sins can lead to cell death. These seven 'sins' are the generation of (1) genetic mutations and (2) secondary structures, the promotion of (3) DNA breakage and (4) DNA recombination, (5) the activation of DNA damage response, (6) the alteration of gene expression by disrupting the methylation pattern, and (7) the alteration of gene expression by disrupting chromatin assembly. This is an appealing vision of both the etiology of cancer and the impact of replication errors, which place tremendous stress on the mechanisms that prevent or repair DNA damage. Examples of the actions of these sins in several diseases are provided (particularly effectively in the case of nucleotide-repeat disorders) and a brief look at medicines targeting DNA replication errors, including antivirals (HIV and HBV therapies) and anti-cancer drugs (topoisomerase inhibitors and others) is described.

The final chapter offers an overview of 'DNA and ability to reproduce: the 'Secret' of evolution', the pivotal theme of the book. This chapter opens novel perspectives on various topics such as the move from RNA to the more complex world of DNA, the role of viruses in the evolution of DNA genomes, the gene invasion from viruses, the intron expansion, the variegate multiple origins of replication in Eukarya, and the deeper significance of viruses (are they fossil records of evolution?).

This book, on the other hand, is very comprehensive and up-to-date, and will be of particular interest to molecular biologists and human geneticists■

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A useful guide for your practice

Practical, brief, yet comprehensive – highly recommended book for practitioners

'Lysosomal Storage Diseases: Early Diagnosis and New Treatments' Edited by Rossella Parini and Generoso Andria
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Published by: Mariani Foundation Paediatric Neurology 23: Montrouge, France, Editions John Libbey Eurotext, 2010, 186pp, Hardback, €70 (approx.)

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I was very pleased to discover a very practical and comprehensive material, containing updated information on basic and clinical research in the field of lysosomal storage diseases, a field that has witnessed an extraordinary evolution in the last 15 years.

The material is structured in four major parts: 'general aspects', 'clinical presentations in detail', 'mucopolysaccharidoses from the specialists' point of view' and 'specific treatments for lysosomal storage diseases', each of them rich in practical data, reflecting the author's experience in the field. A remarkable aspect is that all the presentations concentrate on early recognition and diagnosis, specific therapies being more efficient in incipient stages.

The first part introduces lysosomal storage diseases, discussing common features that suggest the presence of a storage disorder, as well as specific features that make the difference between entities. The next section (epidemiology, biochemistry and genetics) illustrates how to read and interpret biochemical and molecular tests. Suddenly, the complex field of metabolic disorders becomes very understandable after reading this material. This part also presents organizational and ethical aspects of newborn screening, as well as pathophysiological aspects of lysosomal storage disorders in a very comprehensive, yet realistic way. Even if the text is clear, I think some schemes and graphs are necessary to illustrate biochemical pathways, pathophysiology and interpretation of lab results.

The next section remarkably discusses two of the most complex lysosomal storage diseases – mucopolysaccharidoses and Anderson–Fabry disease. Illustrative photographs and imagistic investigations appropriately complete the text for both of them. In my opinion, the value of this material is given by the discussion in detail of early signs and symptoms, separately for different fields involved (dysmorphology, ENT, musculoskeletal, etc). Updated data are included and the most important mechanisms and theories are discussed.

The third part refers to mucopolysaccharidoses from the specialists' point of view. Once you start reading this section, you cannot put it away – you become more and more eager to discover something new about the next specialist's opinion. Classical features are mentioned, but here, the accent is put on practical issues that practitioners have to solve in their daily activity – epilepsy, psychological assessment and support, radiological findings, anesthesia, as well as neurosurgical complications and their management. The rich selection of illustrations makes the material practical and instructive.

Physicians should not miss the last part referring to specific treatments for lysosomal storage diseases. In addition to the presently used enzyme replacement therapy and hematopoetic stem cell transplantation, other therapeutic approaches are discussed (eg, allogeneic stem cell transplantation and hematopoetic stem cell gene therapy). Each of the chapters included in this section is a state-of-the-art description in the field. I am convinced that readers will appreciate the therapeutic goals and the follow-up and management indications added to the classical presentation of therapy.

The bibliography is remarkable – it includes hundreds of very interesting articles, some historical, most of them recent ones.

In conclusion, this is an excellent book that should be available in every medical service that deals with lysosomal storage disorders (pediatric and neurological wards, medical genetics centers, and medical school libraries). The material is dense, but provides you with comprehensive and updated data in the field■

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