Special Issue

Recent Advances in Prenatal Diagnosis and Clinical Management

Message from the Guest Editor

This Special Issue, on advances in prenatal diagnosis, spotlights pivotal innovations in diagnostic technologies and methodologies revolutionizing early fetal health assessment. By integrating non-invasive prenatal testing (NIPT) enhancements (e.g., cfDNA methylation profiling, low-coverage whole-genome sequencing), advanced fetal imaging (AI-driven ultrasound automation, MRI-based volumetric anomaly detection), and multi-omics biomarkers (proteomic, metabolomic, and epigenomic signatures), the collection elevates the detection sensitivity and specificity for chromosomal abnormalities (e.g., aneuploidies, microdeletions), monogenic disorders, structural malformations, and placental dysfunction. Through rigorous evaluation of diagnostic accuracy, cost-effectiveness, and scalability, this Special Issue provides clinicians, geneticists, and public health practitioners with actionable evidence to streamline prenatal screening pathways, minimize diagnostic ambiguity, and align genetic information with culturally sensitive, family-centered care.

Guest Editor

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