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BOOK REVIEW

The Metabolic and Molecular Bases of Inherited Disease

(Scriver, C. R., Beaudet, A. L., Sly, W. S., Valle, D., Childs, B., Kinzler, K. W., and Vogelstein, B., eds., 8th ed., McGraw-Hill, New-York, 2001, 7012 p., $550.00)

The previous seven editions of this book are well known to the large audience of specialists all over the world. Published in 1960, the first edition of the book titled "The Metabolic Basis of Inherited Disease" raised much interest among the specialists in the human inherited diseases field. The current eighth edition is significantly different from its predecessors. First of all the extent of the book is impressive. The book consists of 4 volumes with the total number of over 7000 pages. There are 30 parts, containing 255 chapters. All this material is written by a large international group of authors, with the total number of over 500 contributors. The editorial team of the book also has increased, now including three new editors, Barton Childs, Kenneth Kinzler, and Bert Vogelstein.

Volume I consists of seven parts and includes 76 chapters. Parts 1, 2 and 3 contain "Introduction", "Perspectives" and general themes about the "Human Genome" project, histocompatibility complex, human genetic mutations and models of animal (mice) inherited diseases. There are also chapters devoted to the nature and mechanism of genetic mutations, and biogenesis of membranes and organelles.

Part 4 is devoted to cancer. Reviewed are such questions as main concepts in cancer genetics, chromosome changes, cellular cycle of cancer cells and types cancer tumors: kidney carcinoma, skin cancer, breast cancer, brain, lungs, prostate and other organs and tissues.

Part 5 concludes the data about chromosomes, molecular cytogenetics and chromosome-linked diseases (Down syndrome, syndrome of fragile X chromosome).

Part 6 considers the clinical phenotypes of diseases and their diagnosis and algorithms.

Part 7 analyses disorders of carbohydrate metabolism. Different types of diabetes are characterized as well as diseases consistent with fructose and lactose metabolic disorders, galactosemia, glycogenosis, and diseases associated with disorders of disaccharide metabolism in the colon.

The second volume of the book begins with part 8, which contains the chapters elucidating disorders of amino acid metabolism. Among those diseases the focus is done on hyperphenylalaninemia, hypertyrosinemia, disorders in the metabolism of histidine, proline, oxyproline and ornithine, lysine, and glycine.

Part 9 summarizes the data about disorders of organic acid metabolism. There is information about alcaptonuria, disorders in propionate and methyl malonate metabolism, glutathione-synthetase deficiency and disorders of glycerol metabolism.

Part 10 gives the characteristics of mitochondrial functions disorders.

The chapters in part 11 are focused on the diseases associated with disorders of purine and pyrimidine metabolism. Xanthinuria, Lesch-Nyhan disease, adenine phosphoribosyl transferase and adenosine deaminase deficiency, and other pathologies are also characterized.

Part 12 discusses lipid metabolism under normal conditions and in inherited pathology. Chapters of this part contain the description of blood plasma lipoprotein structure and metabolism, various types of familial lipoprotein lipase deficiencies, diseases caused by bile acids biosynthesis deficiency, and other diseases.

Part 13 characterizes the diseases identified with disorders of porphyrin metabolism. Among those the authors review disorders of heme biosynthesis and disorders of bilirubin metabolism.

Part 14 contains 3 chapters about disorders of metal metabolism and transport. Described are diseases caused by disorders of copper transport, inherited hemochromatosis, and sulfite oxidase and molybdenum cofactor deficiency.

Part 15 includes the chapters concerning the diseases caused by disorders of peroxisome biogenesis, deficiency of a number of enzymes in these cellular organelles. X-Linked adrenoleukodystrophy, Refsum disease and primary hyperoxaluria are also described.

Volume III of the book includes the large part 16, which is dedicated to lysosomal diseases. Such disorders as mucopolysaccharidoses, type II glycogenosis, glycoproteinoses, glycolipidoses, and others are discussed in the 21st chapter.

Parts 17 and 18 describe disorders in vitamin and hormone metabolism accordingly.

Part 19 is about blood and homeostasis. Here are chapters about anticoagulating protein C, the disorders of fibrinogen and factor XIII metabolism, about hemophilia A and B, antithrombin deficiency, disorders in fib-

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rinolytic system, hemoglobinopathies, and other blood diseases.

Part 20 of the book is about the disease developing as a result of disorders of immune and defense systems of the organism.

Part 21 is dedicated to the membrane transport disorders. Among these diseases there are inherited renal glycosuria, cystinuria, cystinosis, cystic fibrosis, immunoglycinuria, and some other pathologies.

Parts 22 and 23 analyze diseases of connective tissue and cardiovascular system accordingly. Comparatively much attention in these chapters is paid to the disorders of collagen metabolism, amyloidosis, various types of achondroplasia, disruptions in blood pressure and cardiomyopathy.

Parts 24 and 25 describe inborn kidney and muscle diseases. Kidney abnormalities and muscular dystrophies are reviewed.

In parts 25 and 27 the diseases of lungs and skin are characterized, among those α-antitrypsin deficiency, albinism, disorders in organization of intermediate filaments and proteins bound to them.

Chapters of part 28 contain the information about the nerve system diseases. Among those Huntington's and Alzheimer's diseases are described, spinocerebral ataxia, inherited epilepsies, spinomuscular atrophy and other disorders of the nervous system.

Part 29 has chapters describing eye diseases. There are retinitis pigmentosa, stationary night blindness, disorders in color vision, inherited disorders of eye-lens functioning, glaucoma, and other diseases.

Part 30 analyzes the diseases with multiple developing processes defects observed.

Being evaluated as a whole, this book can be claimed as a unique and fundamental volume with a wide scope of practically all inherited human diseases. All parts and chapters of this book present disease mechanisms at the molecular and cellular level, and clinical and morphological characteristics of the diseases and approaches to their treatment as well.

The book is a reference aid for a wide range of specialists: biochemists, medical geneticists, molecular and cellular biologists, and physicians.

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