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Molecular Diagnosis in Congenital Fetal Anomalies

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Message from the Guest Editors

Congenital anomalies, also known as birth defects or congenital malformations, are structural or functional anomalies that occur during the intrauterine life and can be identified prenatally at birth or sometimes later in infancy. Recent advances in the field of laboratory genetics, including molecular karyotype and Next-Generation Sequencing (NGS), has allowed for the detection of genetic defects associated with congenital malformations in an increasingly higher number of cases. Genome-wide analysis methods such as Whole Exome Sequencing (WES) and Whole Genome Sequencing (WGS), or Targeted Gene Panels, have made possible the simultaneous investigation of hundreds of candidate genes associated with phenotypes, as indicated by ultrasonographic findings. However, more research is needed to achieve a better classification of genetic variants detected from such methods, to evaluate the impact of environmental factors and understand the exact biological processes behind congenital defects.

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Message from the Editor-in-Chief

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