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Cellular and Molecular Mechanisms of Nephropathic Cystinosis

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Message from the Guest Editor

Dear Colleagues,

Nephropathic cystinosis (MIM # 219800) is a rare autosomal recessive disorder caused by mutations in the lysosomal cystine transporter cystinosin, encoded by the *CTNS* gene (17p13.2). The disease is characterized by lysosomal cystine accumulation in all cells of the body. Cystinosis usually manifests as a general proximal dysfunction, also called renal Fanconi syndrome; however, during the course of the disease, many other organs (eyes, endocrine glands, muscles, central and peripheral neural systems, bones, and liver) become affected.

In this Special Issue, we will highlight the state-of-the-art in our understanding of cellular and molecular mechanisms of nephropathic cystinosis, opening new horizons for innovative treatment strategies for cystinosis and potentially other lysosomal storage diseases.

For further reading, please visit the Special Issue website.









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