

1. Record Nr.	UNINA9910455637703321
Autore	Aaron Daniel <1912->
Titolo	The Americanist [[electronic resource] /] / Daniel Aaron
Pubbl/distr/stampa	Ann Arbor, : University of Michigan Press, c2007
ISBN	1-282-44530-8 9786612445309 0-472-02466-3
Descrizione fisica	1 online resource (209 p.)
Disciplina	973.072/02
Soggetti	Historians - United States Historiography - United States Presidents - United States - History - 20th century Critics - United States American literature - History and criticism Electronic books. United States Study and teaching United States Politics and government 20th century Miscellanea
Lingua di pubblicazione	Inglese
Formato	Materiale a stampa
Livello bibliografico	Monografia
Note generali	Includes index.

2. Record Nr.	UNINA9910139054603321
Titolo	Muscle disease [[electronic resource]] : pathology and genetics // edited by Hans H. Goebel, Caroline A. Sewry, Roy O. Weller
Pubbl/distr/stampa	Chichester, West Sussex ; ; Hoboken, NJ, : Wiley-Blackwell, 2013
ISBN	1-118-63548-5 1-118-63546-9 1-118-63549-3
Edizione	[2nd ed.]
Descrizione fisica	1 online resource (986 p.)
Altri autori (Persone)	GoebelH. -H SewryCaroline A WellerRoy O
Disciplina	616.7/4
Soggetti	Muscles - Diseases Genetics
Lingua di pubblicazione	Inglese
Formato	Materiale a stampa
Livello bibliografico	Monografia
Note generali	Description based upon print version of record.
Nota di bibliografia	Includes bibliographical references and index.
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 muscle Histological 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 changes Fetal 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 Complex Introduction; 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

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#### 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.

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