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Presenting author names **are bolded** in the contributor lists.

E-POSTERS

P01 Reproductive Genetics/Prenatal Genetics

P01.001.A Frequency of Y chromosome microdeletions in Turkish infertile men: Single Center Experience

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Objective: Y chromosome microdeletions are the leading genetic cause of male infertility and their detection is clinically relevant for appropriate genetic counseling. Y chromosome includes genes for testicular development and spermatogenesis. The aim of this study was to establish the frequency of the Y chromosome microdeletions in Turkish infertile men who referred to our center with severe oligozoospermia and azoospermia.

Materials and Methods: In our study, 396 infertile men referred to İstanbul University- Cerrahpaşa, Cerrahpaşa Medical Faculty Department of Medical Genetics (GETAM) between 2016 to 2020 with azoospermia/severe oligospermia. We evaluated microdeletions of the Y-chromosome STS markers AZFa, AZFb and AZFc, ZFX/ZFY, terminal sY160 regions by using DNA Fragment analysis.

Results: Among the 396 infertile men, we determined 30 cases of Y chromosome micro- deletions (7.57%). Among 30 cases, AZFc microdeletions were found in 18 cases (60%), AZFa microdeletions in 4 cases (13.3%), AZFb microdeletions in 1 case (3.3%), AZFa,b,c in 4 cases (13.3%), AZFb,c in 3 cases (10%). Our findings are consistent with the literature.

Conclusion: Our results are similar to the previous studies which have mostly reported a frequency of less than 10% for Y chromosome microdeletions. The etiology of infertility remains unknown and novel genes other than y chromosome microdeletions should be identified with high throughput techniques.

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P01.002.B Serotonin transporter 5-HTTLPR genotypes and trinucleotide repeats of androgen receptor exert a combinatorial effect on hormonal milieu in patients with lifelong premature ejaculation

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Premature ejaculation is one of the most common sexual disorders in men due to the uncontrolled modulation of spinal reflexes. In this study, we investigate the combinatorial effects of trinucleotide repeats of androgen receptor and allelic variants of the 5-HTTLPR gene on sex steroids, hypophyseal hormones, sexual performance, and premature ejaculation assessment parameters among evidence-based lifelong premature ejaculation subjects. A total of 271 patients consulting for evidence-based lifelong premature ejaculatory dysfunction were selected in this study. The control group consists of 155 men with normal IELT (>4 min). The study revealed that the subjects who have the highest (≥26) CAG stretch depicted significantly higher serum oxytocin levels

we describe the specific evolution of dysmorphic features during the different stages of life.

Conclusion: *BRD4*-related phenotype is part of the CdLS spectrum but is characterized by clinically relevant specificities that distinguish it from other cohesinopathies.

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P11.023.A miRNA-free rare pathogenic CNVs could drive toward variable CAKUT phenotypes

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Introduction: Genetic studies of congenital anomalies of the kidney and urinary tract (CAKUT) have demonstrated variable penetrability and expressivity of the associated genetic defects. Previously, it was shown that deletions of 17q12 and 22q11.2 regions were specific for kidney anomalies (KA) while 16p11.2 and 1q21.1 loci showed extensive pleiotropy in CAKUT phenotypes. CNVs affecting miRNA gene dosage have been described to have functional influence on gene expression. We aimed to conduct comprehensive in silico analysis using publicly available databases to analyze miRNA content of CAKUT-associated CNVs in quoted chromosomal loci with regard to pleiotropy.

Methods: Extensive literature review was conducted to collect data about pathogenic rCNVs associated with CAKUT. UCSC genome browser tool was employed for mapping miRNAs onto collected rCNV regions.

Results: Analysis of CNVs in CAKUT included four studies counting more than 2500 patients. In further analysis we included 191 patients harboring pathogenic CNVs. Surprisingly, CAKUT pleiotropic regions (16p11.2, 1q21.2) did not contain any miRNA. 22q11.2 showed the densest miRNAs content (n = 21).

Conclusions: Absence of miRNAs may potentially pronounce the pleiotropy of the CAKUT genetic defects, thus leading to the variety of phenotypes. Contrary, abundancy of miRNAs in 22q11.2 might be associated with reproducible phenotype, such as KA, producing the functional effect when deleted. This assumption agrees with recent results of miRNA expression variability in 22q11.2 deletion syndrome. Acknowledgements: This research was supported by the Science Fund of the Republic of Serbia, PROMIS, # 6066923, miFaDriCa, and Serbian Ministry of Education, Science and Technological development.

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P11.024.B Are miR-548 family members potential genetic drivers of CAKUT

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Introduction: miR-548 family members, located on all human chromosomes except chr19 and chrY, regulate podocyte differentiation in vitro, important for kidney development. Rare copy number variants (rCNVs) are the common genetic cause of Congenital Anomalies of the Kidney and Urinary Tract (CAKUT) and could harbour miRNAs. The aim of this study was to investigate to which extent rCNVs associated with CAKUT harbour miR-548 members.

Materials and Methods: Extensive literature review was conducted to collect data of pathogenic and likely pathogenic rCNVs in CAKUT patients. UCSC genome browser tool was employed for mapping of miR-548 members onto collected rCNV regions and gnomAD SV controls database. Bioinformatic analysis was conducted using miRPathDB2 tool.

Results: We generated CAKUT database of pathogenic CNVs in 79 chromosome regions from 191 patient and likely pathogenic CNVs in 74 regions from 87 patients. Pathogenic rCNVs of seventeen patients, located on 7 chromosomes, contained at least one miR-548 member. Likely pathogenic rCNVs of 4 patients, located on 3 chromosomes, contained one of miR-548 members. Bioinformatic analysis implied the role of mapped miRNAs in the regulation of processes associated with CAKUT. In controls, only hsa-mir-548i-3 (out of 73 precursors) was mapped on polymorphic CNVs (af>1%) and wasn't identified in patients.

Conclusions: miR-548 members located in rCNVs should be investigated in future studies as potential genetic drivers of CAKUT development, beyond protein coding genes. Acknowledgements: This research was supported by the Science Fund of the Republic of Serbia, PROMIS, #6066923, miFaDriCa, and Serbian Ministry of Education, Science and Technological development.

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P11.025.C Preeclampsia as a potential clinical feature in Cantú syndrome

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Introduction: Cantú syndrome (CS) is caused by gain-of-function pathogenic disease-causing variants in the genes coding for ABCC9 and KCNJ8, which together form an ATP-sensitive potassium channel. CS is a rare systemic syndrome with a great clinical variability, characterized by coarse facies, hypertrichosis, osteochondrodysplasia and cardiac anomalies. We present a family with eight individuals with CS for which the proband was initially diagnosed with Beckwith-Wiedemann syndrome. Whole genome trio sequencing revealed a likely pathogenic missense variant in the *ABCC9* gene (NM_005353286.2); c.1745T>A (p.Val582Asp), which the boy shares with seven similarly affected family members (patent ductus arteriosus, pericardial effusion, cardiomegaly, coarse facial features and hypertrichosis). Premature births, polyhydramnios and large for gestational age newborns are perinatal factors also seen in the family. Furthermore, maternal preeclampsia (n = 4) or hypertension during pregnancy (n = 1) is observed in 5 of 6 cases (83%) in this family, when CS mothers carry CS fetuses.

Discussion: Over-activity of the K_{ATP} channel and dysregulation of renin-angiotensin signaling (RAS) triggers cardiac hypertrophy, and dysregulation of RAS is also one of multiple factors thought to contribute to the development of preeclampsia. Here we present