

THE 7th NATIONAL CONGRESS OF MEDICAL GENETICS

WITH INTERNATIONAL PARTICIPATION

17-19 SEPTEMBER 2026
ORADEA

AGENDA



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Congress Venue

Hotel Ramada by Wyndham Oradea

Calea Aradului 9, Oradea, Bihor, 410223, Romania



Dear friends and colleagues,

On behalf of the Organizing Committee, it is our great honor and pleasure to welcome you to ***The 7th National Congress of Medical Genetics with International Participation***, held in **Oradea, Romania, from September 17 to 19, 2026.**

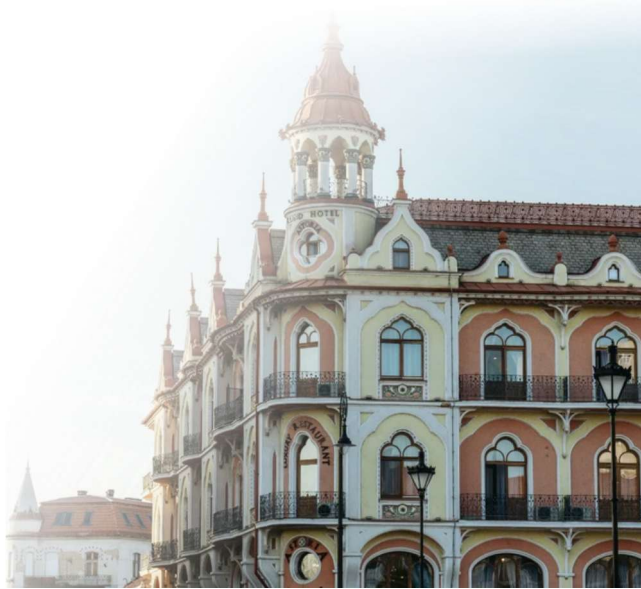
Medical genetics is undergoing a stage of profound development, driven by technological and conceptual advances that have significantly influenced medical practice. Modern genomic analysis methods have contributed to a better understanding of the mechanisms of genetic diseases and have opened new perspectives in diagnosis and therapeutic management.

This year's edition holds special significance, marking the Congress's return to Oradea, the city that hosted the First National Congress of Medical Genetics more than two decades ago, in September 2002. The years since then reflect both the remarkable evolution of the field and the consolidation of an academic community dedicated to research and interdisciplinary collaboration.

Thank you all for being part of this Congress!

Congress Presidents

Prof. Dr. Maria Claudia Jurca & Prof. Dr. Florin Burada



THURSDAY, SEPTEMBER 17, 2026

11:00 – 13:30 – REGISTRATION OF PARTICIPANTS

13:30 – 14:00 – OFFICIAL OPENING OF THE CONGRESS

14:00 – 16:05 – PLENARY SESSION I
Moderators: Florin Burada, Claudia Jurca

14:00 – 14:20 – Viorel Jinga – *The Role of Genetic Testing in Urology: Our Experience from Several Projects*

14:20 – 14:35 – Simona Dima – *The ROGEN Project – Strategic Anchor of the National Genomic Research Infrastructure*

14:35 – 14:50 – Florentina Furtunescu – *National Genomic Medicine Strategy – Challenges and Success Factors*

14:50 – 15:05 – Angela Brand – *Personalised Medicine in Europe*

15:05 – 15:20 – Bogdan Feciche – *Current Trends in the Approach to Oncological Urology From Genetic Mutations to Current Surgery*

15:20 – 15:35 – Ovidiu Cristian Frățilă – *The Genetic Landscape of Inflammatory Bowel Disease*

15:35 – 15:45 – Roxana Măciucă – *IL-6 -174 G/C Polymorphism and Torque Teno Virus Infection in Chronic Glomerulonephritis*

15:45 – 16:05 – Partner Sponsor Symposium AstraZeneca

 **Maria Puiu** – *Advances in the Diagnosis and Treatment of Patients with TWO Rare Metabolic Diseases: HPP, LAL-D*

16:05 – 16:25
COFFEE BREAK & EXHIBITION VISIT & POSTERS

THURSDAY, SEPTEMBER 17, 2026

16:25 – 18:45 – PLENARY SESSION II
Moderators: Adela Chiriță-Emandi, Cristina Rusu

16:25 – 16:50 – Eric Legius – *Inheritance Patterns in Genodermatoses*

16:50 – 17:05 – Cristina Rusu – *ORPHA Codes – Practical Importance and Update in the National and International Context*

17:05 – 17:20 – Adela Chiriță-Emandi – *Genetic Characterization and Genotype - Phenotype Correlations in a Cohort of Romanian Patients with Type 1 Neurofibromatosis: Perspectives on a Clinically Unpredictable Disorder*

17:20 – 17:35 – Camelia Alkhzouz – *Early Clinical Manifestations and Diagnostic Possibilities in Lysosomal Diseases*

17:35 – 17:50 – Raluca Teleanu – *From the Diagnostic Odyssey to the Therapeutic Window: Lessons from Pediatric Neurology Practice*

17:50 – 18:05 – Mihaela Amelia Dobrescu – *Driven by Evidence, Not Preference: Therapeutic Decisions in Genetic Disorders*

18:05 – 18:15 – Iulia-Elena Simina – *Challenges in Patient Access to Innovative Treatments for Rare Diseases: Lessons from a Small Angelman Syndrome Cohort*

18:15 – 18:35 – Partner Sponsor Symposium AstraZeneca

 **Adela Chiriță-Emandi** – *Understanding the Process of Transition from Pediatric to Adult Care: In Focuss NF1 with and Without PN*

18:35 – 18:45 – Gold Sponsor Symposium – Blueprint Genetics

 **Thomas Borge** – *Quality Beyond Sequencing – Why Interpretation and the Clinical Report Matter*

19:30
WELCOME COCKTAIL

FRIDAY, SEPTEMBER 18, 2026

08:30 – 10:30 – PLENARY SESSION III
Moderators: Vlad Gorduza, Mariela Militaru

08:30 – 08:45 – Mariela Sanda Militaru – *Male Infertility in the Genomic Era: Guideline Recommendations Versus Clinical Practice*

08:45 – 09:00 – Florin Burada – *Conflicting NIPT Results: From Causes to Clinical Impact and Management – Lessons from Real Cases*

09:00 – 09:15 – Eusebiu Vlad Gorduza – *Jumping Translocation, a Rare Form of Chromosomal Abnormality – Discussion Based on a Clinical Case*

09:15 – 09:30 – Magdalena Budişteanu – *Rare Genomic Imbalances Encompassing Kinase Genes in a Group of Patients with Autism Spectrum Disorders*

09:30 – 09:40 – Mihai Cucu – *Optical Genome Mapping – First Contact in Craiova*

09:40 – 09:50 – Radu-Alexandru Truica – *Diagnostic Challenges in Sex Chromosome Aneuploidies–Case Reports. When Phenotype and Genotype Do Not Agree*

09:50 – 10:10 – Platinum Sponsor Symposium Novartis

 **NOVARTIS** **Claudia Jurca** – *The Role of Early Diagnosis and the Importance of Genetic Testing in Spinal Muscular Atrophy*

10:10 – 10:30 – Platinum Sponsor Symposium Sanofi

 **sanofi** **Maria Puiu** – *From Genotype to Treatment: Fabrazyme, the Reference Therapy in Fabry Disease for over Two Decades*

10:30 – 10:40
COFFEE BREAK & EXHIBITION VISIT & POSTERS

FRIDAY, SEPTEMBER 18, 2026

10:40 – 12:40 – PLENARY SESSION IV
Moderators: Andreea Cătană, Dragoș Crețoiu

10:40 – 11:05 – Aleš Maver – *Comprehensive Genomic Testing in the Age of Long - Read Sequencing and Artificial Intelligence*

11:05 – 11:20 – Andreea Cătană – *Oncogenetics in the AI Era*

11:20 – 11:35 – Dragoș Crețoiu – *The Geneticist and the Algorithm: Large Language Models in Clinical Variant Interpretation*

11:35 – 11:50 – Vasilica Plăiașu – *Beyond the 'Rigid Textbook Phenotype': AI vs. Human Expertise in Rare Diseases*

11:50 – 12:00 – Gold Sponsor Symposium – Regina Maria



REGINA MARIA

Andreea Cătană – *Beyond the Genetic Code: Genomic Testing in Medical Practice*

12:00 – 12:40 – Partner Sponsor Symposium – Genesis Pharma



Moderator: Maria Puiu

New Insights in the Diagnosis and Management of Rare Diseases

- **Ioana Streață – Vyjuvek (Beremagene Geperpavec)** - *A Topical Gene Therapy Gel for the Treatment of Wounds in Patients with Dystrophic Epidermolysis Bullosa*
- **Niculina Butoianu – Agamree (Vamorolone)** - *A Dissociative Corticosteroid Designed for the Treatment of Duchenne Muscular Dystrophy (DMD)*
- **Diana Miclea – Givlaari (Givosiran)** - *Diagnosis and Management of Acute Hepatic Porphyria*

12:40 – 14:00
LUNCH

FRIDAY, SEPTEMBER 18, 2026

14:00 – 15:55 – PLENARY SESSION V
Moderators: Claudia Bănescu, Nicoleta Andreescu

14:00 – 14:25 – Oliver Bartsch – *Rubinstein-Taybi Syndrome (RSTS) – Natural History and New Drugs for Treatment*

14:25 – 14:50 – Horia Stănescu – *Evolutionary Shaping of Genomic Architecture: Insights for Human Genetics?*

14:50 – 15:05 – Claudia Bănescu – *Integrated Genomic Profiling in Acute Leukemias: The Experience of the Regional Center of Medical Genetics Mures*

15:05 – 15:20 – Nicoleta Andreescu – *Beyond Diagnosis: 10 Years of CRGMT Experience in the Era of Modern Genetics*

15:20 – 15:35 – Romana Vulturar – *Adult Genetics: Old Roads, New Frontiers in Metabolic Medicine*

15:35 – 15:45 – Gold Sponsor Symposium – Biogenetix

 **Virgil Valeriu Vrajmaşu** – *Next Generations Sequencing Laboratory – The Future Was Launched*

15:45 – 15:55 – Gold Sponsor Symposium – Elta 90



Adrian Creţu – *The Role of WES and WGS in the Diagnosis of Genetic Diseases Using Illumina Sequencing Technology*

15:55 – 16:15
COFFEE BREAK & EXHIBITION VISIT & POSTERS

16:00 – 17:00 - SALA B
WORKSHOP FOR YOUNG GENETICISTS
Eric Legius – *Bayes Theorem in Genetic Counselling*

FRIDAY, SEPTEMBER 18, 2026

16:15 – 17:50 – PLENARY SESSION VI
Moderators: Emilia Severin, Camelia Alkhzouz

16:15 – 16:40 – Orly Luminița Levit – *Beyond Lethal: Contemporary Prenatal Counseling and Neonatal Management in Trisomy 13 and 18*

16:40 – 16:55 – Emilia Severin – *Dental Anomalies in Rare Genetic Diseases: From Clinical Recognition to Molecular Diagnosis*

16:55 – 17:10 – Cristina Gug – *When Alzheimer's Mimics Multiple Sclerosis: Unmasking PSEN1-Linked Disease Present Across Four Generations*

17:10 – 17:25 – Claudia Jurca – *Tall Stature: From Clinical Evaluation to Molecular Diagnosis*

17:25 – 17:40 – Diana Miclea – *Genetic Testing in Short Stature*

17:40 – 17:50 – Gold Sponsor Symposium – Medist

 **MEDIST** **Meral Gavaz** – *Advancing Oncology with GeneMind: Comprehensive Next-Generation Sequencing Technologies*

18:00 – 20:00
SRGM MEMBERS' ASSEMBLY

20:30
GALA DINNER



SATURDAY, SEPTEMBER 19, 2026

09:00 – 11:25 – PLENARY SESSION VII
Moderators: Maria Puiu, Elena Braha

09:00 – 09:15 – Maria Puiu – *ExpertRARE and Equitable Genetics – From National Projects to Changing Clinical Practice in Rare Diseases*

09:15 – 09:30 – Dorica Dan – *Complementarity of Health Policies for Rare Diseases*

09:30 – 09:45 – Tudor Pop – *Pediatric Hepatology in the Genomic Era*

09:45 – 10:00 – Ioana Streață – *Skeletal Dysplasias in 2026: From the Revision of the 2023 Nosology to Precision Genetic Diagnosis, Personalized Therapies and the Development of Local Expertise*

10:00 – 10:15 – Corina Paul – *Clinical and Therapeutic Aspects of Noonan Syndrome*

10:15 – 10:30 – Lavinia Caba – *A Century of Genetics in the Light of the Nobel Prizes: Milestones in the Evolution of Modern Genetics*

10:30 – 10:45 – Elena Emanuela Braha – *Multidisciplinary Approach to MEN2 Syndrome – Personalized Medicine and the Impact of Genetic Family Studies*

10:45 – 11:05 – Platinum Sponsor Symposium Merck

 **Claudia Jurca** – *From Gene to Treatment: Genetics of Desmoid Tumors in Clinical Practice*

11:05 – 11:25 – Platinum Sponsor Laboratorium Life Science

 **Damla Bozkulak** – *Elevating Precision Diagnostics: The Integration of SOPHiA DDM Platform and Advanced NGS Assays*

11:25 – 11:40

COFFEE BREAK & EXHIBITION VISIT & POSTER PRESENTATIONS

SATURDAY, SEPTEMBER 19, 2026

11:40 – 13:10 – PLENARY SESSION VIII
Presentation Session for Young Researchers
Moderators: Romana Vulturar, Ioana Streață

- 11:40 – 11:50 – Teodora Barbarii** – *Harmonizing Genetic Testing for Movement Disorders: A Unified Panel Approach and Reanalysis of 4,046 Unsolved Exomes*
- 11:50 – 12:00 – Lorin-Manuel Pîrlog** – *One Family, Two NF1 Variants, Three Phenotypes: Beyond Targeted Cascade Testing*
- 12:00 – 12:10 – Alexandra Zamfirescu** – *A Recurrent Inflammatory Syndrome: More Than Meets the Eye*
- 12:10 – 12:20 – Miruna Gug** – *Couple-Based Reproductive Genetic Carrier Screening in Western Romania: Reproductive Risk and Public Health Implications*
- 12:20 – 12:30 – Iulia Maria Sabau** – *Linear Reference Versus Personalized Pangenome References for Short-Read Whole-Genome Variant Calling*
- 12:30 – 12:40 – Andreea Ardeleanu** – *Beyond the 'Healthy Carrier: When Genotype Demands Functional Confirmation*
- 12:40 – 12:50 – Catalin Vasile Munteanu** – *Mutations We Almost Missed, Reframed: Diagnostic Blind Spots in Short-Read Data Exemplified*
- 12:50 – 13:00 – Silvia Bleahu** – *Bridging the Gap: The Utility of Expanded Carrier Screening in Couples from Central Romania*
- 13:00 – 13:10 – Vlăduț-Ovidiu Geană** – *Intellectual Disability in Neurofibromatosis Type 1: The Experience of a Romanian Clinical Genetics Center*

13:10 – 13:40
AWARDS
CONCLUSIONS
OFFICIAL CLOSING CEREMONY

13:40 – 14:30
LUNCH

SATURDAY, SEPTEMBER 19, 2026

11:25 – 11:40
POSTER SESSION

1. **Simona Ioana Vicas** – *Integrating Genomics and Metabolic Phenotyping in Rare Diseases: The Role of Exogenous Metabolites in Personalized Medicine*
2. **Gabriel Tudor Indig** – *Complete Androgen Insensitivity Syndrome: A Case Highlighting the Importance of Early Genetic Evaluation and Multidisciplinary Care*
3. **Amoașii Anastasia** – *Small Stature, Big Picture: Decoding the Ultra-Rare Genetic Syndromes Masked by Prenatal IUGR*
4. **Alexandra Herlaș-Pop** – *CRGM Bihor – Experience of a Pilot Center in Newborn Screening for Spinal Muscular Atrophy*
5. **Maria-Victoria Corbu** – *Integrating Conventional Karyotyping and FISH in the Cytogenetic Characterization of Acute Leukemias: A Tertiary Center Experience*
6. **Adelina-Maria Cormoș** – *A Case Report of Atypical Alport Syndrome Presenting with Acute Abdominal Pain*
7. **Adela-Daria Neacșu** – *Utility and Limitations of LIRICAL and Phenomiser for Phenotype-Driven Disease Prioritization in an Epilepsy Whole-Exome Sequencing Cohort*
8. **Anca-Lelia Riza (Costache)** – *Perinatal Impaired Cytokine Response in at Risk Pregnant Women. The MERSIN Study (Myeloid dERived Suppressor Cells – Role in Severe Infections)*
9. **Mădălina-Diana Rafa** – *Learning from Family History: A Neonatal Death as a Clue to Inherited Metabolic Diseases—Lessons from Argininosuccinic Aciduria*
10. **Alexandra-Aurora Dumitra** – *Genotype-Phenotype Correlations in Patients with Neurodevelopmental Disorders Investigated by Next-Generation Sequencing (NGS) – Experience of a Regional Center and Implications for Personalized Medicine*
11. **Maria-Liana Marica** – *Psychological Impact and Interdisciplinary Management in Achondroplasia: From Genetics to Quality of Life*
12. **Adriana-Stefania Gidea** – *The Dynamic Genome: Somatic Mosaicism Across the Lifespan – From Early Embryonic Mutagenesis to Age-Related Clonal Selection in Human Disease*
13. **Mitut-Veliscu Andreea** – *Marfan Syndrome: Variable Expressivity and Cascade Testing in a Family – Mother and Two Children*

SATURDAY, SEPTEMBER 19, 2026

**11:25 – 11:40
POSTER SESSION**

14. **Tiberiu-Valentin Pandealea** – *Beyond Common-Mutation Panels: Whole-Gene CFTR Next-Generation Sequencing in a Romanian Cohort Referred for Cystic Fibrosis Suspicion*
15. **Iulia Maria Sabau** – *Temporal Integration of mRNA and lncRNA Expression Identifies Candidate Cis-Regulatory Signatures in Post-Infarction Cardiac Remodeling*
16. **Catalina Muntean-Simona** – *Molecular Spectrum of Polycystic Kidney Disease Diagnosed by NGS Using the TruSight One Panel at the Regional Centre of Medical Genetics Timiș*
17. **Mihai Teodor Barbarii** – *Genetic Counselling in Inherited Cardiomyopathies: Clinical Case Series*
18. **Andrian Fratea** – *Transcriptional Profiling of Immune Cells Reveals a Role for IFN γ /CXCL9/CXCL10 Pathway in Sepsis Patients with Exaggerated Inflammation*
19. **Rebecca-Cristiana Serban** – *Beyond Statural Gain: Evolution of BMI and Anthropometric Profile During Vosoritide Treatment in Patients with Achondroplasia*
20. **Ana-Maria Filip** – *The Immunogenetics of Recurrent Pregnancy Loss: What the Evidence Tells Us*
21. **Lorena-Maria Hanțig** – *Decoding Neonatal Lactic Acidosis: A Biochemical Signature of Mitochondrial Anaplerotic Dysfunction*
22. **Adriana-Stela Crișan** – *MicroRNA Expression in Myeloproliferative Neoplasms*
23. **Raul Andrei Gall** – *Beyond an Adverse Drug Reaction: The Genetic Perspective on Valproate-Induced Hyperammonaemia*
24. **Mihaela-Ștefania Buzdugan** – *Tetrasomy 9p: Phenotypic Correlations and Challenges in Cytogenetic Characterization in a Pediatric Case*
25. **Alina Bogliș** – *From Vascular Phenotype to Molecular Diagnosis: Exome Sequencing Role in Capillary Malformation-Arteriovenous Malformation Syndrome Type 1*
26. **Florica Ramona Dorobantu** – *Severe Delayed Hemolytic Anemia in a Newborn with Maternal–Fetal Rh-c Incompatibility: A Diagnostic Challenge*
27. **Codruta Diana Petchesi** – *Beyond a VUS: A Complex Phenotype*
28. **Olivia Onorica Dumnici** – *Short Stature in Children: A Retrospective Analysis of Cases and Growth Hormone Therapy Outcomes over a 25-Year Period at Regional Center of Medical Genetics Bihor*



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