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Molecular Genetics of Rare Diseases

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Message from the Guest Editor

Dear Colleagues,

A disease affecting fewer than 200,000 people annually is considered a rare disease. Between 5 and 10% of the population is estimated to be affected by rare diseases and a majority (approximately 80%) of the rare diseases, are attributable to genetic abnormalities. The current numbers thus urge the research community to take advantage of new and affordable genome sequencing technologies, and in vitro and in vivo functional assessment strategies, clarifying the etiology of rare diseases. At the same time, developments of new gene therapy and genome editing technologies provide promises for treating these otherwise seemingly untreatable rare diseases. This Special Issue on “Molecular Genetics of Rare Diseases” will highlight recent progress in understanding and treatment of rare diseases.

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Guest Editor



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Special Issue



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Message from the Editor-in-Chief

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