

Special Issue

Application of Next-Generation Sequencing in Genetic Diseases Diagnosis

Message from the Guest Editor

High-throughput sequencing has represented a significant revolution in the field of genetic medicine and the diagnosis of inherited diseases. Currently, we can find highly efficient technical solutions but with great challenges in the clinical interpretation of genetic results, especially since the implementation of whole exome and genome sequencing into routine clinical practice. This great challenge is accompanied by the difficulty of incidental findings interpretation and counseling and also the use of complex samples such as circulating tumor DNA, maternal plasma, and cell-free DNA fetal samples. This Special Issue on Application of Next-Generation Sequencing in Genetic Diseases Diagnosis will provide novel insights and an updated overview of these technology applications and how they can interfere in the pathophysiology, diagnosis, and treatment of inherited disorders. Given the complexity and broadness of these topics, contributions from experts in the field through research papers and reviews are welcome.

Guest Editor

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Deadline for manuscript submissions

closed (20 April 2025)

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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?

Editor-in-Chief

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