

Special Issue

‘Rare Syndromes: Diagnosis and Treatment’ in 2024–2026

Message from the Guest Editors

This Special Issue, related to rare syndromes, provides an open platform for research into all aspects of diseases considered as rare according to US or EU parameters. The goal is to provide an open platform for all health professionals to share knowledge, providing a better understanding of their etiology, prevention, pathogenic mechanisms and clinical aspects.

Accordingly, this could improve the treatment and management of patients suffering from a rare disease. Moreover, we hope that this Special Issue will serve as an additional tool to help in the continuous efforts to tackle difficult-to-diagnose diseases. Rare syndrome topics of interest include, but are not limited to, the following:

- Epidemiology;
- Social aspects;
- Pathogenic mechanisms;
- Genetic aspects;
- Prevention;
- Diagnostic tools;
- Novel approaches: artificial intelligence;
- Patient care;
- Clinical aspects;
- Therapy;
- Patient management;
- Study of animal models;
- In vitro models.

Guest Editors

Prof. Dr. Mojgan Rastegar

Department of Biochemistry and Medical Genetics, Rady Faculty of Health Sciences, Max Rady College of Medicine, University of Manitoba, Winnipeg, MB R3E 0J9, Canada

Dr. Jose Sanchez-Alcazar

Centro Andaluz de Biología del Desarrollo (CABD-CSIC-Universidad Pablo de Olavide), 41013 Sevilla, Spain

Deadline for manuscript submissions

31 December 2026



Diseases

an Open Access Journal
by MDPI

Impact Factor 3.7
CiteScore 4.2
Indexed in PubMed



mdpi.com/si/215434

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

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





Diseases

an Open Access Journal
by MDPI

Impact Factor 3.7
CiteScore 4.2
Indexed in PubMed



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



About the Journal

Message from the Editor-in-Chief

Diseases is an international, peer-reviewed, open access, multidisciplinary journal that focuses on the latest outstanding research concerning diseases and conditions. Research articles, reviews, and other contents are released on the Internet immediately after acceptance. This journal aims to cover international conferences and symposia as new targets.

Diseases is increasingly gaining acceptance and visibility within scholars, and after only a few years of life, *Diseases* is now also covering specific topics with dedicated special issues. We would be pleased to welcome you as one of our authors.

Editor-in-Chief

Prof. Dr. Omar Cauli

Frailty and Cognitive Impairment—FROG Research Group, University of Valencia, 46010 Valencia, Spain

Author Benefits

High Visibility:

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

Journal Rank:

JCR - Q2 (Medicine, Research and Experimental) /
CiteScore - Q1 (General Medicine)

Rapid Publication:

manuscripts are peer-reviewed and a first decision is provided to authors approximately 21.6 days after submission; acceptance to publication is undertaken in 2.6 days (median values for papers published in this journal in the first half of 2026).