:25 **Antti Sajantila** (University of Helsinki, Helsinki, Finland): *Predicting Geolocation from Viral DNA Using AI*

16:00 **Round-Table Discussion**: *Too Much Too Soon?* **Edward Laskowski**, Moderator
(Hall "Mare 1", 10th floor)

19:00 **GALA DINNER, MOSES SCHANFIELD YOUNG INVESTIGATOR AWARD (MSYIA) AND FUTURE SCIENTIST AWARD (FSA) CEREMONY**
(Swimming pool and beach area)

Wednesday, June 17th

AI in Clinical Specialties & Diagnostic Decision Support – Vitaly Herasevich, (Chair (Hall "Mare 1", 10th floor)

08:30 **Dennis Murphree** (Mayo Clinic, Rochester, MN, USA): *An AI-Enabled Automated Triage System for Digital Dermatopathology*

08:55 **Shounak Majumder** (Mayo Clinic, Rochester, MN, USA): *AI Tools for the Diagnosis and Management of Pancreatic Diseases*

09:20 **Mark Waddle** (Mayo Clinic, Rochester, MN, USA): *Real-World Applications of AI in Radiation Oncology: Personalizing Treatment and Improving Efficiency*

09:45 **Juan Retamero** (Tempus AI, Chicago, IL, USA): *Reimagining Precision Oncology Through AI-Native Workflows*

10:10 **Ivana Domljanović** (Beets Health FlexCo, Vienna, Austria): *Beets Health: Framework for Exploring the Future of Preventive Health Using Wearables, AI, and Multi-Omics* (Selected Oral Presentation)

10:20 **Itzhak Attia** (Mayo Clinic, Rochester, MN, USA): From Screening to Action: *AI for Detecting and Managing Asymptomatic Cardiac Disease*

11:00 **Informal networking / Lunch break**

13:00 **NOBEL LAUREATE LECTURE: Aaron Ciechanover** (Technion – Israel Institute of Technology, Haifa, Israel): *TBA – Introduction:* **Stanimir Vuk-Pavlović** (Hall "Mare 1", 10th floor)

14:00 **Afternoon Scientific Session – Natalia Trayanova**, Chair (Hall "Mare 1", 10th floor)

14:00 **Natalia Trayanova** (John's Hopkins University, Baltimore, MA, USA): *Multimodal AI for Predicting Risk of Sudden Cardiac Death*

14:25 **Santiago Romero-Brufau** (Mayo Clinic, Rochester, MN, USA): *Using GenAI and Machine Learning to Identify Surgical Candidacy: An Otolaryngology – Head & Neck Use Case*

14:50 **Mitchell Holland** (Pennsylvania State University, State College, PA, USA): *Probabilistic Intelligence for Mitochondrial DNA Mixture Deconvolution*

15:15 **Eskeatnaf Mulugeta** (Erasmus University Medical Center Rotterdam, Rotterdam, Netherlands): *AI Meets the Non-Coding Genome: Decoding Biological Function +and Disease*

16:30 **NOBEL SPIRIT: A nationally televised discussion with Nobel Laureates participating in the 14th Conference** (by invitation only) (Rector's Palace)

18:00 **MEET THE PROFESSOR Small-group roundtables with conference speakers** (Hall "Mare 2/3/4", 10th floor)

Thursday, June 18th

Genomics, Multi-Omics, Molecular Medicine & Human Evolution: Nataša Pržulj, Chair (Hall "Mare 1", 10th floor)

08:30 **Nataša Pržulj** (Mohamed bin Zayed University of Artificial Intelligence, Abu Dhabi, UAE): *Multi-Omics Data Fusion for Precision Medicine and Precision Therapeutics*

08:55 **Veronique Belzil** (Vanderbilt University Medical Center, Nashville, TN, USA): *Data-Driven Discovery of Latent ALS Phenotypes through ICA and EHR-based AI Modeling*

09:20 **Parth S. Shah** (Dartmouth Health, Lebanon, NH, USA): *Advancing Precision Medicine through Integrative Genomics, Transcriptomics, Epigenomics, and Artificial Intelligence*

09:45 **Dragan Primorac** (St. Catherine Specialty Hospital, Zagreb, Croatia; Medical Schools, Universities of Split, Osijek and Rijeka, Croatia; University of Pittsburgh School of Medicine, Pittsburgh, PA; Eberly College of Science, The Pennsylvania State University, University Park, State College, PA, USA; The Henry C. Lee College of Criminal Justice and Forensic Sciences, University of New Haven, West Haven, CT, USA): *Artificial Intelligence–Driven Multi Omics Comparison of Micro Fragmented Adipose Tissue (MFAT) and Hyaluronic Acid (HA) in Knee Osteoarthritis*

10:10 **Bruce Budowle** (University of Helsinki, Helsinki, Finland): *Artificial Intelligence Enables Scale, Consistency, and Rigor in Forensic Identity Inference*

11:00 **Informal networking / Lunch break**

SATELLITE EVENT: 2nd International Regenerative Medicine Experts Society (IARMES) Conference (by invitation only) (Hall "Kokpit", 9th floor)

13:00 **NOBEL LAUREATE LECTURE: Thomas Südhof** (University of Stanford, Palo Alto, CA, USA): *My Path in Science Before and After the Prize – Introduction:* **Dragan Primorac** (Hall "Mare 1", 10th floor)

14:00 **Afternoon Scientific Session – Mateja Hajdinjak**, Chair (Hall "Mare 1", 10th floor)

14:00 **Johannes Krause** (Max Planck Institute for Evolutionary Anthropology, Leipzig, Germany): *The Genetic History of the Western Balkans and the Impact of Slavic Migrations*

14:25 **Mateja Hajdinjak** (Max Planck Institute for Evolutionary Anthropology, Leipzig, Germany): *Neandertals in Focus: What New Genomes Tell Us About Their Biology and Interactions with Humans*

14:50 **Alex Gotler** (Teva Pharmaceuticals, Tel Aviv, Israel): *Realizing the Promise of Digital Biomarkers: Challenges and Pharma's Role in Development*

15:15 **Richard Braatz** (Massachusetts Institute of Technology, The Broad Institute, Cambridge, MA, USA): *Artificial Intelligence and Machine Learning in Biomanufacturing*

16:30 **POSTER SESSION**
(Hall "Mare 2/3/4", 10th floor)

19:30 **ISABS SPECIAL EVENT: Garry Kasparov** (Office of Garry Kasparov, New York, NY, USA): *The Chessboard of Artificial Intelligence and Human Creativity – Introduction*: **Dragan Primorac** (Hall "Mare 1", 10th floor)

Friday, June 19th

Twenty-Five Years of Inflammaging, Gordan Lauc, Chair
(Hall "Dubrava", 9th floor)

09:00 **Claudio Franceschi** (University of Bologna, Bologna, Italy): *Inflammaging: It Is Time for Clinical Application*

09:25 **Falk Nimmerjahn** (Friedrich-Alexander-Universität Erlangen-Nürnberg, Germany): The Sugar Switch: *Pro- and Anti-Inflammatory Activities of IgG Regulated by Glycosylation*

09:50 **Gordan Lauc** (University of Zagreb, Faculty of Pharmacy and Biochemistry, Zagreb, Croatia; Genos Ltd., Zagreb, Croatia): *Immunoglobulin Glycans as Biomarkers and Functional Effectors of Inflammaging*

10:15 **Samia Mora** (Brigham and Women's Hospital and Harvard Medical School, Boston, MA, USA): *Reducing Inflammation to Prevent Cardiovascular Disease*

10:40 **Andrea Britta Maier** (National University of Singapore, Singapore): *Precision Geromedicine: Changing the Health Narrative*

11:05 **GENERAL DISCUSSION CONFERENCE WRAP-UP – Dragan Primorac and Stanimir Vuk-Pavlović**, Chairs (Hall "Dubrava", 9th floor)

12:00 **Departures**

Saturday, June 20th

Glycan Age
SATELLITE EVENT: Inflammaging in Clinical Practice

08:30 **Arrival & Registration**

09:00 **Opening Remarks: Gordan Lauc** (Glycomics & Immune Aging Expert)

MORNING PLENARY KEYNOTES

09:20 **Plenary Keynote:** *Antibodies as Biomarkers and Effector Molecules in Inflammaging*: **Falk Nimmerjahn** (Antibody and Immune Signalling Researcher)

09:40 **Plenary Keynote:** *Inflmaging Research*: **Claudio Franceschi** (Founder of Inflmaging Research)

10:00 **Coffee Break & Networking**

PLENARY TALKS & PANEL DISCUSSION

10:15 **Plenary Talk:** *Inflmaging as a Modifiable Driver of Aging: Lessons from Five Years of IgG Glycosylation Testing*: **Joseph Rafaelle** (Age-Management Medicine Specialist)

10:55 **Plenary Talk:** *Inflmaging: What's Broken-and How to Correct It:* **Mark Sherwood** (Functional and Metabolic Medicine)

11:15 **Panel Discussion:** *Therapeutic strategies to modify inflmaging – emphasis on sex hormone modulation*: **Flutura Hasa, Joseph Rafaelle, Mark Sherwood**

11:45 **Lunch Break**

CLINICAL CARDIOLOGY & METABOLIC MEDICINE

12:45 **Plenary Talk:** *Lifestyle and Preventive Medicine:* **Jeffrey Egler** (Lifestyle and Preventive Medicine)

13:05 **Plenary Talk:** *How to assess and treat inflmation in cardiovascular clinical practice*: **Samia Mora** (Preventive Cardiology and Metabolic Health)

13:25 **Plenary Talk:** *Rewriting Metabolic Medicine: pigenetics-Driven Precision Care for Cardiometabolic Disease*: **Saima Ajaz (**Metabolic Health and Inflmation)

13:45 **Panel Discussion:** Can weight loss modify inflmaging? Metabolic and immune perspectives: **Saima Ajaz, Samia Mora, Jeffrey Egler**

14:15 **Short Break & Networking**

BIOMARKERS, DIAGNOSTICS & PREVENTION

14:30 **Plenary Talk:** *From concept to clinical target: How do next-generation biomarkers redefine inflmaging in clinical practice?*: **Olga Donica** (Director of Longevity Innovation)

14:50 **Plenary Talk:** *Glycan Biomarkers and Translational Diagnostics*: **Ana Cvetko** (Biomedical Scientist & Glycan Biomarker Specialist)

15:10 **Plenary Talk:** *Inflmaging in Clinical Practice: Emerging Biomarkers and Translational Perspectives*: **Alberto Beretta** (Immunology and Longevity Medicine)

15:30 **Panel Discussion:** *Inflmation as a lifelong signal – From Primary to Secondary Prevention*: **Alberto Beretta, Ana Cvetko, Olga Donica**

16:00 **Short Break & Networking**

16:15 **Plenary Talk:** *From Inflmaging to Bioharmony: How Emotional Regulation Shapes Immune Ageing and Glycan Biology*: **Tamsin Lewis** (Longevity and Performance Medicine)

NOBEL LAUREATE SESSION

MY PATH IN SCIENCE BEFORE AND AFTER THE PRIZE

Südhof Thomas[1]

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In my presentation I will describe my career path, from working as postdoctoral fellow on cholesterol metabolism to studying how synapses manage to release neurotransmitters so precisely and rapidly. I will then describe the work that we have done more recently and that aims to elucidate the mechanisms of synapse assembly. Afterwards, I will discuss challenges that we are currently facing beyond trying to perform scientific studies, challenges that are inherent in the organization of science and amplified by the increased scrutiny of science by non-scientists who are not always valuing science as such. In this latter part of my talk I will in particular discuss how the commercial organization of the scientific publishing industry has led to distortions in science communications that have contributed to the public misperception of science and how science vigilantes who have no interest in scientific results but often pursue commercial interest have created an atmosphere of prosecution in science that has adversely affected in particular junior scientists. Finally, I will try to address true issues of science integrity that persist and that current mechanisms do not address because they are preoccupied with finding formal discrepancies, often of a minor nature, instead of examining the validity of a particular project.

Keywords: synaptic transmission, synapse assembly, cholesterol metabolism, scientific publishing, research integrity

(MY) LIFE WITH OXYGEN

Semenza Gregg[1]

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This is a story about a student inspired by Dr. Rose Nelson, his biology teacher at Sleepy Hollow High School; who learns as much as he can about genetics at Harvard College in between washing pots in the Dunster House kitchen; who tries several times to perform a successful research project at the University of Pennsylvania and obtains psychiatric advice from a classmate's father; who takes his lumps every third night as a pediatric resident at Duke Hospital; who starts an unsponsored research project at Johns Hopkins on this "other gene called *EPO*"; who stumbles upon a DNA binding protein he calls HIF-1; who spends the rest of his life trying to figure out what it does, how it does it, and how it might be targeted therapeutically; and, in his final hours, tries to convince a skeptical world that he might be on to something really important for cancer patients.

Keywords: teacher's influence, Harvard College, *EPO*, HIF-1

ABSTRACTS OF INVITED LECTURES – ISABS

Presentation number: IL 01 Abstract number: 147-ISABS-2026

FROM SCREENING TO ACTION: AI FOR DETECTING AND MANAGING ASYMPTOMATIC CARDIAC DISEASE

Attia Itzhak[1]

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Artificial intelligence (AI) is transforming cardiovascular medicine by expanding the role of the electrocardiogram (ECG) from a tool for rhythm interpretation to a scalable platform for disease detection, risk prediction, and population screening. In this presentation, we showcase how deep learning applied to raw ECG signals enables detection of conditions that are not traditionally identifiable from ECGs. We present AI models capable of identifying left ventricular systolic dysfunction, predicting atrial fibrillation even during normal sinus rhythm, and detecting systemic diseases such as liver cirrhosis. These approaches demonstrate strong diagnostic performance and highlight the ability of AI to extract clinically meaningful signals beyond human perception. We further discuss real-world implementation, including integration into clinical workflows and deployment at scale across primary care, wearable devices, and diverse global settings. Evidence from pragmatic clinical studies suggests that AI-guided screening can improve disease detection while maintaining efficient use of healthcare resources. Finally, we explore future directions, including multimodal AI, point-of-care imaging, and novel data sources such as voice, alongside key challenges related to validation, bias, and ethical deployment. Together, this work illustrates how AI can augment clinical decision-making and enable earlier, more effective detection of disease at scale.

Keywords: artificial intelligence (AI), electrocardiogram (ECG), deep learning, disease detection, risk prediction

Presentation number: IL 02 Abstract number: 161-ISABS-2026

DATA-DRIVEN DISCOVERY OF LATENT ALS PHENOTYPES THROUGH +ICA AND EHR-BASED AI MODELING

Belzil Veronique[1], Hoffman Mariah[1], Hucke Andre[2], Guidubaldi Joseph[2], Peltier Amanda[1], Below Jennifer[1], Landman Bennett[2], Lasko Thomas[1]

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Amyotrophic lateral sclerosis (ALS) is a fatal neurodegenerative disease characterized by progressive motor neuron loss, diagnostic delay, and substantial clinical heterogeneity. Current diagnostic and prognostic approaches rely largely on predefined clinical features and often fail to capture subtle, evolving patterns that precede diagnosis or distinguish biologically meaningful disease subgroups. We apply Independent Component Analysis (ICA) and supervised machine learning to large-scale electronic health record (EHR) datasets to identify latent ALS phenotypes that may improve early diagnosis, prognostic prediction, and patient stratification. We analyze two complementary EHR resources: the Vanderbilt University Medical Center Synthetic Derivative, which includes more than 3.9 million records and over 1,200 clinically confirmed ALS cases, and the NIH All of Us Research Program, a diverse national cohort with harmonized EHR and genomic data. Longitudinal clinical variables, including diagnoses, laboratory values, medications, and other structured features, are transformed into smoothed clinical trajectories suitable for ICA. ICA-derived components are then integrated into machine learning models to predict ALS diagnosis, disease progression, and survival, with model interpretability assessed using feature attribution approaches. Latent phenotypes are evaluated for associations with known clinical and genetic features, including ALS subtype, cognitive involvement, progression rate, and pathogenic variants. This work generates a scalable, data-driven framework for uncovering hidden clinical structure in ALS. By identifying early clinical signatures and biologically meaningful patient subgroups, this approach supports earlier diagnosis, more precise risk assessment, improved cohort enrichment for clinical trials, and more personalized care for individuals living with ALS.

Keywords: amyotrophic lateral sclerosis, electronic health records, independent component analysis, machine learning, precision medicine

Presentation number: IL 03 Abstract number: ABS-183-ISABS-2024

ARTIFICIAL INTELLIGENCE AND MACHINE LEARNING IN BIOMANUFACTURING

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This presentation discusses strategies for the application of artificial intelligence and machine learning (AI/ML) to bioprocess development and manufacturing. While machine learning methods can outperform traditional data analytics tools, a limitation of applying machine learning has been that the selection of the best ML method requires a substantial level of expertise. In practice, methods are chosen based on familiarity, which limits performance, or on cross-validation results from a large candidate model pool, which overfits data. Some reasons are discussed for why commercial Automated ML tools for building models fail when applied biomedical and bioprocess data. Then an AI/ML approach for building data driven models is described that empowers the users to focus on the goals of the intended application rather than on method selection. The approach is based on domain knowledge, the specific data characteristics, and next cross-validation procedures. For some industrial bioprocess datasets, the approach selects symbolic learning methods that produce more accurate models that are also interpretable by humans. The frontier of AI/ML methods, which is the incorporation of mechanistic knowledge, is also briefly discussed.

Keywords: artificial intelligence, machine learning, biomanufacturing, symbolic learning, interpretable machine learning

Presentation number: IL 04 Abstract number: 127-ISABS-2026

FROM CODE TO CLINIC: CHALLENGES AND OPPORTUNITIES IN TRANSLATING AI INTO RADIOLOGY PRACTICE

Conte Gian Marco[1]

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The integration of artificial intelligence into radiology practice presents challenges that extend well beyond model development and validation. This lecture addresses the gap between research-grade AI and clinically deployed tools, focusing on project prioritization, feasibility assessment, and organizational readiness. Drawing from operational experience building and deploying AI solutions in a large radiology department, we examine how the criteria used to evaluate project value differ fundamentally between a research context and a clinical translation context. In research, feasibility centers on data availability, model performance, and scientific novelty; in clinical translation, feasibility must also account for regulatory pathways, system integration, maintenance burden, and sustainable adoption. We describe how misapplying research -oriented prioritization criteria to clinical deployment decisions leads to resource misallocation, project stagnation, and a growing gap between what is technically possible and what is operationally viable. Our experience supports a structural distinction between discovery and validation work on the one hand, and implementation -ready projects on the other, each governed by distinct intake criteria, success metrics, and ownership pathways. We conclude that translating AI into radiology practice requires not only technical rigor but deliberate institutional design, including explicit triage frameworks, defined handoff structures, and a shared organizational language for what clinical readiness truly means.

Keywords: radiology AI, clinical deployment, AI translation, project prioritization, clinical feasibility

Presentation number: IL 05 Abstract number: 138-ISABS-2026

FROM THE DOUBLE HELIX TO THE GENOMICS INFORMATION EXPLOSION

Erlich Henry[1]

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The molecular biology era began with the publication in Nature of the Double Helix model by Watson and Crick and the resulting focus on how biological information is stored and transmitted. PCR uses the same strategy to amplify a target DNA sequence in vitro that the cell uses for DNA replication. The history of PCR development and applications to forensic genetics, medical genetics, and evolutionary genetics will be reviewed. The introduction of a heat stable DNA polymerase made PCR more specific, more efficient, and automatable. The ability to detect a single DNA molecule enabled the analysis of single sperm for measuring recombination, gene conversion, and de novo mutation, and enabled the clonal sequencing property of NGS. The massively parallel aspect of NGS has expanded genomic sequence information, but its clonal feature has made possible the analysis of mixtures. Moving from this historical review, our current project in non invasive prenatal testing by NGS analysis of maternal plasma (a mixture of fetal and maternal DNA) will be discussed. A plasma library encompassing 4 kb of the beta globin (HBB) gene region, prepared by capture hybridization, is sequenced (Illumina MiSeq). Counting single nucleotide polymorphism (SNP) allelic sequence reads yields an estimate for the fetal fraction (FF, typically 5-15%), and expected sequence read ratios at the mutation site for potential fetal genotypes, based on the FF, are compared to observed ratios to predict the genotype. Low FF and based solely on the mutation site limit statistical confidence. Since maternal sequence reads in plasma are slightly longer than fetal reads, we enrich the fetal fraction by excluding longer reads through in silico size selection. By PCR amplifying a 2.2 kb HBB fragment from parental DNA and sequencing the amplicon with the Oxford Nanopore platform, we determine haplotypes and use allelic ratios at linked SNPs to increase statistical confidence in fetal genotype predictions.

Keywords: PCR (Polymerase Chain Reaction), next-generation sequencing (NGS), fetal fraction (FF), haplotypes, beta globin (HBB) gene

Presentation number: IL 06 AAbstract number: 142-ISABS-2026

IMAGING-DRIVEN AI MODELS: PERSONALIZING CARDIOVASCULAR MEDICINE

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Ischaemic heart disease remains the leading cause of mortality worldwide. Coronary computed tomography angiography (CCTA) is the reference non-invasive modality for ruling out obstructive coronary artery disease (CAD) in symptomatic patients. However, in contemporary clinical practice, obstructive CAD is detected in only 15-25% of patients undergoing CCTA, leaving the vast majority classified within a heterogeneous spectrum of non-obstructive or subclinical coronary disease. This large and diverse patient population spans conditions ranging from the complete absence of detectable atherosclerosis to advanced coronary disease characterized by extensive plaque burden, adverse plaque phenotypes, vascular remodeling, and inflammatory activation. Importantly, cardiovascular risk in these patients is highly variable, ranging from negligible levels to a risk approaching or exceeding that of obstructive CAD. Conventional stenosis-based paradigms are insufficient to capture this complexity, underscoring the need for more refined, biologically informed risk stratification. Recent advances in artificial intelligence (AI) applied to CCTA have enabled the extraction of high-dimensional imaging biomarkers, including quantitative plaque burden, plaque composition, vessel wall remodeling, perivascular fat characterization, and hemodynamic descriptors. When integrated with clinical variables, laboratory biomarkers, genetic profiles, and environmental factors, these AI-derived imaging features allow the construction of individualized, predictive models of cardiovascular risk. Such imaging-driven AI frameworks represent a paradigm shift toward truly personalized cardiovascular medicine, enabling risk assessment and preventive strategies to be precisely targeted to the dominant pathophysiological mechanisms driving disease-related risk in each patient. By moving beyond luminal stenosis to a comprehensive, dynamic characterization of coronary atherosclerosis and vascular inflammation, AI-enhanced CCTA has the potential to transform prevention, guide therapy, and improve long-term cardiovascular outcomes.

Keywords: CAD, CCTA, atherosclerotic plaques, AI, prognosis

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INFLAMMAGING: IT IS TIME FOR CLINICAL APPLICATION

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Inflammaging describes a chronic, systemic, low-grade inflammatory state that is recognized as a major risk factor for age-related diseases (ARDs) and a pivotal convergence point of multiple biological mechanisms involved in aging. A major characteristic of inflammaging is its heterogeneity emerging as a consequence of each individual's lifelong exposures to inflammatory stimuli, shaped by a unique combination of genetics, lifestyle, socioeconomic conditions and environmental factors such as infections and pollution. Thus, a crucial problem is how to measure inflammaging. Two clocks addressed this topic. Both the first and the second clock are capable of quantifying inflammatory age and show strong association with ARD incidence, despite taking into account a different set of inflammatory markers. The second clock, based on just 10 immunological parameters (SImAge) and using explainable artificial intelligence methods, is able to explain the model solution for each individual participant. Moreover, we developed EpInflammAge, an explainable deep learning tool that integrates epigenetic and inflammatory markers to create a highly accurate, disease-sensitive biological age predictor. This novel approach bridges two key hallmarks of aging-epigenetic alterations and immunosenescence. Finally, we consider interventions that may counteract inflammaging, including nutritional interventions, physical activity and gerotherapies such as senolytics. We propose that deepening our knowledge of the individual nature of inflammaging stands to enhance our understanding of personalized aging trajectories and inform precision interventions.

Keywords: inflammaging, biological clocks, inflammatory age, explainable artificial intelligence

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REALIZING THE PROMISE OF DIGITAL BIOMARKERS: CHALLENGES AND PHARMA'S ROLE IN DEVELOPMENT

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Across many medical indications, disease diagnosis, monitoring of progression, and evaluation of treatment response are limited by the absence of clear, objective biomarkers. In their place, clinicians rely heavily on patient self report and clinical rating scales that, while indispensable, are subjective, prone to errors, sparsely sampled and present a burden on both patients and clinicians. This presentation examines digital biomarkers as a potential re imagining of how health and disease may be measured. Digital biomarkers leverage widely available technologies and sensors including smartphones, wearables, voice recordings, and video to capture continuous, objective signals of human physiology and behavior in real world settings. The talk introduces what digital biomarkers are, why they may meaningfully complement traditional clinical assessments, and how they may enable more sensitive, frequent, and personalized measurement. At the same time, their promise is balanced by substantial scientific and practical challenges, spanning clinical usability, interpretability, validation, and integration into pharmaceutical development. Rather than viewing digital biomarkers as a single end to end solution, this presentation frames them along a spectrum of clinical proximity, highlighting the importance of aligning specific digital measures with well defined components of the clinical phenomena. The presentation concludes with selected examples from Teva's ongoing digital biomarker initiatives across multiple disease areas, illustrating both tangible progress and the rigor required for successful translation. Together, these examples suggest how digital biomarkers may ultimately support earlier insight, more precise monitoring, and the delivery of better therapies for patients.

Keywords: digital biomarkers, clinical assessment, patient monitoring, digital health, drug development

Presentation number: IL 09 Abstract number: 151-ISABS-2026

FROM PAPER CHARTS TO HOSPITAL-WIDE DETERIORATION DETECTION: AMP, CEDAR, AND SEPSIS SURVEILLANCE

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The transition from paper-based charts to electronic medical records (EMRs) has unlocked unprecedented opportunities to develop real-time systems that continuously monitor hospitalized patients for clinical deterioration. This presentation reviews the historical evolution of electronic surveillance platforms, highlighting the development and implementation of AMP, CEDAR, and the Sepsis Sniffer at Mayo Clinic. Drawing on decades of experience in critical care informatics and clinical event detection, the presentation examines how real-time analytics, automated alerts, and AI-enhanced tools identify physiological decline across the hospital continuum. Given that sepsis remains a leading cause of morbidity and mortality, early recognition and rapid intervention are critical to improving patient outcomes. Finally, we will address the practical limitations of current approaches, focusing on workflow integration, model generalizability, and the barriers to clinician adoption.

Keywords: clinical deterioration, artificial intelligence, clinical surveillance, hospital-wide monitoring, decision support systems

Presentation number: IL 10 Abstract number: 126-ISABS-2026

AI IN PATHOLOGY: USE CASES FOR AND AGAINST – A FRAMEWORK FOR PRESERVING BRAIN CAPITAL IN THE DIAGNOSTIC APEX

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AI integration in pathology is hindered by a significant gap between technical validation and clinical adoption; many tools demonstrate strong performance metrics but fail clinically because they do not account for pathologist cognitive demands. This work presents a five-criterion calibration framework centered on preserving "brain capital" – the finite cognitive resources available to clinicians – and stabilizing the "diagnostic apex," where histological evidence meets clinical judgment. Using cognitive load theory and implementation science, we developed the Pathology AI Diagnostic Balance, a conceptual model mapping the physician-specimen interaction within working memory and IT infrastructure constraints, validated through case studies of implemented pathology AI tools. The framework identifies five evaluation criteria: Contextual Literacy (integration of clinical history), Responsibility (preservation of signing authority and prevention of chronic doubt through spatially localized outputs), Evidence Gap Analysis (closure of diagnostic uncertainty), Verification (feasibility and speed of quality control), and Opportunity Cost (net return on cognitive investment). Successful tools – such as Ki-67 quantification and breast cancer screening AI – reduce cognitive burden by automating routine tasks or enabling rapid verification; conversely, tools requiring exhaustive re-review or producing opaque recommendations deplete cognitive bandwidth and cause decision fatigue. This framework enables pathologists to assess whether AI tools genuinely enhance diagnostic capability or merely redistribute burden, shifting evaluation from purely technical metrics to clinically relevant measures of cognitive impact. By prioritizing cognitive sustainability and clinical utility over technical performance alone, the profession can ensure AI serves as a genuine diagnostic force multiplier rather than an additional burden.

Keywords: artificial intelligence, cognitive load theory, diagnostic workflow, pathology, precision medicine

Presentation number: IL 11 Abstract number: 123-ISABS-2026

SPATIALLY INTELLIGENT ONCOLOGY: INTEGRATING MULTIMODAL AI TO DECODE AND TREAT TUMORS

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Spatially resolved transcriptomics and proteomics are transforming oncology by revealing how tumor ecosystems are organized across tissue architecture, immune niches, stromal programs, and treatment-associated states. Yet extracting insight from these multimodal data remains challenging, requiring programming expertise, spatial statistics, visual interpretation, and iterative hypothesis refinement. We present spatiAlytica, a viewer-centric multi-agent AI system embedded in the Napari spatial viewer that enables natural-language, stateful, and image-grounded spatial biology analysis. spatiAlytica coordinates agents for data resolution, code generation, error recovery, spatial visual question answering, and biological interpretation, supported by memory across multi-turn workflows. Across benchmarks in single-turn code generation, sequential multi-turn analysis, and image-grounded reasoning, spatiAlytica outperformed existing agentic baselines. In oncology case studies, spatiAlytica decoded tumor biology across modalities and disease contexts. In Kaposi's sarcoma, it mapped stage-dependent tissue remodeling, viral tropism, niche composition, and immune dysfunction, revealing progressive CD8 T-cell transition from early immune surveillance to tissue-retained dysfunction. In colorectal cancer CODEX data, it identified subtype-specific immune niches and linked reduced PD1 -positive CD4 T-cell localization in granulocyte-enriched neighborhoods to poorer overall survival. In high-grade serous ovarian cancer Visium da ta, it characterized chemotherapy-associated cancer-associated fibroblast remodeling, showing how treatment reshapes the spatial tumor microenvironment. Together, these results support spatially intelligent oncology, where multimodal AI helps decode tumor ecosystems, identify clinically relevant spatial immune states, and generate hypotheses for therapeutic stratification and intervention.

Keywords: spatial biology, multi-agent system, multi-modal AI, Kaposi's sarcoma, colorectal cancer

Presentation number: IL 12 Abstract number: 140-ISABS-2026

TREATING THE COMPLEX BRAIN: HOW AI ENHANCES EXPERTISE IN DEMENTIA CARE

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Dementia diagnosis remains one of the most demanding tasks in clinical neurology. Overlapping syndromes, heterogeneous trajectories, and protein-centric diagnostic frameworks frequently obscure the underlying network failures that drive symptoms, leading to delayed or imprecise diagnosis, particularly outside specialized academic centers. The goal of this work is to demonstrate how AI can enhance, rather than replace, clinical expertise across the full dementia spectrum by reframing diagnosis around brain network dysfunction. We present StateViewer, an FDG-PET-based AI platform developed at Mayo Clinic and now advancing through an FDA Software as a Medical Device pathway. StateViewer was trained and validated on a large multi -center cohort spanning 14 dementia phenotypes including Alzheimer disease, dementia with Lewy bodies, frontotemporal dementia variants, and related disorders. The platform analyzes regional metabolic patterns and maps them onto phenotype -specific network signatures, producing interpretable outputs that support differential diagnosis at the point of care. Across validation cohorts, StateViewer achieved over 90 percent diagnostic accuracy, with performance stable across sites and scanners. Reader studies demonstrate that AI-assisted interpretation improves accuracy and confidence among both subspecialists and general neurologists, with the largest gains in atypical and early-stage cases. We further situate these findings within the Stochastic Latent Oscillatory Diffusion framework, which models dementia subtypes as selective failures of distinct computational components of a generative brain system, providing a mechanistic foundation for precision phenotyping. AI tools grounded in network neuroscience can extend specialist-level diagnostic reasoning into broader clinical practice and accelerate access to disease-modifying therapies.

Keywords: dementia, FDG-PET, Alzheimer's, brain networks, AI

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THE CHESSBOARD OF ARTIFICIAL INTELLIGENCE AND HUMAN CREATIVITY

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The ancient boardgame of chess has been considered a bellwether of intelligence, both human and machine, both rightly and wrongly, for centuries. It has been used as a testing ground and model for generations of psychologists and computer scientists, from Alfred Binet to Alan Turing. Chess has continued to be a useful drosophila melanogaster of cognition even today, almost 30 years since Garry Kasparov lost his famous rematch against the IBM supercomputer Deep Blue in 1997. One insight was that humans working together with intelligent machines using better collaborative processes produced superior outcomes compared to more knowledgeable humans or higher-powered computers. This was demonstrated in the Kasparov invention of Advanced Chess. In 2017, Demis Hassabis at Deep Mind's chess program AlphaZero demonstrated that reinforcement learning and virtualized data could produce an expert system – designs that went into AlphaFold, for which Hassabis received a 2024 Nobel Prize. With LLMs becoming the dominant face of AI and spreading with unprecedented speed, it is critical to keep humans in the chain of accountability and decision-making. Like any tech, from electricity to nuclear power, AI is not good or evil; it reflects its human creators and users. Human leadership and creativity are more important than ever in using these remarkable new tools to achieve great things. We must use it to augment our ambition and intelligence, not replace them, and to open up new challenges instead of simply performing old tasks more efficiently. In education, business, and everyday life, we must demand more from AI and its creators to emphasize human-centric goals and results.

Keywords: AI, artificial intelligence, chess, Deep Blue, cognition

Presentation number: IL 14 Abstract number: 142-ISABS-2026

AI FOUNDATION MODELS FOR GENOMIC MEDICINE, DRUG DEVELOPMENT, PROTEIN STRUCTURE-TO-FUNCTION, AND PERSONALIZED THERAPEUTICS

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Generative AI is fundamentally reshaping our understanding of biology, medicine, and therapeutics elevating AI from an analytical tool to a true discovery partner. Here, I will present our work building foundational AI models spanning chemistry, protein structure, gene function, patient states, and therapeutic interventions, towards an integrated agentic-AI platform for precision medicine. In the domain of protein structure to function, I will introduce ProCyon, our multimodal foundation model that combines protein sequences, molecular functions, disease associations, therapeutic mechanisms, and structural information into unified representations, which enable zero-shot phenotype annotation, drug-binding prediction, and functional interpretation of disease variants, opening the door to functionally annotating the dark proteome and guiding therapeutic targeting. In the field of chemical space modeling, I will describe our embedding of molecular structures, integrated with global patent databases, drug-target interaction knowledge, and protein function, to create a chemically and functionally interpretable drug landscape, enabling generation of novel molecules, functional annota-tion of the chemical landscape using co-embedded drug patents, and discovery of structure function relationships for drug discovery. In the area of patient trajectory modeling, I will present our latent embedding of patient states, built from multimodal data spanning omics, clinical records, imaging, and treatment histories, inferring representations that enable improved diagnosis, detection of misdiagnosed cases through cross modal consistency, and personalized intervention strategies, tailored to individual patients' trajectories relative to previously treated populations. In the domain of personalized therapeutics, I will describe our work leveraging foundational models connecting molecular, cellular, and patient-level phenotypes to predict therapeutic responses and uncover mechanistic disease subtypes, by integrating molecular function, single cell expression, protein structure function, and chemical spaces, and constructing AI-driven models capable of proposing causally grounded therapeutic recommendations at patient specific resolution. Finally, I will present our AI-powered, human directed visual data science workbench, Mantis, that enables scientists to seamlessly navigate and interrogate these latent embedding landscapes across proteins, chemicals, patients, and therapies, through interactive and interpretable visualizations, agentic workflows where human intuition guides AI actions, uncovering latent patterns and steering the next generation of generative models for biomedicine. Together, these efforts are driving a new paradigm for AI-enabled science, combining mechanistic interpretability, predictive power, and human-centered discovery to transform biomedical research and precision therapeutics.

Keywords: generative AI, precision medicine, foundation models, protein structure–function modeling, personalized therapeutics

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ARTIFICIAL INTELLIGENCE AND ECHOCARDIOGRAPHY FOR CARDIOVASCULAR PROCEDURE PLANNING: FROM AUTOMATED QUANTIFICATION TO PATIENT-SPECIFIC DIGITAL TWINS

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Goal: To synthesize how artificial intelligence (AI) applied to echocardiography can strengthen cardiovascular procedure planning by improving measurement standardization, risk stratification, multimodal model building, and device-aware simulation. Material and methods: We propose a procedural-planning pipeline for AI-enabled echocardiography encompassing acquisition support and image quality assessment, automated segmentation and standardized quantification from 2D/3D echo, multi-view and multimodal fusion with CT/MRI to improve geometric fidelity, machine learning-based phenotype discovery and outcome prediction to inform patient selection, and patient-specific 3D modeling with virtual device placement to enable "what-if" testing. Key implementation requirements include cross-vendor data harmonization, uncertainty quantification, prospective validation in clinical workflows, and governance addressing privacy, safety, bias, and regulatory constraints. Results: Across valve and heart failure interventions, AI-based echo analysis demonstrates feasibility for rapid, reproducible chamber and valve quantification and supports risk stratification and patient selection through phenogrouping and outcome prediction. Automated 3D segmentation from TEE/ICE can reduce analysis time and variability for structural planning (e.g., annular sizing) and enables patient-specific digital models. Device-aware simulation can quantify procedure-relevant risks such as neo-left ventricular outflow tract reduction during mitral interventions by evaluating virtual prosthesis position within individualized anatomy. Data quality remains a critical limiter; multimodal fusion and standardized acquisition improve planning fidelity. Conclusion: AI-enabled echocardiography is moving from workflow automation toward decision-grade procedural planning. By combining robust segmentation, standardized measurements, predictive modeling, and multimodal digital twins with rigorous validation and governance, AI can improve patient selection, procedural strategy, and safety for complex cardiovascular interventions.

Keywords: echocardiography, artificial intelligence, procedure planning, digital twin, multimodal imaging fusion

Presentation number: IL 16 Abstract number: 132-ISABS-2026

TOO MUCH, TOO SOON? THE IMPACT OF AI ON CLINICAL REASONING AND THE PHYSICIAN-PATIENT RELATIONSHIP

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Artificial intelligence (AI) is being rapidly integrated into medical practice across diagnostic, administrative, and documentation workflows. While AI promises efficiency gains and improved clinician experience, real - world evidence demonstrates that benefits are at times modest and highly dependent on governance, clinician oversight, and contextual integration. Recent multi-site observational and randomized studies of ambient AI documentation systems show limited time savings but substantial reductions in clinician burnout, cognitive load, and after-hours work. At the same time, technical limitations such as biased data, overfitting, and model drift intersect with cognitive risks including automation bias, alert fatigue, and erosion of clinical judgment. These challenges introduce ethical, legal, and regulatory complexities related to accountability, transparency, privacy, and patient autonomy. In addition, human factors research has shown that higher confidence in generative AI is associated with reduced critical thinking effort. Increased use of AI in clinical medicine also has the potential to reduce human interaction, increase reliance on AI for information gathering, and de-emphasize the physical examination, all of which can impact the physician patient relationship and the healing process in significant ways. This review synthesizes contemporary evidence on clinical AI applications, outlines major categories of risk, and provides best-practice recommendations for responsible deployment. Emphasis is placed on human-in-the-loop design, continuous validation, and professional stewardship, underscoring that AI in medicine should augment – rather than replace – clinical judgment.

Keywords: AI, critical thinking, clinical impact

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IMMUNOGLOBULIN GLYCANS AS BIOMARKERS AND FUNCTIONAL EFFECTORS OF INFLAMMAGING

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In the context of healthy aging, disease resilience – the capacity to withstand and rapidly recover from physiological stressors such as infection, injury, or chronic disease flares – emerges as a stronger determinant of outcome than disease prevention alone. As complete avoidance of such challenges becomes biologically implausible with advancing age, the difference between robust and frail trajectories hinges on an individual's ability to maintain physiological homeostasis under stress. Chronic low-grade inflammation ("inflammaging") is a central hallmark of diminished resilience. Glycosylation, the enzymatic attachment of complex glycans to proteins and lipids, is a key post-translational regulator of inflammatory pathways, immune function, and cellular stress responses. The human glycome integrates genetic, epigenetic, and environmental signals through hundreds of genes involved in glycan biosynthesis, exhibiting substantial heritability as a complex trait. Alternative glycosylation at individual sites dynamically modulates protein stability, receptor signalling, and immune recognition, thereby actively driving transitions from health to disease and frailty. Using high-throughput glycomics in >250,000 individuals across the lifecourse, we identified glycan signatures that robustly predict resilience to common age-related stressors. These signatures enable personalized risk stratification and reveal novel targets for pharmacological and lifestyle interventions aimed at preserving physiological reserve, attenuating inflammaging, and extending health span. Our findings position the glycome as a clinically actionable biomarker and therapeutic target for enhancing disease resilience and achieving compression of morbidity in aging populations.

Keywords: disease resilience, inflammaging, glycosylation, glycome, healthy aging

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PRECISION GEROMEDICINE: CHANGING THE HEALTH NARRATIVE

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Precision geromedicine is reshaping medicine by positioning the biology of aging, geroscience, as a primary, modifiable clinical target. Rather than focusing solely on the treatment of established disease, this emerging medical discipline aims to optimize function and intrinsic capacity across the adult life course through mechanism-based gerodiagnostics and matched gerointerventions. The field integrates advances in geroscience, systems biology, digital health, and clinical medicine into a translational framework capable of addressing multimorbidity before overt disease develops. Central to precision geromedicine is the development of robust gerodiagnostics. These include molecular, clinical and digital biomarkers of aging that together characterise individual ageing trajectories and underlying biological mechanisms. Epigenetic clocks, inflammatory signatures, clinical measures of all organs, physical and psychological performance metrics and longitudinal digital phenotyping are increasingly being integrated to define "gerotypes" and stratify individuals according to biological rather than chronological age. However, the rapid clinical adoption of these tools requires rigorous evidence building, analytical validation, standardisation, and regulatory oversight. Gerointerventions in precision geromedicine extend beyond single-drug approaches. Multimodal interventions combining exercise, nutrition, sleep optimisation, behavioural strategies, nutraceuticals, and repurposed pharmacological agents are being evaluated to target shared ageing pathways and improve multi-system outcomes. Importantly, the field is moving towards adaptive and personalised intervention strategies informed by longitudinal gerodiagnostic profiling. To establish precision geromedicine as a recognised medical speciality, evidence generation frameworks must evolve. Conventional randomised controlled trials remain essential, but need to be complemented by longitudinal registries, registry-based randomised clinical trials, and real-world evidence platforms embedded within clinical care. Such infrastructures enable continuous learning, evaluation of long-term outcomes, and assessment of interventions across diverse populations. The establishment of international standards, accredited education pathways, clinical guidelines, and dedicated regulatory frameworks will be critical to ensure scientific rigour, patient safety, and responsible implementation. Precision geromedicine therefore represents not only a new clinical discipline, but a fundamental shift in the health narrative: from reactive disease management towards proactive maintenance of lifelong optimized health and function.

Keywords: geroscience, precision, geromedicine, aging, gerodiagnostics

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AI TOOLS FOR THE DIAGNOSIS AND MANAGEMENT OF PANCREATIC DISEASES

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AI is rapidly transforming the diagnosis and management of pancreatic diseases. Early advances are most notable in the domain of pancreatic imaging, where CT-based radiomics models have shown potential for improving early pancreatic cancer detection, risk stratification of pancreatic cystic lesions and enhancing the identification of small, visually occult tumors. In one study, researchers trained a 3 -dimensional convolutional neural network (CNN) for detection of pancreatic cancer in diagnostic CT scans and evaluated its ability to detect cancer before a visible tumor was clinically diagnosed. The model could detect occult pancreatic cancer on prediagnostic CTs with AUROC 0.91 at a median 475 days prior to clinical diagnosis. AI-based imaging tools have also enabled automated analysis of body composition and pancreatic fat. For example, a CNN-based model for quantification of whole-pancreas fat, has overcome the limitation posed by the heterogeneous distribution of pancreatic fat, which renders partial sampling susceptible to error and subjectivity. More recent developments involve AI applications within the electronic health record for individualized extraction of specific disease -related risk factors. In one study, researchers developed a rule -based natural language processing algorithm to identify familial and germline pancreatic cancer risk from unstructured clinical notes. In a test set of 807 subjects, it achieved an F1-score of 0.85 for detecting family history of pancreatic cancer. Currently ongoing work in this domain focuses on the use of longitudinal health records to predict future disease trajectories. Despite rapid progress in the technological aspects, significant challenges remain regarding clinical implementation, adoption, and regulatory approval. As the field continues to advance, it will be crucial to ensure the use of high -quality input data for training and validating models and focus on robust evaluation frameworks and explainability.

Keywords: artificial intelligence, pancreatic cancer, pancreatic fat, natural language processing, image analysis

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FROM IMAGES TO INSIGHTS: USING AI TO IDENTIFY THE RIGHT CELLS

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The development of new medicines is a multi-stage R&D process that begins with early drug discovery, continues through preclinical animal testing, and advances to clinical trials in humans. In the early discovery stage, large numbers of potential drug candidates are explored using in silico and in vitro techniques, even though only a small fraction will ultimately progress to later pipeline stages. This creates a highly selective filtering process in which most of the time, labor, and cost are invested in candidates that will eventually be eliminated. In this work, we demonstrate how artificial intelligence (AI) can be used to improve efficiency in early drug development, with a focus on antibody discovery. We present a tool designed to support the early identification of antibody producing B -cells that are predicted to yield active and developable antibodies in later stages. The tool applies computer vision and machine learning (ML) techniques to analyze cell and assay images and rank candidate cells based on their predicted likelihood of downstream success. Deployed within Teva's drug development workflow, the system prioritizes candidate cell subsets enriched for higher predicted likelihood of downstream success. This enables earlier focus on promising candidates and reduces the time and effort spent on low potential options, improving timelines and supporting more efficient allocation of resources in early-stage antibody discovery.

Keywords: drug discovery, antibody discovery, artificial intelligence, machine learning, biologic therapeutics

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REDUCING INFLAMMATION FOR THE PREVENTION OF CARDIOVASCULAR DISEASE

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Inflammation is a key element of atherosclerotic cardiovascular disease (CVD), contributing to all of its stages, from plaque initiation to subsequent growth and eventual rupture. Circulating downstream biomarkers of systemic inflammation (such as the acute phase reactant C-reactive protein (CRP)) are strongly associated with atherothrombosis, risk factors, and CVD events, but these biomarkers are largely reflective of the overall inflammatory state, and may not drive CVD -associated inflammation. This presentation will provide an overview of the role of inflammation in cardiovascular events and discuss biomarkers of inflammation and CVD risk. Recent evidence demonstrating that the resolution of inflammation is not a passive process but occurs in an active coordinated process will be reviewed, along with a focus on important signalling molecules and mediators. Human experimental and observational studies have shown that glycan posttranslational protein modifications and bioactive lipid mediators are key factors and signaling molecules that initiate and sustain CVD-associated inflammation. Certain protein glycan modifications as well as bioactive lipid mediators (e.g. specialized pro-resolving mediators and omega-3 derived mediators) play a pivotal role in CVD-associated inflammation and could have important public health significance since residual inflammatory risk is common and remains undertreated with current therapeutics.

Keywords: glycans, glycomics, bioactive lipids, specialized pro-resolving mediators

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AI MEETS THE NON-CODING GENOME: DECODING BIOLOGICAL FUNCTION AND DISEASE

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The non-coding genome, which comprises ~98% of human DNA, was long considered "junk." This view has shifted substantially, as non-coding regions are now known to harbor diverse functional elements, including enhancers, silencers, and multiple classes of non-coding RNAs. These elements play central roles in gene regulation, shaping development, cellular identity, and disease states. Remarkably, up to 75% of the genome is transcribed, underscoring the extensive activity of the non-coding landscape. Despite advances in multi-omics technologies, identifying functional non-coding regions and interpreting the impact of variation within them remain major challenges. Genome-wide association studies (GWAS) have identified thousands of disease-associated single nucleotide polymorphisms (SNPs), the majority of which lie in non-coding regions and lack clear mechanistic links to gene regulation. Current diagnostic pipelines, including whole-genome sequencing, still prioritize protein-coding variants, leaving most non- coding genes and variants unexplored. Addressing this gap requires systematic functional annotation and computational frameworks capable of predicting regulatory activity. Our work aims to decode the functional non-coding genome using integrated multi-omics approaches combined with artificial intelligence. Using these strategies, we have identified novel long non-coding RNAs, mapped regulatory elements across cellular contexts, predicted enhancer activity directly from DNA sequence features, and pinpointed critical nucleotides required for regulatory function. Collectively, these efforts uncover previously unrecognized genetic mechanisms underlying development and disease, providing a framework for interpreting non- coding variation and helping to resolve the missing heritability of complex diseases.

Keywords: non-coding genome, long non-coding RNA, regulatory elements, machine learning, artificial intelligence

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AN AI-ENABLED AUTOMATED TRIAGE SYSTEM FOR DIGITAL DERMATOPATHOLOGY

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The rise of digital pathology and the routine digitization of histopathology slides has ushered in a new era of modern practice, creating opportunities for computational approaches that can support diagnostic workflows. This presentation will describe work developing an AI-enabled triage system that can categorize WSIs into broad diagnostic groups in order to improve case prioritization and efficiency. We constructed a dataset of 40,684 whole slide images (WSIs) from routine dermatopathology practice, categorized into nine physician-developed diagnostic classes. A pipeline leveraging the Atlas v1.1 foundation model generated embeddings from 224×224 pixel tissue tiles exhaustively cropped at 10x magnification (1.05 µm/pixel). A slide-level classifier employing attention-based multiple instance learning (ABMIL) was trained using the tile-level embeddings as input. The current best model achieves 80% overall accuracy and 0.74 macro F1- score on the held-out test set. Performance varied by category, with Basaloid (F1=0.90) and Squamous (F1=0.87) leading while Inflammatory conditions trailed (F1=0.65). This study demonstrates the feasibility of foundation-model-based triage systems for dermatopathology. The pipeline shows promise for optimizing case read efficiency and highlights the benefits of using AI-based tools to improve pathology workflow.

Keywords: digital pathology, dermatopathology, multiple instance learning (MIL), foundation models, whole slide imaging (WSI)

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THE SUGAR SWITCH: PRO- AND ANTI-INFLAMMATORY ACTIVITIES OF IGG REGULATED BY GLYCOSYLATION

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IgG antibodies are major drivers of inflammation but are also used in the form of intravenous immunoglobulins (IVIg) to suppress inflammatory processes. A key factor in modulating both, the pro- as well as the anti-inflammatory activity of IgG is the sugar domain attached to each IgG Fc-domain. The talk will discuss how specific IgG glycosylation variants affect IgG function and how this knowledge can be used to optimize the therapeutic activity of IgG antibodies in humans.

Keywords: IgG glycosylation, IVIg, therapeutic antibodies, inflammation, immunomodulation

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ARTIFICIAL INTELLIGENCE–DRIVEN MULTI OMICS COMPARISON OF MICRO FRAGMENTED ADIPOSE TISSUE (MFAT) AND HYALURONIC ACID (HA) IN KNEE OSTEOARTHRITIS

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Osteoarthritis (OA) is the most prevalent musculoskeletal disorder worldwide, affecting more than 528 million individuals. Although traditionally regarded as a cartilage disease driven by mechanical overload, OA is now recognized as a whole joint disorder involving coordinated pathological changes in cartilage, subchondral bone, synovium, and periarticular tissues, resulting in profound alterations in tissue architecture, metabolism, and function. The knee is the most commonly affected joint, followed by the hip and hand. Previous investigations at St. Catherine Specialty Hospital have demonstrated the therapeutic potential of mechanically micro fragmented adipose tissue (MFAT) in knee osteoarthritis (KOA). MFAT is enriched with CD45 negative cell populations, including endothelial progenitors, mature endothelial cells, pericytes, transitional pericytes, and supra adventitial adipose stromal cells, supporting angiogenic, immunomodulatory, and regenerative processes within the joint microenvironment. In this study, we evaluated the clinical, radiological, and biological effects of MFAT in patients with KOA. Clinical outcomes were assessed using validated pain and function questionnaires (VAS, WOMAC, and KOOS). Structural cartilage changes were investigated indirectly through assessment of glycosaminoglycan (GAG) content using delayed gadolinium enhanced magnetic resonance imaging of cartilage (dGEMRIC). In parallel, comprehensive molecular profiling of plasma and synovial fluid was performed, including analysis of cytokines, chemokines, N glycans, and microRNAs. Given the lack of disease modifying therapies for KOA, MFAT was further compared with intra articular hyaluronic acid (HA), a widely used standard treatment primarily providing mechanical and symptomatic benefits. From a master cohort, individuals with an inflammatory KOA phenotype were selected and treated with MFAT or with follow up at baseline, one month, and six months. Blood samples were analyzed using a multi omics framework, and the resulting datasets were integrated using an interpretable artificial intelligence model to identify treatment specific biological pathways. At six months, HA treated patients demonstrated persistent molecular signatures associated with inflammation and cartilage degradation. In contrast, MFAT treatment was associated with activation of pathways related to tissue repair, inflammation resolution, and cartilage protection. These findings suggest that, beyond symptomatic relief, MFAT may favorably remodel the biological milieu of the osteoarthritic joint, supporting its potential role as a disease modifying therapeutic strategy in inflammatory KOA.

Keywords: artificial intelligence; knee osteoarthritis; micro fragmented adipose tissue; hyaluronic acid; multi omics data integration; regenerative medicine

Abstracts of Invited Lectures

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MULTI-OMICS AI-DRIVEN DATA FUSION REVEALS DIVERGENT MOLECULAR SIGNATURES OF INTRA-ARTICULAR MICRO-FRAGMENTED ADIPOSE TISSUE AND HYALURONIC ACID TREATMENT IN INFLAMMATORY-PHENOTYPE KNEE OSTEOARTHRITIS

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Knee osteoarthritis (KOA) is a chronic, progressive joint disease with limited disease-modifying treatment options. In current clinical practice, intra-articular hyaluronic acid (HA) is widely used as a standard, primarily for its lubricating and shock-absorbing properties, providing symptomatic relief but with ongoing debate regarding its true biological impact on disease progression. In contrast, regenerative approaches such as micro-fragmented adipose tissue (MFAT), which contains mesenchymal stem cells, have shown superior clinical outcomes in selected patient groups. However, the biological mechanisms underlying these differences remain insufficiently understood. In this study, we aimed to compare the molecular effects of MFAT and HA in patients with the inflammatory phenotype of KOA, independent of clinical outcome scores. Out of a master cohort of over 18,000 patients, a total of 53 participants were selected based on specific inflammatory phenotype and treated with either MFAT (n=35) or HA (n=18) with a 6-month follow-up. Blood samples were analyzed using a multi-omics approach, including whole genome sequencing, microRNA expression profiling, proteomic analysis, and glycomic profiling. These data were integrated using an interpretable artificial intelligence framework to identify biological pathways that differ between treatments. At six months, clear differences emerged between the two groups. Patients treated with HA continued to show molecular patterns associated with ongoing joint degeneration and inflammation, including pathways linked to cartilage breakdown and inflammatory signaling. In contrast, MFAT-treated patients showed activation of pathways related to tissue repair, reduced inflammation, and cartilage protection. These findings were consistent across both gene-level and microRNA analyses. In conclusion, while HA remains a commonly used treatment for symptomatic management of KOA, our results suggest that its effects are largely mechanical and do not substantially alter the underlying molecular processes driving disease progression. In contrast, MFAT appears to shift the biological environment of the joint toward regeneration and inflammation resolution, supporting its potential as a disease-modifying therapy.

Keywords: artificial intelligence; knee osteoarthritis; multi-omics data fusion; regenerative medicine

Abstracts of Invited Lectures

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MULTI-OMICS DATA FUSION FOR PRECISION MEDICINE AND PRECISION THERAPEUTICS

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Large amounts of multi-omic data are increasingly becoming available. They provide complementary information about cells, tissues and diseases. We need to utilize them to better stratify patients into risk groups, discover new biomarkers and targets, re-purpose known and discover new drugs to personalize medical treatment. This is nontrivial, because of computational intractability of many underlying problems on large interconnected data (networks, or graphs), necessitating the development of new algorithms for finding approximate solutions (heuristics). We develop versatile artificial intelligence (AI) frameworks for multi-omics data fusion, constrained by the state-of-the-art network science methods, to address key challenges in precision medicine and pharmacology from time-series, multi-omics data, including patient-derived single-cell data, to better stratify patients, predict new biomarkers and targets, re-purpose existing and discover new drugs; we apply these to different types of cancer, Covid-19, Parkinson's, osteoarthritis, other diseases and longevity. Our new methods stem from graph-regularized non-negative matrix tri-factorization (NMTF), a machine learning (ML) technique for dimensionality reduction, inference, fusion and co-clustering of heterogeneous datasets, coupled with novel graphlet-based network science algorithms and generative models. We utilize our new frameworks for improving the understanding of the molecular organization of life and diseases from multi-omics data and to generate new precision therapeutics. The aim is to develop an overreaching framework encompassing all multi-omics data towards consumer-facing precision medicine and precision therapeutics products.

Keywords: multi-omics data fusion, precision medicine and therapeutics, network-based machine learning, biomarker and drug discovery, graph-regularized NMTF

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REIMAGINING PRECISION ONCOLOGY THROUGH AI-NATIVE WORKFLOWS

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Precision oncology has advanced rapidly through genomics and targeted therapies, yet clinical workflows remain fragmented, inefficient, and vulnerable to delays. A central challenge is transforming vast, heterogeneous data into timely, actionable decisions. AI in medicine is now evolving from isolated, single task tools into a connected infrastructure that supports the full continuum from diagnosis to treatment. Computational pathology and multimodal models are enabling integrated systems that span the entire cancer pathway. Emerging pathology foundation models can assist pathologists in diagnosis while simultaneously guiding oncologists by inferring molecular alterations, predicting prognosis, and estimating treatment response directly from tumor morphology on routine H&E slides. The lecture will explore the human-AI collaboration and examine how AI could function as the organizational operating system for cancer care by coordinating data, decisions, and clinical action across the diagnostic and therapeutic continuum.

Keywords: precision oncology, artificial intelligence, computational pathology, foundation models, clinical decision support

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USING GENAI AND MACHINE LEARNING TO IDENTIFY SURGICAL CANDIDACY: AN OTOLARYNGOLOGY – HEAD & NECK USE CASE

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Surgical departments at tertiary referral centers face a persistent mismatch between referral volume and clinical capacity. Efficient triage – matching the right patient to the right provider – is essential but labor- intensive, requiring clinician review of heterogeneous records. AI-based systems offer an opportunity to support this process at scale. We developed and deployed two AI-driven triage systems in the Department of Otorhinolaryngology at a tertiary academic medical center. For Head & Neck Surgery, a large language model identifies and extracts relevant documentation from the patient record, then evaluates findings against a physician-authored policy document specifying criteria for surgical consultation. The system outputs a triage recommendation including appointment appropriateness, prerequisite or completed workup, and supporting evidence. For Rhinology, an LLM first extracts structured clinical findings from cross-sectional imaging reports (CT/MRI); these extracted features then serve as inputs to a supervised machine learning model trained to predict whether the patient will undergo surgery within six months of imaging. Both systems are in clinical production, with prospective evaluation underway. We present retrospective validation results demonstrating strong performance. These deployments demonstrate that existing generative AI models, when paired with well-defined clinical criteria, are capable of meaningfully supporting clinical decision-making – and that the optimal integration pattern may vary by subspecialty context and available data.

Keywords: otolaryngology, large language models (LLMs), surgical decision support, machine learning, healthcare workflow optimization

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ADVANCING PRECISION MEDICINE THROUGH INTEGRATIVE GENOMICS, TRANSCRIPTOMICS, EPIGENOMICS, AND ARTIFICIAL INTELLIGENCE

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Advances in next-generation sequencing (NGS) and computational biology are rapidly redefining precision medicine across oncology, inherited disorders, and complex human diseases. The clinical integration of whole-exome sequencing (WES), long-read whole-genome sequencing (lrWGS), and RNA sequencing enables comprehensive characterization of somatic and germline variation, transcriptomic landscapes, and epigenetic alterations with unprecedented resolution. An integrative multi-omic perspective is transforming molecular diagnostics and personalized therapeutic decision-making. Emphasis is placed on the combined application of WES/WGS and transcriptome sequencing for improved detection of clinically actionable variants, fusion transcripts, and pathway dysregulation, as well as the growing role of methylome profiling using long-read WGS technologies capable of simultaneously resolving structural variation and native epigenetic signatures. A central challenge in precision oncology remains tumor heterogeneity, which drives disease evolution, therapeutic resistance, and variable clinical outcomes. Integrating genomic and epigenomic data with advanced computational approaches is increasingly essential for overcoming these limitations and refining patient stratification. The expanding role of artificial intelligence and machine learning in the interpretation of large-scale multi-omic datasets is enabling improved biomarker discovery, predictive modeling, and clinically actionable insights that are shaping the future of precision and personalized medicine.

Keywords: precision medicine, multi-omics, whole-genome sequencing, tumor heterogeneity, artificial intelligence

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AI-ASSISTED BIOMARKER DEVELOPMENT IN HEAD AND NECK CANCER

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Apart from human papillomavirus (HPV), head and neck squamous cell carcinoma (HNSCC) has few biologically driven biomarkers that guide curative-intent treatment. Standard clinicopathologic features explain only part of the variability in response and outcome, leaving patients and physicians to make difficult treatment decisions while balancing cure, toxicity, and treatment burden. AI-based methods may help address this problem by integrating clinical, omic, and imaging data into more useful biomarkers and decision-support tools. Here, we describe linked clinical and translational projects in HNSCC. Clinically, we are harmonizing patient-level data across existing cohorts and using prognostic and treatment-effect modeling to estimate expected outcomes after curative radiation-based therapy while incorporating patient preferences. Translationally, LLM-assisted analysis of omic datasets has identified candidate biomarkers connecting tumor-intrinsic metabolism with antitumor immunity and therapeutic response. Early validation studies have generated new hypotheses about how tumor metabolic state influences treatment vulnerability. Together, these projects support a practical path toward more personalized therapy in HNSCC.

Keywords: head and neck squamous cell carcinoma, precision oncology, multi-omics integration, artificial intelligence, biomarker discovery

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CUTTING-EDGE TOOLS FOR CRITICAL DIAGNOSES: DEEP LEARNING AND LARGE LANGUAGE MODELS IN SPINAL METASTASIS DETECTION

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The spine is the third most common site for metastatic disease, and radiographic detection remains challenging, with missed lesions risking pathologic fractures and spinal cord compression. Artificial intelligence (AI), particularly deep learning, has emerged as a promising decision-support tool by enabling automated lesion detection and segmentation, improving radiologist sensitivity while reducing interpretation time, differentiating metastatic from osteoporotic vertebral fractures, and facilitating early detection of metastatic spinal cord compression, with models reducing diagnostic delays by approximately 20 days. Meta-analytic data across imaging modalities report a pooled sensitivity of 88%, a specificity of 89%, and an AUC of 0.95. Despite these results, significant challenges remain: external validation AUC drops substantially compared to internal validation (0.819 vs. 0.947), and over half of published studies carry a high risk of bias. Multicenter prospective validation and standardized reporting frameworks are needed before widespread clinical adoption.

Keywords: artificial intelligence, spine metastasis, deep learning, imaging, diagnostic accuracy

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MULTIMODAL AI FOR PREDICTING RISK OF SUDDEN CARDIAC DEATH

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Sudden cardiac death (SCD) remains a leading cause of mortality worldwide, yet current risk stratification tools are inadequate, failing to identify a large proportion of individuals who would benefit from implantable cardioverter-defibrillator (ICD) therapy while exposing low-risk patients to unnecessary device implantation. Our team at Johns Hopkins University has pioneered a multimodal artificial intelligence framework that integrates complementary data streams to achieve unprecedented accuracy in predicting individualized SCD risk. Central to this approach is the fusion of cardiac imaging, electroanatomic data, and clinical variables within deep learning architectures. Late gadolinium enhancement MRI provides high-resolution characterization of myocardial scar geometry and heterogeneity, which serves as the structural substrate for life-threatening arrhythmias. These imaging features are combined with patient-level clinical data to train models that capture the multidimensional nature of arrhythmic risk. Validation across multicenter cohorts has demonstrated that the multimodal AI framework significantly outperforms established clinical criteria, including ejection fraction thresholds, in identifying patients at genuine risk of SCD. Importantly, the approach also identifies low-risk patients who may safely forgo ICD implantation, with meaningful implications for reducing procedural complications and healthcare costs. This approach represents a shift in cardiac risk stratification, moving from population-level guidelines toward precision, patient-specific prediction that integrates the full complexity of arrhythmic substrate.

Keywords: multimodal AI, imaging, sudden cardiac death, clinical variables

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APPLICATION OF AI TO PATHOLOGY

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The goal of this presentation is to demonstrate the applicability of artificial intelligence (AI) to the practice of pathology with an emphasis on anatomical pathology, whole slide images and multi -modal datasets. The presentation will review the conceptual space of digital whole slide images (WSI) and explore several diverse applications which have been developed at Dartmouth Health. The tasks which can be modeled using the features contained in WSI are surprisingly broad, despite the limitations of anatomic pathology tissue preparation techniques and WSI creation as they relate to dynamic biological processes. It is reasonable to attempt to create models for virtually any task which may be driven by tissue morphology regardless of whether the task is performable by a human.

Keywords: digital pathology, whole slide images, morphometry, multi-modal, machine learning, artificial intelligence

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AI WON'T CURE CHILDHOOD CANCER, BUT IT WILL HELP THE PEOPLE WHO DO

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Artificial intelligence is reshaping oncology – not as a distant promise, but as a set of tools already changing how tumors are detected, how patients are matched to therapies, and how clinicians make decisions under uncertainty. The goal of this presentation is to provide a clinically-grounded assessment of where AI is delivering impact in cancer care today and where critical gaps remain. Drawing on real-world deployments across radiology, pathology, and clinical decision support, as well as data from the Pediatric Cancer Data Commons, a harmonized repository spanning 46,000 patients across 200 institutions globally, this talk examines three domains: early detection, where deep learning models match or exceed specialist performance in identifying malignant findings; precision oncology, where integration of genomic and clinical data at scale reveals patient subgroups whose outcomes differ in ways invisible to single-institution analysis; and workflow automation, where continuously-running predictive algorithms flag deterioration and treatment toxicity before clinical thresholds are crossed. Results from deployed systems demonstrate both the potential and the limits of these tools: adversarial vulnerabilities, dataset shift, and cognitive biases baked into training data remain real constraints. The conclusion is that AI will not replace the clinicians who cure childhood cancer, but it will make them faster, more accurate, and better equipped to act on the right information at the right time.

Keywords: AI, pediatric oncology, bioinformatics, machine learning, deep learning

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REAL-WORLD APPLICATIONS OF AI IN RADIATION ONCOLOGY: PERSONALIZING TREATMENT AND IMPROVING EFFICIENCY

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Artificial intelligence (AI), particularly large language models (LLMs), is rapidly transforming radiation oncology by enabling more efficient workflows, enhanced data synthesis, and personalized patient care. This work highlights real-world clinical implementations of AI tools within a large academic radiation oncology practice, focusing on the impact on efficiency, quality, and decision-making. RadOnc-GPT, a Mayo Clinic AI-powered platform designed to address multiple data streams and "noise" present in modern electronic health records. By using RadOnc-GPT to access longitudinal oncology records with a multi-stage architecture, we can identify and synthesize clinically relevant information. This system supports multiple applications, including automated patient summaries, treatment documentation, and clinical decision support. One key implementation, "Daily Dose," generates concise, structured patient summaries by integrating data from electronic health records, treatment planning systems, and imaging platforms. Following deployment across more than 500 users, the system produces hundreds of summaries daily and saves an average of 11 minutes per clinician per day at low cost. Additionally, automated radiation treatment summary notes have eliminated manual workflows, reducing documentation burden while maintaining clinical accuracy. We further demonstrate AI applications in toxicity assessment through automated extraction of CTCAE adverse events from clinical notes and ambient audio transcripts. These approaches achieve performance comparable to human reviewers, with improved consistency and reduced inter-observer variability. Emerging tools such as OncoTimeline integrate AI-driven data visualization and interaction to support longitudinal patient management and decision-making. These real-world implementations illustrate how AI is enhancing efficiency, reducing administrative burden, and enabling personalized care in radiation oncology.

Keywords: artificial intelligence, radiation oncology, clinical workflows, large language models, personalization

ABSTRACTS OF INVITED LECTURES – MOSES SCHANFIELD MEMORIAL SYMPOSIUM

Presentation number: IL 37 Abstract number: 124-ISABS-2026

ARTIFICIAL INTELLIGENCE ENABLES SCALE, CONSISTENCY, AND RIGOR IN FORENSIC IDENTITY INFERENCE

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For more than two decades forensic science took a targeted approach of typing relatively small panels of short tandem repeat (STR) markers coupled with capillary electrophoresis for human identification purposes. However, this approach has limitations such as sensitivity of detection, particularly with highly degraded DNA samples, and resolution power only for direct comparisons and kinship analyses typically with first degree relationships. Massively parallel sequencing (MPS) and dense single nucleotide polymorphisms (SNPs) analyses greatly extends human identification capabilities. MPS coupled with forensic genetic genealogy (FGG) overcomes many of the limitations of STR typing. To establish potential kinship relationships, dense SNP data are searched against a database(s) of reference samples from consented volunteers. Associations are made primarily on identity -by-descent segment analysis, with the amount and total size of shared segments serving as indicators of genetic relationships. By FGG, searching for near and distant relatives greatly expands the range of cases in which DNA evidence can generate investigative leads. With these capabilities there is a need to go beyond predominantly human-centered workflows and limited hypothesis testing and instead embrace automation and capabilities to reason consistently , transparently, and at scale over increasingly complex genetic, genealogical, and contextual information. Artificial intelligence (AI) will be an enabling layer which is particularly suited for FGG as a computational decision-support system(s) that structures, prioritizes, and documents reasoning over genetic associations, genealogical structures, and investigative context during identity hypothesis development in a sustainably scaling manner. Incorporation of FGG and AI requires governance mechanisms that ensure transparency, accountability, privacy protection, and human oversight.

Keywords: FGG, forensic genetic genealogy, artificial intelligence, single nucleotide polymorphisms, kinship

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AI AS A TIME MACHINE: INFERRING ANCIENT MOLECULAR PHENOTYPES

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The sequencing of genomes from archaic hominins and modern humans has transformed our understanding of recent human history. However, due to the difficulty of inferring phenotypes from genotypes, ancient DNA has yielded few insights into the traits of ancient individuals. In this talk, I will describe how my group is using powerful machine learning methods to infer molecular phenotypes from ancient genetic sequences. I will illustrate how leveraging sequence-based deep neural networks can quantify protein structures, gene expression, splicing, and genome three -dimensional (3D) structure in archaic individuals. These analyses have revealed substantial similarities and differences between modern and ancient individuals that highlight molecular divergence in systems relevant to known phenotypic differences in the immune, metabolic, and skeletal systems. We identify specific loci where modern Eurasians have inherited novel molecular phenotypes from Neanderthal ancestors and show that these provide putative molecular mechanisms for phenotypes associated with the introgressed haplotypes. In summary, deep learning applied to ancient DNA sequences has great potential to reveal previously unobservable molecular differences between humans and our closest relatives.

Keywords: AI, human evolution, Neanderthals, ancient DNA, gene regulation

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NEANDERTALS IN FOCUS: WHAT NEW GENOMES TELL US ABOUT THEIR BIOLOGY AND INTERACTIONS WITH HUMANS

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Despite genome-wide data being recovered from close to 20,000 ancient humans to date, genomic data from Neandertals, our closest evolutionary relatives, are still comparatively sparse. Thus far, nuclear genetic data have been recovered from 35 Neandertals from 17 archaeological sites, spanning large parts of their history and geographical range. These data have offered a broad overview of Neandertal populations, indicating the existence of multiple distinct groups, as well repeated population turnovers. Archaeological and genetic evidence suggests that Neandertals lived in small groups, however, less is known if these groups were part of isolated communities or belonged to larger, well-connected populations. Here, I will present our recent efforts in reconstructing a more fine-scale view of Neandertal populations, focusing on some of the very last Neandertals from ten archaeological sites in Belgium and France. We used minimally destructive sampling of 36 skeletal remains which were radiocarbon dated to between ~52 and ~36 kya. Out of those, 27 contained enough endogenous DNA to generate autosomal, mitochondrial and Y-chromosomal data, including a new high-coverage genome from a 45,000-year-old Neandertal from Goyet. We found that late Neandertals from west Eurasia had much higher genetic diversity than the Neandertals from the Altai mountains, which lived between ~130 and 60 kya, suggesting that they lived in larger or better-connected groups. They also had fewer long tracts of homozygosity, comparable to those found in the Upper Palaeolithic humans present in Europe around the same time. And although these Neandertals overlapped temporally with early modern humans, we find no evidence of recent gene-flow from humans in their genomes. Moreover, we find no evidence for the accumulation of the deleterious mutations in the genomes of late Neandertals and no increase in genetic load over time, suggesting that genetics played a minor role in Neandertal extinction.

Keywords: neandertals, ancient DNA, admixture

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PROBABILISTIC INTELLIGENCE FOR MITOCHONDRIAL DNA MIXTURE DECONVOLUTION

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MixtureAce MT™ is a user environment for analysis of PCR-targeted mtDNA sequence data. The software runs in a cloud/Windows environment and accommodates fastq or bam files from MPS systems. Noise from sequencing error and sequences arising from nuclear mtDNA segments (NUMTs) are reduced. Contributor proportions and haplogroups are generated using the probabilistic technique of Vohr et al. [FSIG 2017]. Both the contributor proportions and haplogroup of each contributor is estimated simultaneously. Files previously generated were combined from two sources to form in-silico mixtures. GeneMarker HTS generated alignment files (bam files) for each sole-source contributor were used to create mixtures, combined in various ratios (1:1, 1:3, 1:9, 1:49, and 1:99) with each mixture consisting of a total 0.5, 1, 2, 5, or 9K read fragments. Contributor proportions and haplogroups were estimated simultaneously. The process is phylogenetically directed in that individual reads are assigned likelihoods of belonging to each individual haplogroup. The software identifies the best-supported haplogroups as those receiving the most support from read likelihoods and counts. MixtureAce MT™ reports the top-most supported haplogroups and the contribution proportion of the best-supported haplogroups. The haplogroup calls are substantiated by a listing of the phylogenetically diagnostic variant positions (diagnostic SNPs, single nucleotide polymorphisms) that point to the haplogroup call and provide a partial haplotype from the haplogroup defining SNPs. The points of view are those of the authors and do not reflect the views of Penn State University, West Virginia University, or the National Institute of Justice. Any mention of commercial products is done for scientific transparency and should not be viewed as an endorsement of the product or manufacturer.

Keywords: forensic genetics, mitochondrial DNA, haplogroup classification, MPS, probabilistic modeling

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AI-DRIVEN INSIGHTS INTO THE GENETIC BASIS OF HUMAN FACIAL VARIATION

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The enormous – near-individual-specific – variability of human faces has long intrigued researchers across diverse disciplines and the broader public. Although facial morphology is highly heritable, elucidating its genetic determinants is challenging due to the large phenotypic and underlying genetic complexity. In this presentation, I will summarize a series of collaborative efforts aimed at characterizing the genetic basis of human facial appearance, spanning from the first genome-wide association study (GWAS) that identified five genetic loci, via our recently published GWAS reporting almost 200 loci, to ongoing efforts, including those based on AI. I will demonstrate that, beyond the expected gains from increased sample sizes and expanded phenotypic characterization, the Combined GWAS (C-GWAS) framework we developed substantially enhanced locus discovery. I will present how these genetic insights can be leveraged to predict facial appearance from genetic data. I will show how AI-based facial phenotyping and C-GWAS of AI-derived facial traits enables the identification of additional loci not detectable from conventionally collected phenotypes. I will highlight how explainable AI allows the visualization of the identified genetic effects on the facial image. Finally, I will outline ongoing efforts to further refine the genetic understanding of facial variation through large-scale C-GWAS encompassing nearly one thousand facial traits in more than 50,000 individuals.

Keywords: face, genome-wide association study, genetic face prediction, AI-derived facial traits, explainable AI

Presentation number: IL 42 Abstract number: 166-ISABS-2026

THE GENETIC HISTORY OF THE WESTERN BALKANS AND THE GENETIC IMPACT OF THE SLAVIC MIGRATIONS

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The second half of the first millennium CE was a period of profound cultural, demographic, and political transformation across Central and Eastern Europe, commonly associated with the emergence and expansion of Slavic-speaking populations. While textual and archaeological evidence has long suggested large-scale changes during this period, the extent to which these developments were driven by migration, cultural diffusion, or Slavicisation has remained debated. Here, we synthesize recent genome-wide data from 555 ancient individuals, including 359 individuals from Slavic-associated archaeological contexts dating as early as the seventh century CE, to investigate the demographic impact of the Slavic migrations on the Western Balkans and wider Central-Eastern Europe. The results demonstrate substantial population movement originating from Eastern Europe between the sixth and eighth centuries CE, leading to the replacement of more than 80% of the local gene pool in regions such as Eastern Germany, Poland, and Croatia. Genetic evidence further reveals strong biological links among Slavic-period populations across geographically distant regions, including Croatia, Eastern Germany, and Poland–Northwestern Ukraine. These groups share extensive identical-by-descent (IBD) segments with one another while showing minimal genetic continuity with preceding local populations, supporting a recent shared origin and rapid demographic expansion. In contrast, Roman and Migration Period individuals from Croatia cluster genetically with present-day Italian and Eastern Mediterranean populations, highlighting the scale of the demographic transformation associated with the Slavic expansion. Despite this broad pattern of genetic turnover, significant regional variability is evident. The absence of strongly sex-biased admixture suggests that migration involved entire communities rather than predominantly male warrior groups, while varying levels of admixture indicate differing degrees of assimilation with local autochthonous populations. In Eastern Germany, genetic changes coincided with shifts in social organization, including intensified inter and intra-site relatedness and increased patrilocality. However, Slavic-period societies in the Northern Balkans appear to have retained aspects of earlier Migration Period social structures, suggesting that pre existing cultural practices persisted despite large-scale demographic and linguistic change. Overall, the integration of archaeological and genetic evidence supports the interpretation that the widespread cultural and linguistic transformations observed across Europe between the sixth and eighth centuries CE were closely linked to large-scale population movements associated with the Slavic migrations.

Keywords: population genetics, slavic migrations, ancient DNA, demographic transformation, central and eastern Europe

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BRIDGING GENOMIC AND EPIGENOMIC DATA VIA MACHINE LEARNING FOR FORENSIC DNA PHENOTYPING

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Forensic DNA Phenotyping (FDP) serves as an intelligence tool for perpetrator profiling when there are no suspects and no matches in national forensic DNA databases. The success of FDP became remarkable with the development of prediction models for eye, hair, and skin color based on the analysis of a finite set of SNP markers. Significant progress has also been achieved in predicting height, freckles, hair shape, and alopecia. However, the prediction of certain progressive appearance traits depends on age, which is an important factor in the models being developed. Notably, age can now be effectively estimated using DNA methylation analysis. In a study of ~1,000 individuals, we showed that incorporating age significantly improves hair graying prediction compared to SNP -only approaches. Integration of genetic and epigenetic data is also essential for facial skin aging traits, which are only partly heritable (~50%), with the remaining variation likely driven by epigenetic factors. Within the EPIGENOME project, using genome-wide SNP and methylation microarray data from >700 Polish individuals, we developed predictive models for wrinkle formation and perceived age (i.e., age estimated from facial appearance). These achieve an accuracy of ±0.4% of facial surface area for wrinkles and 3.3 years for perceived age. Since DNA methylation reflects both genetic and environmental influences, it also enables behavioral profiling. A promising example is BMI prediction. While existing models often rely on hundreds of variables, we developed a compact, forensically oriented model combining SNPs and CpG methylation, achieving an accuracy of ±2.9 BMI units. In summary, combining genomic and epigenomic data provides substantial opportunities for further development of FDP. At the same time, the role of machine learning methods, capable of handling large datasets in the context of relatively small sample sizes, is crucial for marker selection and model development.

Keywords: forensic DNA phenotyping, epigenetic and genetic data, facial skin aging, hair greying, prediction modelling

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PREDICTING GEOLOCATION FROM VIRAL DNA USING AI

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Persistent human DNA viruses are globally distributed, typically acquired early in life, and leave stable genomic traces in host tissues, thereby illustrating viral evolution and reflecting relationships among human populations. This viral “fingerprint” may complement human genomic data as a biogeographic marker, particularly in settings where reference datasets are limited or fragmented. We assembled 24,441 near-full-length genomes from NCBI GenBank, representing 29 human DNA viruses across five families (Polyomaviridae, Herpesviridae, Papillomaviridae, Parvoviridae, and Hepadnaviridae) with metadata extracted from database records and primary literature. Phylogenetic analyses were conducted using BEAST2, and machine -learning models were implemented through the mGPS framework to infer the geographic origin of viral sequences. Phylogenetic reconstructions revealed continent- and country-level clustering for numerous viruses, including BKPyV, JCPyV, HHV1, HHV2, HHV3, HHV6B, HCMV, HPV16, HPV18, HPV31, HPV45, and HBV. Geographic structure was strongest in regions with sufficient sequence representation, whereas global data availability remained highly uneven. Sixteen viruses met the sample-size criteria for mGPS modelling, yielding weighted F1 scores from 0.7663 to 0.9980, indicating robust continent-level predictivity despite dataset imbalance. Overall, several persistent human DNA viruses (14 of 29) show promise as biogeographic markers, offering a complementary tool for forensic identification and population studies. Integrating phylogenetics and machine learning enables inference of host geographic origin, although broader global sequence representation, improved metadata standards, and appropriate ethical frameworks will be essential for future applications.

Keywords: forensic genetics, geolocation, human DNA viruses, persistent DNA viruses, ancestry markers

ABSTRACTS OF INVITED LECTURES – AMERICAN ACADEMY OF FORENSIC SCIENCES

Presentation number: IL 45 Abstract number: 129-ISABS-2026

REVISITING THE CT-43 CRASH NEAR DUBROVNIK AFTER THREE DECADES: FORENSIC INQUIRY, DIPLOMATIC LEGACY, AND CROATIAN–AMERICAN PARTNERSHIP

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On 3 April 1996, a United States Air Force CT-43A aircraft carrying 35 occupants crashed into the mountainous terrain above Dubrovnik while on approach to the airport, resulting in the deaths of all those on board. For the United States, the accident claimed the life of the Secretary of Commerce, along with twelve members of an official government delegation, thirteen representatives of the business community, six members of the flight crew, and one journalist. For Croatia, the event meant the loss of a Croatian interpreter and journalist and transformed what had been planned as a diplomatic visit symbolizing partnership and support into a tragedy on Croatian territory. This article considers the CT43A accident not solely as an aviation disaster, but as a historical and forensic case positioned at the intersection of international diplomacy, national remembrance, investigative practice, and collective mourning. Using official aviation investigation findings, Croatian police and forensic records, U.S. governmental and congressional documentation, and contemporary commemorative materials, the study reconstructs the objectives of Secretary Brown's mission, the circumstances leading up to the crash, and the coordinated Croatian efforts in search, recovery, and victim identification. The article argues that the CT43A tragedy should be viewed beyond the technical and operational factors that led to the accident. Equal attention should be given to the broader diplomatic context of the mission and to the professional, humane, and organized response of Croatian institutions. On the occasion of its thirtieth anniversary, the event emerges as a significant chapter in Croatian-American relations and shows that forensic history can serve not only analytical purposes but also as a form of commemoration.

Keywords: CT-43A crash, aircraft accident investigation, DVI, Croatian-American relations, forensic history

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THE RON BROWN FORENSIC CONTROVERSY

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The April 3, 1996 crash of a U.S. Air Force CT-43 aircraft near Dubrovnik, Croatia resulted in the deaths of U.S. Secretary of Commerce Ron Brown and 34 others during an official trade mission. Although the official investigation concluded that pilot error, navigational deficiencies, and adverse environmental conditions caused the crash, unresolved forensic concerns regarding the postmortem investigation continue to generate discussion. The goal of this presentation is to retrospectively evaluate forensic procedures performed following the disaster and examine unresolved medicolegal questions surrounding the death investigation. Materials reviewed include publicly available aviation investigation findings, military forensic documentation, pathology reports, congressional correspondence, media archives, and records related to victim identification. Special attention is directed toward forensic odontology procedures used in victim identification, reported cranial injuries, the absence of a complete autopsy, missing radiographic evidence, and chain-of-custody concerns. Review of the available evidence demonstrates that while victim identification was completed and the official cause of death was reported as blunt force trauma, unanswered questions remain regarding documentation standards and forensic transparency. This case emphasizes the importance of comprehensive autopsy protocols, preservation of radiographic evidence, multidisciplinary forensic collaboration, and independent review in high-profile mass fatality investigations.

Keywords: Ron Brown, forensic odontology, aviation disaster, death investigation, mass fatality identification

ABSTRACTS SELECTED FOR ORAL COMMUNICATION

Presentation number: OC 01 Abstract number: 35-ISABS-2026

ARTIFICIAL INTELLIGENCE IN SKELETAL BIOLOGICAL PROFILING: TOWARD AN INTERPRETABLE AND AUTOMATED FORENSIC ANTHROPOLOGY

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The study aims to demonstrate a unified AI strategy for skeletal biological profiling across multiple anatomic elements and modalities. It addresses gaps in previous approaches, focuses on model performance and interpretability in real-world forensic applications, and bypasses the lack of documented skeletal collections in Europe. The main focus is biological profiling of the modern Croatian population using models developed from clinical MSCT images across 1,500+ specimens, including skulls, mandibles, hyoids, clavicles, and sterna. Deep learning segmentation enables automated extraction and organization of skeletal elements into structured digital datasets that mimic documented skeletal collections. AI models are trained on 2D skeletal images and 3D skeletal models, using convolutional neural networks to capture shape, size, and surface texture, while visualization tools (e.g., Grad-CAM activation maps, saliency maps) are systematically evaluated against domain expertise to confirm model reliance on anatomically meaningful regions. Sex estimation using fine-tuned VGG-16 and ResNet50V2 on 3D skull renders achieved accuracies of 93.5% and 92.5%, respectively (n=233). An adapted PointNet++ network applied to 3D hyoid point clouds achieved 88.7% test-set accuracy (MCC=0.77, n=202); mandibular models reached 92% test-set accuracy (n=254). Age estimation from medial clavicle images correctly classified the forensically critical 30-year threshold in 82.5–92.5% of cases. The models are deployed as open- access web applications for external validation and direct practitioner use. Ongoing work compares MSCT-derived models with smartphone photogrammetry and LiDAR-based alternatives to support field deployment without specialized equipment, with direct implications for forensic identification contexts where triage is required before considering confirmatory genetic evidence.

Keywords: artificial intelligence, forensic anthropology, MSCT, three-dimensional imaging, skeletal biological profiling

Presentation number: OC 02 Abstract number: 83-ISABS-2026

PREPARING FOR PRECISION MEDICINE AI DEPLOYMENT: A FUNDAMENTAL RIGHTS IMPACT ASSESSMENT FRAMEWORK FROM A CROATIAN GENERAL HOSPITAL

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Hospitals preparing to deploy AI-powered diagnostic imaging, clinical triage algorithms, and pharmacogenomic decision support – the precision medicine tools at the centre of this conference – face a regulatory prerequisite that remains largely unaddressed: the Fundamental Rights Impact Assessment (FRIA). Article 27 of the EU Artificial Intelligence Act (Regulation 2024/1689) requires every public hospital deploying high-risk clinical AI to assess its impact on patients' right to health, non - discrimination, human dignity, and equitable access to care before first use, with compliance required by August 2, 2026. No operational methodology for conducting FRIA in hospital settings has been published. We developed a six-phase framework at General Hospital Dubrovnik (308 beds, 18 departments), classified as an essential entity under the Croatian Cybersecurity Act (NIS2 transposition). The framework addresses: (1) regulatory classification of AI systems under Annex III, (2) identification of affected patient populations and vulnerability factors, (3) mapping fundamental rights at risk per clinical AI category, (4) algorithmic bias assessment against hospital-specific demographics, (5) definition of human oversight mechanisms preserving clinician decisional authority, and (6) documentation and reassessment governance. By mapping Article 27 requirements against existing institutional roles (CISO, DPO, clinical department heads, Cybersecurity Board), we demonstrated that 78% of FRIA operational elements can be fulfilled through governance structures already mandated by NIS2 and GDPR, without establishing a dedicated AI ethics committee. This pre-deployment framework offers a replicable model for secondary hospitals across Europe seeking to move precision medicine AI from research validation to compliant clinical deployment.

Keywords: fundamental rights impact assessment, EU AI Act, precision medicine, clinical AI governance, hospital cybersecurity

Presentation number: OC 03 Abstract number: 10-ISABS-2026

DECODING BREAST CANCER THROUGH GENE EXPRESSION: PRELIMINARY RESULTS FROM THE CANCER TRANSCRIPTOMICS INTELLIGENCE FRAMEWORK (CTIF)

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Breast cancer (BC) is a clinically and biologically heterogeneous disease characterized by marked variability in prognosis, treatment response, and molecular subtype. Hormone receptor status, including estrogen receptors (ER) and progesterone receptors (PR), as well as HER2 expression, play a central role in therapeutic decision-making and prognostics. Transcriptomic tumor profiling provides a comprehensive view of gene expression patterns, enabling deeper molecular characterization. In this study, machine learning (ML) was applied to high-dimensional transcriptomic data to predict BC clinical outcomes and immunophenotype, thereby advancing precision oncology and individualized risk stratification. This in silico study was conducted on transcriptomic BC data from TCGA, including 1215 samples and 20532 features. Based on transcriptomic features, unsupervised ML (UMAP + HDBSCAN) classified the samples into three distinct clusters. Supervised ML predictive modeling was conducted on raw transcriptomic features using the XGBoost algorithm, as well as on the three derived clusters using logistic regression (LR). Where clinical outcomes were concerned, XGBoost and LR achieved high accuracy scores for 5-year overall survival (88.1% vs. 89.3%), 5-year disease-specific survival (93.7% vs. 93.7%), 5-year disease-free interval (92.3% vs. 92.3%), and 5-year platinum-free interval (88.1% vs. 88.5%). Where hormone receptor status and HER2 expression were concerned, XGBoost and LR achieved high accuracy scores for ER positivity (92.6% vs. 89.6%), PR positivity (85.7% vs. 82.7%) and HER2 positivity (95.5% vs. 85.2%). Both XGBoost-based and LR-based predictive models performed well across all tasks, with XGBoost outperforming LR on immunophenotype prediction, and LR outperforming XGBoost on clinical outcome prediction. The results demonstrate a clear potential for BC clinical outcome and immunophenotype prediction through ML analysis of transcriptomic data.

Keywords: breast cancer, transcriptomic profiling, machine learning, artificial intelligence, outcome prediction

Presentation number: OC 04 Abstract number: 1-ISABS-2026

BEETS HEALTH: FRAMEWORK FOR EXPLORING THE FUTURE OF PREVENTIVE HEALTH USING WEARABLES, AI, AND MULTI-OMICS

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Digital biomarkers derived from wearables offer scalable opportunities for preventive health, yet their utility is limited by fragmented interpretation and weak integration with established biomedical reference frameworks. We present Beets Health, an AI-driven multi-omics platform designed to support precision preventive health by integrating 75 physiological, behavioral, cognitive, environmental, and psychosocial variables into a unified and interpretable health profile. The platform aggregates continuous wearable-derived data spanning cardiovascular function, metabolic health, autonomic regulation, sleep, physical activity, cognitive performance, lifestyle behaviors, and environmental exposure. Variables are mapped to age- and sex-adjusted reference ranges and categorized into risk strata based on epidemiological evidence, clinical guidelines, and large population datasets. These features are computationally combined into four resilience domains – Calm, Fuel, Perform, and Recharge – to summarize multidimensional health patterns and generate a composite health score, with an associated confidence metric that reflects data completeness and signal stability. In addition to physiological measures, the platform incorporates psychosocial indicators, including mood, social connectedness, and reflective behaviors, which are increasingly recognized as biologically relevant contributors to cardiometabolic and neurocognitive risk. Beets Health is designed for iterative multi-omics expansion, enabling the future integration of blood-based and epigenetic biomarkers to improve alignment between wearable-derived digital phenotypes and laboratory-based measures. This scalable AI framework aims to support early risk identification, personalized prevention strategies, and longitudinal health monitoring, facilitating the translation of continuous real-world data into clinically meaningful preventive insights.

Keywords: multi-omics, wearable biosensors, blood biomarkers, systems biology, artificial intelligence

Presentation number: OC 05 Abstract number: 68-ISABS-2026

INTEGRATION OF DIETARY AND MULTI-OMICS DATA REVEALS MICROBIOME METABOLISM INTERACTIONS IN ANKYLOSING SPONDYLITIS

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Ankylosing Spondylitis (AS) is a chronic inflammatory disease influenced by complex interactions between host genetics, environmental factors, and gut microbiota. While dietary modulation of the microbiome is increasingly recognized, the mechanistic links between diet, microbial metabolism, and immune regulation remain poorly understood. This study aims to leverage AI powered multi-omics integration to uncover microbiome diet host interactions and identify precision nutrition strategies in AS. We integrate metagenomic sequencing, dietary intake data derived from validated food frequency questionnaires, and genome-scale community metabolic models. The data integration frameworks are employed to link microbial composition, metabolic potential, and dietary patterns. Constraint-based modeling approaches, including flux balance analysis (FBA), flux variability analysis (FVA), and flux sampling, are used to simulate microbial metabolic responses to diet. Correlation and machine learning-based analyses are applied to identify associations between nutrients, microbial taxa, metabolic flux distributions, and inflammatory markers. Distinct microbial compositions and metabolic profiles were observed between AS patients and healthy controls. AI driven integration revealed key associations between specific dietary components, microbial metabolic pathways, and inflammation related features. Model based simulations suggested that diet dependent shifts in microbial metabolism may influence immune relevant metabolites, highlighting candidate pathways for intervention. This study demonstrates the potential of AI powered multi-omics integration combined with genome scale metabolic modeling to elucidate complex microbiome diet host interactions in AS. Our findings support the development of precision nutrition strategies targeting microbial metabolism to modulate inflammation. This integrative framework provides a scalable approach.

Keywords: ankylosing spondylitis, genome scale metabolic models, microbiota, precision nutrition

Presentation number: OC 06 Abstract number: 139-ISABS-2026

EXPLOITING THE LINEARITY OF JOINT EMBEDDING SPACES TO MINE CANCER PRECISION MEDICINE KNOWLEDGE

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Cancer is a leading cause of death worldwide. It is a multifactorial disease that results from multiple alterations, which are only partially captured by any biological data type studied in isolation (e.g., somatic mutation profiles or dysregulated genes). Hence, novel multi-omics data-fusion and analytics methods are needed to holistically mine the wealth of all available clinical and molecular data for new cancer precision medicine discovery. Network embedding is a cornerstone of modelling and analysis of such complex, biological datasets. In the field of NLP, it was observed that the embeddings of words capture semantic relationships linearly, allowing for efficient mining using simple linear vector operations rather than by using computationally intensive, black-box downstream analysis methods. Since then, this observation has been made in other fields, yielding the so-called linear representation hypothesis in vision language models. The question is whether this observation holds and can be exploited in the context of multi-omics data-fusion for cancer precision medicine. To assess this, we defined a Non-Negative Matrix Tri-Factorization based framework to jointly embed the multi-omics datasets of all cancer patients from TCGA (somatic mutation profiles and gene expression profiles of 9,134 cancer patients from 33 TCGA projects), the protein interactions from BioGRID and the drug data from DrugBank into the same embedding spaces. By comparing the performances of linear and non-linear methods for downstream classification and clustering tasks, we demonstrate that our joint embedding spaces are indeed linearly organized. Then, we define linear vector operations that enable prioritizing new pan-cancer or cancer-specific genes and drug repurposings, paving the way to new cancer therapies.

Keywords: AI/ML, multi-omics data-fusion, precision medicine, cancer gene prioritization, drug repurposing

MOSES SCHANFIELD YOUNG INVESTIGATOR AWARD PRESENTATIONS

Presentation number: MSYIA 01 Abstract number: 67-ISABS-2026

DECODING TUMOR ORIGIN USING ARTIFICIAL INTELLIGENCE: TRANSFORMING DIAGNOSTIC STRATEGIES IN PRECISION ONCOLOGY

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Cancers of unknown primary (CUP) remain a major clinical challenge due to limited diagnostic accuracy and the inability to apply optimal site-specific therapies, as most oncological treatment protocols are defined according to the primary tumor origin. In the era of precision oncology, characterized by the increasing use of whole-genome sequencing and artificial intelligence, we aimed to develop and validate an AI-driven approach for predicting the primary tumor site based on comprehensive genomic profiling, with the objective of improving diagnostic accuracy and supporting clinical decision-making. An in silico diagnostic study was conducted using publicly available datasets comprising over 20,000 metastatic tumor samples with annotated clinical variables and mutational profiles across more than 600 cancer-related genes. Multiple machine learning algorithms were trained and evaluated for multi-class tumor site classification, with model performance assessed using cross-validation and an independent test cohort. The optimal model was implemented into a software platform with an integrated graphical user interface (GUI) to facilitate clinical application. Among the evaluated models, the XGBoost-based classifier demonstrated superior performance, achieving a top-2 accuracy of 0.91 and an ROC-AUC of 0.97, with consistent generalizability across datasets. Feature importance analysis identified biologically relevant genomic patterns contributing to tumor classification. This approach enables more accurate tumor origin prediction and supports more informed, site-directed therapeutic decision-making, particularly in diagnostically challenging cases such as CUP. The developed platform represents a step toward translating complex genomic data into clinically actionable insights, while future integration with deep whole-genome sequencing and liquid biopsy approaches may further improve tumor origin identification and advance precision oncology.

Keywords: artificial intelligence, cancer of unknown primary, machine learning, next-generation sequencing, precision oncology

Presentation number: OP 02 Abstract number: 82-ISABS-2026

TRANSFORMING CANCER DIAGNOSIS: QUANTUM AI FOR GLOBAL LEUKEMIA SCREENING

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Acute lymphoblastic leukemia (ALL) is the most prevalent childhood malignancy, yet survival rates diverge sharply between high-income (>90%) and low-income settings (<50%), largely due to delayed diagnosis. Flow cytometry and cytogenetic analysis remain inaccessible across 70% of facilities in low - and middle-income countries. Classical AI approaches for automated blood smear analysis achieve limited accuracy (75-82%) with classical ML and require substantial computational resources for deep learning alternatives. We present QUANT-HemoDx, a hybrid quantum-classical framework integrating ResNet-18 convolutional feature extraction with an 8-qubit variational quantum circuit (VQC). Classical features (512-dimensional) are compressed and encoded into quantum states via parameterized RY rotations. Quantum superposition enables parallel morphological feature evaluation, entanglement captures higher-order cellular correlations, and interference reinforces diagnostically meaningful patterns. The model was trained on the C-NMC 2019 dataset (15,114 images, 118 patients) and independently validated at Ahsania Mission Cancer & General Hospital, Dhaka, Bangladesh. Hardware validation was performed on IBM's ibm_kyoto 127-qubit quantum processor. QUANT-HemoDx achieved 96.7% accuracy (ROC-AUC 0.991; 95% CI: 0.981-0.998), sensitivity 98.1%, and specificity 95.2% on the benchmark test set. Clinical validation yielded 94.2% accuracy on real quantum hardware. Compared to ResNet-50, the architecture requires 266× fewer parameters (96,000 vs. 25.6 million) and trains 1.68× faster, reducing per-patient diagnostic cost to $0.08. QUANT-HemoDx demonstrates that quantum AI architectures can achieve clinically competitive ALL detection with dramatically reduced computational overhead - a meaningful advantage for resource-limited screening contexts. Prospective multicenter validation is ongoing.

Keywords: quantum AI, leukemia detection, cancer prevention, early diagnosis, health equity

Presentation number: MSYIA 03 Abstract number: 27-ISABS-2026

FROM SPEED TO TRUST: THE GOVERNANCE OF ARTIFICIAL INTELLIGENCE-ASSISTED KNOWLEDGE PRODUCTION IN PRECISION MEDICINE

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Artificial Intelligence (AI) transforms knowledge-intensive work by generating outputs faster than they can be verified. The goal of this study was to examine the consequences of this asymmetry in science production, with a focus on its implications for precision medicine and biomedical research. In this paper, we analyze current evaluation systems reward speed instead of validation and deliver outputs at a pace that exceeds verification. Our analysis identifies structural deficiencies in institutions that may lead unverified AI-assisted claims to enter translational pipelines and clinical practice. In precision medicine, the risk of unverified AI claims increases, as AI-assisted research processes tend to produce statistically probable, consensus-aligned outputs that can suppress the biological heterogeneity precision medicine is designed to address. We propose a system that checks AI-assisted claims, which includes built-in staged review points and separation of teams that generate theories, and conduct empirical research, from those that evaluate their results. This challenge extends beyond technical issues. It requires organizational solutions and the formulation of an early model for structuring knowledge work in the AI era.

Keywords: AI governance, evaluation infrastructure, precision medicine, organizational design, knowledge production

THE ISABS FUTURE SCIENTIST AWARD PRESENTATIONS

Presentation number: FSA 01 Abstract number: 169-ISABS-2026

INVESTIGATION OF GENOTOXICITY AND CYTOTOXICITY OF UV NAIL LAMP RADIATION

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The aim of the research was to determine toxicity (genotoxicity and cytotoxicity) of radiation emitted by UV, LED, and combined UV/LED nail lamps using the Allium cepa test. The experiment investigated the effects of different radiation types and exposure durations on the growth and genetic material of onion root tips. Germinated seeds of Allium cepa L. were exposed to UV (36W), LED (5W), and UV/LED (54W) lamps for intervals of 2.5, 5, 7.5, and 10 minutes. Root length was measured to assess cytotoxicity, while the mitotic index, chromosomal aberrations, and micronuclei were analyzed via microscopy to evaluate genotoxic effect. The results showed that UV lamp radiation caused significant growth inhibition of the roots, particularly at the 10-minute exposure mark ($p=0.02$). Combined UV/LED radiation resulted in growth inhibition and morphological changes during longer exposure intervals, whereas LED radiation showed no significant impact on root length under the experimental conditions. While the mitotic index did not show statistically significant deviations, a significant increase in chromosomal aberrations and micronuclei was observed in the UV and UV/LED treatment groups. Specifically, UV radiation for 7.5 minutes induced aberrations in 70.84% of dividing cells ($p=0.00002$). These findings confirm the genotoxic potential of UV and UV/LED nail lamps. Given that an accompanying survey revealed that over 80% of users apply no skin protection during treatments, further education on the risks and preventive measures is essential.

Keywords: genotoxicity, cytotoxicity, UV radiation

Presentation number: FSA 02 Abstract number: 171-ISABS-2026

USING THE ALPHAGENOME AI MODEL TO INTERPRET A GENETIC VARIANT LINKED TO FAMILIAL HYPERCHOLESTEROLAEMIA

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To test whether the AlphaGenome AI model can quickly and accurately analyse a known harmful genetic variant in the LDLR gene – a gene linked to Familial Hypercholesterolaemia (FH), a hereditary condition causing dangerously high cholesterol – and to explore whether this kind of AI analysis could help doctors make faster diagnostic decisions. AlphaGenome (developed by Google DeepMind, published in Nature, 2026) was used through its Python programming interface in Google Colab. A 1-megabase stretch of DNA around the variant position was analysed across four biological readouts: RNA-seq, DNase-seq, CHIP-TF and splice site signals. A 'local effect score' was calculated to measure how strongly the variant disrupts each readout near its exact position. Statistical significance was tested using the Mann-Whitney U test. A gene ranking tool (score_variant) was also used to compare the LDLR gene against all 57 other genes in the region. All four readouts showed signs of harmful impact. The effect scores were: RNA-seq 7.8σ, DNase 72.1σ, CHIP-TF 51.5σ, and splice sites 477.6σ. The Mann-Whitney U test gave U= 0 and p ≈ 0, meaning the result is highly statistically significant. LDLR ranked 6th most affected out of 57 genes. AlphaGenome produced strong, consistent evidence that the variant rs121908027 in the LDLR gene is harmful – in under five minutes. Standard genetic testing for the same conclusion takes four to eight weeks. These results suggest that AI tools like AlphaGenome could help doctors reach faster diagnoses, as long as they are used responsibly and always reviewed by qualified medical professionals.

Keywords: familial hypercholesterolaemia, AlphaGenome, artificial intelligence, genetic variant interpretation

Presentation number: FSA 03 Abstract number: 170-ISABS-2026

APPLICATION OF ARTIFICIAL INTELLIGENCE IN THE DIAGNOSIS AND TREATMENT OF RARE GENETIC DISEASES

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To date, approximately 8,000 rare diseases have been identified. Although each affects a small number of individuals, collectively they represent a major global health challenge, impacting nearly 400 million people worldwide (about 5.7% of the population). Around 80% have a genetic origin, with symptoms often emerging in early childhood. Despite this, diagnosis often takes years or even decades, and about 95% of rare diseases remain without approved treatments. In this context, this study explores the potential of artificial intelligence as a transformative tool for improving the diagnosis and treatment of rare diseases. This study is based on a review of scientific literature, primarily utilizing the PubMed database along with additional relevant sources, focusing on genetics, bioinformatics, and the clinical application of artificial intelligence. A review of the literature indicates that artificial intelligence enables rapid and highly accurate analysis of complex genetic and clinical data, identifying pathogenic variants among hundreds of thousands of alterations generated through whole-genome sequencing. Machine learning algorithms can detect subtle patterns often missed by traditional methods. Advanced AI systems such as DeepRare demonstrate high diagnostic accuracy, frequently outperforming conventional approaches while significantly reducing time to diagnosis. By integrating genetic, clinical, and phenotypic data, artificial intelligence accelerates diagnosis and supports the development of targeted, personalized therapies. Overall, it is redefining rare disease care by enabling earlier detection, more precise treatment, and advancing precision medicine.

Keywords: artificial intelligence, machine learning, rare genetic diseases, next-generation sequencing

Presentation number: FSA 04 Abstract number: 168-ISABS-2026

EFFECT OF HYDROGEN PEROXIDE ON Na-K ATPASE ABUNDANCE IN NEURO2A CELLS

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The aim of this study is to investigate the effect of different concentrations of hydrogen peroxide on the abundance of sodium-potassium ATPase in a mouse neural cell line, Neuro2a, in order to further determine to what extent oxidative stress induces changes in gene expression in addition to its known effects on enzyme activity. Neuro2a cells were exposed to a range of H_2O_2 concentrations (10 nM to 10 mM) for 15 minutes to model both physiological and oxidative stress conditions. Following overnight incubation, cells were lysed and protein extracts were analysed using Western blotting. Na-K ATPase abundance was quantified and normalised to beta-actin. Data was analysed using one-way ANOVA to assess statistical significance between treatment groups. No statistically significant differences in Na-K ATPase abundance were observed across the tested H_2O_2 concentrations, as □□=0.82. High variability between replicates and a low coefficient of determination, □□2=0.0259, indicated the absence of a clear relationship between H_2O_2 concentration and protein abundance. No consistent trend was observed across treatment groups. These findings suggest that short-term oxidative stress does not significantly affect Na-K ATPase abundance in Neuro2a cells. The results support the idea that hydrogen peroxide primarily influences enzyme activity through rapid posttranslational mechanisms rather than inducing changes in gene expression under acute conditions. Further studies incorporating longer exposure times and complementary functional assays are required to determine whether sustained oxidative stress leads to transcriptional regulation of Na-K ATPase

Keyword: Neuro2a cells, Na-K ATPase, hydrogen peroxide effect, beta-actin

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ARTIFICIAL INTELLIGENCE IN PRECISION ONCOLOGY: ADVANCING CANCER DIAGNOSIS AND TREATMENT

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The aim of this work is to explore how artificial intelligence (AI) can transform precision oncology. It examines the biological complexity of cancer and compares traditional therapies and AI-driven tools in diagnosis, treatment selection, and patient outcomes. This study is based on a review of scientific literature, with the PubMed database used as the primary source. The selected literature includes studies focusing on artificial intelligence, cancer biology, precision medicine, and targeted therapy in oncology. Analysis shows that AI facilitates precision medicine by tailoring treatment to the specific genetic profile of a patient and the unique heterogeneity of their tumor. Furthermore, by utilizing AI-assisted predictions for immunotherapy and targeted therapies, clinicians can identify the most effective drugs for individual patients. These AI applications demonstrate significant potential in minimizing treatment side effects, detecting tumors at earlier stages, and ultimately improving patient survival rates. While cancer remains a complex disease requiring highly individualized approaches, AI significantly improves the precision of data analysis and clinical decision-making. The integration of AI into oncology promises a revolution in medicine through advancements such as AI-assisted drug discovery and real-time monitoring of tumor evolution. In the new era of precision medicine, artificial intelligence will not replace oncologists, but oncologists who use artificial intelligence will likely replace those who do not.

Keywords: artificial intelligence, precision oncology, cancer genomics, machine learning, personalized medicine

ABSTRACTS OF POSTER PRESENTATIONS

AI FOR CLINICAL DECISION SUPPORT

Presentation number: PP 01 Abstract number: 7-ISABS-2026

INTEGRATING 3D SIMULATION AND PSYCHOLOGICAL ASSESSMENT IN POST-BARIATRIC BREAST RESHAPING: A PROSPECTIVE CONTROLLED STUDY IN A PUBLIC HEALTH SETTING

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Post-bariatric women frequently develop severe breast deformities following massive weight loss, with significant physical discomfort and psychosocial distress. Optimizing preoperative counseling is crucial to align expectations and improve postoperative satisfaction. The aim of this prospective controlled study is to evaluate the clinical and psychological impact of integrating three-dimensional (3D) simulation into preoperative counseling for breast reshaping within a public healthcare system. One hundred consecutive post-bariatric patients undergoing mastopexy, reduction mammaplasty, or mastopexy with or without implants are enrolled at a university-affiliated tertiary referral center and randomized 4:1 to standard counseling or counseling enhanced with 3D simulation. Baseline evaluation includes comprehensive clinical assessment, anthropometric and surgical data collection, and validated psychological and body image questionnaires. Primary outcome is postoperative satisfaction at 6 weeks. Secondary outcomes include expectation–outcome alignment, decisional conflict reduction, changes in body image perception, rate of surgical plan modifications after counseling, treatment postponement or refusal, and perceived correspondence between simulated and surgical results. Preliminary findings indicate improved expectation realism, greater patient engagement, and reduced decisional uncertainty in the 3D simulation group, without increased operative time or complication rates. The integration of structured 3D simulation into preoperative counseling may represent a valuable tool to enhance psychological preparedness, shared decision-making, and early satisfaction in post-bariatric breast reshaping within a public health framework.

Keywords: breast, mastopexy, massive weight loss, counseling, prothesis

Presentation number: PP 02 Abstract number: 66-ISABS-2026

AI DECODING DCIS: IMAGING SIGNATURES ON CONTRAST-ENHANCED MAMMOGRAPHY

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The aim of this study was to assess the potential of contrast-enhanced mammography (CEM) combined with artificial intelligence (AI) for non-invasive characterization of tumor biology, with a particular focus on ductal carcinoma in situ (DCIS) as a distinct entity. In this retrospective single-center study, 654 women underwent breast imaging and histopathological evaluation, with 113 lesions analyzed, including 10 cases of DCIS (9%). Imaging features on CEM, AI-derived malignancy scores (iCAD ProFound AI®), and pathological characteristics were assessed. Compared with invasive cancers, DCIS lesions demonstrated significantly lower conspicuity ($p<0.001$), more frequent association with suspicious microcalcifications (86% vs 36–57%, $p=0.02$), and a higher prevalence of partial enhancement (57% vs 5%, $p=0.005$). Most DCIS lesions did not present as a distinct mass, corresponding to a non-mass imaging phenotype. No DCIS cases showed axillary lymph node involvement, in contrast to invasive cancers. AI-derived malignancy scores were lower in DCIS compared to invasive tumors, without statistical significance. These findings indicate that DCIS exhibits a specific imaging signature characterized by low conspicuity, limited enhancement, and strong association with microcalcifications. The integration of functional imaging and AI provides measurable signals that may reflect tumor behavior beyond conventional assessment. This approach supports imaging-based phenotyping and highlights the potential for non-invasive risk stratification and personalized management in early breast cancer.

Keywords: breast cancer, ductal carcinoma in situ, contrast-enhanced mammography, artificial intelligence, personalized medicine

Presentation number: PP 03 Abstract number: 64-ISABS-2026

FROM PIXELS TO PHENOTYPE: CAN AI-ENHANCED MAMMOGRAPHY DECODE BREAST CANCER BIOLOGY?

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The aim of this study was to evaluate whether tumor biology can be inferred non-invasively using contrast-enhanced mammography (CEM) and artificial intelligence (AI). This retrospective single-center study included 399 patients with abnormal screening findings (BI-RADS 0), with 113 analyzed lesions, including 76 malignant and 37 benign lesions. All lesions were assessed using CEM and AI-based malignancy scoring (iCAD ProFound AI®). Tumors were classified according to molecular subtypes and grouped as luminal versus HER2-positive and triple-negative. CEM demonstrated subtype-related imaging phenotypes, with luminal tumors more frequently appearing as irregular, spiculated masses with heterogeneous enhancement, while non-luminal tumors more often presented as round lesions with rim or homogeneous enhancement. AI analysis showed significantly higher malignancy scores in malignant compared to benign lesions (70.5% vs 38.0%, $p < 0.001$), with good diagnostic performance (AUC = 0.744, sensitivity 71%, specificity 70%). Although AI scores varied across molecular subtypes, these differences were not statistically significant. These findings suggest that while tumor biology is not fully captured by conventional morphology, the integration of functional imaging and AI moves imaging beyond detection toward biological characterization and supports the development of non-invasive biomarkers in precision breast cancer diagnostics.

Keywords: breast cancer, contrast-enhanced mammography, artificial intelligence, molecular subtypes, precision medicine

Presentation number: PP 04 Abstract number: 15-ISABS-2026

EGFR-AI: A MACHINE LEARNING MODEL FOR PREDICTING EARLY POSTOPERATIVE RENAL FUNCTION

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Acute kidney injury is a frequent and serious postoperative complication, making early identification of patients at risk essential for optimizing clinical management. This study aimed to develop a machine learning–based predictive model capable of estimating renal function in the early postoperative period using routinely collected perioperative data. A retrospective cohort of 200 surgical patients admitted to the intensive care unit of the University Hospital Centre Zagreb between January and July 2024 was analyzed. Preoperative and intraoperative variables were used to predict postoperative estimated glomerular filtration rate (eGFR), calculated using the CKD-EPI equation and categorized into three classes (G1, G2, and G3+). A two-layer machine learning architecture ("eGFR-AI") combining XGBoost classifiers and logistic regression was developed to predict postoperative eGFR categories. The final model achieved an accuracy of 0.75 and ROC-AUC of 0.92. Feature importance analysis identified chronic kidney disease, arterial hypertension, age, and sepsis or shock as key predictors of postoperative renal function. These results demonstrate that machine learning models based on routinely available clinical data may support early risk stratification and personalized postoperative management of surgical patients.

Keywords: postoperative kidney function, machine learning, artificial intelligence, glomerular filtration rate, personalized postoperative care

Presentation number: PP 05 Abstract number: 102-ISABS-2026

DIAGNOSTICS OF LUPUS ANTICOAGULANT IN COAGULATION LABORATORY

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Lupus anticoagulant is a heterogeneous group of antibodies that interfere with phospholipid-dependent coagulation pathways in vitro, resulting in prolongation of APTV, PV or dRVVT (dilute Russell vipervenom time). The purpose of testing lupus anticoagulant is in suspected primary or secondary antiphospholipid syndrome, together with aCL and anti-β2-GPI, in elderly patients with deep vein thrombosis or embolism and in case of repeated miscarriages and suspected thrombophilia. At the Clinical Hospital Center Zagreb, Laboratory of Hematology and Coagulation performs lupus anticoagulant test on a daily basis. Blood is collected in a coagulation tube with 3.2% sodium citrate anticoagulant. From the plasma sample first are performed screening tests: diluted APTV and dRVVT (diluted Russell Viper Venom Test, LA1) and if necessary, confirmatory tests: LA2 Confirmation Reagent and Lupus anticoagulant test (immuno test). For reliable interpretation of LA test results, samples should not be taken from patients under anticoagulant therapy. In 2025, 1000 samples were processed, of which 134 (13.4%) tested positive for lupus anticoagulant and 788 (78.8%) were negative. Part of the patients, 78 (7.8%) of them were on direct oral anticoagulant therapy and such samples are not processed in the confirmatory tests. Lupus anticoagulant is estimated to be present in 2 to 4% of the general population, but true prevalence is unclear. Among the different types of antiphospholipid antibodies, the lupus anticoagulant antibodies, characterized by their interference with clotting assays with low phospholipid content, show the strongest association with both thromboembolic and obstetric complications.

Keywords: lupus anticoagulant, coagulation, APTV, antiphospholipid, plasma

Presentation number: PP 06 Abstract number: 38-ISABS-2026

MULTI-BAND REHABILITATION OF TDCS AT BAIHUI (GV20) FOR ABNORMAL POST-STROKE NEURAL SYNCHRONIZATION: A PHASE-AMPLITUDE JOINT VALIDATION

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This study investigates the promoting effect of transcranial direct current stimulation (tDCS) at the Baihui acupoint (GV20) on the restoration of brain functional networks in stroke patients and its regulatory mechanism on abnormal synchronization. Through a randomized controlled trial, phase lag index (PLI) was employed to analyze multi-band brain networks on 14 subjects. Combined with weighted phase lag index (wPLI) and amplitude envelope correlation (AEC) methods, a dual-dimensional evaluation system for phase synchronization and amplitude coupling was constructed. The results showed that the experimental group exhibited a significant increase in whole-brain PLI values post-intervention ($p=0.016$), indicating enhanced global phase synchronization. wPLI analysis revealed a 12.6% increase in gamma-band synchronization, while theta-band synchronization was significantly suppressed by 36.9%, demonstrating its ability to correct abnormal low-frequency hypersynchronization post-stroke. AEC analysis further indicated that tDCS induced a 26.3% suppression of amplitude synchronization in the theta band, with varying degrees of reduction in the alpha and gamma bands. Graph theory analysis showed that the brain network tended toward decentralization, sparsity, and higher efficiency, supporting the transition from pathological coupling to functional integration in neural networks. The study confirms that tDCS at Baihui can effectively improve pathological brain network synchronization post-stroke by modulating cross- frequency neural oscillation patterns. The mechanism involves restoring phase coordination in impaired brain regions and bidirectionally regulating abnormal synchronization across different frequency bands.

Keywords: tDCS, Baihui (GV20), stroke rehabilitation, brain network synchronization, phase-amplitude coupling

Presentation number: PP 07 Abstract number: 21-ISABS-2026

SARCOPENIA AND MENOPAUSE

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The goal of this review was to synthesize current evidence on the impact that menopause has on sarcopenia and to explore artificial intelligence (AI) driven approaches for early detection alongside risk stratification. Sarcopenia, defined most commonly as age-related loss of muscle mass and function, is a major concern in menopause stemming from hormonal shifts – particularly oestrogen decline as well as changes in testosterone, IGF-1 with a rise in inflammatory markers. Menopause is clinically defined as a cessation of menses for over 12 months consecutively. Materials and methods used include an integrative analysis of observational studies, clinical trials along with meta-analyses on menopausal women – later combined with AI-based evaluations of clinical, functional and laboratory parameters from multiple datasets to identify patterns associated with muscle loss. Results consistently show that postmenopausal women exhibit reduced muscle mass, strength, and functional performance compared to premenopausal counterparts, whereas lifestyle factors such as physical activity, protein intake, and resistance training significantly mitigate risk. AI models, by integrating multidimensional data, demonstrate high accuracy in predicting sarcopenia thus allowing early identification of high-risk individuals. To conclude, menopause displays a critical window for sarcopenia onset, thereby by combining traditional clinical assessment tests with AI analytics, a newfound promising strategy offers for more personalized interventions. They must include changes in exercise, nutrition - with special emphasis on superior protein intake and hormonal considerations, as to preserve muscle function and improve long-term health outcomes.

Keywords: AI, menopause, proteins, sarcopenia, women

Presentation number: PP 08 Abstract number: 22-ISABS-2026

LIBIDO IN MENOPAUSE

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The goal of this summary is to identify influences and explore changes in libido during menopause. Menopausal transition can last a few months or years, typically starting in mid-40s/50s with an age average of 51,3 years. In every individual's life sexual activity plays an important role. Materials and methods used include artificial intelligence (AI) assessment in hormonal change, lifestyle and relationship dynamics then applied to detect patterns as well as predictive contributors towards sexual dysfunction. Women's sex lives can be subjected to numerous changes that arise due to hormonal transitions, psychosocial factors alongside cultural influences. Main symptoms that women report in menopause are vaginal dryness, dyspareunia, stunted libido and difficulties achieving an orgasm. Results show that approximately 62% of women report noticeable changes, whereas reduced oestrogen and testosterone values, stress, sleep depletion with relational issues prove to be strongly associated. AI modelling highlights that the combination of hormonal decline and psychosocial factors has the highest predictive value for decreased libido. Treatment options are standard and innovative. Standard option is a hormone replacement therapy in the form of oestrogen, progesterone and/or testosterone. Oestrogen is most often used as a low-dose topical therapy for vaginal dryness thereby reducing discomfort during intercourse, although systemic application also shows improvement. Innovative therapies may also include psychological treatment options since libido is not only a physiological problem, but has psychological, sociocultural and intrapersonal relationships influence as well. Female sexual dysfunction (FSD) requires a holistic approach. Ultimately, menopause-related reduction in libido is multifactorial and integrating AI- assisted assessments with individualized interventions addressing both biological and psychosocial contributors can support sexual wellbeing and improve quality of life.

Keywords: AI, FSD, hormone replacement therapy, libido, menopause

Presentation number: PP 09 Abstract number: 23-ISABS-2026

TESTOSTERONE'S ROLE IN WOMEN'S LIVES

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This study was made to review the role that testosterone has across women's lifespan as well as to explore the contribution of artificial intelligence (AI) in advancing research and clinical decision-making regarding female androgen health. A narrative review of latest scientific literature, randomized controlled trials, observational studies and international clinical guidelines was performed focusing on female androgen physiology, age-related hormonal changes and implications of testosterone deficiency. Additionally, the application of AI-driven analytic tools for large-scale biomedical data interpretation along knowledge synthesis was considered. Testosterone contributes towards sexual function, mood regulation, cognitive performance, maintenance of bone turnover and muscle mass. Its levels peak during early reproductive years and progressively decline with ageing, with a more pronounced decrease after menopause or bilateral oophorectomy. In polycystic ovary syndrome testosterone is greatly elevated thereby producing symptoms such as seborrhoea, alopecia, hirsutism, etc. On the other hand, reduced androgen levels are associated with decreased libido, fatigue, inferior wellbeing and changes in body composition. Among clinical indications, the strongest evidence for testosterone therapy is the treatment of hypoactive sexual desire disorder, quite often seen in (peri)menopausal women – randomized controlled trials demonstrated improvements in both libido and overall sexual satisfaction. AI-based approaches may support the identification of complex hormonal patterns, integration of multi-source clinical data and development of predictive models that could enable more personalized therapeutic strategies. Testosterone's role represents an important yet often underestimated factor in women's health hence why the integration of AI-assisted analytical frameworks may enrich understanding of female androgen physiology and facilitate advanced precision medicine approaches.

Keywords: AI, menopause, polycystic ovary syndrome, testosterone, women

Presentation number: PP 10 Abstract number: 84-ISABS-2026

FUNCTIONALLY DISTINCT MONOCYTE PHENOTYPES PROPAGATE PSORIATIC ARTHRITIS AND ANKYLOSING SPONDYLITIS

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Monocytes play a crucial role in the pathogenesis of spondyloarthropathies (SpA), such as ankylosing spondylitis (AS) and psoriatic arthritis (PsA), by differentiating into key inflammatory cells. Among these, monocyte-derived dendritic cells (moDCs) are of particular interest due to their close interaction with T- cells via the IL-12-23/IL-17 axis. This study aims to analyze the expression of DC markers on blood monocytes, evaluate changes upon their differentiation into DCs, and test their subsequent functionality. PBMCs were isolated from patients with active PsA, AS, and healthy controls. Using flow cytometry, we analyzed a panel of markers for monocyte lineage (CD14, CD16), DC differentiation (CD1a, CD1c, CD141, CD206, CD209), antigen presentation (CD40, MHCII), and costimulation (CD80, CD86). Sorted classical monocytes were cultured with DC-inducing factors, IL-17, or both; a control group underwent spontaneous differentiation. Phagocytic ability was tested via pHrodo assay, and antigen-presenting capacity was assessed in a moDC:T-cell co-culture by measuring cytokine production. Fresh PsA monocytes expressed higher levels of CD206, CD209 and CD1a. In vitro, spontaneously differentiated AS monocytes showed lower CD141 and higher CD40. AS-derived moDCs had heightened phagocytosis but produced less IL-12/23, resulting in lower T-cell IFN- and TNF-α production. Conversely, PsA-derived moDCs showed higher expression of costimulatory (CD86) and dendritic (CD1c) markers. Across all groups, IL-17 supplementation increased CD209 while decreasing CD141 and CD1a expression. Conclusion: Our findings reveal distinct pathological pathways in SpA. In psoriatic arthritis, monocytes and moDCs show a phenotype skewed towards heightened antigen presentation. In contrast, cells in ankylosing spondylitis display a profile characterized by enhanced phagocytosis but a reduced capacity to stimulate T-cell responses, suggesting different primary mechanisms of immune dysfunction.

Keywords: autoimmune diseases, peripheral blood monocytes, phenotype, phagocytosis, antigen presentation

Presentation number: PP 11 Abstract number: 17-ISABS-2026

WHEN A DESMOID IS NOT A DESMOID: NODULAR FASCIITIS REVEALED BY MOLECULAR DIAGNOSTICS

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Nodular fasciitis is a benign self-limited myofibroblastic/fibroblastic tumor of uncertain etiology. It is often misdiagnosed as a malignant tumor due to its rapid growth and increased mitotic activity. Aggressive fibromatosis/desmoid tumor is a rare fibroblastic disease belonging to low-grade malignant tumors since they show high potential for infiltrating surrounding tissues. Due to high recurrence rate, these tumors require strict follow-up. Given the histological overlap between both entities, molecular diagnostics play a crucial role in establishing the diagnosis and course of treatment. A 42-year-old patient presented with recently worsened long-standing limb pain and tingling. Upon admission, magnetic resonance imaging (MRI) of the cervical spine revealed an expansive lesion at the C1-C2 level. The patient underwent surgical treatment, after which she reported symptoms regressing. Initial histopathological and immunohistochemical testing corresponded to a primary mesenchymal neoplasm of the aggressive fibromatosis type. However, further molecular genetic analysis revealed a *MYHg::USP6* gene fusion, a characteristic molecular feature of nodular fasciitis that supports the distinction of this benign, self-limited lesion from other mesenchymal neoplasms, including aggressive fibromatosis. Accordingly, the biological nature of the tumor was reinterpreted as a more benign, self-limited proliferation that can morphologically mimic a desmoid-type tumor. This finding is clinically significant because it substantially changes the therapeutic approach, preventing unnecessary aggressive treatments. This case highlights the role of molecular diagnostics in refining and, in some cases, redefining the initial diagnosis. Identification of specific genetic alterations can influence clinical decision-making and guide appropriate therapeutic management therefore preventing unnecessary aggressive treatment while ensuring that patients receive appropriate and individualized care.

Keywords: desmoid tumors, molecular tumor profiling, precision oncology, next-generation sequencing, whole exome sequencing

Presentation number: PP 12 Abstract number: 59-ISABS-2026

AI-DRIVEN HRD DETECTION USING SHALLOW WGS: A COMPARATIVE STUDY FROM UNIVERSITY HOSPITAL OF SPLIT

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Homologous recombination deficiency (HRD) is a key biomarker for predicting response to PARP inhibitor therapy in precision oncology. While *BRCA1/2* mutations represent the canonical mechanism of HRD, many tumors exhibit HRD without detectable mutations. The goal of this report was to evaluate AI-driven detection of HRD using shallow whole -genome sequencing (sWGS) in a real -world clinical setting. Two tumor samples were analyzed using sWGS combined with targeted *BRCA1/2* panel sequencing. Genomic instability was quantified using large genomic alterations (LGA) and loss of parental copy (LPC), and an ensemble of 100 machine learning classifiers was applied to estimate genomic instability probability (GI), with HRD defined as GI > 0.5. Both samples were classified as HRD-positive. The *BRCA*-mutated sample showed GI 0.84, with dominant structural alterations (LGA 28.96, LPC 1) and a pathogenic *BRCA2* mutation. The *BRCA*-wild-type sample showed higher GI 0.99, characterized by extensive copy number loss (LGA 20.83, LPC 22) without detectable pathogenic variants. Model dispersion was lower in the *BRCA*-wild-type case, indicating higher prediction stability. These results demonstrate that HRD represents a genomic phenotype detectable independently of *BRCA* mutation status. AI-driven analysis of sWGS data enables robust identification of HRD across diverse molecular contexts and supports improved patient stratification for targeted therapies in routine clinical practice.

Keywords: HRD, shallow WGS, artificial intelligence, genomic instability, precision oncology

Presentation number: PP 13 Abstract number: 112-ISABS-2026

EFFECTS OF GLP-1 AND GLP-1/GIP AGONIST THERAPY ON IGG N-GLYCOSYLATION IN OVERWEIGHT INDIVIDUALS

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Protein glycosylation is a key post-translational modification that modulates protein function. Immunoglobulin G (IgG) N-glycans are established biomarkers, with specific patterns associated with immune function and inflammatory processes. These patterns can be integrated into GlycanAge, a composite metric of biological age reflecting immune system status and response to interventions. GLP-1 and GIP receptor agonists, including semaglutide (Wegovy) and tirzepatide (Mounjaro), are widely used for obesity treatment and have demonstrated systemic metabolic and anti-inflammatory effects. However, their impact on IgG glycosylation and GlycanAge remains insufficiently characterized. Twenty-four individuals with obesity undergoing GLP-1/GIP-based therapy were included. Plasma samples were collected at baseline, 3, and 6 months. IgG N-glycans were enzymatically released, labelled, and analysed using capillary gel electrophoresis with fluorescence detection (CGE-LIF). A total of 27 glycan peaks were quantified and used to derive glycan traits, including galactosylation, sialylation, and bisection. Biological age was calculated using the GlycanAge algorithm. Treatment was associated with a shift in IgG glycosylation towards a less inflammatory profile, with increased anti-inflammatory and reduced pro-inflammatory glycan features. This was accompanied by a decrease in GlycanAge, indicating improved biological age, viewed through the lens of the immune system. However, individual responses varied, with some participants showing minimal or adverse changes. GLP-1/GIP-based therapies are associated with favourable changes in IgG glycosylation and biological age, consistent with reduced chronic inflammation. IgG glycan profiling may serve as a sensitive biomarker for monitoring treatment effects, while highlighting interindividual variability.

Keywords: IgG glycosylation, glycanage, GLP-1 receptor agonists, obesity, biological age

Presentation number: PP 14 Abstract number: 94-ISABS-2026

ASSOCIATION OF IL-1β RS16944 POLYMORPHISM WITH COAGULATION PARAMETERS IN COVID-19 PATIENTS

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COVID-19 is frequently associated with coagulation abnormalities that contribute to disease progression and clinical outcomes. The aim of this study was to investigate the association of IL-1β polymorphism (rs16944) with coagulation parameters and clinical severity in COVID-19 patients. The study included 750 patients from the Bosnian population, stratified into three groups according to disease severity. Coagulation parameters, including prothrombin time (PT), activated partial thromboplastin time (aPTT), platelet count, international normalized ratio (INR), and D-dimer, were analyzed using standard IFCC protocols. Statistical analysis was performed using linear regression models to assess differences between genotypes, while logistic regression adjusted for age and sex was used to evaluate the association with disease severity. In patients with mild clinical presentation, significant differences were observed in coagulation parameters, with AA genotype carriers showing higher PT (p=0.010) and aPTT (p=0.030) values compared to GG and GA genotypes. In patients with moderate disease, significant differences were found in platelet count (p=0.046), with higher values observed in AA genotype carriers. No statistically significant associations were observed in patients with severe clinical presentation. Logistic regression analysis showed no significant association between rs16944 polymorphism and disease severity. These findings suggest that the IL-1β rs16944 polymorphism influences coagulation pathways, particularly in earlier stages of COVID-19, but is not an independent predictor of disease severity. The results highlight the importance of genetic variability in modulating host response and support further investigation of IL-1β as a potential biomarker in clinical risk assessment.

Keywords: COVID-19, disease severity, polymorphisms, coagulation, biomarkers

Presentation number: PP 15 Abstract number: 106-ISABS-2026

MACHINE LEARNING EXPLORATION OF MATERNAL CORTISOL VARIABILITY ACROSS ENVIRONMENTAL AND BIOLOGICAL DETERMINANTS IN PREGNANCY

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Cortisol, a primary biomarker of the hypothalamic–pituitary–adrenal axis, reflects maternal physiological adaptation to pregnancy-related stress. Increasing evidence suggests that cortisol regulation is influenced by complex interactions between environmental and biological factors, highlighting the relevance of integrative approaches. This study aimed to investigate differences in maternal cortisol levels according to geographic environment (island vs mainland), parity (primipara vs multipara), body mass index (BMI), fetal sex. A cross-sectional analysis on a sample of 337 pregnant women was therefore performed, applying standard statistical methods and exploratory machine learning models to evaluate group differences, potential nonlinear relationships and relative importance of predictors. Results indicated modest variability in cortisol levels across geographic and parity groups with BMI showing a weak association with cortisol variability. Machine learning models suggested that combinations of biological and environmental factors contributed more to cortisol variation than individual predictors; however, the overall explained variance remained low. These findings suggest that maternal cortisol regulation in the second trimester is shaped by multifactorial interactions, while the examined variables alone have limited predictive value. Machine learning approaches may provide a useful framework for exploring complex physiological patterns during pregnancy, although their contribution to individualized risk prediction in this context appears modest.

Keywords: cortisol, pregnancy, machine learning, integrative approach, multifactorial interactions

Presentation number: PP 16 Abstract number: 41-ISABS-2026

INTELLIGENCE-DRIVEN PRECISION SONOTHROMBOLYSIS: ADVANCED DEVICE DESIGN AND PREDICTIVE RBC REPAIR

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Thrombotic diseases are a leading cause of global mortality. Traditional interventions face limitations such as prolonged duration and vascular injury. This study advances precision medicine by developing an intelligent dual-frequency sonothrombolysis system and ultrasound-responsive nanocarriers, while addressing red blood cell (RBC) hemolysis through predictive modeling. Intelligent Interventional System: A dual-frequency ultrasound device was engineered for targeted thrombosis treatment. Utilizing a microfluidic thrombus chip and extracorporeal platforms, we performed data-driven optimization of parameters (frequency, drug concentration). Animal experiments validated its superior efficacy, providing a foundation for intelligence-powered clinical decision support in interventional therapy. Precision Drug Delivery: To enhance penetration, we developed ultrasound-responsive carriers featuring phase-change nanodroplet-coated microbubbles. Fluorescence imaging elucidated the cavitation-mediated thrombolytic mechanism. This approach aligns with intelligence-enhanced drug development, ensuring high-precision delivery and safety through real-time blood flow monitoring. Predictive RBC Repair Model: To mitigate hemolysis, we investigated RBC fatigue under cyclic stress. A microfluidic device revealed morphological transitions, leading to the establishment of an empirical fatigue model. Furthermore, a regenerative strategy using glucose/oxygen-carrying nanodroplets was proposed. By simulating cytoskeletal extension, a critical threshold model for RBC repair was established, offering a theoretical framework for protecting blood cells during therapy. This research integrates intelligent hardware with predictive biological modeling, providing a comprehensive solution for precision sonothrombolysis. It offers a robust technical foundation for the design of future intelligence-optimized medical devices and regenerative therapies.

Keywords: intelligence-driven precision sonothrombolysis, advanced interventional device, precision drug delivery, predictive model of RBCs, microfluidics

Presentation number: PP 17 Abstract number: 5-ISABS-2026

AI-POWERED DETECTION OF FOOD CONTAMINANTS: A PREVENTIVE APPROACH WITHIN THE PRECISION MEDICINE FRAMEWORK – HEAVY METAL DETECTION IN FISH AND MARINE ORGANISMS

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Food contamination with heavy metals represents a significant challenge for food safety and poses serious risks to human health, particularly through the consumption of fish and marine organisms. Traditional analytical methods for heavy metal detection are often time-consuming, costly, and require extensive sample preparation, highlighting the need for faster and more efficient screening approaches. In this context, artificial intelligence (AI) has emerged as a promising tool for improving contaminant detection and supporting preventive food safety strategies.This review evaluates current AI-based approaches for the detection of heavy metal contamination in fish and marine organisms, with a focus on their application in food safety monitoring. Various machine learning and deep learning models, combined with non-destructive analytical techniques such as hyperspectral imaging, have been applied to classify contaminated and non-contaminated samples of marine organisms and to predict heavy metal concentrations based on different types of input data. The reviewed studies demonstrate that AI-driven methods achieve high accuracy, sensitivity, and rapid data processing, often outperforming conventional analytical and statistical approaches. Deep learning models have proven to be excellent for rapid, non-destructive detection, while machine learning algorithms have demonstrated reliable performance even with limited datasets. Overall, AI-powered detection systems represent an effective complementary tool for enhancing heavy metal detection and monitoring in food production systems. Although further efforts are required for standardization and the collection of reference data, AI-based detection systems have strong potential to contribute to early detection and prevention of exposure, thereby enhancing food safety and supporting both public and individual human health within a preventive and precision medicine framework.

Keywords: artificial intelligence, heavy metals, food contamination, food safety, precision medicine

Presentation number: PP 18 Abstract number: 6-ISABS-2026

AI-BASED PREDICTIVE TOXICOLOGY FOR FOOD ADDITIVES: TOWARDS PRECISION RISK ASSESSMENT IN NUTRITION AND HEALTH- APPLICATION TO ARTIFICIAL SWEETENERS

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Artificial food additives, particularly sweeteners, are widely used in the food industry, and their safety assessment represents an important challenge in food toxicology. Traditional toxicological evaluation is time-consuming and resource-intensive, highlighting the need for alternative approaches that enable earlier and more efficient risk assessment. In this context, artificial intelligence (AI) has emerged as a powerful tool in predictive toxicology.This review explores the application of AI-based predictive toxicology approaches for food additive safety assessment, with a particular focus on artificial sweeteners, including aspartame, sorbitol and xylitol as a case example. Different computational strategies, such as quantitative structure–activity relationship (QSAR) models and machine learning algorithms, have been applied to predict toxicological endpoints based on molecular descriptors and available experimental data. The reviewed studies indicate that AI-driven models can achieve high predictive accuracy for several toxicity endpoints, including genotoxic and other biologically relevant effects, supporting their potential use in early hazard identification and prioritization of substances for further testing. Evidence from the literature further suggests that AI-supported analyses are effective in screening artificial sweeteners and identifying potential risks under different exposure scenarios, while also highlighting limitations related to data availability, model interpretability, standardization, and the need for experimental validation. Overall, AI-based predictive toxicology represents a promising complementary approach in food safety assessment, contributing to preventive risk management and supporting the broader goals of precision and preventive medicine.

Keywords: artificial intelligence, predictive toxicology, food additives, artificial sweeteners, food safety

Presentation number: PP 19 Abstract number: 52-ISABS-2026

EXPLAINABLE AI IN PRECISION MEDICINE FOR DIAGNOSING AND TREATING RARE DISEASES

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GenAI (Generative AI) shows great promise in precision medicine, but hallucination and high computational demands remain a problem. We present a novel Neuro-Symbolic AI approach, which utilizes GenAI, Knowledge Graphs (KG), and GraphRAG (Graph Retrieval-Augmented Generation) to address these challenges. We used PubMed, FAERS (FDA Adverse Event Reporting System), UMLS (Unified Medical Language System), NORD (National Organization for Rare Disorders), NCIt (National Cancer Institute Thesaurus), and GO (Gene Ontology) as our knowledge sources. We used GenAI to mine PubMed for 18 entity types and 24 relationships. We mined FAERS for Drug-Drug interactions and Disease-Drug contraindications. We constructed a knowledge graph (KG) with all these entities and relationships and added NCIt and GO into it. We embedded the NORD and UMLS knowledge source as GraphRAG. Natural language user input is passed through named entity recognition (NER) and named entity normalization (NEN) to give us UMLS concept IDs (CUIs). These CUIs are used to fetch the immediate neighbours in the biomedical KG and supplementary knowledge from RAG. The results are passed through fine-tuned SLM GenAI for human understandable language. The integration of Knowledge Graphs and GraphRAG enables AI systems to become explainable. The hallucinations of AI have reduced substantially, reasoning is explainable, accuracy is improved and the system can run on desktop. We do not have sufficient data to quantify these results yet. Neuro-Symbolic AI with integrated GraphRAG offers explainability and reduced hallucination with higher accuracy, critical features that are required for medical applications of AI.

Keywords: neuro-symbolic AI, GraphRAG, GenAI, explainability

Presentation number: PP 20 Abstract number: 75-ISABS-2026

NOCTURNAL HYPOCALCEMIC SEIZURES AS THE FIRST MANIFESTATION OF PSEUDOHYPOPARATHYROIDISM TYPE IA IN A YOUNG CHILD

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Pseudohypoparathyroidism type Ia (PHP-Ia) results from maternal inactivating *GNAS* mutations on chromosome 20q13.3 that impair the stimulatory G protein (Gsα) signaling. Tissue-specific paternal imprinting leads to markedly reduced Gsα expression in renal proximal tubules, thyroid and pituitary, resulting in PTH resistance with hypocalcemia and hyperphosphatemia. Patients also exhibit Albright hereditary osteodystrophy (AHO) features including brachydactyly, round face, short stature and ectopic ossifications. We report a case of hypocalcemic seizures in a pediatric patient diagnosed with PHP-Ia, emphasizing diagnostic challenges of epileptic events in children. A 5-year-old boy presented with nocturnal tonic-clonic seizures preceded by hand/foot/face spasms over 3 weeks. History revealed infantile diarrhea, speech delay and previous subcutaneous osteoma removal. The patient presented with macrocephaly (+2.85Zcore), stocky habitus (height +1.20Zscore, weight +1.75Zscore), round face, ataxia and subcutaneous osteoma. Laboratory findings included hypocalcemia (Ca^{2+} 0.56 mmol/L, Ca 1.33 mmol/L), elevated PTH (60.89 pmol/L), hyperphosphatemia (3.16 mmol/L) and low vitamin D levels. Brain CT revealed symmetric basal ganglia calcifications. The diagnosis of PHP-Ia was established based on clinical and laboratory criteria. Treatment with calcitriol and calcium carbonate normalized calcium and improved neurological findings (spasms and ataxia). With its evolving phenotype, the PHP-Ia diagnosis relies on a combination of clinical and biochemical findings. While molecular testing is definitive, the complex *GNAS* locus inheritance requires a multi-modal approach including DNA sequencing, methylation studies and copy number variant analysis. Hypocalcemic seizures, as the initial manifestation of PHP-Ia, necessitates differentiation from other causes of epilepsy. Therefore, low calcium should prompt immediate PTH screening for timely detection of this rare but intricate disease.

Keywords: albright hereditary osteodystrophy, basal ganglia calcification, hypocalcemia, pseudohypoparathyroidism, seizures

Presentation number: PP 21 Abstract number: 39-ISABS-2026

A FLEXIBLE CONFORMAL NEAR-FIELD RESONANT SENSING INTERFACE FOR CONTINUOUS MONITORING OF PERIPHERAL ARTERIAL HEMODYNAMICS

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The early detection and proactive management of cardiovascular diseases depend critically on the long-term, continuous monitoring of peripheral arterial hemodynamics. However, conventional non-invasive modalities, such as photoplethysmography (PPG), are fundamentally limited by shallow optical penetration, which constrains their ability to reliably capture transient hemodynamic signatures in deep arteries. To address this challenge, we present a near-field resonant sensing system featuring a flexible, conformal interface for continuous arterial monitoring. The platform integrates a planar spiral single-coil sensor fabricated using flexible printed circuit (FPC) technology with a continuous-tracking readout architecture, enabling stable conformal coupling to the radial artery region and continuous acquisition of resonance-frequency shifts. Comparative experiments with a conventional rigid FR-4 interface demonstrated that the flexible FPC interface substantially improved coupling stability, reduced baseline drift induced by motion and deformation, and enhanced the beat-to-beat repeatability of cardiac-cycle waveforms. The resonance-derived signals showed strong agreement with synchronized PPG reference signals (Pearson's $r = 0.909$, $P < 0.001$) while preserving subtle morphological features, including the dicrotic notch, that are essential for hemodynamic characterization and AI-based cardiovascular assessment. These findings identify conformal mechanical coupling as a key determinant of signal fidelity and robustness in near-field physiological sensing. Collectively, this work establishes a flexible physical front end for the continuous, non-invasive monitoring of arterial hemodynamics and provides a high-quality data foundation for AI-enabled cardiovascular screening and personalized intervention in precision medicine.

Keywords: cardiovascular diseases, arterial hemodynamics, near-field resonant sensing, continuous monitoring, flexible interface

Presentation number: PP 22 Abstract number: 2-ISABS-2026

ARTIFICIAL INTELLIGENCE–BASED FUNCTIONAL CLASSIFICATION OF PARA-SWIMMERS WITH ACHONDROPLASIA: A BIOMECHANICAL APPROACH FROM EULERIA LAB

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Artificial Intelligence–Based Functional Classification of Para-Swimmers with Achondroplasia: A Biomechanical Approach from EULERIA LAB. The goal of this study was to develop and validate an artificial intelligence–based model for objective functional classification of para-swimmers with achondroplasia using biomechanical and anthropometric parameters. A cohort of competitive para- swimmers with achondroplasia underwent three-dimensional kinematic analysis of stroke cycles, assessment of joint range of motion, limb proportions, and propulsion-related variables, combined with wearable sensor and video-based data acquisition in controlled swimming trials. Machine learning algorithms were trained to identify movement patterns and functional limitations relevant to swimming performance and to predict optimal sport class allocation. Model performance was evaluated against expert-based classification outcomes and competition results. The results demonstrated that the AI model could accurately discriminate functional profiles and showed high agreement with current classification decisions, while additionally revealing subtle biomechanical determinants of propulsion efficiency, stroke symmetry, and start and turn performance specific to the morphological characteristics of achondroplasia. Feature importance analysis indicated that upper-limb lever arm length, shoulder range of motion, and trunk stability were key predictors of functional capacity in water. In conclusion, the integration of artificial intelligence and biomechanical analysis provides an objective, reproducible, and transparent framework for functional classification of para-swimmers with achondroplasia, with potential to reduce subjectivity, improve fairness in competition, and support evidence-based refinement of classification systems, as well as individualized training and rehabilitation strategies developed within the EULERIA LAB.

Keywords: artificial intelligence, para-swimming, achondroplasia, functional classification, biomechanics

Presentation number: PP 23 Abstract number: 43-ISABS-2026

FROM PLANNING TO ROUTINE PRACTICE: DIGITAL TRANSFORMATION OF A PATHOLOGY DEPARTMENT AT UNIVERSITY HOSPITAL OF SPLIT

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The digitalization of pathology departments represents a major step toward improving diagnostic efficiency, data accessibility, and the integration of advanced technologies in routine clinical practice. This study presents the implementation of a fully digital workflow at the Clinical Department of Pathology, University Hospital of Split. The digitalization process began in December 2023, following a structured planning phase that included defining the implementation timeline and technical requirements, and was completed in May 2025, after which the system has been continuously used in routine practice. The goal was to optimize workflow, reduce turnaround time (TAT), enable remote consultations, and establish a foundation for the application of AI tools. Materials and methods included the introduction of whole slide imaging (WSI), integration with PACS and LIS systems, and the reorganization of laboratory workflow starting from grossing and slide preparation, followed by prescan, selection of areas of interest, and scan quality control. This approach resulted in improved quality of hematoxylin and eosin (H&E) slides, which directly enhanced the quality of scanned WSI and contributed to overall laboratory quality improvement. The implementation required close collaboration between pathologists, the hospital IT department, the PACS provider, and scanner vendors, along with continuous staff training. Results demonstrated a significant improvement in workflow performance and TAT. The implementation also enabled improved collaboration, remote diagnostics, and increased satisfaction among staff. In conclusion, the digitalization of a pathology department is a complex, multidisciplinary process that extends beyond the mere acquisition of scanning equipment. It requires thorough assessment of technical infrastructure, coordinated involvement of multiple stakeholders, and continuous education to ensure successful implementation and long-term sustainability.

Keywords: digital pathology, whole slide imaging (WSI), workflow optimization, telepathology, artificial intelligence

AI-POWERED MULTI-OMICS INTEGRATION FOR PRECISION MEDICINE

Presentation number: PP24 Abstract number: 49-ISABS-2026

TOWARDS CLINICAL INTEGRATION OF CARDIOVASCULAR PHARMACOGENOMICS: THE CARDIOPHARMAGENET APPROACH

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Cardiovascular diseases remain the leading cause of mortality worldwide, while the integration of pharmacogenomics (PGx) into clinical practice is still fragmented and slow across Europe due to the lack of harmonized guidelines, limited clinical implementation, and inconsistent policy frameworks. Furthermore, the absence of centralized data infrastructures and insufficient use of advanced digital tools, including AI, further hinder the translation of PGx research into routine care. COST Action CA24165 Network for Cardiovascular Pharmacogenomics and Precision Medicine (CardioPharmaGENET) addresses these challenges by establishing a multidisciplinary, pan-European network aimed at harmonizing the generation, analysis, storage, and clinical use of cardiovascular PGx data, while creating a centralized knowledge hub and delivering evidence-based recommendations for policymakers and clinicians. The main objective of the Action is to enable the equitable and consistent implementation of PGx in cardiovascular medicine through coordinated research, capacity building, and stakeholder engagement. The work is structured into five dedicated Working Groups: WG1 Current landscape and status of PGx in cardiovascular medicine; WG2 Pharmacogenetic guideline development and optimization; WG3 AI/ML use in PGx of cardiovascular medicine; WG4 Policy regulation of PGx guidelines and impact analysis across Europe; and WG5 Communication, dissemination and exploitation. By integrating expertise from diverse disciplines and fostering collaboration across countries and sectors, the Action aims to bridge the gap between research and clinical practice, ultimately improving patient outcomes and reducing healthcare inequalities. Within only five months of implementation, CardioPharmaGENET has already engaged over 230 participants from 38 countries, demonstrating strong community uptake and positioning the network as a key driver of future advancements in personalized CVD medicine in Europe.

Keywords: pharmacogenomics, personalized medicine, cardiovascular medicine, artificial intelligence, COST Action

Presentation number: PP25 Abstract number: 28-ISABS-2026

IDENTIFICATION AND CHARACTERIZATION OF RARE DE NOVO CODING MUTATIONS IN CONGENITAL ANOMALIES OF THE KIDNEY AND URINARY TRACT (CAKUT) USING WHOLE EXOME SEQUENCING

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Congenital anomalies of the kidney and urinary tract (CAKUT) represent a group of developmental disorders and are one of the leading causes of chronic kidney disease in children. The aim of this study was to identify rare de novo coding mutations associated with CAKUT and highlight their importance in understanding disease mechanisms and improving genetic diagnostics. Samples are obtained from human peripheral blood, DNA is extracted, and Whole Exome Sequencing (WES) is performed to generate genetic data. The study focused on 13 affected probands, comparing their genotypes with those of their unaffected family members to identify de novo mutations. Variants that were common, non-coding, or clinically benign/uncertain were excluded to focus on rare, potentially pathogenic changes. Filtered de novo mutations were then analyzed by mutation type to characterize the genetic alterations in the cohort. Mutation counts per gene and proband were visualized using a CDS-length–normalized heatmap to allow comparison across genes of different sizes. In the cohort of 13 probands, missense mutations were the most frequent type, followed by other functional variants such as frameshift and splice-site mutations. SNVs were significantly more common than indels, and base substitution analysis showed a predominance of specific transitions (e.g., C>T) across the cohort. Using a CDS-length–normalized heatmap, the distribution of mutations in the top 20 genes was visualized for each proband, highlighting which genes were proportionally most affected by de novo mutations. The identified candidate genes represent a valuable resource for future functional studies and may contribute to improved diagnosis and personalized clinical management.

Keywords: CAKUT, whole exome sequencing, de novo variants, SNV, genetic analysis

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FROM BIOMARKER DISCOVERY TO REGENERATIVE THERAPY: THE DUAL POTENTIAL OF MENSTRUAL BLOOD-DERIVED MSCS AND EXTRACELLULAR VESICLES

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The identification of novel mesenchymal stromal cell (MSC) sources for biomarker discovery and regenerative application represents a key priority in translational medicine. Menstrual blood is a unique, non-invasive, and ethically acceptable source with utility in both diagnostics and regenerative medicine. Menstrual blood-derived mesenchymal stromal cells (MenSCs) and their extracellular vesicles (EVs) have emerged as promising tools in reproductive medicine, where both applications are urgently needed. Proteomic profiling of menstrual blood serum EVs reveals distinct molecular signatures in unexplained infertility compared to fertile controls, capturing alterations in cell adhesion, immune response, apoptosis, fibrosis, metabolism, and oxidative stress. These profiles enable patient stratification into endotypes, supporting personalised diagnostic strategies. Transcriptomic analysis further confirmed changes in unexplained infertility. From a regenerative perspective, MenSCs exhibit high proliferative capacity, multipotency, and an immunomodulatory secretome. MenSC-derived EVs might hold potential to restore endometrial receptivity and uterine microenvironments central to implantation failure – a key unmet need in infertility management. Beyond reproductive medicine, MenSC paracrine factors may stimulate regenerative responses in different tissues. We demonstrated that MenSC-EVs enhance extracellular matrix production, chondrogenic differentiation, and protect cartilage from degradation, extending their therapeutic reach to musculoskeletal disorders. The translational potential of MenSC-derived products as accessible biomarker sources and cell-free therapeutics positions menstrual blood as a compelling precision medicine resource for reproductive and musculoskeletal conditions alike. Funded by HORIZON-WIDERA-2021-ACCESS-03-01 program 382 project No. 101079489-TWINFLAG and EP Permed call 2024, project PERFERT 2025-2028

Keywords: menstrual blood, extracellular vesicles, proteomic profiling, unexplained infertility, cartilage regeneration

Presentation number: PP27 Abstract number: 109-ISABS-2026

A MACHINE-LEARNING-GUIDED LIPIDOMICS PIPELINE FOR IDENTIFYING CANDIDATE LIPID METABOLIC CHANGES ASSOCIATED WITH ACQUIRED VEMURAFENIB RESISTANCE IN BRAFV600E-MUTANT COLORECTAL CANCER

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BRAFV600E-mutant metastatic colorectal cancer is a clinically aggressive subgroup with limited benefit from vemurafenib monotherapy because of rapid development of resistance. The aim of this study was to identify lipidomic features linked with acquired resistance to BRAF inhibition and to build an analysis pipeline for selecting candidate lipid metabolic pathways for further studies. Vemurafenib-resistant RKO BRAFV600E colon cancer cells were generated by gradual exposure of parental cells to increasing drug concentrations over 6 months until stable resistance to 11.52 μM was achieved. Resistance was confirmed by MTT assay and morphological evaluation. Lipids were extracted using the Matyash protocol and analyzed by UHPLC-QTOF-MS. Untargeted lipidomics data from parental and resistant cells, under treated and control conditions, were processed and explored using L1/L2 logistic regression and linear SVM models. Internal cross-validation showed good separation between parental and resistant cells and treatment subgroups, indicating group-related biological differences in the lipidomic data. HexCer 34:0;2O, LPI 20:4 and FA 18:1 changed with vemurafenib treatment in both parental and resistant cells, indicating a shared response to drug exposure. In contrast, many PC, PI and LPC species showed different treatment- related changes between parental and resistant cells, suggesting a possible link to acquired resistance. To extend these findings, BioPAN analysis was performed on the same experimental contrasts. Combining BioPAN outputs with lipids identified by our ML analysis highlighted recurrent changes in membrane lipid remodeling and lipid storage pathways, with candidate genes including PLA2, LPCAT, MBOAT7, DGAT2, PEMT and CHPT1. These findings do not identify definitive targets, but they provide a practical framework for generating hypotheses and prioritizing lipid-related pathways for follow-up validation in BRAFV600E-mutant colorectal cancer.

Keywords: BRAFV600E, colorectal cancer, lipidomics, vemurafenib resistance, machine learning

Presentation number: PP28 Abstract number: 80-ISABS-2026

FC-RECEPTOR AFFINITY CHROMATOGRAPHY: A TOOL FOR IVIG THERAPEUTICS PROFILING

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Intravenous immunoglobulin (IVIg) therapeutics are complex biological mixtures of immunoglobulin G (IgG) requiring robust analytical methods to resolve their functional and structural heterogeneity. First established for immunodeficiencies, IVIg concentrates are widely used for immunomodulation in diverse immune-mediated diseases. While IVIg actions are multifactorial, key therapeutic pathways rely on direct, structure-sensitive binding events between IgG and Fc receptors. In this study, we evaluated the chromatographic performance of receptor affinity columns utilizing FcγRIIIa and FcRn ligands to profile two IVIg products. Using pH-gradient elution on analytical nonporous resin (NPR) columns, we demonstrated that two receptors display distinct separation selectivities toward specific IgG Fc-glycoforms. Retention of the IgG glycoforms on the FcγRIIIa column was consistent with sensitivity to steric constraints imposed by both core fucosylation and galactosylation. Conversely, the FcRn column displayed a separation mechanism likely driven by charge heterogeneity. Despite the distal location of the glycosylation site relative to the binding interface with the IgG, the pH-gradient elution resolved sialylated subpopulations based on pI-mediated retention shifts. Both receptor columns yielded highly reproducible, product-specific fingerprints that enabled differentiation of the studied commercial preparations. Distinct fingerprints validate receptor affinity chromatography as a versatile tool for IVIg quality control. Furthermore, isolating distinct high-affinity subpopulations offers a preparative strategy for manufacturing next-generation IVIg therapeutics with tailored profiles. Specifically, FcγRIIIa-based enrichment could yield formulations with enhanced effector functions, such as increased antibody-dependent cellular cytotoxicity (ADCC), while FcRn-mediated fractionation could produce variants with a prolonged IgG half-life to extend biotherapeutic activity.

Keywords: affinity chromatography, immunoglobulin G, N-glycosylation, biotherapeutics, quality control

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AI-DRIVEN ECHOCARDIOGRAPHIC ASSESSMENT OF INTRACARDIAC MASSES

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Accurate detection and characterization of intracardiac masses remain a persistent challenge in cardiovascular imaging, primarily due to overlapping morphological and functional features among neoplastic and non-neoplastic entities. Recent advances in artificial intelligence (AI) are redefining the echocardiographic assessment of cardiac masses, with increasing impact on diagnostic accuracy and clinical decision making. Contemporary machine learning (ML), deep learning (DL), and multimodal AI approaches demonstrate that AI driven models, particularly convolutional neural networks and radiomics based techniques, can achieve high performance in detection, segmentation, and classification tasks. These methods also enable automated image acquisition and more standardized interpretation, reducing operator dependency. Furthermore, integration of echocardiographic data with complementary imaging modalities and clinical parameters enhances diagnostic precision and supports a more comprehensive evaluation framework. Despite these advances, several barriers continue to limit broader clinical adoption, including small and imbalanced datasets, heterogeneity of imaging data, limited external validation, and challenges related to model interpretability and generalizability. Taken together, current evidence positions AI assisted echocardiography as a key driver toward more objective, reproducible, and efficient cardiovascular diagnostics. Future progress will depend on large scale multicenter validation, development of explainable models, and seamless integration into routine clinical workflows.

Keywords: echocardiography, artificial intelligence, cardiac tumors, deep learning, machine learning

Presentation number: PP30 Abstract number: 90-ISABS-2026

LONG-TERM STABILITY ASSESSMENT OF IGG N-GLYCANS IN LYOPHILIZED PLASMA FOR GLYCOMICS

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High-throughput glycomics relies on the availability of stable and reproducible samples for use as internal standards and in large-scale clinical cohorts. While previous literature has established the longitudinal stability of IgG N-glycans in regular frozen plasma, lyophilization offers superior advantages for global shipping and long-term storage logistics. This study evaluated a new lyophilization protocol to verify the structural integrity of the plasma glycome post-reconstitution. Human plasma samples were lyophilized and stored at room temperature (RT), +4˚C, and -20˚C, with stability assessed at baseline, 1 week, 1 month, 3 months, and 6 months. Analytical characterization was performed using Capillary Gel Electrophoresis with Laser-Induced Fluorescence (CGE-LIF). Our results demonstrate that IgG N-glycan profiles in lyophilized plasma remain stable for at least six months, with glycan concentrations and profiles matching those of non-lyophilized controls. While stability was maintained across all conditions, storage at -20˚C provided the most robust results, yielding the highest peak intensities and superior analytical consistency. Conversely, samples stored at room temperature exhibited the highest Coefficients of Variation (CVs). These findings confirm that lyophilization is a highly effective preservation method that maintains the biochemical integrity of IgG N-glycans. Beyond its utility as a stable internal standard, this approach facilitates the simplified distribution of samples for multi-center studies, ensuring high-quality glycomic data while reducing the dependency on continuous cold-chain logistics.

Keywords: glycosylation, electrophoresis, plasma, lyophilization, immunoglobulin

Presentation number: PP31 Abstract number: 55-ISABS-2026

PHYSIOXIA-DRIVEN FUNCTIONAL AND SECRETOME PROFILING OF MENSTRUAL BLOOD-DERIVED STROMAL CELLS FOR PRECISION BIOMARKER DISCOVERY IN REPRODUCTIVE BIOCOMPATIBILITY AND INFERTILITY

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Infertility affects ~17.5% of couples worldwide, with up to 30% of cases remaining unexplained (uI), highlighting the need for precision medicine approaches and novel biomarker discovery. Human menstrual blood-derived mesenchymal stromal cells (MenSCs) provide a non-invasive model to study uterine biology, implantation, and partner-dependent reproductive compatibility. However, standard in vitro culture conditions fail to replicate the physiologically low oxygen environment of the endometrium. This study evaluates the effect of physioxia (5% O_2) versus normoxia (21% O_2) on MenSC functional, metabolic, and secretory profiles, and explores their relevance for identifying biomarkers linked to infertility and reproductive biocompatibility. MenSCs were assessed for proliferation (spectrophotometry), migration (holomonitor), immunophenotype and intracellular calcium (flow cytometry), metabolic activity (metabolism analyser), and secretome composition (multiplex assay). In parallel, extracellular vesicles (EVs) derived from menstrual blood, MenSCs, and semen are being investigated to characterize partner-specific molecular interactions (flow cytometry). Physioxia significantly enhanced MenSC proliferation, glycolytic capacity, and intracellular calcium signaling, while modulating surface marker expression (↓CD73; ↑CD105, CD90, CD13). Secretome analysis revealed increased production of growth factors and chemotactic mediators alongside reduced pro-inflammatory cytokines, indicating a microenvironment supportive of implantation. Ongoing multi-omic profiling (transcriptomics, proteomics) of EV cargo and MenSC–semen EV interactions aims to identify coordinated molecular signatures associated with altered reproductive compatibility. These findings support physioxia as a critical parameter for physiologically relevant in vitro modeling and highlight MenSC-derived systems as a platform for discovering couple-specific biomarkers.

Keywords: precision medicine, menstrual blood stromal cells, extracellular vesicles, unexplained infertility, multi-omics

Presentation number: PP32 Abstract number: 69-ISABS-2026

HEREDITARY TRANSTHYRETIN CARDIAC AMYLOIDOSIS IN A 50-YEAR-OLD FORMER PROFESSIONAL ATHLETE: A CASE REPORT

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Transthyretin amyloidosis is a progressive systemic disorder associated with noteworthy mortality and morbidity, commonly in older adults. TTR is a protein synthesized in the liver that functions in the transport of thyroxine and retinol, which consists of four monomers that assemble into a tetramer. It is characterized by structural and functional abnormalities caused by extracellular deposition of misfolded transthyretin protein as amyloid fibrils in multiple organs, with clinical manifestations typically dominated by cardiomyopathy and/or polyneuropathy. We present to you the case of a 50-year-old former professional athlete who was evaluated in the cardiology department with a family history of sudden cardiac death and amyloidosis. The patient complained of activity intolerance, irritable bowel syndrome, and symptoms of peripheral neuropathy. ECG findings included biatrial enlargement, increased myocardial wall thickness, and reduced longitudinal strain from base apex. Particularly, the apical strain was presented with a „cherry on top" appearance, specific for cardiac amyloidosis. Serum protein electrophoresis showed no abnormalities, excluding the AL amyloidosis. Bone scintigraphy showed reduced bone uptake and increased cardiac uptake, consistent with ATTR amyloidosis. The diagnosis of hereditary ATTR amyloidosis was confirmed through genetic testing, which has also provided prognostic and therapeutic guidance. Following heart failure therapy showed modest symptomatic improvement and activity resumption. At one-year follow-up, disease progression and heart failure exacerbation led to the addition of tafamidis to the therapeutic regimen. Despite its rarity, cardiac amyloidosis should be considered in the differential diagnosis of a patient with heart failure, particularly when accompanied by a positive family history and systemic symptoms. Furthermore, this consideration allows for the timely introduction of treatment like tafamidis.

Keywords: transthyretin amyloidosis, cardiac amyloidosis, genetic testing, precision medicine, tafamidis

Presentation number: PP33 Abstract number: 110-ISABS-2026

ASSOCIATIONS BETWEEN INDOOR POLYCYCLIC AROMATIC HYDROCARBON EXPOSURE AND SERUM LIPIDOME PROFILES IN CHILDREN

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Exposure to indoor air pollutants, particularly polycyclic aromatic hydrocarbons (PAHs), represents a significant environmental health risk for children, yet the underlying biological mechanisms remain insufficiently understood. To quantify the association between indoor PAH exposure and the serum lipidome, an unsupervised principal component analysis (PCA) approach was used in 99 school-aged children from the Evidence Driven Indoor Air Quality Improvement (EDIAQI) project asthma cohort. Indoor air samples were analyzed for 11 PAHs, while serum lipid profiles were measured using high-throughput Nightingale NMR spectroscopy. Component scores were compared between high- and low-exposure groups (based on median splits) using Mann-Whitney U-tests and regression models adjusted for age, sex, and body mass index. PC1, PC2, and PC3 captured atherogenic lipoprotein burden, and HDL-versus-VLDL contrast, and triglyceride enrichment, respectively. Children with higher PAH exposure consistently showed higher PC1 and lower PC2 scores across 10 of the 11 PAHs, suggesting a subtle pro-atherogenic lipidome profile. The largest centroid separations occurred for benzo[g,h,i]perylene ($\Delta PC1 = 3.42$) and fluoranthene ($\Delta PC1 = 3.21$). A trend toward a positive association was found between benzo[a]pyrene and PC4 ($\beta = 0.35$, $p = 0.067$). Indoor PAH exposure appears to be modestly but consistently associated with a pro-atherogenic lipid profile in children.

Keywords: polycyclic aromatic hydrocarbons (PAHs), lipidomics, principal component analysis (PCA), indoor air pollution, environmental health

Presentation number: PP34 Abstract number: 42-ISABS-2026

FAMILIAL PORPHYRIA CUTANEA TARDA TYPE II ASSOCIATED WITH A NOVEL UROD I334T VARIANT AND HFE H63D

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Porphyria is a group of rare metabolic disorders characterised by the buildup of porphyrins due to an abnormal heme biosynthesis pathway. Porphyria cutanea tarda (PCT) type II manifests as light-sensitive blistering cutaneous lesions and is caused by a heterozygous mutation in the *UROD* gene, which leads to a 50% reduction in uroporphyrinogen decarboxylase activity. However, symptoms develop when residual enzyme activity drops below a threshold of 25%. Some factors that contribute to such reduction of UROD activity are alcohol use, hepatitis C and genetic hemochromatosis. Genetic hemochromatosis is commonly caused by a homozygous C282Y mutation in the *HFE* gene. The H63D variant, which is prevalent in Mediterranean populations, does not cause significant iron overload on its own, but it may contribute to some abnormalities in its metabolism and, as such, is found to contribute to PCT onset and severity, especially when combined with other risk factors. We present the case of a 43-year-old male who was referred due to abnormal porphyrin and bilirubin findings. O/E the patient had vesicles and bullae, which had recently become less pronounced. His brother was diagnosed with porphyria, but genetic analysis was not conducted. The patient's father also reported he'd had similar hand lesions in his thirties, which spontaneously resolved after a few years. Genetic testing was performed, and the report showed both a heterozygous variant of uncertain significance (VUS) c.1001T>C (I334T) in the *UROD* gene and a heterozygous pathogenic variant c.187C>G (H63D) in the HFE gene. To clarify the clinical significance of the VUS in the *UROD* gene, segregation analysis was recommended, and subsequently, the patient's father and brother were tested. The same *UROD* VUS was detected in both. In this case, segregation of the *UROD* VUS in multiple family members provided evidence supporting its pathogenicity and highlighted its potential as a novel disease-causing variant in PCT type II.

Keywords: porphyria, porphyria cutanea tarda, *HFE*, *UROD*, whole-genome sequencing

Presentation number: PP35 Abstract number: 92-ISABS-2026

PERSONALIZED HEALTH AND PERFORMANCE OPTIMIZATION IN PROFESSIONAL ATHLETES: INSIGHTS FROM ACE AND ACTN3 GENETIC POLYMORPHISMS

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Athletic performance arises from a complex interplay of genetic, physiological and environmental determinants. Among the most extensively investigated genetic contributors are polymorphisms in the *ACE* and *ACTN3* genes, which have been linked to endurance and power-oriented phenotypes. This study aimed to examine the distribution of *ACE* and *ACTN3* polymorphisms and their association with sport type in a cohort of 85 professional athletes from Bosnia and Herzegovina. Genomic DNA was analyzed using PCR-based genotyping methods, followed by appropriate statistical analyses. The *ACE* ID and *ACTN3* RX genotypes were the most prevalent across the cohort, indicating a predominance of mixed performance profiles. The*ACTN3* XX genotype was underrepresented, consistent with its lower frequency among elite athletes. A higher prevalence of the *ACTN3* RR genotype and R allele was observed in power-oriented athletes, supporting its role in fast-twitch muscle performance. In contrast, *ACE* genotype distribution did not show significant variation between sport types. Notably, combined genotype analysis revealed that the *ACTN3* RR and *ACE* DD "power" genotype combination was more frequent among power athletes, suggesting potential additive effects of multiple polymorphisms. However, logistic regression analysis did not identify any genetic markers as significant predictors of sport type, underscoring the multifactorial nature of athletic performance. Based on these findings, a preliminary framework for personalized training recommendations was developed, integrating genotype profiles with performance-related characteristics. These results support the potential of data-driven approaches, with future work focused on the development of artificial intelligence–based models to enhance predictive accuracy and enable scalable personalization in athlete training and performance optimization.

Keywords: *ace*, *actn3*, personalized health, polymorphisms, performance optimization

Presentation number: PP36 Abstract number: 100-ISABS-2026

INTEGRATED CLINICAL, BIOMECHANICAL AND MOLECULAR PROFILING OF GENERALIZED JOINT LAXITY AS A RISK FACTOR FOR ACL INJURY: PRELIMINARY RESULTS

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This study aimed to systematically investigate clinical, biomechanical, and molecular correlates of generalized joint laxity as a potential risk factor for anterior cruciate ligament (ACL) injury. A total of 200 participants aged 18–50 years without prior ACL rupture were prospectively enrolled and stratified according to Beighton score (≥6 and <6). All participants underwent bilateral knee magnetic resonance imaging to exclude ACL pathology, comprehensive orthopedic examination, and objective biomechanical assessment, including instrumented Lachman testing and rotational stability analysis performed using the KiRA device. Peripheral blood and urine samples were obtained from all participants for targeted genetic, and glycomic analyses, as well as quantification of collagen degradation products, with analyses currently underway. Each group comprised 100 participants. The mean Beighton score was higher in females (5.86) compared to males (4.51). No statistically significant differences were observed between groups in anterior tibial translation. However, the Beighton <6 group demonstrated significantly lower rotational instability values, indicating greater rotational knee stability compared to the hypermobile cohort. These findings suggest that generalized joint laxity may preferentially influence rotational knee stability, thereby contributing to ACL injury susceptibility. Ongoing molecular analyses, including genetic profiling, glycan characterization, and assessment of collagen turnover biomarkers, are expected to further elucidate the underlying pathophysiological mechanisms and support the development of predictive, personalized risk stratification models.

Keywords: ACL, Beighton score, joint laxity, glycans, genetics

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GLYCOLYMPUS: A MODULAR FRAMEWORK FOR INTEGRATED PROCESSING AND VISUALIZATION OF GLYCOMICS DATA

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Glycans are complex carbohydrate structures attached to proteins and lipids that play essential roles in cellular communication, immune response, and disease processes. Their comprehensive study, glycomics, is increasingly important for understanding biological systems and identifying biomarkers. However, glycomics data analysis typically involves multiple sequential steps, including raw data processing, electropherogram visualization, quantitative comparison, and report generation. These steps are often performed using separate tools, resulting in fragmented workflows, reduced reproducibility, and increased potential for user-induced errors. To address these challenges, we developed GlycOlympus, a modular desktop platform designed to integrate key components of glycan data analysis within a single, interactive environment. The platform consolidates essential functionalities, including input/output generation, electropherogram visualization, glycan data export, comparative analysis, and automated report generation. It supports workflows commonly used in capillary gel electrophoresis (CGE) and ultra-performance liquid chromatography (UPLC), enabling seamless transitions between different stages of analysis without the need for external tools. The application features a graphical interface that enables structured interaction with glycomics data across multiple stages of analysis, with an emphasis on consistent data organization and workflow continuity. Its modular design allows extensibility and facilitates the incorporation of additional analytical packages. By unifying multiple analytical steps into a coherent framework, GlycOlympus improves workflow efficiency and promotes reproducibility by reducing manual data handling and tool-switching. Overall, it provides a scalable, user-oriented solution for glycomics data analysis, contributing to more standardized and efficient analytical pipelines.

Keywords: glycomics, data interpretation, UHPLC, CGE, bioinformatics

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AI-DRIVEN IDENTIFICATION OF POLLUTION-ASSOCIATED TRANSCRIPTOMIC SIGNATURES IN ALZHEIMER'S DISEASE USING RNA-SEQ DATA

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Air pollution, particularly fine particulate matter (PM2.5 and PM10), has been increasingly associated with neurodegenerative diseases, including Alzheimer's disease. However, the molecular mechanisms linking environmental exposure to neurodegeneration remain insufficiently understood. In particular, the integration of transcriptomic data with AI methods offers a promising approach for identifying disease-associated molecular signatures. In our study, we employed a machine learning-based framework to analyze publicly available RNA sequencing data from human hippocampal tissue (GEO accession: GSE161199). The dataset includes transcriptomic profiles from Alzheimer's disease patients and age-matched controls. After preprocessing and normalization, feature selection was performed to identify the most informative genes associated with disease status, with a specific focus on genes previously reported to be responsive to oxidative stress, inflammation and environmental pollutants. Supervised ML models (Random Forest classifier and Support Vector Machine) were trained to distinguish between Alzheimer's disease and control samples. Model performance was evaluated using cross-validation. To enhance interpretability, XAI methods, including SHAP, were applied to identify key genes contributing to model predictions. The analysis revealed a subset of genes involved in neuroinflammation, mitochondrial dysfunction and epigenetic regulation, which are also implicated in responses to air pollution exposure. These findings suggest a potential mechanistic link between environmental pollutants and transcriptomic alterations observed in neurodegenerative diseases. This study demonstrates the feasibility of integrating AI with publicly available multi-omics datasets to uncover environmentally relevant molecular signatures in Alzheimer's disease. The proposed approach highlights the potential of AI-driven models in identifying early biomarkers associated with environmental risk factors.

Keywords: air pollution, PM, Alzheimer's disease, ecogenotoxicology, neurodegenerative disorders

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OMICS APPROACH FOR DISCOVERING IMMUNE CANDIDATE GENES IN MULTIPLE SCLEROSIS

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Multiple sclerosis (MS) is a complex immune-mediated disorder with a polygenic architecture. Identification of causal genes remains challenging due to the indirect nature of genome-wide association studies (GWAS). We propose an integrative multi-omics framework for prioritizing immune-relevant candidate genes and supporting targeted panel design. Candidate genes are derived from published literature, GWAS datasets, and validated susceptibility variants from the International Multiple Sclerosis Genetics Consortium (IMSGC). Gene prioritization was performed using locus-to-gene (L2G) scores from Open Targets Genetics, integrating genetic distance, eQTL/sQTL colocalization, chromatin interaction data, and predicted functional impact of coding variants. Immune relevance is refined using curated pathway databases (Reactome, KEGG, ImmPort). Multi-layered evidence from transcriptomic, epigenomic, and proteomic datasets is integrated into a unified scoring framework to stratify genes into high- and medium-confidence categories numbering 100-150 genes in Next Generation Sequencing (NGS) panel. The study cohort includes 300 MS patients, stratified into 100 familial cases, 150 sporadic cases, and 50 individuals from the Gorski Kotar region, which represents a region of particular epidemiological interest for MS research in Croatia. The integrative framework is expected to prioritize immune-related genes supported by convergent genetic and functional evidence. High-confidence candidates are anticipated to converge on key biological pathways, including cytokine signaling, lymphocyte activation, and neuroinflammatory regulation. This approach enables systematic gene prioritization and supports the design of a targeted NGS panel, facilitating downstream sequencing and comparative analysis across familial, sporadic, and regionally enriched MS subgroups.

Keywords: genome-wide association studies, immunology, multi-omics, multiple sclerosis, next generation sequencing panel

Presentation number: PP40 Abstract number: 26-ISABS-2026

LIFESTYLE AND DIETARY INFLUENCES ON GUT MICROBIOTA DIVERSITY AND COMPOSITION IN A HEALTHY CROATIAN COHORT

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The human gut microbiota plays a key role in health and is influenced by different environmental factors. This study examined associations between microbial diversity and composition in the gut with lifestyle and dietary factors in a healthy Croatian cohort (N=60). 16S rRNA gene sequencing was performed on the Illumina platform, following standard procedures. Genetic data were processed in R using the phyloseq package. Alpha diversity was analyzed using linear models and Kruskal–Wallis tests, while beta diversity (Bray–Curtis) was assessed using PERMANOVA. Genus-level relative abundance profiles were generated to characterize microbial composition. Beta diversity analysis revealed significant differences in microbial community structure associated with gender (p=0.024), physical activity (p=0.013), bloating frequency (p=0.002), and weekly wine consumption (p=0.047). In contrast, only age showed a marginal association with alpha diversity. Wine consumption was most significantly associated with variation in Faecalibacterium (p=0.011) and Ruminococcus (p=0.007), physical activity with [Eubacterium] hallii group (p=0.004, q=0.031) and Bifidobacterium (p=0.003, q=0.031), and meat consumption with Anaerostipes (p=0.003, q=0.025) and [Eubacterium] hallii group (p<0.001, q=0.005). Soft drink consumption showed limited associations. Relative abundance profiles showed consistent but modest shifts across lifestyle categories, including trends toward higher proportions of short-chain fatty acid–producing genera (e.g., Faecalibacterium, Roseburia) with increased physical activity and lower processed beverage intake. These taxa are commonly considered beneficial due to their role in short-chain fatty acid production, maintenance of gut health and healthy aging in general. These findings suggest that while lifestyle and dietary factors may not substantially alter microbial richness, they are associated with compositional shifts in specific bacterial taxa.

Keywords: gut microbiota, lifestyle, diet, health, bacterial taxa

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INTEGRATIVE EPIGENETIC AND TRANSCRIPTOMIC PROFILING REVEALS DNA METHYLATION–GENE EXPRESSION RELATIONSHIPS IN IDIOPATHIC ALS

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Epigenetic modifications capture gene–environment interactions and reflect biological processes associated with various diseases and aging. These features offer a potential molecular record of tissue injury accessible through peripheral biofluids. In idiopathic amyotrophic lateral sclerosis (ALS) cases, noninvasive biomarkers that reflect neuronal pathology remain limited. Recent studies have explored the circulating cell-free DNA (cfDNA) methylome, linking epigenetic alterations to neurodegenerative pathways, such as endocytosis. However, the cfDNA methylome in idiopathic ALS, derived from dying neurons and glia, and its relationship to brain transcriptional dysregulation remain unclear. To address this gap, we generated high-resolution DNA methylation profiles from cfDNA and matched genomic DNA (gDNA) in 48 blood samples, including 24 individuals with idiopathic ALS and 24 controls. Enzymatic methylation sequencing was employed to preserve DNA integrity and maximize signal recovery from low-input cfDNA. Differentially methylated regions were identified using a tiling window approach ($q \leq 0.01$; ≥10% methylation difference). Comparisons between cfDNA- and gDNA-derived methylation profiles were used to distinguish injury-associated epigenetic signals from stable blood cell–intrinsic methylation states, and CelFiE-based deconvolution was applied to infer cellular contributors to cfDNA. Circulating methylation features were integrated with brain-derived bulk and single-nucleus RNA sequencing data to prioritize pathways relevant to neurodegeneration and metabolic dysregulation. This analysis identified concordant methylation and transcriptional alterations near key metabolic regulators, including ACSS2, PCCB, and ABAT. Epigenetic age estimates derived from cfDNA were not directly comparable to clocks calculated from matched gDNA, highlighting limitations of existing epigenetic aging models when applied to cfDNA and underscoring the need for cfDNA-optimized clocks.

Keywords: dna methylation, amyotrophic lateral sclerosis, epigenetic aging, multi-omics integration

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PHYSICAL ACTIVITY AND IgG N-GLYCOMIC PROFILE: A CROSS-SECTIONAL STUDY AMONG MEDICAL STUDENTS

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The aim of this study was to examine physical activity (PA) among medical students, to measure their IgG N-glycan composition and determine differences in the IgG N-glycan profile according to PA category, and to explore associations between specific types of PA and IgG N-glycan traits. This cross-sectional study was conducted in December 2023 among first- and second-year medical students at the University of Osijek, Croatia. PA was assessed using the International Physical Activity Questionnaire - Short Form (IPAQ-SF) questionnaire. IgG was isolated from plasma, and its N-glycan composition was analyzed by capillary gel electrophoresis (27 IgG N-glycan peaks, and eight derived glycan traits representing shared structural features). Results: A total of 79 students (23 males), median age 20 years (interquartile range 19 – 20), completed the questionnaire. According to the IPAQ-SF, 23 participants were classified as health-enhancing physical activity (HEPA)-active, while 56 were categorized as non–HEPA-active. After Benjamini–Hochberg false discovery rate (FDR) correction, no significant differences were found in IgG N- glycan peaks or traits between groups. Initial correlations were observed between vigorous PA and monogalactosylated glycan traits (G1) ($\rho = -0.24$, $P = 0.034$), total PA and digalactosylated glycan traits (G2) ($\rho = -0.31$, $P = 0.005$) and G1 ($\rho = 0.25$, $P = 0.024$), and walking and G2 ($\rho = -0.23$, $P = 0.046$), but none remained significant after FDR correction ($P \geq 0.218$). PA in medical students was not prevalent. No significant differences or associations between PA and the IgG N-glycan profile were identified after multiple testing correction in this young, homogeneous population. Larger longitudinal and interventional studies with objective PA assessment are needed. In this context, artificial intelligence–based multi-omics integration may help identify complex PA–IgG glycosylation interactions.

Keywords: glycomics, immunoglobulin G, medical students, N-glycans, physical activity

AI IN PRECISION ONCOLOGY

Presentation number: PP43 Abstract number: 18-ISABS-2026

CIRCULATING TUMOR DNA AND MULTIPARAMETRIC MRI BIOMARKERS FOR MONITORING RESPONSE TO STEREOTACTIC RADIOTHERAPY IN RENAL CELL CARCINOMA: A REVIEW OF EVIDENCE AND PERSPECTIVES

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This review synthesizes current evidence on circulating tumor DNA (ctDNA) and multiparametric MRI biomarkers (ADC, K^trans, perfusion/excretion parameters) for assessing response and monitoring patients with renal cell carcinoma (RCC) after stereotactic body radiotherapy (SBRT). It also highlights opportunities for integrating these biomarkers to enable earlier detection of progression, with emphasis on the limitations of existing literature, particularly for ctDNA, where data are heterogeneous and mostly derived from small or extrapolated cohorts. A systematic search of PubMed, EMBASE, and Web of Science identified studies evaluating ctDNA and MRI biomarkers in RCC after SBRT. Key aspects reviewed include ctDNA detection approaches (tumor informed vs. tumor agnostic), detectability and temporal association with imaging findings, correlations of diffusion and perfusion MRI parameters with volumetric response and clinical outcomes, and concepts for combining ctDNA and MRI signals in predictive models. Due to methodological variability and few studies evaluating both biomarker types concurrently, a qualitative synthesis was performed. Ultrasensitive ctDNA assays detect molecular residual disease (MRD) in ~60% of oligometastatic RCC patients, predicting progression with a lead time of 13 to 14 weeks. DCE MRI parameters show strong correlations with volumetric response at 12–24 months, while volumetry outperforms RECIST and transient enlargement occurs in ~20% of cases. Integrating ctDNA and MRI biomarkers improves prognostic accuracy: serial ctDNA negativity with favorable MRI parameters identifies low risk patients, whereas persistent ctDNA positivity indicates need for earlier intervention. Combined ctDNA–MRI assessment represents a promising strategy for personalized post SBRT monitoring, though prospective studies are needed to standardize integration protocols and define robust biomarker thresholds.

Keywords: precision oncology, multiparametric MRI, circulating tumor DNA, stereotactic radiotherapy, renal cell carcinoma

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CLINICAL UTILITY OF TUMOR MOLECULAR PROFILING IN BILATERAL CLEAR CELL RENAL CELL CARCINOMA

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The Von Hippel-Lindau (VHL) protein is an E3 ligase that functions as a tumor suppressor, clinically associated with clear-cell renal cell carcinoma (ccRCC). Loss of VHL function leads to activation of hypoxia-inducible transcription factors (HIF), which can promote tumorigenesis. Inactivation of the *VHL* gene, located on chromosome 3p25, is the most common abnormality in ccRCC. This case report aims to highlight the importance of tumor molecular profiling in identifying actionable mutations and enabling the initiation of targeted therapy in patients with ccRCC. We present a 51-year-old male patient with bilateral ccRCC detected during routine ultrasound examination. Imaging revealed a mass in the upper pole of the left kidney measuring 27 x 26 x 22 mm and a smaller mass of 12 mm in the upper pole of the right kidney. MSCT confirmed hypervascular lesions suspicious of RCC. The patient underwent surgical resection of the left renal tumor and histopathological examination confirmed ccRCC. Tumor molecular profiling was subsequently performed using next-generation sequencing to identify potential actionable genomic alterations. Tumor molecular profiling identified a pathogenic *VHL G144** mutation, with a variant allele frequency of 30.03%. Copy number analysis additionally revealed 3p and 3q chromosome deletions. The identified alteration represents a potential therapeutic target, as inhibitors targeting vascular endothelial growth factor receptors and HIF proteins may be efficient in treating tumors with *VHL* alterations. Belzutifan is a HIF-2 alpha inhibitor approved for the treatment of adult patients with ccRCC associated with *VHL* mutation, which could therefore be considered as a potential therapeutic option in this case. This case highlights the clinical value of tumor molecular profiling in RCC. Identification of actionable mutations can facilitate personalized treatment strategies and expand therapeutic options in patients with ccRCC.

Keywords: renal cell carcinoma, next-generation sequencing, targeted molecular therapy, Von Hippel- Lindau, precision oncology

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PROGRAMMABLE DNA ORIGAMI BIOSENSORS FOR RAPID MICRORNA-BASED CANCER BIOMARKER DETECTION IN PRECISION ONCOLOGY

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Rapid and accurate detection of cancer-associated biomarkers is essential for precision oncology, particularly for early detection, diagnosis, therapy monitoring, and relapse. Circulating microRNAs (miRNAs) are powerful liquid-biopsy biomarkers, but their clinical utility is limited by detection technologies that require amplification, centralized laboratories, and complex workflows.Therefore, there is a critical need for rapid, low-cost, and highly sensitive detection. We introduce a DNA-origami "book" biosensor that detects breast cancer–associated miRNAs in biological fluids. It features a reconfigurable DNA nanostructure that opens upon miRNA binding, producing an optical signal. Readouts based on FRET and fluorescence quenching rely on optimized donor–acceptor fluorophore pairs for robust, high–dynamic- range detection. To enhance sensitivity in biofluids, the lock mechanism was optimized using fluorine- modified bases and locked nucleic acids, while diethylene glycol improved stability and target accessibility. These features enable specific miRNA detection in buffer, human serum, and plasma. Using breast cancer–associated miR-21, we achieve detection limits of 0.69 pM in biofluids within 10 min. The platform is validated using plasma samples from primary and metastatic breast cancer patients and benchmarked against qPCR. Multiplexed detection is demonstrated by measuring miR-21 and miR-7a in plasma samples. In parallel, a qPCR-validated miRNA expression database from breast cancer patients is being developed to train artificial intelligence models for miRNA signature discovery. This AI-based framework identifies miRNA patterns linked to cancer subtype, stage, and metastasis, enabling data-driven patient stratification. The integration of DNA nanotechnology and AI establishes a new paradigm for precision oncology through rapid, amplification-free liquid biopsies, improving early detection, relapse prediction, therapy monitoring, and personalized care via tests.

Keywords: miRNA, DNA origami, AI, breast cancer, biosensing

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MOLECULAR CHARACTERIZATION OF COLON ADENOCARCINOMA USING WES/WTS TO GUIDE TARGETED TREATMENT STRATEGIES

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In the era of precision medicine, oncology is increasingly based on the understanding of molecular tumor traits, enabling the use of targeted therapies directed at specific genetic alterations. This is made possible by advances in medical genetics and next-generation sequencing (NGS). The aim of this abstract is to highlight the importance of tumor molecular profiling in order to enable effective targeted therapy with minimal adverse effects. A 60-year-old woman with a diagnosis of medium grade mucinous colon adenocarcinoma o(pT3N1b, G II), without extramural vascular invasion, presented to the St. Catherine Hospital for molecular tumor profiling. The analysis was performed using whole exome sequencing (WES) and whole transcriptome sequencing (WTS), taking into account 1190 clinically relevant genes linked to approved therapies. A *KRAS G13D* mutation was identified, which is associated with treatment options using MEK- or ERK-targeted inhibitors such as Binimetinib, Cobimetinib, and Trametinib. Additionally, sequencing identified a *PIK3CA E545K* mutation, which may respond to selective PI3K inhibitors. A third alteration was found in the tumor suppressor gene *FBXW7*. Several promising clinical studies support the use of the PKMYT1 inhibitor Lunresertib in combination with the ATR inhibitor Camonsertib in patients with *FBXW7*-mutated solid tumors. For the alterations detected in the *SMAD2* and *APC* genes, there were currently no FDA-approved therapies or NCCN-compendium-listed treatment options. In summary, close and effective collaboration between oncology and medical genetics holds the potential for targeted therapy application and improved treatment outcomes for patients with malignancies. Genetic counseling is crucial, as it places sequencing results into a clinical context and guides the selection of targeted therapy based on identified molecular alterations.

Keywords: colon adenocarcinoma, genetic counseling, gene expression profiling, molecular targeted therapy, precision oncology

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MULTI-MODAL GENOMIC PROFILING IN CANCER DIAGNOSTICS: INTEGRATING GERMLINE AND LIQUID BIOPSY FINDINGS IN OVARIAN CANCER

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Comprehensive genomic profiling is increasingly transforming the management of cancer. By integrating data from germline and somatic genomic profiles, physicians are offered a more complete understanding of tumor biology, enabling risk assessment and targeted treatment. We present the case of a 50-year-old female patient diagnosed with ovarian cancer who underwent a comprehensive genomic evaluation, including liquid biopsy, whole genome sequencing (WGS), as well as pharmacogenomic and nutrigenomic testing to support a personalized approach to care. Initial liquid biopsy identified two pathogenic variants: a frameshift variant in the *BRCA1* gene (c.1938_1947del; p.Ser646ArgfsTer2) with a variant allele frequency (VAF) of 45.30%, and a splice-site variant in the *RB1* gene (c.607+1G>C, VAF 2.27%). The *BRCA1* alteration is associated with homologous recombination deficiency and has therapeutic implications, including eligibility for PARP inhibitor therapy. Subsequent germline WGS analysis from peripheral blood, targeting hereditary cancer, cardiometabolic, and carrier screening panels confirmed the presence of a heterozygous pathogenic *BRCA1* variant, despite variable representation in circulating tumor DNA. Additional findings included carrier status for pathogenic variants in the *SBDS, MYO7A, UGT1A1* and *HFE* genes, as well as a likely pathogenic variant in the *HAVCR2* gene. This case underscores the clinical value of combining liquid biopsy with germline WGS to distinguish somatic versus inherited variants and to refine risk assessment. Furthermore, integration of pharmacogenetic and nutrigenetic profiling enabled a personalized approach to patient management. Genetic counseling and targeted therapeutic recommendations, including consideration of PARP inhibitors, were provided based on the comprehensive genomic findings.

Keywords: ovarian cancer, *BRCA1* gene, liquid biopsy, whole-genome sequencing, precision oncology

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SMARCB1 LOSS GUIDES TARGETED THERAPY IN PEDIATRIC DEDIFFERENTIATED CHORDOMA

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Chordoma is a rare primary malignant bone tumor of the axial skeleton that arises from remnants of the fetal notochord. The World Health Organization (WHO) classifies chordomas into three subtypes: conventional, dedifferentiated, and poorly differentiated, which all differ in histopathology and prognosis. A six-year-old girl diagnosed with dedifferentiated chordoma was referred for molecular tumor profiling to identify potential mutations for targeted therapy. She initially presented to her physician with persistent coccygeal pain following trauma that occurred five months earlier. A comprehensive diagnostic workup was performed, including ultrasound, magnetic resonance imaging, and biopsy. Biopsy confirmed the diagnosis of chordoma and showed both dedifferentiated and conventional components. The disease was classified as stage IV with suspected pulmonary metastases. Molecular analysis showed low tumor mutational burden, a *TERT* promoter mutation (c.-124C>T), and a *SMARCB1* deletion (exons 1–7). The tumor was *BRAF* wild-type, microsatellite stable, and negative for homologous recombination deficiency signature (HRDsig). Although no standard actionable genomic alterations were identified, the *SMARCB1* loss suggested potential sensitivity to EZH2 inhibition. Based on these findings, a multidisciplinary tumor board recommended surgical resection followed by adjuvant radiotherapy, with evaluation of pulmonary lesions. In parallel, systemic therapy with an EZH2 inhibitor was proposed as a targeted approach informed by the molecular profile. This case demonstrates that molecular tumor profiling can uncover biologically actionable vulnerabilities even in the absence of canonical targetable mutations. Specifically, *SMARCB1* deletion may serve as a predictive biomarker for EZH2 inhibition, highlighting the clinical value of comprehensive genomic profiling in guiding precision oncology strategies for rare pediatric tumors.

Keywords: chordoma, EZH2, molecular profiling, precision oncology, *SMARCB1*

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PCDH9 EXPRESSION AND PROGNOSTIC SIGNIFICANCE IN CLEAR CELL RENAL CELL CARCINOMA

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Clear cell renal cell carcinoma (ccRCC) is the most prevalent subtype of renal cancer, frequently diagnosed at advanced stages and associated with poor prognosis. Protocadherin 9 (PCDH9) is an emerging tumor suppressor increasingly recognized across multiple malignancies, yet its role in ccRCC and its relationship with epithelial-mesenchymal transition (EMT) remain poorly characterized. This study aimed to investigate PCDH9 expression, its co-regulation with epithelial adhesion molecules, and its prognostic significance in ccRCC. Immunofluorescence analysis was performed on FFPE tissue sections from 48 ccRCC patients (31 low-grade, 17 high-grade) and adjacent normal renal cortex, and findings were validated using the TCGA-KIRC dataset via GEPIA2/GEPIA3. PCDH9 mRNA was significantly downregulated in ccRCC tumors compared to normal kidney tissue. At the protein level, a strong trend toward reduced PCDH9 immunoexpression was observed in tumor tissue. A strong positive correlation between PCDH9 and CDH1 (E-cadherin) present in normal kidney was completely lost in ccRCC, indicating disruption of epithelial adhesion molecule co-regulation during tumorigenesis. PCDH9 downregulation may be mechanistically linked to VHL inactivation, which occurs in the majority of sporadic ccRCC cases. Unexpectedly, PCDH9 showed positive correlations with EMT-inducing transcription factors in tumor tissue, supporting a partial rather than classical EMT phenotype in ccRCC. In univariate survival analysis, high PCDH9 expression was significantly associated with improved overall survival, though this association was lost in multivariate analysis, likely reflecting its co-regulation with β-catenin. These findings collectively support a tumor-suppressive role for PCDH9 in ccRCC and suggest its potential utility as a prognostic biomarker, warranting further functional and prospective clinical validation.

Keywords: PCDH9, ccRCC, EMT, prognostic biomarker, clear cell renal cell carcinoma

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MOLECULAR PROFILING IN ENDOMETRIAL SARCOMAS: IDENTIFYING POTENTIAL THERAPEUTIC TARGETS

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Next-generation sequencing (NGS) is a modern technology for parallel DNA and RNA sequencing that enables detailed molecular profiling of malignant tumors. It provides key information for personalized therapy particularly in rare, aggressive tumors such as endometrial stromal sarcoma (ESS). We present the case of a 53-year-old woman who was diagnosed with high-grade ESS, confirmed by pathohistological findings after hysterectomy and bilateral adnexectomy. Chemotherapy included six cycles of the AI protocol but PET/CT demonstrated progression so second-line treatment with trabectedin via an elastomeric pump was initiated. Tumor genetic profiling using NGS was performed (whole exome and whole transcriptome sequencing), which revealed an oncogenic variant in the *PTEN* gene (c.402_404del, p.Met-134del), a homozygous deletion of *CDKN2A/B*, and *ZC3H7B::BCOR* gene fusion. Considering that chemotherapy/radiotherapy do not demonstrate improved survival rates and overall survival ranging 9-24 months, a new approach is needed. Although therapies targeting *PTEN* mutations are approved in breast cancer, their applicability in ESS is unknown. Laboratory data suggest that beta isoform-selective PI3K/mTOR inhibitors may be effective against *PTEN*-mutant ESS. Considering BCOR fusions have been identified in ESS and BCOR is involved in epigenetic regulation, it is possible that epigenetic inhibitors (EZH2/HDAC inhibitors) could be a therapeutic option but there is no clinical evidence to support this. *CDKN2A* deletion is common and cells harboring this deletion could potentially be sensitive to CDK4/6 inhibitors (palbociclib/ribociclib/abemaciclib). We highlight the importance of tumor molecular profiling and precision oncology, especially in patients diagnosed with aggressive tumors with high mortality. With the relevant genetic mutations known, future research should focus on exploring potential therapies to address the gap in clinical evidence for targeted therapy application.

Keywords: tumor molecular profiling, high-grade endometrial stromal sarcoma, next-generation sequencing, precision oncology, targeted tumor therapy

Presentation number: PP51 Abstract number: 74-ISABS-2026

MOLECULAR PROFILING IDENTIFIES TARGETABLE *CHEK2* MUTATION IN A METACHRONOUS DUCTAL CARCINOMA IN SITU

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Ductal carcinoma in situ (DCIS) is the most common form of non-invasive breast cancer and is typically identified through mammographic screening programmes. Despite multiple therapeutic options, there is a significant over-treatment risk. Genomic profiling using next-generation sequencing (NGS) has become increasingly utilized in clinical oncology, detecting target mutations and creating personalized treatment strategies. We present a case of a 53-year-old woman diagnosed with DCIS in her right breast. The patient underwent skin-sparing mastectomy followed by endocrine therapy (ET) (tamoxifen, letrozole, exemestane and anastrozole). Three years after her initial diagnosis, she underwent prophylactic contralateral skin- and nipple-sparing mastectomy with histopathological examination identifying subsequent DCIS. Due to metachronous disease, molecular profiling using next- generation sequencing (Illumina NovaSeq/NextSeq platform) was performed, enabling the detection of key solid tumor genetic variants. Profiling identified a likely pathogenic mutation in the *CHEK2* gene, a checkpoint kinase responsible for DNA repair. This finding provided biological justification for the use of olaparib or talazoparib, PARP inhibitors currently indicated as targeted therapy in other malignancies. This case highlights the potential for expanding therapeutic strategies in DCIS treatment, suggesting possible wider implementation of personalized medicine through genomic profiling in clinical decision-making.

Keywords: ductal carcinoma in situ, next-generation sequencing (NGS), *CHEK2* mutation, personalized oncology, PARP inhibitors

Presentation number: PP52 Abstract number: 85-ISABS-2026

USING DEEP LEARNING TO PREDICT DUSP1 EXPRESSION DYNAMICS IN GEFITINIB-SENSITIVE AND -RESISTANT LUNG CANCER CELLS: TOWARD PRECISION ONCOLOGY

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The goal of this study was to investigate dynamic gene expression patterns of DUSP1, a stress response gene implicated in drug resistance, in gefitinib-sensitive PC9 and gefitinib-resistant PC9GRM2 lung cancer cells and to evaluate deep learning models for predicting temporal expression trends. The data were obtained from the Gene Expression Omnibus (GSE34228), focusing on cells treated with Epidermal Growth Factor (EGF) and a combination of EGF and an EGFR inhibitor (IRS) over 24 hours. Data for DUSP1 were extracted and filtered to create subsets for specific cell line and treatment conditions (PC9 + EGF, PC9 + EGF + IRS, PC9GRM2 + EGF, PC9GRM2 + EGF + IRS). Two models were compared: Long Short-Term Memory (LSTM) network with an input window of 8 and a Transformer with an input window of 20, optimized on the volatile PC9_EGF_IRS dataset. Hyperparameter tuning showed that the optimal Transformer configuration used a learning rate of 0.001 and four attention heads. Data were split 80/20 for training and testing. Results showed that sensitive PC9 cells exhibited more dynamic and volatile DUSP1 expression than resistant PC9GRM2 cells, suggesting distinct cellular responses. Combined EGF+IRS treatment induced sharper fluctuations than EGF alone. LSTM models captured general trends, but struggled with sharp peaks, showing overfitting in volatile PC9 datasets. The tuned Transformer provided smoother and more accurate predictions while still struggling with the timing and magnitude of sharp DUSP1 changes, reflecting biological complexity. On less variable PC9GRM2 data, both models performed comparably. In conclusion, our ability to predict DUSP1 expression dynamics, even with some limitations, represents a meaningful step toward applying gene expression trajectories as predictive tools. DUSP1 not only reflects cellular states under treatment and resistance, but also shows promise as a biomarker for anticipating therapeutic response in precision oncology.

Keywords: DUSP1, lung cancer, gefitinib resistance, deep learning, precision oncology

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MOLECULAR PROFILING IDENTIFIES ACTIONABLE *PTEN* MUTATION IN RECURRENT UTERINE LEIOMYOSARCOMA: A CASE REPORT SUPPORTING TARGETED THERAPY APPROACH

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Uterine leiomyosarcoma is a rare and highly aggressive malignancy with limited treatment options in the recurrent setting. Comprehensive genomic profiling using next-generation sequencing (NGS) is increasingly used to identify actionable mutations and guide precision oncology. We present a 47-year-old female patient diagnosed with uterine leiomyosarcoma (FIGO stage IC) in October 2022. The patient underwent multiple surgical procedures, including myomectomy followed by hysterectomy with bilateral adnexectomy and repeated cytoreductive surgeries due to recurrent disease. Systemic treatment included adjuvant chemotherapy (AI protocol), gemcitabine due to hypersensitivity to docetaxel, and subsequent targeted therapy with pazopanib. Despite treatment, the disease progressed with a pelvic recurrence measuring 29 × 29 × 37 mm. Due to further progression, molecular profiling was performed using hybrid- capture-based NGS (Illumina NovaSeq platform), enabling simultaneous DNA and RNA analysis from metastatic tissue. Molecular profiling results identified a pathogenic mutation in the *PTEN* gene, a key regulator of the PI3K/AKT signaling pathway. This finding provided a biologically rational therapeutic option using targeted therapy with the AKT inhibitor capivasertib in combination with fulvestrant. This case demonstrates the clinical value of repeat molecular profiling in advanced uterine leiomyosarcoma. Genomic analysis may significantly influence treatment decisions and represents an important step toward precision oncology in rare gynecologic malignancies.

Keywords: uterine leiomyosarcoma, next-generation sequencing (NGS), *PTEN* gene mutation, precision oncology, capivasertib therapy

AI IN DRUG DISCOVERY AND DEVELOPMENT

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PRESERVING HYPOXIC CONDITIONS FOR ACCURATE DRUG RESPONSE ASSESSMENT: AN OPTIMIZED FLOW CYTOMETRY APPROACH

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Hypoxia critically influences physiological and pathological processes, particularly cancer progression and drug response, yet standard in vitro analyses often fail to maintain physiological oxygen conditions, limiting translational relevance. This study aimed to develop a method for accurate cellular analysis under controlled oxygen environments using a tubing-based interface connected to a high-performance flow cytometer, which enabled more reliable investigation of hypoxia-driven cellular processes relevant to drug discovery and development. A549 lung epithelial cancer cells were cultured under hypoxic (1% O_2) and normoxic (21% O_2) conditions. A flow cytometry system with direct sampling from a hypoxia chamber was established to prevent reoxygenation. Cell viability, mitochondrial membrane potential, and intracellular hypoxia were assessed using DRAQ7, JC-1, and HypoxiTRAK probes. Results showed stable cell viability (80-95%) across conditions, while HypoxiTRAK signals confirmed that hypoxic status remained unchanged during short handling but increased significantly after 60 minutes under hypoxia and decreased upon reoxygenation. Mitochondrial membrane potential rapidly responded to oxygen shifts, with a significant increase observed within 15-60 minutes, particularly after hypoxia-to-normoxia transition, indicating fast metabolic adaptation. Even brief exposure to normoxia altered hypoxia-associated readouts. These findings demonstrate that maintaining oxygen conditions throughout analysis is essential for reliable data. The proposed approach improves physiological relevance in vitro and enhances the predictive power of drug screening models targeting hypoxia-related pathways. This research was funded by HORIZON- WIDERA-2021-ACCESS-03-01 program 382 project No. 101079489-TWINFLAG.

Keywords: hypoxia, flow cytometry, cancer, jc-1, hypoxiTRAK

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FIRST CRISPR/DCAS9 B-CELL BASED PLATFORM TO STUDY REGULATORY MECHANISMS OF IGG GLYCOSYLATION

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Immunoglobulin G (IgG) is the most abundant antibody in human blood, secreted by plasma cells, and plays a central role in adaptive immunity. Its function is regulated by glycosylation, a post-translational modification in which glycans are attached to proteins. These glycans influence IgG stability, conformation, half-life, and are critically involved in inflammatory responses. Numerous studies have shown that IgG glycosylation changes across pathological conditions, but the molecular mechanisms governing this process remain poorly understood. Insights have been gained through genome-wide association studies (GWAS) of the IgG glycome, which identified genes with previously unknown roles in glycosylation pathways. To functionally validate these genes, we developed the first CRISPR/dCas9 B-cell platform with stably integrated CRISPRa and CRISPRi tools. Using PiggyBac transposition, we generated IgG-secreting stable cell lines expressing either dCas9-VPR for transcriptional activation or dCas9-KRAB for transcriptional silencing. Taking advantage of the compatibility of Sleeping Beauty transposase with this system, we established a robust protocol for gRNA addition, achieving strong activation and silencing of gene expression. As proof of concept, we targeted key glycosyltransferases, B4GALT1 and ST6GAL1, responsible for adding galactose and sialic acid, and showed that their modulation leads to significant changes in IgG galactosylation and sialylation. Using this platform, we manipulated multiple GWAS candidate genes and demonstrated their involvement in IgG glycosylation for the first time. This CRISPR/dCas9 B-cell system enables identification of novel regulatory pathways. Since glycosylation is a critical quality attribute of IgG therapeutics, these findings may support advanced glycoengineering and future drug development.

Keywords: glycosylation, IgG, CRISPR, epigenetics, glycoengineering

Presentation number: PP56 Abstract number: 57-ISABS-2026

PATIENT-SPECIFIC TRANSCRIPTOMIC RESPONSES TO CONNEXIN-43 INHIBITION IN OSTEOARTHRITIS

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Osteoarthritis (OA) is a degenerative joint disease marked by cartilage degradation, cellular senescence, and chronic inflammation, with limited treatment options. Connexin-43 (Cx43), a gap junction protein upregulated in OA, promotes harmful intercellular communication driving senescence and tissue damage, making it a promising therapeutic target. This study evaluates TUB1, a Cx43-inhibitory peptide, in human OA chondrocytes cultured under normoxia (21% O_2) or hypoxia (2% O_2) for 48 h with or without TUB1. RNA sequencing was performed; differential expression was defined as $|log2FC| \geq 1$ and adjusted $p \leq 0.05$. Oxygen alone altered 106 genes (92 upregulated, 14 downregulated), with GO enrichment indicating response to endogenous stimulus and regulation of cell proliferation, confirming oxygen shapes the transcriptome. Pooled TUB1 analysis showed limited changes; however, patient-level analysis revealed 61–1,000 differentially expressed genes per donor, indicating high variability. Under normoxia, TUB1 reduced inflammatory and chemokine signaling and increased DNA damage response and cell cycle genes. Under hypoxia, it affected collagen metabolism and cell–matrix adhesion. GO and KEGG analyses confirmed consistent effects across senescence, immune, and extracellular matrix pathways. These findings show TUB1 induces oxygen-dependent and individualized transcriptomic changes, supporting Cx43 inhibition as a candidate for personalized OA therapy.
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Keywords: osteoarthritis, senescence, inflammation, connexin 43, hypoxia

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DEVELOPMENT OF CRISPR-DCAS9 EXPRESSION SYSTEM TO INVESTIGATE ABERRANT IGA GLYCOSYLATION

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IgA is the second most abundant immunoglobulin class in serum. In addition to providing passive immunity by neutralising pathogens at mucosal sites, it actively controls immune responses, and can drive the pathogenesis of autoimmune diseases, such as IgA nephropathy and rheumatoid arthritis. Like other antibodies, it is subject to glycosylation, a complex posttranslational modification orchestrating profound changes of protein function in health and disease. IgA1, the most abundant subclass, has two N- glycosylation and six O-glycosylation sites on each heavy chain. In cell culture, glycosylation can be modified by manipulation of gene expression using CRISPR technology. This provides a framework to test whether a gene implicated in a certain disease affects glycosylation. To study the effect of disease- associated genes on IgA1 glycosylation, we developed a system for IgA1 secretion in FreeStyle™ 293-F cells and utilised CRISPR-dCas9 to upregulate and downregulate genes of interest. We successfully designed the expression plasmid for IgA1 production, which yields enough IgA1 for LC-MS glycan analysis. Glycoprofiling of cell-secreted IgA revealed N- and O-glycosylation profiles similar to those of human derived IgA. To validate the established framework, we targeted glycosyltransferases directly involved in the process of glycosylation. We successfully upregulated and downregulated the expression of target genes which resulted in specific changes of IgA glycome composition in accordance with the function of each enzyme. Our platform represents the first cell-based CRISPR-dCas9 system to study the glycosylation of IgA. This technology will enable precise studies of aberrant IgA glycosylation, the main driver of IgA nephropathy, a debilitating condition that affects millions of people worldwide.

Keywords: IgA, glycosylation, CRISPR-dCas9, LC-MS, Golden Gate cloning

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EFFECT OF EMPAGLIFLOZIN ON PANNEXIN 1 AND CONNEXIN EXPRESSION IN THE KIDNEY

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Sodium-glucose cotransporter-2 inhibitors (SGLT2i), including empagliflozin (EMPA), revolutionized diabetes treatment, yet their effects on renal intercellular communication pathways remain unexplored, potentially influencing therapeutic benefits and adverse events. This study aimed to investigate EMPA's impact on pannexin 1 (Px1) and connexin (Cx43, Cx40, Cx45) expression in key renal cell populations. Thirteen C57BL/6J mice were divided into EMPA-treated (15 mg/kg/day in 0.2% DMSO via drinking water for 4 weeks; N=7) and control (0.2% DMSO; N=6) groups. Px/Cx expression in proximal tubule epithelium (megalin+, M+), podocytes (podocin+, POD+), and endothelium (CD31+) was quantified by flow cytometry. EMPA significantly increased Cx43 in CD31+ cells ($P<0.05$) but did not affect other Px/Cx. Px1 expression was highest in PT (M+; $P<0.001$ compared to POD+ cells). Cx43 expression was significantly higher in M+ and CD31+ cells compared to POD+ ($P<0.01$ both); Cx40 expression was highest in CD31+ cells ($P<0.01$ compared to M+ and POD+), while Cx45 expression was higher in M+ ($P<0.05$) and CD31+ ($P<0.01$) compared to POD+. In M+ PT epithelium, the highest Cx43 expression was recorded ($P<0.05$ to $P<0.0001$); POD+ and CD31+ cells showed the highest expression of Cx43 and Cx40 ($P<0.01$ to $P<0.0001$ compared to Px1 and Cx45). Enhanced endothelial Cx43 may modulate vascular function/permeability, contributing to EMPA's renal benefits and risks. These findings advance understanding of SGLT2i in renal physiology/pathology for optimized therapy. This work was funded by the Croatian Science Foundation under the projects number [HRZZ-IP-2022-10-7321] and [DOK-2025-02-5516].

Keywords: empagliflozin, SGLT2 inhibitors, connexins, pannexin 1, kidney

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CONNEXIN 43 TARGETING FOR PRECISION BIOMARKER DISCOVERY AND REGENERATIVE MODULATION IN OSTEOARTHRITIS

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Osteoarthritis (OA) is the most prevalent joint disease worldwide, characterized by impaired cartilage regeneration, chronic inflammation, and cellular senescence. Advancing precision medicine in OA requires identification of molecular regulators and biomarkers reflecting disease progression and treatment response. Connexin 43 (Cx43), a key mediator of intercellular communication and senescence, may influence OA pathology and extracellular vesicle (EV) signaling. This study evaluates the effects of a Cx43 inhibitory peptide (TUB1) on cartilage- and synovium-derived EVs to identify regenerative and biomarker- related signatures. Cartilage and synovium tissues obtained post-surgery were cultured as explants and treated with TUB1. Cellular proliferation, glycosaminoglycan (GAG) deposition, cytokine secretion, and gene expression were analyzed. EVs were isolated by size-exclusion chromatography and characterized for protein markers (Cx43, Pan1, CypA) using flow cytometry. TUB1 treatment reduced cellular senescence, confirmed by decreased β-galactosidase activity. Regenerative effects included reduced GAG release and improved extracellular matrix retention. Cytokine profiling indicated modulation of inflammatory signaling. Importantly, Cx43 expression decreased in EVs, suggesting that EV cargo reflects treatment-induced molecular changes and may serve as a biomarker of therapeutic response. Targeting Cx43 modulates senescence, inflammation, and EV composition, supporting its role in OA pathogenesis and regeneration. EV-based biomarker analysis combined with targeted intervention represents a promising precision medicine approach for patient stratification and monitoring treatment efficacy. The work was supported by the European Social Fund supported project according to the activity HORIZON- WIDERA-2021-ACCESS-03, title: "Twinning for Promoting Excellence, Ability and Knowledge to develop novel approaches for targeting inflammatory and degenerative age-related joint diseases".

Keywords: osteoarthritis, precision medicine, connexin43, biomarkers, extracellular vesicles

AI-DRIVEN INNOVATION IN TISSUE ENGINEERING AND REGENERATIVE THERAPIES

Presentation number: PP60 Abstract number: 58-ISABS-2026

THE DEVELOPMENT OF LOAD-RESPONSIVE BIOMATERIAL SCAFFOLDS FOR TARGETED EXTRACELLULAR VESICLE DELIVERY AS A NOVEL CARTILAGE REGENERATION THERAPY

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Osteoarthritis (OA) is a major cause of disability worldwide, lacking effective regenerative therapies. Precision medicine approaches in cartilage repair require controlled, localized delivery of bioactive factors adapted to the joint microenvironment. This study aimed to develop a load-responsive biomaterial scaffold enabling targeted delivery of extracellular vesicles (EVs) derived from menstrual blood–derived stromal cells (MenSCs). MenSCs were isolated and expanded in vitro, and their EVs were incorporated into surface-engineered polycaprolactone scaffolds designed to mimic cartilage structure and mechanical properties. Human bone marrow–derived mesenchymal stromal cells and chondrocytes were seeded onto scaffolds to evaluate cytocompatibility and regenerative potential. Constructs were subjected to dynamic compression to simulate the OA joint environment. Under mechanical stimulation, EV-loaded scaffolds demonstrated enhanced and sustained release compared to static conditions. The system supported high cytocompatibility and promoted chondrogenic differentiation, as evidenced by increased expression of cartilage-specific markers in mesenchymal stromal cells and chondrocytes. Notably, this effect was further enhanced in EV-functionalized scaffolds under mechanical load. These findings demonstrate that biomechanical cues can regulate therapeutic delivery and enhance regenerative outcomes. This study presents a smart, adaptive platform integrating biomaterials and EV-based therapy, advancing precision regenerative medicine. Load-responsive scaffolds represent a promising strategy for personalised cartilage repair with controlled, stimulus-driven therapeutic activity. Funding: Lithuanian Research Council, Postdoctoral Programme (P-PD-24-167, agreement No. S-PD-24-114).

Keywords: precision medicine, regenerative medicine, biomaterials, extracellular vesicles, osteoarthritis

Presentation number: PP61 Abstract number: 50-ISABS-2026

CHLOROGENIC ACID PRESERVES CHONDROCYTE VIABILITY BY MODULATING BCL-2 DEPENDENT APOPTOSIS AND INFLAMMASOME ACTIVATION

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Cartilage degeneration in growth plates arises from uncontrolled apoptosis and inflammasome activation of chondrocytes. We examined whether pharmacological reinforcement of the anti-apoptotic regulator B cell lymphoma-2 (Bcl-2) could break this pathogenic loop. In vivo, thiram fed broiler chickens were randomized to control, toxin, or chlorogenic-acid (CGA) rescue groups, while in vitro primary chondrocytes were transfected with miR-460a mimic/inhibitor or siBcl-2 and exposed to CGA. Bcl-2 axis modulation was confirmed by RT-qPCR, Western blotting, and immunofluorescence; mitochondrial integrity, cytochrome-c efflux, caspase-3/-7 cleavage, and IL-1β maturation were quantified, and apoptosis was profiled by Annexin V/PI flow cytometry. Thiram suppressed Bcl-2 and obliterated vascularization of the hypertrophic zone, doubling apoptotic chondrocytes ($p < 0.01$). Genetic silencing of Bcl-2 or over expression of miR- 460a replicated this phenotype in culture, elevating cytochrome c release by 2.4 fold and caspase-3/-7 activity by 2.1 fold, whereas Bcl-2 over expression blunted these signals. CGA restored Bcl-2 protein both in vivo and in vitro, reduced mitochondrial permeabilization, and cut apoptosis by 55% while halving IL-1β maturation ($p < 0.01$). The co-delivery of CGA with siBcl-2 markedly attenuated the siRNA's pro-apoptotic effect, implicating CGA-driven post transcriptional stabilization of the Bcl-2 pathway. Collectively, these data identify CGA as a potent small molecule shield that preserves chondrocyte viability by reinforcing Bcl- 2 dependent control of the mitochondrial apoptosis and inflammasome axis. Which offers a tractable, low toxicity strategy for precision regenerative therapy in growth plate disorders.

Keywords: chlorogenic acid, Bcl-2 signaling, chondrocyte apoptosis, inflammasome, regenerative therapy

Presentation number: PP62 Abstract number: 97-ISABS-2026

GRADED MODULATION OF *HNF1A* REVEALS DOSE-DEPENDENT CHANGES IN THE PANCREATIC BETA CELL N-GLYCOME

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HNF-1α is an essential transcription factor in the development and regulation of beta cell functions. Beta cells are key for proper glucose metabolism regulation and their impaired function or cell death leads to the development of diabetes, a disease now impacting almost 10% of the global population. Mutations in *HNF1A* cause the monogenic diabetes MODY3, but also contribute to type 2 diabetes risk. HNF-1α regulates a wide range of key pathways, including glycosylation, a post-translational modification that affects protein stability, localization, and function. Due to its complex regulatory network and analytical challenges, beta cell glycosylation has been severely understudied. To fill this gap, we created a beta cell model based on EndoC-βH1 cells stably integrating dCas9-KRAB or dCasMINI-VPR fusions. This system enables us to precisely modulate the expression of genes of interest. By targeting *HNF1A* with varying numbers of gRNAs, we generated a gradient of expression ranging from mild knockdown to almost complete silencing. Interestingly, mild silencing of *HNF1A* led to increased sialylation and complex branching, pointing toward a potential cell defense mechanism as a first response to stress. Similar increases in sialylation have been previously described in other cell types as a protective response to inflammation and environmental stress. In contrast, more severe silencing did not further increase these levels, but rather led to slight decrease compared to control levels. This trend may suggest that beta cells attempt to protect themselves by modifying their glycome during early stress exposure; however, as stress becomes more severe (notably *HNF1A* knockout causes EndoC-BH1 apoptosis) this protective signature appears to diminish. These preliminary observations demonstrate how the same gene can have profoundly different effects on glycosylation depending on its expression level.

Keywords: *HNF1A*, pancreatic beta cells, EndoC-βH1, CRISPRi/a, N-glycosylation

Presentation number: PP63 Abstract number: 103-ISABS-2026

STANDARDIZED SINGLE-INJECTION LEUKOCYTE-RICH VERSUS LEUKOCYTE-POOR PLATELET-RICH PLASMA FOR MILD-TO-MODERATE KNEE OSTEOARTHRITIS

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Current literature suggests that leukocyte-poor platelet-rich plasma may provide better clinical outcomes than leukocyte-rich formulations in knee osteoarthritis, yet uncertainty remains when both products are prepared as standardized, high-dose PRP. The aim of this randomized double-blind study was to compare standardized leukocyte-rich PRP (LR-PRP) and leukocyte-poor PRP (LP-PRP) in patients with mild-to- moderate knee osteoarthritis and to determine whether leukocyte content influences outcome when platelet dose is quantified. Adults with symptomatic Kellgren-Lawrence grade 1-3 knee osteoarthritis received a single ultrasound-guided intra-articular injection of LR-PRP or LP-PRP and were followed for 12 months. Clinical outcomes were assessed at baseline and at 1, 3, 6, and 12 months using VAS, WOMAC, and KOOS scores. Both groups showed significant longitudinal improvement in pain, stiffness, function, sport/recreation, and knee-related quality of life. The greatest improvement was observed early after treatment and was maintained through 12 months. No statistically significant differences were found between LR-PRP and LP-PRP at any follow-up time point for the principal outcomes. Exploratory analyses also showed no significant difference in thrombocyte values between groups and no significant difference in responder rates between LR-PRP and LP-PRP. In conclusion, when administered as a single injection using a standardized and quantified protocol, both LR-PRP and LP-PRP provided significant and durable clinical benefit in mild-to-moderate knee osteoarthritis, without evidence of superiority of either leukocyte profile. These findings highlight the importance of PRP standardization and platelet-dose reporting in future clinical studies.

Keywords: knee osteoarthritis, platelet-rich plasma, leukocyte-rich PRP, leukocyte-poor PRP, orthobiologics

Presentation number: PP64 Abstract number: 78-ISABS-2026

DEVELOPMENT OF A PATIENT-SPECIFIC MECHANICAL FINGER PROSTHESIS BASED ON MULTI-MATERIAL ADDITIVE MANUFACTURING

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This work presents the design and fabrication of a personalized mechanical finger prosthesis developed based on the patient's body geometry. The device was designed as a tendon-driven system, enabling finger motion through a cable (tendon) mechanism that translates external actuation into functional grasping movement. The prosthesis was manufactured using multi-material additive manufacturing, combining a rigid nanocomposite polylactic acid (PLA) reinforced with copper (PLA-Cu) for structural components and thermoplastic polyurethane (TPU) for flexible functional elements. This combination enabled the integration of stiffness and elasticity within a single fabrication process, improving both structural stability and user interaction. The proposed design methodology emphasizes patient-specific geometry acquisition and direct digital adaptation of functional prosthetic systems, enabling rapid customization without the need for traditional tooling. Particular attention was given to the integration of compliant elements to enhance grip performance and user comfort. The developed prosthesis was tested in real conditions by the end user, confirming its functional effectiveness and satisfactory usability. This work was created as a result of the research project no. 2024/53/N/ST8/01291, titled Experimental analysis of manufacturing methods for flexible coatings used to enhance the performance of hand prosthesis components, funded by the National Science Centre, Poland.

Keywords: finger prosthesis, patient-specific design, additive manufacturing, tendon-driven mechanism, TPU

Presentation number: PP65 Abstract number: 114-ISABS-2026

STEM CELL THERAPY IN PATIENTS WITH PERIPHERAL ARTERIAL DISEASE

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Peripheral arterial disease (PAD) affects over 65% of patients over 60 and results mainly from atherosclerosis of lower limb arteries, causing obstructed blood flow. Clinical manifestations range from intermittent claudications to critical limb ischemia (CLI), with 10-40% of CLI patients undergoing major limb amputation within six months of diagnosis. Revascularization including surgery or endovascular treatment is mandatory, whilst 20-45% of CLI patients are ineligible, defining the "no-option" group with poor prognosis. This highlights the need for novel therapies, with stem cell therapy emerging as a potential option. This review aimed to assess whether stem cell therapy can enhance quality of life in no-option patients. A narrative review of relevant biomedical literature was conducted using predefined search terms. Stem cells exert therapeutic effects primarily through paracrine signaling, promoting angiogenesis, modulating inflammation, and enhancing tissue repair via secretion of growth factors such as VEGF and cytokines. They may also improve microvascular perfusion and endothelial function in ischemic tissues. The literature indicates autologous stem cell therapy benefits no-option patients, outperforming conventional treatment in ulcer healing, pain-free walking distance, rest pain reduction, and hemodynamic measures such as ankle-brachial index and transcutaneous oxygen pressure. In conclusion, stem cell therapy shows potential in improving outcomes for no-option PAD patients. Nevertheless, long-term benefits remain unconfirmed, and evidence quality is low, influenced by cell type, dose, and administration route. Given the high morbidity and mortality in this population, discontinuing this therapy would be premature. Further placebo-controlled, multicenter phase 3 trials with long-term follow-up are needed to establish stem cell therapy as standard treatment for CLI in no-option patients.

Keywords: angiogenesis, critical limb ischemia, limb amputation, peripheral arterial disease, stem cell therapy

THE FUTURE OF AI IN PRECISION MEDICINE: ETHICAL, LEGAL, AND SOCIAL IMPLICATIONS

Presentation number: PP66 Abstract number: 117-ISABS-2026

CRANIAL EPIGENETIC TRAIT FREQUENCIES IN CROATIA: COMPARING MODERN AND ARCHAEOLOGICAL POPULATIONS

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To quantify cranial epigenetic (non-metric) traits in a contemporary Croatian MSCT sample and compare trait frequencies with an archaeological cranial series, overall and by sex. The modern dataset comprised 390 adult MSCT scans from Split (n=196) and Zagreb (n=194) and was balanced by sex (195 males and 195 females). The archaeological dataset comprised 140 adult crania (71 females, 69 males); trait-specific sample size varied due to missing observations. Traits were scored as present/absent (bilateral traits by side). Group differences were tested using χ^2/Fisher's exact tests, with Benjamini–Hochberg FDR adjustment (q-values). In the modern sample, 6/74 traits differed between Split and Zagreb, and 10/74 differed by sex after FDR correction ($q<0.05$), with higher frequencies consistently observed in Split and in males. In the archaeological series, no sex differences remained significant after FDR correction. Modern– archaeological comparisons across overlapping traits identified 35/72 traits with significant differences ($q<0.05$), consistent with secular change in cranial epigenetic trait frequencies. Contemporary MSCT- based profiling provides population-specific epigenetic trait reference data and, relative to archaeological material, indicates limited regional differentiation but pronounced sex-associated patterning in modern samples and substantial secular change across time.

Keywords: MSCT, cranial epigenetic traits, modern Croatian population, medieval period, secular changes

Presentation number: PP67 Abstract number: 48-ISABS-2026

BRIDGING THE GAP: A NATIONAL ROADMAP FOR IMPLEMENTING EHDS STANDARDS IN A NON-EU COUNTRY – THE CASE OF SERBIA

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The development of AI in precision medicine increasingly depends on access to high-quality, interoperable health data, positioning the European Health Data Space (EHDS) as a key regulatory and technological framework. While primarily designed for EU Member States, its implications for non-EU countries remain underexplored. This study aims to develop a national-level roadmap for aligning a non-EU healthcare system with EHDS standards, using Serbia as a case study, and to assess its broader ethical, legal, and social implications for AI deployment. The research applies a qualitative methodology combining normative legal analysis, comparative assessment with selected European models, and policy mapping of Serbia's existing digital health ecosystem, including legislation on medical documentation, national registries, and eHealth platforms. Findings indicate that Serbia has established a foundational legal and technical infrastructure, yet faces challenges related to interoperability, data governance, institutional capacity, and alignment with EU data protection and AI regulations. The proposed roadmap outlines phased alignment across legal harmonization, institutional design, and technical standardization, enabling integration into cross-border data ecosystems such as HealthData@EU. Importantly, the study highlights that such alignment extends beyond technical readiness, raising critical questions around data sovereignty, trust, algorithmic accountability, and equitable access to AI-driven healthcare. The paper concludes that adopting EHDS-compatible standards in non-EU contexts can accelerate innovation and international integration but must be accompanied by robust ethical and governance frameworks to ensure responsible and inclusive use of AI in precision medicine.

Keywords: EHDS, artificial intelligence, health data governance, digital health, Serbia

Presentation number: PP68 Abstract number: 120-ISABS-2026

MAPPING THE CROATIAN OPHTHALMIC DATA LANDSCAPE FOR INHERITED RETINAL DISEASES: A STEP TOWARD A NATIONAL FRAMEWORK

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To evaluate the availability of diagnostic procedures, access to genetic testing, and data management practices in tertiary ophthalmology centers in Croatia, with the goal of identifying needs for establishing a national framework for the care of patients with inherited retinal diseases (IRDs). A cross -sectional survey was conducted among ophthalmologists actively involved in IRD patient care across tertiary centers. Data were collected on the availability of diagnostic procedures, access to genetic testing, and local practices for data collection, storage, and integration of clinical and genetic information. Considerable variability was identified between centers in the availability of diagnostic methods and access to genetic testing. While core imaging modalities are widely available, advanced retinal function testing and genetic diagnostics are not uniformly accessible. Data are predominantly stored within local hospital systems, with limited standardization and minimal integration of clinical and genetic information. This study provides the first national overview of IRD -related diagnostic and data management practices in Croatia. Despite the establishment of a national Reference Center for inherited retinal dystrophies in 2022, its potential to coordinate care and enable centralized data collection remains underutilized. The identified gaps highlight the need for improved coordination, standardization, and data integration. Establishing a structured national framework, including a hub-and- spoke model and centralized data collection, could enhance patient care and support future therapeutic and research initiatives.

Keywords: inherited retinal diseases (IRDs), genetic testing, diagnostic techniques, data management, health care organization

Presentation number: PP69 Abstract number: 98-ISABS-2026

EVALUATION OF TRUST AND PERSPECTIVES ON ARTIFICIAL INTELLIGENCE INTEGRATION AMONG MEDICAL AND PHARMACEUTICAL PROFESSIONALS: A SURVEY STUDY

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This study evaluated the level of trust, risk perception, and expectations of the medical and pharmaceutical community regarding the implementation of artificial intelligence (AI) tools in professional practice. A cross-sectional survey was conducted among 71 healthcare professionals, including 36 pharmacists (50.7%) and 35 physicians (49.3%). The questionnaire addressed AI tool usage frequency, trust in generated recommendations measured on a 5-point Likert scale, and assessment of privacy risks. Statistical analysis involved calculating mean values and frequency distributions. The results demonstrated an average level of professional trust in AI recommendations of 3.14 points. The highest level of concern related to patient confidentiality and data security risks, which were rated at 3.24 points. While the hypothesis of complete human replacement by AI technology was decisively rejected (1.8 points), forecasts for deep AI integration into professional workflows over the next decade remained very high (3.89 points). A significant "educational vacuum" was identified, with 67.6% of respondents acquiring AI skills through self-study, whereas university curricula lacked such knowledge for all participants. In conclusion, specialists view AI as an intelligent digital assistant rather than a replacement for human expertise. Despite a high readiness for professional transformation, barriers such as data security risks and the absence of systemic education must be addressed through secure local AI solutions and modernized medical curricula.

Keywords: artificial intelligence, digital medicine, healthcare, professional trust, medical education

Presentation number: PP70 Abstract number: 116-ISABS-2026

DEVELOPING REFERENCE DATA BASES FOR MEGALOPOLISES BY DNA-MARKERS TAKING INTO ACCOUNT DYNAMICS OF GENE POOL UNDER ACTION OF MIGRATION

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Megalopolises represent specific historically new type of population structure for which adequate models and methods of study have not yet been developed. Megalopolises are characterized by ethnically mixed population (1 million or over) with territorial subdivision, high level of migration and reproduction of population due to migration preferentially. Migration is the main factor of gene pool dynamics of megalopolises. The goal of the study was to characterize the gene pool of three megalopolises by, DNA markers of Y-chromosome and mtDNA, by autosomal STR and to estimate influence of migration on their dynamics using standard statistical approaches, Material and methods. In the samples from Saint- Petersburg, senior generation of Novosibirsk, senior and young generations from Moscow, the genetic differentiation by 18 STR and haplogroups of Y-chromosome and haplogroups (detected by 18 STR haplotypes by Internet-predictors) of mtDNA detected by Internet-predictor basing on nucleotide sequences of HVSI and HVSII (Moscow samples by 27 autosomal STR, additionally) was studied. Migration parameters were calculated by analysis of questionnaire data. Results. Differentiation by DNA markers of Y-chromosome demonstrates a more prominent influence of migration processes compared to other markers. Accumulation of "southern origin" haplogroups of Y-chromosome in the gene pool of Moscow was demonstrated under action of modern migration flows. Autosomal STR frequencies demonstrate the less influence of migration in generations. Territorial ethnic subdivision of the megalopolises population was demonstrated by 2010 Census data. Conclusion. Observed statistically significant differences between samples by DNA-markers mostly mediated by modern migration (especially from North Caucasus. Transcaucasia and Middle Asia) evidence of necessity of developing separate reference data bases for each megalopolis (considering territorial subdivision) including molecular and genetic demographic data.

Keywords: megalopolis, population, migration, reference database

MOSES SCHANFIELD MEMORIAL SYMPOSIUM – POSTER PRESENTATIONS

Presentation number: PP71 Abstract number: 40-ISABS-2026

AN EXPLORATORY AI APPROACH COMBINING INTERPRETABLE MACHINE LEARNING (ML) AND SELF-SUPERVISED DEEP LEARNING FOR DIFFERENTIATING NAIL SCRATCH PATTERNS ON PORCINE SKIN

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The study explored the identifiability of nail-shape-related signals using interpretable ML and AI-based image analysis. The study included 8 participants: 4 with rounded and 4 with square gel nails. Each participant performed 5 controlled scratch strokes on a different porcine skin piece. The length and maximum width of each of the three individual traces per stroke were measured using a digital caliper, and the mean width/ length ratio was calculated. For each trace, edge morphology and line continuity were assessed and photographed. A regularized logistic regression (LR) model with class balancing was trained and evaluated using leave-one-participant-out cross-validation (LOPO-CV) to account for intra-participant dependency. To independently assess whether shape-related information was present in the photographs, a self-supervised vision transformer (DINO ViT-S/8) was used with CLAHE contrast enhancement and attention-weighted patch feature extraction and evaluated under identical LOPO-CV conditions. The morphological LR achieved an accuracy of 0.76 (AUC 0.83), with a sensitivity of 0.84 for rounded nails and a specificity of 0.68 for square nails. Scratch width was the most influential predictor: wider traces were associated with square nails, whereas smooth edges and continuous lines were associated with rounded nails. The DINO classifier achieved an accuracy of 0.74 (AUC 0.83, sensitivity 0.68, specifically 0.79). The results suggest that scratch morphology encodes nail shape information detectable by both feature-engineered and pixel-level methods. The differences likely reflect contact mechanics: square nails (flatter edges, sharper lateral transitions) distribute force more broadly, producing wider and irregular traces, while rounded nails concentrate force, resulting in narrower, smoother, and more continuous marks. The comparable AUCs across methods support signal consistency, though the findings are exploratory and require further validation.

Keywords: scratch marks, nail morphology, porcine skin model, interpretable machine learning, self-supervised deep learning

Presentation number: PP72 Abstract number: 121-ISABS-2026

WHEN ALGORITHMS TESTIFY: AIDRIVEN DNA ANALYSIS, EVIDENTIARY STANDARDS, AND CRIMINAL JUSTICE REFORM

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Forensic DNA analysis has already influenced criminal justice, and serves as a powerful tool and double-edged sword for both conviction and exoneration. Despite its scientific foundations and wide application, DNA evidence is vulnerable to factors as interpretive error, methodological limitations, and cognitive bias, as demonstrated by numerous wrongful convictions identified through the Innocence Project. Recent advances in artificial intelligence (AI) promise to improve the interpretation of complex DNA samples, including mixed, low template, and degraded profiles. At the same time, the growing use of AI driven forensic analysis raises significant legal, ethical concerns, as well as procedural challenges in terms of whether it can be used as a direct evidence in the procedure. This article examines the implications of AI based DNA interpretation for criminal justice, with particular attention to evidentiary reliability, due process, institutional accountability, and emerging policy responses in the U.S. and Europe. Further it draws on parallels with clinical genomics and documented forensic applications of AI, where the authors argue that AI can enhance forensic accuracy and fairness only if integrated within transparent, validated, and ethically governed frameworks that respect fundamental legal protections.

Keywords: probabilistic genotyping, artificial intelligence, innocence, fair trial rights

Presentation number: PP73 Abstract number: 89-ISABS-2026

HELIXCROSS – AN AI-POWERED CROSS-EXAMINATION SIMULATION FOR FORENSIC EXPERT TESTIMONY TRAINING

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Forensic expert testimony faces intensified scrutiny as methods have grown more complex, probabilistic reasoning has become central to evidence interpretation, and admissibility standards have strengthened. Testimony preparation, however, remains largely informal – conducted in-house, lacking standardization, and rarely providing the adversarial practice needed to develop reliable courtroom competence. HelixCross is a prompt-engineered large language model (LLM) agent designed to address this gap. It simulates defense counsel cross-examination and provides structured, on-demand coaching for forensic practitioners in a controlled, repeatable environment at no cost. The flagship module targets forensic DNA interpretation, focusing on areas that routinely generate courtroom challenges: probabilistic genotyping, likelihood ratio communication, mixture interpretation assumptions, number-of-contributors sensitivity, and admissibility framing under legally permissible standards. The curriculum is discipline-agnostic and intended for extension to other forensic domains. HelixCross uses a Retrieval-Augmented Generation (RAG) architecture, anchoring questioning and coaching to a curated knowledge base of forensic science standards and legal frameworks rather than relying on general training data alone. The curriculum spans nine progressive phases, from voir dire and foundations through to reliability and disclosure challenges at peak difficulty. The simulation alternates between two modes: In-court mode simulates adversarial questioning, while Coach mode delivers phase-by-phase debriefs with exploitable-statement analysis, court-safe rewrites, and a grading rubric across ten performance dimensions. The tool is freely deployed on OpenAI ChatGPT and Google Gemini. Future work will evaluate educational effectiveness and scoring reliability through a controlled empirical study.

Keywords: expert testimony, simulation-based education, large language model, probabilistic genotyping, artificial intelligence

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DEEP LEARNING-BASED AUTOMATED SKELETAL SEGMENTATION FOR FORENSIC RECONSTRUCTION: A CRITICAL EVALUATION OF AI RELIABILITY AND HUMAN-IN-THE-LOOP NECESSITY

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Automated AI segmentation offers a promising approach for building virtual skeletal reference datasets where osteological collections are absent, yet systematic assessment of its forensic limitations remains sparse. This study evaluated the reliability and failure modes of Total-Segmentator, a MONAI-based deep learning plugin for 3D Slicer, applied to lower-extremity segmentation of clinical MSCT data. Ninety-one MSCT angiography scans (UH Merkur Zagreb; UHC Split) were processed on a Ryzen 9 8945HX (8 GB GDDR6 VRAM), with mean runtimes of 108.63 s (femur, cropped protocol) and 75.62 s (tibia/patella). The femur was the primary target given its preservation frequency, pronounced sexual dimorphism, and utility for stature estimation. Automated segmentation generated STL surface meshes reviewed for anatomical validity. Of 91 cases, 79 (86.8%) produced valid models; of 16 failures, 8 were due to incomplete acquisition, 1 to non-skeletal protocol scope, 3 to unexplained cortical artifacts, and 4 to voxel-level metal- bone misclassification in endoprosthetic cases (25% of such cases; 4.4% of total), producing STL models fusing bone and implant geometry. The key finding is not proportion but the invisibility: endoprosthetic hardware was not consistently detectable at pre-selection, so corrupted models entered the pipeline undetected, producing systematically erroneous bone anatomy. No DICOM metadata differences (acquisition parameters, reconstruction kernel, or protocol descriptors) were identified between cases with and without endoprosthetic hardware, confirming implant-related errors cannot be anticipated or filtered by metadata alone. These findings confirm that, despite its efficiency for virtual collection building, deep learning-based segmentation requires structured human-in-the-loop validation. Implant-related misclassification represents a non-obvious but forensically consequential risk to data integrity that pre- screening alone cannot eliminate.

Keywords: MSCT, automatic segmentation, deep learning, skeletal collections, femur

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ANALYSIS OF POST-MORTEM BLOOD MICROBIOME: HIGH-RESOLUTION IDENTIFICATION OF PSEUDOMONAS TOLAASII USING PACBIO 16S FULL-LENGTH SEQUENCING

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Accurate identification of the thanatomicrobiome is crucial for estimating the post-mortem interval (PMI) and understanding decomposition processes. Traditional culture-based methods, such as MicroSeq ID, often fail to detect non-culturable or fastidious microorganisms, limiting the comprehensive profiling of the post-mortem microbial community. In this study, we analyzed DNA extracted from post-mortem blood samples (n=20) using third-generation sequencing (TGS). We employed the PacBio Sequel system for 16S rRNA full-length sequencing to achieve species -level resolution. The results were compared with previous data obtained via culture-based MicroSeq ID analysis to evaluate the diagnostic improvement. Peripheral blood samples were collected from 20 cadavers during autopsies conducted within a post-mortem interval (PMI) of 1 to 3 days. To ensure analytical consistency, the extracted DNA underwent PacBio 16S full-length sequencing across two independent runs. Among the 20 samples analyzed, *Pseudomonas tolaasii* was identified in 5 cases, while it remained undetected. The consistent detection of this species in a significant majority of samples collected within a narrow, early PMI window suggests that *P. tolaasii* may be a characteristic component of the early-stage thanatomicrobiome in peripheral blood, a finding that was previously unattainable through conventional culture-based MicroSeq ID systems. *Pseudomonas tolaasii* was identified in 5 out of 13 samples (38.5%) through PacBio full-length sequencing. Notably, this species had never been detected in the same samples using the MicroSeq ID system. The high-fidelity long-read sequencing provided sufficient phylogenetic markers to distinguish *P. tolaasii* from other closely related Pseudomonas species, which is often challenging with short-read NGS or phenotypic assays. The detection of *P. tolaasii* highlights the superior sensitivity and resolution of third-generation sequencing in forensic investigations.

Keywords: thanatomicrobiome, *pseudomonas tolaasii*, forensic microbiology, post-mortem interval (PMI)

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INTERPRETATION OF TWO INDEPENDENT AUTOSOMAL STR ANOMALIES IN A COURT-ORDERED PATERNITY TESTING CASE

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This case study describes a court-ordered paternity investigation involving two children, in which an exceptionally rare discordance was observed in one confirmed father-child pair. The anomaly consisted of two independent autosomal STR inconsistencies, highlighting challenges in interpreting atypical inheritance patterns in paternity testing. DNA from buccal swabs was analyzed using a 24 STR loci system. To exclude technical artifacts (e.g., allele dropout, amplification failure, or sample mix-up), additional testing with alternative STR kits and sequence-based analysis was performed, confirming the initial findings. For the first child, full concordance was observed among the child, mother, and alleged father across all loci. The second child matched the mother but showed discordance with the alleged father at two loci, sharing 21 of 23 obligate alleles. Despite this, the probability of paternity was 99.85%. Comparison between siblings showed concordance at 19 of 23 loci, supporting a full sibling relationship (99.84%). At the vWA locus (chromosome 12), the second child was homozygous for the maternal allele, lacking the paternal allele. A similar pattern was observed at D7S820 (chromosome 7), without broader chromosomal abnormalities. These findings suggest rare biological mechanisms beyond simple STR mutations, including multistep germline mutation, gene conversion, or localized loss of heterozygosity. Sequence analysis of vWA showed the child's allele matched the maternal microvariant, supporting a locus-specific anomaly. Analysis of D7S820 confirmed previously identified alleles, consistent with mutation or gene conversion. This rare case of dual STR anomalies in a confirmed father-child pair highlights the complexity of STR interpretation and the importance of confirmatory and sequence-based analyses in resolving atypical results.

Keywords: paternity testing, autosomal STR, sequence-based analysis, inheritance anomalies

Presentation number: PP77 Abstract number: 93-ISABS-2026

PRELIMINARY VALIDATION OF THE A27PLEX STR DETECTION KIT AND CONCORDANCE STUDY OF THE NEW GENERATION MULTIPLEX STR KITS

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Short tandem repeats (STRs), also referred to as microsatellites, are polymorphic genetic markers consisting of short repetitive sequences, typically 2-7 base pairs in length, that are consecutively repeated a variable number of times at a given locus. The number of repeats varies greatly among individuals, making STRs ideal markers for DNA profiling and a powerful tool for forensic identification, as well as for paternity and relatedness testing. With the expansion of STR marker use in forensic identification, commercial multiplex STR kits have become widely available. These kits are designed to analyze multiple autosomal STR loci in a single PCR reaction. The purpose of this study was to validate a novel A27Plex Fluorescence Detection Kit, produced by Nanjing Superyears Gene Technology Co., Ltd., and to conduct a concordance study comparing this commercial kit with those currently used in forensic analyses in Croatia. The A27Plex Fluorescence Detection Kit is a 6-dye STR multiplex assay that amplifies 25 autosomal STR loci, one Y-chromosome STR locus, and an insertion-deletion (indel) sex-determining marker. In this preliminary study, standard samples with previously determined DNA profiles were analyzed. Detected loci, intra-locus balance, interlocus balance, stutter ratios, and concordance with other kits used in Croatia were evaluated. Based on the obtained results, it can be concluded that this kit is suitable for forensic identification in the Croatian population and produces results comparable to those of other kits routinely used in this region. This study represents the first validation of the A27Plex Fluorescence Detection Kit in Croatia.

Keywords: short tandem repeats (STRs), DNA profiling, forensic identification, multiplex PCR, concordance

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PRELIMINARY VALIDATION OF THE Y41SE STR DETECTION KIT AND CONCORDANCE STUDY OF THE NEW GENERATION MULTIPLEX STR KITS

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Y-chromosome short tandem repeats (Y-STRs) are genetic markers consisting of short repetitive sequences, typically 1 to 6 base pairs in length, distributed throughout the Y chromosome and inherited as a haplotype along the paternal line. They are primarily used to trace male lineage in forensics, anthropology, and genealogy. Commercial multiplex Y-STR kits are designed to analyze multiple Y- chromosome STR loci in a single PCR reaction. The aim of this study was to validate the novel Y41se STR Fluorescence Detection Kit, produced by Nanjing Superyears Gene Technology Co., Ltd., and to perform a concordance study comparing this kit with those currently used in forensic analyses in Croatia. The Y41se STR Fluorescence Detection Kit is a 6-dye multiplex assay that amplifies 40 Y-chromosome STR loci (33 low-mutation-rate and 7 rapidly mutating loci) and one Y-chromosome insertion–deletion (INDEL) marker. In this preliminary study, selected samples with previously determined Y-STR profiles were analyzed. Detected loci, stutter ratios, and concordance with other kits used in Croatia were evaluated. Based on the obtained results, it can be concluded that this kit is suitable for use in the Croatian population and produces results comparable to those of other kits routinely used in this region. This study represents the first validation of the Y41se STR Fluorescence Detection Kit in Croatia.

Keywords: Y-chromosome STRs (Y-STRs), forensic genetics, male lineage, multiplex PCR, concordance

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IN HOUSE OPTIMISED METHOD OF NON-DESTRUCTIVE AUTOMATED DNA EXTRACTION FROM FORENSIC TOOTH SAMPLES

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At the Forensic Science Centre "Ivan Vucetic," routine methods for DNA extraction from dental/skeletal remains often involve time-consuming procedures and the destructive grinding of samples into powder. However, recent forensic guidelines for the identification of human remains prioritize less invasive methods. To address this shift, previously published protocols for DNA extraction from ancient/archaeological intact tooth samples were reviewed and optimized, to establish a rapid, effective, and non-destructive method for obtaining short tandem repeat (STR) profiles. Tooth samples from five cases of unidentified human remains received in 2025 were processed using the optimized protocol: chemical cleaning, washing, drying, UV irradiation, followed by demineralization and lysis. The last two steps of the described protocol were achieved by immersing the intact teeth in 0.5 M EDTA at pH 8, supplemented with proteinase K, under continuous mixing for 24 hours. The resulting supernatants were centrifuged at 5000 rpm using Amicon Ultra 15 filter units until a lysate volume of approximately 300 µl was reached. Automated purification was performed on a BioRobot EZ1 using a large-volume protocol, yielding 50 µl of eluate. DNA quality and quantity were evaluated via Real-Time PCR, followed by STR typing using the GlobalFiler™ PCR Amplification Kit resulting in complete DNA profiles for all samples. Subsequently, to validate the efficacy of this non-destructive approach, the same samples were grounded into powder and processed according to the laboratory's validated protocol, which produced consistent results. Comparison with reference DNA profiles obtained from biological relatives and personal belongings further confirmed the accuracy of the findings. Overall, this study demonstrates the applicability of the established automated, non-destructive extraction method for the forensic identification of human remains, ensuring the preservation of evidence, resulting in a usable STR profile.

Keywords: forensic identification, intact tooth samples, automated DNA extraction, STRs analysis

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ANALYSIS OF DEGRADED FORENSIC SAMPLES USING MASSIVELY PARALLEL SEQUENCING: ADVANTAGES AND CHALLENGES

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Forensic casework samples are often challenging. PCR- and capillary electrophoresis (CE)-based analysis methods may result in partial DNA profile or no profile at all when analyzing inhibited and/or degraded samples. Massively parallel sequencing (MPS) offers analysis of many different types of loci, short tandem repeats (STRs) and single nucleotide polymorphisms (SNPs), in a single reaction, thus conserving the sample while extracting the maximum amount of information. In this study, degraded samples from commercial test kit (containing 500, 300 and 150 bp fragments; Qiagen) were amplified in duplicate using Primer Mix B from ForenSeq™ DNA Signature Prep (Illumina®/Verogen). One replicate of samples underwent library preparation according to manufacturer's instructions, while preparation of the other replicate was modified thusly: 30 µL of purified libraries (instead of 20 µL) were added into normalization, with proportionately increased magnetic beads volume. The total of 18 casework samples (tissue, bone, teeth, blood) were also prepared for sequencing using unmodified protocol. Libraries were sequenced on the MiSeq FGx® instrument (Illumina/Verogen). ForenSeq® Universal Analysis Software (Verogen) and Microsoft Excel were used for data analysis. Detection of STR loci (particularly long allele variants) was considerably reduced in 150bp-samples, which was expected. Impact of degradation on detection of SNPs was not significant, due to their short loci lengths. Most prominent differences between 20µL- and 30µL-libraries were observed in numbers of imbalanced heterozygous loci and detected alleles above analytical threshold, with small gain in favor of 30µL-libraries. For casework samples, only 4/18 resulted in poor MPS profiles, while others benefited from larger number of detected markers and additional information of ancestry and phenotype, clearly underlining the advantages of MPS in comparison to CE-based STR genotyping.

Keywords: degraded forensic samples, MPS preparation modification

Presentation number: PP81 Abstract number: 33-ISABS-2026

EVALUATION OF SIMULATED FORENSIC MIXED SAMPLES: COMPARISON AND ADVANTAGES OF MASSIVELY PARALLEL SEQUENCING TECHNOLOGY OVER CAPILLARY ELECTROPHORESIS

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Forensic samples from multiple donors present significant challenges in evaluation and interpretation. Massively parallel sequencing (MPS) enables simultaneous analysis of many genetic markers, including short tandem repeats (STRs) and single nucleotide polymorphisms (SNPs), while also providing detailed sequence information for each allele. The ability to distinguish alleles with identical STR repeat numbers but different sequences enhances the resolution and interpretation of mixed DNA profiles. In this study, we aimed to evaluate the analytical performance of an MPS kit in comparison to capillary electrophoresis (CE) results for simulated forensic mixtures. Samples were prepared by mixing the male positive control 2800M and the female positive control 9947A at ratios of 1:1, 1:2, 1:5, 1:10, and 1:20. Each mixture was amplified in triplicate using Primer Mix B from the ForenSeq® DNA Signature Prep kit (Verogen), targeting 231 loci. Libraries were subsequently prepared, pooled, and sequenced on the MiSeq FGx® platform (Illumina/Verogen) according to the manufacturer's instructions. Data analysis was performed using the ForenSeq® Universal Analysis Software (Verogen) and Microsoft Excel. Mixture ratios calculated for STR and identity SNP loci without shared alleles generally showed lower female-to-male average values than expected theoretical values and CE results. Identity SNPs were consistently more reliable than other loci. Significant dropout of male alleles was observed across all loci at ratios of 1:10 and 1:20. For certain loci (e.g., D1S1656, vWA, DXS10103, and four Y-STRs), allele dropout occurred even at a 1:1 ratio, with D22S1045 being particularly problematic, showing markedly deviating average ratios. In conclusion, MPS offers advantages over CE in mixture analysis; however, results should still be interpreted cautiously, as sequencing artefacts may overlap with minor contributor ratios.

Keywords: mixed samples, evaluation, massively parallel sequencing, ForenSeq, MiSeq FGx

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DNA-BASED PREDICTION OF EYE, HAIR, AND SKIN COLOR IN MEDIEVAL INDIVIDUALS FROM BOSNIA AND HERZEGOVINA

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Human skeletal remains provide valuable biological information, but many aspects of an individual's physical traits remain unknown without genetic analysis. Recent developments in DNA phenotyping now allow researchers to reconstruct key pigmentation features from ancient remains. In that regard, we analyzed DNA from ten skeletal remains recovered from three medieval necropolises in Bosnia and Herzegovina, alongside five contemporary individuals used as controls. A set of 40 SNP markers linked to pigmentation was examined using the HIrisPlex-S system, and the results were expressed as probabilities of specific traits. The predictions for the modern samples matched their known physical characteristics, supporting the reliability of the method when applied to ancient remains. Among the medieval individuals, blue eyes were predicted in four, and brown eyes in six. Hair color was mostly brown, with some individuals showing darker or lighter shades. Most were also predicted to have intermediate skin pigmentation, while a smaller number likely had lighter skin. Overall, this study offers the first genetic-based insight into the pigmentation traits of medieval individuals from Bosnia and Herzegovina, improving our understanding of their biological characteristics.

Keywords: DNA phenotyping, medieval Bosnia, ancient DNA, pigmentation traits

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MOLECULAR APPROACH TO AGE ESTIMATION: EXPLORING THE POTENTIAL OF TELOMERE LENGTH IN FORENSICS

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With the growing application of age estimation in modern forensics, the potential of telomere length (TL) as a biomarker of molecular age should not be overlooked, given the continuous shortening of telomeres throughout life. This study aimed to establish and refine methodological approaches for TL assessment in human samples, evaluate its applicability in a forensic context, and investigate its correlation with chronological age, as well as the potential influence of sex on this relationship. A total of fifty unrelated, healthy adult non-smokers (aged 22-73 years) provided buccal epithelial cells and peripheral venous blood samples, along with relevant personal, lifestyle, and health information. Genomic DNA was subjected to absolute TL estimation using quantitative Real-Time PCR. Absolute telomere length per diploid genome was determined using standard curves, with the 36B4 gene serving as the reference marker. In addition, Fluorescence In Situ Hybridization (FISH) was employed to visualize telomere structure, length, and potential chromosomal aberrations in metaphase spreads from selected samples representing the highest and lowest TL values. Statistical analysis revealed significant differences in telomere length between lymphocytes and buccal epithelial cells, while no significant associations were observed with age or sex, regardless of the tissue type used for DNA extraction. FISH analysis demonstrated stronger fluorescent signals in samples with previously determined longer telomeres, supporting the reliability of qPCR-based estimation. Although telomere length alone may have limited reliability as a predictor of age, it may still serve as a complementary biomarker alongside epigenetic and other molecular markers. Future studies with larger and more diverse cohorts, as well as integrated multi-marker approaches, are needed to improve the accuracy and practical applicability of molecular age prediction in forensic settings.

Keywords: telomeres, absolute telomere length, molecular age, Real-Time qPCR, FISH

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AN ANALYSIS OF THE EFFICIENCY AND RELIABILITY OF PHOTOGRAPH-BASED LINEUP RECOGNITION WITH AI-GENERATED IMAGES

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This study examined the reliability of photograph-based lineups composed of AI-generated fillers, as well as the relationship between recognition accuracy and characteristics of the event, the witness, and the identification procedure. The study included 161 participants who were shown four video recordings of simulated criminal acts, two violent and two non-violent. After viewing each video, participants were asked to identify the perpetrator from a five-person photographic lineup consisting of one real perpetrator and four AI-generated fillers, while control questions were used to introduce a short delay between observation and identification. Overall identification accuracy across all scenarios was 25%, with participants correctly identifying the perpetrator in an average of one video. Accuracy was significantly higher in scenarios involving violent events (39.13%) compared to non-violent events (12.42%). The highest accuracy was observed in simulated robbery (44.10%) and aggravated theft (34.16%), whereas identification in bicycle theft (16.77%) and drug delivery (8.07%) remained at or near chance level. Multivariate analysis showed that identification accuracy was influenced by the presence of violent elements, the order of video presentation, the position of the perpetrator in the lineup, participant age, and prior police-related experience. The results indicate that AI-generated images can be effectively used to construct fair lineup fillers, as participants did not treat them differently from real faces, supporting their application in forensic practice. However, overall identification accuracy remains limited and is influenced by both procedural and psychological factors, which should be considered in future applications.

Keywords: eyewitness identification, lineup recognition, photo lineup, ai-generated images, recognition accuracy

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KIT-DEPENDENT ALLELIC DISCORDANCE AT THE D1S1656 STR LOCUS: A FORENSIC CASE REPORT

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In forensic science, human identification relies on DNA profiles. Short Tandem Repeats (STRs) are repetitive DNA sequences that vary in length among individuals, and are widely used for identification purposes e.g. in criminal investigations and paternity testing. Misinterpretation of alleles at any locus may lead to incorrect conclusions and have significant downstream consequences. Here we report a case of discordant results at D1S1656 STR locus obtained with different PCR kits. Genomic DNA was extracted from a buccal swab sample using two methods: EZ1 Investigator Kit (Qiagen) and Chelex® 100. Real-time PCR with Quantifiler® Human DNA Quantification Trio Kit (Applied Biosystems) and Investigator Quantiplex Pro RGQ Kit (Qiagen) were used for quantification. STR amplification was performed using GlobalFiler™ PCR Amplification Kit (Thermo Fisher Scientific), PowerPlex® Fusion 6C System (Promega), and Investigator 24plex Kit (Qiagen), with subsequent fragment separation by capillary electrophoresis on 3500 Genetic Analyzer (Applied Biosystems). Data analysis was performed in GeneMapper® ID-X v1.7. The resulting DNA profile from Investigator 24plex kit exhibited a tri-allelic pattern at D1S1656 and a single peak at D12S391. In contrast, bi-allelic peaks at D1S1656 and D12S391 were obtained with GlobalFiler™ and PowerPlex® Fusion 6C kits. The remaining profile was concordant across kits, no contamination was detected in negative controls, and positive controls were as expected. Re-extraction from the same sample yielded consistent results. These findings highlight the advantage of using multiple STR amplification kits: in cases of extremely short or extremely long allele variants appearing at loci that are closely located in one kit, which may cause misinterpretation, it is greatly beneficial to use another kit with different loci arrangement. Any atypical patterns need to be carefully evaluated to prevent erroneous interpretation of forensic DNA evidence.

Keywords: short tandem repeats, human identification, misinterpretation, allelic discordance, tri-allelic peak

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TRUSTWORTHY AI IN FORENSIC GENETICS STARTS WITH CONTAMINATION GOVERNANCE: A EUROPEAN EDB PERSPECTIVE

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Artificial intelligence (AI) applications in forensic and anthropological genetics increasingly depend on the quality, traceability, and governance of upstream laboratory and casework data. Yet discussions on AI readiness often underemphasize a foundational issue: contamination control. This presentation argues that elimination DNA databases (EDBs) should be understood not only as quality assurance tools, but as core components of trustworthy AI-enabling infrastructure in forensic genetics. Building on a narrative review with scoping elements and a policy-focused synthesis of empirical contamination studies, accreditation practice, and European legal frameworks, we identify harmonized minimum requirements for EDB operation. The analysis integrates forensic contamination evidence, ISO/IEC 17025-aligned quality management principles, and European data protection constraints (including strict purpose limitation and safeguards against function creep). Particular attention is given to governance separation from criminal/intelligence databases, auditable workflows, role-based inclusion and retention, and monitoring metrics that improve comparability across jurisdictions. The central claim is that robust EDB governance improves not only contamination detection and incident handling, but also the reliability of downstream data ecosystems on which AI-supported forensic decision systems may rely. In the context of expanding cross-border forensic data exchange in Europe, harmonized EDB standards can therefore be framed as both a quality assurance priority and a precondition for ethically and legally trustworthy AI deployment in forensic genetics.

Keywords: forensic genetics, elimination DNA database (EDB), DNA contamination control, trustworthy AI

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FORENSIC INVESTIGATION OF INTENTIONAL ANIMAL POISONING: AN INTEGRATED APPROACH IN CRIME PREDICTION AND RESOLUTION

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Animal poisoning, particularly in urban environments, frequently presents a significant challenge for investigative authorities due to the difficulty of linking the toxic agent with the perpetrator's intent. This paper presents a forensic investigation of a case involving the serial poisoning of cats, where a combination of forensic necropsy, toxicology, and molecular identification played a pivotal role. Following several suspicious deaths of cats, a forensic examination of one carcass was performed. The necropsy raised reasonable suspicion of carbofuran poisoning (a carbamate pesticide) based on external findings of vomit around the mouth, the presence of bluish granules with a distinctive odour in the digestive tract, and systemic findings such as generalised extensive congestion and haemorrhage within parenchymal organs. Toxicological analysis using gas chromatography-mass spectrometry confirmed carbofuran in the gastric contents and its derivatives in the liver. To establish the biological origin of the partially digested bait, DNA barcoding was performed targeting four mitochondrial markers (cyt b, COI, 12S, and 16S rRNA). This molecular evidence created a critical link between the preparation of the poison and the criminal act. Proving the method of poisoning and identifying the biological origin of the bait are essential steps in establishing the elements of the criminal offense "Killing or Torturing Animals." The synergy of these techniques, further enhanced by artificial intelligence for predictive mapping and pattern recognition, not only elucidates the cause of death but provides irrefutable evidence of a deliberate act, thereby facilitating the identification of perpetrators and prevention of future incidents.

Keywords: animal poisoning, forensic necropsy, forensic toxicology, DNA barcoding, crime prediction

Presentation number: PP88 Abstract number: 76-ISABS-2026

AI ASSISTED CLASSIFICATION OF HUMAN, DOG AND CAT HAIR USING MICROSCOPY IMAGES: A PILOT STUDY

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The aim of this study was to evaluate the performance of a simple, freely available AI-based image classification tool for differentiating human, dog, and cat hair. Hair samples were mounted in glycerol and analysed under 400× magnification using a DigiCyte NE930 microscope. Four images were captured from different regions of each hair shaft using focus stacking. Medulla type was recorded, and the medullary index (MI) was calculated as the ratio of medulla width to total hair width. A total of 120 images (10 hair samples per category, 4 images per sample) were used to train the model using the Teachable Machine platform. Model performance was evaluated on an independent test set of 60 images (15 hair samples; 5 per category). The model achieved an overall accuracy of 85%. Human hair was correctly classified in all cases (20/20), while cat hair was correctly classified in 19 out of 20 cases. Dog hair showed lower classification accuracy (12/20), with all misclassified dog samples assigned to the cat category. Correct classifications were associated with higher confidence values (mean 0.97), whereas misclassified samples showed lower confidence (mean 0.78). Analysis of misclassified samples indicated that darker hair colour and borderline medullary index values may contribute to classification errors. Human hair predominantly exhibited absent or fragmented medulla and lower MI values (mean 0.24), whereas animal hair showed continuous and wider medulla, with higher values in dog (0.41) and cat hair (0.69). Dog hair exhibited intermediate medullary index values, which may explain lower classification accuracy and increased variability in model confidence. These results demonstrate that simple AI models can effectively differentiate human from animal hair while classification between animal species remains challenging, likely due to overlapping morphological features. Future studies should include larger datasets and additional species to improve model robustness.

Keywords: forensic hair analysis, microscopy, artificial intelligence, machine learning, image classification

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INTERNAL VALIDATION AND COMPARATIVE ANALYSIS OF OSIRIS AND GENOPROOF SOFTWARE FOR STR PROFILE INTERPRETATION

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Accurate interpretation of STR profiles is essential for reliable forensic DNA analysis. The evaluation of a broad spectrum of forensic samples can be complicated by the presence of artifacts such as stutter peaks, pull-up peaks, dye blobs, off-ladder alleles, and others. This study presents an independent validation and comparative assessment of the open-source software OSIRIS v2.4 and the commercial platform GenoProof Suite, using GeneMapper® v5 as the reference standard. A total of 113 human DNA profiles (comprising approximately 3400 alleles), including both single-source and mixed samples, were analyzed using raw .fsa data generated on a SeqStudio™ Genetic Analyzer (Applied Biosystems) with the NGM Select™ kit. The dataset consisted of 68 concordant profiles and 45 problematic cases ("flags") involving artifacts, mixed samples, or degraded DNA. All profiles were independently interpreted in the three software platforms. Results showed over 99% concordance in allele calling, with no significant differences in allele designation between the tools. However, OSIRIS consistently reported higher RFU values across all loci compared to GeneMapper and GenoProof. This behavior is attributed to OSIRIS's use of adaptive baseline modeling and mathematical curve fitting, which normalize raw data to a zero baseline and apply iterative fitting of idealized parametric curves to the signal. In contrast, GeneMapper and GenoProof utilize fixed-window baseline estimation, which may result in lower peak intensities or uncalled alleles. These findings suggest that OSIRIS is a highly effective and cost-efficient alternative for allele designation and artifact identification, potentially reducing interpretation errors in forensic casework. However, as all profiles were manually reviewed, this study does not validate any automated expert system functionality.

Keywords: GeneMapper, GenoProof Suite, OSIRIS, STR profiling, artifact interpretation

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COMPARISON OF CONVENTIONAL AUTOSOMAL STR ANALYSIS AND NEXT-GENERATION SEQUENCING (NGS) IN DEGRADED BONE SAMPLES FROM MASS GRAVE VICTIMS AND BLOOD SAMPLES FROM PHENOTYPICALLY KNOWN INDIVIDUALS

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Identification of degraded human remains represents one of the greatest challenges in forensic genetics, particularly in samples obtained from mass graves and other aging environments, where degradation and inhibitors compromise conventional STR genotyping methods. Previous studies have shown that numerous PCR inhibitors, including humic acids, heavy metals (iron, copper, cadmium, and lead), and microbial contamination, may interfere with DNA analysis and reduce amplification efficiency. The aim of this study was to compare the results obtained by capillary electrophoresis and next-generation sequencing (NGS) in degraded bone samples from mass grave victims and blood samples of persons with known phenotypes. The previous study included 30 bone samples from war crime mass graves analyzed in 2002, 2003, and 2007. In the initial analysis, DNA was extracted using a phenol-chloroform-based protocol, followed by autosomal STR genotyping using the PowerPlex® 16 System on an ABI Prism 310 capillary electrophoresis platform. In the present study, the same samples were reprocessed, and DNA was re-isolated using the PrepFiler BTA Forensic DNA Extraction Kit (Thermo Fisher Scientific) following the manufacturer's protocol. Subsequent genotyping was conducted using next-generation sequencing (NGS) after library preparation with the MiSeq FGx System Identification Library Prep Kit, targeting forensic identification markers. Blood samples of persons with known phenotypes were also analyzed using the same methodology (NGS).

Keywords: forensic genetics, next-generation sequencing (NGS), degraded bone samples, mass grave identification, STR genotyping

ABSTRACTS OF THE 10TH CROATIAN HUMAN GENETICS CONFERENCE & 3RD CROATIAN PERSONALIZED AND PRECISION MEDICINE CONFERENCE

ORAL PRESENTATIONS

Presentation number: CSHG/CSPPM OP 01 Abstract number: 104-ISABS-2026

CROATIA'S PIONEERING ROLE FIRST SYSTEMATIC DNA-BASED IDENTIFICATION OF MASS GRAVE VICTIMS

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The application of large-scale DNA-based identification in the context of armed conflict emerged in the early 1990s in response to unprecedented forensic challenges posed by mass graves and severely degraded human remains following the wars in Croatia and Bosnia and Herzegovina. This historical perspective examines the origins, methodological innovations, and lasting scientific impact of the first systematic uses of DNA technology for the identification of victims recovered from mass-fatality sites during active conflict and post-conflict period. These pioneering efforts were initiated between 1993 and 1994 at the Department of Pathology and Forensic Medicine of the University Hospital in Split, representing one of the earliest organized forensic DNA identification programs. By integrating classical forensic disciplines with early PCR based DNA profiling methods, and by establishing a structured kinship reference database, Croatian scientists developed a comprehensive and robust framework for victim identification. This work was carried out in close collaboration with leading forensic experts from the USA, enabling the transfer and adaptation of emerging molecular technologies to an active wartime environment. On December 6, 1995, during a Congressional Hearing on Intelligence and Security entitled "Mass Graves and Other Atrocities in Bosnia", Dr. Barbara Wolf formally acknowledged the Croatian DNA identification efforts, an exceptional endorsement of a forensic initiative developed outside established Western forensic systems. As part of this presentation, we will also examine the Duboki Jarak case, the first instance in Croatia in which a court (Military Court in Zagreb, April 18, 1994) formally mandated DNA analysis for the identification of victims of a mass-fatality event occurring under active wartime conditions. Notably, these applications preceded the formal incorporation of DNA analysis into international DVI guidelines later in the decade.

Keywords: forensic genetics, DVI, DNA identification, military compound explosion

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DYSPLASIA EPIPHYSEALIS HEMIMELICA: PERSONALIZED AND MULTIPLE TREATMENT MODALITIES FOR A SINGLE RARE DISEASE

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Dysplasia epiphysealis hemimelica (DEH), also known as Trevor's disease, is a non-hereditary developmental bone disease mostly diagnosed in the first decade of a patient's age. Genetic tests for *EXT1* and *EXT2* gene expression can be used for diagnosis, as in multiple osteochondromas. The clinical experience with this condition is limited due to its very rare incidence (1:1,000,000). DEH is characterized by a benign, abnormal growth of the joint cartilage that produces an irregular intra-articular mass within the joint, causing limited function and deformity of the joint and limb length discrepancy. Clinical and radiological features are generally sufficient for a correct diagnosis if one has an early suspicion. If left untreated, degenerative arthritis with all its consequences will occur. This report aims to underline a personalized treatment approach and to present mid-term outcomes of the clinical and therapeutic course in three children. A single bone or multiple bones in a single limb may be affected. In a generalized form, it usually involves multiple bones and joints in two or more limbs or even the axial skeleton. A retrospective clinical and radiological analysis of three children with different challenging localizations of the disease was performed, i.e., lateral femoral condyle, scaphoid bone, and generalized form. We used different surgical modalities, e.g., arthroscopic resection, temporary hemi-epiphysiodesis, and corrective osteotomy as treatment methods. Ten-year clinical follow-up results were good and very good. Functional and radiological outcomes in two patients (scaphoid bone, femoral condyle) were very good. In generalized DEH, outcomes were less satisfactory due to multiple affected sites and more frequent surgeries. In children with symptomatic DEH, treatment is individualized based on lesion location, size, and age. Clinical observation until puberty is recommended, as lesions may grow until skeletal maturity.

Keywords: dysplasia epiphysealis hemimelica, Trevor's disease, osteochondral lesions, personalized surgical treatment

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GENETIC DIAGNOSIS OF RARE NEUROLOGICAL DISORDERS: CURRENT TRENDS AND FUTURE DIRECTIONS

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Rare neurological disorders are predominantly genetic and represent a major diagnostic challenge. Ongoing European initiatives are targeting current limitations in genetic diagnosis of rare neurological disorders, including technology limitations, challenges in variant interpretation, lack of standardisation, insufficient integration of multi-omics approaches, inequalities in access to testing and specialised knowledge. The main focus of European collaborative frameworks is on advancing and standardisation of diagnostic approaches, pathways, and healthcare system factors. As a full member of European Reference Networks for Rare Neurological and Neuromuscular Disorders we will showcase the use of advanced diagnostic strategies, including exome and genome sequencing, cross-border data sharing, expert consultation via the Clinical Patient Management System, RNA sequencing for functional validation, and iterative reanalysis of genomic data. These approaches led to the resolution of previously unsolved cases and refinement of diagnostic pathways. Our experience highlights that the key determinants of successful diagnosis are not only technological advances but also access to collaborative networks, data integration, and multidisciplinary expertise. Ongoing efforts, including participation in European registries and expansion of multi-omics approaches, effective implementation and validation of artificial intelligence- based interpretation tools, and the development of learning healthcare systems aim to further improve diagnostic equity and efficiency. In conclusion, combining state-of-the-art genomic methods with structured European collaboration represents a critical step toward standardized, more accurate and timely diagnosis of RNDs.

Keywords: rare diseases, neurological disorders, neuromuscular disorders, genetic diagnosis, European Reference Networks

Presentation number: CSHG/CSPPM OP 04 Abstract number: 16-ISABS-2026

THE HIDDEN LANGUAGE OF CANCER: PREDICTING CLINICAL OUTCOMES THROUGH GENOMIC SIGNALS

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Cancer is increasingly recognized as a dynamic and heterogeneous biological system shaped by interactions between germline predispositions and somatic genomic alterations. Advances in high- throughput sequencing, combined with artificial intelligence (AI), have enabled the extraction of clinically relevant patterns from complex genomic data, moving oncology from descriptive classification toward predictive modeling of disease behavior. Germline variants influence susceptibility, DNA repair capacity, and pharmacogenomic response, while somatic alterations define tumor phenotype, clonal evolution, and therapeutic vulnerabilities. AI-driven approaches, including machine learning and deep learning models, provide powerful tools for integrating these genomic layers with clinical data, capturing nonlinear relationships and improving predictive performance beyond traditional biomarker-based strategies. Recent developments have demonstrated that AI-based genomic models can stratify patients according to survival outcomes, predict recurrence risk, and anticipate response to targeted therapies and immunotherapy. In addition, circulating tumor DNA enables dynamic monitoring and early detection of minimal residual disease, further enhancing predictive accuracy. The integration of multi-omics data within AI frameworks represents a key step toward more precise and individualized prediction. Despite these advances, challenges remain, including data heterogeneity, limited generalizability, and issues of interpretability and clinical validation. Addressing these barriers is essential for successful implementation. Integrating germline and somatic genomic information through AI-driven models represents a critical step toward predictive oncology, enabling more accurate forecasting of clinical outcomes and more personalized cancer management.

Keywords: next-generation sequencing, precision oncology, artificial intelligence, machine learning, molecular tumor profiling

Presentation number: CSHG/CSPPM OP 05 Abstract number: 72-ISABS-2026

IMPLEMENTATION OF NEXT-GENERATION SEQUENCING IN ROUTINE CARDIOLOGY PRACTICE: A RETROSPECTIVE STUDY FROM CLINICAL HOSPITAL CENTER RIJEKA

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To assess the diagnostic yield, clinical indications, and utility of next-generation sequencing in adult cardiology patients at the Clinical Hospital Center Rijeka. This preliminary retrospective study included adult patients referred for genetic testing for cardiac diseases between 2023 and 2025. Genetic testing was offered to all patients with suspected inherited cardiovascular disease referred by the Clinic for Cardiovascular Diseases, including cardiomyopathies and cardiac channelopathies. Whole exome sequencing with phenotype-driven bioinformatic gene panels tailored to the specific cardiac conditions was applied. Medical data were collected from the hospital information system. There were 189 adult participants included in the study, consisting of 65 female (34.39%) and 124 male (65.61%). Regarding clinical indications, 15 (7.94%) had arrhythmogenic cardiomyopathy, 76 (40.21%) had dilated cardiomyopathy, 59 (31.22%) had hypertrophic cardiomyopathy, 6 (3.17%) had noncompaction cardiomyopathy, 1 (0.53%) had restrictive cardiomyopathy, 8 (4.23%) were classified as cardiomyopathy, and 24 (12.70%) were referred as suspected cardiac channelopathy. We successfully identified a causative genetic variant in 36 of the 189 patients, corresponding to an overall diagnostic yield of 19.05%. Causative variants were detected in the following genes: *MYBPC3, TNNT2, LMNA, MYL2, MYH7, KCNH2, TTN, PKP2, DSP, TPM1,* and *DMD*. In addition, a pathogenic heterozygous trinucleotide repeat expansion in the *DMPK* gene was detected in one patient. Our preliminary results support the integration and clinical utility of whole exome sequencing into routine practice for accurate diagnosis, prognosis, and management of inherited cardiac disorders. While genetic testing provides meaningful diagnostic insights, its effectiveness in routine clinical practice remains highly dependent on careful patient selection and detailed phenotyping, underscoring the growing role of medical genetics specialists.

Keywords: genetic testing, cardiomyopathies, cardiac channelopathies, heart rhythm disorders, whole exome sequencing

Presentation number: CSHG/CSPPM OP 06 Abstract number: 60-ISABS-2026

FROM SEQUENCING TO THERAPY: IMPLEMENTING COMPREHENSIVE GENOMIC PROFILING IN ROUTINE CLINICAL PRACTICE

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The goal of this report was to evaluate the implementation of comprehensive genomic profiling (CGP) in routine diagnostics at the Department of Pathology, University Hospital Split. Material and methods included analysis of 1,093 samples (FFPE, blood and cfDNA) using next-generation sequencing (NGS)-based platforms, covering tumor-specific panels, homologous recombination repair/deficiency (HRR/HRD), myeloid and hereditary panels, and combined DN A/RNA CGP approaches. Results demonstrated successful detection of a wide range of genomic alterations, including SNVs, indels, CNVs, translocations, and gene fusions, as well as key immuno -oncology biomarkers such as tumor mutational burden (TMB), microsatellite instability (MSI), and homologous recombination deficiency (HRD). Integration of HRD testing enabled patient selection for PARP inhibitor therapy, while bioinformatics tools improved interpretation in low tumor content samples. Implementation also resulted in optimized workflows and high reproducibility. The introduction of CGP into routine diagnostics represents a significant advancement toward precision and personalized medicine, improving tumor characterization, therapeutic decision-making, and patient outcomes.

Keywords: comprehensive genomic profiling, next-generation sequencing, HRD, precision medicine, molecular diagnostics

Presentation number: CSHG/CSPPM OP 07 Abstract number: 86-ISABS-2026

Y CHROMOSOME STORY AND CROATIAN HERITAGE: MODERN CROATIAN GENETIC POOL VS ANCIENT GENETIC DATA

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Due to its turbulent demographic history, marked by extensive settlement and gene flow from diverse regions of Eurasia, Southeastern Europe (SEE) has consistently served as a genetic crossroads between East and West and a junction for the migrations that reshaped Europe's population. SEE, including modern Croatian territory, was a crucial passage from the Near East and even more distant regions and human populations in this region, as almost any other European population represents a remarkable genetic mixture. Our views of its history have been mostly renewed in the last few decades by extraordinary data obtained from Y-chromosome studies. A substantial majority, exceeding three-quarters of the present-day Croatian male population likely traces its ancestry to "Old Europeans" who came here before and after the Last Glacial Maximum. The remaining part of the population is the offspring of the people arriving in this part of Europe through the southeastern migratory route in the last 10,000 years, mainly during Neolithization. In recent times, the international research community, bringing together geneticists and archaeologists, has steadily released a growing number of ancient genomes from this region, shedding more light on its complex past population dynamics and shaping the genetic pool in Croatia and this part of Europe. We believe this presentation will help us to summarize almost three decades of scientific efforts within the analysis of the genetic structure of the Croatian population in more detail, as well as the aspects of its demographic history.

Keywords: Y chromosome, old Europeans, present-day population, ancient data

Presentation number: CSHG/CSPPM OP 08 Abstract number: 51-ISABS-2026

GENETIC CAUSES OF LISSENCEPHALY SPECTRUM: INSIGHTS FROM CHROMOSOMAL MICROARRAY AND CLINICAL/WHOLE-EXOME SEQUENCING

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Lissencephaly is a rare malformation of cortical development caused by abnormal neuronal migration. It represents a spectrum that includes agyria, pachygyria, and subcortical band heterotopia. Lissencephaly is considered as a predominantly genetic disorder, with at least 31 genes currently known to be associated with the condition. Our aim was to determine the diagnostic yield of comprehensive genetic testing in a cohort of patients with neuroimaging findings suggestive of the lissencephaly spectrum disorders and to characterize novel pathogenic variants contributing to the genetic architecture of the spectrum. We reviewed clinical and genetic findings in 23 patients with neuroimaging features suspected of the lissencephaly spectrum, seen at the Children's Hospital Zagreb between 2016 and 2025. Clinical data were obtained from medical records and outpatient assessments by clinical geneticists. Genetic testing included chromosomal microarray, clinical exome sequencing and whole-exome sequencing. A molecular diagnosis was established in 16 of 23 patients (69.5 %). The pathogenic variants involved genes related to microtubule function (*PAFAH1B1, DCX, TUBA1A, TUBB2B, DYNC1H1*), and variants in transcriptional and regulatory genes (*FOXG1, WDR62*). Five novel variants were detected in well-established lissencephaly genes (*DCX, PAFAH1B1, TUBA1A, FOXG1* and *WDR62*). Although most cases involved single-gene variants, three patients had pathogenic copy number variants (1q43q44, 22q11.21 and Xq22.3q23 deletions). Exome sequencing when used as a first-line test, complemented by chromosomal microarray, provides high diagnostic yield in patients with lissencephaly spectrum disorders. This integrated approach facilitates precise diagnosis, informs prognosis, enables targeted follow-up, and supports comprehensive genetic counselling.

Keywords: lissencephaly, pachygyria, subcortical band heterotopia, copy number variants, exome sequencing

Presentation number: CSHG/CSPPM OP 09 Abstract number: 62-ISABS-2026

ARTIFICIAL INTELLIGENCE IN CYTOGENETICS: PROGRESS IN AUTOMATED KARYOTYPE ANALYSIS

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Computer systems for karyotype analysis have evolved in parallel with advances in digital microscopy, image processing, and artificial intelligence. In the 1980s and 1990s, early digital systems enabled storage and visualization of chromosome images, but analysis remained largely manual, requiring cytogeneticists to crop, arrange, and classify chromosomes with minimal software support. From the 1990s to around 2010, semi-automated systems introduced automated metaphase detection, chromosome segmentation, and preliminary classification based on morphological features such as length, centromere position, and banding patterns. However, expert validation was still necessary to confirm final results. After 2010, major improvements in image analysis and machine learning enabled advanced cytogenetic workstations capable of rapid slide scanning and efficient analysis of large numbers of metaphase spreads. More recently, deep learning methods, particularly convolutional neural networks, have enabled highly accurate automatic detection, segmentation, and classification of chromosomes. These developments have significantly improved the speed, reproducibility, and standardization of karyotype analysis in clinical and research cytogenetics. The aim of this study is to outline the evolution of computer systems for karyotype analysis and to highlight the role of artificial intelligence in improving efficiency, accuracy, and standardization.

Keywords: computer system detection, deep learning methods, karyotype analysis methods, metaphase chromosome

Presentation number: CSHG/CSPPM OP 10 Abstract number: 53-ISABS-2026

CLINICAL FEATURES, GENETIC LANDSCAPE AND MANAGEMENT OF OSTEOGENESIS IMPERFECTA PATIENTS IN CROATIA

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The aim of the study is to evaluate clinical and molecular features and treatment of osteogenesis imperfecta patients (OI) in a cohort of Croatian patients. For this study, we made a retrospective analysis of OI patients that were diagnosed and treated in the Children's Hospital Zagreb. The clinical evaluation encompassed detailed phenotype assessment and stratification according to the modified Sillence classification system. We analyzed the total number of fractures, presence of associated anomalies and developmental abnormalities and use of bisphosphonate therapy. The molecular genetic testing was performed using the NextSeq Illumina exome sequencing (ES) and chromosomal microarray (CMA) using the Agilent 60K oligonucleotide. Variants were classified according to the ACMG/AMP guidelines. In the period 1998-2026 there were 22 patients with clinical diagnosis of OI. The most common OI type was type I detected in 12 patients, type III was present in four patients, type IV in two, type VI in three and type II in one patient. The total number of fractures ranged from 1 fracture to more than 100 fractures. Severe deformities were present in five patients. Mitral valve abnormalities were present in three patients. Three patients had neurodevelopmental abnormalities. The genetic basis of OI in these patients were alterations in *COL1A1* (12), *COL1A2* (2), *SERPINF* (3) and *IFITM5* (1). Fourteen patients received bisphosphonate therapy with variable response. Our study represents the largest cohort of OI patients in Croatia. The clinical presentation showed variable disease severity ranging from the sporadic fractures to severe bone disease with multiple deformities and loss of ambulation. The elucidation of genetic basis in these patients revealed the autosomal dominant and recessive form of disease. The variable response to bisphosphonate therapy underscores the necessity of additional treatment approaches.

Keywords: osteogenesis imperfecta, genetic testing, clinical features, therapy

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LEGAL DETERMINANTS IN MEDICAL GENETICS

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Every occurrence and every phenomenon, as a rule, possesses various aspects including a legal aspect. This equally applies to medicine in general (indeed, it is precisely on this basis that a distinct branch of law, namely medical law), has emerged and consequently to its numerous specialized fields, including medical genetics. Medical genetics is a scientific discipline characterized by continuous and almost extraordinary progress. Medical (health) law represents a significant challenge for the average legal practitioner (and non-lawyers), primarily due to the complexity and constant expansion of medical terminology, the scope of which is directly proportional to the overall advancement and increasing specialization within medicine. In this context, the criminal law dimension of these issues is of particular importance, as is the role of court-appointed medical experts in criminal proceedings. Criminal offences against human health are regulated in Chapter XIX "Criminal Offences Against Human Health" of the Criminal Code of the Republic of Croatia. The protected legal interest underlying these offences is human health itself. Considering the significance and specificity of such offences in the contemporary world, the legislature was fully justified in regulating criminal offences against human health within a separate chapter of the Criminal Code of the Republic of Croatia. Within the framework of this topic, particular attention is devoted to the criminal offence of Unauthorized Removal and Transplantation of Human Body Parts, as prescribed under Article 182 of the Criminal Code. It is indisputable that medicine and medical research are developing on a daily basis, thereby simultaneously creating opportunities for various forms of human health abuse. For this reason, by prescribing this and other criminal offences within the aforementioned chapter, the legislature sought to provide the highest possible level of protection for human health and bodily integrity.

Keywords: medical law, people's health, criminal law aspects, removal and transplantation of human body parts, court experts

POSTER PRESENTATIONS

Presentation number: CSHG/CSPPM PP 01 Abstract number: 101-ISABS-2026

FROM KNOWLEDGE TO ACTION: EMPOWERING FUTURE HEALTHCARE PROFESSIONALS IN COLORECTAL CANCER PREVENTION

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Colorectal cancer (CRC) is a major public health burden, and healthcare students are future promoters of prevention and screening. This study assessed knowledge, attitudes, and behaviours related to CRC prevention and screening among healthcare students. A cross-sectional study included 160 nursing, physiotherapy, and dental medicine students. Overall knowledge was good, with a median of 11 correct answers out of 13. Nursing students had higher knowledge scores than other groups (P=0.03). Formal education on CRC during studies was strongly associated with better knowledge (P<0.001), while information from healthcare professionals was also linked to higher scores (P=0.02). Attitudes toward prevention were generally positive, especially regarding its importance, screening benefits, trust in the healthcare system, and intention to participate in screening. Formal education was associated with more positive attitudes, including perceived benefits, self-efficacy, trust, and social influence. Nursing students showed stronger intention to participate in screening (P=0.003), while senior students perceived higher personal risk (P=0.003). Knowledge was positively correlated with information-seeking and discussing prevention with others. Main barriers were lack of knowledge, fear, and embarrassment. These findings highlight formal education as a key driver of knowledge, attitudes, and preventive behaviours. Better integration of prevention and screening into healthcare curricula may strengthen promotion of early detection. Acknowledgement: Funded by the European Union – NextGeneration EU through programme funding of the University of Osijek (UNIOS) – Institutional Project (Project code: 581-UNIOS-46) to R.S. The views and opinions expressed are solely those of the authors and do not necessarily reflect the official positions of the European Union or the European Commission. Neither the European Union nor the European Commission can be held responsible for them.

Keywords: colorectal cancer, healthcare students, knowledge, attitudes, screening

Presentation number: CSHG/CSPPM PP 02 Abstract number: 36-ISABS-2026

ASSOCIATION OF *MTHFR C677T* AND *A1298C* POLYMORPHISMS WITH OVARIAN RESERVE AND OVARIAN STIMULATION OUTCOMES IN INFERTILE WOMEN WITH AUTOIMMUNE THYROIDITIS

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Variability in the methylenetetrahydrofolate reductase (*MTHFR*) gene has been implicated in reproductive dysfunction and may influence ovarian physiology in women with autoimmune thyroiditis (AIT), a population in which infertility is increasingly recognized. This prospective genotyping study aimed to evaluate the frequency of the *MTHFR C677T* and *A1298C* polymorphisms and their association with ovarian reserve (OR) and response to controlled ovarian hyperstimulation (COH) in infertile women with AIT. Genomic DNA was extracted from peripheral blood samples, amplified by polymerase chain reaction (PCR), and genotyped using restriction fragment length polymorphism (RFLP) analysis. Associations between *MTHFR* polymorphisms and OR in AIT patients undergoing assisted reproductive technology were assessed. In addition, differences in COH outcomes across genotypes were evaluated, including the proportion of MII oocytes, fertilization rate, total number of retrieved oocytes, and the proportion of day 3 and day 5 embryos, using linear regression and group comparison analyses. A statistically significant difference was observed in both genotype distribution and allele frequency of the *MTHFR C677T* polymorphism ($p=0.00065$ and $p=0.00006$, respectively) between infertile AIT patients with diminished OR and those with preserved OR. No significant differences were found in the distribution of *MTHFR A1298C* genotypes between the same patient groups ($p>0.05$). Furthermore, the combined genotypes of both polymorphisms did not show a significant association with COH outcomes. The present results suggest that the *MTHFR C677T* polymorphism may represent a potential factor associated with diminished OR in infertile women with AIT. Accordingly, this polymorphism may have clinical relevance in the diagnostic and prognostic evaluation of infertility associated with AIT.

Keywords: autoimmune thyroiditis, gene, infertility, methylenetetrahydrofolate reductase, polymorphism

Presentation number: CSHG/CSPPM PP 03 Abstract number: 77-ISABS-2026

MILD PHENOTYPIC PRESENTATION OF RUBINSTEIN–TAYBI SYNDROME TYPE 1 IDENTIFIED BY WHOLE EXOME SEQUENCING: A CASE REPORT

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Rubinstein-Taybi syndrome type 1 (RTS1) is a rare genetic disorder caused by pathogenic variants in the *CREBBP* gene, characterized by distinctive facial features, broad thumbs and halluces, growth restriction, and varying degrees of intellectual disability. However, phenotypic expression can be highly variable, and mild presentations may remain unrecognized without advanced genetic testing. We report a female patient referred for genetic evaluation at 7 years of age following whole exome sequencing (WES) performed due to speech delay and developmental cognitive delay. Previous genetic investigations, including karyotype, *FMR1* testing, and chromosomal microarray, were normal. The patient had been followed by a pediatric neurologist since early childhood due to speech delay. Electroencephalography revealed paroxysmal irregular spike-wave complexes and treatment with lamotrigine was initiated. Brain MRI and an extensive metabolic workup were unremarkable. Developmental history showed mild global delay, with relatively preserved motor milestones but persistent speech and cognitive impairment. Multidisciplinary assessment identified behavioral immaturity and autism-like features. WES identified a heterozygous likely pathogenic variant in *CREBBP* (c.3832G>A, p.Glu1278Lys) gene, confirming RTS1. Physical examination revealed only subtle dysmorphic features, including highly arched eyebrows, high-arched palate, and mild micrognathia without classic hallmark features such as broad thumbs or halluces. This case highlights a mild and atypical presentation of RTS1, emphasizing the importance of genomic testing in patients with unexplained developmental delay – even in the absence of classic phenotypic features. The findings support the concept of variable expressivity in *CREBBP*-related disorders and expand the phenotypic spectrum of RTS1. Early genetic diagnosis is essential for appropriate management, surveillance, and genetic counseling.

Keywords: whole exome sequencing, *CREBBP*, Rubinstein-Taybi syndrome type 1, mild phenotype, developmental delay

Presentation number: CSHG/CSPPM PP 04 Abstract number: 108-ISABS-2026

ROLE OF MITOCHONDRIAL DNA MUTATIONS IN ACUTE MYELOID LEUKEMIA TREATMENT RESPONSE: A REVIEW OF THE PATHOPHYSIOLOGICAL MODEL

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Mitochondrial DNA (mtDNA) mutations have emerged as important regulators of cancer cell metabolism and survival, particularly in acute myeloid leukemia (AML), a heterogeneous hematologic malignancy characterized by variable treatment outcomes. Increasing evidence suggests that mitochondrial dysfunction contributes to leukemogenesis and influences sensitivity to chemotherapy. This abstract aims to synthesize recent findings on the role of mtDNA mutations in AML treatment response, with emphasis on specific mitochondrial genes and their functional consequences. Recent studies demonstrate that mtDNA mutations in AML frequently involve genes encoding components of the oxidative phosphorylation system, including *MT-ND1, MT-ND4*, and *MT-ND5* (Complex I), *MT-CYB* (Complex III), and *MT-CO1* (Complex IV). Mutations in MT-ND genes impair electron transport chain efficiency, leading to reduced ATP production and increased reactive oxygen species (ROS), which promote genomic instability and survival signaling. Alterations in *MT-CYB* and *MT-CO1* further disrupt mitochondrial respiration and enhance metabolic reprogramming toward glycolysis. Additionally, mutations in *MT-ATP6* (Complex V) affect ATP synthesis, contributing to altered energy homeostasis and resistance to apoptosis. Variations in mtDNA copy number and heteroplasmy levels modulate the severity of these effects and correlate with treatment response. These mitochondrial alterations support leukemic cell adaptation to chemotherapeutic stress and are particularly relevant in leukemic stem cells, which rely on oxidative phosphorylation. Importantly, targeting these dysfunctional pathways with mitochondrial inhibitors has shown potential to restore chemosensitivity and overcome resistance. In conclusion, mutations in key mtDNA-encoded respiratory genes play a critical role in AML treatment response by driving metabolic adaptation and resistance mechanisms. These alterations represent promising biomarkers and therapeutic target.

Keywords: apoptosis, gene, leukemia, mutation, mitochondrial DNA

Presentation number: CSHG/CSPPM PP 05 Abstract number: 29-ISABS-2026

FETAL GENETICS AND SPONTANEOUS PRETERM BIRTH: A SYSTEMATIC REVIEW AND META-ANALYSIS

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Spontaneous preterm birth (sPTB), defined as delivery before 37 weeks of gestation without medical induction, remains a major contributor to perinatal morbidity and mortality worldwide. The role of fetal genetic predisposition in sPTB is increasingly recognized but not yet fully elucidated. The aim of this study was to systematically evaluate the role of fetal genetic variations in sPTB. A comprehensive literature search of PubMed and Scopus databases was conducted up to June 2025 following PRISMA 2020 guidelines. Eligible studies included case-control, cross-sectional, and cohort studies assessing associations between fetal genetic variants and sPTB. Data extraction and risk of bias assessment were performed using Cochrane criteria. Meta-analyses were conducted for variants investigated in at least three independent studies using Comprehensive Meta-Analysis Software. A total of 52 studies were included in the qualitative synthesis, of which 15 met criteria for meta-analysis. Across studies, over 145 SNPs in 62 genes were reported as significantly associated with sPTB. Meta-analysis identified a statistically significant association only for the gene Vitamin D Receptor (*VDR*) FokI (C>T) polymorphism under dominant (CC vs CT+TT) and overdominant (CC+TT vs CT) models, while *TNF*-α, *IL6*, and *TLR4* variants showed no significant associations. Genome-wide studies identified several loci reaching genome- wide significance, but replication across populations was limited. These findings suggest that fetal genetic factors contribute modestly to sPTB risk, with vitamin D–related pathways representing a potential mechanism. However, the overall evidence remains inconsistent due to methodological variability and limited replication. Future large-scale, multi-ethnic studies integrating fetal and maternal genetics are needed to clarify genetic contributions and improve risk prediction.

Keywords: fetal blood, genetic association studies, genetic variation, meta-analysis, preterm birth

Presentation number: CSHG/CSPPM PP 06 Abstract number: 113-ISABS-2026

SEMAGLUTIDE ATTENUATES STEATOSIS AND PROINFLAMMATORY SIGNALING ASSOCIATED WITH MASLD PROGRESSION AT THE HEPATOCYTE LEVEL

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Metabolic dysfunction-associated steatotic liver disease (MASLD) represents a major global health burden, affecting approximately 25-40% of the adult population worldwide. The complex pathophysiology of MASLD is closely associated with obesity, insulin resistance, and type 2 diabetes. Current therapeutic options remain limited, with pharmacological treatment largely restricted to advanced stages such as metabolic dysfunction-associated steatohepatitis (MASH) and fibrosis. This study investigated the effects of semaglutide in an in vitro MASLD model using HepG2 cells exposed to oleic acid. Semaglutide (2-20 nM) significantly improved cell viability ($p < 0,01$), reduced intracellular lipid accumulation ($p < 0,01$), and decreased triglyceride levels across all concentrations compared to the MASLD model. At the protein level, semaglutide significantly reduced PPAR-α levels at 10 and 20 nM ($p < 0,05$), while TNF-α concentrations were significantly decreased at all tested concentrations ($p < 0,05$) relative to oleic acid treatment. At the gene expression level, semaglutide significantly downregulated PPAR-γ expression at all concentrations ($p < 0,05$), and reduced TNF-α expression at 5 and 10 nM ($p < 0,05$). These findings demonstrate that semaglutide attenuates both steatotic and inflammatory alterations in an in vitro MASLD model. Modulation of TNF-α suggests a key role in reducing early inflammatory signalling, which may be critical in limiting disease progression. Overall, the results support the potential of semaglutide to act at the hepatocyte level in mitigating early-stage MASLD and preventing progression toward MASH, highlighting its relevance beyond currently approved indications.

Keywords: semaglutide, MASLD, MASH, HepG2, hepatic steatosis

Presentation number: CSHG/CSPPM PP 07 Abstract number: 87-ISABS-2026

COMPUTATIONAL DRUG DISCOVERY ASSESSMENT OF CARVACROL AGAINST *PSEUDOMONAS AERUGINOSA* USING BIOINFORMATICS AND CHEMINFORMATICS

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Natural products often exhibit pleiotropic and multi-target effects, making them promising candidates for antibacterial drug discovery against antibiotic-resistant bacteria such as *Pseudomonas aeruginosa*, whose carbapenem-resistant strains are listed in the 2024 World Health Organization Bacterial Priority Pathogens List. This study evaluated carvacrol as a potential antibacterial agent against *P. aeruginosa* using an in silico drug discovery framework integrating bioinformatics and cheminformatics approaches for early-stage drug development. Physicochemical descriptors, pharmacokinetic properties, and drug-likeness were assessed with the Swiss Absorption, Distribution, Metabolism, and Excretion web tool (SwissADME). P. aeruginosa protein structures were obtained from the Research Collaboratory for Structural Bioinformatics Protein Data Bank (RCSB PDB) based on literature relevance, binding pockets were identified with the Computed Atlas of Surface Topography of Proteins (CASTp), and molecular docking was performed in triplicate using AutoDock Vina 1.1.2. Binding poses and intermolecular interactions were further analysed using the Python Molecular Graphics System (PyMOL 3.1.6.1), University of California, San Francisco Chimera (UCSF Chimera 1.19), and BIOVIA Discovery Studio Visualiser 2025. Carvacrol demonstrated a favourable in silico profile, met key drug-likeness criteria consistent with Lipinski's Rule of Five, and showed reproducible interactions with the selected protein targets. These findings support prioritising carvacrol as a potential antibacterial agent against P. aeruginosa, justifying further preclinical evaluation. They also highlight the effectiveness of bioinformatics-guided target selection, cheminformatics profiling, and structure-based computational chemistry as a resource-efficient approach for hit identification, lead prioritisation, and candidate profiling, potentially reducing early attrition before in vitro and in vivo assessments.

Keywords: antibacterial agent, antibacterial drug resistance, carvacrol, drug discovery, *Pseudomonas aeruginosa*

Presentation number: CSHG/CSPPM PP 08 Abstract number: 54-ISABS-2026

THE ROLE OF (−)-EPICATECHIN IN OXIDATIVE STRESS MODULATION IN MASLD

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The aim of this study was to evaluate the role of (-)-epicatechin (EPI) in modulating oxidative stress and lipid accumulation in an in vitro model of metabolic dysfunction-associated steatotic liver disease (MASLD). The levels of glutathione (GSH), lipid hydroperoxides (LOOHs), conjugated dienes (CDs), and intracellular triacylglycerols were determined to assess oxidative stress and lipid accumulation. MASLD was induced in HepG2 cells using oleic acid. Preventive and therapeutic experimental models were established by pre- and post-treatment with EPI (10-50 μM). Oxidative stress was assessed by measuring intracellular GSH levels and lipid peroxidation products (CDs and LOOHs) using spectrophotometric methods. Lipid accumulation was quantified by determining intracellular triacylglycerol concentrations using the enzymatic GPO-PAP method. Oleic acid treatment caused an increase in the CD and LOOH concentrations, along with increased triacylglycerol accumulation. Treatment with EPI resulted in a dose-dependent preservation of GSH levels and a reduction in lipid peroxidation markers, indicating attenuation of oxidative stress. Additionally, EPI decreased intracellular triacylglycerol content, thereby alleviating MASLD progression. (-)-Epicatechin mitigates oxidative stress and lipid accumulation in steatotic HepG2 cells by increasing antioxidant capacity and reducing lipid peroxidation. Therefore, these results support its potential role as a modulator of redox balance and lipid metabolism in MASLD. Acknowledgement: Funded by the European Union – NextGeneration EU through programme funding of the University of Osijek (UNIOS) – Institutional Project (Project code: 581-UNIOS-47) to M.S. The views and opinions expressed are solely those of the authors and do not necessarily reflect the official positions of the European Union or the European Commission. Neither the European Union nor the European Commission can be held responsible for them.

Keywords: (-)-epicatechin, MASLD, GSH, conjugated dienes, lipid hydroperoxides

Presentation number: CSHG/CSPPM PP 09 Abstract number: 65-ISABS-2026

SELECTIVE CYTOTOXIC EFFECTS OF CHAGA EXTRACT ON CANCER CELLS

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The aim of this study was to evaluate the anticancer activity of Inonotus obliquus (chaga) extract in different human cancer cell lines compared to normal cells. Chaga extract was prepared using 80% ethanol extraction from chaga powder, followed by filtration and drying. Antioxidant capacity was assessed using the DPPH assay. Human cancer cell lines (CLS-354, Detroit-562, Huh-7, HepG2) and normal human fetal lung fibroblasts (MRC-5) were cultured under standard conditions and treated with chaga extract (5-100 μg/mL) for 24 and 48 hours. Metabolic activity was determined using the MTS assay, while apoptotic changes were evaluated by propidium iodide (PI) and Hoechst staining using fluorescence microscopy. Chaga extract demonstrated antioxidant activity in the DPPH assay. Treatment for 48 hours resulted in a dose-dependent decrease in metabolic activity in cancer cell lines, indicating cytotoxic effects, while minimal effects were observed after 24 hours. The extract showed selective toxicity toward cancer cells compared to normal MRC-5 cells. Fluorescence staining revealed morphological changes characteristic of late apoptosis in treated cancer cells, particularly at higher concentrations. Chaga extract exhibits selective anticancer effects in vitro, reducing metabolic activity and inducing apoptosis in cancer cell lines, while showing no significant effects in normal cells. Therefore, these results support further investigation of chaga extract as a source of bioactive compounds with potential applications in cancer therapy. Acknowledgement: Funded by the European Union – NextGeneration EU through programme funding of the University of Osijek (UNIOS) – Institutional Project (Project code: 581-UNIOS-47) to M.S. The views and opinions expressed are solely those of the authors and do not necessarily reflect the official positions of the European Union or the European Commission. Neither the European Union nor the European Commission can be held responsible for them.

Keywords: chaga extract, Inonotus obliquus, anticancer activity, cancer cells, natural compounds

Presentation number: CSHG/CSPPM PP 10 Abstract number: 99-ISABS-2026

SGLT2 INHIBITORS: A NEW ALLY AGAINST MASLD IN TYPE 2 DIABETES

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Type 2 diabetes mellitus (T2DM) is often associated with metabolic dysfunction–associated steatotic liver disease (MASLD), for which effective treatments remain limited. This study evaluated the effects of sodium–glucose cotransporter 2 inhibitors (SGLT2i) on liver fibrosis, steatosis, and metabolic biomarkers in patients with T2DM-related MASLD. A total of 67 patients were followed for six months after initiating SGLT2i therapy. Serum PPARα, PPAR, SREBP1, and MTTP levels were measured, while liver fibrosis and steatosis were assessed using 2D-SWE and UGAP. After six months, MTTP levels significantly increased (198.4 → 428.9 ng/L; $p<0.001$), while PPAR levels decreased (895.0 → 565.4 ng/L; $p<0.001$). Liver stiffness improved, as indicated by reduced 2D-SWE values (6.22 → 6.10 kPa; $p=0.001$), which showed an inverse correlation with MTTP ($\rho=-0.268$; $p=0.03$). UGAP values were positively correlated with 2D- SWE ($\rho=0.365$; $p=0.002$) and with leukocyte count, triglycerides, and liver enzymes, while demonstrating a negative association with HDL cholesterol ($p\leq0.01$). Significant improvements were also observed in fasting glucose, HbA1c, INR, waist circumference, and BMI ($p\leq0.04$). SGLT2i therapy was associated with early improvements in liver steatosis, fibrosis, and metabolic control, accompanied by favorable changes in MTTP and PPAR, suggesting improved hepatic lipid metabolism. Larger controlled studies are needed to confirm these findings. Acknowledgement: Funded by the European Union – NextGeneration EU through programme funding of the University of Osijek (UNIOS) – Institutional Project (Project code: 581- UNIOS-46) to R.S. The views and opinions expressed are solely those of the authors and do not necessarily reflect the official positions of the European Union or the European Commission. Neither the European Union nor the European Commission can be held responsible for them.

Keywords: SGLT2 inhibitors, MASLD, T2DM, PPARs, MTTP

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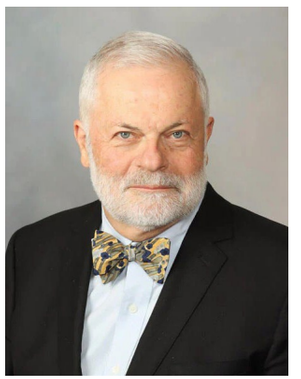
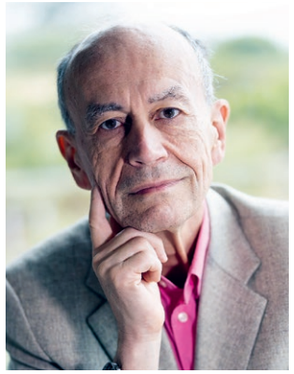
Dragan Primorac, M.D., **Ph.D.**, is a pediatrician, forensic expert and geneticist. He is the first recipient of the title "Global Penn State University Ambassador" and currently he serves as the Chair of the International Affairs Committee of the American Academy of Forensic Sciences. He is professor at Eberly College of Science, The Pennsylvania State University, and Henry C. Lee College of Criminal Justice and Forensic Sciences, University of New Haven, in the United States and as professor at Medical Schools in Split, Osijek and Rijeka as well as at Department of Biotechnology, University of Rijeka, in Croatia. In October of 2016 he was appointed as a visiting professor at the College of Medicine and Forensics, Xi'an Jiaotong University, People's Republic of China. Dr. Primorac is one of the pioneers in DNA identification of skeletal human remains from mass graves. Currently, he has particular interest in metabolic bone and cartilage disorders, pain treatment, sports medicine as well as in personalized and regenerative medicine. Dr. Primorac was an invited speaker at more than 150 conferences all around the world. His work was published in most cited journals including Science and Nature and his papers have been cited more than 4300 times (Google Scholar) while h-index is 29. Currently, he is a team leader of the Croatian partner in the international consortium within EU FP7 project entitled "Multi-dimensional OMICS approach to stratification of patients with low back pain", worth 7.6 million euros. He is the co-founder and the President of the International Society of Applied Biological Sciences (www.isabs.hr). In 2017 he was elected president of The Croatian Society for Human Genetics. Dr. Primorac received 21 domestic and international awards. From 2003 to 2009 he served as Minister of Science, Education and Sports of the Republic of Croatia.



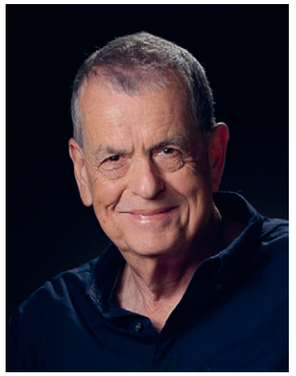
Stanimir Vuk-Pavlović, **Ph.D.**, is Professor Emeritus of Biochemistry and Molecular Biology at the Mayo Clinic College of Medicine and Science, as well as Director Emeritus of the Stem Cell Laboratory, Mayo Clinic Comprehensive Cancer Center in Rochester, Minnesota. He is a native of Zagreb where he graduated from the School of Science, University of Zagreb. He obtained his Ph.D. in biophysics from the same University and received his postdoctoral training at the Weizmann Institute of Science, Rehovot, Israel. After a stint at the Institute of Immunology in Zagreb, he joined the Mayo Clinic, where he has remained for the rest of his career. For his contribution to the Croatian war of independence, he was awarded two presidential medals. Prof. Vuk-Pavlović is the corresponding (foreign) member of the Croatian Academy of Science and Arts. He has been involved with ISABS from the very beginning, joining Dragan Primorac and the late Moses Schanfield in organizing ISABS conferences. He initiated the collaboration of ISABS with the Mayo Clinic and has served as program director for several past conferences, including the current one.

ISABS 2026
ABOUT NOBEL LAUREATES



Thomas Christian Südhof, **M.D.**, **Ph.D.**, was born in Göttingen, Germany in 1955 and obtained his M.D. and doctoral degrees from the University of Göttingen in 1982. He performed his doctoral thesis work at the The Max Planck Institute for Biophysical Chemistry in Göttingen with Prof. Victor P. Whittaker on the biophysical structure of secretory granules. From 1983 to 1986, Südhof trained as a postdoctoral fellow with Drs. Mike Brown and Joe Goldstein at UT Southwestern in Dallas, TX, and elucidated the structure, expression and cholesterol-dependent regulation of the LDL receptor gene. Südhof began his independent career in 1986 at UT Southwestern, where he stayed until 2008 and, among others, was the founding chair of the Department of Neuroscience. In 2008, Südhof moved to Stanford, and became the Avram Goldstein Professor in the School of Medicine at Stanford University. In addition, Südhof has been an Investigator of the Howard Hughes Medical Institute since 1986. Prior to becoming a neuroscientist, Südhof was trained in the biophysics of subcellular organelles at the The Max Planck Institute for Biophysical Chemistry and in cholesterol metabolism at UT Southwestern. When Südhof started his laboratory, he decided to switch to neuroscience to study synapses because of their central, as yet incompletely understood role in brain function. Südhof's work initially focused on the mechanism of neurotransmitter release, which is the first step in synaptic transmission that accounts for the speed and precision of information transfer in the brain. It was for this work that Südhof was awarded in 2013 the Albert Lasker Basic Medical Research Award (with Richard Scheller) and the Nobel Prize in Physiology or Medicine (with James Rothman and Randy Schekman). In the last decade, Südhof's research emphasis has switched to focus on a different unsolved problem in neuroscience that regards synapses, namely how synapses are established specifically between defined pre- and postsynaptic neurons, and how such connections are endowed with specific properties by these neurons. Addressing this fundamental question is essential for understanding how circuits are wired and how they process information, but the basic rules that govern synapse formation and specification are only now beginning to emerge. Elucidating these rules is the goal of Südhof's present work.

Gregg L. Semenza M.D., **Ph.D.**, received his B.S. degree in genetics from Harvard University and the M.D. and Ph.D. degrees from the University of Pennsylvania. He completed an internship and a residency in pediatrics at Duke University, and postdoctoral studies in medical genetics at Johns Hopkins University. He joined the Johns Hopkins faculty where he established his own laboratory. He later served as director of the Vascular Program at the Johns Hopkins Institute for Cell Engineering. Dr. Semenza is known for his investigations of how cells use and regulate oxygen and for his discovery of hypoxia-inducible factor (HIF), a molecule that is activated by reduced oxygen availability in cells and that plays a critical role in enabling cells to survive in certain disease states. Semenza's research opened a door for the investigation and development of novel treatments for diseases such as cancer and ischemic cardiovascular disease, in which reduced oxygen availability is a major feature of disease. Dr. Semenza was recognized for his work with multiple awards throughout his career. He shared the 2010 Canada Gairdner International Award and the 2016 Albert Lasker Basic Medical Research Award with the American William G. Kaelin, Jr., and the British Sir Peter J. Ratcliffe. He is a member of the National Academy of Sciences and the National Academy of Medicine. For his discoveries he was awarded the 2019 Nobel Prize for Physiology or Medicine (shared with Kaelin and Ratcliffe).



Aaron Ciechanover, **M.D.**, **Ph.D.**, was born in Haifa, Israel in 1947. He is currently a Distinguished Research Professor in the Faculty of Medicine at the Technion - Israel Institute of Technology in Haifa, Israel. He received his M.Sc. (1971) and M.D. (1973) from the Hebrew University in Jerusalem. He then completed his national service (1973-1976) as military physician and continued his studies to obtain a doctorate in biological sciences in the Faculty of Medicine in the Technion (D.Sc.; 1982). There, as a graduate student with Dr. Avram Hershko and in collaboration with Dr. Irwin A. Rose from the Fox Chase Cancer Center in Philadelphia, USA, they discovered that covalent attachment of ubiquitin to a target protein signals it for degradation. They deciphered the mechanism of conjugation, described the general proteolytic functions of the system, and proposed a model according to which this modification serves as a recognition signal for a specific downstream protease. As a post-doctoral fellow with Dr. Harvey Lodish at the M.I.T., he continued his studies on the ubiquitin system and made additional important discoveries. Over the years it has become clear that ubiquitin-mediated proteolysis plays major roles in numerous cellular processes, and aberrations in the system underlie the pathogenetic mechanisms of many diseases, among them certain malignancies and neurodegenerative disorders. Consequently, the system has become an important platform for drug development. Among the numerous prizes Ciechanover received are the 2000 Albert Lasker Award, the 2002 EMET Prize, the 2003 Israel Prize, and the 2004 Nobel Prize (Chemistry; shared with Drs. Hershko and Rose). Among many academies, Ciechanover is member of the Israeli National Academy of Sciences and Humanities, The European Molecular Biology Organization (EMBO), the American Academy of Arts and Sciences (Foreign Fellow), the American Philosophical Society, the National Academies of Sciences (NAS) and Medicine (NAM) of the USA (Foreign Associate), the Pontifical Academy of Sciences at the Vatican, the Chinese Academy of Sciences (CAS; Foreign Member), the Russian Academy of Sciences (Foreign Member), and the German Academy of Sciences (Leopoldina).

ISABS 2026
ABOUT INVITED SPEAKERS

Zachi I. Attia, Ph.D., is the co-director of Artificial Intelligence for Cardiovascular Medicine at Mayo Clinic, where he manages multiple teams of AI researchers developing innovative diagnostic tools for early detection of heart diseases. Under his leadership, several AI algorithms have been FDA-approved, demonstrating their clinical effectiveness and impact on patient care. These models, including those that detect conditions like left ventricular dysfunction and cardiac amyloidosis from ECG data, are transforming how clinicians identify and manage cardiac conditions, often using data from everyday devices such as Apple Watches. His work focuses on integrating these advanced AI solutions into clinical practice, making diagnostics faster, more accurate, and accessible to patients globally. He also holds an Executive M.B.A. from MIT, which supports his efforts in translating research into clinical practice and navigating operational aspects of healthcare innovation. Dr. Attia directs efforts across multiple research teams to develop, validate, and implement AI models that go beyond traditional cardiology, including detecting non-cardiac conditions like cirrhosis using ECG findings. His multidisciplinary approach includes conducting digital clinical trials and advancing explainable AI to ensure that these technologies are understandable and trusted by healthcare providers. With numerous patents and multiple FDA-approved AI models already in use, Dr. Attia's work continues to lead the transformation of cardiovascular and broader medical diagnostics through cutting-edge artificial intelligence.

Dr. Veronique Belzil is an Associate Professor of Neurology and Genetic Medicine at Vanderbilt University Medical Center and Director of the Vanderbilt ALS Research Center, whose mission is to accelerate discoveries in ALS into better diagnostics, biomarkers, and treatments through collaborative, patient-centered translational research. Her commitment to ALS is deeply personal, as a family member's diagnosis motivated her to pursue a PhD in Neuroscience focused on ALS genetics at the University of Montreal under the mentorship of Dr. Guy A. Rouleau. She then completed postdoctoral training at Mayo Clinic in Florida with Dr. Leonard Petrucelli, where she established the foundation of the translational research program she now leads at Vanderbilt.

Richard D. Braatz, **Ph.D.**, received a B.S. in chemical engineering from Oregon State University and an M.S. and Ph.D. from the California Institute of Technology. He joined the faculty at the University of Illinois at Urbana-Champaign where he rose up the ranks to Millennium Chair and Professor of Chemical and Biomolecular Engineering. After spending a year at Harvard University as a Visiting Scholar, he moved to the Massachusetts Institute of Technology (MIT) where he is the Edwin R. Gilliland Professor of Chemical Engineering and the Faculty Director of the Center for Continuous mRNA Manufacturing and the Center for Biomedical Innovation. Most of the research in his laboratory concerns the development of advanced manufacturing technologies for genomic medicines and vaccines. He serves on various Scientific Advisory Boards, and has collaborated with numerous companies including Novartis, Pfizer, Merck, Biogen, Amgen, Takeda, and Abbott Labs. His startup activities include cofounding BioCurie Inc. where he serves as the Chief Scientist and on the Board of Directors. Dr. Braatz has been recognized for his work with multiple awards throughout his career, including the AIChE PD2M Award for Outstanding Contribution to QbD for Drug Substance, the AIChE Excellence in Process Development Research Award, the ISA Technical Innovation Award, and the IEEE Control Systems Society Transition to Practice Award. Dr. Braatz is a member of the National Academy of Engineering.

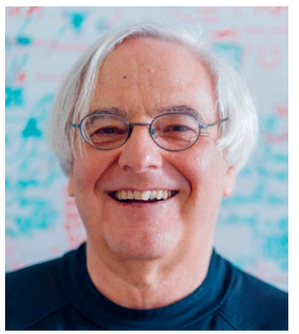
Andrea Britta Maier is Oon Chiew Seng Professor in Medicine, NUS Academy of Healthy Longevity, National University of Singapore and Professor of Gerontology, Human Movement Sciences VU Amsterdam after serving from 2016 to early 2021 as Divisional Director at the Royal Melbourne Hospital, Australia, and as Professor of Medicine and Aged Care at the University of Melbourne, Australia. She graduated in Medicine from the University of Lübeck, Germany, was registered 2009 in The Netherlands as Specialist in Internal Medicine-Geriatrics and was appointed Full Professor of Gerontology at Vrije Universiteit Amsterdam (The Netherlands) in 2013. Andrea B. Maier is an authority in human cohort studies and randomized controlled trials with more than 500 peer reviewed publications (H index 90, i10 index 374). She is an invited member and advisor of international academic and health policy committees and funding agencies, including the World Health Organization evaluating the United Nations Decade of Healthy Ageing and Hevolution. She is the past Founding President of the Healthy Longevity Medicine Society and serves as a selected Member of The Royal Holland Society of Sciences and Humanities, Fellow of the Atria Academy of Science and Medicine, and Board of the Academy for Health and Lifespan Research. In 2023, she co-founded the NUS Academy for Healthy Longevity to conduct, teach

and disseminate Geroscience and Precision Geromedicine. To bring highest evidence based Precision Geromedicine to clinical practice, she founded Chi Longevity – Asias first evidence based Precision Geromedicine clinics – in 2022 and established Maier's Longevity in 2025 in the Netherlands.

Dr. Gian Marco Conte is an AI Scientist, Senior Associate Consultant, and Assistant Professor of Radiology at the Mayo Clinic in Rochester, Minnesota, USA. In his current role as an AI Scientist in the Department of Radiology, Dr. Conte focuses on integrating AI into clinical practice, bridging the gap between research and healthcare applications. In addition to his clinical and research work, Dr. Conte is part of the editorial boards of "Radiology: Artificial Intelligence", and the "Journal of Imaging Informatics in Medicine", serving as Associate Editor for both journals. Dr. Conte earned both his MD and Ph.D. from Vita-Salute San Raffaele University in Milan, Italy, where his early research centered on the application of perfusion MRI in neuroimaging. Following his Ph.D., Dr. Conte pursued postdoctoral research at Mayo Clinic, focused on integrating machine learning techniques with medical imaging- particularly in the areas of neuroimaging and neuro-oncology.

Dr. Henry Erlich's laboratory has been involved for over 40 years in the development and application of molecular genetic technology such as PCR and DNA-based HLA typing for the analysis of infectious diseases and autoimmune diseases. They have also focused on the development of diagnostic tests for monogenic disease, such as cystic fibrosis and the hemoglobinopathies, as well as the application of PCR in forensic genetics. His lab performed the first forensic DNA case in the US in 1986 (Penn. Vs. Pestinikas) and the first post-conviction review. They were also applying the HLA typing system to the analysis of type 1 diabetes, inflammatory bowel disease, and many other autoimmune diseases. They have pioneered the application of next generation sequencing (NGS) to the analysis of the highly polymorphic HLA class I and class II genes as well as to the analysis of mitochondrial DNA for forensic analyses. They applied HLA NGS systems to the analysis of cell free DNA in plasma for detecting donor DNA in recipient plasma for early detection of graft rejection. Recently, they are using a probe capture/NGS strategy to analyze fetal DNA in the maternal plasma to develop a non-invasive prenatal test (NIPT) for the beta-hemoglobinopathies.

Antonio Esposito is a Full Professor of Radiology at the University Vita - Salute San Raffaele and Deputy Scientific Director of IRCCS San Raffaele Hospital. He also serves as Director of the Center for Experimental Imaging, driving innovation in advanced imaging technologies. A leading expert in MRI and CT, based cardiovascular and oncologic imaging, Prof. Esposito has been at the forefront of integrating Artificial Intelligence into radiology, enhancing diagnostic precision and predictive analytics. He oversees the Advanced Imaging Unit for Personalized Medicine, performing thousands of diagnostic procedures annually, and directs the Preclinical Imaging Facility, advancing disease research through state-of-the-art imaging modalities. With over 300publicationsin high-impact scientific journals, he has led and contributed to numerous national and international research projects. He also founded and coordinates the inter-university master's program in Health Informatics, a collaboration between UniSR and Politecnico di Milano,shaping the next generation of digital health leaders. As Co-Director of the Strategic Artificial Intelligence Program at San Raffaele, he spearheads initiatives that integrate AI into medicine, fostering personalized, data-driven, and more sustainable healthcare solutions.



Claudio Franceschi is an immunologist and Professor Emeritus of General Pathology at the University of Bologna. He is internationally recognized for proposing the theory of inflammaging, a hallmark concept describing chronic, low grade inflammation as a driver of aging. His career includes professorships in Padua, Modena, and Bologna, and he currently leads a laboratory in Russia. Franceschi's research has shaped modern biogerontology through pioneering studies on immunosenescence and the biology of centenarians. Author of roughly 900 scientific publications, he has coordinated major European research projects and received numerous international awards and honorary degrees.

Alex Golter education is in the field of cognitive neuroscience. He has a Ph.D from Ben-Gurion University and subsequent postdoc at Vanderbilt University involved the study of executive functions and the role of intention and consciousness in the control of behavior. Currently is leading digital markers development for psychiatry at Teva pharmaceuticals.

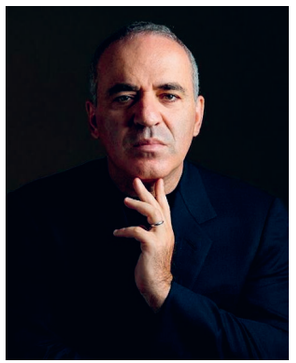
Dr. Steven Hart is a Consultant in Artificial Intelligence and Associate Professor of Biomedical Informatics at Mayo Clinic, where his research bridges computational innovation and clinical translation. His work focuses on advancing digital pathology, medical imaging, and precision oncology through state-of-the-art machine learning and computer vision. He develops large-scale frameworks for analyzing whole slide images, integrating advanced architectures such as Vision Transformers and self-supervised learning to characterize tumor micro-architecture, heterogeneity, and disease progression. His research also emphasizes adaptive exploration strategies for computationally efficient image analysis, with applications in biomarker discovery and integration of pathology-derived insights into clinical decision support. In addition to imaging, Dr. Hart has contributed to understanding genetic predisposition to hereditary cancers, linking molecular and imaging biomarkers with patient outcomes. His work is driven by the goal of accelerating discovery-to-practice translation, enabling data-driven diagnostics, and supporting the next generation of precision medicine. Through leadership in biomedical informatics, AI, and clinical research, Dr. Hart's scientific contributions highlight how advanced computation can transform the practice of medicine-bringing scalable, intelligent, and interpretable solutions to complex biomedical challenges.

Vitaly Herasevich, **MD**, **PhD**, **MSc** is a Professor of Anesthesiology and a Professor of Medicine in the Department of Anesthesiology and Perioperative Medicine, Division of Critical Care, Mayo Clinic, Rochester, Minnesota. He has been involved in medical informatics for over 25 years, with a specific concentration on applied clinical informatics in critical care and the science of health care delivery. He joined Mayo Clinic in 2006 and co-direct of the Mayo Clinic Clinical Informatics in Intensive Care Laboratory. Dr. Herasevich's applied clinical informatics in critical care work included clinical data representation, ambient decision support and alerting systems for early detection of critical syndromes and hospital-wide surveillance. He architected data warehouses in support of clinical decision-making quality and outcomes research. With longstanding interest in information security and more recent in computer vision Dr. Herasevich has coauthored over 150 scientific articles and book "Health Information Evaluation Handbook" (now in second edition). Dr. Herasevich has a long mentorship record, appointed with Full faculty privileges in clinical research and also artificial intelligence at Mayo Graduate School, developed Informatics Curriculum for Mayo Medical School, CCaTS and Master in Medical AI programs. Dr. Herasevich contributed as principal investigator, co-investigator and informatics expert for a number of past and ongoing federally and industry funded projects totaling more than $115 millions in research support. Dr. Herasevich is Fellow of American College of Critical Care Medicine (FCCM), Fellow of HIMSS (FHIMSS), Fellow of American Medical Informatics Association (FAMIA) and President of Minnesota HIMSS Chapter.

Yufei Huang received his Ph.D. in Electrical and Computer Engineering from the State University of New York at Stony Brook. He is currently a Professor at the Department of Medicine, School of Medicine, University of Pittsburgh and Leader in AI for Cancer Research at UMPC Hillman Cancer Center. He also holds joint appointments at the Electrical and Computer Engineering, Intelligent Systems Program, and Pharmaceutical Sciences at the University of Pittsburgh. Prior to that, he was Professor and Associate Chair of Research of the Department of Electrical and Computer Engineering at the University of Texas at San Antonio, and an adjunct professor at the Department of Population Health Science at the University of Texas Health San Antonio. He has been a visiting professor at the Center of Bioinformatics, Harvard Center for Neurodegeneration & Repair. Dr. Huang has multi-disciplinary expertise in cancer genomics, clinical informatics, and AI/machine learning. His current research focuses on developing AI and informatics tools for epitranscriptomics and spatially resolved single-cell transcriptomics analysis and using these tools to study cancer and cancer viruses. He was a recipient of the National Science Foundation (NSF) CAREER Award, UTSA Presidential Achievement Award on Research Excellence, UPMC Hillman Senior Fellow for Innovative Cancer Research, and multiple best paper awards. His research has been supported by NSF, NIH, Air Force Office of Scientific Research, Army Research Lab, and DOD. He is a member of the National Cancer Institute Informatics (NCI) Technology for Cancer Research Program and an associate member of the NCI Cancer System Biology Consortium. He has served as Associate Editor for multiple journals including IEEE Reviews in Biomedical Engineering, IEEE Journal on Biomedical Health Informatics, and Frontiers Genetics. Currently, he serves as the Chair of the IEEE Engineering in Medicine and Biology Society, Biomedical and Health Informatics Technical Committee. He was the General Chair of the 2023 IEEE-EMBS International Conference on Biomedical Informatics (BHI 2023) and Technical Committee Co-Chair of BIBM '2021.

Dr. David T. Jones is a behavioral neurologist and Founding Director of the Neurology Artificial Intelligence Program at Mayo Clinic. He is internationally recognized for his work at the intersection of clinical neurology, quantitative neuroimaging, and artificial intelligence. Dr. Jones specializes in cognitive and behavioral neurology, with a focus on early-onset and atypical Alzheimer's disease, normal pressure hydrocephalous, frontotemporal dementia, and limbic-predominant syndromes. He developed the first clinical criteria for dysexecutive Alzheimer's disease and introduced a novel limbic-predominant amnestic neurodegenerative syndrome. His research combines advanced PET and MRI techniques with computational modeling to uncover functional brain disruptions across the spectrum of neurodegeneration. A clinician-scientist by training, Dr. Jones has led major efforts in translational neuroimaging, computational brain modeling, and data-driven biomarker development,

including the design and deployment of StateViewer, a graph-based diagnostic AI system integrated across Mayo Clinic's clinical enterprise. His work aims to build validated, scalable, and equitable neurotechnology platforms that bring precision medicine to neurological care. Dr. Jones has authored over 200 publications and contributed foundational insights into computational models of brain function and dementia, the use of FDG-PET in clinical practice, and the application of machine learning to multimodal brain data. His work has been recognized by the Alzheimer's Association, Mayo Clinic, and multiple national news outlets and research programs.



Garry Kasparov. Born in Baku, Azerbaijan, in the USSR in 1963, Garry Kasparov became the youngest world chess champion in history in 1985. He frequently spoke out against the Soviet Communist regime, which accustomed him at an early age to being in political hot water, as when he refused to play under the Soviet flag in his 1990 world championship match against Karpov. His 1996-97 matches against the IBM supercomputer Deep Blue put chess and artificial intelligence on front pages all over the world. In 2005, still ranked #1, he retired to form the Russian pro-democracy opposition against the rising dictatorship of Vladimir Putin. Facing imminent arrest during Putin's crackdown, Kasparov entered exile in New York and became a Croatian citizen, reflecting his long-time support and time there. In 2017, he founded the Renew Democracy Initiative (RDI) in New York, dedicated to promoting the principles of the free world at home and fighting authoritarianism abroad. Kasparov later co-founded the World Liberty Congress, which aims to unite dissidents from around the world to fight against authoritarian regimes. The Kasparov Chess Foundation sponsors chess education and many of the world's top young talents. Kasparov's post-chess career also includes a busy schedule of writing, advocacy, and public speaking on foreign policy, AI, and achieving peak mental performance. His 2007 book How Life Imitates Chess on strategy and decision-making is available in over 20 languages. His prescient 2015 book, Winter Is Coming: Why Vladimir Putin and the Enemies of the Free World Must Be Stopped, warned against the increasing belligerence of Russia and other illiberal regimes. His 2017 book on the inside story of his matches against Deep Blue and his groundbreaking theories on human-plus-machine and artificial intelligence is Deep Thinking: Where Machine Intelligence Ends and Human Creativity Begins. Kasparov's latest book (May 2026) is The World of Fake Values, which updates and unifies his critical insights into democracy, tech and AI, and geopolitics. He is also the author of two acclaimed series of chess books, My Great Predecessors and Modern Chess. Kasparov is a regular contributor to many major publications and publishes The Next Move Substack with RDI. In 2025, he hosted the Autocracy in America podcast for The Atlantic magazine, and his active social media presence boasts over 1.5 million followers.

Manolis Kellis is a professor of computer science at MIT, a member of the Broad Institute of MIT and Harvard, a principal investigator of the Computer Science and Artificial Intelligence Lab at MIT, and head of the MIT Computational Biology Group (compbio.mit.edu). His research includes disease circuitry, genetics, genomics, epigenomics, coding genes, non-coding RNAs, regulatory genomics, and comparative genomics, applied to Alzheimer's Disease, Obesity, Schizophrenia, Cardiac Disorders, Cancer, and Immune Disorders, and multiple other disorders. He has led several large-scale genomics projects, including the Roadmap Epigenomics project, the ENCODE project, the Genotype Tissue-Expression (GTEx) project, and comparative genomics projects in mammals, flies, and yeasts. He received the US Presidential Early Career Award in Science and Engineering (PECASE) by US President Barack Obama, the Mendel Medal for Outstanding Achievements in Science, the NIH Director's Transformative Research Award, the Boston Patent Law Association award, the NSF CAREER award, the Alfred P. Sloan Fellowship, the Technology Review TR35 recognition, the AIT Niki Award, and the Sprowls award for the best Ph.D. thesis in computer science at MIT. He has authored over 325 journal publications cited more than 190,000 times. He has obtained more than 20 multi-year grants from the NIH, and his trainees hold faculty positions at Stanford, Harvard, CMU, McGill, Johns Hopkins, UCLA, and other top universities. He lived in Greece and France before moving to the US, and he studied and conducted research at MIT, the Xerox Palo Alto Research Center, and the Cold Spring Harbor Lab. For more info, see: compbio.mit.edu

Dr. Shelby Kutty is a pediatric and congenital cardiologist, physician-scientist, and academic leader with extensive international experience in cardiovascular medicine, imaging, and translational research. He completed his medical education in India and pursued advanced clinical and research training in Australia, Canada, and the United States. Dr. Kutty earned a PhD from the University of Nebraska Medical Center and a Master of Healthcare Management degree from Harvard University. Throughout his career, he has held senior academic and clinical positions at leading institutions, including the University of Nebraska Medical Center and Johns Hopkins Medicine, where he served as Director of Pediatric and Congenital Cardiology and co-director of the Blalock-Taussig-Thomas Heart Center. His research focuses on congenital heart disease, cardiovascular imaging, myocardial function, therapeutic ultrasound, and the application of artificial intelligence in cardiovascular medicine. Dr. Kutty has authored hundreds of peer-reviewed scientific publications and has contributed extensively to international collaborations in healthcare innovation and medical research. He currently serves as Vice President and Chief Academic Officer at BayCare Health System in Florida.

Dr. Edward Laskowski has been recognized for exceptional contribu-tions to fitness and exercise science and to sports and musculoskeletal medicine and rehabilitation, being named among "America's Top Physicians" and annually acknowledged as one of the "Best Doctors in America," a national award selected by his peers. He was selected to serve as an Olympic Polyclinic physician at the 2002 Olympic Winter Games in Salt Lake City. From 2006 to 2010, he was appointed by President George W. Bush to serve as a member of the President's Council on Physical Fitness and Sports, serving under both Presidents Bush and Obama, and he received special commendation by the U.S. Department of Health and Human Services for his service to the Council. He has also received the Distinguished Public Service award from the American Academy of Physical Medicine and Rehabilitation, and he was voted by his colleagues to receive the Mayo Clinic Distinguished Clinician Award, the highest clinical honor for Mayo Clinic physicians. The Mayo Clinic Sports Medicine Symposium bestowed the honor of naming the keynote lecture of the symposium the "Edward R. Laskowski, M.D. Honorary Keynote," and he also recently was awarded the "Frank Krusen Visiting Professorship" by the Department of Physical Medicine and Rehabilitation, Mayo Clinic Rochester. He also has received one of the American College of Sports Medicine's highest honors, the Citation award. Dr. Laskowski's research has garnered funding from competitive granting organizations, including the National Institutes of Health (NIH), and he has been invited to present his work nationally and internationally. His interests and research focus include general fitness, strength training, stability training, performance enhancement, injury rehabilitation and prevention, musculoskeletal rehabilitation, exercise for population health and fitness, combating obesity and inactivity, sports for the disabled, and wellness program design, implementation, and efficacy.

Prof. Gordan Lauc, **Ph.D.,** is a Professor of Biochemistry and Molecular Biology at the University of Zagreb, Director of the National Centre of Scientific Excellence in Personalised Healthcare, Honorary professor at the Kings College London and member of the Johns Hopkins Society of Scholars. He graduated Molecular Biology in 1992 and got PhD in Biochemistry in 1995 at the University of Zagreb. In 2017 he initiated the launch of the Human Glycome project and is one of its two co-directors. His research team is pioneering high throughput glycomic analysis and the application of glycan biomarkers in the field of precision medicine. By combining glycomic data with extensive genetic, epige-netic, biochemical, and physiological data on over 200,000 individual they are trying to understand the role of glycans in normal physiology and disease. Professor Lauc co-authored over 300 research articles that are cited over 15,000 times in Google Scholar. In 2007 he founded Genos, a biotech company that is currently global leader in high throughput glycomics. Research in Genos led to the development of the GlycanAge test of biological age.

Shounak Majumder, MD is an Associate Professor of Medi-cine and Consultant in the Division of Gastroenterology and Hepatology at Mayo Clinic, Rochester, MN. He serves as Chair of Innovation for the GI Division and Program Director for the Pancreas Fellowship Program. Shounak is a board-certified gastroenterologist and fellowship-trained in me-dical pancreatology. He is a clinical expert in the manage-ment of pancreatic diseases and a nationally and inter-nationally recognized key opinion leader in the field of pancreatology. Shounak is a NIH-funded physician scientist and leads the Pancreatic Cancer Early Detection Research Program at Mayo Clinic. His research program focuses pri-marily on developing novel molecular approaches and digital tools for early detection of pancreatic cancer. Signi-ficant scientific contributions of his program include discovery and validation of methylated DNA biomarkers in pancreatic juice, cyst fluid and blood for detecting pancreatic cancer and advanced precancer. His ongoing research studies include estimating pancreatic cancer risk in patients with chronic pancreatitis, pancreatic cysts and pancreatic steatosis, prospective validation of biomarkers for early detection of pancreatic cancer in high-risk individuals and developing artificial intelligence-based applications for pancreatic cancer screening. Shounak has published extensively in the field of pancreatology including high-impact journals such as Lancet, JAMA Oncology and Gastroenterology. He also serves as the Vice Chair for the Pancreatic Disorders (PAN) section of the American Gastroenterological Association (AGA) Institute Council.

Michal Melamed, **Ph.D.**, is an Associate Director of Data Science who develops AI tools for biomedical research. Her current work focuses on improving early-stage antibody discovery, where scientists must rapidly review thousands of images to identify promising antibody-secreting cells. Michal leads the development of a real-time AI system that supports this process by highlighting cells with a higher likelihood of downstream success, helping teams make decisions more efficiently and with greater confidence. Michal's broader experience includes applying AI to biolog-ical signals, clinical research, and digital health. She holds a Ph.D. from the Technion - Israel Institute of Technology.

Dr. Samia Mora is a cardiologist and molecular epidemiologist who has advanced research on lipids, inflammation, and cardiometabolic diseases. She is Professor of Medicine at Harvard Medical School, the Director of the Center for Lipid Metabolomics and Director of the Biorepository, Divisions of Preventive and Cardiovascular Medicine, Brigham and Women's Hospital, Mass General Brigham, Boston, MA. The author of over 200 peer-reviewed publications, Dr. Mora's research focuses on risk factors and prevention of cardiovascular disease. Dr. Mora is a highly sought-after and exemplary mentor and teacher. She received her undergraduate degree from Harvard University, and her medical degree from Harvard Medical School. She completed an internal medicine residency at Massachusetts General Hospital and cardiovascular disease fellowship at Johns Hopkins Hospital, where she also obtained an MHS degree (Epidemiology) from the Johns Hopkins Bloomberg School of Public Health. Dr. Mora is board certified in cardiovascular disease and echocardiography, and serves on the Editorial Boards of JAMA Internal Medicine and Associate Editor of Atherosclerosis. Dr. Mora is a Fellow of the American Heart Association and the American College of Cardiology. She is an Elected Member of the American Society for Clinical Investigation and the Association of University Cardiologists, and a 2024 Honorary Lifetime Member of the National Lipid Association. She currently serves as Executive Board Member and President-Elect of the International Society of Atherosclerosis.

Dr. Ir. Eskeatnaf Mulugeta is an assistant professor at Erasmus University Medical Center Rotterdam (Erasmus MC) in The Netherlands. He has a multidisciplinary educational background with a Bachelor's degree in Pharmacy, followed by three Master's degrees in Biotechnology, Bioinformatics, and Molecular Medicine. He performed his PhD research at Erasmus MC in the lab of Prof. Dr. Joost Gribnau, where he investigated the evolution of mammalian sex chromosomes (X&Y) during evolution and development. He then moved to the Institut Curie (Paris, France) for his postdoctoral research (with Prof. Edith Heard), focusing on cancer genomics and epigenomics, and also adapting Naked mole rats as model animals for aging and cancer research. After completing his postdoctoral research, he started his research group at Erasmus MC in the Department of Cell Biology. Eskeatnaf's current research focus includes deciphering and understanding gene regulatory networks that orchestrate normal and diseased development, understanding the role of the non-coding genome, developing novel single-cell omics techniques and analysis methods, and understanding the molecular mechanisms of longevity and cancer resistance in Naked mole rats. Additionally, in collaboration with Prof. Dr. Manfred Kayser (Department of Genetic Identification, Erasmus MC), he adapts single-cell omics

techniques and analysis methods for forensic applications. Eskeatnaf is also a coordinator of the genetics and genomics course at a graduate school and a lecturer in computational biology.

Dennis H. Murphree, **Ph.D.** is Scientific Director of the Digital Health & Artificial Intelligence Program in Dermatology at Mayo Clinic, and a Consultant with joint appointments in both the departments of Dermatology and AI&I (Artificial Intelligence and Informatics). Dr. Murphree's work focuses on building practically useful AI-enabled systems that directly improve patient care. Before joining Mayo Clinic, Dennis spent six years in quantitative finance where he was responsible for training, deploying, monitoring and updating a variety of AI-driven commodities and energy strategies, giving him significant experience and insight into real world use of predictive models. Prior to this he earned a BS in Physics (with Honors) from Stanford and a PhD in Physics from Yale, spending a year in between as a Fulbright Fellow. More recently Dennis was a finalist for best paper at the International Conference on Artificial Intelligence in Medicine 2020. Dennis' lifelong love of rock climbing, including climbing El Capitan in Yosemite National Park, has been tragically curtailed by a career-ending love of cheese.

Falk Nimmerjahn performed his undergraduate studies in Biology at the Universities of Bayreuth and Erlangen. He moved to Ludwig-Maximilians University and the Helmholtz-Center Munich for his PhD thesis where he focused on understanding the role of autophagy on antigen presentation and was awarded the prize for the best PhD thesis at the Helmholtz Centre in 2002. After a short postdoc period at the Helmholtz Centre Munich, he joined the laboratory of Jeff Ravetch at Rockefeller University in New York as a postdoc in 2004 and investigated how IgG antibodies mediate their effects in vivo. After being awarded a grant of the Bavarian Genome Network he returned to Germany in 2007 as an associate professor (W2) for Immunology and Immunotherapy at the Department of Medicine III (Rheumatology and Immunology) at the University Hospital Erlangen. Since 2010 Falk Nimmerjahn heads the Division of Genetics at the Department of Biology of the Friedrich-Alexander University in Erlangen as a full professor (W3) and has been the Speaker of the Department of Biology and currently is the Vice Dean for Research at the Faculty of Natural Sciences at FAU (since 2019). For his work he was awarded several prestigious prizes, including the Paul-Ehrlich and Ludwig Darmstaedter award for young scientist and the Research Award of the European Macrophage and Dendritic Cell Society (EMDS).

Nataša Pržulj is a Professor of Computational Biology at Mohamed bin Zayed University of Artificial Intelligence (MBZUAI) in Abu Dhabi. She has been a Full Professor of Computer Science at University College London since 2016. She is a leader in biological network analysis and a pioneer in network biology geometry applied to personalized (precision) medicine. She introduced graphlets to extract biomedical knowledge from molecular networks, revolutionizing the field. Graphlets are now widely built upon and utilized to produce feature vectors capturing network topology that are input into many AI algorithms for network data analytics in various domains. Her research has been supported by over €27 million in competitive research funding. Notably, she received three prestigious, single PI, European Research Council (ERC) grants: ERC Consolidator (2018-2025), ERC Proof of Concept (2020-2023) and ERC Starting (2012-2017). She is in the top 0.5% of the world's best scientists according to Stanford University's list of the World's Top 2% Scientists. She was elected into many academies, including: Vice-President of the International Academy of Artificial Intelligence Sciences (AAIS), in 2026; Fellow of the AAIS, in 2025; Fellow of the International Society for Computational Biology (ISCB), in 2024; Fellow of The European Laboratory for Learning and Intelligent Systems (ELLIS), in 2024; Fellow of the British Computer Society (BCS) Academy of Computing, in 2013; Academia Europaea, The Academy of Europe, in 2017; Scientific Advisor of the Mathematics Institute of the Serbian Academy of Sciences and Arts (SANU), in 2012; The Serbian Royal Academy of Scientists and Artists (SKANU), in 2019; etc. In 2014, she received the British Computer Society Roger Needham Award, sponsored by Microsoft Research, in recognition of the potential her research has to revolutionize health and pharmaceutics. She obtained a PhD in Computer Science from the University of Toronto.

Dr. Juan Antonio Retamero is an anatomical pathologist whose work has focused on the digital transformation of pathology and the integration of artificial intelligence into clinical diagnostics. He played a leading role in the implementation of a fully digital pathology workflow at Granada University Hospitals in Spain, one of the earliest large-scale examples of digital pathology adoption in routine practice. His experience spans digital pathology, computational diagnostics, and the deployment of AI technologies in healthcare. Currently serving as Medical Vice President, Pathology Operations for Diagnostic Products at Tempus, Dr. Retamero collaborates with healthcare institutions worldwide to accelerate the adoption of digital and AI-enabled diagnostic solutions. Through his clinical, scientific, and industry activities, he continues to contribute to the advancement of pathology as a data-driven discipline, supporting more accurate diagnoses and improved patient care.

Santiago Romero-Brufau is the Director of Artificial Intelligence for the Department of Otolaryngology at Mayo Clinic, where he leads the integration of advanced AI and machine learning into clinical practice. With a deep background in AI, medical informatics and systems engineering, his current work focuses on developing agentic LLM pipelines and predictive models to improve patient outcomes and operational efficiency. In addition to his clinical leadership, he is a dedicated educator, teaching courses on Deep Learning and AI at the Harvard T.H. Chan School of Public Health, where he also serves in the Executive Committee of the Health Data Science Program. His research often explores the practical implementation of AI in healthcare settings, bridging the gap between theoretical AI and real-world clinical application.

Dr.Parth Shah is the Director of Genome Informatics at Dartmouth Hitchcock Medical Center and a physician specializing in Bone Marrow Transplantation, Cellular Therapy, and Multiple Myeloma. He has led several transformative initiatives at Dartmouth, including spearheading the development of the Dartmouth Cloud, a fully integrated HIPAA-compliant environment seamlessly connected with conventional institutional data centers. In his role as Director of Genome Informatics, he has been instrumental in developing cloud infrastructure that supports both clinical and research operations. He has also developed and implemented a broad portfolio of advanced molecular assays, including somatic sequencing, germline sequencing, and cell-free DNA analysis, while designing and deploying large-scale distributed graph-based databases capable of supporting real-time genomic query and analytics at enterprise scale.

Heath D. Skinner, **MD**, **PhD** is Professor and Claude Worthington Benedum Chair of the Department of Radiation Oncology in the University of Pittsburgh School of Medicine, and Chief Medical Officer of the UPMC Hillman Cancer Center Radiation Oncology Network. He specializes in the study and treatment of head and neck cancer and is a board-certified radiation oncologist as well as a physician-scientist. His focus remains on providing excellent and compassionate patient care and seeking to integrate clinical trials into clinical practice to improve the lives of his patients.

Dr. Carmen Terzic has been a member of the Mayo Clinic staff since 1992 and was the Chair of the Department of Physical Medicine and Rehabilitation from 2011-2020. She holds the academic rank of Professor. She is co-Director of the Rehabilitation Medicine Research Center and she is also the Director of the Cardiovascular Rehabilitation Program, Mayo Clinic, Rochester, MN. Dr. Terzic earned her medical degree from Universidad Centro Occidental Lisandro Alvarado, in Barquisimeto, Venezuela and a PhD. from Mayo Graduate School, Mayo Clinic, Rochester, MN. She has more than 100 publications in the areas of regenerative medicine and stem cell-based cardiac repair, nuclear transport, cardiovascular rehabilitation, and disparities in health care. Her work has been published in leading journals, such as Science, Nature Cell Biology, Translational Medicine as well as in specialized journals, including AJPMR, Journal of Cardiopulmonary Rehabilitation and Prevention, Circulation, Circulation Research, Clinical Pharmacology and Therapeutics, Genome Biology and Stem Cells. She is active in a number of national and international specialty societies and is recognized globally for her groundbreaking research and contributions to clinical guidelines and practice in the area of cardiovascular prevention and rehabilitation. Dr. Terzic is a recipient of numerous awards – including the National Association of Academic Physiatrists (AAP) Distinguished Academician Award and more recently The Mayo Clinic Alumni Association Mid-Career Award for 2024. She is an editorial board member of journals including American Journal of Physical Medicine & Rehabilitation. Mayo Clinic Proceedings and Journal of Cardiopulmonary Rehabilitation and Prevention. Dr. Terzic was also a competitive fencer from the age of 8 and a member of the Team Venezuela during the years 1978-1992. She entered The Sport Hall of Fame in Lara, Venezuela in 2012.

Dr. Natalia Trayanova holds the Murray B. Sachs Professorship in the Department of Biomedical Engineering at Johns Hopkins University. She is also a Professor of Medicine at the Johns Hopkins School of Medicine, and a Professor of Applied Mathematics and Statistics. She directs the Alliance for Cardiovascular Diagnostic and Treatment Innovation, and is also the Director for AI Research in Health and Medicine in the Data Science and AI Institute. Trayanova has pioneered the development and use of patient heart digital twins of representing the functioning of the patient's organ. Using her first-of-their-kind personalized heart digital twins and deep learning approaches, Dr. Trayanova has developed new technologies for accurately predicting risk of cardiac arrest and for the precise delivery of catheter ablation therapies in patients with heart rhythm disorders. For her pioneering work in computational cardiology, she has received NIH Director's Pioneer Award. Her research output includes 450 published papers and book chapters. Dr Trayanova is the inventor on numerous patents and patent applications filed world-wide, and was named Fellow of the National Academy

of Inventors in 2020. Trayanova's work has received world-wide recognition, and she is the recipient of numerous honors and awards. In 2019, Dr. Trayanova was inducted in the Women of Technology International Hall of Fame, an honor conferred only on 5 women each year from around the world. She has received the Distinguished Scientist Award and the Zipes Distinguished Award from Heart Rhythm Society, and the Gordon Moe Award by the Cardiac Electrophysiology Society. This year, she is the recipient of the Hodgkin-Huxley-Katz Award by the Physiological Society and the James T. Willerson Award in Clinical Science by AHA. Trayanova has been named a Fellow of every American and European clinical cardiology society, testifying to her impact in clinical practice. She is also a Fellow of AIMBE, BMES, IAMBE, IUPS, and IEEE.

Louis Vaickus is a pathologist board-certified in anatomic pathology, cytopathology and clinical informatics. His scientific work focuses on the use of programmatic data analysis, machine learning and digital imaging to address "low hanging fruit" problems which are common to pathology and laboratory services. His current focus is urine cytopathology, advanced imaging techniques and the digital pathology transition.

Dr. Samuel Volchenboum, **Ph.D.**, is a professor of pediatrics and the associate chief research informatics officer for the Division of Biological Sciences at the University of Chicago. He is the Dean of Masters Programs, and he designed and launched the UChicago Master's in Biomedical Informatics. His clinical specialty is pediatric hematology / oncology, caring for kids with cancer and blood diseases. His research group includes the University of Chicago's Data for the Common Good (D4CG), dedicated to building communities, platforms, and ecosystems that maximize the potential of data to drive discovery and improve human health. D4CG's flagship project, the Pediatric Cancer Data Commons is dedicated to liberating and democratizing international data for pediatric malignancies. He is the director of the Informatics Core for the Clinical and Translational Science Award (CTSA), and he is director of the UChicago Clinical Informatics fellowship program.

Dr. Mark Waddle is an Associate Professor of Radiation Oncology at Mayo Clinic and a nationally recognized leader in the practical application of artificial intelligence in radiation oncology. He serves as Physician Chair of the Artificial Intelligence Task Force for the American Society for Radiation Oncology (ASTRO) and represents Radiation Oncology on the American Medical Association's (AMA) AI Specialty Society Collaborative. At Mayo Clinic, Dr. Waddle leads departmental initiatives integrating advanced machine learning, data-driven decision support, and workflow optimization into clinical cancer care. He is a key contributor to RadOncGPT and to the application of large language models to improve clinical efficiency, enable standardized data capture, and streamline documentation. Dr. Waddle plays a pivotal role in shaping policy, safety standards, and best practices for the responsible use of AI in medicine. He is a frequent invited speaker at national and international forums on artificial intelligence in oncology.

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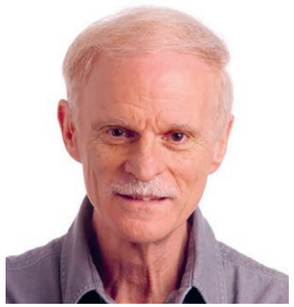
Prof. Šimun Anđelinović is a Croatian physician, forensic pathologist, and full professor at the University of Split. He currently serves as Head of the Clinical Department of Pathology, Forensic Medicine, and Cytology at the University Hospital of Split and previously served as Rector of the University of Split (2014-2018). His work has focused on pathology, forensic medicine, forensic genetics, and biological anthropology. He graduated from the School of Medicine at the University of Zagreb, where he also earned an MSc and a PhD in Medical Sciences. During his career, he advanced from assistant at the Institute of Pathology in Zagreb to full professor at the School of Medicine, University of Split. He completed professional training in pathology and forensic sciences at the University of Connecticut Health Center and the Connecticut State Police Forensic Science Laboratory in the United States. Professor Anđelinović is internationally recognized for pioneering work in DNA-based identification of war victims from mass graves during and after the Homeland War. He co-authored landmark forensic DNA publications and has published extensively in forensic genetics, pathology, molecular diagnostics, bioarchaeology, and forensic anthropology, including studies on osteometric sex estimation, archaeological populations, and forensic genomics.



Dr. Bruce Budowle received a PhD in Genetics in 1979 from Virginia Polytechnic Institute and State University. From 1979-1982, Dr. Budowle was a postdoctoral fellow at the University of Alabama at Birmingham. Working under a National Cancer Institute fellowship, he carried out research predominately on genetic risk factors for diseases such as insulin dependent diabetes mellitus, melanoma, and acute lymphocytic leukemia. From 1983-2009, Dr. Budowle worked at the FBI's Laboratory Division to carry out research, development, and validation of methods for forensic biological analyses. He has published more than 700 articles, made more than 800 presentations, and testified in well over 300 criminal cases in the areas of molecular biology, population genetics, statistics, quality assurance, and forensic biology. In addition, he has authored or co-authored books on molecular biology techniques, electrophoresis, protein detection, forensic genetics, and microbial forensics. Dr. Budowle recently retired as Director of the Center for Human Identification and Regents Professor at the University of North Texas Health Science Center at Fort Worth, Texas where his efforts focused on the areas of human forensic identification, microbial forensics, and emerging infectious disease with substantial emphasis in genomics and next generation sequencing. He continues to research and work in the areas of forensic genomics and contributes to supporting humanitarian efforts via human identification. He currently is a visiting professor in the Department of Forensic Medicine at the University of Helsinki and an adjunct professor in the Forensic Science Institute at Radford University.

John A. (Tony) Capra is the Harry Wm. and Diana V. Hind Distinguished Professor in the Department of Bioengineering and Therapeutic Sciences at the University of California, San Francisco. He is also Head of the Division of Bioinformatics in the Department of Epidemiology and Biostatistics, Associate Director for Discovery AI and Education in the Bakar Computational Health Sciences Institute, and the Co-Director of the UCSF Biological and Medical Informatics PhD program. His research group uses the tools of computer science and statistics to address problems in genetics, evolution, and biomedicine.

Dr. Mateja Hajdinjak (Dr. rer. nat.) is a Croatian molecular biologist using ancient DNA to study human evolutionary history. She is currently a Max Planck Research Group Leader of the Hominin Palaeogenomics group (HOPE) at the Max Planck Institute for Evolutionary Anthropology (MPI EVA) in Leipzig, Germany. She completed her PhD at the MPI EVA under the supervision of Dr. Matthias Meyer and Prof. Dr. Svante Pääbo, recovering and analysing genome-wide data of some of the last Neandertals and some of the earliest modern humans in Eurasia. As a Marie Skłodowska Curie Individual Fellow she then went to the Ancient Genomics Laboratory of Dr. Pontus Skoglund at the Francis Crick Institute in London, United Kingdom, where she continued working on human evolutionary genomics and tracing origins of modern human ancestry using ancient DNA.

Mitchell Holland, **Ph.D.**, is a Fellow in the American Academy of Forensic Sciences, has served as an associate professorial lecturer and adjunct faculty member at various colleges and universities. Dr. Holland has been on the Editorial Board of the Journal of Forensic Sciences and a member of the Advisory Board of the International Journal of Legal Medicine. Prior to being asked in early 2005 to help develop the Forensic Science Program at Penn State, Dr. Holland was the Senior Vice President of Operations and Laboratory Director of The Bode Technology Group. At Bode, Dr. Holland led the efforts to produce DNA profiles from victim remains recovered from Ground Zero (World Trade Centers) following the terrorist attacks of 9/11. His group is currently leveraging

the power of massively parallel sequencing (MPS) to measure rates of mtDNA heteroplasmy in different population groups; evaluate the transmission of heteroplasmic variants between maternal relatives and tissue types; assess the impact of damage on the interpretation of low-level heteroplasmic variants; and develop best practices for the application of MPS approaches in forensic casework. In addition, members of Dr. Holland's group are exploring ways to extract small fragments of DNA from highly degraded skeletal material for STR and SNP analysis on an MPS platform.

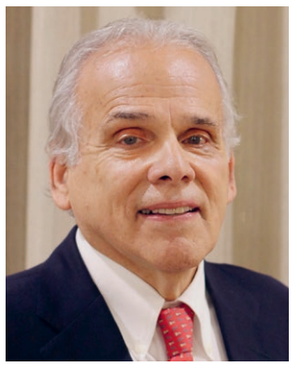
Prof. Manfred Kayser is a molecular biologist and geneticist with a broad interest in human variation. Since 2004, he is (full) Professor of Forensic Molecular Biology at Erasmus MC University Medical Center Rotterdam, where he chairs the Department of Genetic Identification. His scientific interest is in understanding human (epi)genetic and (epi) genomic variation and applying it to address forensic, anthropological, and medical questions. During the last 20 years, his ground-breaking fundamental and applied research has innovated the field of forensic genetics in several subfields such as forensic DNA phenotyping, forensic Y-chromosome analysis, forensic tissue identification, forensic time estimation and he additionally published on forensic epigenetics, forensic microbiome analysis, and forensic massively parallel sequencing. He published over 300 articles in peer-reviewed scientific journals. He is the most cited scientist worldwide in the field of forensic genetics and second most cited in forensic medicine 1960-2022. He received several research awards such as from the International Society for Forensic Genetics (ISFG). In 2023, he was elected member of the European Molecular Biology Organisation (EMBO).

Johannes Krause, **Ph.D.**, earned his Ph.D. in Genetics at Leipzig University. He was appointed junior professor for Paleogenetics at the University of Tübingen in 2010, and subsequently full professor for Archaeo- and Paleogenetics at the same university in 2013. In 2014, he became founding director of the Max Planck Institute for the Science of Human History in Jena, heading the Department of Archaeogenetics. In 2018 he became full professor at the Friedrich Schiller University Jena. He is one of the founding directors of the Max Planck-Harvard Research Center for the Archaeoscience of the Ancient Mediterranean (MHAAM), established in 2017. In 2020 he was reappointed to the Max Planck Institute for Evolutionary Anthropology and his department moved

to Leipzig. Prof. Dr. Krause focuses on the analysis of ancient DNA to investigate such topics as pathogens from historic and prehistoric epidemics, human genetic history and human evolution. He contributed substantially to deciphering the Neanderthal genome and the shared genetic heritage of Neanderthals and modern humans. In 2010, while working at the Max Planck Institute for Evolutionary Anthropology in Leipzig, he discovered the first genetic evidence of the Denisovans, an extinct hominin discovered in Siberia. His recent work includes revealing the genetic heritage of ancient Egyptians, reconstructing the first Pleistocene African genomes, uncovering the source of the epidemic plague bacteria that periodically caused historic and prehistoric epidemics in Europe, or clarifying the complex history of Europe's prehistoric mass migrations.

Ewelina Pośpiech holds an associate professorship at the Department of Genomics and Forensic Genetics at the Pomeranian Medical University in Szczecin, Poland. She completed her studies in medical biotechnology at the Jagiellonian University in Kraków and obtained her PhD in 2013 for research on the genetic prediction of human pigmentation traits. Continuing her work in the field of Forensic DNA Phenotyping, she has made significant contributions to the development of predictive genetic models for other externally visible characteristics (EVCs), including hair shape, baldness, and hair greying. Currently, her research focuses on the mechanisms of epigenetic aging and their practical applications in public health, precision medicine, and forensic science. She is actively engaged in the field of lifestyle epigenetics, with a particular emphasis on developing predictive models to support the design of health programs and guide interventions aimed at improving quality of life in aging populations, as well as enabling behavioral profiling in forensic contexts. In 2017, she received the Foundation for Polish Science Prize for outstanding young researchers. In recent years, she has also been honored with an award from the Polish Society of Forensic Medicine and Criminology, a prize from the Scientific Council of the Institute of Forensic Research in Kraków, and a distinction from the Polish Society of Genetics. For her research, she was also recognized by the Japan Society of Human Genetics. She has been recognized among the top 2% of the world's most cited scientists.

Prof. Antti Sajantila, **M.D.**, **Ph.D.**, works as a professor of forensic medicine in the Department of Forensic Medicine, at the University of Helsinki, and as a senior medical examiner in the Forensic Medicine Unit at the Finnish Institute of Health and Welfare. He is an Honorary Professor of Pontificia Universidad Católica del Perú in Lima. He received the ISFG Scientific Prize in 1997. Professor Sajantila is a member of the executive board of European Council of Legal Medicine and the Steering Committee of the Independent Forensic Expert Team hosted by the International Rehabilitation Council for Torture Victims. He has served in the Advisory Board for the United Nations Human Rights Special Rapporteur for the Minnesota Protocol on the Investigation of Potentially Unlawful Death. Professor Sajantila has participated in many international medico-legal investigations, implemented on the requests of various international communities. Professor Sajantila has published over 230 articles in peer-reviewed scientific journals and co-authored books in forensic genetics, pathology, and pharmacogenetics. His current research interests are ancient DNA, archaeovirology, forensic genetics and forensic pathology.



Dr. Ira R. Titunik has been in forensic dentistry approximately forty years. He is the Chief Forensic Dental Consultant, Bergen County Medical Examiner, New Jersey, Adjunct Professor, Henry C. Lee Institute of Forensic Science, University of New Haven, former Adjunct Professor, John Jay College of Forensic Science, New York, former Forensic Dental Consultant, Medicolegal Investigations Unit, New York State Police, Albany, NY, past President, USA Section, International College of Dentists.

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Exhibition Announcement and presentation of the monograph „Names in the Silence“

Šimun Anđelinović, Željana Bašić, Ivan Jerković, Ivana Kružić, Dragan Primorac

At the upcoming ISABS Conference, we are honored to present the exhibition and monograph “Names in the Silence”, a profound testimony to science, humanity, and international collaboration. The exhibition traces the first systematic applications of DNA technology in the identification of victims from mass graves discovered in Croatia and Bosnia and Herzegovina. This pioneering work – launched during the Homeland War in Croatia – emerged under extraordinary conditions and represents one of the earliest and most consequential integrations of modern DNA science into postconflict humanitarian and forensic practice. It was developed through an unprecedented collaboration between Dragan Primorac, Šimun Anđelinović, and leading forensic authorities from the United States, at a time when neither the institutional infrastructure nor the technological framework for such work yet existed in the region. Among the American partners were some of the most distinguished figures in global forensic science, including Henry C. Lee, Moses Schanfield, Michael Baden, Barbara Wolf, and Mitchell M. Holland. In parallel, close cooperation with Bruce Budowle and his team resulted in the creation of the first Croatian population DNA database, based on seven PCRbased genetic loci – an achievement that laid the scientific foundation for modern forensic identification and population genetics in Croatia and the wider region. The complex international collaboration was strategically coordinated by Dragan Primorac, who led and integrated the contributions of what came to be known as the “U.S. Forensic Dream Team,” ensuring the rapid transfer of cuttingedge molecular methods and ethical standards. At the national level, Šimun Anđelinović directed and coordinated all victim identification activities in Croatia, overseeing the operational and humanitarian dimensions of the work with unwavering commitment. Integral to this historic Croatian-American effort were Marija Definis, Jakša Ivanović, Irena Drmić, and Boja Režić, whose expertise and dedication formed the operational backbone of the project. Together, this multidisciplinary team transformed scientific innovation into acts of justice and human dignity – restoring names to the nameless, truth to families, and credibility to institutional processes in the aftermath of war. A defining moment in the global recognition of this work came with Barbara Wolf’s testimony before the U.S. Congress in 1995, which formally acknowledged its international scientific and humanitarian significance. The enduring contributions of those who are no longer with us – most notably Moses Schanfield, and more recently Henry C. Lee – remain permanently embedded in the foundations of contemporary forensic science. The enduring impact of Kupres on Henry C. Lee’s life and career is perhaps best symbolized thousands of kilometers away, in Nantong, China, within one of the largest forensic science museums in the world. There, an entire crime scene – faithfully reconstructed to the scale and reality of the Kupres mass graves – stands permanently on display, accessible to visitors from all continents. It is not presented as a spectacle, but as an education and a memorial: a place where science confronts humanity, and where the silence of mass graves is transformed into testimony. Through original documents, photographs, and personal testimonies, the exhibition reveals not only a scientific breakthrough but also a deeply human mission: the restoration of identity and dignity to those who had been lost in silence. A central visual component of the exhibition consists of photographs taken by Phil Farnsworth, whose work documented these events in real time and preserves an authentic record of the forensic and humanitarian efforts on the ground. Alongside the exhibition, the monograph „Names in the Silence“ will be formally presented and made available during the “14th ISABS and Mayo Clinic Conference: Advances in the Application of Artificial Intelligence in Precision Medicine.” The monograph brings together scientific, historical, and personal narratives documenting the creation of the Croatian model of DNA identification of war victims – one of the earliest such models globally – developed in collaboration with leading American experts and institutions. From the first laboratory steps undertaken in Clinical Hospital Split to international recognition, including acknowledgment in the United States, the monograph captures a unique intersection of science, war, and humanity. It honors all those who made this work possible. More than a scientific record, Names in the Silence is a testimony to people and to science – and to a world that came together to return names to the nameless.

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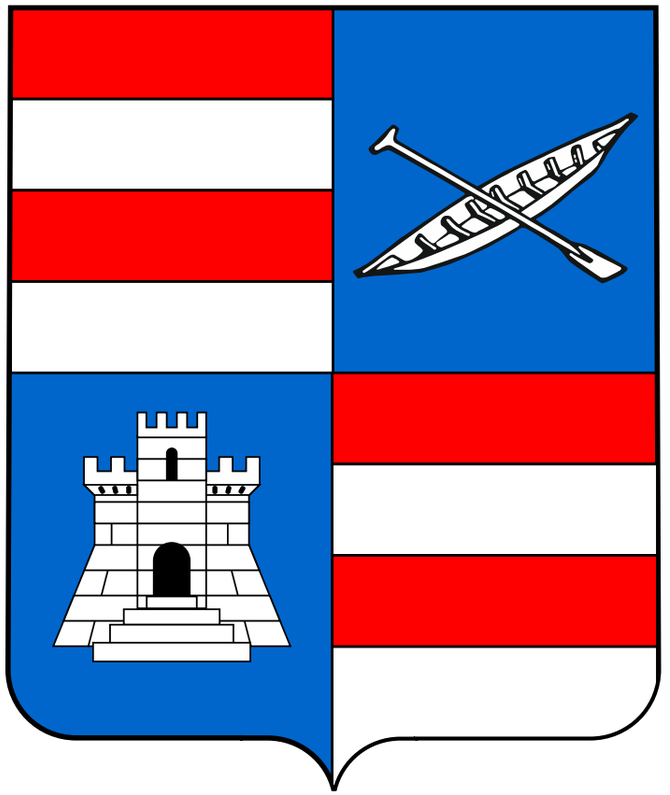

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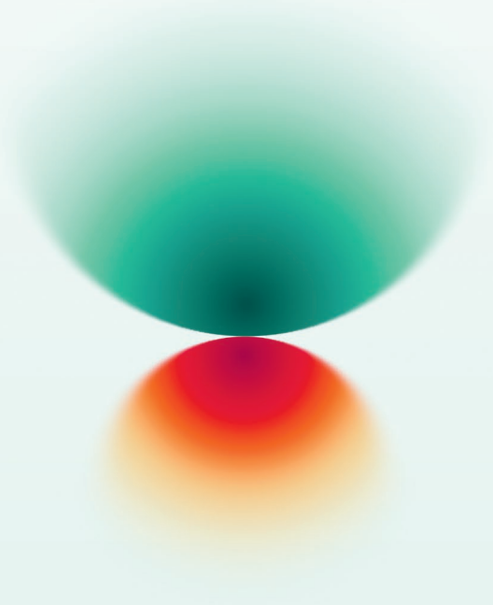

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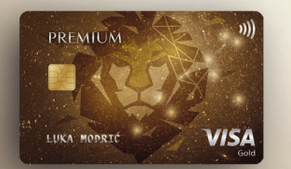

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