

Special Issue

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

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.

Guest Editor

Prof. Dr. Antonio Pizzuti

Department of Experimental Medicine, Faculty of Medicine and Dentistry, Sapienza University of Rome, 00161 Rome, Italy

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Editorial Office
MDPI, Grosspeteranlage 5
4052 Basel, Switzerland
Tel: +41 61 683 77 34
diagnostics@mdpi.com

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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

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