Pompe Disease

Martina Baethmann, Volker Straub, Arnold J.J. Reuser


Pompe disease is a rare lysosomal storage disorder with limited therapeutic options. With rising awareness of Pompe disease over the past few years less well known clinical features have come to light and more patients have been diagnosed, and there is increasing knowledge about pathophysiology and therapeutic approach. This second edition considers the significant advances in the understanding of disease mechanisms, in delineating the phenotypic spectrum of Pompe disease, in developing diagnostic algorithms and in improving patient care. It also addresses new technologies concerning their supposed significant impact on disease diagnosis, predictive genetic testing and therapy development in the coming years. This book was updated especially regarding enzyme replacement therapy, diet, respiratory care and physiotherapy and provides helpful information to healthcare professionals as well as to interested lay people on all aspects of Pompe disease.