Special Issue

Genetics of Human Disease: Emphasis on X-Linked Diseases and Clinical Practice

Message from the Guest Editor

The susceptibility of an individual to almost all human diseases is influenced, to some degree, by genetic variation. There are different patterns of genetic inheritance, including X-linked diseases. For almost all X-linked diseases, clinical manifestation is more severe in males than in females. In carrier women, the presence of cellular mosaicism is considered an advantage because the presence of wild-type alleles in at least 50% of the cells allows them to manifest no clinical symptoms. Furthermore, the interaction between the cells expressing the wild-type or mutant X chromosome determines metabolic cooperation, which leads to the correction, at least in part, of the defect in the mutant cells. As a result, there is greater difficulty in the diagnosis of rare X-linked diseases, with a consequent delay in the choice of treatment options. The aim of this Special Issue is to summarize recent advances in genetics/epigenetics and related research on genotype–phenotype correlations in women with X-linked disorders, with the identification of screening techniques for diagnosis and treatment.

Guest Editor

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