CROATIAN SOCIETY OF HUMAN GENETICS

Croatian Medical Association

Croatian Society of Human Genetics Croatian Medical Association

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THE FIFTH DAYS OF HUMAN GENETICS - PROF. LJILJANA ZERGOLLERN-ČUPAK

Dubrovnik Palace Hotel/Dubrovnik, June 20, 2017

SCIENTIFIC PROGRAMEE

8:00 a.m.-11:30 a.m Co-Chairs: Dragan Primorac and Ingeborg Barišić

Oral session

8:00-8:20 Alenka Gagro, Martina Krmek, Goran Roić, Amarela Lukić-Grlić, Ana Budimir, Goran Tešović: **MOLECULAR CHARACTERIZATION OF PANTON-VALENTINE LEUKOCIDIN-POSITIVE METHICILLIN-SUSCEPTIBLE STAPHYLOCOCCUS AUREUS IN A CHILD WITH OSTEOMYELITIS COMPLICATED BY DEEP VENOUS THROMBOSIS OF POPLITEAL VEIN**

8:20-8:40 Adriana Bobinec, Ana-Maria Ivankov, Ljubica Boban, Ivona Sansović, Ingeborg Barišić: **PHENOTYPIC FEATURES OF 15q11.2-q13.3 COPY NUMBER VARIATIONS**

8:40-9:00 Lucija Ružman, Jelena Radić Nišević, Igor Prpić: **VANISHING WHITE MATTER DISEASE: FROM CLINICS TO GENETICS**

9:00-9:20 Ana-Maria Ivankov, Adriana Bobinec, Ivona Sansović, Ljubica Boban, Ingeborg Barišić: **EPILEPSY AND COPY NUMBER VARIANTS: INDICATIONS FOR CHROMOSOMAL MICROARRAY ANALYSIS**

9:20-9:40 Ljubica Boban, Adriana Bobinec, Ana-Marija Ivankov, Ivona Sansović, Ingeborg Barišić: **REGISTERY OF PATIENTS WITH AUTISM SPECTRUM DISORDERS**

9:40-10:00 **Discussion**

10:00-11.30 a.m. Poster session

Ingeborg Barišić, Joan K. Morris, Maria Loane, Ester Garne, Judith Rankin, James Densem, Amanda J. Neville, Anna Pierini, Hermien de Walle: EUROlinkCAT - **ESTABLISHING A LINKED EUROPEAN COHORT OF CHILDREN WITH CONGENITAL ANOMALIES**

Ljubica Boban, Ingeborg Barišić, EUROCAT Working group: **WEB SITE OF CROATIAN EUROCAT REGISTRY OF CONGENITAL ANOMALIES**

Adriana Bobinec, Ana-Maria Ivankov, Ljubica Boban, Ivona Sansović, Ingeborg Barišić: **GENOTYPE-PHENOTYPE CORRELATION IN RARE 17q24.1-q24.3 MICRODELETION**

Kristina Crkvenac Gornik, Ivana Tonkovic Durisevic, Sanda Huljev Frkovic, Iva Kolombo, Ela Brgodac, Vjekoslav Krzelj, Bernarda Lozić: **RUSSELL - SILVER SYNDROME – CLINICAL AND MOLECULAR PERSPECTIVE**

Alenka Gagro, Goran Roić, Ivona Sansović, Ana-Maria Ivankov, Adriana Bobinec, Darko Antičević, Ingeborg Barišić**: INFLAMMATORY POLYARTHRITIS IN PATIENT WITH 18q DELETION SYNDROME**

Sanda Huljev Frković, Daniel Franjić, Jadranka Kelečić, Mia Šalamon Janečić, Kristina Crkvenac Gornik, Marijan Frković: **NIJMEGEN BREAKAGE SYNDROME**

Ana-Maria Ivankov, Adriana Bobinec, Ivona Sansović, Ljubica Boban, Ingeborg Barišić: **A 5.5-YEAR OLD BOY WITH A *DE NOVO* COMPLEX 2q32q35 CHROMOSOMAL REARRANGEMENT**

Valentina Karin, Vedran Kardum, Mislav Glibo, Anita Skrtic,Tamara Nikuseva-Martic, Nermina Ibisevic, Faruk Skenderi, Semir Vranic, Ljiljana Serman: **METHYLATION-ASSOCIATED SILENCING OF *SFRP1* GENE IN HIGH-GRADE SEROUS OVARIAN CARCINOMAS**

Mijana Kero, Maja Tomasovic, Mirjana Jukica, Futoshi Sekiguchi, Noriko Miyake, Naomichi Matsumoto, Bernarda Lozic: **HEREDITARY SPASTIC PARAPLEGIA TYPE 4 WITH A NOVEL SPAST MUTATION WITH CLINICAL ONSET IN MALE INFANT**

Ružica Lasan-Trčić: **CLINICAL CYTOGENETICS FROM KARYOTYPE TO GENOME SEQUENCE**

Ivan Lekić, Ivana Škrlec, Sanela Štibi, Jasenka Wagner: **MOLECULAR GENETIC ANALYSIS OF *AR* GENE AMONG PATIENTS WITH ANDROGEN INSENSITIVITY SYNDROME**

Sergej Nadalin, Jelena Rebić, Klementina Ružić, Ante Prpić, Miljenko Kapović, Alena Buretić-Tomljanović: **POLYMORPHISMS IN THE *PLA2G4A* AND *PLA2G6* GENES AND NICOTINE DEPENDENCE IN SCHIZOPHRENIA PATIENTS**

Silvija Pušeljić, Višnja Tomac, Nora Pušeljić, Diana Milas, Vesna Milas: **MARSHALL SYNDROME AUTISM, PROFUND INTELECTUAL DISABILLITY, HEARING LOSS AND AMBLIOPIA - CASE REPORT**

Smiljana Ristić, Nada Starčević Čizmarević, Polona Lavtar, Luca Lovrečić, Olivio Perković, Juraj Sepčić, Saša Šega Jazbec, Miljenko Kapović , Borut Peterlin: **ANGIOTENSIN-CONVERTING ENZYME (ACE) I/D GENE POLYMORPHISM AND IFN-β TREATMENT RESPONSE IN MULTIPLE SCLEROSIS PATIENTS**

Ivana Skrlec, Višnja Tomac, Silvija Pušeljić, Mia Galić, Karmela Barišić, Jasenka Wagner: **VALIDATION OF THE SCREENING METHOD FOR DYNAMIC MUTATIONS IN THE *FMR1* GENE**

Višnja Tomac, Silvija Pušeljić, Jasenka Wagner, Ivana Škrlec, Mirna Anđelić, Dunja Čokolić-Petrović, Martina Kos, Nora Pušeljić, Diana Milas, Vesna Milas: **A RARE *DE NOVO* DUPLICATION 21q22.3 SYNDROME - CASE REPORT**

Jadranka Vraneković, Ivana Babić Božović, Bojana Brajenović Milić: **METHYLENETETRAHYDROFOLATE REDUCTASE DIMER CONFIGURATION AND CHROMOSOME 21 NONDISJUNCTION**