

Χαρακτηριστικά:

- Reflects the importance of **genetic understanding and genetic diagnosis** as the current approach to rare diseases
- Organizes content around anatomical systems, with **concise chapters** that cover discrete disorders and conditions
- Focuses on **diagnosis and management**, describing the **clinical, laboratory, imaging, and genetic diagnostic features** in every chapter to help differentiate disorders with similar symptoms or phenotypes
- Contains numerous figures, algorithms, tables, photographs, and radiographic images for **enhanced visual guidance**
- Includes chapters devoted to topics such as Ciliopathies, Neurodegeneration with Brain Iron Accumulation, Cancer Susceptibility Syndromes, Mitochondrial Disorders, Interferonopathies, and Epigenomic and Imprinting Syndromes, and others covering dysmorphology, neurologic, metabolic, genetic, and immune disorders
- Shares the knowledge and experience of editors who are **leaders in the field of rare diseases** in both the U.S. and Europe. Among the talented editors, Dr. Palau is editor-in-chief of *Orphanet Journal of Rare Diseases* and the first scientific director of CIBERER, the Spanish network of excellence in rare diseases. Dr. Basel is the medical director of the Genetics and Genomics Program at Children's Wisconsin, named a Center of Excellence by the National Organization for Rare Disorders (NORD)
- A **unique, comprehensive resource** for pediatric specialists, pediatric hospitalists, pediatric fellows, geneticists, and general pediatricians, and an ideal companion volume to *Nelson Pediatric Symptom-Based Diagnosis: Common Diseases and their Mimics* and *Nelson Textbook of Pediatrics*
- **An eBook version is included with purchase.** The eBook allows you to access all of the text, figures, and references, with the ability to search, make notes and highlights, and have content read aloud. Additional digital ancillary content may publish after the publication date

Περιεχόμενα:

Section I Introductory Chapters

1. Finding the Rare Among the Common: Diagnosis, Diagnostic Error, and When to Suspect a Rare Disease
2. Diagnostic Methods: Genomics, Omics, Biomarkers, Imaging, and Other Technologies
3. The Approach to Rare Diseases: From Local to Global

Section II Developmental Structural Disorders

4. Epigenetics, Genomic Imprinting, and Imprinting Disorders

5. Dysmorphology and Phenotyping
6. Ciliary Signaling and Dysmorphology
7. Ciliopathies: Clinical Presentations and Syndromes
8. Mosaic Overgrowth Syndromes
9. Ectodermal Dysplasia
10. Heritable Disorders of Connective Tissue
11. Genetic Disorders of Bone

Section III Neuro-Sensory Disorders

12. Rare Causes of Autistic Spectrum-Like Disorders and Syndromes With Autistic-Like Behaviors
13. Neurodegeneration With Brain Iron Accumulation
14. Disorders of Movement
15. Fever-Associated Seizures and Epilepsies
16. Nonfebrile Epilepsy Syndromes Including Epileptic Encephalopathies
17. Hereditary Motor-Sensory Neuropathies or Charcot-Marie-Tooth Disease and Related Neuropathies
18. Sensory and Autonomic Neuropathies Including Familial Dysautonomia and Small Fiber Neuropathies
19. Metabolic Myopathies
20. Skeletal Muscle Channelopathies: Periodic Paralysis and Nondystrophic Myotonias
21. Congenital Blindness
22. Late-Onset Blindness
23. Sensorineural Hearing Loss

Section IV Cardiopulmonary Disorders

24. Interstitial Lung Diseases of Childhood
25. Primary Ciliary Dyskinesia
26. Pulmonary Alveolar Proteinosis
27. Respiratory and Autonomic Disorders of Infancy, Childhood, and Adulthood (RADICA): Congenital Central Hypoventilation Syndrome (CCHS) and Rapid-Onset Obesity With Hypothalamic Dysfunction, Hypoventilation, and Autonomic Dysregulation (ROHHAD)
28. Pediatric Cardiomyopathies
29. Congenital/Familial Arrhythmia Syndromes
30. Vascular and Lymphatic Malformations

Section V Gastrointestinal and Hepatic Disorders

31. Very Early Onset Inflammatory Bowel Disease and Congenital Diarrheal Disorders
32. Pediatric Intestinal Pseudo-Obstruction
33. Rare Inborn Defects Causing Malabsorption
34. Genetic Etiologies of Neonatal and Infantile Cholestasis

Section VI Renal Disorders

35. Congenital Nephrotic Syndromes
36. Bartter and Gitelman Syndromes

- 37. Monogenic Etiologies of Hypertension
- 38. Genetic Etiologies of Hemolytic Uremic Syndrome

Section VII Endocrine Disorders

- 39. Autoimmune Polyglandular Syndromes and Other Disorders Associated With Immune-Related Endocrinopathies
- 40. Cancer Predisposition Syndromes in Children
- 41. Precocious and Delayed Puberty
- 42. Monogenic Diabetes: MODY and Other Rare Genetic Etiologies

Section VIII Metabolic Disorders

- 43. An Approach to Inborn Errors of Metabolism
- 44. Genetic Disorders of Neurotransmitters
- 45. Metabolic Crisis With Inborn Errors of Metabolism
- 46. Differentiating Features of Storage Diseases
- 47. Congenital Disorders of Glycosylation
- 48. Mitochondrial Disorders

Section IX Hematologic Disorders

- 49. Nonimmune Hemolytic Anemias
- 50. Lymphoproliferative Disorders
- 51. Hereditary Bone Marrow Failure Syndromes
- 52. Hereditary Etiologies of Thrombosis

Section X Immune/Inflammatory Disorders

- 53. When to Consider a Primary Immune Deficiency Disorder: Pathogens Associated With Specific Immune Defects
- 54. Primary T Cell Immunodeficiencies
- 55. Innate Defects in Host Defenses Against Infections
- 56. Rare and Unusual Etiologies of Atopic Diseases (Eczema, Urticaria)
- 57. Hereditary Autoinflammatory Disorders
- 58. Type I Interferonopathies
- 59. Disorders of Immune Regulation
- 60. Complement Deficiencies
- 61. Antibody Deficiencies