Bioinformatic Analysis for Rare Diseases

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Deadline for manuscript submissions:
closed (15 June 2019)

Message from the Guest Editors

Dear colleagues,

Rare diseases, especially Mendelian and monogenic, have played a critical role in elucidating the genetic basis of human diseases. The development of modern techniques such as next generation sequencing (NGS) combined with the increase of computing power and bioinformatic software development has enabled broader-scale research of rare diseases to better understand their biological mechanisms, genomic and proteomic basis, environment, and the combination of these different contributing factors. Therefore, especially in the light of incomplete clinical penetrance, some rare diseases are increasingly viewed as complex diseases. Large volumes of biological data at various levels have been exponentially accumulated over the last decade, including NGS (whole genome/exome sequencing, RNA-seq, DNA methylation), proteomics, and metabolomics. These complex and large data pose challenges for bioinformatic analyses, especially in the interpretation and understanding of biological mechanisms underlying diseases. We welcome submissions of reviews, research articles, short communications, and concept papers.

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Guest Editors

Special Issue

Editor-in-Chief

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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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