THE 13th MEDICAL GENETICS CONFERENCE WITH INTERNATIONAL PARTICIPATION

TIMIȘOARA, 28th – 30th SEPTEMBER 2023

AGENDA

SRGM
Societatea Romana
de Genetica Medicala

A XIII - A CONFERINȚĂ DE GENETICĂ MEDICALĂ CU PARTICIPARE INTERNAȚIONALĂ

Timișoara, 28 - 30 SEPTEMBRIE 2023

ORGANIZATORI

SRGM
Societatea Romana
de Genetica Medicala

UNIVERSITATEA
DE MEDICINĂ ȘI FARMACIE
„VICTOR BABEȘ“ DIN TIMIȘOARA

PARTENER

Alianța Națională
pentru Boli Rare ROMÂNIA

THURSDAY, 28th SEPTEMBER 2023

09:00–10:30 – PARTICIPANTS REGISTRATION

10:30–11:00 – OFFICIAL OPENING OF THE CONFERENCE
Representatives of MoH, UMFT, SRGM, local administration

KEYNOTES SPEAKERS

11:00–11:40 - *From rare diseases to Public Health Genomics* – Borut PETERLIN
11:40–12:20 - *Diagnosis and follow-up of children and adolescents with neurofibromatosis type 1* – Eric LEGIUS

12:20–12:35 - *Increasing life expectancy for patients with Niemann Pick disease* - Maria PUIU – SANOFI Symposium
12:35–12:50 - *Phenotypic spectrum of lysosomal acid lipase deficiency and experience with enzyme replacement therapy* – Adela CHIRIȚĂ-EMANDI –ASTRAZENECA Symposium
12:50–13:05 – *Management of patients with hypophosphatasia* – Adela CHIRIȚĂ-EMANDI – ASTRAZENECA Symposium

13:05–14:00 – LUNCH

14:00–16:10 – PLENARY SESSION - CARDIOGENETICS
Chairmen: Dragoș COZMA, Adela CHIRIȚĂ-EMANDI

14:00–14:30 - *Cardiac Genetic Testing from an UK perspective* – Claire HOPTON
14:30–14:40 - *Genetic testing in cardiomyopathies - when and how?* – Ruxandra JURCUȚ
14:40–14:50 - *Diagnosis in arrhythmogenic heart disease: from the first cases described until today* – Dragoș COZMA
14:50–15:00 - *Monogenic causes of dilated cardiomyopathy in Romanian adults* – Adela CHIRIȚĂ-EMANDI
15:00–15:10 - *Genetics of cardiomyopathy: advances and pitfalls of molecular diagnosis* – Anca-Lelia RIZA
15:10-16:10 - Innovations in the diagnosis and management of rare diseases - GENESIS PHARMA Symposium
Moderator: Maria PUIU
***15:10–15:20 - siRNA therapy in Rare Diseases* - Maria PUIU**
***15:20–15:30 - Fabry disease - Experience of the Expertise Center for Rare Genetic Cardiovascular Diseases in Bucharest* - Ruxandra JURCUȚ**
***15:30–15:40 - Discovering Acute Hepatic Porphyria* - Mirela DĂNILĂ**
***15:40–15:50 - A New Perspective in the Management of hATTR Amyloidosis* - Diana-Luisa LIGHEZAN**
15:50–16:00 - *New Scientific Evidence in hATTR Amyloidosis* - Robert ADAM
***16:00–16:10 - When nephrolithiasis can be a sign of primary hyperoxaluria type 1* - Adela CHIRIȚĂ-EMANDI**

16:10 – 16:25 – COFFEE BREAK. POSTERS & EXHIBITION VIEWING

16:25–18:15 - PLENARY SESSION - ONCOGENETICS
Chairmen: Șerban NEGRU, Adrian TRIFA

16:25–16:40 - *The essential role of genetics in the practice of clinical oncology* – Șerban NEGRU
16:40–16:50 – *Germline molecular testing in the management of oncological patients* – Andreea CĂTANĂ
16:50–17:00 – *The role of comprehensive somatic tests in the management of oncological patients* – Adrian TRIFA
17:00–17:10 - *Oncofertility in patients with germline pathogenic variants predisposing to breast and ovarian cancer* – Cristina DAMIAN
17:10–17:20 - *Where dMMR Cases Continue Their Diagnostic, Therapeutic, and Screening Journey* – Oana VOINEA
17:20–17:30 - *Updates in risk-stratification in acute myeloid leukemia* – Claudia BĂNESCU
17:30–17:40 - *Lynch syndrome – the experience of Filantropia Hospital, Bucharest* – Florina NEDELEA
17:40–17:50 - *The road between Germline and Somatic variants. An CRGM Dolj update in oncogenetics testing and counseling* – Răzvan PLEȘEA
17:50–18:15 – *Chromosomal Microarray Analysis applications using Cytoscan* – Monique OLIMA – ANTISEL Symposium

18:15–19:30 - PLENARY SESSION - BIOINFORMATICS
Chairmen: Anca-Lelia RIZA, Cristian ZIMBRU

18:15–18:45 - *Long-read genomic technologies for rare disease research* – Bart van der SANDEN
18:45–19:00 – *Single-cell RNA sequencing for clinical applications* – Cristian ZIMBRU
19:00–19:10 – *Data sharing and mining - European initiatives* – Alexandru PRICĂ
19:10–19:30 – *The impact of early diagnosis on the quality of life of patients with alpha mannosidosis* – Ioana STREAȚĂ - CHIESI Symposium

20:30 – DINNER - CURTEA BERARILOR

FRIDAY, 29th SEPTEMBER 2023

09:00–10:45 – PLENARY SESSION - GENOMICS AND PERSONALIZED MEDICINE
Chairmen: Claudia BĂNESCU, Horia STĂNESCU

09:00–09:30 – ***Genomics for Medicine: Practical clinical application for cancer and rare disease*** **– Michel HUNBANK**
09:30–09:50 - ***Development of the National Genomic Medicine Network*** **– Octavian BUCUR**
09:50–10:05 - ***Ancient DNA, Modern Phenotype*** **- Horia STĂNESCU**
10:05–10:25 – ***Shortening the diagnostic odyssey with advanced genomics solutions*** **– Jeroen ADEMA – Illumina - Simpozion ELTA 90 MR**
10:25–10:40 – ***Next-Generation Sequencing in pathology: Enabling personalized medicine*** **– Agnieszka GRYBOS-GAJNIAK – Illumina - Simpozion ELTA 90 MR**
10:40–10:55 – ***Cystinosis - causes, diagnosis, and treatment*** **- Flavia CHIȘAVU - CHIESI Symposium**

10:55–11:00 – COFFEE BREAK. POSTER & EXHIBITION VIEWING

11:00–13:30 - PLENARY SESSION – CLINICAL GENETICS, OPHTHALMOGENETICS
Chairmen: Maria PUIU, Marius BEMBEA

11:00–11:20 – ***Genetics of the circadian rhythm*** **– Marius BEMBEA**
11:20–11:40 - ***Modeling human de novo heterozygosity using mosaic gene dosage differences in mice*** **- Tudor BADEA**
11:40–11:55 – ***Congenital fibrosis of the extraocular muscles or congenital myasthenic syndrome? When genetic testing is essential*** **- Andreea CIUBOTARU**
11:55–12:10 – ***2q37 microdeletion/deletion syndrome -CRGM Iasi experience*** **– Cristina RUSU**
12:10–12:20 – ***RLBP1 novel variant in a patient with cone/rod retinopathy*** **- Raluca PASCALAU**
12:20–12:30 – ***The diagnostic odyssey for the 3 most difficult ophthalmogenetics cases within CRGM Timiș*** **– Costela ȘERBAN**
12:30–12:40 – ***Expert eye" for "rare eyes": Inherited retinal diseases (IRDs) – a case series*** **– Vasilica PLĂIAȘU**
12:40–12:50 – ***Clinical and molecular findings in autism spectrum disorders*** **– Magdalena BUDIȘTEANU**
12:50–13:00 – ***Clinical efficacy and the diversity of genetic diagnosis using the TruSightOne panel. The experience of the Parhon National Institute of Endocrinology*** **– Elena Emanuela BRAHA**
13:00–13:10 – ***Genetic pathologies under the mask of congenital glycosylation disorders*** **– Daniela BLĂNIȚĂ**
13:10–13:40 – ***The role of the geneticist in the multidisciplinary management of the patient with cystic fibrosis*** **– Ioana STREAȚĂ – MEDISON Symposium**

13:40 – 14:30 – LUNCH

14:30–16:00 - PLENARY SESSION – GENETIC AND RARE DISEASES – ORGANIZATION AT THE NATIONAL LEVEL IN THE EUROPEAN CONTEXT - JOINT ACTION, ERNS, AND NATIONAL EXPERTISE CENTERS - I
Chairmen: Nicoleta ANDREESCU, Dorica DAN

14:30–14:45 – ***The role of Norwegian programs in the progress of rare diseases*** **– Dorica DAN**
14:45–15:00 – ***Screening of Inborn Errors of Metabolism by tandem MS/MS tool: piloting METABO MS*** **– Cristina-Adela IUGA**
15:00–15:10 - ***Together for a better future*** **– Iuliana DUMITRIU**
15:10–15:20 – ***Living the Transformation: The Patient's Experience during the limb lengthening operation*** **– Alina TĂTUCU**
15:20–15:30 - ***Newborn screening for spinal muscular atrophy in Romania: 1 year experience of a pilot project at Robanescu Center*** **– Elena NEAGU**
15:30–15:40 - ***Surmounting Challenges: Implementing Newborn Genetic Screening for Spinal Muscular Atrophy in Republic of Moldova*** **– Iulia COLIBAN**
15:40–15:50 - ***Genetic Testing for Cystic Fibrosis in the Regional Center of Medical Genetics Cluj-Napoca*** **– Florina – Victoria NAZARIE**
15:50–16:00 - ***Current strategies for the genetic diagnosis of Mitochondrial DNA disorders in Republic of Moldova*** **– Doina SECU**
16:00–16:20 – NF1 – A rare condition that hides ”in plain sight” – Maria PUIU – ASTRAZENECA Symposium
16:20–16:35 – ***Pompe disease: a clinical, diagnostic, and therapeutic overview*** **– Iulia SIMINA – SANOFI Symposium**

16:35–16:45 – COFFEE BREAK. POSTER & EXHIBITION VIEWING

16:45–18:00 - PLENARY SESSION - GENETIC AND RARE DISEASES – ORGANIZATION AT THE NATIONAL LEVEL IN THE EUROPEAN CONTEXT - JOINT ACTION, ERNS, AND NATIONAL EXPERTISE CENTERS - II
Chairmen: Cristina RUSU, Claudia JURCĂ

16:45–17:20 – ***Regional Medical Genetics Centers - present and future*** **- Florin BURADA, Maria PUIU, Cristina RUSU, Claudia JURCA, Vasilica PLĂIAȘU, Camelia AL-KOZHUZ, Claudia BĂNESCU**
17:20–17:30 – ***Centers of expertise, Romania - versus Europe*** **– Maria PUIU**
17:30–17:50 - ***Joint Action in rare genetic diseases - Romanian Medical Genetics Centers activity within European Reference Networks*** **– Ioana STREAȚĂ**
17:50–18:00 – ***AGMP The Genetics and Personalized Medicine Association*** **– Andreea CĂTANĂ**

18:00 – 19:00 – SRGM GENERAL ASSEMBLY

20:30 – DINNER – CONTINENTAL HOTEL

SATURDAY, 30th SEPTEMBER 2023

09:00 11:00 - PLENARY SESSION – PRENATAL GENETICS
Chairmen: Florin BURADA, Vlad GORDUZA

09:00–09:10 – ***The limits of prenatal genetics*** **- Bogdan – Marius MUREȘAN**
09:10–09:20 – ***Genetic counselling after discordant noninvasive prenatal test results*** **- Florin BURADA**
09:20–09:30 – ***Prenatal diagnosis: from imaging to genetics or vice versa?*** **– Adrian RAȚIU**
09:30–09:40 – ***Prenatal screening and diagnosis strategies - comparative strategies and point of view*** **– Anca-Lelia RIZA**
09:40–09:50 – ***Congenital abnormalities and genetic associations. When should we start and stop counseling?*** **- Ștefania TUDORACHE**
09:50–10:00 - ***Trisomy 12 in mosaic - a challenge of prenatal diagnosis. Case presentation*** **– Mariela MILITARU**
10:00–10:10 - ***Genetic causes of early menopause*** **– Flavia Elena HARADJA**
10:10–10:20 - ***Prenatal WES in a case with a family history of lissencephaly*** **– Cristina GUG**
10:20–10:40 - BLUE PRINT GENETICS Symposium
10:40–11:00 - ***News in Fabry syndrome*** **– Ioana STREAȚĂ – TAKEDA Symposium**

11:00 – 11:20– COFFEE BREAK. POSTER & EXHIBITION VIEWING

11:20–12:40 - PLENARY SESSION – YOUNG GENETICISTS & RESEARCHERS SESSION
Chairmen: Ioana STREAȚĂ, Emilia SEVERIN

11:20–11:30 - ***Congenital Cutis Laxa: A Complex Diagnostic Case Involving Multiple Clinical Manifestations*** **- Baczoni BALAZS**
11:30–11:40 - ***The utility of Whole Genome Sequencing in the diagnosis of Retinitis Pigmentosa*** **– Iulian-Andrei HOTINCEANU**
11:40–11:50 - ***Diagnosis of imprinting disorders using MS-MLPA*****– Lucian-Mihai ANTOCI**
11:50–12:00 - ***Lynch syndrome - Oncohelp experience, Timisoara*** **- Iulia SIMINA, Iulia PERVA**
12:00–12:10 - ***A new KAT6A heterozygous mutation - case report*** **– Alexandru CĂRĂMIZARU**
12:10–12:20 - ***Characterization of sepsis inflammatory endotypes using circulatory proteins in patients with severe infection: the FUSE study*** **– Andra GRIGORESCU**
12:20–12:30 - ***Hereditary optic atrophy associated with OPA1 gene - NGS genetic testing for 7 members of a family*** **- Miruna GUG**

12:30–13:00 AWARD CEREMONY. CONCLUSIONS.
OFICIAL CLOSING CEREMONY

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