

Special Issue

Rare and Orphan Disorders: An Emerging Challenge

Message from the Guest Editor

The rapid development of genomic techniques such as exome, genome, and RNA sequencing has advanced the field of genetic diagnosis and personalized therapy for the rare and orphan disease cohort. The *Journal of Personalized Medicine* aims to publish a collection of articles that address the utility and efficiency of these diagnostic tools and provide fresh insights to ongoing discussion and debate in genomic medicine. We will consider original research articles, systematic reviews, and well-designed case studies and analyses that report empirical work that presents experiences and perspectives from the US and abroad. The included topics in this Issue are as follows:

- Advancements in genomic techniques for diagnosing and treating rare and orphan diseases;
- Studies of novel disease-causing genes, phenotype-genotype relationships, etc.;
- Bioinformatics including exome data reanalysis, machine learning, etc.;
- Personalized and individualized therapy for rare and orphan disease.

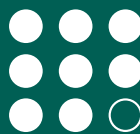
Guest Editor

Dr. Shiyu Luo

Division of Newborn Medicine, Boston Children's Hospital and Harvard Medical School, Boston, MA, USA

Deadline for manuscript submissions

closed (10 April 2023)



Journal of Personalized Medicine

an Open Access Journal
by MDPI

CiteScore 6.0
Indexed in PubMed



mdpi.com/si/98525

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

mdpi.com/journal/

jpm





Journal of Personalized Medicine

an Open Access Journal
by MDPI

CiteScore 6.0
Indexed in PubMed



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



About the Journal

Message from the Editor-in-Chief

Journal of Personalized Medicine (JPM), ISSN 2075-4426) is an international, open access journal aimed at bringing all aspects of personalized medicine to one platform. *JPM* publishes cutting edge, innovative preclinical and translational scientific research and technologies related to personalized medicine (e.g., precision medicine, pharmacogenomics/proteomics, systems biology, 'omics association analysis). *JPM* is covered in Scopus, the Science Citation Index Expanded (SCIE), PubMed, PMC, Embase, and other databases.

Editor-in-Chief

Prof. Dr. Kenneth P.H. Pritzker

Department of Laboratory Medicine and Pathobiology, Department of Surgery, University of Toronto, 6 Queens Pk Crescent W,F, Toronto, ON M5S 3H2, Canada

Author Benefits

High Visibility:

indexed within Scopus, PubMed, PMC, Embase, and other databases.

Journal Rank:

CiteScore - Q1 (Medicine (miscellaneous))

Rapid Publication:

manuscripts are peer-reviewed and a first decision is provided to authors approximately 21.5 days after submission; acceptance to publication is undertaken in 3.5 days (median values for papers published in this journal in the first half of 2025).