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Genetics and Genomics of Skeletal Disorders

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Deadline for manuscript submissions:

closed (20 August 2022)

Message from the Guest Editors

The identification of the mechanisms underlying the pathophysiology and therapies of skeletal disorders continues to be an area of significant research. More than 460 different disorders are included under the definition of skeletal disorders, divided in 42 distinct groups according to their phenotype and genotype. Furthermore, these entities present a high level of clinical and genetic heterogeneity; in many cases, mutations in a single gene may lead to a broad variability in clinical expression intra- and extra-familially. Over the past two decades, advances in genetic and genomic technologies have strengthened the field of skeletal disorders, increasing the molecular knowledge and discovering the genetic defects underlying many of these diseases. There are still many challenges regarding disease etiologies, pathways, natural history and possible new targets for future therapies; thus, research is highly needed. We welcome original research articles, case reports, reviews and mini-review articles addressing the aims in skeletal disorders. And we also welcome papers from ERN-BOND focusing on rare skeletal diseases.



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Special Issue



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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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