



## Molecular Genetics of Disorders of Sex Development

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### Message from the Guest Editors

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Disorders (or differences) of sex development (DSD) are congenital conditions characterized by the atypical development of genetic, gonadal or phenotypic sex. DSDs include a wide spectrum of conditions mostly due to genetic variants, altered hormonal secretion or abnormal peripheral sensitivity to gonadal hormones, which are all able to change the typical male or female foetal development. Today, the improvement of genetic and endocrinological technologies permits the etiological diagnosis in 50–80% of individuals with DSD. However, an exact diagnosis remains unknown in some patients, in whom variants of uncertain pathogenicity or endocrine data of uncertain significance are found. In addition, rare patients with new or unusual laboratory or clinical findings are observed in practice.





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## Message from the Editor-in-Chief

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