Special Issue

The Genotype/Phenotype Relationship and Therapy Optimization in a Monocentric Fabry Disease Cohort

Message from the Guest Editor

In Fabry disease (FD), genetic factors play a key role both in early diagnosis in potentially affected patients and in prognosis by optimizing the specific therapy timing. The data are ambiguous and limited only to male patients in the FOS database registry. However, histopathological lesions from renal biopsies have revealed similar features in both sexes and in early phase. Extending the analysis to women and children will further increase our knowledge of clinical and pathological findings of Fabry disease. Critical points to elucidate: the high proportion of de novo mutations of uncertain significance; the variable phenotype in members of the same family with the same mutation, suggesting underlying modifying factors that influence clinical presentation; familial pedigrees, to date not implemented in clinical routine. This Special Issue represents an useful "tune-up" on the current state of knowledge on FD, in light of the suggestion that FD patients' life expectancy may increase as more patients are beginning targeted treatment at an earlier age. In addition, FD therapeutic choices extend beyond enzyme therapy, substrate reduction therapy, and gene therapy.

Guest Editor

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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?

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