Cover image Title page Table of Contents Half Title Copyright Preface to the Fifth Edition Acknowledgements List of Abbreviations List of Gene Symbols Section A. The Biopsy: Normal and Diseased Muscle 1. The Procedure of Muscle Biopsy The Evolving Role of Muscle Pathology The Procedure of Muscle Biopsy Selection of the Patient Selection of the Muscle Technique of Biopsy Preparation of Specimen Cutting the Sections Electron Microscopy Specimen Preparation for Electron Microscopy Resins Immunohistochemistry 2. Histological and Histochemical Stains and Reactions Histological Stains Histochemical Reactions Histological and Histochemical Methods 3. Normal Muscle Histological Structure Muscle Fibre Types Histochemical Identification of Muscle Fibre Types Ultrastructure of the Myofibre Development of Human Muscle

4. Histological and Histochemical Changes Changes in Fibre Shape and Size Changes in Fibre Type Patterns Changes in Sarcolemmal Nuclei Degeneration and Regeneration Fibrosis and Adipose Tissue Cellular Reactions Changes in Fibre Architecture and Structural Abnormalities Deficiencies of Enzymes Accumulation of Glycogen or Lipid Accumulation of Amyloid Common Artefacts in Muscle Biopsies 5. Ultrastructural Changes Sarcolemma Myofibrils and Associated Cytoskeleton Z Line Intermediate Filaments Nucleus Mitochondria Membrane Systems **Deposits and Particles Other Unusual Structures** 6. Immunohistochemistry and Immunoblotting Immunohistochemistry Methods for Immunohistochemistry Baselines for Interpretation Use of Tissues Other Than Muscle Pathological Features of Diseased Muscle Immunohistochemistry of Other Tissue Components Immunoblotting Quantification Panel of Antibodies to Apply

7. How to Read a Biopsy

Part I

Part II

Part III

Part IV

Part V

Section B. Pathological Muscle: Individual Diseases

8. Classification of Neuromuscular Disorders

9. Neurogenic Disorders

General Pathological Features of Denervated Muscle

Spinal Muscular Atrophy

Histology and Histochemistry

10. Muscular Dystrophies and Allied Disorders I: Duchenne and Becker

Muscular Dystrophy

Background

Clinical Features

Histology and Histochemistry

Immunohistochemistry

Carriers of Duchenne and Becker Muscular Dystrophy

Therapies for Duchenne Muscular Dystrophy

11. Muscular Dystrophies and Allied Disorders II: Limb-Girdle Muscular

Dystrophies

History and Background

Histology and Histochemistry

Immunohistochemistry

12. Muscular Dystrophies and Allied Disorders III: Congenital Muscular

Dystrophies and Associated Disorders

History and Background

General Pathological Features of Congenital Muscular Dystrophies

Congenital Muscular Dystrophies Associated with Sarcolemmal Proteins

Congenital Muscular Dystrophies Associated with Abnormal Glycosylation of α -Dystroglycan

Rigid Spine with Muscular Dystrophy (RSMD1) Congenital Muscular Dystrophies Associated with Nuclear Membrane Proteins Additional Forms of Congenital Muscular Dystrophy 13. Muscular Dystrophies and Allied Disorders IV: Emery–Dreifuss Muscular Dystrophy and Similar Syndromes Clinical Features Molecular Genetics **Biochemistry** Histopathology Immunohistochemistry Electron Microscopy Other Emery–Dreifuss-Like Syndromes 14. Muscular Dystrophies and Allied Disorders V: Facioscapulohumeral, Myotonic and Oculopharyngeal Muscular Dystrophies Facioscapulohumeral Muscular Dystrophy Myotonic Dystrophies Oculopharyngeal Muscular Dystrophy 15. Congenital Myopathies and Related Disorders Introduction Myopathies with Structural Defects Core Myopathies Nemaline Myopathies Myotubular/Centronuclear Myopathies Titin-related Congenital Myopathy Sarcotubular Myopathy Surplus Protein Myopathies Cap Disease Spheroid Body Myopathy Reducing Body Myopathy Congenital Fibre Type Disproportion Congenital Myopathies with Other Ultrastructural Abnormalities Novel Rare Congenital Myopathies

Congenital Myopathies Characterized by Distal Involvement and/or Distal Arthrogryposis 16. Myofibrillar Myopathies and Other Myopathies with Rimmed Vacuoles Introduction Myofibrillar Myopathies Other Myopathies with Autophagic Vacuoles and/or Protein Aggregates 17. Metabolic Myopathies I: Glycogenoses Type II Glycogenosis (Pompe Disease, Acid Maltase Deficiency) *Type III Glycogenosis (Debrancher Enzyme Deficiency) Type IV Glycogenosis (Branching Enzyme Deficiency)* Type V Glycogenosis (McArdle Disease) *Type VII Glycogenosis (Phosphofructokinase Deficiency) Type XIV Glycogenosis (Phosphoglucomutase 1 Deficiency)* Type 0 Glycogenosis (Glycogen Synthase Deficiency) *Type XV Glycogenosis (Glycogenin-1 Deficiency)* RBCK1 (HOIL-1) Deficiency Other Glycogenoses with Neuromuscular Symptoms 18. Metabolic Myopathies II: Lipid-Related Disorders and Mitochondrial **Myopathies** Disorders of Muscle Lipid Metabolism Mitochondrial Myopathies 19. Myopathies Associated with Systemic Disorders and Ageing Endocrine Disorders Disorders of the Thyroid Disorders of the Pituitary and Adrenals Insulin-Associated Disorders Disorders of the Parathyroids, Osteomalacia and Vitamin Deficiencies Malignancy Amyloidosis Ageing and Cachexia 20. Ion Channel Disorders Syndromes with Non-Dystrophic Myotonia

Periodic Paralysis Syndromes Disorders Associated with Calcium Homeostasis Malignant Hyperthermia Myoglobinuria/Rhabdomyolysis 21. Myasthenic Syndromes Myasthenia Gravis Lambert–Eaton Syndrome Acquired Neuromyotonia Congenital Myasthenic Syndromes 22. Inflammatory Myopathies Dermatomyositis Antisynthetase Syndrome Inclusion Body Myositis Immune-Mediated Necrotizing Myopathy Granulomatous Myositis Focal Myositis Macrophage Myofasciitis 23. Toxic and Drug-Induced Myopathies Classification Necrosis and Rhabdomyolysis Inflammation Mitochondrial Myopathy Myosin Heavy Chain Loss (Critical Illness Myopathy) *Type 2 Fibre Atrophy* Vacuolar Myopathies Neuromyopathy Focal Myopathy Appendix 1. Glossary of Genetic Terms Appendix 2. Useful Websites Index