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ABSTRACTS COLLECTION

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e-Posters

E-P01 Reproductive Genetics/Prenatal Genetics

E-P01.07
Epidemiological monitoring of congenital malformations in Yakutia from 2007 to 2018

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Introduction: Congenital malformations represent an urgent problem that affects the structure of infant and child mortality. In Yakutia, the prevalence of congenital malformations is recording in Register of Medical Genetic Center, Republican Hospital №1 - "National Medical Center". The purpose of epidemiological monitoring is to improve the prevention and treatment of congenital malformations.

Material and methods: The data from Medical Genetic Center Register was analyzing by the MedCalc 15.8 program.

Results: The incidence rate of congenital malformations over a 12-year period averaged 29.4 cases per 1000 newborns with a standard deviation of 2.9. It's high indicator In Russia (>20 ‰). At this period, the average annual number of births was 15725.7 (SD 1174.3). The high frequencies of malformations in 2012, 2017 and 2018 was observing (Table 1). The observed differences are statistically significantly ($p < 0.05$).

Table 1. Congenital malformations prevalence

Years	Incidences with 95% CI
2007	0.0296 (0.02694-0.03246)
2008	0.02734 (0.02479-0.03008)
2009	0.02743 (0.02492-0.03012)
2010	0.02893 (0.02636-0.03168)
2011	0.02981 (0.02723-0.03258)
2012	0.03336 (0.03067-0.03622)

E. Gumuslu[1], *K. Karaer*[2], *E. Gumus*[3], *A. Ekici*[1], *C. Kraus*[1], *A. Reis*[1]

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Autosomal recessive neurodevelopmental disorders are extremely heterogeneous and for many only few affected individuals have been described hampering delineation of the full spectrum of clinical presentation. In a large study of Turkish consanguineous families we identified a family with 3 affected siblings (two males 11 and 8 y.o. and one 5 y.o. female) all presenting with intellectual disability, severe verbal deficiency (2-3 words), short stature (<3p), delayed walking, seizures and dysmorphic features including malar hypoplasia, broad nose, short philtrum, wide mouth and full lips. After excluding aneuploidies exome sequencing revealed a novel frameshift insertion c.475_478dup, p.(Lys160Ilefs*2) in the *LARP7* gene. *LARP7* is a negative transcriptional regulator of polymerase II, acting by means of the 7SK RNP system.

Frameshifting variants in *LARP7* have been associated with Alazami Syndrome (OMIM 615071) characterized by ID, severe growth restriction and characteristic facial dysmorphisms. To date, 15 patients were reported. By comparing this family with the previously reported cases, we strengthen the key features of AS: ID, growth restriction, and highlight the facial features including malar hypoplasia, broad nose, full lips and teeth abnormalities. Interestingly, no cases with seizures were described, so far. As we failed to identify in exome sequencing any variant in seizure associated genes and all three siblings are concordant we wondered if this symptom is part of the clinical spectrum of AS. Although we cannot exclude an undetected cause independent of *LARP7*, we propose that seizures are a so far undescribed symptom associated with Alazami Syndrome.

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E-P08.03
Genomic and clinical characterisation of microduplications in a patient with developmental delay

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Introduction: The implementation of array Comparative Genomic Hybridization (aCGH) allows efficient genetic diagnosis of pathological conditions before their full clinical manifestation. Application of novel "genotype-first" diagnosis is of great importance to the patient's health and medical outcome due to higher rates of chromosomal abnormalities detection compared to conventional karyotyping and multiple ligation-dependent probe amplification (MLPA) analysis.

Materials and methods: The aCGH technique was used to determine the genetic background of developmental abnormalities in a 8-year-old female Macedonian patient. The blood-derived DNA sample was analyzed using the Affymetrix® CytoScanTM 750K Array (Applied Biosystems) that includes 550 k non-polymorphic and 200 k SNP markers. The data were interpreted by using Chromosome Analysis Suite (ChAS) Software (v4.0).

Results: The patient showed the following clinical conditions: motor delay, deafness, lack of concentration and several dysmorhological features (blepharophymosis, telecanthus, narrow palpebral fissures, wide nose, low set ears, small mouth and arched eyebrows). Noteworthy, the girl was operated from atrial septal defect and was previously diagnosed with Ohdo Sy. Karyotyping results revealed a normal female 46, XX karyotype, while the MLPA analysis for microdeletions was negative. The aCGH analysis detected a pathological microduplication occurring at the 10q26.3 cytoregion (787 kb, 26 genes included), as well as mosaic presence of 3 copies of the Xq26.3q28 cytoregion (15,864 kb, 115 genes included).

Conclusion: The present chromosomal abnormalities detected by aCGH analysis could be responsible for the developmental delay of the patient, considering their pathological expression according to ClinVar database. Further studies are needed to confirm these preliminary findings.

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E-P08.05
BRAT1-related neurodevelopmental disorders: two unrelated patients - one previously undescribed mutation

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BRAT1 gene mutations cause lethal neonatal rigidity and multifocal seizure syndrome (RMFSL) or

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