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Rare Diseases Associated with SNPs and Protein Structure Modifications

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Deadline for manuscript submissions:

closed (31 August 2022)

Message from the Guest Editors

Dear Colleagues,

Insufficient data limits our knowledge of rare diseases. Genetic disorders often manifest in mutated proteins with modified structure and functionality, and finally lead to changes in metabolic pathways and organism homeostasis. The discovery of the linkage between single-nucleotide modification and the observed dysfunction of the organism is a challenging task, and requires the cooperation of genetics, biochemists, structural biologists, and many other specialists.

The aim of the current Special Issue is to provide substantial contributions to the field examining the structure–function relationship of the changes in proteins caused by single-nucleotide polymorphisms or their combination. We welcome original research papers presenting novel computational/experimental studies, as well as reviews, in themes including, but not limited to:

- Newly identified disorders modifying proteins' function;
- Studies of the linkages of mutations in proteins with metabolic pathways;
- Structural analysis of the molecular basis of protein disfunction.

Prof. Dr. Sabina Wallmark Dr. Artur Góra Guest Editors













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