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BOOK REVIEW

Gaucher Disease

(A. Futerman and A. Zimran, eds., CRC Press, Taylor & Francis Group, Boca Raton, FL, USA, 2007, 528 p., $179.95)

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The book summarizes the latest achievements in the study of Gaucher disease, which was first described by the French physician Philippe Gaucher in 1882. Various chapters of the book were written by internationally known experts from many countries who made significant progress in the study of different aspects of Gaucher disease on clinical, biochemical, molecular biological, and genetic levels.

The book consists of 29 chapters, subject index, and bibliography to each chapter.

Chapter 1 (R. Brady) presents a short historical overview of basic steps in the study of Gaucher disease starting with description of the first 23 year old patient with significant increasing spleen size. Later accumulating products, glucocerebroside and glucosphingosine, were identified, and this observation was critical for discovery of enzymatic abnormality in Gaucher disease which is a hereditary deficiency of lysosomal enzyme β-glucocerebrosidase. Enzyme replacement therapy (ERT) for patients with Gaucher disease was approved for clinical practice in 1991. The biotechnological company Genzyme (USA) had significant impact in ERT of Gaucher disease.

Chapter 2 (K. Hruska et al.) highlights data about molecular biology and genotype–phenotype correlations in Gaucher disease. The glucocerebrosidase gene and identification of mutant alleles in Gaucher disease are characterized in detail. A special part of this chapter deals with genetic counseling in Gaucher disease.

Chapter 3 (G. Grabowski et al.) is devoted to cell biology and biochemistry of glucocerebrosidase. The chapter contains data about enzyme expression in normal tissues, peculiarities of enzyme biogenesis and lysosomal localization, effect of detergents, bile salts, and negatively charged phospholipid modifiers on enzyme activity. There are also data about glucocerebrosidase substrate specificity and it inhibitors and natural activators, e.g. saposin C.

Chapter 4 (S. Hoops et al.) deals with activator proteins (saposin C and others) increasing glucocerebrosidase activity. One part of this chapter is devoted to characterization of storage diseases developing due to hereditary deficiency of various types of saposin.

Chapter 5 (L. Premkumar et al.) discusses the X-ray structure of human glucocerebrosidase and implication of these data for second-generation ERT.

Chapter 6 (A. Futerman) summarizes cellular pathology in Gaucher disease. There is information about accumulation of glucosylceramide in cells and pathology of macrophages and neurons.

Chapter 7 (J. Shayman) deals with biochemistry and cellular biology of sphingolipids and glucosylceramide in the norm and in Gaucher disease.

Chapter 8 (E. Schuchman, S. Muro) discusses some problems in enzyme correction of Gaucher disease. Significant attention is devoted to the targeting of glucocerebrosidase by specific markers, which increase binding and delivery of this enzyme into lysosomes. Delivery into lysosomes of others enzymes, β-glucuronidase, α-L-iduronidase, α-glucosidase, and sphingomyelinase are also discussed.

Chapter 9 (Y. Sun et al.) deals with animal models of Gaucher disease. Major attention is directed to characterization of mouse models of Gaucher disease with various types of mutation in the glucocerebrosidase gene.

Chapter 10 (P. Mistry, A. Zimran) describes clinical features of Gaucher disease type 1. There are numerous data about changes in various organs, tissues, and cells.

Chapter 11 (R. Schiffmann, A. Vellodi) highlights data about a neuronopathic form of Gaucher disease. There is characterization of abnormalities in many body systems and severe clinical syndromes including seizures, mental retardation, serious ocular problems, enlarged organs, and abnormalities of the skeleton.

Chapter 12 (R. Lee) summarizes data about pathologic anatomy in Gaucher disease. There are discussions of abnormalities in spleen, liver, bones, lung, and brain. A special part of this chapter presents numerous pictures of tissue sections from various organs and description of electron microscopic photos.

Chapter 13 (K. Wong) highlights neuropathological aspects of Gaucher disease.

Chapter 14 (C. Hollak, J. Aerts) is devoted to diagnosis of Gaucher disease based not only on observation of β-glucocerebrosidase deficiency and specific storage products, but also based on various biochemical abnormalities. Clinical applications of surrogate biochemical markers of Gaucher disease, e.g. chitotriosidase enzyme activity which significantly increases in Gaucher disease, is also discussed.

Chapter 15 (M. Maas, E. Akkerman) focuses on bone pathology in Gaucher disease and application of magnetic resonance imaging as the most sensitive technique to evaluate bone marrow invasion.

Chapter 16 (G. Mariani, P. Erba) presents data about radionuclide evaluation of Gaucher disease.

Chapter 17 (P. Meikle et al.) deals with epidemiology and screening policy of Gaucher disease.

Chapter 18 (A. Zimran et al.) summarizes data about enzyme replacement therapy for type I Gaucher disease. Doses of enzyme and changes after it is injected including biomarkers level are discussed.

Chapter 19 (F. Platt, T. Cox) considers a comparatively new approach in Gaucher disease treatment based on use of specific inhibitors of biosynthesis of storage products with following decrease in their concentration e.g. under inhibition of imino sugars.

Chapter 20 (R. Desnick, J.-Q. Fan) deals with pharmacological chaperone therapy for lysosomal diseases: Fabry, Gaucher, Tay-Sachs, GM1 gangliosidosis, and other pathologies.

Chapter 21 (D. Begley) discusses the significance of the blood–brain barrier for treatment of Gaucher disease and other lysosomal storage diseases.

Chapter 22 (C. Peters, W. Krivit) characterizes some approaches for correction of Gaucher disease (type 1, 2, and 3) by hematopoietic stem cell transplantation, stem cells, and gene therapy.

Chapter 23 (D. Elstein, A. Steinberg) summarizes data about ethical concerns in treating rare diseases with expensive therapy.

In chapter 24 (D. Elstein, A. Israeli), societal aspects in treating rare diseases with expensive therapy are discussed. There is analysis of governmental health policy and role of the private sector in financing of treatment of patients.

Chapter 25 (J. Waalen, E. Beutler) deals with medical and economics aspects of Gaucher disease as a model for an orphan disease.

Chapter 26 (D. Meeker, H. Termeer) describes the first steps of Genzyme Corporation in preparation of glucocerebrosidase from human placenta and subsequent infusion into patients with Gaucher disease. The authors point out that enzyme replacement therapy is not considered as the final treatment procedure for Gaucher and others rare diseases. They believe that soon highly effective methods for treatment of Gaucher disease, including gene therapy, will be developed.

Chapter 27 (S. Lewis et al.) highlights Gaucher disease from the patients' perspective. There is characterization of centers and funds specialized in diagnosis, treatment, and financing of patients with Gaucher disease.

Chapter 28 (A. Alpert et al.) focuses on societal perspectives in organization of treatment of Gaucher disease.

Chapter 29 (Gaucher Associations) consists of a list of Gaucher associations around the world, which are involved in diagnosis and treatment of Gaucher disease; this includes their phone numbers and mail and website addresses.

The book is a very valuable source for specialists interested in various aspects of Gaucher disease: from physicians to biochemists, molecular biologists, geneticists, and biotechnologists. There is no doubt that this book will be very useful for medical schools and university students and their teachers involved in study of rare human diseases.

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