

Special Issue

Discovering the World of Rare Diseases: 2nd Edition

Message from the Guest Editors

Rare diseases, although individually uncommon, can affect a significant proportion of the population (5–8% of the global population, with 7.9% of those younger than 25). In Europe, the European Medicines Agency specifies that rare disorders include those with a prevalence of less than 5 in 10,000 people (equivalent to less than 1 in 2,000). The exact number of rare diseases is variable, ranging from 5,000 to 8,000, of which 80% have a genetic etiology. A report from the 2005 European Conference on Rare Diseases, evaluating the life expectancy of patients affected with 323 rare diseases, concluded that 25.7% of rare diseases could be considered potentially lethal before 5 years of age, and that a further 36.8% lead to a reduced life expectancy. Conversely, only about a third (37.5%) are associated with a normal lifespan. We are pleased to invite you and your coworkers to contribute to this Special Issue with original research reports, reviews, or meta-analyses on the topics of rare diseases, evaluating the incidence and prevalence of rare diseases in different countries, diagnostic approaches and delays, standard protocols of care, and national legislation and registries.

Guest Editors

Dr. Filippo Manelli

Emergency Department, ASST-Valcamonica, Esine, Brescia, Italy

Dr. Maria Sofia Cotelli

Neurology Unit, Vallecamonica Hospital, Esine, Brescia, Italy

Deadline for manuscript submissions

closed (30 June 2024)



Medicina

an Open Access Journal
Published by MDPI

Impact Factor 2.4
CiteScore 4.1
Indexed in PubMed



mdpi.com/si/194050

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

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





Medicina

an Open Access Journal
Published by MDPI

Impact Factor 2.4
CiteScore 4.1
Indexed in PubMed



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



About the Journal

Message from the Editor-in-Chief

You are invited to contribute a research article or a comprehensive review for consideration and publication in *Medicina* (ISSN: 1648-9144). *Medicina* is an open access, peer-reviewed scientific journal that publishes original articles, critical reviews, research notes, and short communications on medicine. The scientific community and the general public can access the content free of charge as soon as it is published.

Editor-in-Chief

Prof. Dr. Edgaras Stankevičius
Medical Academy, Lithuanian University of Health Sciences, Kaunas,
Lithuania

Author Benefits

Open Access:

free for readers, with article processing charges (APC) paid by authors or their institutions.

High Visibility:

indexed within Scopus, SCIE (Web of Science), PubMed, MEDLINE, PMC, and other databases.

Journal Rank:

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