BOOK REVIEW

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Organelle Diseases. Edited by Derek A. Applegarth, James E. Dimmick, and Judith G. Hall. London: Chapman & Hall Medical. Pp. 454. £150. (Available in North America)

Inherited metabolic diseases now represent an enormous field within medicine. Recent biochemical and molecular-genetic advances have revolutionized our understanding of these disorders. Although fundamental knowledge and classification have improved, this wealth of new information is not always packaged into a coherent form for the clinician who is faced with an ill patient. This is an even greater challenge for the junior trainee trying to establish a practically useful framework upon which such diseases may be approached.

The editors of *Organelle Diseases* have taken on the challenge of providing a comprehensive account of the clinical features, diagnosis, and management of selected inherited metabolic diseases. So how does this newcomer differ from the well-established texts on metabolic disease? The answer lies in the interesting and fundamentally biological approach that has been taken. Rather than choosing a particular metabolic pathway as their starting point, the authors have elected to describe metabolic diseases on the basis of the organelle. This is a logical division of metabolic diseases, from a functional point of view, and I think it works well.

The editors have chosen to consider diseases of the lysosome, the peroxisome, and the mitochondrion. The endoplasmic reticulum, the golgi apparatus, and the nucleus could arguably also have a rightful place in such a disease taxonomy, but the editors point out that knowledge about disease due to dysfunction of these latter organelles is still relatively underdeveloped. Furthermore, according to data obtained in the editors' pediatric practice, metabolic disease due to dysfunction of the three organelles under consideration might account for as much as 50% of cases, once phenylketonuria and galactosemia are excluded. Since the latter two disorders are routinely screened for in most Western countries, it follows that diseases of these three organelles represent a significant proportion of newly presenting undiagnosed metabolic disease.

Given this disease taxonomy, the book's layout is logical, and it is lucidly written and well illustrated. Repetition, often a problem in mutiauthor texts, is minimal, although the chapters can each be read individually. For each organelle, basic biology, clinical features, laboratory diagnosis, and disease pathogenesis are considered. Forty lysosomal and 15 peroxisomal storage diseases are covered, in addition to all the well-recognized diseases associated with mitochondrial respiratory-chain dysfunction. Non respiratory–chain mitochondrial diseases are not considered.

As a physician, I found the clinical chapters particularly informative. I believe that this material should address most of the practical and genetic questions that arise in the clinic. The mitochondrion is privileged in having two chapters devoted to laboratory diagnosis, one detailing biochemical investigations and the other considering mtDNA analysis. The clinical chapter on mitochondrial disease discusses the relative merits of these two approaches to mitochondrial disease and indicates that not all cases require both levels of diagnosis. The general principle that a detailed clinical evaluation will allow the most efficient and directed use of diagnostic tests is emphasized throughout.

Following the three main sections, which consider each organelle in turn, a fourth section, entitled "Practical Issues," considers the still-challenging area of therapy, the importance of the family history, and the physical examination and how it may provide important clues to diagnosis and the metabolic autopsy. Finally, the appendices include information about DNA banking, useful addresses for patients, useful Websites, and a detailed glossary.

It is clear that the editors are active clinicians, since, although the basic science and pathogenesis of organelle diseases are up to date and comprehensively considered, they have ensured that this is an eminently practical text. I have little doubt that *Organelle Diseases* will be of considerable interest to pediatricians dealing with metabolic disease, and I think that it will also be useful to clinical neurologists, who often encounter the forms of these disorders that present in adolescence and adulthood. Basic scientists interested in developing biochemical and genetic diagnostic services and those who wish to learn more about the clinical aspects of these diseases will also find much of interest. The editors should be congratulated, and I look forward to their sequel covering the other organelles!

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