International Journal of
Molecular Sciences

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Molecular Effects of Mutations in Human Genetic Diseases

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Deadline for manuscript submissions:
closed (30 June 2021)

Message from the Guest Editors

The aim of this Special Issue is to attract high-quality studies describing computational and experimental approaches for investigating the molecular effects of novel genetic mutations, and providing a useful framework for understanding the molecular defects underlying human diseases. Contributors are also encouraged to submit articles describing use cases, models, and methodological innovations.

The Special Issue will include (1) human genetics studies on genome/exome or targeted sequencing panels that allow either identification of disease–gene associations, characterization of rare diseases with significant genetic heterogeneity, or differential clustering of disease mutations associated with distinct phenotypes; (2) experimental studies investigating how a genetic variant causes disease at the molecular, cellular, and organismal levels; and (3) computational methods devised to predict the impact of genetic variations and their assessment, large-scale statistical studies dissecting key features of disease mutations, or well-curated data repositories of genetic variation and/or disease associations.

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Message from the Editor-in-Chief

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