

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 pathogenesis 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

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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?

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