

FIFTH SCIENTIFIC CONFERENCE

GENETICS IN CLINICAL PRACTICE

WITH INTERNATIONAL SPEAKERS

05–08 June 2025

Sol Luna Bay Hotel, Obzor



05.06.2025 /Thursday/

12.00–15.30 REGISTRATION

15.30–17.00 SCHOOL OF GENETICS – MITOCHONDRIAL DISEASES

MODERATOR: PROF. ILIANA PACHEVA

15.30–15.50 Clinical manifestations in patients with mitochondrial diseases

PROF. ILIANA PACHEVA

15.50–16.10 Imaging studies in the diagnosis of mitochondrial diseases

ASSOC. PROF. MARIN PENKOV

16.10–16.30 Genetic studies in mitochondrial diseases

PROF. SNEZHINA MIHAYLOVA-KANDILAROVA

16.30–16.50 Treatment for patients with mitochondrial diseases

DR. TSVETINA VELEVA

16.50–17.00 Discussion

17.00–17.10 BREAK

17.10–17.20 OPENING

Mythology and genetics

PROF. DANIELA AVDJIEVA-TZAVELLA

17.20–17.40 Dysmorphology

IN MEMORY OF PROF. EMIL SIMEONOV

Genetics of Intellectual Disability

DR. IRENA BRADINOVA

17.40–18.00 IN MEMORY OF DR. ADIL KADIM

Gastrointestinal manifestations in children with congenital metabolic disorders

ASSOC. PROF. IVAN YANKOV

18.00–20.10 DIAGNOSTIC DILEMMAS – PART 1

MODERATORS: PROF. IVAN IVANOV, ASSOC. PROF. ATANAS BANCHEV

18.00–18.15 The well-known beta-thalassemia – the surprises it may bring

DR. SILVIA ANDONOVA

18.15–18.30	3M syndrome DR. TODOR RUSKOV, DR. MARIA SREDKOVA
18.30–18.45	Exome sequencing in the diagnosis of a complex pediatric case: The journey from an unclear prenatal diagnosis to an accurate postnatal genetic diagnosis ASSOC. PROF. MARGARITA PANOVA, DR. CHRISTO IVANOV, DR. IGLIKA SOTKOVA
18.45–19.00	Cerebral Palsy – Where and When Does Genetics Fit In? An Instructive Case ASSOC. PROF. PETYA DIMOVA, MILA SLEPTSOVA, TIHOMIR TODOROV, PROF. ALBENA TODOROVA
19.00–19.15	Clinical case of infantile cortical hyperostosis in infancy DR. KATYA TEMELKOVA
19.15–19.30	Genetic diagnosis: (im)possible? ASSOC. PROF. MARIA LEVKOVA
19.30–19.45	Late but on time – the role of pre-analytical genetic counseling DR. MILENA STOYANOVA
19.45–20.00	Microcephaly and epilepsy DR. DIMITAR STAMATOV AND TEAM
20.00–20.10	Discussion

06.06.2025 /Friday/

08.30–10.00	MADE IN BULGARIA MODERATORS: PROF. IVANKA DIMOVA, PROF. SNEZHINA MIHAYLOVA- KANDILAROVA
08.30–08.50	"Over-diagnosis" in the era of genomic technologies. When can it put us in a deadlock situation? TIHOMIR TODOROV
08.50–09.10	The dark side of the genome DR. LYUBOMIR BALABANSKI
09.10–09.30	Genetic enigmas in frequently ill children in Bulgaria DR. POLINA KOSTOVA-SHAHID, DR. SILVIA ANDONOVA, TIHOMIR TODOROV, PROF. ALBENA TODOROVA, PROF. GERGAN PETROVA

09.30–09.50	New genetic forms of intellectual deficiency syndromes DR. NEVYANA IVANOVA
09.50–10.00	Discussion
10.00–11.30	A SENSE OF THE FUTURE MODERATORS: PROF. ALBENA TODOROVA, PROF. RADKA KANEVA
10.00–10.20	In the mysterious labyrinth of intronic sequences PROF. ALBENA TODOROVA
10.20–10.40	Genetics on the other side of the Atlantic DR. PLAMENA SIMEONOVA
10.40–11.00	Artificial intelligence in the diagnosis of congenital metabolic disorders ASSOC. PROF. MARIA IVANOVA
11.00–11.20	Genomic project How will the project support the development of genomic medicine in Bulgaria? PROF. RADKA KANEVA
11.20–11.30	Discussion
11.30–13.30	Scientific Session GENETICS AND NEUROLOGY – PART 1 MODERATORS: PROF. VENETA BOZHINOVA, PROF. IVAYLO TANEV
11.30–11.50	Hereditary spastic paraplegia in children ROF. IVAYLO TARNEV
11.50–12.10	Clinical aspects in the etiology of Global Developmental Delay and intellectual disability PROF. IVAN IVANOV
12.10–12.30	Epileptic and developmental encephalopathies PROF. VENETA BOZHINOVA
12.30–13.00	Scientific Symposium
12.30–12.50	Gene therapy for AADC deficiency – first Bulgarian patient ASSOC. PROF. PETYA DIMOVA, PROF. KRASIMIR MINKIN
12.50–13.00	Phenylketonuria – Unmet Needs. What's New? PROF. DANIELA AVDJIEVA-TZAVELLA

13.00–13.20	Scientific Symposium Genetics of Porphyrias and Other Rare Diseases in the Light of New Therapeutic Approaches PROF. ALBENA TODOROVA
13.20–13.30	Discussion
13.30–14.30	LUNCH
14.30–16.30	Scientific Session GENETICS AND NEUROLOGY – PART 2 MODERATORS: PROF. IVAN LITVINENKO, PROF. PETYA DIMOVA
14.30–14.50	Mutations in focal cortical dysplasias: germline, somatic, or both? ASSOC. PROF. PETYA DIMOVA
14.50–15.10	Genetic and Clinical Heterogeneity of POLR3A Defects: From Hypomyelinating Leukodystrophy to Peripheral Polyneuropathies PROF. ALBENA YORDANOVA
15.10–15.30	Congenital Myasthenia PROF. IVAN LITVINENKO
15.30–16.00	Scientific Symposium MODERATOR: PROF. IVAN LITVINENKO Our Experience with Gene Therapy in Children Diagnosed with Spinal Muscular Atrophy DR. GENOVEVA TACHEVA
16.00–16.20	Scientific Symposium Acid Sphingomyelinase Deficiency PROF. IVAYLO TURNEV
16.20–16.35	Scientific Symposium Current Perspectives on the Diagnosis and Treatment of Dravet Syndrome PROF. ILIYANA PACHEVA
16.35–16.50	COFFEE BREAK

16.50–18.30	Scientific session CLINICAL GENETICS MODERATORS: PROF. SAVINA HADZHIDEKOVA, ASSOC. PROF. RADOSLAVA VAZHAROVA
16.50–17.10	PGT and hereditary breast cancer – can and should we break the chain? PROF. SAVINA HADZHIDEKOVA
17.10–17.30	When genomics takes center stage – reflections from a genetic counselor DR. MARI HACHMERYAN
17.30–17.50	The path of a patient with a rare disease PROF. IVANKA DIMOVA
17.50–18.10	Telomere biology disorders ASSOC. PROF. RADOSLAVA VAZHAROVA
18.10–18.30	Scientific Lecture Blueprint Genetic WES future with genotyping first approach MURAT TOPAL
18.30–20.10	Scientific Session – GENETICS AND ONCOHEMATOLOGY MODERATORS: PROF. BORIANA AVRAMOVA, ASSOC. PROF. MAYA YORDANOVA
18.30–18.45	Scientific Symposium Genotyping in Hemophilia – Beyond Prenatal Diagnosis ASSOC. PROF. ATANAS BANCHEV
18.45–19.05	Genotyping in hemophilia beyond prenatal diagnosis ASSOC. PROF. ATANAS BANCHEV
19.05–19.25	The Critical Role of Genetics: Guidelines for Managing CMMRD in Clinical Practice DR. CHRISTO IVANOV
19.25–19.45	Genetic syndromes associated with agranulocytosis ASSOC. PROF. BORIANA AVRAMOVA
19.45–20.05	Genetics in hematological aplastic conditions in pediatrics ASSOC. PROF. MAYA YORDANOVA
20.05–20.10	Discussion

07. 06. 2025 /Saturday/

08.30–11.00 DIAGNOSTIC DILEMMAS – PART 2

MODERATORS: PROF. IVAN LITVINENKO, ASSOC. PROF. IVAN YANKOV

08.30–08.45 Visceral myopathy associated with ACTG2 mutation
DR. NADEZHDA DOLASHKOVA, DR. CHRISTO NAYDENOV,
DR. EMILIYA NEDEVA, PROF. ALBENA TODOROVA, TIHOMIR TODOROV,
DR. MILA BAYCHEVA

08.45–09.00 Rare genetic form of protein-losing enteropathy in DGAT1 deficiency
DR. PETAR NIKOLOV, DR. CHRISTO NAYDENOV, DR. EMILIYA NEDEVA,
PROF. ALBENA TODOROVA, TIHOMIR TODOROV, DR. MILA BAYCHEVA

09.00–09.15 Variable phenotype of intrahepatic cholestasis in TJP2 heterozygotes
DR. EMILIA NEDEVA, DR. NADEZHDA DOLASHKOVA,
DR. CHRISTO NAYDENOV, PROF. ALBENA TODOROVA, TIHOMIR TODOROV,
DR. MARLENA PANAYOTOVA, DR. MILA BAYCHEVA

09.15–09.30 Diagnostic and therapeutic approach in glycosylation disorder type 2
DR. VALENTINA VARBANOVA, DR. GENOVEVA TACHEVA,
DR. TEODORA PANEVA, DR. MARINA KRSTeva, DR. DIMITAR STAMATOV,
PROF. IVAN LITVINENKO

09.30–09.45 Biotinidase deficiency – is the path from symptoms to diagnosis long?
DR. IGLIKA YORDANOVA, DR. BILYANA GEORGIEVA

09.45–10.00 Hypertrophic obstructive cardiomyopathy, congenital heart malformation,
and genetics – a clinical case
ASSOC. PROF. ANNA DASHEVA-DIMITROVA, PROF. ALBENA TODOROVA

10.00–10.15 From a single syndrome to the discovery of two rare diseases: The role of
exome sequencing
DR. IGLIKA SOTKOVA, DR. MARGARITA PANOVA, PROF. ILIANA PACHEVA,
ASSOC. PROF. RALITSA VASILEVA, DR. HRISTO IVANOV

10.15–10.30 From clinical presentation to genetic diagnosis: A case of PIGN-associated
encephalopathy
DR. MARIANA GEORGIEVA, DR. HRISTO IVANOV

10.30–10.45 Rahman syndrome with new features of phenotypic expression
DR. YULIA BAZDARSKA

10.45–11.00	Scientific Lecture 21st Century Cancer Genetic Diagnostics DR. DMITRI OSSIPOV
11.00–11.30	COFFEE BREAK
11.30–13.10	GENETICS, ENDOCRINOLOGY, AND METABOLISM MODERATORS: PROF. VIOLETA YOTOVA, PROF. DANIELA AVDJIEVA-TZAVELLA
11.30–11.50	Diagnosis of lysosomal storage diseases in the era of new technologies DR. IVANKA SINIGERSKA
11.50–12.10	The role of quality control in the diagnosis of inborn errors of metabolism VIKTORIA BICHEV
12.10–12.30	Definition of tall stature in childhood and adolescence. Differential diagnosis and management PROF. VIOLETA YOTOVA
12.30–12.45	Scientific Symposium Personal experience with the application of rchRH in patients with malformative syndromes DR. MARIA SREDKOVA
12.45–13.00	Scientific Lecture Children with Fabry Disease PROF. DANIELA AVDJIEVA-TZAVELLA
13.00–13.10	Discussion
13.10–14.30	LUNCH

14.30–16.50	Scientific session GENETICS AND GASTROENTEROLOGY MODERATORS: PROF. MIGLENA GEORGIEVA, DR. MILA BAYCHEVA
14.30–14.50	Hepatic tumors in children with congenital metabolic disorders PROF. MIGLENA GEORGIEVA
14.50–15.10	Genetic aspects in severe gastro-intestinal dysmotility and chronic intestinal pseudo-obstruction DR. MILA BAYCHEVA
15.10–15.30	Hepatic Ciliopathies DR. TRAYAN DELCHEV
15.30–16.00	Scientific Symposium Supported by EXCEED ORPHAN/REIMEX PHARMA Genetic and clinical heterogeneity of intrahepatic cholestasis MODERATOR: PROF. MIGLENA GEORGIEVA
15.30–15.45	Symptoms, Biomarkers, and Prognostic Challenges DR. MILA BAYCHEVA
15.45–16.00	Innovations and Traditions in Genetic Diagnostics PROF. ALBENA TODOROVA
16.00–16.10	Discussion
16.10–16.40	COFFEE BREAK
16.40–18.40	Scientific Session TREATMENT OF PATIENTS WITH RARE DISEASES IN BULGARIA MODERATORS: PROF. TEODORA CHAMOVA, PROF. GERGANA PETROVA
16.40–16.55	Cerebrotendinous xanthomatosis DR. DINA ATOVA, PROF. DANIELA AVDJIEVA-TZAVELLA
16.55–17.10	Cerebral folate deficiency DR. TEODORA ALEKSANDROVA, PROF. DANIELA AVDJIEVA-TZAVELLA
17.10–17.25	Results from pharmacological treatment in neurotransmitter disorders DR. GENOVEVA TACHEVA

17.25–17.40	On some new aspects of the diagnosis and treatment of tuberous sclerosis – before and after birth! ASSOC. PROF. RUMEN MARINOV
17.40–17.55	New developments in the treatment of dystrophinopathies PROF. TEODORA CHAMOVA
17.55–18.10	Results from treatment in cystic fibrosis PROF. GERGANA PETROVA
18.10–18.30	Scientific Lecture Epidermolysis bullosa – challenges and new therapeutic options DR. TEODOR VASILEV
18.30–18.40	Discusson
18.40–19.30	GENETICS FOR ENTHUSIASTS MODERATOR: PROF. ALEXEY SAVOV
18.40–19.00	The genetic memory PROF. ALEXEY SAVOV
19.00–19.20	Paleogenomics – What can the future learn from the past? STOYAN BITCHEV
19.20–19.30	Discusson
19.30–19.40	CLOSING
08. 06. 2025 /Sunday/	
09.00	DEPARTURE