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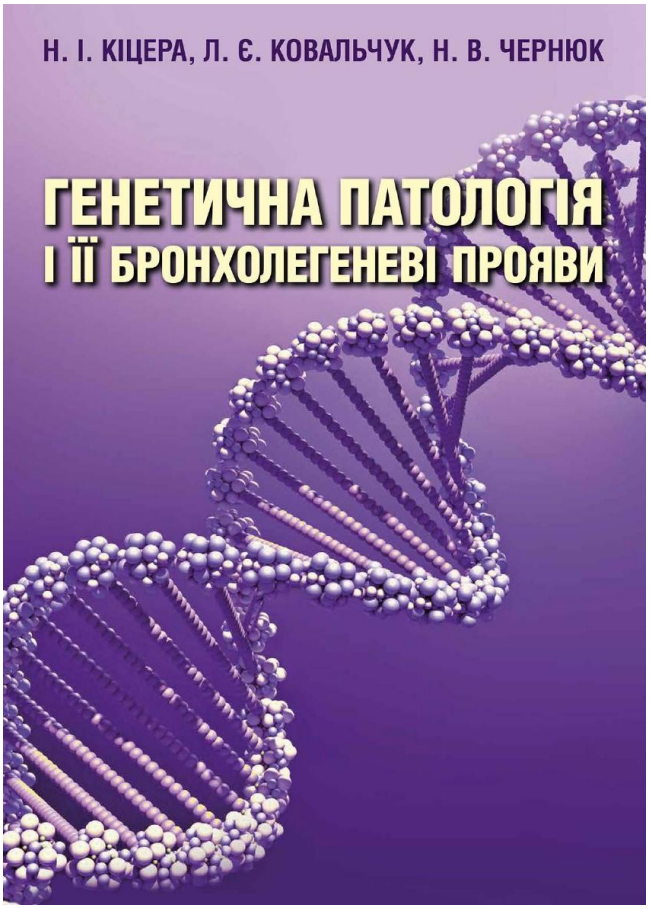
Book review



The textbook "Genetic Pathology and Its Bronchopulmonary Manifestations"

Kitsera N.I., Kovalchuk L.Y., Cherniuk N.V. Lviv: Kameniar; 2024. 464p. Illustrated.

The efficacy and efficiency of medical genetics depend on the level of knowledge among physicians, the thoroughness of analysis conducted, and the diagnostic capabilities concerning pathologies that are relatively uncommon within the population. This allows maximum assistance to patients with congenital and hereditary diseases and improves their quality of life. In recent years, despite the undeniable achievements of medical genetics, there has been an increase in genetically determined pathologies, a trend often linked to the intensification of the mutagenic influence of a polluted environment, the increase in the diagnosis of genetic diseases, and the development of new methods for diagnosing hereditary pathologies. Therefore, the relevance and significance of this work are beyond doubt.

The textbook authored by Doctors of Medical Sciences and professors Kitsera N.I., Kovalchuk L.Y., and Cherniuk N.V., titled "Genetic Pathology and Its Bronchopulmonary Manifestations," is dedicated to advancing the knowledge of bronchopulmonary pathology in hereditary diseases among physicians and medical students. This book represents the first scholarly work to comprehensively elucidate the essence of various chromosomal (such as Patau syndrome and Edwards syndrome), monogenic (such as Ehlers-Danlos syndrome and Osler-Weber-Rendu syndrome), X-linked, and multifactorial syndromes encountered in children, as well as describes pulmonary and bronchial complications associated with these conditions.

The textbook focuses on the understudied bronchopulmonary pathology within these syndromes. It provides a thorough analysis of the historical discovery of specific syndromes and incredibly intriguing biographical facts about the authors of the described syndromes. I have never encountered such a comprehensive approach to presenting the material about genetically determined pathology, rendering the textbook engaging and enlightening. The unconventional presentation of professional information intended for medical professionals and students creates a new paradigm for integrating foundational knowledge in the realm of genetics into the daily practice of the medical community.

The textbook is logically structured and aimed at achieving its stated goal – to describe the genetic basis of the most common hereditary diseases and syndromes, providing a detailed analysis of modern scientific research on bronchopulmonary disorders, their diagnostic features, and treatment options. It comprises six sections, each

containing information on bronchopulmonary manifestations of syndromes based on their mode of inheritance: in chromosomal disorders; in genetic diseases with autosomal dominant and autosomal recessive inheritance patterns (such as cystic fibrosis, Kartagener syndrome, Mounier-Kuhn syndrome, and ataxia-telangiectasia); in X-linked disorders (such as Rett syndrome, Duchenne muscular dystrophy, and loss of the SRY gene); in mitochondrial pathology (such as Kearns-Sayre syndrome); and multifactorial diseases (such as bronchial asthma, sarcoidosis, and chronic obstructive pulmonary disease). Such a structure of the book is entirely justified and raises no objections. The work spans 464 pages.

A positive aspect is that the textbook's content is enriched with graphical representation, incorporating photographs of individuals exhibiting clinical and phenotypic manifestations of syndromes and photographs of the authors who described syndromes and after whom they were named. Based on systematic and comprehensive approaches, the reviewed textbook thoroughly and comprehensively elucidates the genetic foundations of inheritance of specific syndromes and the underlying changes in the human bronchopulmonary system associated with the described syndromes.

The authors provide comprehensive insights into the historical exploration of syndromes and clinical manifestations of diseases, along with their prevalence in the population, mode of inheritance, risk factors for affected offspring, and diagnostic methods, including both prenatal and postnatal approaches, with a focus on bronchopulmonary manifestations. The potential treatment options and surgical interventions for congenital malformations associated with genetically determined syndromes are substantiated. Each section is abundantly illustrated with color drawings containing detailed annotations, computer tomograms, and radiograms, which facilitates enhanced understanding and retention of clinical manifestations associated with various genetic syndromes.

The textbook is noteworthy for its practical and constructive approach, designed to suit a wide range of medical professionals, including geneticists, pulmonologists, family physicians, pediatricians, and medical students. The authors highlight the importance of accurately diagnosing congenital genetic syndromes while also stressing the significance of early detecting bronchopulmonary pathology, which markedly complicates the course of any syndrome and frequently impacts the ultimate prognosis of the disease.

The textbook reviewed represents the culmination of extensive research conducted by Professor Kitsera N.I. at the Institute of Hereditary Pathology of the National Academy of Medical Sciences of Ukraine, focusing on studying the frequency and structure of congenital and hereditary pathologies, as well as examining the specific features of genealogical history associated with genetically determined conditions. The outcome explains genetically determined pathology as the underlying basis for all human diseases.

The most prevalent genetic syndromes and their bronchopulmonary manifestations are described using contemporary methodology. The research is characterized by its comprehensive nature, systematic approach, and practical orientation. An advantage of the textbook is its close integration of theoretical principles with the specific clinical presentation of pathology.

The textbook "Genetic Pathology and Its Bronchopulmonary Manifestations" logically concludes with two tables summarizing the presented material. Table 1, titled "Bronchopulmonary Manifestations in Patients with Genetic Syndromes," presents information on the mode of inheritance, mutation of specific genes, their location on the chromosomes, and the phenotype of the patient with a particular syndrome. Table 2, titled "Risk of Affected Offspring According to the Mode and Mechanism of Inheritance of Traits," delineates the probability of affected offspring alongside the birth of healthy children and carriers of the mutation responsible for a specific syndrome. Notably, the manifestation of each syndrome is delineated according to various modes of inheritance, indicating whether it occurs in every generation, exclusively in paternal lineage, transmitted solely by females, irrespective of gender, etc.

Having extensive experience in teaching medical genetics and pulmonology, counseling patients with various genetic syndromes, and treating bronchopulmonary manifestations in diverse genetic pathologies, the authors of the textbook - Kitsera N.I., Kovalchuk L.Y., and Cherniuk N.V., have synthesized their profound knowledge, conducted a thorough literature review, and briefly presented the most important information on genetic syndromes that are accompanied by bronchopulmonary complications. This knowledge is necessary for further study and understanding various syndromes' phenotypes.

The proposed textbook organically and practically integrates all components of the multifaceted process of providing

medical care for bronchopulmonary pathology to patients with genetically determined disorders.

The textbook is crafted to a high standard, printed on coated paper by the "Kameniar" in 2024, and supplemented with vibrant colored illustrations. It exemplifies an exhaustive exploration of the subject matter. The extensive reference list comprises 922 sources, with 846 authored by international scholars.

All the above highlights that the recommended textbook will benefit geneticists, pulmonologists, family physicians, healthcare professionals from diverse specialties, medical students, and individuals interested in the multifaceted issues pertaining to the manifestations of genetic syndromes.

Further investigation into the manifestations affecting other body systems in genetic pathology holds promise.

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