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Molecular Genetics of Retinal Dystrophies

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Deadline for manuscript submissions:
closed (31 March 2020)

Message from the Guest Editors

Massive parallel sequencing has completely revolutionized the field of genetic diagnosis in rare diseases, such as inherited retinal dystrophies. Although the information gathered has greatly increased the number of causative genes (now over 300 genes), a considerable number of cases still remain unsolved due to technical limitations. New strategies to unveil “hidden” genetic variants and assign pathogenicity to variants of unknown significance are required to increase the diagnostic yield, pave the way for precision medicine, and inspire effective therapeutic approaches for these severe visual disorders.

This Special Issue focuses on the identification and pathogenic evaluation of novel mutations in known or unreported genes, phenotype-genotype correlations, and functional and NGS strategies for the identification of “hidden mutations” in the genome (e.g., SNVs, deep intronic mutations, and regulatory mutations). We welcome submissions of reviews, research articles, or small focused reviews in IRDs.

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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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