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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)

patients only 1 proband had both mutations. Our results demonstrate that patients with suspected CF and undetected mutations in the *CFTR* gene are necessary to differential diagnosis with a SDS.

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E-P03.08
Thyroid peroxidase (TPO) mutations in Macedonian patients with congenital hypothyroidism

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Introduction: Congenital hypothyroidism (CH) due to thyroid dyshormonogenesis is a heterogenic disorder caused by impairment in any stage of the thyroid hormone biosynthesis pathway. Mutations in seven genes may be associated with thyroid dyshormonogenesis: *SLC5A5 (NIS), SCL26A4 (PDS), TG, TPO, DUOX2, DUOXA2, and IYD (DHEAL1)*. Defects in thyroid peroxidase (TPO) gene are reported as the most frequent cause of dyshormonogenesis with permanent CH characterized with permanent total iodide organification defect.

Methods: A comprehensive, phenotype-driven, approach was used to identify underlying mutations in Macedonian cohort of CH patients, by sequentially screening known dyshormonogenesis-associated genes. Genomic DNA was extracted from peripheral blood leukocytes, and Sanger sequencing was used to screen for *TPO* gene mutations in all coding exons and exon/intron boundaries amplified by PCR specific primers.

Results: Analysis of the *TPO* gene revealed variants in 3 cases including 2 siblings who each harboured a heterozygous frameshift variant, p.A397Pfs*77, inherited from a euthyroid father. An unrelated case was compound heterozygous for a maternally inherited missense mutation, p. R438H, and a paternally inherited frameshift variant, p. E17Dfs*77. All 3 cases exhibited severe CH at diagnosis, with significantly elevated serum TSH values (mean: 103mU/L, range >75-159mU/L) and low fT4 values (mean<4ug/dl). Goitre was diagnosed on the neonatal thyroid ultrasound and during foetal ultrasound examination in the third case.

Conclusion: Finding *TPO* gene mutations in a neonate with CH indicates that the subject will require life-long treatment with thyroid hormone and that future pregnancies should be carefully monitored for the presence of prenatal goiter.

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E-P03.10
Molecular investigation of Cystic Fibrosis: CFTR mutation spectrum among Greek population

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Introduction: Cystic Fibrosis (CF) represents the second most common congenital disorder among Greek population. Approximately 4-5% of Greek population represents asymptomatic CF mutation carriers. This in conjunction with the limited knowledge about this disease necessitates the implementation of carrier screening programs.

Materials and methods: The study population consisted of 485 Greek individuals screened for CF mutations. We used a targeted NGS to sequence all CFTR gene coding regions, intron/exon splice sites and UTRs, combined with MLPA analysis.

Results: Molecular analysis showed that 96 individuals (19.8%) represent CF mutation carriers, consisting of 24 individuals (4.75%) carrying typical CF-causing mutations, 38 individuals (7.52%) carrying CFTR-RD (CFTR-related disorders) mutations and 38 individuals (7.52%) carrying variants of uncertain significance (VOUS). Seven cases of complex alleles that carry two mutations were also observed, a factor that can complicate CFTR diagnosis and clinical evaluation. The most common typical mutation identified in 15 carriers was ΔF508, followed by 621+1G>T mutation occurring in 3 carriers. Likewise, R75Q mutation represents the most common CFTR-RD mutation identified (8 carriers), followed by L997F and R1162L (found in 6 and 4 carriers respectively). Lastly, c.2620-15C>G mutation was identified in 11 carriers, representing the most common VOUS mutation in the population studied.

Discussion: Aim of the current study was to estimate the CFTR mutation spectrum and the prevalence of CF mutation carriers among Greek population. Accurate knowledge of CF mutation spectrum provides information for CF prevention programs and may prove particularly important for providing more sufficient genetic counselling to the Greek population.

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For the organiser
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