

Special Issue

Genomic Insights: Exploring the Applications of Sequencing Technologies in Genetics

Message from the Guest Editor

Next-Generation Sequencing technologies can improve our knowledge of the molecular genetics of human diseases, but may also lead to uncertainty during the interpretation of the results. Providing a definitive genetic diagnosis is critical in prenatal testing or management of severely affected paediatric patients and can be especially challenging in affected individuals in whom a broad unbiased sequencing approach such as whole-exome sequencing or whole-genome sequencing is potentially detrimental. Molecular studies are warranted to clarify the genetic basis of human diseases and emphasize the importance of functional studies in the diagnostic variant reclassification process.

Guest Editor

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