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## Human Genome Sequencing in Health and Disease

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Deadline for manuscript  
submissions:

**closed (31 January 2024)**

### **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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# Special Issue



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## Message from the Editor-in-Chief

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