Special Issue

22q11.2 Deletion Syndrome

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 22a11.2DS, 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.

Guest Editors

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Deadline for manuscript submissions

closed (24 September 2022)

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About the Journal

Message from the Editor-in-Chief

Genes is central to our understanding of biology, and modern advances such as genomics and genome editing have maintained genetics as a vibrant, diverse and fast-moving 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

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