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Advances in Fabry Disease: From Molecular Insights to Innovative Therapeutics

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Message from the Guest Editors

Fabry disease, a rare X-linked lysosomal storage disorder, results from mutations in the GLA gene, leading to deficient α-galactosidase A enzyme activity. This deficiency causes glycosphingolipids to accumulate in various tissues, affecting the kidneys, heart, skin, and nervous system. Recent research has advanced our understanding of Fabry disease's pathophysiology, clinical manifestations, and treatment options, including enzyme replacement therapy (ERT) and chaperone therapy. However, challenges remain in early diagnosis, disease management, and the development of more effective adjuvants and specific treatments.

This Special Issue aims to consolidate cutting-edge research and foster multidisciplinary collaboration to address these challenges. Original research articles and reviews that explore novel diagnostic biomarkers, genetic and molecular basis, and innovative therapeutic approaches. Clinical studies that offer new insights and advancements in imaging findings, patient-reported outcomes, and the long-term efficacy of current treatments are also encouraged.

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Message from the Editor-in-Chief

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