

Special Issue

Molecular Defects of Naturally Occurring Mutations

Message from the Guest Editor

Naturally occurring mutations are nature-made experiments that yield novel insights into the physiological functions of the proteins encoded by these mutated genes. Functional studies of the mutant proteins provide important structure–function insights. These studies can be more fruitful than traditional mutagenesis experiments because the mutant proteins have important phenotypes. Mutant proteins can have either loss-of-function or gain-of-function. The studies of these mutant proteins can also point to potential strategies for precision medicine. For example, loss of function mutations can be potentially treated with pharmacological chaperones that correct the misfolding. This Special Issue will provide examples of these aspects of human genetics. Manuscripts dealing with mutations in other animals that have relevance to humans are also welcome.

Guest Editor

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