

OXFORD MONOGRAPHS ON MEDICAL GENETICS NO. 25

The Molecular Genetics of Haemostasis and its Inherited Disorders

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OXFORD MEDICAL PUBLICATIONS

Molecular Genetics Of Haemostasis And Its Inherited Disorders

**Philip R. Beales, I. Sadaf
Farooqi, Stephen O'Rahilly**



Molecular Genetics Of Haemostasis And Its Inherited Disorders:

The Molecular Genetics of Haemostasis and Its Inherited Disorders Edward G. D. Tuddenham, David Neil Cooper, 1994 This work uniquely collates and summarizes our current knowledge of the structure and function of the genes and proteins involved in blood coagulation and its regulation For the first time the vast rapidly growing and scattered literature has been comprehensively analyzed The book presents critical reviews covering the entire field of the hereditary disorders of haemostasis and thrombosis An introductory chapter outlines the most recent theories of blood coagulation and the state of the art of the analysis of human gene mutation followed by 32 chapters each devoted to a single gene or small group of genes and their corresponding disorders As well as detailing each gene's structure chromosomal location and regulation the authors provide concise but comprehensive summaries of each protein product and of the deficiency disorders associated therewith whether leading to hemophilia or thrombophilia Appendices contain essential practical information on genotype and phenotype analysis and on the epidemiology of the disorders of blood coagulation This is an invaluable text for clinicians and laboratory scientists who want to unravel the molecular basis of inherited disease presented with a patient with either a bleeding or thrombotic disorder

A Short History of Medical Genetics Peter S. Harper, 2008 This book traces the development of genetics in medicine from the first descriptions of inherited diseases more than 300 years ago to the new applications resulting from mapping and sequencing the human genome It follows both the scientific and the medical advances focusing especially on those of the past 50 years which have seen the field of medical genetics emerge as one of the foremost and most rapidly changing medical specialties now influencing the whole of medicine It also examines the ethical challenges faced by those working in the field and describes some of the past disasters that have resulted from these being ignored notably the abuses of eugenics and the catastrophic destruction of genetics in Soviet Russia This is the first book of its kind it is clearly and simply written and will be valuable to all those who have an interest or concern in the development of medical genetics as well as those actually working in the field Historians and social scientists will likewise find this book an important foundation for future detailed studies which are urgently needed **BOOK JACKET** *Genetic Diseases of the Eye* Elias I. Traboulsi, 2012-01-12 This book takes a clinical approach to the patient with a genetic disease that affects the eye The chapters on particular types of diseases follow the same organizational format covering history pathogenesis and etiology epidemiology classification clinical manifestations and diagnosis and treatment The recent progress achieved in the molecular genetics of eye disease is fully reflected throughout the book It is written by leading experts in the field and provides clinical molecular genetic and management information on common and rare diseases The chapters are heavily illustrated and provide a good Atlas for the practicing ophthalmologist or geneticist

Genomic Medicine Dhavendra Kumar, Charis Eng, 2015 The first edition of Genomics and Clinical Medicine provided an overview of genomics based advances in disease susceptibility diagnosis and prediction of treatment outcomes in various areas of

medicine Since its publication the science of genomics has made tremendous progress and exciting new developments in biotechnology and bioinformatics have created possibilities that were inconceivable only a few years ago This completely revised second edition of Genomic Medicine reflects the rapidly changing face of applied and translational genomics in the medical and health context and provides a comprehensive coverage of principles of genetics and genomics relevant to the practice of medicine

Huntington's Disease Gillian Bates, Sarah Tabrizi, Lesley Jones, 2014-03-20 This fourth edition of Huntington's Disease presents a comprehensive summary of the current knowledge of this disease including the major scientific and clinical advances that have occurred since publication of the third edition in 2002 Completely updated and expanded chapters in this volume are organized in five sections Clinical aspects of Huntington's disease including updated chapters on historical perspectives neurological neuropsychiatric and neuropsychological aspects and new chapters on juvenile Huntington's and the premanifest and early stages The genetics of Huntington's disease including new information on its epidemiology discussions of new testing guidelines Neurobiology including recent insights into correlations between pathology and symptoms and a new chapter on neuronal circuitry The molecular biology of Huntington's disease including new chapters on the normal function of huntingtin the molecular pathogenesis of Huntington's disease and the peripheral pathology of the disorder and an extensively updated chapter on its structural biology An updated description of the comprehensive care for Huntington's disease featuring a new chapter on preclinical therapeutics and a completely rewritten chapter on the state of the art of experimental therapeutics and clinical trials

Hemostasis and Thrombosis Protocols David J. Perry, K. John Pasi, 2008-02-02 Laboratory studies in hemostasis have traditionally focused on abnormalities of platelet function or the quantitative and qualitative disorders that affect the proteins involved in blood coagulation However over the last 10 years there has been an explosion in our understanding of the molecular bases that underlie many of the inherited and acquired disorders of hemostasis Many of these disorders are now routinely diagnosed and assessed by methods that involve genotypic analysis Indeed in the late 1990s the distinction between molecular methods for research and for routine diagnosis is becoming increasingly blurred The techniques and approaches that are used in hemostasis are manifold and published in isolation in a variety of publications The aim therefore of this volume Hemostasis and Thrombosis Protocols is to pull together into a single volume the variety of techniques that are frequently used in the field of hemostasis We have targeted this volume at laboratories who wish to move into the field of molecular hemostasis or who may already have some experience in this area but wish to develop new areas of research and diagnosis The chapters are wide ranging and hopefully provide a broad overview of the differing applications in which these standard techniques can be used Though the articles may appear relatively specific the techniques contained within them are applicable to the study of many different disorders and we hope that they provide a series of ideas and concepts well suited to problem solving

Genetic Disorders of Human Sexual Development Leonard Pinsky, Robert P. Erickson, R. Neil Schimke, 1999 There have been many advances

in understanding mammalian sex determination and differentiation during the last decade Using these advances to elucidate clinical conditions of abnormal sexual development the authors of this book bring together expertise in molecular endocrinology molecular genetics and dysmorphology This is not a conventional textbook It is seldom dogmatic frequently presents alternatives highlights speculation raises questions and attempts to provide answers This book will be welcomed by medical geneticists genetic counselors endocrinologists gynecologists urologists and students who need fully referenced information about the genetic aspects of human sexual maldevelopment in order to better manage their patients and their patients families

Genetics of Obesity Syndromes Philip R. Beales,I. Sadaf Farooqi,Stephen O'Rahilly,2008-08-29

Obesity is one of the most important contributing factors to disease throughout the world and is an area of great current interest among researchers and clinicians The genetics of common obesity is complex and an important thread through this labyrinth is the study of genetic syndromes in which obesity is a major component By examining the genetic mechanisms of obesity in these syndromes the authors will shed new light on the genetics of common obesity This is the first book on this important and exciting new area and addresses both the molecular and clinical features of the obesity syndromes providing hard core information for researchers and practical guidelines for clinicians caring for obese patients The book is divided into three sections the first covers approaches for assessing and investigating the obese individual the second describes nondysmorphic monogenic forms of obesity and the third documents key multisystem obesity syndromes with various genetic etiologies It is as much a reference book as it is a manual and will appeal to clinical geneticists obesity researchers endocrinologists nutritionists and medical biologists

Hereditary Diseases and Blood Transfusion C.Th. Smit Sibinga,P.C. Das,E. Briët,2012-12-06 Proceedings of the Nineteenth Annual International Symposium on Blood Transfusion Groningen 1994 organized by the Red Cross Blood Bank Groningen Drenthe

Landmarks in Medical Genetics Peter S. Harper,2004-03-11 Advances in genetics over the past 50 years have been dramatically changed the understanding and management of inherited disorders and are beginning to have a major impact on the practice of medicine overall The rapidity of these advances means that clinicians and scientists in the field are often unfamiliar with the key research that has led to many developments that now are accepted and familiar Few have time to search or the original papers which are scattered and often difficult to obtain This collection has been edited mainly for medical geneticists and genetics researchers who wish to learn more about how their field originated and developed Brief clearly written commentaries on each paper and section place the work in its current context and serve to unify the different parts of the book They also help make it a readable and authoritative source of information The papers chosen fall into several groups First are classic descriptions of important genetic disorders often from the pre mendelian era The following sections deal with the definition of human mendelian inheritance the origins of human cytogenetics the early development of the human gene map and the transition from biochemical genetics to human molecular genetics the relatively recent studies that have shown how mendelian principles

are increasingly modifiable and finally advances in the treatment and management of genetic disorders which are placed in their social context

Human Malformations and Related Anomalies Roger E. Stevenson, 2016 The central theme of this text is to provide information on individual anomalies et to connect these anomalies to the malformation syndromes et associated problems primarily through the use of differential diagnostic tables

Syndromes of the Head and Neck Robert J. Gorlin, M. Michael Cohen Jr., D.M.D., Ph.D., Raoul C.M. Hennekam M.D., Ph.D., 2001-09-27 This classic text one of the true anchors of our clinical genetics publishing program covers over 700 different genetic syndromes involving the head and neck and it has established itself as the definitive comprehensive work on the subject The discussion covers the phenotype spectrum epidemiology mode of inheritance pathogenesis and clinical profile of each condition all of which is accompanied by a wealth of illustrations The authors are recognized leaders in the field and their vast knowledge and strong clinical judgment will help readers make sense of this complex and burgeoning field Dr Gorlin retires as editor in this edition and co editor Raoul Hennekam takes over Dr Hennekam is regarded as one of the top dysmorphologists and indeed one of the top clinical geneticists in the world Judith Allanson is new to the book but is a veteran OUP author and a widely respected geneticist and Ian Krantz at Penn is a rising star in the field Dr Gorlin's name has always been closely associated with the book and it has now become part of the title As in all fields of genetics there has been an explosion in the genetics of dysmorphology syndromes and the author has undertaken a complete updating of all chapters in light of the discoveries of the Human Genome Project and other ongoing advances with some chapters requiring complete rewriting Additional material has been added both in terms of new syndromes and in updating information on existing syndromes The book will appeal to clinical geneticists pediatricians neurologists head and neck surgeons otolaryngologists and dentists The 4th edition which published in 2001 has sold 2 600 copies

Gardner and Sutherland's Chromosome Abnormalities and Genetic Counseling R. J. McKinlay Gardner, David J. Amor, 2018 Even as classic cytogenetics has given way to molecular karyotyping and as new deletion and duplication syndromes are identified almost every day the fundamental role of the genetics clinic remains mostly unchanged Genetic counselors and medical geneticists explain the unexplainable helping families understand why abnormalities occur and whether they're likely to occur again Chromosome Abnormalities and Genetic Counseling is the genetics professional's definitive guide to navigating both chromosome disorders and the clinical questions of the families they impact Combining a primer on these disorders with the most current approach to their best clinical approaches this classic text is more than just a reference it is a guide to how to think about these disorders even as our technical understanding of them continues to evolve Completely updated and still infused with the warmth and voice that have made it essential reading for professionals across medical genetics this edition of Chromosome Abnormalities and Genetic Counseling represents a leap forward in clinical understanding and communication It is as ever essential reading for the field

Inherited Metabolic Disease in Adults Carla E. M. Hollak, Robin Lachmann, 2016 As clinical management of inherited

metabolic diseases IMDs has improved more patients affected by these conditions are surviving into adulthood This trend coupled with the widespread recognition that IMDs can present differently and for the first time during adulthood makes the need for a working knowledge of these diseases more important than ever Inherited Metabolic Disease in Adults offers an authoritative clinical guide to the adult manifestations of these challenging and myriad conditions These include both the classic pediatric onset conditions and a number of new diseases that can manifest at any age It is the first book to give a clear and concise overview of how this group of conditions affects adult patients a that topic will become a growing imperative for physicians across primary and specialized care

The Genetic Basis of Common Diseases Richard A.

King,Jerome I. Rotter,Arno G. Motulsky,2002-10-17 Since the first edition of this highly acclaimed text was published in 1992 much new knowledge has been gained about the role of genetic factors in common adult diseases and we now have a better understanding of the molecular processes involved in genetic susceptibility and diseases mechanisms The second edition fully incorporates these advances The entire book has been updated and twelve new chapters have been added Most of these chapters deal with diseases such as gallstones osteoporosis osteoarthritis skin cancer other common skin diseases prostate cancer and migraine headaches that are seen by all physicians Others address the genetic and molecular basis of spondylarthropathies lupus hemochromatosis IgA deficiency mental retardation hearing loss and the role of mitochondrial variation in adult diseases Chapters on the evolution of human genetic disease and on animal models add important background on the complexities of these diseases Unique clinical applications of genetics to common diseases are covered in the additional new chapters on genetic counseling pharmacogenetics and the genetic consequences of modern therapeutics

Genetic Disorders Among Arab Populations Ahmad S. Teebi,Talaat I. Farag,1997 Few regions of the world have a more varied physiography or a richer ethnic religious social and cultural mix than the Arab world As a consequence Arabs are genetically diverse despite their linguistic and religious cohesion High but variable rates of inbreeding prevail in all Arab countries with numerous examples of isolates among them These include some Bedouin groups Nubians Druze Jews and others Arab populations have high frequencies of autosomal recessive disorders homozygosity of autosomal dominant and X linked traits and a wealth of new syndromes and variants the majority of which are autosomal recessive Genetic disorders that cause major health problems include hemoglobinopathies neurogenetic disorders inherited metabolic diseases and inborn error of morphogenesis Because of their characteristically high prevalence some of these disorders are considered markers for Arab populations This book presents the unique profile of genetic disorders and variants in Arabs In addition it describes their unusual demographic pattern including fertility rates and other population dimensions family structure magnitude and effects of consanguineous marriages and indicators of health and disease Genetic counseling and the distinctive cultural and religious attitudes towards various genetic issues are also presented Disorders that have increased tribal occurrences or are limited to large kindreds as well as small geographic or religious isolates are highlighted to

facilitate their recognition study and management

Transcription Factors and Human Disease Gregg L. Semenza, 1998

Several general principles have emerged from the study of human transcription factors First germline mutations in genes encoding transcription factors result in malformation syndromes in which the development of multiple body structures is affected Second somatic mutations involving many of the same genes contribute to tumorigenesis Third transcriptional regulatory mechanisms demonstrate remarkable evolutionary conservation Fourth prenatal development and postnatal physiology are unified by the demonstration that a single transcription factor can control the proliferation of progenitor cells during development and the expression within the differentiated cells of gene products that participate in specific physiologic responses Transcription Factors and Human Disease presents the basic science of transcriptional regulation and then describes inherited human diseases attributable to mutations in DNA sequences encoding transcription factors or their cognate binding sites The involvement of transcription factors in somatic cell genetic diseases cancer and epigenetic disease teratogenesis is briefly discussed The effect of specific mutations on transcription factor activity and the relationship between transcriptional dysregulation dominant or recessive inheritance patterns and disease pathogenesis are also explored This book thus provides a direct connection between molecular defects in transcriptional regulation and human

pathophysiology **Hereditary Hearing Loss and Its Syndromes** Robert J. Gorlin, Helga V. Toriello, 1995 This definitive reference work replaces Konigsmark and Gorlin's Genetic and Metabolic Deafness 1976 Whereas the earlier volume covered 151 genetic conditions related to hearing loss this work covers 435 As before the authors first discuss isolated hereditary hearing loss and then present hearing loss syndromes such as those involving the nervous system eye external ear and musculoskeletal system The discussions are authoritative practical and well illustrated and those of the most important syndromes are very detailed Introductory chapters deal with the history of the field clinical approach embryology of the ear tooth anomalies and hearing loss and related endocrine and metabolic disorders Throughout the authors pay careful attention to nomenclature and classification This will be an invaluable resource for all professionals concerned with genetic hearing loss including medical geneticists audiologists and otolaryngologists

Hereditary Hearing Loss and Its Syndromes Helga V. Toriello, Shelley D. Smith, 2013-06-20 This is the third edition of the foremost medical reference on hereditary hearing loss Chapters on epidemiology embryology non syndromic hearing loss and syndromic forms of hearing loss have all been updated with particular attention to the vast amount of new information on molecular mechanisms and chapters on clinical and molecular diagnosis and on genetic susceptibility to ototoxic factors have been added As in previous editions the syndromes are grouped by system visual metabolic cardiologic neurologic musculoskeletal endocrine etc with each chapter written by a recognized expert in the field Written for practicing clinicians this volume is an excellent reference for physicians audiologists and other professionals working with individuals with hearing loss and their families and can also serve as a text for clinical training programs and for researchers in the hearing sciences

Pharmacogenetics Wendell

Weber, 2008-04-02 Drugs are administered to patients with the intention of achieving an expected therapeutic response Yet drug therapy remains a medical art because of the wide variation in individual responses to standard drug doses Pharmacogenetics emerged as a field aimed at identifying heritable factors that are predictive of individual variations in drug efficacy and toxicity from the dedicated work of a small group of investigators who in the 1950s began to enrich the discipline of pharmacology by integrating it with human genetics Pharmacogenetics provides the experimental framework to understand variation in human reactions to drugs and other exogenous substances as a function of inborn human genetic variability Recently pharmacogenetics has experienced a period of rapid growth and redefinition The human genome initiative has provided a wealth of primary genetic information and functional genomic data to fuel the understanding of genetic polymorphism and its functional consequences on a genomic scale Today the field of pharmacogenetics is a well integrated worldwide network engaging a vast community of academic institutional and industrial scientists The second edition of this highly regarded book provides a systematic and comprehensive account of the foundations fundamentals and futures of the field of pharmacogenetics and serves as introduction to all facets of pharmacogenetics for physicians pharmacists epidemiologists and other professionals in the life sciences Dr Wendell Weber has completely revised and updated each chapter in this new edition to reflect the enormous changes in this field

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