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22q11.2 Deletion Syndrome

Guest Editors:

Dr. Beata Nowakowska
Instytut Matki I Dziecka, 01-211 Warsaw, Poland

Prof. Dr. Donna M. McDonald-McGinn
The Children's Hospital of Philadelphia, Philadelphia, PA 19104, USA

Deadline for manuscript submissions:
closed (24 September 2022)

Message from the Guest Editors

Editors of *Genes* call for submissions for a Special Issue on chromosome 22q11.2 deletion syndrome, in particular studies presented at the 12th Biennial International 22q11.2 Conference in Split, Croatia.

Chromosome 22q11.2 deletion syndrome (22q11.2DS) is the most common genomic disorder, with a prevalence of 1 in 2148 livebirths and 1 in 992 unselected pregnancies. Despite the well-characterized primary cause of the disease, the clinical variability is extremely high. Numerous features have been described, but no single finding occurs in 100% of patients. Phenotypic variability is a major source of misdiagnosis in individuals with 22q11.2DS, and despite its frequency the condition is still unfamiliar to many specialists. Recent research has shown the genetic complexity of the condition including the influence of genetic modifiers outside the chromosome 22q11.2 region on such phenotypic variability.

Special Issue will include reviews and original research manuscripts, providing an overview of current knowledge on diagnostics, treatment, and management of 22q11.2DS, as well as highlighting cutting edge clinical and basic science research.

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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.

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

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Contact Us

Genes Editorial Office
MDPI, St. Alban-Anlage 66
4052 Basel, Switzerland

Tel: +41 61 683 77 34
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𝕏@Genes_MDPI