Special Issue

Cytogenetics and Cytogenomics in Clinical Diagnostics: Innovations and Applications

Message from the Guest Editor

Chromosomes serve as both structural and regulatory units of the human genome. Clinical cytogenetics and cytogenomics enable the diagnosis of constitutional and oncological chromosomal abnormalities. While conventional karyotyping provides genome-wide analysis at low resolution, the integration of fluorescence in situ hybridization (FISH), microarray analysis, and optical genome mapping (OGM) since the 1990s has significantly enhanced diagnostic precision and sensitivity.This Special Issue invites submissions on chromosomal abnormalities and submicroscopic genomic variations in developmental disorders, neuropsychiatric diseases, and neoplastic conditions. We welcome research on mechanistic insights, genotype–phenotype correlations, technological innovations, and clinical applications. Studies on three-dimensional chromosome organization in human diseases and AI-driven advancements in cytogenomics are particularly encouraged.

Guest Editor

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Deadline for manuscript submissions

closed (15 January 2026)

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Genes is central to our understanding of biology, and modern advances such as genomics and genome editing have maintained genetics as a vibrant, diverse and fast-moving 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. Why not consider *Genes* for your next genetics paper?

Editor-in-Chief

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