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Recent Trends in Prenatal Genetic Screening and Testing

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Deadline for manuscript submissions:
closed (30 April 2022)

Message from the Guest Editors

Dear Colleagues,

Noninvasive prenatal diagnosis (NIPD) has become routine test in the last years. It is achieved by studying cell-free DNA (cfDNA) in the maternal plasma, which contains fetal cfDNA (cffDNA) derived from the placenta. NIPD is frequently limited to the screening of chromosomal anomalies, fetal sex determination and Rhesus D genotyping.

Actually, about 25-30% of genomic rearrangements visible through molecular cytogenetics are underlying congenital anomalies and 10-20% of isolated or syndromic anomalies can be associated with monogenic diseases, whose diagnosis is often established based on a family history, clinical examination, and confirmed through genetic tests.

In our study, the challenge was to apply trio-Whole Exome Sequencing (trio-WES) strategy using cffDNA, in order to implement NIPD to cases of fetal ultrasound malformations.

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Editors-in-Chief

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