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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)

developmental delay. Short stature and facial dysmorphisms were noted and, at 24 months, she developed generalized seizures. Currently, she does not walk unaided, says only one word, and has poor visual contact and marked stereotypic behavior with bruxism. Whole exome sequencing identified a *de novo* pathogenic variant in *COL4A3BP* (S260L).

Conclusions: This missense variant was previously found in 4 out of 5 known patients, suggesting a mutational hotspot in this gene. Former functional evidence indicates it results in intracellular imbalances in ceramide, pivotal to the synthesis of sphingolipids. Our case has significant overlap with clinical data from previous patients. This report adds evidence to an emerging consistent phenotype associated with *COL4A3BP* mutations, characterized by severe ID, postnatal microcephaly, epilepsy, autistic behavior including striking bruxism, and unspecific dysmorphic features.

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E-P08.09
Paediatric patient with deletion on chromosome 10q11.22 diagnosed by aCGH

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Introduction: The microchip-based diagnostic practices become widely introduced in postnatal diagnosis of individuals with developmental delay and dysmorphism. Array Comparative Genomic Hybridization (aCGH) technology to whole human genome screening revealed an unexpectedly large number of deletions and duplications.

Materials and methods: aCGH analysis was perform in pediatric patient with dysmorphia and motor delay using the CytoScan_750k Array platform (Affymetrix) which comprises 550 k non-polymorphic and 200 k SNP markers by the Chromosome Analysis Suite (ChAS) Software (v4.0).

Results: Our patient was born with small birth weight with idiopathic thrombocytopenia during the neonatal period. At the age of 4 years, she has short stature and following sings of dysmorphia - sparse hair, baldness in infancy, slow growing hair, wide fontanel, dry skin, no sweating, small teeth, deep set eyes, wide nasal tip, smooth philtrum, up slanted palpebraes, protruded tongue in infancy, prominent forehead and some missing teeth-upper incisors. Hydronephrosis on the left kidney, reflux vesicaurinaria, atrial septal defect, dilatated pulmonary artery and CNS plexus hypertrophy were noticed by ultrasonography. Also, subclinical hypothyreosis was present. She has mild motor delay, started to walk at 20 months, first words spoken at 2.5 years with poor vocabulary at the moment. Good social skills were present. Karyotype was normal. aCGH analysis showed 1 pathogenic copy of the segment on the chromosome 10q11.22 (437 kb) including GPRIN2(611240) gene associated with brain development, according to aDGV and ClinVar databases.

Conclusions: The application of microarray will expand the spectrum in diagnosing cytogenomic abnormalities by including complex structural variants.

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E-P08.10
Deletion 7q31.2q31.31 segregating in a family with speech and language deficiencies

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Deletions of chromosome 7q31 have been described in individuals with variable neurodevelopmental disorders and speech impairment. These deletions usually contain *FOXP2*, in which loss-of-function mutations represent a major cause for specific language and speech phenotypes. Recently, a single patient with developmental delay as well as speech and language impairment carrying a 3.2 Mb deletion in 7q31.2q31.31 excluding *FOXP2* was reported. By chromosomal microarray analysis we now identified a similar microdeletion in three family members presenting with variable speech, language and neurodevelopmental phenotypes. The index patient presented with normal motor milestones but delayed speech development, reduced comprehension and an incomplete sentence structure at age 5 years. At age 10 years he attended a school for children with special needs and presented with microcephaly, attention deficits and slurred speech. His sister was referred at age 3 years with normal first milestones but a delay in speech development. Linguistic testing revealed impairment of all language and speech levels with limited vocabulary and slurred and imprecise pronunciation. The mother had attended a school focusing on speech and language deficiencies and now presented with slurred speech. Segregation analysis in the healthy, maternal grandparents did not detect the microdeletion, thus indicating de novo occurrence in the mother. The deletion contains 17 genes and is located

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