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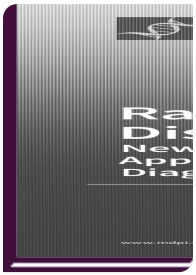
Special Issue Reprint

Rare Kidney Diseases



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Rare kidney diseases comprise a large group of different life-threatening or chronically debilitating disorders that affect very small numbers of people (<1 in 2000 individuals in Europe and <200,000 in USA) with local or systemic manifestations. For several years, the research and development of treatments in this field have been neglected in favor of more common diseases. The main reasons for the lack of interest in rare kidney diseases seem to be the small numbers of patients and limited epidemiological data on the natural history of many of these diseases. Rare diseases can affect people differently. Even patients with the same condition can exhibit very different signs and symptoms, or there may be many subtypes of the same condition. This diversity constitutes a significant challenge to healthcare practitioners and scientists alike, in terms of being able to acquire sufficient experience for the most appropriate and timely definition, diagnosis, and management. Fortunately, in the last ten years, concerted efforts have led to a marked improvement in the understanding of these disorders. In particular, an important step forward has been taken with the employment of innovative technologies (including next-generation sequencing), in order to replace obsolete phenotypic classifications and to discover new useful diagnostic biomarkers. These new tools are, in fact, becoming part of routine clinical practice, increasing diagnostic accuracy and facilitating genetic counseling. Moreover, biomedical research, providing insights into the pathologies of these rare diseases and elucidating their underlying mechanisms, is revealing new therapeutic avenues and driving the industry to develop safer and more effective orphan drugs. Finally, in this field, it is desirable that, in the future, closer cross-talk between basic scientists and clinicians could achieve a great clinical result, improving the quality of life of these patients as well. This Special Issue welcomes original contributions and critical reviews describing new pathogenetic insights, reporting novel and specific disease biomarkers, and underlying new pharmacological targets or therapies for rare diseases of the kidney and urinary tract.

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