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Advancements in Morphological and Molecular Prenatal Diagnosis: European Practice and Future Perspectives

Guest Editor:

Prof. Dr. Antonio Pizzuti

Department of Experimental Medicine, "Sapienza" University of Rome, 00100 Rome, Italy

Deadline for manuscript submissions: closed (31 August 2022)

Message from the Guest Editor

Prenatal diagnosis is an ever-growing field with continuous innovation. Advances in morphological and molecular approaches provide valuable new tools, resulting in remarkable achievements, but also increasing difficulties in the interpretation and management of such findings.

Invasive genetic testing, either for morphological anomalies or anamnestic indications, has undergone radical changes. Molecular cytogenetics is a staple in prenatal diagnosis, and the recent availability of broad cohorts can offer precious insights and prompts to fetal medicine specialists. The advent of Next Generation Sequencing (NGS) made timely prenatal molecular diagnoses possible for varied indications, with different approaches available, such as targeted panels, exome sequencing or even genome sequencing. The diagnostic yield is encouragingly high, but specific indications of each approach, as well as the management of incidental and uncertain findings, are yet to be defined.

This Special Issue aims to collect and integrate advancements in morphological and molecular prenatal diagnosis, depicting the state-of-the-art and future perspectives in European practice.

Specialsue



mdpi.com/si/112537





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Editor-in-Chief

Prof. Dr. Andreas Kjaer

Department of Clinical Physiology, Nuclear Medicine & PET National University Hospital, Rigshospitalet, University of Copenhagen, Blegdamsvej 9, DK-2100 Copenhagen, Denmark

Message from the Editor-in-Chief

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Diagnostics Editorial Office MDPI, St. Alban-Anlage 66 4052 Basel, Switzerland Tel: +41 61 683 77 34 www.mdpi.com mdpi.com/journal/diagnostics diagnostics@mdpi.com X@diagnostic_mdpi