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Phenotype and Pathogenetic Mechanisms in 22q11.2 Deletion/DiGeorge Syndrome

Guest Editor:

Prof. Dr. Vera Lúcia Gil-da-Silva-Lopes
Department of Translational Medicine—Medical Genetics and Genomic Medicine, School of Medical Sciences, State University of Campinas, Campinas 13083-887, SP, Brazil

Deadline for manuscript submissions:
closed (10 September 2024)

Message from the Guest Editor

Despite the 22q11.2 deletion syndrome (22q11.2DS) being the most common microdeletion in humans, it is a challenging condition to diagnose, as the phenotype is widely heterogeneous. Most individuals with 22q11.2DS have the typical ~3 Mb deletion on chromosome 22, but smaller deletions and atypical deletions of varying sizes can also be present in a minority of patients. However, the size of the deletion seems not to interfere with the phenotype. In addition, this condition includes several comorbidities throughout life, and its clinical management could be improved based on an enhanced understanding of its pathogenicity.

There is evidence that clinical heterogeneity underlies complex genetic mechanisms, including variants in other regions of the genome. Therefore, this Special Issue aims to search for studies that may contribute to the genesis of the genetic heterogeneity of the 22q11.2 deletion syndrome and its pathogenetic mechanisms. Clinical, molecular, experimental, and reviews papers are welcome.

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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

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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Contact Us

Genes Editorial Office
MDPI, Grosspeteranlage 5
4052 Basel, Switzerland

Tel: +41 61 683 77 34
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