

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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Deadline for manuscript submissions

20 September 2025



International Journal of Molecular Sciences

an Open Access Journal
by MDPI

Impact Factor 4.9
CiteScore 9.0
Indexed in PubMed



mdpi.com/si/217091

*International Journal of
Molecular Sciences*
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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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