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Genetics and Genomics for Clinical Monitoring and Diagnosis 2023

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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.



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Special Issue



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Editor-in-Chief

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Message from the Editor-in-Chief

Genes are central to our understanding of biology, and modern advances such as genomics and genome editing have maintained genetics as a vibrant, diverse and fastmoving 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.

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