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Marfan Syndrome and Related Disorders: Genetic Basis, Molecular Mechanisms, and Genotype–Phenotype Correlations

Guest Editor:

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Deadline for manuscript submissions:
closed (20 April 2021)

Message from the Guest Editor

Marfan syndrome belongs to the group of rare heritable connective tissue disorders and syndromic and non-syndromic hereditary thoracic aortic aneurysms/dissections. The clinical and genetic features that allow the diagnosis of Marfan syndrome are aneurysm/dissection of thoracic aorta at the Valsalva's sinus; subluxation/luxation of the eye lenses; and presence of systemic features with a score ≥ 7 .

This Special Issue will focus on the current state-of-the-art and novel research findings concerning the molecular basis and pathogenesis of Marfan syndrome and related disorders, with particular interest in news regarding genotype–phenotype correlation, the discovery and characterization of modifier genes, the patterns of mutations/genes associated to a clinical phenotype and the techniques applied to these studies. A review regarding the aspects of hereditary transmission, of the genes associated with several pathologies or of the single gene underlying each pathology is also welcome, as well as a review regarding the cell-molecular physiopathology of Marfan syndrome and related disorders.



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Special Issue



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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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