European Journal of Human Genetics (2020) 28:798–1016
https://doi.org/10.1038/s41431-020-00741-5

ABSTRACTS COLLECTION

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

Volume 28 | Supplement 1

Virtual Conference

June 6-9, 2020

Sponsorship: Publication of this supplement was sponsored by the European Society of Human Genetics. All content was reviewed and approved by the ESHG Scientific Programme Committee, which held full responsibility for the abstract selections.

Disclosure Information: In order to help readers form their own judgments of potential bias in published abstracts, authors are asked to declare any competing financial interests. Contributions of up to EUR 10 000.- (Ten thousand Euros, or equivalent value in kind) per year per company are considered "Modest". Contributions above EUR 10 000.- per year are considered "Significant".

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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-P09.39
Correlation of genetic analysis and evolution of MRI changes leading to diagnosis of Labrune syndrome

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Progressive leukoencephalopathy with calcification and cysts was first reported by Labrune in 1996. The pathology underlying the disease is a diffuse cerebral microangiopathy with development of micro-, macrocysts, tumour-like vascular hyperplasia, calcification, glial proliferation, demyelination, necrosis, iron deposition and haemorrhage. Mutations in the *SNORD118* gene were associated with Labrune syndrome in 2012. Since then only individual cases have been presented.

We report a normally developing 10-years-old girl suffering from chronic headaches without neurological symptoms. Initially, Aicardi-Goutieres syndrome was suspected. We performed retrospective analysis of three annual brain MRI with MR angiography. In all MRI we found diffuse symmetrical white matter areas of increased signal with U-fibers sparing. Deep nuclei were affected. Cystic areas, enlarging in consecutive examinations were observed in thalami, with mass effect in last MRI. Multiple signals with blooming effect were spread both supra- and infratentorially. Partial peripheral contrast enhancement was seen in supratentorial changes. A suspicion of Labrune syndrome has arose after progression of cysts in degenerated white matter and appearance of calcifications. Complex heterozygous mutations 17:008173448-G>C and 17:008173570-G>C in the *SNORD118* gene encoding small nuclear RNA were found in patient using whole exome sequencing. Both parents are carriers of identified mutations.

Conclusion: Follow up MRI examinations in normally developing child and correlation with in-depth genetic analysis led to diagnosis of a rare genetic metabolic syndrome. Collaboration between radiologists, geneticist and clinicians was crucial for establishing final diagnosis.

J. Pilch: None. M. Machnikowska-Sokołowska: None. M. Rydzanicz: None. A. Pollak: None. J. Kosińska: None. P. Gasperowicz: None. K. Gruszczynska: None. E. Emich-Widera: None. R. Płoski: None.

E-P09.42
Early onset of complex seizures as a first sign of 16p11.2 deletion syndrome

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Background: Recent technologies enabled clarification of some previously undetected causes of intellectual disability and autism spectrum disorders. There are reports of 16p11.2 deletion in the literature, describing variable clinical presentation in patients. Most of them describe developmental delay, autism and seizures, however phenotypic pattern is undistinguishable and variable. There is still no sufficient clinical data of this CNV in babies.

Case report: We report on a patient with developmental delay and complex seizures. This is a second child in a family, the pregnancy and delivery was uneventful. Microcephaly, foramen ovale and generalized hypotonia were noticed shortly after birth. At the age of 3 months he developed infantile spasms, followed by profound developmental delay within the next several months. He has microcephaly, early closure of the fontanel, high forehead, bitemporal narrowing, wide nasal root, narrow palpebral features, upper lip notch, short neck, cryptorchidism. Karyotype was normal. aCGH analysis (using Affymetrix® CytoScan™ 750K Array) showed deletion of 16p11.2 (1.430 kb), also deletion on chromosome 14q11.2 (433 kb), both are considered as pathogenic according aDGV database.

Discussion and conclusion: Deletion of the 16p11.2 is mostly described in older children that developed autistic spectrum of disorder, ADHD, etc. Wide clinical variability is described between patients, mostly due to the variations on size and breakpoint of the deleted region. There are some inconsistencies in phenotype described in literature, however this could be a part of a changing phenotype with age. Since the baby had two pathogenic microdeletions, it is difficult to distinguish between two phenotypes.

E. Sukarova-Angelovska: None. V. Anastasovska: None. F. Duma: None. L. Muaremovska: None. D. Nestoroska: None. G. Ilieva: None. M. Pesevska: None. M. Velkov: None.

E-P09.45
Whole Exome Sequencing of consanguineous families of clinically diagnosed with Neurodevelopmental Disorders

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