

## Special Issue

# Prenatal Diagnosis: State of the Art and Future Directions

### 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 mosaicism 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

---

### Guest Editors

Dr. Edoardo Errichiello

1. Department of Molecular Medicine, University of Pavia, Pavia, Italy  
2. IRCCS Mondino Foundation, Pavia, Italy

Dr. Elena Rossi

Medical Genetics, Department of Molecular Medicine, University of Pavia, 27100 Pavia, Italy

---

### Deadline for manuscript submissions

closed (15 March 2023)



## Medical Sciences

---

an Open Access Journal  
by MDPI

---

Impact Factor 4.4  
CiteScore 4.6  
Indexed in PubMed



[mdpi.com/si/84467](https://mdpi.com/si/84467)

*Medical Sciences*  
Editorial Office  
MDPI, Grosspeteranlage 5  
4052 Basel, Switzerland  
Tel: +41 61 683 77 34  
[medsci@mdpi.com](mailto:medsci@mdpi.com)

[mdpi.com/journal/](https://mdpi.com/journal/)

[medsci](https://medsci)





# Medical Sciences

---

an Open Access Journal  
by MDPI

---

Impact Factor 4.4  
CiteScore 4.6  
Indexed in PubMed



[mdpi.com/journal/  
medsci](https://mdpi.com/journal/medsci)



## About the Journal

### Message from the Editor-in-Chief

---

#### Editor-in-Chief

Prof. Dr. Antoni Torres

1. Institut D investigacions August Pi I Sunyer (IDIBAPS), Department of Pulmonology, Hospital Clinic de Barcelona, Universidad de Barcelona, 08036 Barcelona, Spain

2. Centro de Investigación Biomédica en Red en Enfermedades Respiratorias (CIBERES), Instituto de Salud Carlos III, 28029 Madrid, Spain

---

#### Author Benefits

##### High Visibility:

indexed within Scopus, ESCI (Web of Science), PubMed, PMC, MEDLINE, CAPlus / SciFinder, and other databases.

##### Journal Rank:

JCR - Q1 (Medicine, General and Internal) / CiteScore - Q1 (General Medicine)

##### Rapid Publication:

manuscripts are peer-reviewed and a first decision is provided to authors approximately 18.7 days after submission; acceptance to publication is undertaken in 2.8 days (median values for papers published in this journal in the second half of 2025).