Special Issue

Genetics and Genomics for Clinical Monitoring and Diagnosis 2023

Message from the Guest Editor

As a result of the introduction of high-throughput techniques, the number of studies investigating the genomic pathogeneses of human diseases is increasing. The more our ability to discover new genetic/genomic variants increases, the more distant the concept of a “Mendelian disease” becomes. Thanks to high-throughput DNA/RNA sequencing, we have indeed discovered that each individual carries thousands of single nucleotides as well as copy-number variants. Genetic/genomic testing to identify the molecular basis of a disease in a clinical context is generally aimed to pick up the “major pathogenic variant” that can explain a patient’s phenotype. However, how closely the patient’s observed phenotype corresponds to the phenotype described for a gene can affect the sensitivity of the molecular test.

The aim of this Special Issue is to collect evidence that could help in establishing the sensitivity of current genetic/genomic tests in the clinical assessment and clinical monitoring of patients.

Guest Editor

Dr. Irene Bottillo

Division of Medical Genetics, Department of Molecular Medicine, Sapienza Università di Roma, San Camillo-Forlanini Hospital, 00152 Rome, Italy

Deadline for manuscript submissions

closed (15 November 2023)

GCAT
TACG
GCAT

Genes

an Open Access Journal by MDPI

Impact Factor 2.8
CiteScore 5.5
Indexed in PubMed

mdpi.com/si/161086

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

mdpi.com/journal/genes

MDPI

GCAT
TACG
GCAT

Genes

an Open Access Journal by MDPI

Impact Factor 2.8
CiteScore 5.5
Indexed in PubMed

mdpi.com/journal/genes

MDPI

About the Journal

Message from the Editor-in-Chief

Genes is central to our understanding of biology, and modern advances such as genomics and genome editing have maintained genetics as a vibrant, diverse and fast-moving field. There is a need for good quality, open access journals in this area, and the *Genes* team aims to provide expert manuscript handling, serious peer review, and rapid publication across the whole discipline of genetics. Starting in 2010, the journal is now well established and recognised. Why not consider *Genes* for your next genetics paper?

Editor-in-Chief

Prof. Dr. Selvarangan Ponnazhagan
Department of Pathology, The University of Alabama at Birmingham, 1825 University Blvd, SHEL 814, Birmingham, AL 35294-2182, USA

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, Embase, PubAg, and other databases.

Journal Rank:

JCR - Q2 (Genetics and Heredity) / CiteScore - Q2 (Genetics (clinical))