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Complex Molecular Mechanism of Monogenic Diseases 2.0

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Message from the Guest Editor

Monogenic diseases are defined as genetic disorders caused by mutations in single genes. Therefore, one could assume that their mechanisms might be relatively simple, as a defect in one gene should cause dysfunction of just one protein or functional RNA molecule. However, recent studies have indicated that molecular mechanisms of monogenic diseases are significantly more complicated. Dysfunction of one gene product results not only in the inactivation of just one biochemical reaction, but a network of various reactions is affected. Then, secondary and tertiary effects sometimes lead to dysregulation of various cellular processes, including the up- or down-regulation of the expression of many genes, and disturbance of the physiology of cells, tissues, organs, and whole organisms. We are only at the beginning of understanding the complicated molecular mechanisms of monogenic diseases.