



Genetic Testing for Rare Diseases

Guest Editor:

Dr. José M. Millán

Health Research Institute La Fe,
Valencia, Spain

Deadline for manuscript
submissions:

closed (31 December 2021)

Message from the Guest Editor

Rare diseases are defined by their low prevalence but, as a whole, they affect 8–10% of the population in Western countries. Six to seven thousand rare diseases have been reported in the literature, and the number is regularly increasing. There are many differences among them in terms of etiopathology, age of onset, affected organs and tissues, etc., but about 80% of them have a genetic cause.

Therefore, the genetic testing of rare diseases is a key point for confirming the clinical diagnosis, offering the correct genetic counseling to the patients and their relatives and, more recently, the possibility of receiving a

This Special Issue will be focused on clinical genetics, genetic counselling, rare metabolic diseases, sensorineural disorders, neuromuscular diseases, and bioinformatics, among other topics.





an Open Access Journal by MDPI

Editor-in-Chief

Prof. Dr. Andreas Kjaer

Department of Clinical
Physiology, Nuclear Medicine &
PET National University Hospital,
Rigshospitalet, University of
Copenhagen, Blegdamsvej 9, DK-
2100 Copenhagen, Denmark

Message from the Editor-in-Chief

You are cordially invited to submit research articles, short communications, comprehensive reviews, case reports or interesting images for consideration and publication in *Diagnostics* (ISSN 2075-4418). *Diagnostics* is published in open access format – research articles, reviews and other contents are released on the Internet immediately after acceptance. The scientific community and the general public have unlimited and free access to the content as soon as it is published. We would be pleased to welcome you as one of our authors.

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](#), [PMC](#), [Embase](#), [Inspec](#), [CAPus / SciFinder](#), and [other databases](#).

Journal Rank: JCR - Q2 (*Medicine, General & Internal*)

Contact Us

Diagnostics Editorial Office
MDPI, St. Alban-Anlage 66
4052 Basel, Switzerland

Tel: +41 61 683 77 34
www.mdpi.com

mdpi.com/journal/diagnostics
diagnostics@mdpi.com
[X@diagnostic_mdpi](#)