# 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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The International Journal of Molecular Sciences (*IJMS*, ISSN 1422-0067) is an open access journal, which was established in 2000. The journal aims to provide a forum for scholarly research on a range of topics, including biochemistry, molecular and cell biology, molecular biophysics, molecular medicine, and all aspects of molecular research in chemistry. *IJMS* publishes both original research and review articles, and regularly publishes special issues to highlight advances at the cutting edge of research. We invite you to read recent articles published in *IJMS* and consider publishing your next paper with us.

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