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A Multi-faceted Approach for Understanding Czech Prenatal Cases

July 3, 2021| 10:40 - 11:35 CEST

A Multi-faceted Approach for Understanding Czech Prenatal Cases

Improvements in first-trimester screening of prenatal syndromic cases using a combination of biochemistry, ultrasound and non-invasive prenatal testing have become much more precise, reaching >99% accuracy in some cases. However, there is still a need to confirm or clarify the results of the screening methods by analysis of fetal material obtained by invasive procedures.

Molecular technologies, such as QF-PCR, array-CGH and NGS, bring accuracy with rapid turn-around time and improved clinical results in the prenatal setting, aiding both clinicians in clinical interpretation and parents to decide on the fate of the pregnancy.
The first method of choice is excluding common aneuploidies (13, 18, 21, X and Y) by QF-PCR in pregnancies at high risk for these chromosomal abnormalities. This can be obtained within the same day as the invasive procedure.

The second method of choice is array-CGH (array comparative genomic hybridization) which can provide a much higher resolution than conventional karyotyping, revealing severe microdeletion and microduplication syndromes. Moreover, this can be achieved in rapid turn-around time and from very little clinical material (starting at 50 ng of DNA).

We will present our experience with array-CGH in the clarification of high-risk prenatal cases using both legacies 8x60K ISCA and recently released GenetiSure Cyto microarrays with increased resolution in clinically relevant target regions. In addition, we will also discuss the use of NGS in a prenatal setting for identifying clinically relevant events for severe congenital heart disorders and RASopathies by leveraging the power of customized NGS gene panels.

Dr. Diana Nikulenkov Grochova, Ph.D.

Cytogenetica laborator Brno, Czech Republic

Dr. Diana Nikulenkov Grochová has a broad experience in the field of molecular genetics with a focus on human genetics and prenatal diagnostic.

Event Information

Date:
July 3, 2021

Time:
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Speaker:
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