Special Issue

Molecular Discoveries, Clinical Diagnostics, and Personalized Treatments for Human Genetic Diseases

Message from the Guest Editor

Collaborations between scientists and clinicians have helped genetic medicine to make significant advances in recent years. The latest human genome assembly from Ensembl! (GRCh38.p14) has identified over 1 billion short variants and 7.8 million structural variants in humans; yet, for most of these variants, we have no data on whether they are silent or result in rare phenotypes or life-threatening genetic diseases. The use of in silico tools and laboratory studies of engineered polymorphisms in cells or animals can lead to the creation of clinical diagnostic tools, and eventually to personalized treatments for human carriers. In this Special Issue, we invite researchers and clinicians to contribute high-quality original papers describing new polymorphisms contributing to human genetic disease, new or improved molecular diagnostic tests, new pharmacogenetic findings that define treatment strategies and novel genetic discoveries from all areas of medicine. Papers including methodologies such as whole genome sequencing, cell and animal models, in silico prediction tools, and clinical trials are welcome.

Guest Editor

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Deadline for manuscript submissions

closed (25 July 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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