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Nota di contenuto	Cover; Advisory Editors; Title page; Copyright page; List of Contributors; Preface; List of Abbreviations; 1: Introduction to Muscle Disease: Pathology and Genetics; Introduction; Structure of the book; Conclusion; Section 1: Assessment of Muscle Disease; 2: Clinical Features of Muscle Disease; Introduction; Clinical history and examination; Conclusion; 3: General Pathology of Muscle Disease; Introduction; Selection of muscle to biopsy; Biopsy technique; Tissue preparation; Use of other tissues for diagnosis; Development of human muscle Histological and histochemical features of normal muscleHistological and histochemical defects in pathological muscle; Structural abnormalities; Absence of an enzyme; Storage of a product; Immunohistochemistry; Electron microscopy; Future perspectives; 4: Genetics of Muscle Disease; Introduction; Discovery of genes causing muscle disease; Benefits of finding the mutation causing a disease in a patient; Types of DNA mutations; Blurring of traditional clinical classifications - parallel nosologies Mutations within the same gene can cause a spectrum of phenotypes

(as classified by classic nosology)One disease-related pathology can be caused by mutations in different genes; Some genes are to date only implicated in one disease; Some muscle proteins have not yet been associated with human disease; Interesting recent developments; The way forward, addressing the grand challenges in the genetics of muscle disease; Conclusions and future perspectives; Section 2: Neurogenic Muscle Disease; 5: Neurogenic Muscle Pathology; Defining denervation; Clinical features; Pathophysiology
Pathological changesFetal and infantile denervation; Differential diagnosis; Section 3: Diseases of Neuromuscular Transmission; 6: Autoimmune Myasthenias; Introduction; Incidence; Clinical features; Investigations; Treatment and prognosis; Pathology; Genetics; Differential diagnosis; Animal models; Lambert-Eaton myasthenic syndrome; 7: Congenital Myasthenic Syndromes; Introduction; The neuromuscular junction; Clinical features of congenital myasthenic syndromes; Conclusions and future perspectives; Section 4: Sarcolemma: Muscular Dystrophies and Related Disorders
8: Dystrophin and Its Associated Glycoprotein ComplexIntroduction; Incidence; Clinical features; Pathology; Genetics; Animal models; Conclusions and future perspectives; 9: Proteins of the Extracellular Matrix; Introduction; Congenital muscular dystrophies; Laminin 2 primary deficiency; Collagen VI-related myopathies; Perlecan-related disorders; Laminin 2 and agrin-related disorders; Animal models for extracellular matrix-related disorders; Conclusions and future perspectives; 10: Plasma Membrane Proteins: Dysferlin, Caveolin, PTRF/Cavin, Integrin 7, and Integrin 9; Introduction
Dysferlin

Sommario/riassunto

This book clarifies the pathology and genetics of muscle disease for pathologists, clinicians, geneticists and researchers to aid in the diagnosis and management of patients. Organized around the 'motor unit' concept, this book presents the latest understanding of muscle disease, and how this can help identify new treatments.