SRGM
Societatea Romana
de Genetica Medicala

George Emil Palade 50 de ani

UNIVERSITATEA DE MEDICIN
FARMACIE, ȘTIINȚE ȘI TEHN
„GEORGE EMIL PALADE"
DIN TÂRGU MUREȘ

A XIV-A CONFERINȚĂ DE GENETICĂ MEDICALĂ CU PARTICIPARE INTERNAȚIONALĂ

AGENDA

XIVth MEDICAL GENETICS

CONFERENCE WITH INTERNATIONAL PARTICIPATION

Târgu Mureș, 03-05 Octombrie 2024

COMITET DE ORGANIZARE / ORGANIZING COMMITTEE

Claudia Bănescu	Florin Tripon	Florin Burada	Ioana Streață
Alina Bogliș	Andrei Crauciuc	Beata Balla	Adriana Crișan
Camelia Chirteș	Anamaria Todoran-Butilă	Balazs Baczoni	Damiana Codați
Corina Gutman	Vasile Păcurar	Adriana Bota	Alexandrina Dragomir
Daniela Vacariu			

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COMITET ȘTIINȚIFIC / SCIENTIFIC COMMITTEE

Maria Puiu	Mircea Covic	Marius Bembea	Florin Burada
Vlad Gorduza	Cristina Rusu	Claudia Bănescu	Vasilica Plăiașu
Emilia Severin	Camelia Al-Khzouz	Radu Popp	Adrian Trifa
Claudia Jurcă	Nicoleta Andreescu	Mariela Militaru	Adela Chiriță-Emandi
Andreea Cătană	Ioana Streață	Elena Braha	Diter Atasie
Georgeta Cozaru	Teodora Barbarii	Katalin Csép	

JOI, 3 Octombrie / THURSDAY, October 3rd

10:00 – 14:00 – ÎNREGISTRAREA PARTICIPANȚILOR / REGISTRATION OF PARTICIPANTS

14:00 – 14:30 – DESCHIDEREA OFICIALĂ A EVENIMENTULUI / OFFICIAL OPENING OF THE EVENT - Oficialități ale Universității de Medicină, Farmacie, Științe și Tehnologie "George Emil Palade" Târgu Mureș și ale Societății Române de Genetică Medicală

14:30 – 16:15 – SESIUNE PLENARĂ / PLENARY SESSION

Moderatori / Moderators: Florin Burada, Claudia Bănescu

14:30 – 14:50 – Vlad Gorduza - Importance of genetic heterogeneity in practice of medical genetics - Craniosynostosis

14:50- 15:10 - Emilia Severin - Oro-dental anomalies in patients with rare monogenic disorders

15:10- 15:30 - Dorica Dan - Why do patients need to shape the rare diseases ecosystem?

15:30 – 15:45 - Simpozion BioMarin. Adela Chiriță-Emandi - The multidisciplinary management of patients with Morquio A syndrome

15:45 - 16:00 - Simpozion Antisel- Butnaru Cristian. Rethink sequencing - the affordable solution for your genetic testing is here.

16:00 - 16:15 Simpozion BIOgenetiX - Florina Raicu- The 4th generation sequencing-from applications in research to clinical diagnosis

16:15– 17:00 PAUZĂ DE CAFEA / COFFEE BREAK

17:00 – 18:40 – SESIUNE PLENARĂ / PLENARY SESSION

Moderatori / Moderators: Maria Puiu, Bogdan Mirăuță

17:00 – 17:20 - Michael Baudis **-** Data discovery in biomedical genomics - time for a new paradigm

17:20 – 17:40 - Maria Puiu **-** European policies and regulations in rare diseases and genetics

17:40 – 18:00 - Bogdan Mirăuță **-** Romania Joins ELIXIR- next steps and impact on genomics research

18:00 – 18:20 - Simpozion ELTA90 MR. Dumitru Jardan **–** TruSight oncology 500 – a comprehensive genomic profiling test – what it can do for you?

18:20 – 18:40 - Simpozion Medist. Raymond Gochuico **–** Revolution in liquid handling - Opentrons Flex

19.00- 23.00 - CINA / DINNER - Restaurant MAZA (Mihai Viteazul street, no 60, Târgu Mureș 540098)

VINERI, 4 Octombrie / FRIDAY, October 4th

9:00 – 11:00 – SESIUNE PLENARĂ / PLENARY SESSION

Moderatori / Moderators: Cristina Rusu, Claudia Jurcă

9:00 – 9:20 - Cristina Rusu- Classic and modern in RASopathies

9:20 – 9:40 - Vasilica Plăiașu - Long experience in solving rare and undiagnosed diseases – what have we learnt? what about those left behind?

9:40 – 9:50 - Elena Emanuela Braha - Co-occurrence of two genetic diseases - a clinical challenge for the medical team: four unrelated cases

9:50 – 10:00 - Codruța Diana Petcheși - Double trouble: Beckwith-Wiedemann syndrome associated with familial Long QT syndrome type I

10:00 - 10:10 - Florin Tripon - Genetic counseling - challenges, opportunities and perspectives

10:10 - 10:20 - Răzvan Pleșea - Clinically significant germline variants in hereditary cancers in south-west Romania - a preliminary study

10:20-11:00- Simpozion Genesis Pharma. "New insights in the diagnosis and management of rare diseases" - ***Moderator Maria Puiu***

10:20-10:30 ***Maria Puiu*** - siRNA therapy in rare diseases' management – mechanism of action

10:30-10:45 ***Claudia Bănescu*** - The importance of early diagnosis in acute hepatic porphyria

10:45-11:00 ***Carmen Muntean***- When kidney stones may be a sign of primary hyperoxaluria type 1

11:00 – 11:30 PAUZĂ DE CAFEA / COFFEE BREAK

11:30 - 14:00 SESIUNE PLENARĂ / PLENARY SESSION

Moderatori / Moderators: Emilia Severin, Nicoleta Andreescu

11:30 - 11:50 - Marius Bembea - Applications of genomics in population genetics

11:50 - 12:10 - Richard Constantinescu - Heredity and genetics in Romanian Medicine: a journey from Cantemir to DNA

12:10 – 12:30 - Adela Chiriță-Emandi - Landscape of disease causing variants in Romanian children with inborn errors of immunity

12:30 – 12:50 - Claudia Jurcă - Wolfram Syndrome: A journey from gene to phenotype

12:50 – 13:00 - Mariana Sprincean - Autism spectrum disorders in Kleefstra syndrome: clinical case report

13:00 – 13:10 - Magdalena Budișteanu - Autism spectrum disorder in males with sex cchromosome aaneuploidy

13:10 – 13:20 - Elena Neagu - Neonatal screening and presymptomatic intervention in spinal muscular atrophy, genetic disorder with multisystemic involvement

13:20 – 13:30 - Carmen Muntean - DICER1 syndrome - a surprise diagnosis starting from cystic nephroma

13:30 – 13:45 - Simpozion Roche. Andreea Cătana - The role of comprehensive genomic profiling in personalized medicine

13:45 – 14:05 - Simposion Genesis. "New insights in the diagnosis and management of rare diseases"- Sesiunea 2. ***Moderator Florin Burada***

13:45-13:50 Florin Burada - Introduction

13:50-14:05 Ioana Streață - Chaperone therapy in the management of Fabry disease

14:05 – 15:30 PAUZĂ DE PRÂNZ / LUNCH BREAK (Microcantină HESTIA, UMFST G.E. Palade Târgu Mureș) & VIZITARE STANDURI SPONSORI

15:30 – 17:15– SESIUNE PLENARĂ / PLENARY SESSION

Moderatori / Moderators: Vlad Gorduza, Adela Chiriță-Emandi

15:30 - 15:50 – Simpozion Medist. Chris Sale- Expanding the exome: unveiling disease in non-coding regions with Nexome

15:50 - 16:00 - Vanesa Larisa Bloaje Florică - Prenatal identification of chromosomal abnormalities: karyotyping in various tissue samples - Participant competiție tineri cercetători / Young researchers competition participant

16:00 - 16:10 - Teodora Barbarii - Caveats in rare disease diagnostics: alternative transcripts and alternative haplotypes - Participant competiție tineri cercetători / Young researchers competition participant

16:10 - 16:20 - Alexandru Caramizaru - Genetic testing in Romania through an international collaboration – Expanding the accessibility to whole exome sequencing- Participant competiție tineri cercetători / Young researchers competition participant

16:20 - 16:35 - Simpozion Takeda. Irena Nedelea - Integrated management of patients with hereditary angioedema

16:35 – 17:20 - Simpozion AstraZeneca. Moderator Maria Puiu

16:35 - 16:50 **Adela Chiriță-Emandi** - Challenges in the diagnosis and management of patients with hypophosphatasia (HPP)

16:50 - 17:05 **Ioana Streață** - Lysosomal acid lipase deficiency (LAL-D): from theory to practice.

17:05 - 17:20 - **Q&A.** ***Moderator Maria Puiu***

17:20 – 17:45 – PAUZĂ DE CAFEA / COFFEE BREAK

17:45-18:05 - RAPORT ACTIVITATE / ACTIVITY REPORT

Moderatori / Moderators: Florin Burada, Claudia Bănescu

17:45 - 17:55- Florin Burada - Romanian Society of Medical Genetics- annual activity report

17:55-18:05 - Teodora Barbarii - Romanian Young Geneticists activity report for 2022-2024

18:05- 18:30- Adunare generală membrii SRGM / SRGM Members' Assembly

19:00 -23:00 – CINA / DINNER - RESTAURANT HOTEL PLAZA (Piața Trandafirilor street, no. 46-47, Târgu Mureș 540053)

SÂMBĂTĂ, 5 Octombrie / SATURDAY, October 5th

9:00 – 11:05 – SESIUNE PLENARĂ / PLENARY SESSION

Moderatori / Moderators: Ioana Streață, Horia Stănescu

9:00 – 9:15 - Claudia Bănescu - Genomic landscape of myelodysplastic syndrome and its clinical implication

9:15 – 9:35 - Horia Stănescu - An interesting renal phenotype (Distal Renal Tubular Acidosis) - and a mysterious gene (*WDR72*)

9:35 - 9:50 - Andreea Cătană - Genetic predisposition assessment in Hereditary Breast Cancer – IOCN Cluj-RPS Regina Maria Collaborative Study

9:50 - 10:05 - Ioana Streață - Genetic testing strategy in epilepsy

10:05 - 10:20 - Lavinia Caba - How important are genetics and epigenetics in Ulcerative colitis

10:20 - 10:35 - Simpozion CHIESI. Maria Puiu - Management of alpha-mannosidosis

10:35 - 10:50 - Simpozion AstraZeneca. Claudia Bănescu - Management of plexiform neurofibromas in NF1

10:50 - 11:05 - Simpozion Medison Pharma. Andrei Teodorescu - Applied Genetics in practice: Products for the Future of Medicine

11:05– 12:10 - PAUZĂ DE CAFEA & PREZENTARE POSTERE / COFFEE BREAK & POSTER PRESENTATIONS

12:10- 14:00 SESIUNE PLENARĂ / PLENARY SESSION

Moderatori / Moderators: Vlad Gorduza, Mariela Sanda Militaru

12:10– 12:25 - Florin Burada - Discordant non-invasive prenatal testing- a case series

12:25-12:40 - Mariela Sanda Militaru - The importance of genetic testing in establishing the optimal treatment method for infertile couples

12:40 - 12:50 - Loredana Valentina Nemțanu – The role of genetic testing for In Vitro Fertilization

12:50 - 13:00 - Ina Ofelia Focșa - Preimplantation Genetic Testing: A chance for life

13:00 - 13:15 - Simpozion Sanofi. Maria Puiu - Olipudase alfa (Xenpozyme) the first and only treatment available for patients with Niemann-Pick disease, type A/B and B.

13:15 - 13:30 - Simpozion Dexter. Alex Kurze and Morgan Thenoz. Accelerating NGS analysis.

13:30 - 13:45 - Simpozion CeGaT. Gözdenur Schmelzer- How ExomeXtra® Solves Patient Cases?

13:45 - 14:00 - Simpozion Molecular Genomics. Miloš Šakan - Beyond the Needle in the Haystack: Unlocking Rare Disease Identification Using Hi-Fi Sequencing in Dark Genomic Regions

14:00 – 15:00 - PAUZĂ DE PRÂNZ / LUNCH BREAK (Microcantina HESTIA, UMFST G.E. Palade Târgu Mureș)

15:00 - 16:00 SESIUNE PLENARĂ / PLENARY SESSION

Moderatori / Moderators: Andreea Cătană, Katalin Csep

15:00 – 15:10 - Anca Costache (Riza) - Exome analysis pitfalls and perspectives

15:10 - 15:20 - Alina Bogliș - Maternally inherited diabetes and deafness syndrome: the applicability of the MLPA method in early diagnosis

15:20 - 15:30 - Mihai Cucu - MLPA genetic testing: available kits and positive cases, an update at CRGM Dolj

15:30 - 15:40 - Beata Balla - The use of MLPA technique in genetic investigation of Romanian CLL patients

15:40 - 15:50 - Adriana-Stela Crișan - Testing options for target and fast investigation of myeloproliferative neoplasms

ÎNCHIDEREA OFICIALĂ A CONFERINȚEI ȘI CEREMONIA DE PREMIERE / OFFICIAL CLOSING OF THE CONFERENCE & AWARDING CEREMONY

SESIUNE POSTERE / POSTER SESSION

Moderatori / Moderators: Claudia Bănescu, Alina Bogliș, Florin Tripon

1. Andreea Ardeleanu - Von Hippel-Lindau syndrome – multidisciplinary assessment. Case report

2. Anca Florentina Mitroi - Rare deletions and duplications of chromosome 4 - case reports and literature review

3. Claudia Ștefania Bala - Genetics roles in finding personalized treatment in oncology

4. Stela Racoviță - Cytogenetic evaluation in male infertility with azoospermia

5. Adelina Glangher - PPP2 syndrome type R5D- clinical variability- 2 cases

6. Vanesa Larisa Bloaje Florică - To know or not to know? – prevalence of carrier status for cystic fibrosis and non-syndromic hearing loss in a romanian adult population

7. Vanesa Larisa Bloaje Florică - Comprehensive therapies for inherited metabolic diseases: molecular mechanisms and innovations in treatment

8. Miruna Gug - De novo deletion *RAI1* gene - first case report of Smith–Magenis syndrome in Romania

9. Cristina-Loredana Pantea - Congenital neutropenia due to JAGN1 deficiency in roma ethnicity

10. Cristina-Adriana Buzlea-Zaha - What is wrong with *POLG* gene? Quick view upon myocerebrohepathopathy

11. Mihaela Ștefania Buzdugan - X linked intellectual disability associated with the *CASK* gene

12. Enikő Kutasi - The role of telomere biology in early aging: how an extra copy of chromosome 21 contributes to early-onset Alzheimer's disease

13. Monica-Cristina Panzaru - Chromosomal anomalies and disorders of the corpus callosum

14. Ramona Vulturar - Unraveling the spectrum: cystinuria and the contiguous gene deletion syndrome hypotonia-cystinuria syndrome – a journey from cases to pathogenesis

15. Florina-Victoria Nazarie - Decoding the complexity: from RASopathy genes to clinical implications in Noonan and Noonan-like syndromes

16. ***Bogdan Borzei*** **-** Genetic links: uncovering the hidden connection between telomeres and metabolism

17. Elena Maria Pîrlici - Axonal guardians: neurofilaments as pioneering biomarkers in cell biology and neuromuscular diseases

18. Darius Lotrean **-** Generation and use of synthetic data using AI, machine learning and artificial neural networks in rare genetic disease research: explorative study on ankylosing spondylitis patients from Romania

19. Camelia-Maria Chirteș **-** Renal MicroRNA as possible biomarkers and therapeutic targets in numerous kidney cystic diseases

20. Katalin Csép **-** Bioelectrical impedance analysis of body composition in normal and over-weight/ obese parents and their adult offspring

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CeGaT

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Dialab Solutions

Genekor

Molecular Genomics

synevo

WATT

ACCELA

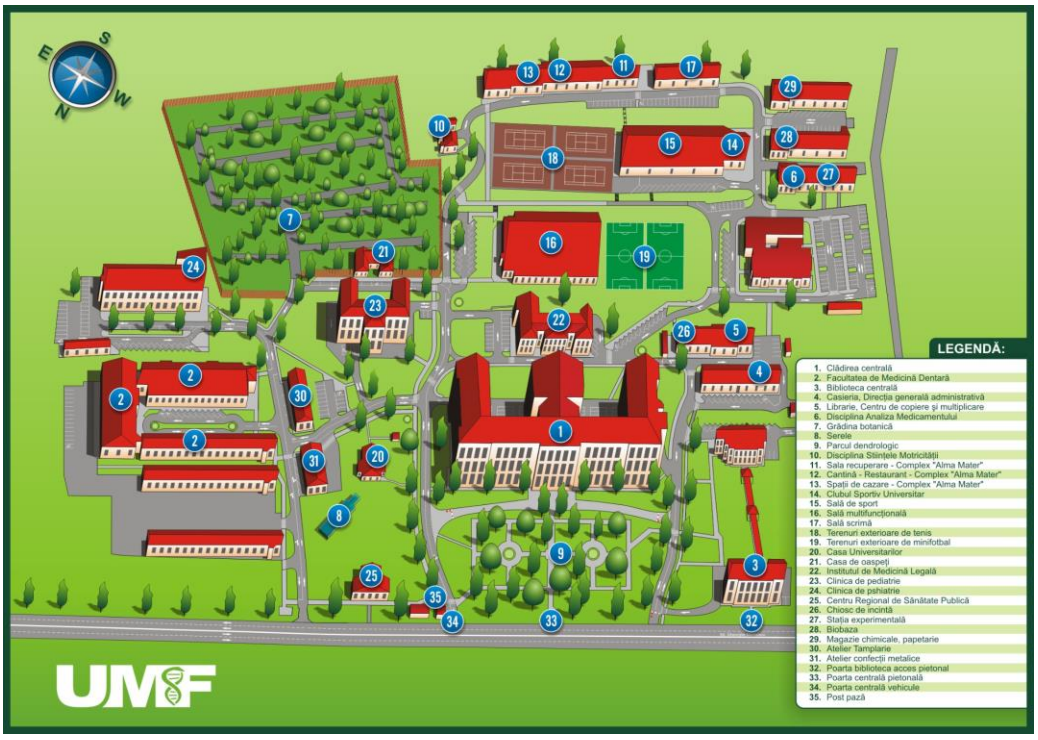
ZEISS

LOCAȚII / LOCATIONS

CAMPUSUL UMFST G. E. PALADE / UMFST G. E. PALADE CAMPUS

(1) CLĂDIREA PRINCIPALĂ ETAJ II: SALA FESTIVĂ - SALA SESIUNILOR PLENARE; STANDURI SPONSORI; PAUZĂ DE CAFEA - DISCIPLINA DE GENETICĂ /
MAIN BUILDING, SECOND FLOOR: FESTIVE HALL - PLENARY SESSIONS ROOM SPONSORS BOOTHS; COFFEE BREAKS - GENETICS DISCIPLINE

(12-13) PAUZĂ DE PRÂNZ - MICROCANTINA HESTIA (FOSTA ALMA MATER) - CAZARE HYPNOS / LUNCH BREAK - HESTIA MICROCANTEEN (former Alma Mater) - ACCOMODATION HYPNOS.



RESTAURANT MAZA	***RESTAURANT HOTEL PLAZA***	***HOTEL CONTINENTAL***
Mihai Viteazul street, no. 60 Târgu Mureș	*Piața Trandafirilor 46-47, Târgu Mureș 540053*	*Piața Teatrului 5, Târgu Mureș 540046*