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Changing Diagnosis, Treatment and Monitoring of Rare Genetic Disorders in the -Omics Era

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Message from the Guest Editors

To date, thousands of rare genetic disorders have been identified. Individuals with these disorders have a significantly increased risk of morbidity and mortality. The complex nature of these disorders, combined with a lack of knowledge and expertise, often results in a deficiency of adequate screening methodologies and/or delayed or inconclusive diagnoses. Although a genomic diagnosis may be definitive, the absence of appropriate diagnostic, prognostic, or treatment-responsive biomarkers (or biosignatures) significantly impacts a patient's quality of life and causes substantial burden to carers, the health system, and the economy.

Understanding disease pathology and being able to screen for and diagnose these disorders through the discovery of novel biomarkers is essential. Furthermore, the application of innovative data analytics and predictive neural networks (combined with metabolomic, proteomic, transcriptomic, lipidomic, and other -omic findings) are accelerating the discovery of rare disease biomarkers and biosignatures.

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Message from the Editor-in-Chief

The metabolome is the result of the combined effects of genetic and environmental influences on metabolic processes. Metabolomic studies can provide a global view of metabolism and thereby improve our understanding of the underlying biology. Advances in metabolomic technologies have shown utility for elucidating mechanisms which underlie fundamental biological processes including disease pathology. *Metabolites* is proud to be part of the development of metabolomics and we look forward to working with many of you to publish high quality metabolomic studies.

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