Special Issue

Breaking the Code: How Genome Sequencing Is Revolutionizing the Diagnosis of Rare Diseases

Message from the Guest Editors

Recent advances in genomic sequencing are transforming rare disease diagnosis and comprehension, giving patients and their families new hope around the world. More than 300 million people worldwide are affected by uncommon diseases, many of which are genetically based. Accurate and prompt diagnosis is still a major obstacle. Whole genome sequencing (WGS), next-generation sequencing (NGS), and other advanced genomic technologies have made it possible to identify disease-causing gene variations more quickly and accurately, opening the door to more targeted therapies and enhanced clinical outcomes. Original research and reviews examining the most recent developments in rare disease genome sequencing are requested for this Special Issue. We are looking for submissions on a variety of subjects, such as new diagnostic techniques, developments in bioinformatics, and the practical implications of genomic results. This Special Issue aims to highlight the revolutionary potential of genome sequencing in rare disease research and patient treatment by bringing together a variety of viewpoints from genetics, bioinformatics, and clinical practice.

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Life (ISSN 2075-1729) is an international, peer-reviewed open access journal that publishes scientific studies related to fundamental themes in life sciences. Some papers are published individually, while others are submitted for inclusion in special issues with guest editors. You are invited to contribute a research article, essay, or a review to be considered for publication.

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