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Genetics of Tubulopathies

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Message from the Guest Editors

One of the key tasks of the kidney is homeostasis, the maintenance of the extracellular fluid in which all cells are bathed. Ensuring the proper composition and volume of this “milieu interieur” or internal environment is critical for the function of all cells. Homeostasis is mainly provided by the renal tubules and involves a large number of different transport proteins and regulators. Defects in these tubular transport processes are collectively referred to as tubulopathies. The associated clinical phenotype can vary enormously in severity: from minor abnormalities in urine composition, to kidney stones, hypertension, chronic kidney disease, or life-threatening acid-base and electrolyte abnormalities. Genetic investigations into these rare disorders have clarified the aetiology of most tubulopathies and provided tremendous insights into their underlying pathophysiology.

This Special Issue focuses on the genetics and genomics underlying the diseases of tubulopathy, to better understand the disease causes and mechanisms and to identify and develop better treatments.



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Special Issue



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Message from the Editor-in-Chief

Genes are central to our understanding of biology, and modern advances such as genomics and genome editing have maintained genetics as a vibrant, diverse and fastmoving field. There is a need for good quality, open access journals in this area, and the *Genes* team aims to provide expert manuscript handling, serious peer review, and rapid publication across the whole discipline of genetics. Starting in 2010, the journal is now well established and recognised.

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