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

JOI, 3 Octombrie / Thursday

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 UMFST George Emil Palade și ale societății SRGM /

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

Moderatori: Burada Florin, Bănescu Claudia

14.30 – 14.50 – Gorduza Vlad - Importance of genetic heterogeneity in practice of medical genetics - Craniosynostosis

14.50- 15.10 - Severin Emilia - 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. Chiriță Emandi Adela- The multidisciplinary management of patients with Morquio A syndrome

15.45 - 16.00 - Simpozion Antisel- Rethink sequencing - the affordable solution for your genetic testing is here.

16.00-16.15 Simpozion BIOgenetiX- The 4th generation sequencing-from applications in research to clinical diagnosis

16:15– 17.00 PAUZĂ DE CAFEA / COFFEE BREAK

17:00 – 19:15 – SESIUNE PLENARĂ / PLENARY SESSION

Moderatori: Puiu Maria, Mirăuță Bogdan

17:20 – 17:40 - Michael Baudis - Data discovery in biomedical genomics - time for a new paradigm

17:40 – 18:00 - Puiu Maria - European policies and regulations in rare diseases and genetics

18:00 – 18:20 - Mirăuță Bogdan - Romania Joins ELIXIR- next steps and impact on genomics research

18:20 – 18:40 - Simpozion ELTA90 MR.

18:40 – 19.00 - Simpozion Medist.

19.30- 23.00 - CINA- DINNER - Restaurant MAZA (Strada Mihai Viteazul 60, Târgu Mureș 540098)

VINERI, 4 Octombrie / Friday

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

Moderatori: Rusu Cristina, Jurcă Claudia

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

9.20 – 9:40 - Plăiașu Vasilica- Long experience in solving rare and undiagnosed diseases – what have we learnt? what about those left behind?

9:40 – 9.50 - Braha Elena Emanuela - Co-occurrence of two genetic diseases - a clinical challenge for the medical team: four unrelated cases

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

10.00- 10.10 - Tripon Florin - Genetic counseling - challenges, opportunities and perspectives

10.10- 10.20 - Pleșea Răzvan - 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 Puiu Maria**

10.20-10.30 ***Puiu Maria*** - siRNA therapy in Rare Diseases' management – Mechanism of Action

10.30-10.45 ***Bănescu Claudia***- The importance of early diagnosis in Acute Hepatic Porphyria

10.45-11.00 ***Muntean Carmen***- 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: Bembea Marius, Severin Emilia

11.30- 11.50 - Bembea Marius - Applications of genomics in population genetics

11.50- 12.10 - Constantinescu Richard - Heredity and Genetics in Romanian Medicine: A Journey from Cantemir to DNA

12.10 – 12.30 - Chirița Emandi Adela - Landscape of disease causing variants in Romanian children with inborn errors of immunity

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

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

13:00 – 13:10 - Budișteanu Magdalena - Autism Spectrum Disorder in Males with Sex Chromosome Aneuploidy

13.10 – 13.20 - Neagu Elena - Neonatal screening and presymptomatic intervention in spinal muscular atrophy, genetic disorder with multisystemic involvement

13.20 – 13.30 - Muntean Carmen - DICER1 syndrome - a surprise diagnosis starting from cystic nephroma

13.30 – 13.45 - Simpozion Roche. Cătana Andreea - 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 Burada Florin***

13.45-13.50 Burada Florin- Introduction

13.50-14.05 Streață Ioana- Chaperone therapy in the management of Fabry disease

14:05 – 15:00 PAUZĂ DE PRÂNZ / LUNCH BREAK (Microcantina HESTIA, UMFST G.E. Palade)

15:00 – 17:00 – SESIUNE PLENARĂ / PLENARY SESSION

Moderatori: Gorduza Vlad, Chiriță Emandi Adela

15.00-15.10 - Vanesa Larisa Bloaje Florică - Prenatal identification of chromosomal abnormalities: karyotyping in various tissue samples - Participant competiție Tineri cercetători

15.10- 15.20 - Barbarii Teodora - Caveats in rare disease diagnostics: alternative transcripts and alternative haplotypes - Participant competiție Tineri cercetători

15.20-15.30 - Caramizaru Alexandru- Genetic testing in Romania through an international collaboration – Expanding the accessibility to whole exome sequencing- Participant competiție Tineri cercetători

15.30- 15.50 - Simpozion Medist.

15.50- 16.05 - Simpozion Takeda. **Nedelea Florina**- Integrated management of patients with hereditary angioedema

16:05 – 16:50 - Simpozion AstraZeneca. Moderator Puiu Maria

16:05 - 16:25 **Chiriță Emandi Adela**- Challenges in the diagnosis and management of patients with hypophosphatasia (HPP)

16:25 - 16:45 **Streață Ioana**- Lysosomal acid lipase deficiency (LAL-D): from theory to practice.

16:45 - 16:50 - **Q&A**

16.50 – 17.30 – PAUZĂ DE CAFEA / COFFEE BREAK

17.30-17.40- Burada Florin- Romanian Society of Medical Genetics- annual activity report

17.40-17.50 - Teodora Barbarii- Romanian young geneticists activity report for 2022-2024

17.50- 18.15- Adunare generală membrii SRGM

19.00 -23.00 – CINA / DINNER - RESTAURANT HOTEL PLAZA (Piața Trandafirilor 46-47, Târgu Mureș 540053)

SÂMBĂTĂ, 5 Octombrie / Saturday

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

Moderatori: Streață Ioana, Stănescu Horia

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

9.15 – 9.35- Stanescu Horia - An interesting renal phenotype (Distal Renal Tubular Acidosis) - and a mysterious gene (WDR72)

9.35- 9.50 - Cătana Andreea - Genetic Predisposition Assessment in Hereditary Breast Cancer – IOCN Cluj-RPS Regina Maria Collaborative Study

9.50-10.05 - Streață Ioana- Genetic testing strategy in epilepsy

10.05-10.20-Caba Lavinia - How important are genetics and epigenetics in Ulcerative colitis

10.20-10.35- Simpozion CHIESI. Puiu Maria- Management of alpha-mannosidosis

10.35-10.50- Simpozion AstraZeneca. Bănescu Claudia- Management of plexiform neurofibromas in NF1

10.50-11.05- Simpozion Medison Pharma.

11:05– 12.10 PAUZĂ DE CAFEA & PREZENTARE POSTERE / COFFEE BREAK & POSTER PRESENTATIONS

12.10- 14.00 SESIUNE PLENARĂ / PLENARY SESSION

Moderatori Gorduza Vlad, Militaru Mariela Sanda

12.10– 12.25 - Burada Florin - Discordant non-invasive prenatal testing- a case series

12.25-12.40 - Militaru Mariela Sanda - The importance of genetic testing in establishing the optimal treatment method for infertile couples

12.40-12.50 - Nemțanu Loredana Valentina – The role of genetic testing for In Vitro Fertilization

12.50- 13.00 - Focșa Ina Ofelia - Preimplantation Genetic Testing: A chance for life

13.00-13.15 - Simpozion Sanofi.

13.15 -13.30 - Simpozion Dexter.

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)

15.00- 16.00 SESIUNE PLENARĂ / PLENARY SESSION

Moderatori Cătană Andreea, Andreescu Nicoleta

15.00 – 15.00 - Costache (Riza) Anca- Exome analysis pitfalls and perspectives

15.10- 15.20 - Bogliș Alina- Maternally inherited diabetes and deafness syndrome: the applicability of the MLPA method in early diagnosis

15.20- 15.30 - Cucu Mihai- MLPA genetic testing: available kits and positive cases, an update at CRGM Dolj

15.30- 15.40 - Balla Beata- The use of MLPA technique in genetic investigation of Romanian CLL patients

15.40- 15.50 - Crișan Adriana- 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

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

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

5. Glangher Adelina- 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. Gug Miruna- De novo deletion RAI1 gene - first case report of Smith–Magenis syndrome in Romania

9. Pantea Cristina-Loredana - Congenital neutropenia due to jagn1 deficiency in roma ethnicity

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

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

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

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

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

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

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

17. ***Borzei Bogdan*****-** Genetic Links: Uncovering the Hidden Connection between Telomeres and Metabolism

18. Elena Maria Pîrlici - Axonal Guardians: Neurofilaments as Pioneering Biomarkers in Cell Biology and Neuromuscular Diseases

19. Lotrean Darius - 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

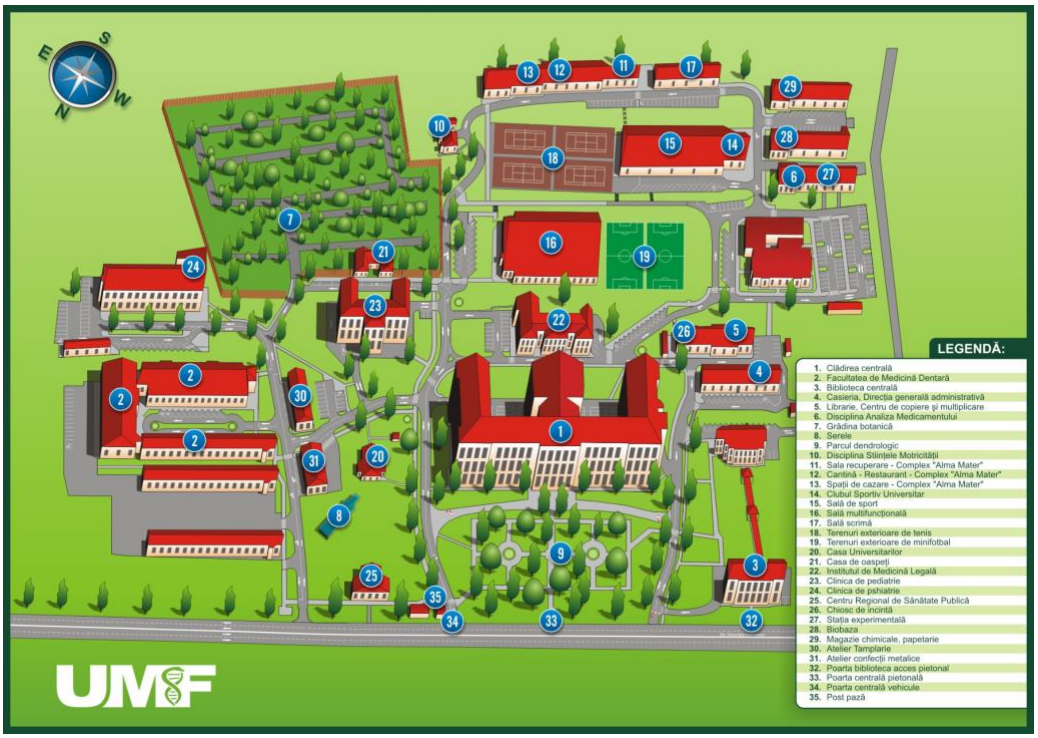
20. Chirteș Camelia-Maria-Renal MicroRNA as possible biomarkers and therapeutic targets in numerous kidney cystic diseases

LOCAȚII

CAMPUSUL UMFST G. E. PALADE

(1) CLADIREA PRINCIPALĂ ETAJ II: SALA FESTIVĂ- SALA SESIUNILOR PLENARE; STANDURI SPONSORI; PAUZĂ DE CAFEA- DISCIPLINA DE GENETICĂ

(12-13) PAUZĂ DE PRÂNZ- MICROCANTINA HESTIA (FOSTA ALMA MATER) CAZARE HESTIA



RESTAURANT MAZA	*RESTAURANT HOTEL PLAZA*	*HOTEL CONTINENTAL*
Str. Mihai Viteazul nr. 60 Târgu Mureș	*Piața Trandafirilor 46-47, Târgu Mureș 540053*	*Piața Teatrului 5, Târgu Mureș 540046*