

Special Issue

Application of Genome-Wide Association Studies in Rare Diseases Research

Message from the Guest Editors

Genome-wide association studies (GWASs) have revolutionized our understanding of genetic contributions to complex traits and diseases. This Special Issue aims to highlight innovative methodological approaches and applications of GWASs in rare disease contexts. We welcome original research articles and reviews addressing novel statistical frameworks, meta-analytic approaches, integration of multi-omics data, and innovative study designs that overcome sample size limitations. This Special Issue will showcase advances in polygenic risk scores for rare conditions, cross-ancestry GWAS applications, and gene–gene interaction (epistasis) and gene–environment interaction findings that may have outsized effects in rare disease etiology. We particularly encourage submissions exploring how environmental factors may modify genetic risk in rare diseases and methodological advances for detecting epistatic effects despite limited sample sizes. Contributions demonstrating successful translation of GWAS findings into clinical applications for rare disease diagnosis, prognosis, and treatment are also highly welcomed.

Guest Editors

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Deadline for manuscript submissions

closed (25 April 2026)

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About the Journal

Message from the Editor-in-Chief

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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