

Foreword

Over the past few decades we have seen remarkable progress in our understanding of the lysosomal storage diseases (LSDs) thanks to many physicians and scientists who have devoted their careers to unraveling the clinical, biochemical and molecular intricacies of these rare disorders. Dr Pastores has been both a witness and a contributor to the emergence of this field close to the forefront of medical genetics. This concise guide is a distillation of his experiences caring for patients, teaching medical students, residents and fellows, conducting clinical trials and participation in national and international meetings devoted to progress in the LSDs.

He begins by emphasizing that these disorders involve all age groups and multiple organ systems so that physicians caring for both children and adults and from all specialties need to be informed. Historical information of each disorder and vignettes liven up the text. The complexity of diagnosis is simplified by an emphasis on clinical signs and a paradigm for diagnostic testing. Guidance is given on biochemical and molecular testing and prenatal diagnosis and screening for carriers. Laboratory pitfalls such as pseudo-deficiency alleles and activator protein deficiencies are addressed in this well-referenced and up-to-date monograph. Disease mechanisms, a relatively new field of inquiry, are nicely summarized yet there is much about pathogenesis of the LSDs that remains unknown. However, what makes the LSDs of such great interest today are the multiple approaches to therapy, some already in use and others in clinical trials and potentially promising. To measure treatment progress, disease-specific scoring systems are needed as well as quality of life measures and these are also well-covered. The many tables in this manual allow even

the beginning student convenient shortcuts to differentiate the various LSDs on clinical grounds.

Even though rare, the LSDs continue to spawn new insights into basic molecular processes with the potential for wider applications to other categories of genetic disease. Both new and established students of the LSDs will find this book a roadmap not only for better case finding and management but also a stimulus to continue the tradition of discovery that is the lifeblood of academic medicine.

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