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

Guest Editor:

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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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Special Issue



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Message from the Editor-in-Chief

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