Special Issue

Multiple Molecular Diagnoses in Rare Disease through Massive Parallel Sequencing Approach

Message from the Guest Editors

More than 6000 rare diseases have been described so far. On the whole, 80% of rare diseases are of genetic origin and are often chronic and life-threatening. Rare diseases are characterised by a wide diversity of symptoms and signs not only among different conditions but also between patients suffering from the same disease. Due to the low prevalence of each disorder, medical expertise is rare, knowledge is scarce, care opportunities are inadequate, and research is limited. Whole exome sequencing (trio-WES) analysis has substantially improved the chance of obtaining a genetic diagnosis in rare and ultra-rare diseases. The purpose of this Special Issue is to host particularly interesting complex case reports solved by WES with final multiple molecular diagnoses, as well as research and review papers on rare diseases and complex phenotypes in order to assess the real effectiveness of WES in solving complex diagnoses and to eventually stress pitfalls in this approach solved by alternative strategies (WGS, mDNA signature). Great attention will be given to accurate genotype–phenotype correlation in multiple molecular diagnoses in solving complex phenotypes.

Guest Editors

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Deadline for manuscript submissions

closed (25 November 2022)

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Genes

an Open Access Journal by MDPI

Impact Factor 2.8
CiteScore 5.5
Indexed in PubMed

mdpi.com/si/101583

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Message from the Editor-in-Chief

Genes is central to our understanding of biology, and modern advances such as genomics and genome editing have maintained genetics as a vibrant, diverse and fast-moving 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. Why not consider *Genes* for your next genetics paper?

Editor-in-Chief

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