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ABSTRACTS COLLECTION

# Abstracts from the 53rd European Society of Human Genetics (ESHG) Conference: e-Posters



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Presenting authors are indicated with bold typeface in the contributor lists.

## 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) |

Chromosomal band 17q12 is a gene rich region flanked by segmental duplications which make the region prone to deletions and duplication by NAHR mechanism (non-allelic homologous recombination). While the deletions cause well described clinical unit with specific phenotype called RCAD (renal cyst and diabetes mellitus), the phenotype caused by reciprocal duplications of the same region still remains unclear especially due to variable expressivity and incomplete penetrance and fact, that they are often detected in healthy patients. Here we present unusual case of family where the mother is carrier of duplication (paternally inherited) and also deletion (inherited from mother with RCAD) of the identical 17q12 region. All of her children were diagnosed with 17q12 microduplication syndrome. Except of the mother and grandfather, all her children carrying duplication express variable degree of neurodevelopmental problems, such as epilepsy, mild intellectual disability, delayed speech development or attention deficit disorder, that correlate with the hypothesis of incomplete penetrance and variable phenotype published by many studies. As a potential causative genes are considered *LHX1* for neurodevelopmental problems and gene *ACACA* for epilepsy. The simultaneous occurrence of deletion and duplication of the same chromosomal region in one family is very rare. This case supports hypothesis that 17q12 duplications are stable and may segregate in a family for several generations. Supported by: 00064203, NF-CZ11-PDP-3-003-2014, AZV17-29423A

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## E-P14.05
## Duplication of 10q22.2q23.1 as a cause for severe hypotonia in a child

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**Introduction:** Array comparative genomic hybridization (aCGH) technique enables high-resolution screening of the genome for segmental genomic copy number variations (CNVs). The technique today represents an unsurpassed tool for detecting minor chromosomal aberrations in children with expressed pathological clinical features, such as hypotonia, dysmorphia, motor and mental delay.

**Materials and methods:** aCGH analysis was performed in a child with clinical signs of dysmorphia and severe hypotonia using the Affymetrix® CytoScan™ 750K Array (Applied Biosystems). Each array was consisted of 200 k SNP and 550 k non-polymorphic markers. The data was analysed and interpreted using Chromosome Analysis Suite (ChAS) Software (v4.0).

**Results:** 7-month-old child had hypotonia, was unable to hold his head, to sit and did not develop fine motor skills. The clinical conditions included asthenia, frequent respiratory infections and atypical convulsion. Dysmorphological features followed wide forehead, protruded eyes, long eyelashes, triangular face, slender extremities and diminished fat tissue. Karyotype results were typical for a normal male (46, XY). The array CGH analysis revealed a pathological microduplication of a 10q segment - arr 10q22.2q23.1(75,709,593-80,912,470)x3 that was 5230 kb long and includes 44 genes.

**Conclusion:** Application of array Comparative Genomic Hybridization (aCGH) in patient with severe hypotonia, dysmorphia and neurological disability represents a great diagnostic marker compared to conventional karyotyping. The results suggest that the main reason for the various clinical and dysmorphological features of the child has genetic background. Keywords: array Comparative Genomic Hybridization, karyotype, dysmorphia, hypotonia

G. Ilieva: None. E. Sukarova-Angelovska: None. D. Nestoroska: None. S. Naunova-Timovska: None. M. Pesevska: None. V. Anastasovska: None.

## E-P14.07
## Chromosome instability index: a potential human cancer biomarker?

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**Introduction:** Chromosome Instability Index (CII) is a biomarker that has been remarkably increased in cancer patients and it consists of three parameters: Sister Chromatid Exchange (SCE) frequencies, Proliferating Rate Index (PRI) and Mitotic Index (MI). The most important of the above is SCEs because they showed the most significant difference between patients and controls. The aim of the study was to investigate the CII as possible biomarker in different types of cancer.

**Material and Method:** We used our data of the SCE methodology as it is a simple, sensitive and rapid detection technique used to evaluate chromosomal fragility. Increased SCEs mean increased unspecific damage to the DNA molecule, which was caused by various mutants in lymphocyte cultures.

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