Special Issue

Diagnosis, Management and Therapy of Rare Diseases

Message from the Guest Editor

More than 8500 rare diseases have been described to this date, and the advances in genomic analyses have substantially improved the chance of achieving a genetic diagnosis of these disorders. Of note, only approximately 5% of these conditions recognize an approved tailored treatment, limiting the possibilities to provide truly personalized management. This Special Issue aims at focusing on the role of clinical and molecular genetics in clinically complex cases, solved by applying the most advanced sequencing technologies, for which molecular diagnosis deeply modified the treatment and care of patients. In particular, this Special Issue includes, but it is not limited to, the following topics:

- Molecular mechanisms behind rare and ultrarare genetic diseases;
- Diagnosis, clinical features, and expansion of the phenotypic spectrum of rare and ultrarare genetic diseases;
- The clinical and molecular characterization of family members;
- Ethnic variability in rare and ultrarare genetic diseases;
- Gene-tailored therapeutic options (including pharmacogenomics);
- Clinical trials with which to develop new treatments.

Guest Editor

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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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