Pompe Disease

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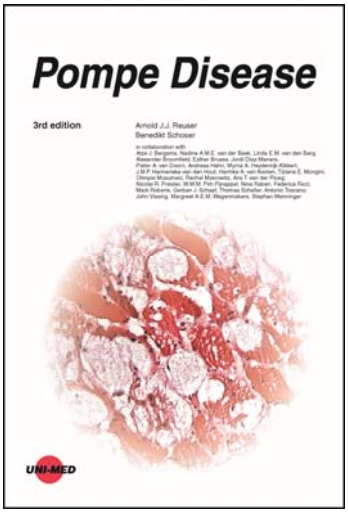

Pompe disease is a rare lysosomal storage disorder with growing therapeutic options. Because of rising awareness of Pompe disease over the past decades, less well-known clinical features have emerged, and the number of diagnosed patients has increased around the world. Furthermore, embedded research is turning into an increased understanding of the pathophysiology and has opened the way for novel therapeutic approaches.
This edition highlights the significant advances in the understanding of disease mechanisms, in describing the phenotypic spectrum of Pompe disease, in developing diagnostic algorithms and in improving patient care. We summarized the current techniques contributing to disease diagnosis, predictive genetic testing and therapy development.
This third edition of 'Pompe Disease' was updated particularly with regards to the clinical spectrum, the diagnostic procedures, the efficacy of enzyme replacement therapy, and future gene therapy. It provides helpful information to healthcare professionals as well as to interested lay people on all aspects of Pompe disease.

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