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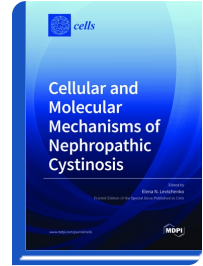
*Special Issue Reprint*

## Cellular and Molecular Mechanisms of Nephropathic Cystinosis

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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). This devastating condition initially affects kidneys and subsequently many other organs including eyes, thyroid, pancreas, muscles, and brain. While lysosomal cystine storage is a key feature of the disease and the main target of current therapy, recent groundbreaking research has revealed that cystinosin has diverse functions in cells, being involved in vesicle trafficking, energy homeostasis, and cell death mechanisms. These discoveries deepen our insights into the mechanisms of cystinosis and of lysosomal biology in general. In this Special Issue dedicated to the pioneer of cystinosis research Dr. Jerry Schneider, we highlight the state-of-the-art understanding of cellular and molecular mechanisms of various disease features, opening new horizons for innovative treatment strategies for cystinosis and potentially other lysosomal storage diseases.



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