

Protein Dysfunction in Human Genetic Disease

Edited by D.M. Swallow and Y.H. Edwards



Protein Dysfunction In Human Genetic Disease

Marcel A. Müller



Protein Dysfunction In Human Genetic Disease:

Protein Dysfunction in Human Genetic Disease D M Swallow,Y H Edwards,2024-11-15 Recent years have seen rapid advances in our understanding of genetic diseases in terms of their molecular origins The book focuses on DNA mutations which give rise to abnormalities of protein structure and function in genetic disease *Protein Dysfunction in Human Genetic Disease* D. M. Swallow,Dallas M. Swallow,Yvonne H. Edwards,1997 Recent years have seen rapid advances in our understanding of genetic diseases in terms of their molecular origins In almost all cases where disease genes have been cloned the mutations in the DNA sequence which are responsible for the clinical phenoty **Human Blood Cells:**

Consequences Of Genetic Polymorphisms And Variations May-jean King,2000-07-04 This important book uses selected molecules expressed on erythrocytes lymphocytes platelets and granulocytes to illustrate how genetic polymorphisms and variations in these molecules can affect their structure and function in mature human blood cells The examples described tend to have a clinical association Human blood group antigens and HLA antigens are classic examples of genetic polymorphism and they are important in blood transfusion and organ transplantation respectively In common with the blood group antigens the polymorphic and variant antigens on platelets and granulocytes can be targets for antibodies in feto maternal antigen incompatibility and transfusion reactions Certain inherited haemolytic anaemias can be attributed to some of the polymorphic and variant forms of erythrocyte anion transport protein spectrin and glucose 6 phosphate dehydrogenase which exhibit abnormal structural or functional properties Similarly the study of cytokine gene polymorphism can provide a further understanding of the immune inflammatory diseases and allogeneic transplantation a **Disorders of Voluntary Muscle** George Karpati,David Hilton-Jones,Robert C. Griggs,2001-07-12 Rewritten and redesigned this remains the one essential text on the diseases of skeletal muscle *Analysis of Triplet Repeat Disorders* Michael Hayden,Dr David Rubinsztein,2020-07-26 Analysis of Triplet Repeat Disorders is aimed at clinicians and scientists who work with these diseases or who have an interest in the field Using the clinical picture of these diseases as a starting point the book reviews and integrates the current understanding of their molecular pathologies the genotype phenotype relationships the mutational processes of trinucleotide repeats and the laboratory and clinical issues relating to genetic testing for these disorders

Motor Disorders David S. Younger,2021-12-22 The fourth edition of Motor Disorders reinforces the reputation gained by earlier editions The book gives a detailed and practical account of motor disorders that will appeal to specialists as well as to general physicians and trainees while providing a convenient but thorough summary of advances in the field The book brings together the entire spectrum of motor disorders and thereby aids in the evaluation diagnosis and distinction of these various disorders Through the perspectives of many subspecialties this book provides accounts of all disorders with motor manifestations providing a truly comprehensive but concise account of the subject matter *Analysis of Multifactorial Diseases* D T Bishop,Pak Sham,2024-12-06 Analysis of Multifactorial Diseases examines the evidence of genetic factors that

increase susceptibility to a range of complex diseases and describes the approaches taken to identify these genes It reviews the successes and failures and examines whether there are any general conclusions to be drawn from this current range of approaches

Motor Disorders David S. Younger, MD, 2014-10-01 This preeminent book in the field of Neurology has been extensively updated and expanded by 70 leading authorities providing a single encyclopedic summary the scientific advances and new clinical practices that can be immediately considered for your patients It brings together nearly the entire spectrum of motor disorders and Neurology into one convenient resource to aid in a comprehensive evaluation diagnosis distinction and treatment of various disorders Additionally by using the perspectives of different subspecialties the book also provides a comprehensive yet concise account of any disorder with motor manifestations This new edition now includes the following content and features Edited by David S Younger MD a highly respected practitioner and researcher with scholarly chapters by leaders in the field and more international authors than other texts Up to date articles on highly changing subjects related to Neurogenetics Chapters on Neurorehabilitation coverage not usually found in other neurological textbooks Advances in Alzheimer Disease Parkinson Disease Amyotrophic Lateral Sclerosis Both adult and pediatric topics in one book High quality figures of neuroimaging and neuropathology

Diagnostics and Gene Therapy for Human Genetic Disorders K.V. Chaitanya, 2022-12-30 Diagnostics and Gene Therapy for Human Genetic Disorders provides an integrative and comprehensive source of information blending classical human genetics with the human genome It provides a multidisciplinary overview of Mendelian inheritance and multifactorial inheritance genetic variations polymorphisms chromosomal multifactorial and mitochondrial disorders PCR electrophoresis cytogenetics prenatal and HPLC based techniques applied for diagnosing genetic disorders are discussed with applications Symptoms etiology diagnosis treatment of 14 major and 5 minor genetic disorders are discussed in detail Methods employed for the preparation of kits for the diagnosis of diseases are provided The role of gene therapy in the amelioration of genetic disorders and the methodology employed are discussed The success of gene therapy in controlling various disorders such as immune system disorders neurodegenerative disorders cardiovascular disorders eye diseases and cancer has been described along with type studies Features A blend of classical human genetics with molecular and genome based applications Techniques applied for the diagnosis of genetic disorders Diagnostics of 19 genetic disorders including symptoms etiology diagnosis and treatment Role of gene therapy in the amelioration of disorders Type studies describing the role of diagnostics in conserving the human health This book attempts to connect all the information about classical and modern human genetics genetic disorders and gene therapy to all types of diseases in one place This work provides a comprehensive source of information that can serve as a reference book for scientific investigations and as a textbook for the graduate students

Molecular Genetics of Cancer John Cowell, 2001-06-15 Molecular Genetics of Cancer Second Edition provides an authoritative and up to date review of the key genes known to be critical in the development or progression of cancer Throughout the book scientific advances and

their clinical relevance are covered in detail particularly in the light of findings concerning the inheritance of genes predisposing to tumorigenesis The book is therefore a valuable source of reference for clinicians and genetic counsellors as well as researchers

Metabolic Aspects of Chronic Liver Disease Ami Schattner,Hilla Knobler,2008 This book discusses in detail new aspects of the metabolic basis of important chronic liver diseases Entities such as non alcoholic fatty liver disease fatty liver and Non Alcoholic SteatoHepatitis NASH diabetes in chronic hepatitis C hemochromatosis Wilson's disease Gaucher disease porphyria as well as liver cirrhosis and its metabolic consequences will be discussed in detail These clinical conditions are highly prevalent and affect millions of patients in the USA and world wide For example non alcoholic fatty liver disease is the most common cause of elevated liver enzymes in the general population This field has been practically transformed over the last few years with many new insights gained regarding both pathogenesis and effective novel treatments

Rosenberg's Molecular and Genetic Basis of Neurological and Psychiatric Disease, Seventh Edition Roger N. Rosenberg,Juan M. Pascual,2024-10-07 Rosenberg's Molecular and Genetic Basis of Neurologic and Psychiatric Disease Seventh Edition provides a comprehensive introduction and reference to the foundations and practical aspects relevant to the majority of neurologic and psychiatric disease This updated volume focuses on degenerative disorders movement disorders neuro oncology neurocutaneous disorders epilepsy white matter diseases neuropathies and neuronopathies muscle and neuromuscular junction disorders stroke psychiatric disease and a neurologic gene map This volume includes new chapters on Von Hippel Lindau disease antisocial and violent behavior and Autism A favorite of over four generations of students clinicians and scholars this new edition retains and expands on the informative concise and critical tone of the previous edition This is an essential reference for general medical practitioners neurologists psychiatrists geneticists related professionals and for the neuroscience and neurology research community at large Both volumes combined provide a comprehensive coverage on the neurogenetic foundation of neurological and psychiatric disease This volume presents detailed coverage of disease mechanisms and management for degenerative disorders movement disorders and muscle and neuromuscular junction disorders Includes new chapters on the pharmacogenomics of Alzheimer's Disease and Epilepsy and the most recent updates in molecular genetics focusing on pain genetics and muscular dystrophy

Genotype to Phenotype J. J. Goodship,S. Malcolm,2003-12-16 This new edition builds on the success of the first by reviewing the increased understanding of the mechanisms of gene action in humans focusing particularly on those derived from the study of genetic diseases It deals mainly with the fundamental aspects of gene arrangement and expression rather than mutation As well as updating and revising material from the first edition it covers methods of exploring gene function and contains a range of chapters on specific systems which raise issues of special interest such as imprinting or homologous genes within clusters

Transcription Factors Joseph Locker,2003-12-16 Transcription factors are important in regulating gene expression and their analysis is of paramount interest to molecular biologists studying this area This book

looks at the basic machinery of the cell involved in transcription in eukaryotes and factors that control transcription in eukaryotic cells It examines the regulatory systems that modulate gene expression in all cells as well as the more specialized systems that regulate localized gene expression throughout the mammalian organism Transcription Factors updates classical knowledge with recent advances to provide a full and comprehensive coverage of the field for postgraduates and researchers in molecular biology involved in the study of gene regulation Venous Thrombosis D.N. Cooper,M. Krawczak,2024-12-06 Thrombotic disease is a major cause of death and disablement in Western society The most common causes is an inherited defect in one of the many genes encoding a protein which is involved in clotting It s regulation or the process of clot dissolution Fibrinolysis Venous Thrombosis from genes to clinical medicine presents our current knowledge of potentially prothrombotic protein at modalities together with the clear and concise views of the roles these proteins play in haemostasis and the use of molecular techniques in the analysis and diagnosis of inherited defects Each chapter describes a specific protein deficiency with information on protein structure function and biochemistry gene structure and expression as well as epidemiological and molecular genetic aspects of the deficiency state This multifaceted approach aims to examine the possible causes of venous thrombosis to explain the underlying molecular defects and to explore the genotype phenotype relationship in this complex multi gene disorder The potential roles of both aesthetic interactions and environmental risk factors are also discussed This book will be of value to haematologists genetic counsellors and clinical geneticists **From Genotype to Phenotype** Sue Malcolm,2001-11-19 This volume of the Human Molecular Genetics series covers such genotype phenotype correlations as clinical and environmental aspects gene structure expression and mutation Also discussed are models of certain diseases and future prospects for treatment and prevention This book provides the reader with a basic overview of the physical expression of genetic disease before discussing in detail the most recent research and therapeutic developments **Medical Genetics** Ian D Young,2010-09-02 Medical Genetics provides medical and biomedical students with an understanding of the basic principles of human genetics as they relate to clinical practice showing how our genome lies at the heart of our health and well being Molecular Endocrinology Gill Rumsby,Dr Sheelagh Farrow,2020-07-26 This volume in the Human Molecular Genetics series is an invaluable text for endocrinologists wishing to update their knowledge It also provides an excellent grounding in the basic genetics of molecular endocrinology and relevant analytical techniques Gene Therapy David Cooper,Prof Nick Lemoine,2020-09-10 Gene Therapy describes the delivery systems now available to target a given tissue with specific gene or oligonucleotide sequences and explores the utility of animal modules as test systems In the context of selected disease states it summarises in vitro and in vivo studies and clinical trials performed to date **Giant Molecules** ,2011-05-26 Our lives are dominated by giant molecules which have remarkable properties some of which are only just being discovered and exploited by science though many have long been exploited far more effectively by Nature Giant molecules dominate our lives from the proteins and DNA within us to the man

made fibres of our clothes and the many plastics that we use every day And they are set to have an enormous impact on the future as scientists and engineers learn from nature biomimetics and utilize the full potential of tiny carbon nanotubes The possibilities may seem like science fiction a space station tethered to Earth by cables of giant molecules tiny molecular vehicles carrying and dispensing drugs in our bodies smart materials that adjust automatically to optimize our comfort minute computers utilizing the information storage capacity of DNA but they are the subjects of cutting edge research Walter Gratzer gives a fascinating account of the discovery and variety of giant molecules how they come to have their remarkable properties and how these are used by Nature and increasingly by us pausing now and again to tell of some of the remarkable characters involved in their discovery and development

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