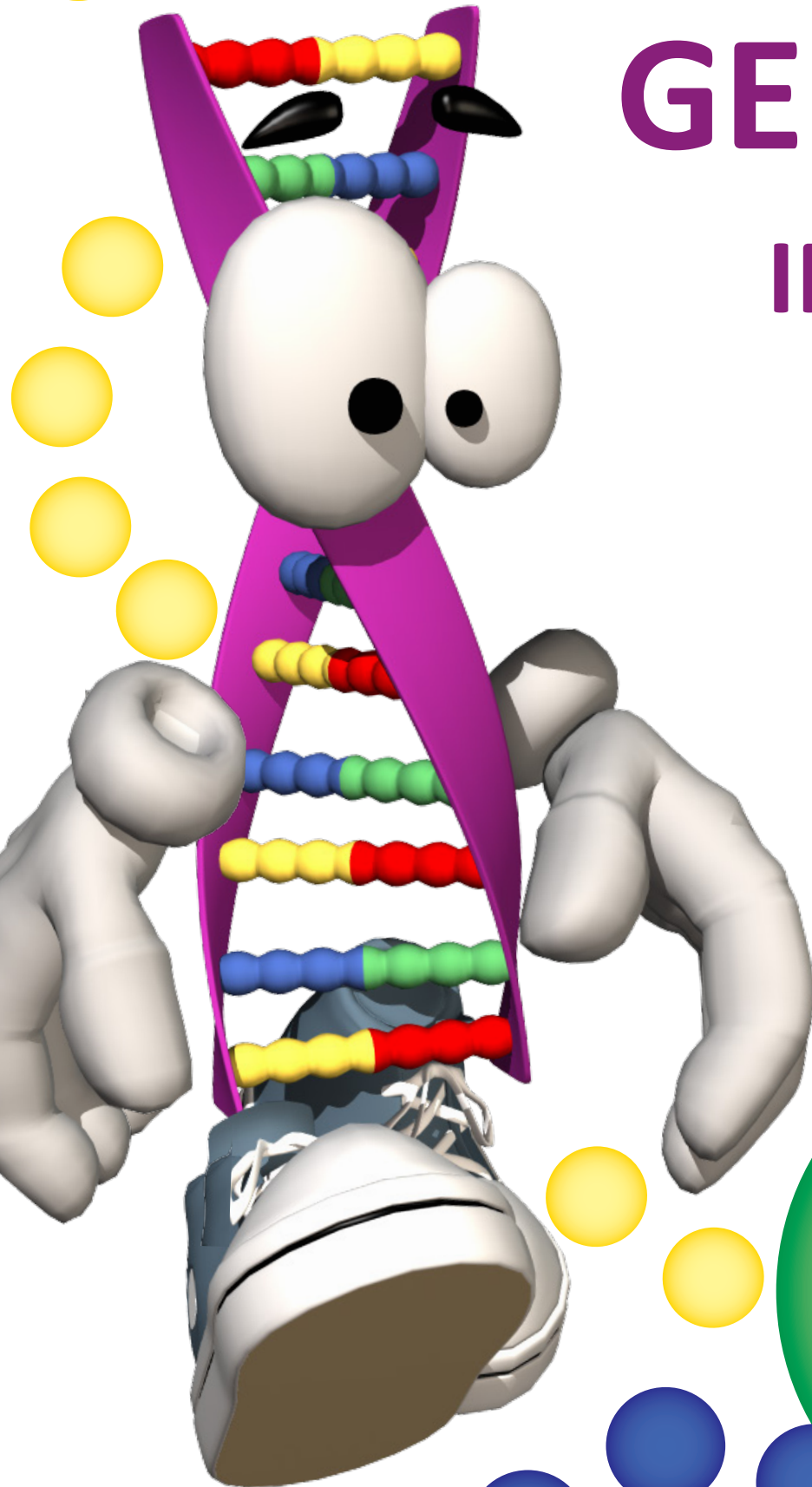


THIRD SCIENTIFIC CONFERENCE

GENETICS IN CLINICAL PRACTICE



25 – 28 MAY 2023

Hotel Belleville,
Duni Royal Resort

SCIENTIFIC PROGRAM

25. 05. 2023 /Thursday/

15:30–17:00	Clinical Genetics School: WHAT CLINICIANS NEED TO KNOW ABOUT GENETIC TESTING <i>MODERATOR: PROF. ALBENA TODOROVA</i>
15:30–15:50	Chromosomal abnormalities and dysmorphology: Conventional and molecular cytogenetics, chromosomal sequencing, Optical Genome mapping ASSOC. PROF. RADOSLAVA VUZHAROVA
15:50–16:10	DNA methylation profiling in the human genome. Epigenetic modifications: MLID (multi-locus imprinting disorders) DR. TIHOMIR TODOROV
16:10–16:30	Analysis of whole exome/genome sequencing data – challenges, RNA sequencing DR. SLAVENA ATEMIN, DR. LYUBOMIR BALABANSKI
16:30–16:50	Transcriptome analysis: protein biomarkers for diagnosis and monitoring of treatment response PROF. ALBENA TODOROVA
16:50–17:00	Discussion
17:00–17:10	Break
17:10–17:20	OPENING Genetics in Art <i>PROF. DANIELA AVDZHIEVA-TZAVELLA</i>
17:20–17:35	In memory of Dr. Adil Kadum DOPA-responsive dystonias DR. GENOVEVA TACHEVA
17:35–17:45	Genetics and clinical spectrum of neurofibromatosis type 1 PROF. DANIELA AVDZHIEVA-TSAVELLA
17:45–20:00	Scientific session: GENETICS AND ONCOHEMATOLOGY/IMMUNOLOGY <i>Moderators: ASSOC. PROF. BORYANA AVRAMOVA, ASSOC. PROF. SNEZHINA KANDILAROVA</i>
17:45–18:00	From accurate diagnosis to targeted therapy – a single-center clinical experience in children with aHUS DR. LILIYA IVANOVA
18:00–18:20	Combined immune deficiencies with syndromic features – algorithms for diagnosis, monitoring, and therapy ASSOC. PROF. SNEZHINA KANDILAROVA

18.20–18.40	Genetic basis of immune deficiencies in Bulgaria DR. POLINA KOSTOVA, DR. ATANAS BANCHEV, DR. VERA PAPOCHIEVA, DR. D. MITEVA, DR. S. MILEVA, DR. MARTIN SHAHID, PROF. ALBENA TODOROVA, ASSOC. PROF. GERGAN PETROVA
18.40–19.00	Fanconi anemia – clinical and genetic approaches to diagnosis DR. NADEZHDA YURUKOVA
19.00–19.20	Genetic predisposition to childhood and adolescent malignancies ASSOC. PROF. BORYANA AVRAMOVA
19.20–19.40	Epigenetics in pediatric oncology DR. IVAN BORUNSUZOV
19.40–20.00	What we don't know about beta-thalassemia – from a genetic perspective PROF. ALEXEY SAVOV

26. 05. 2023 /Friday/

08.30–12.15	Scientific session GENETICS AND NEUROLOGY <i>MODERATORS: PROF. IVAN LITVINENKO, PROF. IVAYLO TARNEV</i>
08.30–08.50	Clinical and genetic spectrum of congenital myopathies PROF. IVAN LITVINENKO
08.50–09.10	New horizons for precision medicine in epilepsy – therapy based on genetically determined functional impairment PROF. ILIYANA PACHEVA
09.10–09.30	New therapies in patients with progressive muscular dystrophies ASSOC. PROF. TEODORA CHAMOVA
09.30–09.50	Leukodystrophies and ceroid lipofuscinoses – clinical and genetic characteristics, differential diagnosis PROF. VENETA BOJINOVA
09.50–10.10	Inherited peripheral neuropathies in childhood PROF. IVAYLO TARNEV
10.10–11.10	Scientific symposium <i>MODERATOR: PROF. DANIELA AVDZHIEVA-TZAVELLA</i>
10.10–10.30	Gene therapy for aromatic L-amino acid decarboxylase (AADC) deficiency PROF. DANIELA AVDZHIEVA-TZAVELLA
10.30–10.50	The role of NGS panels in the diagnosis of early childhood developmental disorders PROF. ILIYANA PACHEVA
10.50–11.10	Challenging clinical case > AADCd? PROF. ALBENA TODOROVA

11.10–11.40	Treating spinal muscular atrophy with SPINRAZA® - from clinical trials to real-world clinical practice PROF. IVAN IVANOV
11.40–11.55	How to wake up a child with narcolepsy? DR. TEODORA PANEVA
11.55–12.15	New diagnostic and therapeutic possibilities in Leber's hereditary optic neuropathy PROF. SILVIYA CHERNINKOVA
12.15–12.45	Coffee break
12.45–14.00	Scientific session GENETICS AND PSYCHIATRY <i>MODERATORS: PROF. VENETA BOJINOVA, PROF. IVAN IVANOV</i>
12.45–13.00	Attention Deficit Hyperactivity Disorder as a manifestation of congenital inborn errors of metabolism DR. TSVETINA VELEVA
13.00–13.15	Genetics of sleep disorders DR. IRENA BRADINOVA
13.15–13.30	Genetics of eating disorders DR. MARI HACHMERIAN
13.30–13.45	Genetics of alcohol and drug addiction STOYAN BICHEV
13.45–14.00	Discussion
14.00–15.00	Lunch break
15.00–16.10	Scientific session: GENETICS AND GASTROENTEROLOGY <i>MODERATOR: PROF. MIGLENA GEORGIEVA</i>
15.00–15.20	Genetic basis of Wilson's disease PROF. MIGLENA GEORGIEVA, DR. NIA RASHEVA, ASSOC. PROF. KRASIMIRA KOLEVA, ASSOC. PROF. ANTONIA ATANASOVA, DR. MILENA STOYANOVA, PROF. LYDMILA ANGELOVA
15.20–15.40	Intrahepatic cholestasis of pregnancy DR. MILA BAICHEVA

15.40–16.00	Clinical and genetic spectrum of congenital diarrhea and enteropathies ASSOC. PROF. EMILIA PANTELEEVA
16.00–16.10	Discussion
16.10–16.30	Progressive familial intrahepatic cholestasis – the possibilities of personalized medicine DR. MILA BAYCHEVA
16.30–17.00	Coffee break
17.00–19.00	Scientific session GENETICS AND RHEUMOCARDIOLOGY <i>MODERATORS: PROF. STEFAN STEFANOV, ASSOC. PROF. ANNA DASHEVA</i>
17.00–17.20	Genetic basis of congenital heart malformations DR. DIMITAR PECHILKOV
17.20–17.40	Genetic basis of congenital heart malformations: cardiomyopathies, aortopathies, channelopathies DR. ELISAVETA LEVUNLIEVA
17.40–18.00	Genetics and hypertrophic cardiomyopathy in children ASSOC. PROF. ANNA DASHEVA
18.00–18.20	Monogenic vasculitis and vasculitis mimickers ASSOC. PROF. MARGARITA GANEVA
18.20–18.40	Genetics in pediatric rheumatology PROF. STEFAN STEFANOV
18.40–19.00	The Unknown UFMilation in norm and in pathology PROF. IVAN IVANOV
19.00–19.10	„Maternal and Child Health – benefits for children of Bulgaria“ Program ARETA ZUMEROVA
19.10–20.00	General Meeting of the Bulgarian Pediatric Association (BPA)
27. 05. 2023 /Saturday/	
08:30–10:10	DIAGNOSTIC DILEMMAS /Part 1/ <i>MODERATORS: ASSOC. PROF. GERGANA PETROVA, ASSOC. PROF. IVAN YANKOV</i>
08:30–08:45	Jarcho-Levin syndrome ASSOC. PROF. IVAN YANKOV
08.45–09.00	Sotos syndrome DR. YOANA IVANOVA, PROF. DANIELA AVDZHIEVA-TZAVELLA

09:00–09:15	Clinical case of a child with glycogen storage disease type IXc DR. DENITSA KOFINOVA, DR. PETYO HADJIYSKI, DR. RAYNA SHENTOVA-ENEVA, DR. MILA BAICHEVA, ASSOC. PROF. RADOSLAVA VUZHAROVA, DR. KOSTADIN KETEV, ASSOC. PROF. NARTSIS KALEVA-HODZHEVA, PROF. IVAN IVANOV, PROF. DANIELA AVDZHIEVA-TZAVELLA
09:15–09:30	Trichothiodystrophy – an (im)possible clinico-genetic diagnosis DR. MILA BAICHEVA, PROF. ALBENA TODOROVA
09:30–09:45	Schaaf-Yang Syndrome DR. TODOR RUSKOV, DR. MARIA SREDKOVA
09:45–10:00	Cystic fibrosis obscured by a predisposition to celiac disease ASSOC. PROF. GERGAN PETROVA
10:00–10:10	Discussion
10.10–10.40	Early diagnosis of cystic fibrosis in the era of CFTR modulators ASSOC. PROF. GERGAN PETROVA
10.40–10.50	Provoked epileptic seizures as a reason to delay the diagnosis of a treatable disease – CLN2 DR. GENOVEVA TACHEVA
10.50–11.20	Coffee break
11.20–13.30	Scientific Session METABOLIC DISEASES <i>MODERATORS: ASSOC. PROF. MARIA IVANOVA, DR. IVANKA SINIGERSKA</i>
11.20–11.40	Dysmorphology and congenital inborn Errors of Metabolism DR. TRAYAN DELCHEV
11.40–12.00	Endocrine Disorders in congenital inborn Errors of Metabolism DR. MARIA SREDKOVA
12.00–12.20	Biomarkers in Lysosomal Diseases DR. IVANKA SINIGERSKA
12.20–12.40	Cytokine regulation – normal ranges and pathology; therapeutic challenges ASSOC. PROF. ANNA VALERIEVA
12.40–13.00	Acute liver failure and congenital inborn errors of metabolism ASSOC. PROF. YORDANKA UZUNOVA

13:00–13:20	Metabolomics ASSOC. PROF. MARIA IVANOVA
13:20–13:30	Discussion
13:30–14:30	Lunch break
14:30–16:00	Scientific session GENETICS AND ENDOCRINOLOGY <i>MODERATORS: PROF. VIOLETA YOTOVA, PROF. IVA STOEVA</i>
14:30–14:50	Skeletal dysplasias in the differential diagnosis of disproportional short stature DR. VESELIN BOYADZHIEV, DR. YULIYA BAZDARSKA, DR. NIKOLINKA YORDANOVA
14:50–15:10	Contemporary treatment of patients with Turner syndrome PROF. VIOLETA YOTOVA, DR. VILHELM MLADENOV, DR. KAMELIA RANKOVA
15:10–15:30	5-Alpha-Reductase deficiency in Bulgaria – the new territory for the (un)known diagnosis DR. SILVIA ANDONOVA
15:30–15:50	Multiple endocrine neoplasia: When to consider it? PROF. IVA STOEVA
15:50–16:00	Discussion
16:00–16:20	Growth hormone treatment in patients with syndromic short stature PROF. DANIELA AVDZHIEVA-TZAVELLA
16:20–16:40	X-linked hypophosphatemic rickets: challenges and opportunities DR. MARIA SREDKOVA
16:40–16:50	New therapies in achondroplasia PROF. DANIELA AVDZHIEVA-TZAVELLA
16:50–17:20	Coffee break
17:20–17:40	Genetic mutations in Bulgaria Diagnosis of rare genetic diseases in Bulgaria. Opportunities and challenges in the era of genomic medicine PROF. RADKA KUNEVA
17:40–19:30	Scientific session CLINICAL GENETICS <i>MODERATOR: PROF. SAVINA HADZHIDEKOVA</i>
17:40–18:00	Facial phenotype: studying the correlation between genes and the human face DR. HRISTO IVANOV

18.00–18.20	Homozygous NDUF56 splice mutation highlights the importance of peripheral neuropathy in the clinical spectrum of primary mitochondrial disorders PROF. ALBENA YORDANOVA
18.20–18.40	Possibilities for neonatal SMA screening in Bulgaria DR. VICTORIA YORDANOVA, STOYAN BICHEV
18.40–19.00	Molecular genetics in a complicated clinical phenotype DR. NEVYANA IVANOVA
19.00–19.20	Which “Genes” you are from? PROF. SAVINA HADZHIDEKOVA
19.20–19.30	Discussion

28 May 2023, Sunday

09:00–10:55 **DIAGNOSTIC DILEMMAS /Part 2/**

***MODERATORS: PROF. DANIELA AVDZHIEVA-TZAVELLA,
ASSOC. PROF. YORDANKA UZUNOVA***

09:00–09:15	Cavitating encephalopathy in a child with methylmalonic acidemia and homocysteinemia DR. RALITSA YORDANOVA, PROF. ILIANA PACHEVA, DR. KATERINA GABEROVA, DR. IGLIKA SOTKOVA-IVANOVA, DR. DANIELA CHECHEVA, M. IVANOVA, PROF. IVAN IVANOV
09:15–09:30	Classical form of Pelizaeus-Merzbacher disease with a mutation in the PLP1 gene DR. KATERINA GABEROVA, PROF. ILIANA PACHEVA, PROF. IVAN IVANOV
09:30–09:45	Lesch-Nyhan syndrome DR. BORYANA HRISTOVA, PROF. DANIELA AVDZHIEVA-TZAVELLA
09:45–10:00	Loeys-Dietz syndrome DR. VIOLETA KOLEVA
10:00–10:15	Difficulties in the path of diagnosis after whole-exome sequencing DR. MARIA LEVKOVA
10:15–10:30	Fatty acid oxidation defect – clinical case DR. ANDREY GONCHAROV, ASSOC. PROF. YORDANKA UZUNOVA
10:30–10:45	Role of genomic testing to refine the diagnosis of prenatally diagnosed fetal polycystic kidney disease DR. SLAVENA NIKOLOVA, PROF. KATYA KOVACHEVA, DR. ZORNITSA KAMBUROVA, DR. MAGDALENA IVANOVA, DR. IVANA STANKOV
10:45–10:55	Discussion

10:55 **Closing**