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Functional Studies for Interpreting Genetic Variants Associated with Genetic Disorders

Guest Editor:

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Deadline for manuscript submissions:
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Message from the Guest Editor

Dear Colleagues,

The new high performant sequencing techniques have generated a lot of data about human genome variation. In terms of pathogenicity or benignity, the classification of all gene variants is an important future goal for medical genetics and relevant efforts should be pursued to interpret gene variants associated with genetic disorders. The American College of Medical Genetics and Genomics (ACMG) and the Association for Molecular Pathology (AMP) established guidelines for variant interpretation to help variant classification. Variant type, familial inheritance, variant frequency, and prediction tools are essential to assess a gene variant classification. One of the strong criteria included in the ACMG guidelines is based on the characterisation of gene variants by functional studies. Therefore, studies reporting experimental evidence that helps the interpretation and classification of gene variants are critical.

This Special Issue aims to collect research articles and reviews based on functional studies that support the classification of germline gene variants detected in human genetic diseases.

Prof. Dr. Rodolfo Iuliano
Guest Editor

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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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