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Characterization of Molecular Mechanisms and Tailored Correction Approaches for Inherited Disorders

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Message from the Guest Editors

Mutations affecting coding and non-coding gene regions result in the alteration of the normal flow of genetic information at different levels. To this purpose, the identification of novel mutations through next-generation or whole-exome sequencing and case reports in the context of rare disorders, as well as experimental tools such as cellular and animal models of disease, provide relevant qualitative and quantitive information.

Moreover, the knowledge of disease mechanisms paves the way for the development of tailored correction approaches, ranging from genome-oriented strategies to intervention at the transcriptional/post-transcriptional (e.g., promoter activation or splicing modulation) or translational/post-translational (e.g., modulation of ribosome activity) levels.

Overall, the identification and characterization of new disease-causing molecular mechanisms, the widening of gene mutational patterns, particularly for rare diseases, and/or the possibility to correct a specific gene defect, thus counteracting the detrimental effects of mutations, represent important issues to address in order to improve and deepen our knowledge of human genetic disorders.

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Message from the Editor-in-Chief

As the world of science becomes ever more specialized, researchers may lose themselves in the deep forest of the ever increasing number of subfields being created. This open access journal Applied Sciences has been started to link these subfields, so researchers can cut through the forest and see the surrounding, or quite distant fields and subfields to help develop his/her own research even further with the aid of this multi-dimensional network.

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