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Utilizing RNA-Seq and Genome Sequencing to Uncover Complexities of Genetic Disorders

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Message from the Guest Editors

Mining this data by investigating mechanisms and variations in the genome can help us better understand how certain variants contribute to a phenotype and the genetic pathways involved to determine the underlying causes of a genetic disorder.

Various NGS applications, particularly whole-genome (WGS) and exome sequencing, are powerful tools for diagnosing the underlying causes of genetic disorders. However, despite the recent advances in NGS technologies, their diagnostic rate is limited. Augmenting it with RNA-Seq can facilitate in resolving the genetic basis of unsolved diseases, narrowing the diagnostic gap. Additionally, RNA-Seq can detect a wide variety of RNA species, including pre-mRNA, mRNA, and non-coding RNAs (ncRNAs), allowing for a deeper understanding of their role in the progression of a genetic disorder.

This Special Issue invites original research and reviews on topics utilizing the power of RNA-Seq in addition to other sequencing methods to determine causative variants in different diseases, including cancer, in research as well as in clinical applications.

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Message from the Editor-in-Chief

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