



Human Genome Sequencing in Health and Disease

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Message from the Guest Editors

This Special Issue on "Human Genome Sequencing in Health and Disease" will mainly focus on the following trends, addressed as reviews, new methodological approaches and original research:

- Whole genome sequencing developments in the identification of CNVs and trinucleotide repeat expansion
- Improving the interpretation of genomic data
- Genomic-based cancer diagnosis and treatment
- Diagnosing neurodevelopment disorders
- RNA vs. DNA sequencing: strategies to improve diagnostic yield
- Diagnosis and treatment of rare hereditary diseases
- Unraveling missing heritability with genomic sequencing
- Neomutations, gonadal mosaicism or parent low-level mosaicism: new perspectives from deep sequencing
- Pharmacogenomics in clinical practice: are we ready?
- Liquid biopsy: current state and future perspectives
- Transcriptomic applications in cancer research and in clinical practice





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Message from the Editor-in-Chief

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