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Revealing the Genomic Big Data: How Much Information Is Ready for Clinical Application?

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Message from the Guest Editors

In the last 10 years, the molecular approach for the study of genetic disorders has completely changed. Molecular diagnosis is based on the identification of the causative variant that allows genetic counseling, prenatal diagnosis, understanding of pathological mechanisms, personalized therapeutic approaches. This Special Issue aims to summarize our current abilities in the investigation of the human exome/genome as well as the clinical utility and applicability of the knowledge about coding and non-coding genomic regions. This Issue intends to cover: (i) the analytical sensitivity of different DNA sequencing technologies for the identification of both single-nucleotide and copy number variants; (ii) methods for investigating the functional roles of synonymous, 5'UTR and 3'UTR variants; (iii) the possibility of establishing a frequency threshold of variants' allele frequency in different populations, and how referring to a "pan-genome" would increase our power to connect variation to human diversity and disease; and (iv) the current application of multi-omics approaches and how they could improve the diagnostic power of genetic testing.

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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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