



12th BALKAN CONGRESS
OF HUMAN GENETICS

8th NATIONAL CONFERENCE
FOR RARE DISEASES

RARE DISEASES – NEW HORIZONS FOR SCIENTIFIC RESEARCH

8 - 10 September 2017
Grand Hotel Plovdiv, Bulgaria

September 8 (Friday)					
12.00 – 19.00	Registration				
15.00 – 17.30	<p>OPENING SESSION Moderators: Draga Toncheva, Rumen Stefanov</p> <p><i>Official messages (to be confirmed)</i></p> <p>Plenary lectures: Draga Toncheva. Development of genomic medicine in Bulgaria Rumen Stefanov. State-of-art of rare disease policy in Bulgaria and the EU Aspazija Sofijanova. Rare diseases in Macedonia: from in the middle of nowhere to the point of a part of national programme Vladimir Tomov. Rare diseases and genomic medicine – patients’ perspective Dragan Primorac. Stem cell application for osteoarthritis in the knee join</p>				
17.30 – 18.00	Coffee break				
18.00 – 19.30	<table border="1"> <thead> <tr> <th>SESSION 1 Moderators: Draga Toncheva, Rumen Stefanov</th> <th>PARALLEL SYMPOSIUM 1 Systemic lupus erythematosus (supported by GSK)</th> </tr> </thead> <tbody> <tr> <td> <p>Plenary lectures: Borut Peterlin. Genomic medicine in health care systems: the time for change Kanay Yararbas. Interpreting next generation sequencing and array data of patients with rare conditions Ugur Ozbek. Rare diseases in Turkey Karin Writzl. Next generation clinical genetics: genotype first or phenotype first approach Jasmina Koeva. Precision and personalized medicine – a promise or an imperative</p> </td> <td> <p>Oral presentations: Velichka Popova. Mixed diseases of the connective tissue – systemic lupus erythematosus [in Bulgarian]</p> </td> </tr> </tbody> </table>	SESSION 1 Moderators: Draga Toncheva, Rumen Stefanov	PARALLEL SYMPOSIUM 1 Systemic lupus erythematosus (supported by GSK)	<p>Plenary lectures: Borut Peterlin. Genomic medicine in health care systems: the time for change Kanay Yararbas. Interpreting next generation sequencing and array data of patients with rare conditions Ugur Ozbek. Rare diseases in Turkey Karin Writzl. Next generation clinical genetics: genotype first or phenotype first approach Jasmina Koeva. Precision and personalized medicine – a promise or an imperative</p>	<p>Oral presentations: Velichka Popova. Mixed diseases of the connective tissue – systemic lupus erythematosus [in Bulgarian]</p>
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20.30 – 23.00	Gala dinner				

September 9 (Saturday)	
8.30 – 16.00	Registration
9.00 – 10.30	<div style="display: flex; justify-content: space-between;"> <div style="width: 48%;"> <p style="text-align: center;">SESSION 2</p> <p>Moderators: Rada Staneva, Ralitsa Yordanova</p> <p>Plenary lectures: Giovanni Neri. RASopathies: clinical and molecular correlations Emilia Severin. Cherubism – a rare genetic disorder Anastas Pashov. Rational design of mimotope library for IgM repertoire studies</p> <p>Oral presentations: Olena Grechanina. A rare disease of the mitochondrial respiratory chain – 3-methylglutaconic aciduria. Approach to diagnosis and rehabilitation Yulia Grechanina. Selective screening of mitochondrial dysfunction in a region with a high level of neurological diseases</p> </div> <div style="width: 48%;"> <p style="text-align: center;">PARALLEL SYMPOSIUM 2</p> <p style="text-align: center;">Rare lysosome storage diseases (supported by Sanofi/Genzyme) Moderators: Ivaylo Tournev</p> <p>Oral presentations: Teodora Chamova. Pompe disease – a treatable myopathy Emil Paskalev. Fabry disease – 10 years of experience of the Bulgarian expert centre [in Bulgarian] Zlate Stojanoski. Myeloproliferative neoplasm in patients with Gaucher disease</p> </div> </div>
10.30 – 11.00	Coffee break
11.00 – 12.30	<div style="display: flex; justify-content: space-between;"> <div style="width: 48%;"> <p style="text-align: center;">SESSION 3</p> <p style="text-align: center;">From cost to value (supported by Novartis) Moderators: Ana Pejčić</p> <p>Plenary lectures: Stamen Popov. From cost to value Silvia Vandeva. Quality of life in acromegaly Natalia Grigorova. Acromegaly – psychological consequences for the patient</p> </div> <div style="width: 48%;"> <p style="text-align: center;">PARALLEL SYMPOSIUM 3</p> <p style="text-align: center;">Hemophilia (supported by Shire) Moderators: Valeria Kaleva</p> <p>Oral presentations: Valeria Kaleva. New opportunities for personalized prophylaxis in patients with hemophilia A with Advate [in Bulgarian] Stefan Goranov. Feiba – review of clinical trials (Pro-FEIBA, PROOF, FENOC) [in Bulgarian] Denka Stoyanova. Overview of thromboembolic events in congenital hemophilia, reported with Feiba over the last four decades [in Bulgarian]</p> </div> </div>
12.30 – 13.30	Lunch break
13.30 – 14.30	Poster session 1
	Poster jury: Dijana Plaseska Karanfilaska, Borut Peterlin, Savina Hadjidekova
14.30 – 16.00	<div style="display: flex; justify-content: space-between;"> <div style="width: 48%;"> <p style="text-align: center;">SESSION 4</p> <p>Moderators: Vily Stoyanova, Dimitrina Konstantinova</p> <p>Plenary lectures: Radoslava Vazharova. Application of targeted NGS in diagnostic work-up of patients with rare diseases Savina Hadjidekova. Pre-implantational</p> </div> <div style="width: 48%;"> <p style="text-align: center;">PARALLEL SYMPOSIUM 4</p> <p style="text-align: center;">Rare immune-mediated diseases – new horizons (in collaboration with BACI) Moderators: Mariana Murdjeva, Marta Baleva</p> <p>Plenary lectures: Mariana Murdjeva, Marta Baleva. Rare</p> </div> </div>

	<p>genetic testing for monogenic and chromosomal disorders: the experience in Bulgaria</p> <p>Oral presentations: Maya Atanasoska. Pre-implantation genetic diagnosis in a family with Waardenburg syndrome Momchil Rizov. Application of next-generation sequencing for balanced translocations in pre-implantation embryos Trifon Chervenkov. Stem cell transplantation – present and future Desislava Nesheva. Ancient mtDNA studies on Thracian and Proto-Bulgarian samples: new perspectives on the origin of contemporary Bulgarians</p>	<p>immune-mediated diseases in Bulgaria – the challenge continues [in Bulgarian] Snezhina Mihaylova. Molecular diagnostics and genetic screening of primary immune deficiencies [in Bulgarian] Nevena Gesheva. Study of the signaling pathways of T- and B-cell activation in common variable immunodeficiency - sight to the pathogenesis of disease [in Bulgarian]</p> <p>Oral presentations: Todorka Beleva-Popova. Firazyr (icatibant) for treatment of hereditary angioedema (supported by Shire) [in Bulgarian] Petya Yankova. Ataxia telangiectasia – case report [in Bulgarian] Neofit Spasov. Homozygous MyD88 deficiency – case report and literature review [in Bulgarian]</p>
16.00 – 16.30	Coffee break	
16.30 – 18.00	<p style="text-align: center;">SESSION 5-I</p> <p>Moderators: Ugur Ozbek, Dragomira Nikolova</p> <p>Plenary lectures: Dijana Plaseska Karanfilska. Phenotype and genotype heterogeneity of thalassemia intermedia Valeria Kaleva. Non-transfusion-dependent thalassaemia Galina Kurteva. Colorectal carcinoma – from genetic markers, response predictors for the treatment to personalized therapy and new genetic classification</p> <p>Oral presentations: Emilija Shukarova Stefanovska. Molecular defects determined among hemophilia patients in Republic of Macedonia Radka Kaneva. Molecular profiling of Hereditary Breast and Ovarian Cancer in Bulgaria Katerina Popovska-Jankovic. Differential expression of 12 microRNAs in breast cancer and their potential use as markers for different clinicopathologic features</p>	<p style="text-align: center;">SESSION 5-II</p> <p>Moderators: Emilia Severin, Giovanni Neri</p> <p>Oral presentations: Ivaylo Tournev. TTR FAP in Balkan countries Stayko Sarafov. TTR FAP in Bulgaria Mariana Gospodinova. Cardiac involvement in patients with hereditary transthyretin amyloidosis associated with Glu89Gln mutation and its impact on prognosis Andrey Kirov. Genetic screening for transthyretin amyloidosis in Bulgaria. Genetic profile of TTR-FAP: Glu89Gln founder effect Albena Todorova. Genetic profile of TTR-FAP in Bulgaria: parent-of-origin difference in penetrance</p>

September 10 (Sunday)		
9.00 – 10.30	<p style="text-align: center;">SESSION 6-I</p> <p style="text-align: center;">Moderators: Albena Todorova, Dimitrina Konstantinova</p> <p>Plenary lectures: Ivaylo Tournev. Neuromuscular disorders in Roma (Gypsies) Robert Penchovsky. Engineering integrated digital circuits with allosteric ribozymes for scaling up molecular computation and diagnostics of rare diseases</p> <p>Oral presentations: Katya Kovacheva. Pleven registry of congenital anomalies in the EUROCAT network Sara Radunovic. Genetic study of achondroplasia in Serbian population Hristo Ivanov. Dysregulated pathways in autism spectrum disorder Angelina Mandadzhieva. Genetically proven cases of Wolman disease in Bulgaria and mutation screening of two presumable endemic regions Marija Brankovic. NPC1 and NPC2 gene analysis in Serbian patients with Niemann-Pick disease type C.</p>	<p style="text-align: center;">SESSION 6-II</p> <p style="text-align: center;">Moderators: Karin Writzl, Olga Boyanova</p> <p>Plenary lectures: Sena Karachanak-Yankova. Genome structure of modern Bulgarians Mariya Petrova. Medical legal challenges in genetic research Ana Pejic. Orphan drugs in Serbia: evaluation of market authorization, pricing, reimbursement and expenditure</p> <p>Oral presentations: Borislav Popov. Genetic testing and insurance Veselina Petrova-Tacheva. Genetic research, family and family relations Rada Staneva. The era of precision medicine – a basic guide of how to navigate the field</p>
10.30 – 11.00	Coffee break	
11.00 – 12.30	<p style="text-align: center;">Session 7-I</p> <p style="text-align: center;">Moderators: Dijana Plaseska Karanfilaska, Ugur Ozbek</p> <p>Plenary lectures: Gueorgui Balatzenko. Monitoring of molecular response during the therapy with nilotinib as first-line tyrosine kinase inhibitor in patients with chronic myeloid leukemia.</p> <p>Oral presentations: Zora Hammoudeh. Pharmacogenetic studies in patients with cancer Marica Pavkovic. Association of Fc gamma receptor polymorphisms with autoimmune hemolytic anaemia Marija Dusanovic Pjevic. Analysis of the association between PAI-1 gene 4G/5G polymorphism and efficacy of thrombolytic therapy in patients with ischemic stroke Milka Grk. Association of ADORA2A gene rs2298383 polymorphism with efficacy/toxicity of MTX</p>	<p style="text-align: center;">Session 7-II</p> <p style="text-align: center;">Moderators: Ivaylo Tournev, Radostina Simeonova</p> <p>Oral presentations: Teodora Chamova. Autosomal recessive neurologic disorders among Bulgarian Muslims Sashka Zhelyazkova. Clinical and genetic study of Huntington’s disease: the 9 years of experience of our team Stayko Sarafov. Genetic forms of amyotrophic lateral sclerosis in Bulgaria Kristina Kastreva. Charcot-Marie-Tooth: ethnic differences, genetic and clinical spectrum in Bulgaria. Jean Samuel. The genetic epidemiology of hereditary spastic paraplegias in Bulgaria Tihomir Todorov. NGS approach in cases with congenital neuromuscular disorders Olga Antonova. Screening for SNPs in a set of drug-metabolizing enzymes in Bulgarian population</p>

	Suada Mucaj. Identification of CYP2C19*2 allelic variant in healthy Albanian population	Darina Kachakova. NGS sequencing in service of neurogenetics in Bulgaria
12.30 – 13.30	Lunch break	
13.30 – 14.30	Poster session 2 Poster jury: Dijana Plaseska Karanfilska, Borut Peterlin, Savina Hadjidekova	
14.30 – 16.00	<p style="text-align: center;">Session 8-I</p> <p>Moderators: Katya Kovacheva, Borislav Popov</p> <p>Oral presentations: Slavica Josifovska. Genetic heterogeneity of cardio vascular diseases associated with pathology of great vessels. Mariana Gospodinova. Genetics and cardio-vascular diseases Elena Sukarova Angelovska. Recognition of syndromic forms of disorders of sexual differentiation Valentin Penchev. Genetic background of steroid-resistant nephrotic syndrome in Bulgaria Olga Beltcheva. Molecular basis of developmental disorders: a view through the kidney filter Mila Baycheva. Diagnostic and therapeutic approach in children with biliary atresia</p>	<p style="text-align: center;">Session 8-II</p> <p>Moderators: Lyudmila Angelova, Trifon Chervenkov</p> <p>Oral presentations: Zoran Gucev. Rare Diseases, new genes, molecular mechanisms and treatments Ana Marjanovic. Study of ATXN2 repeat length in C9ORF72 expansion Grigor Zoraqi. Spectrum of mutations in the CFTR gene of Albanian cystic fibrosis patients Mariya Glushkova. Genetically verified tuberous sclerosis complex in a cohort of fifteen Bulgarian families Dragomira Nikolova. Molecular profiling of papillary thyroid cancer by RNA expression and NGS sequencing platforms Silva Giragosyan. Genetic profiling of advanced laryngeal carcinoma by NGS</p>
16.00 – 17.00	Closing session Moderators: Draga Toncheva, Rumen Stefanov	

September 9 (Saturday)	
13.30 – 14.30	Poster session 1 Poster jury: Dijana Plaseska Karanfilska, Borut Peterlin, Savina Hadjidekova

PP-01. Kavcan I et al. MULTIDISCIPLINARY APPROACH AND SPEECH, OCCUPATIONAL, AND PHYSICAL THERAPY IN A CASE OF JOUBERT SYNDROME

PP-02. Kulbalaeva Sh et al. THE CASE OF THE TERMINAL DELETION OF LONG ARM OF THE CHROMOSOME 3

PP-03. Koprululu G et al. MOLECULAR ANALYSIS OF AZF (AZOOSPERMIA FACTOR) GENE MICRODELETIONS WITH QUADRUPLEX REAL TIME POLYMERASE CHAIN REACTION METHOD IN INFERTILE TURKISH MALES

PP-04. Pavkovic M et al. FCGR2A AND FCGR3A VARIANTS ARE NOT ASSOCIATED WITH RESPONSE TO RITUXIMAB IN PATIENTS WITH B-CELL CHRONIC LYMPHOCYTIC LEUKEMIA

PP-05. Pavkovic M et al. CYTOTOXIC T-LYMPHOCYTE ANTIGEN-4 GENE POLYMORPHISM AND THE RISK FOR CHRONIC LYMPHOCYTIC LEUKEMIA IN MACEDONIA POPULATION

PP-06. Pavkovic M et al. TUMOR NECROSIS FACTOR GENE POLYMORPHISMS AND THE RISK FOR CHRONIC LYMPHOCYTIC LEUKEMIA IN MACEDONIA POPULATION

PP-07. Gok I et al. BETA-1 ADRENERGIC RECEPTOR GENE POLYMORPHISMS WITH OBSTRUCTIVE SLEEP APNEA SYNDROME IN EAST OF TURKEY

PP-08. Gatseva P et al. SHORT-BOWEL SYNDROME IN THE NEONATAL PERIOD

PP-09. Angelovic R et al. DIAGNOSIS OF MYELOPROLIFERATIVE NEOPLASMS USING MOLECULAR METHODS FROM SINGLE CENTRE EXPERIENCE

PP-10. Arangelovic E et al. CYTOTOXIC T-LYMPHOCYTE ANTIGEN-4 GENE POLYMORPHISM AND THE RISK FOR TYPE 2 DIABETES IN MACEDONIA POPULATION

PP-11. Biletska S. METABOLISM OF VITAMIN D SYSTEM IN CHILDREN WITH AUTISM SPECTRUM DISORDERS

PP-12. Grechanina O et al. THE CASE OF A COMBINATION OF FOLATE TRANSPORTER, METHYLENETETRAHYDROFOLATE REDUCTASE AND PYRIDOXINE DEFICIENCY IN CHILD WITH EPILEPSY AND PSYCHOMOTOR AND SPEECH RETARDATION

PP-13. Popovic B. ESTIMATION OF RELATIVE TELOMERE LENGTH IN DIFFERENT MSCS

PP-14. Petrova-Tacheva V et al. COMPARATIVE STUDY OF THE EFFECT OF TOTAL EXTRACT FROM HABERLEA RHODOPENSIS LEAVES AND VITAMIN C ON THE CELL VIABILITY OF LYMPHOCYTES ISOLATED FROM PERIPHERAL HUMAN BLOOD

PP-15. Mazurenko L et al. THE MLL REARRANGEMENTS WITH ADDITIONAL CHROMOSOMAL ABBERATIONS IN PATIENTS UNDER ONE YEAR OLD WITH AML (TWO CASES)

PP-16. Kovacheva K et al. RING CHROMOSOME 15 SYNDROME IN A NEWBORN BABY FROM BULGARIA – A CLINICAL CASE

PP-17. Kovacheva K et al. CHROMOSOMAL ABNORMALITIES AND Y CHROMOSOME MICRODELETIONS IN BULGARIAN MALE WITH AZOOSPERMIA OR SEVERE OLIGOSPERMIA

PP-18. Ковачева К и съавт. КЛИНИЧЕН СЛУЧАЙ НА НОВОРОДЕНО МОМИЧЕ С ICHTHYOSIS CONGENITA, РЕЗУЛТАТ ОТ МУТАЦИЯ В ALOX12B ГЕНА

PP-19. Pesic M et al. THE PRESENCE OF PREMUTATION IN THE FMR1 GENE IN PATIENTS WITH CLINICAL PICTURE OF DEGENERATIVE ATAXIA, TREMOR AND PARKINSONISM

PP-20. Maksimovic N et al. THE ANALYSIS OF ENOS 4A/B POLYMORPHISM IN SURGICAL PATIENTS WITH SECONDARY PERITONITIS

PP-21. Tsaneva S et al. THE ROLE OF MOLECULAR GENETIC ANALYSIS IN THE DIAGNOSTICS OF CONGENITAL HEART DISEASES

PP-22. Kavrakova A et al. PRESENCE OF HIGH RISK HUMAN PAPILOMA VIRUSES IN NONINVASIVE URINE SAMPLES WITH MOLECULAR PROFILE FLUCTUATIONS FOR PROSTATE CANCER, CYTOLOGICALLY CONFIRMED

PP-23. Tincheva S et al. GENETIC TESTING IN COELIAC DISEASE – OUR EXPERIENCE IN BULGARIA

PP-24. Nikolova E et al. MOLECULAR DIAGNOSTIC APPROACH IN NEURO-ONCOLOGY

PP-25. Yordanova I et al. ARSA GENE TESTING: METACHROMATIC LEUKODYSTROPHY OR ARSA PSEUDODEFICIENCY?

PP-26. Kadiyska T et al. ROLE OF THE FTO GENE POLYMORPHISM RS9939609 IN BULGARIAN OBESE ADULTS IN THE DEVELOPMENT OF PREDIABETES

PP-27. Dodova R et al. SCREENING OF BRCA1 AND BRCA2 GENES IN BULGARIAN OVARIAN CANCER PATIENTS

PP-28. Molodan L et al. THE CASE OF A COMBINATION OF WILSON-KONOVALOV DISEASE AND HEMOCHROMATOSIS, CAUSED BY HETEROZYGOUS CARRIAGE OF THE MUTATIONS C282Y AND H63D OF THE GENE OF HEMOCHROMATOSIS

PP-29. Grechanina O et al. THE CASE OF RARE DISEASE – BENIGN RECURRENT INTRAHEPATIC CHOLESTASIS (BRIC)

PP-30. Peycheva V et al. PRE- AND POSTNATAL DIAGNOSTICS OF GENOMIC DISORDERS IN CHROMOSOMES 17 AND 22

- PP-31. Popov I et al.** MUTATIONAL BURDEN IN BULGARIAN SCHIZOPHRENIA AND BIPOLAR DISORDER PATIENTS: A BIOINFORMATICS ANALYSIS
- PP-32. Ivanov H et al.** BUDD-CHIARI SYNDROME IN A PATIENT WITH MUTATIONS IN JAK-2, FACTOR V AND MTHFR
- PP-33. Ivanov H et al.** DE NOVO C.721A>C, P.(THR241PRO) MUTATION IN BRAF GENE IN PATIENT WITH RASOPATHY
- PP-34. Katarova M et al.** BRUCK TYPE 1 SYNDROME DUE TO A MUTATION C.831DUPC IN FKBP10 GENE IN BULGARIAN ROMA
- PP-35. Chamova T et al.** NOVEL FORM OF COMPLICATED HEREDITARY SPASTIC PARAPLEGIA (SPG78), DUE TO MUTATIONS IN THE ATP13A2/PARK9 GENE
- PP-36. Chamova T et al.** CLINICAL AND GENETIC HETEROGENEITY OF MYOPATHIES IN BULGARIA
- PP-37. Chamova T et al.** AUTOSOMAL-RECESSIVE CONGENITAL CEREBELLAR ATAXIA IS CAUSED BY MUTATIONS IN METABOTROPIC GLUTAMATE RECEPTOR 1
- PP-38. Колева Р.** ЗАХАРЕН ДИАБЕТ И ЛОШ АПЕТИТ – КАКВА Е ДИАГНОЗАТА?
- PP-39. Penchovsky R.** PROGRAMMABLE AND AUTOMATED BEAD-BASED MICROFLUIDICS FOR VERSATILE DNA MICROARRAYS FOR GENOMIC ANALYSES UNDER ISOTHERMAL CONDITIONS
- PP-40. Vozhilova R et al.** RARE GENETIC VARIANTS IDENTIFIED BY NGS CONTRIBUTE TO BOTH SCHIZOPHRENIA AND BIPOLAR DISORDER
- PP-41. Дамянова В и съавт.** КЛИНИЧЕН СЛУЧАЙ НА ОСТЕОПЕТРОЗА С НЕГАТИВЕН РЕЗУЛТАТ ЗА JAK-2 МУТАЦИЯ
- PP-42. Cherninkova S et al.** CLINICAL SYMPTOMATOLOGY AND GENETIC INVESTIGATION IN PATIENTS WITH LEBER'S HEREDITARY OPTIC NEUROPATHY
- PP-43. Emilova R et al.** RESULTS FROM CYTOGENETIC STUDIES IN PATIENTS WITH REPRODUCTIVE FAILURE (A 20-YEAR EXPERIENCE)
- PP-44. Mihaylova M et al.** APPLICATION OF MOLECULAR KARYOTYPING IN THE DIAGNOSIS OF 10Q MICRODELETION SYNDROME
- PP-45. Zoraqi G et al.** GENETIC POLYMORPHISM DATA ON 15 AUTOSOMAL STR MARKERS IN ALBANIANS FROM ALBANIA
- PP-46. Tsvetkova M et al.** CASES OF WOLF-HIRSCHHORN SYNDROME DIAGNOSED THE LABORATORY OF MEDICAL GENETICS – VARNA FOR THE PERIOD 2008-2017
- PP-47. Zoraqi G et al.** GENETIC STRUCTURE OF ALBANIAN GIPSY POPULATION BY 15 STR IDENTIFILER MARKERS

PP-48. Konstantinova D et al. GENETIC MARKERS FOR IMMUNE INTOLERANCE IN REPRODUCTIVE FAILURE – A 5 YEARS' EXPERIENCE IN VARNA

PP-49. Kischeva A et al. GENETIC MARKERS FOR THROMBOPHILIA IN PATIENTS WITH CEREBROVASCULAR ACCIDENT AND PERSISTENT FORAMEN OVALE

PP-50. Zoraqi G et al. PRELIMINARY RESULTS OF GENETIC VARIANT CYP1A2 1*F, INVOLVED IN CAFFEINE INTOLERANCE, IN ALBANIAN POPULATION

September 10 (Sunday)	
13.30 – 14.30	Poster session 2 Poster jury: Dijana Plaseska Karanfilska, Borut Peterlin, Savina Hadjidekova

PP-51. Miteva V et al. BECKWITH-WIEDEMANN SYNDROME IN FOUR UNRELATED CASES – FROM THE GENETIC COUNSELING IN MBAL “ST. MARINA” VARNA

PP-52. Dimova I et al. GENOMIC DISORDERS IN THE CONTEXT OF PRENATAL AND POSTNATAL DIAGNOSTICS IN BULGARIA

PP-53. Stoyanova M et al. GENETIC DIAGNOSTIC SURVEY ON CHILDREN WITH CONGENITAL AND HEREDITARY DISEASES FOR A PERIOD OF TWO YEARS

PP-54. Kastreva K et al. CLINICAL VARIABILITY OF CONGENITAL MYASTHENIC SYNDROME TYPE IA DUE TO MUTATION 1267DELG IN 100 CASES

PP-55. Petkova V et al. ANALYSIS OF SOMATIC MUTATIONS IN LUNG ADENOCARCINOMAS AND SQUAMOUS CELL CARCINOMAS WITH TARGETED NGS

PP-56. Atanasoska M et al. A RARE MUTATION OF SCN8A GENE IN A PATIENT WITH EARLY INFANTILE EPILEPTIC ENCEPHALOPATHY

PP-57. Kaneva R et al. MOLECULAR MARKERS WITH DIAGNOSTIC AND PROGNOSTIC VALUE FOR TREATMENT OF PEDIATRIC SOLID TUMOURS

PP-58. Mucaj S et al. IDENTIFICATION OF CYP2C9*2 ALLELIC VARIANT IN HEALTHY ALBANIAN POPULATION

PP-59. Ignatova I et al. CREATING HAEMOPHILIA COMPREHENSIVE CARE CENTRE AT ST. MARINA UNIVERSITY HOSPITAL – VARNA

PP-60. Taneva A et al. CLINICAL AND GENETIC SPECTRUM OF LIMB-GIRDLE MUSCULAR DYSTROPHIES (LGMD) IN BULGARIA

PP-61. Grigorova N. NEUROFORMA – INNOVATIVE APPROACH OF REHABILITATION FOR NEUROLOGICAL DISEASES AND INJURIES

PP-62. Pavel A et al. BLOOD BIOMARKERS FOR RISK PREDICTION OF AGGRESSIVE PROSTATE CANCER PATIENTS – PRELIMINARY STUDY OF A ROMANIAN COHORT

PP-63. Мандова В и съавт. ОРТОДОНТСКО ЛЕЧЕНИЕ ПРИ ПАЦИЕНТ СЪС СИНДРОМ НА ТЪРНЪР: ПРЕДСТАВЯНЕ НА КЛИНИЧЕН СЛУЧАЙ

PP-64. Mihova K et al. FIRST SYSTEMATIC GENETIC STUDY OF BULGARIAN PATIENTS WITH PARKINSON DISEASE

PP-65. Ivanov S et al. WHOLE-GENOME SEQUENCING IN NEWBORN SCREENING – PRELIMINARY RESULTS ON MEDICAL PROFESSIONALS’ ATTITUDES AND OPINIONS IN BULGARIA

PP-66. Anastasovska V et al. MOLECULAR DETECTION OF VIRUS HERPES SIMPLEX TYPE 1 IN PATIENTS WITH PERIODONTAL DISEASE

PP-67. Tsochev K et al. CRITICALLY ILL MSUD PATIENT - CHALLENGES, WAYS TO PREVENT AND OUTCOME

PP-68. Stoycheva R et al. A 17-YEAR-OLD BOY WITH LEOPARD SYNDROME

PP-69. Emilova R et al. RESULTS FROM CYTOGENETIC ANALYSIS IN CHILDREN WITH SUSPECTED CHROMOSOMAL DISORDERS (A 10-YEAR EXPERIENCE). THREE CYTOGENETICS CASE REPORTS

PP-70. Balabanski L et al. THE BULGARIAN CENTENARIAN GENOME PROJECT

PP-71. Вълчева М и съавт. КЛИНИЧНИ СЛУЧАИ НА ПАЦИЕНТИ С МНОЖЕСТВЕНИ МЕТАХРОННИ НЕОПЛАЗИИ

PP-72. Грудева-Попова Ж и съавт. СИНХРОННО И МЕТАХРОННО ПРОТИЧАЩИ НЕОПЛАЗИИ

PP-73. Исмаил Е и съавт. АТИПИЧНА КЛИНИКА ПРИ ИНФЕКЦИОЗНА МОНОНУКЛЕОЗА

PP-74. Chamova T et al. NIEMANN-PICK DISEASE TYPE B AND TYPE C IN BULGARIA – GENETIC AND CLINICAL CHARACTERISTICS

PP-75. Duca S et al. IDENTIFICATION OF HLA B57:01 ALLELE, CONFERRING HYPERSENSIBILITY REACTION TO ABACAVIR, IN HLAB57 INDIVIDUALS OF ALBANIAN POPULATION BY AS-PCR

PP-76. Станчева-Иванова М и съавт. СИНДРОМ НА ПРАЗНАТА СЕЛА – ОПИСАНИЕ НА КЛИНИЧЕН СЛУЧАЙ

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