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Rare Monogenic Diseases: Molecular Pathophysiology and Novel Therapies

Guest Editor:

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Message from the Guest Editor

Most rare diseases we know arise from single gene mutations. In fact, the number of rare monogenic diseases is growing continuously, and to date, near 4000 single-gene inherited disorders have been characterized. Pathogenic mutations typically affect the coding regions, thus resulting in classical amino acid substitutions responsible for loss- or gain-of-function in protein products. However, several disease-causing defects originate from regulatory and non-coding DNA regions, ultimately affecting gene expression by transcriptional and/or post-transcriptional mechanisms.

Understanding the molecular pathophysiology of a rare monogenic disease has a double value. The identification of alterations that occur in specific genes, proteins, and pathways allows the translation of scientific advances into novel therapeutic approaches for these traits. Moreover, the investigation of rare monogenic diseases has the power to reveal fundamental biological mechanisms that would otherwise remain unknown.

This Special Issue will focus on the key molecular mechanisms that are affected within a rare monogenic disorder. New experimental therapies to target specific mechanisms are relevant.

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Message from the Editor-in-Chief

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