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RNA Splicing in Health and Disease

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Deadline for manuscript submissions:
closed (5 December 2023)

Message from the Guest Editor

Large numbers of splicing-related diseases have been documented; however, this number is likely to be substantially underestimated because the effects of mutations on splicing are often not pursued as a primary cause of disease.

In fact, hundreds of thousands of DNA variants are detected in massive sequencing projects of genetic disorders, and interestingly, recent estimations have shown that an unexpectedly large fraction of genetic diseases are caused by variants that disrupt the splicing process. Although the classification of variants located within many canonical splice sites is often straightforward due to clear biological consequences, it is important to identify and functionally study the impact on splicing of sequence variants outside the canonical splice sites.

Moreover, the modulation of splicing provides a potent therapeutic approach. In recent years, there has been emergence in RNA therapeutics to directly target specific RNA molecules, but there are still many challenges that arise. This Special Issue focuses on the impact of RNA splicing in health and disease and the new therapies that are emerging to modulate it.

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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?

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Journal Rank: JCR - Q2 (Genetics and Heredity) / CiteScore - Q2 (Genetics (clinical))

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