Special Issue

Prenatal Diagnosis: From Morphological Evaluation to Genetic Testing

Message from the Guest Editors

Ultrasound evaluations are offered to all pregnant women in order to detect fetal malformations, soft markers, or abnormalities of the amniotic fluid and placenta. Based on ultrasound findings and family history, they can be classified into low- and high-risk pregnancies. Women with low-risk pregnancies can undergo screening evaluations, represented by first-trimester combined testing and noninvasive prenatal screening, while in high-risk pregnancies, an invasive procedure is required to obtain diagnostic results. Soft markers represent relatively frequent transient, non-malformative ultrasound findings that, if numerous or associated with additional features, may be suggestive of specific genetically determined conditions. Cytogenetic analysis, including standard karyotype and chromosomal microarray analysis, can detect imbalances. Exome sequencing may be proposed in selected cases to increase the possibility of identifying causative variants, taking into account the risk of inconclusive results. Additional imaging examinations can play a key role in defining the phenotype and guiding genetic testing and postnatal clinical management.

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