ABSTRACTS COLLECTION

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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

Conclusions: Biallelic loss-of-function *P4HTM* variants were shown to cause HIDEA syndrome. Our findings enable diagnosis of the condition, and highlight the importance of assessing the need for non-invasive ventilatory support in patients. Funding: This study was supported by the Academy of Finland Grants 266719 and 308009, the S. Jusélius Foundation, the Emil Aaltonen Foundation and the Jane and Aatos Erkko Foundation to P.K.

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P08.058.D Phelan-McDermid syndrome: the use of modern methods of cytogenetic examination in the diagnosis of autism spectrum disorders

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Introduction: Phelan-McDermid syndrome (PMD) is one of the autism spectrum disorder (ASD) syndromes caused by the deletion of the terminal or interstitial parts chromosome 22q13.3. In the case of the formation of a circular chromosome without loss of material, the phenotype remains normal, but there is a risk of microdeletion in the offspring. Patients with PMD are usually seen with a diagnosis of undifferentiated mental retardation or autism.

Materials and Methods: a clinical case of Phelan-McDermid syndrome in a child with undifferentiated mental retardation. Clinical genealogical, syndromic, cytogenetic, molecular genetic methods were used.

Results: A six-year-old girl with undifferentiated mental retardation was referred for genetic counseling. Previously observed by a pediatrician, pediatric neurologist, psychiatrist for microcephaly, delayed statokinetic and psychoverbal development. Girl phenotype: dolichocephaly, high forehead, flattening of the middle part of the face, deep-set eyes, full and puffy eyelids, long eyelashes, hypertelorism, full cheeks, enlarged ears. The child exhibits autistic behavior. Genetic testing included determination of the karyotype of the proband and parents by several methods: GTG; FISH with DNA samples WCP1-22, X, Y and FISH with locus specific samples 22*SI LSI* TUPLE1, 22q13 ARSA. Result: 46,XX,r(22)(p11.2q13), Phelan McDermid syndrome, recommendations for the rehabilitation of the child were given. Maternal karyotype: 46, XX. Paternal karyotype: 46,XY,r(22), gene sequencing is recommended SHANK3.

Conclusions: The use of a complex of modern cytogenetic methods, FISH with DNA probes, or SHANK3 gene sequencing can significantly increase the number of diagnosed cases of genetically determined mental retardation and increase the effectiveness of preventive measures.

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P08.063.A Expanding the phenotype of QRICH1 associated neurodevelopmental disorder

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