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Molecular and Cellular Mechanisms of Marfan Syndrome

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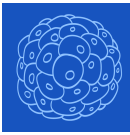
Message from the Guest Editors

Marfan syndrome (MFS) is a rare genetic connective tissue disorder with a prevalence of 1 per 5,000 individuals, and it is caused by variants in the gene encoding for the glycoprotein fibrillin-1 (FBN1). It is inherited in an autosomal dominant manner, but ~25% of the variants are de novo mutations. FBN1 variants induce abnormal or deficient fibrillin-1 fiber formation, affecting the structural integrity of the extracellular matrix (ECM) fibrillary network in a multitude of organs, such as vascular, skeletal, and ocular, hence the occurrence of multisystemic symptoms. More knowledge is essential to understanding the molecular and cellular mechanisms of the different Marfan syndrome symptoms and to improving diagnostic and treatment strategies (pharmacological and gene therapy), which are the topic of this Special Issue on Marfan syndrome.



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Special Issue



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