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Genetic and Phenotypic Correlation: Gene-Disease Validation

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Deadline for manuscript submissions:
closed (20 July 2023)

Message from the Guest Editors

With the increasing use of genomic sequencing technology, a significant number of genes contributing to Mendelian disorders can be rapidly identified. However, the clinical utility of this technology bottlenecks at variant interpretation. Substantial gaps in the knowledge base necessitate more information on gene disease validation, especially genetic or experimental evidence on variants of known genes, which can clarify the correlation between genotype and phenotype.

The “one-gene-one-disease” paradigm has been challenged by multiple disease traits caused by one gene or one locus in one gene. However, variable expressivity and the incomplete penetrance of recurrent variants make genetic diagnosis challenging. More genetic evidence could enhance our understanding of the role of genetic etiology in diseases.

This Special Issue welcomes a variety of research papers, including systematic reviews of the genotype–phenotype correlation, detailed studies on genetic and experimental evidence of gene alteration, and novel insights into the genetic mechanism of rare genetic diseases.

GCAT TACG GCAT genes

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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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