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Prenatal Diagnosis: State of the Art and Future Directions

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Message from the Guest Editors

The purpose of this Special Issue is to summarize the state of the art in prenatal diagnosis with a focus on the diagnostic approaches (invasive and non-invasive) currently available to diagnose genetic conditions in prenatal settings. The journal will also consider paradigmatic single case or few case reports based on the use of the latest techniques in prenatal cytogenomics, expanding our knowledge of early human development and occurrence of genetic diseases during fetal development.

The topics of interest for this Special Issue include:

- prenatal exome and genome sequencing
- use of whole-exome and genome sequencing in fetuses with or without structural anomalies detected by ultrasonography
- discovery of novel prenatal phenotypes and prenatally lethal/developmental genes
- non-invasive prenatal test (NIPT)
- prenatal genetic counselling
- preimplantation genetic testing
- single-cell analysis of human embryos
- low-level mosaicisms detection by ultrasensitive sequencing methods
- fetal and parental incidental findings related to the implementation of NGS platforms in prenatal diagnosis
- implementation of bioinformatic pipelines for fetal CNV detection on WES data