Topical Collection

Study on Genotypes and Phenotypes of Pediatric Clinical Rare Diseases

Message from the Collection Editors

With the arrival and widespread adoption of high-throughput DNA sequencing, genetic discoveries in neurodevelopmental disorders (NDDs) and genetic syndromes are advancing very quickly. The identification of novel genes and of rare, highly penetrant pathogenic variants is helping to enhance our understanding of genotype–phenotype correlations. While most dominant NDD genes are highly intolerant to variation, some exceptions are connected to the presence of variants in transcripts that are not brain expressed and/or genes that demonstrate acquired somatic mosaicism in blood. The study of the genotype–phenotype correlation is not simple in recently-described genetic syndromes, with limited numbers of clinical cases, but it is very important for the clinician, who has to interpret the genetic results and organize the follow-up for children with genetic syndromes. It would be an honour for us if you agreed to be one of the authors of this initiative. We would be happy to accept your suggestion for a title on a subject in which you are an expert.

Collection Editors

Dr. Livia Garavelli
Azienda USL-IRCCS di Reggio Emilia, 42122 Reggio Emilia, Italy

Dr. Stefano Giuseppe Caraffi
Azienda USL-IRCCS di Reggio Emilia, 42122 Reggio Emilia, Italy

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Genes
MDPI, Grosspeteranlage 5
4052 Basel, Switzerland
Tel: +41 61 683 77 34
genes@mdpi.com

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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.
Why not consider *Genes* for your next genetics paper?

Editor-in-Chief

Prof. Dr. Selvarangan Ponnazhagan
Department of Pathology, The University of Alabama at Birmingham, 1825 University Blvd, SHEL 814, Birmingham, AL 35294-2182, USA

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