

Special Issue

Diagnosis and Treatment of Rare Diseases

Message from the Guest Editor

Despite their low prevalence, clinically defined rare diseases (RDs), of which there are over 6000 affecting 300 million people worldwide, share certain characteristics (i.e., they have a significant diagnostic delay, they are usually chronic and incapacitating, and the majority have a genetic basis, high morbidity rate, and limited therapeutic options). Therefore, they usually have a deep impact on the quality of life of patients and caregivers. Some of the priorities that deserve further development include: better knowledge of the natural history and pathophysiological mechanisms of RDs, improving access to multidisciplinary medical care and orphan drugs, increasing awareness among healthcare professionals, and enhancing genetic and molecular characterisation for a large number of undiagnosed RDs. For this Special Issue, we welcome the submission of original research articles and up-to-date reviews on the described topics.

Guest Editor

Dr. Álvaro Hermida-Ameijeiras

Department of Psychiatry, Radiology, Public Health, Nursing and Medicine, University of Santiago de Compostela, 15704 Santiago de Compostela, Spain

Deadline for manuscript submissions

closed (25 January 2024)



Journal of Clinical Medicine

an Open Access Journal
by MDPI

Impact Factor 2.9
CiteScore 5.2
Indexed in PubMed



mdpi.com/si/152956

Journal of Clinical Medicine
Editorial Office
MDPI, Grosspeteranlage 5
4052 Basel, Switzerland
Tel: +41 61 683 77 34
jcm@mdpi.com

mdpi.com/journal/

jcm





Journal of Clinical Medicine

an Open Access Journal
by MDPI

Impact Factor 2.9
CiteScore 5.2
Indexed in PubMed



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



About the Journal

Message from the Editorial Board

There has been an explosion of gene and target based research and therapeutics in the multitude of fields that compose clinical medicine. The *Journal of Clinical Medicine's* (JCM) staff and editorial board are dedicated to providing cutting edge, timely, and peer-reviewed articles covering the diverse subspecialties of clinical medicine. The journal publishes concise, innovative, and exciting research articles as well as clinically significant articles and reviews that are pertinent to the myriad of disciplines within medicine. The articles published are relevant to both primary care physicians and specialists. The journal's full-texts are archived in PubMed Central and indexed in PubMed. Please consider submitting your manuscripts for publication to our journal and check us out on-line!

Editors-in-Chief

Prof. Dr. Emmanuel Andr  s

Internal Medicine Department, University Hospital Strasbourg, 67000
Strasbourg, France

Prof. Dr. Kent Doi

Department of Acute Care Medicine, University of Tokyo, Tokyo, Japan

Author Benefits

High Visibility:

indexed within Scopus, SCIE (Web of Science), PubMed, PMC, Embase, 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 17.7 days after submission; acceptance to publication is undertaken in 2.7 days (median values for papers published in this journal in the first half of 2025).