KNOWLEDGE GRANTS LIFE foundation

FIFTH SCIENTIFIC CONFERENCE

GENETICS IN CLINICAL PRACTICE

WITH INTERNATIONAL SPEAKERS

05–08 June 2025

Sol Luna Bay Hotel, Obzor

PRELIMINARY SCIENTIFIC PROGRAM

05. 06. 2025 /Thursday/	
12.00–15.30	**REGISTRATION**
15.30–17.00	**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** PROF. DANIELA AVDZHIEVA-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
18.00–18.30	**Scientific Symposium**
18.30–20.00	**Scientific Session – GENETICS AND ONCOHEMATOLOGY** MODERATORS: PROF. BORIANA AVRAMOVA, ASSOC. PROF. MAYA YORDANOVA
18.30–18.50	Constitutional mismatch repair deficiency DR. CHRISTO IVANOV
18.50–19.10	Genotyping in hemophilia beyond prenatal diagnosis ASSOC. PROF. ATANAS BANCHEV
19.10–19.30	Genetic syndromes associated with agranulocytosis ASSOC. PROF. BORIANA AVRAMOVA
19.30–19.50	Genetics in hematological aplastic conditions in pediatrics ASSOC. PROF. MAYA YORDANOVA
19.50–20.00	Discussion

06.06.2025 /Friday/

08.30–10.40	**Scientific Session GENETICS AND NEUROLOGY** MODERATORS: PROF. VENETA BOZHINOVA, PROF. IVAYLO TANEV
08.30–08.50	Mutations in focal cortical dysplasias: germline, somatic, or both? ASSOC. PROF. PETYA DIMOVA
08.50–09.10	Expected topic PROF. ALBENA YORDANOVA /Antwerp University, Belgium/
09.10–09.30	Congenital Myasthenia PROF. IVAN LITVINENKO
09.30–09.50	Hereditary spastic paraplegias in children PROF. IVAYLO TANREV

09.50–10.10	Clinical aspects in the etiology of Global Developmental Delay and intellectual disability PROF. IVAN IVANOV
10.10–10.30	Epileptic and developmental encephalopathies PROF. VENETA BOZHINOVA
10.30–10.40	Discussion
10.40–11.10	**Scientific Symposium**
11.10–11.30	**Scientific Symposium**
11.30–12.00	**COFFEE BREAK**
12.00–12.40	**GENETICS FOR ENTHUSIASTS** MODERATOR: PROF. ALEXEY SAVOV
12.00–12.20	The genetic memory PROF. ALEXEY SAVOV
12.20–12.40	Paleogenomics – What can the future learn from the past? STOYAN BITCHEV
12.40–13.00	**Scientific Symposium**
13.00–14.00	**LUNCH**
14.00–16.25	**DIAGNOSTIC DILEMMAS – PART 1** MODERATORS: PROF. IVAN LITVINENKO, ASSOC. PROF. IVAN YANKOV
14.00–14.15	Visceral myopathy associated with ACTG2 mutation DR. NADEZHDA DOLASHKOVA, DR. CHRISTO NAYDENOV, DR. EMILIYA NEDEVA, PROF. ALBENA TODOROVA, TIHOMIR TODOROV, DR. MILA BAYCHEVA

14.15–14.30	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
14.30–14.45	Variable phenotype of intrahepatic cholestasis in TJP2 heterozygotes DR. EMILIYA NEDEVA, DR. NADEZHDA DOLASHKOVA, DR. CHRISTO NAYDENOV, PROF. ALBENA TODOROVA, TIHOMIR TODOROV, DR. MARLENA PANAYOTOVA, DR. MILA BAYCHEVA
14.45–15.00	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
15.00–15.15	Biotinidase deficiency – is the path from symptoms to diagnosis long? DR. IGLIKA YORDANOVA, DR. BILYANA GEORGIEVA
15.15–15.30	Hypertrophic obstructive cardiomyopathy, congenital heart malformation, and genetics – a clinical case ASSOC. PROF. ANNA DASHEVA-DIMITROVA, PROF. ALBENA TODOROVA
15.30–15.45	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
15.45–16.00	From clinical presentation to genetic diagnosis: A case of PIGN-associated encephalopathy DR. MARIANA GEORGIEVA, DR. HRISTO IVANOV
16.00–16.15	Cerebral Palsy – Where and When Does Genetics Fit In? An Instructive Case ASSOC. PROF. PETYA DIMOVA, MILA SLEPTSOVA, TIHOMIR TODOROV, PROF. ALBENA TODOROVA
16.15–16.25	Discussion
16.25–16.35	**Scientific Lecture** The More, The Better? Comparing WES XTRA and WGS in Genetic Diagnostics DR. DMITRI OSSIPOV

16.35–16.55	**Scientific ymposium**
16.55–17.15	**COFFEE BREAK**
17.15–19.25	**DIAGNOSTIC DILEMMAS – PART 2** MODERATORS: PROF. IVAN IVANOV, ASSOC. PROF. ATANAS BANCHEV
17.15–17.30	The well-known beta-thalassemia – the surprises it may bring DR. SILVIA ANDONOVA
17.30–17.45	3M syndrome DR. TODOR RUSKOV AND TEAM
17.45–18.00	Exome sequencing in the diagnosis of a complex pediatric case: The journey from an unclear prenatal diagnosis to an accurate postnatal genetic diagnosis DR. MARGARITA PANOVA, DR. CHRISTO IVANOV, DR. IGLIKA SOTKOVA
18.00–18.15	Rahman syndrome with new features of phenotypic expression DR. YULIA BAZDARSKA
18.15–18.30	Clinical case of infantile cortical hyperostosis in infancy DR. KATYA TEMELKOVA
18.30–18.45	Genetic diagnosis: (im)possible? ASSOC. PROF. MARIA LEVKOVA
18.45–19.00	Late but on time – the role of pre-analytical genetic counseling DR. MILENA STOYANOVA
19.00–19.15	Microcephaly and epilepsy DR. DIMITAR STAMATOV AND TEAM
19.15–19.25	Discussion
19.25–19.45	**Scientific symposium**

07. 06. 2025 /Saturday/	
08.30–09.40	**Scientific session CLINICAL GENETICS** MODERATORS: PROF. SAVINA HADZHIDEKOVA, ASSOC. PROF. RADOSLAVA VAZHAROVA
08.30–08.45	Expected topic PROF. SAVINA HADZHIDEKOVA
08.45–09.00	When genomics takes center stage – reflections from a genetic counselor DR. MARI HACHMERYAN
09.00–09.15	The path of a patient with a rare disease PROF. IVANKA DIMOVA
09.15–09.30	Telomere biology disorders ASSOC. PROF. RADOSLAVA VAZHAROVA
09.30–09.40	Discussion
09.40–10.00	**Scientific lecture** Blueprint Genetic WES future with genotyping first approach
10.00–11.10	**A SENSE OF THE FUTURE** MODERATORS: PROF. ALBENA TODOROVA, PROF. RADKA KANEVA
10.00–10.15	In the mysterious labyrinth of intronic sequences PROF. ALBENA TODOROVA
10.15–10.30	Genetics on the other side of the Atlantic DR. PLAMENA SIMEONOVA /Johns Hopkins Institute, USA/
10.30–10.45	Artificial intelligence in the diagnosis of congenital metabolic disorders ASSOC. PROF. MARIA IVANOVA
10.45–11.00	Genomic project How will the project support the development of genomic medicine in Bulgaria? PROF. RADKA KANEVA
11.00–11.10	Discussion

11.10–11.30	**COFFEE BREAK**
11.30–12.30	**Scientific session GENETICS AND GASTROENTEROLOGY** MODERATORS: PROF. MIGLENA GEORGIEVA, DR. MILA BAYCHEVA
11.30–11.45	Hepatic tumors in children with congenital metabolic disorders PROF. MIGLENA GEORGIEVA
11.45–12.00	Genetic aspects in severe gastro-intestinal dysmotility and chronic intestinal pseudo-obstruction DR. MILA BAYCHEVA
12.00–12.15	Hirschsprung›s disease – genetics and associated syndromes DR. TRAYAN DELCHEV
12.15–12.30	Discussion
12.30–13.00	**Scientific symposium** Genetic and clinical heterogeneity of intrahepatic cholestasis MODERATOR: PROF. MIGLENA GEORGIEVA
12.30–12.45	Symptoms, Biomarkers and Prognostic Challenges DR. MILA BAYCHEVA
12.45–13.00	Innovations and traditions in genetic diagnostics PROF. ALBENA TODOROVA
13.00–14.00	**LUNCH**
14.00–15.15	**GENETICS, ENDOCRINOLOGY, AND METABOLISM** MODERATORS: PROF. VIOLETA YOTOVA, PROF. DANIELA AVDZHIEVA-TZAVELLA
14.00–14.15	Diagnosis of lysosomal storage diseases in the era of new technologies DR. IVANKA SINIGERSKA
14.15–14.30	The role of quality control in the diagnosis of inborn errors of metabolism VIKTORIA BICHEV

14.30–14.45	Definition of tall stature in childhood and adolescence. Differential diagnosis and management PROF. VIOLETA YOTOVA
14.45–15.05	**Scientific lecture** Personal experience with the application of rchRH in patients with malformative syndromes DR. MARIA SREDKOVA
15.05–15.15	Discussion
15.15–15.35	**Scientific symposium**
15.35–16.00	**COFFEE BREAK**
16.00–17.20	**TREATMENT OF PATIENTS WITH RARE DISEASES IN BULGARIA** MODERATORS: PROF. TEODORA CHAMOVA, PROF. GERGANA PETROVA
16.00–16.15	Cerebrotendinous xanthomatosis DR. DINA ATOVA, PROF. DANIELA AVDJIEVA-TZAVELLA
16.15–16.30	Cerebral folate deficiency DR. TEODORA ALEKSANDROVA, PROF. DANIELA AVDJIEVA-TZAVELLA
16.30–16.45	Results from pharmacological treatment in neurotransmitter disorders DR. GENOVEVA TACHEVA
16.45–17.00	On some new aspects of the diagnosis and treatment of tuberous sclerosis – before and after birth! ASSOC. PROF. RUMEN MARINOV
17.00–17.15	New developments in the treatment of dystrophinopathies PROF. TEODORA CHAMOVA
17.15–17.20	Discussion
17.20–17.40	**Scientific lecture** Results from treatment in cystic fibrosis PROF. GERGANA PETROVA

17.40–18.00	**Scientific lecture** Epidermolysis bullosa – challenges and new therapeutic options DR. TEODOR VASILEV
18.00–19.30	**MADE IN BULGARIA** MODERATORS: PROF. IVANKA DIMOVA, PROF. SNEZHINA MIHAYLOVA-KANDILAROVA
18.00–18.20	«Over-diagnosis» in the era of genomic technologies. When can it put us in a deadlock situation? TIHOMIR TODOROV
18.20–18.40	The dark side of the genome DR. LYUBOMIR BALABANSKI
18.40–19.50	Genetic enigmas in frequently ill children in Bulgaria DR. POLINA KOSTOVA-SHAHID, DR. SILVIA ANDONOVA, TIHOMIR TODOROV, PROF. ALBENA TODOROVA, PROF. GERGANA PETROVA
18.50–19.10	New genetic forms of intellectual deficiency syndromes DR. NEVYANA IVANOVA
19.10–19.30	Discussion
19.30–20.00	**Scientific symposium**
08. 06. 2025 /Sunday/	
09.00	**Отпътуване**