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The Value of Genetics in the Identification of Treatable Rare Diseases. The Paradigm of Inborn Errors of Metabolism

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Deadline for manuscript submissions:
closed (20 November 2020)

Message from the Guest Editors

Science can provide some answers regarding all rare diseases, and it is now possible to diagnose hundreds of them through biological sample tests. Among these, the field of inherited metabolic diseases is now a major cause of acute life-threatening but largely treatable diseases.

Inborn errors of metabolism are rare diseases caused by defects of individual genes which encode enzymes that facilitate the conversion of substrates into metabolites, transporters, receptors, or molecules involved in organelles and cell traffic. These diseases can occur at any age and many of them are treatable; therefore, practical knowledge of these diseases, their presentations, and an appropriate diagnostic-therapeutic approach is essential.

The study of these disorders, is within the paradigm of the new precision medicine, which requires customizing diagnoses and treatments assuming participation of the child or adult patient and their family.

We focus on inborn errors of metabolism including clinical trial design, funding for research, digital health, and big data on one hand.

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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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