



EDUCATIONAL CORNER OF THE ISSUE

Basic concepts of medical genetics

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1. Gene function

The genome of each organism, composed of all genes specific to the organism, is responsible for determining, configuring, and regulating all structural and functional aspects of that organism. These activities are actually mediated by the unique proteome of each organism, i.e. the sum of proteins produced in that particular organism by its own genome. The genomes of different organisms are composed of variable numbers of genes, up to tens of thousands as is the case in human, and can produce variable numbers, up to tens of thousands or even millions, of different proteins required for development and maintenance of all life aspects of the organism. So, the proteome of each organism is determined by its genome. Each of the genes of the genome is responsible for controlling a particular biological function through its product, may be a particular protein or a non-mRNA molecule responsible for mediating that particular biological function. Accordingly, the main function of the gene is the production of a unique product responsible for a specific biological function. Each gene carries in its unique base sequence the genetic information necessary

for production of its gene product or its unique protein synthesized in the cell cytoplasm or cytosol [1].

The central dogma of molecular biology, or the frame of mediation of life processes at the gene-protein level, as illustrated in (Fig. 1), entails that genes regulate synthesis of proteins which mediate all life activities of living organisms. Actually, this is the major function of most genes. Genes which directly regulate protein synthesis are called structural genes. The functions of these structural genes are conducted under strict supervision of regulatory genes which regulate switching on, or activation, and switching off, or suppression, of structural genes so that the proper amount of proteins necessary for mediating physiological processes is synthesized at proper times. Proper control of these temporal aspects of gene function is of utmost importance for fine control, regulation and cooperation of the very large number of metabolic networks of the cell. Still, a third group of genes, master genes or housekeeping genes, function in a continuous manner to ensure maintenance and permanence of life-dependant processes. Examples of these processes include cell growth, cell division, ATP production and apoptosis. Continuous monitoring of these life-dependant activities is mandatory to keep life activities in living organisms [1].

Structural genes regulate protein synthesis via the genetic information, or the genetic code, which is the information contained within the specific arrangement of the nucleotides, or bases, of the gene that determine the gene function(s). It is designed to function in a specific way so that each three nucleotides in sequence along the gene, known as the codon, which is the functional unit of the gene, define a single specific amino acid in the protein synthesized under regulation of the gene. There are 64 known codons: 61 functional codons which define specific amino acids in the proteins, and three termination or stop codons that do not define any amino acids in the protein (Fig. 2). Rather, they determine the end point of the synthesized protein.

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Basic Concepts In Medical Genetics

**Marshall Horwitz, Mary Beth
Dinulos, Kris Carroll**



Basic Concepts In Medical Genetics:

Basic Concepts in Medical Genetics Marshall Horwitz, Mary Beth Dinulos, Kris Carroll, 2000 Medical Genetics E-Book
Lynn B. Jorde, John C. Carey, Michael J. Bamshad, 2019-09-27 Up to date and extensively revised to reflect recent advances in the genetics of common diseases as well as current progress in gene therapy Medical Genetics 6th Edition delivers easy to read highly visual coverage of this rapidly changing field This accessible practical text integrates key concepts with clinical practice highlighted by numerous illustrations tables concept summaries and more all designed to enhance effective learning and retention of complex material Discusses current topics including polygenic risk scores and their potential applications for diabetes cancer and heart disease and the latest sequencing technologies and their clinical application in genetic testing and diagnosis Offers a completely updated discussion of genetic testing modalities and applications Includes convenient concept summaries more than 230 photographs illustrations and tables as well as patient family vignettes that present valuable perspectives on disease and treatment Features Clinical Commentary boxes that demonstrate how the hard science of genetics has real applications to everyday patient problems preparing you for problem based integrated courses Illustrates key concepts with disease examples to demonstrate relevance to medicine Provides study questions for self assessment as well as 200 additional USMLE style questions online Enhanced eBook version included with purchase Your enhanced eBook allows you to access all of the text figures and references from the book on a variety of devices *Introduction To Basic Concepts Of Medical Genetics* Mohammad Salem, 2012 Medical genetics a branch of human genetics spans a wide spectrum of sub branches including basic clinical diagnostic therapeutic prophylactic and applied genetics This booklet I hope it be the first in a series is confined to two disciplines of basic genetics viz molecular genetics and pathogenetics that are concerned with studying the structure and function of the genetic material in health and disease states It aims at offering the basic concepts of these two subjects to clinicians and research workers in the field For clinicians beginning their specialization as medical geneticists I tried my best to offer as simplified as possible definitions and illustrations of the main points of both subjects concerning the structure and function of genes mutagens and disease causing mutations as well as the anti mutation mechanisms of the human genome For research workers I pointed out some of the most enigmatic aspects of the field that still await more research to disclose in order to go a step further in our way to understand what we think we know about the role of our genetic constitution in shaping our life Medical Genetics Lynn B. Jorde, PhD, John C. Carey, MD, MPH, Michael J. Bamshad, MD, 2015-09-04 Popular for its highly visual clinical approach Medical Genetics delivers an accessible yet thorough understanding of this active and fast changing field Key updates in this new edition cover the latest developments which are integrated with clinical practice to emphasize the central principles and how they apply to practice Photographs illustrations and tables along with boxes containing patient family vignettes demonstrate clinical relevance and enhance visual impact of the material for easier and more effective learning and retention Includes access to the complete text and

images online at studentconsult.com along with 200 additional USMLE style questions for self assessment Mini summaries study questions suggested reading and a detailed glossary supplement and reinforce what you learn from the text More than 230 photographs illustrations and tables along with patient family vignettes clarify difficult concepts and demonstrate clinical significance Clinical Commentary Boxes help demonstrate how the hard science of genetics has real applications to everyday patient problems and prepare you for problem based integrated courses The latest knowledge and research on gene identification cancer genetics gene testing and gene therapy common disorders ethical and social issues and much more so you can keep up with current developments in genetics Student Consult eBook version included with purchase This enhanced eBook experience allows access to 200 additional USMLE questions as well as new materials outlined above designed to produce a more rounded learning experience

Handbook of Neurodevelopmental and Genetic Disorders in Children, 2/e Sam Goldstein, Cecil R. Reynolds, 2010-10-19 Recognized as the definitive reference in the field this book addresses a broad range of biologically based disorders that affect children's learning and development Leading authorities review the genetics of each disorder its course and outcome associated developmental cognitive and psychosocial challenges and what clinicians and educators need to know about effective approaches to assessment and intervention Coverage encompasses more frequently diagnosed learning and behavior problems with a genetic component as well as numerous lower incidence neurodevelopmental disabilities Illustrations include 12 color plates

Medical Genetics for Healthcare Professionals Pasquale De Marco, Medical genetics is a rapidly evolving field that is having a profound impact on healthcare This book provides a comprehensive overview of medical genetics for healthcare professionals covering the basics of molecular and clinical genetics as well as the latest advances in the field Written in a clear and engaging style this book is accessible to readers with a variety of backgrounds from those with no prior knowledge of genetics to those who are already familiar with the field The book is divided into 10 chapters each of which focuses on a different aspect of medical genetics The first few chapters introduce the basic concepts of genetics including DNA structure and function gene expression and genetic variation The book then discusses the role of genetics in health and disease and how genetic testing can be used to diagnose and treat genetic disorders The book also explores the ethical and social implications of medical genetics such as genetic discrimination and the use of genetic information in law enforcement The latter chapters of the book focus on specific areas of medical genetics such as cancer genetics cardiovascular genetics and neurogenetics The book also discusses the role of medical genetics in public health including genetic screening and prevention programs This book is an essential resource for healthcare professionals who want to learn more about medical genetics and its applications in clinical practice It is also a valuable resource for students and researchers in the field of genetics If you like this book write a review

Medical Genetics: First South Asia Edition - E-Book Lynn B. Jorde, John C. Carey, Michael J. Bamshad, 2017-01-20 Popular for its highly visual clinical approach Medical Genetics First South Asia Edition delivers an accessible yet thorough

understanding of this active and fast changing field Key updates in this new edition cover the latest developments which are integrated with clinical practice to emphasize the central principles and how they apply to practice Photographs illustrations and tables along with boxes containing patient family vignettes demonstrate clinical relevance and enhance visual impact of the material for easier and more effective learning and retention Mini summaries study questions suggested reading and a detailed glossary supplement and reinforce what you learn from the text More than 230 photographs illustrations and tables along with patient family vignettes clarify difficult concepts and demonstrate clinical significance Clinical Commentary Boxes help demonstrate how the hard science of genetics has real applications to everyday patient problems and prepare you for problem based integrated courses Human Genetics ,1980 The Era of Precision Medicine: Genetics, Pharmacy, and Personalized Health Edenilson Brandl, This book explores the fascinating field of precision medicine and its role in understanding and treating genetic diseases in various systems and organs of the human body Throughout the chapters topics such as precision medicine in the context of developmental diseases nutrigenomics and its relationship to diet personalization pulmonary diseases musculoskeletal diseases genetic diseases of the cardiovascular system and genetic diseases of the hepatic system are explored In addition subjects such as the application of genetic tests in precision medicine analysis of genetic variants precision medicine in women s health rare genetic diseases dermatological diseases nervous system diseases renal system diseases and metabolism diseases are addressed The book also explores the role of epigenetics in precision medicine highlighting how modifications to the genetic environment can influence disease development and progression In addition topics such as allergic diseases genetic diseases of the respiratory system and genetic diseases of the musculoskeletal system are discussed As a comprehensive guide this book provides valuable insights into precision medicine and its impact on clinical practice in the quest for more accurate diagnoses and personalized treatments It is essential reading for healthcare professionals researchers and students interested in the field of medical genetics and its applications in the era of precision medicine *Human Genetics, Informational and Educational Materials* National Clearinghouse for Human Genetic Diseases (U.S.), **Histology and Cell Biology: An Introduction to Pathology - E-Book** Abraham L. Kierszenbaum, Laura Tres, 2025-03-25 The first text to integrate microscopic anatomy with cell biology general pathology and physiology Histology and Cell Biology An Introduction to Pathology Sixth Edition continues to set the standard for studying today s rapidly evolving molecular oriented medicine Drs Abraham L Kierszenbaum and Laura L Tres present essential concepts in an accessible easy to understand manner with full color illustrations diagrams photomicrographs and pathology photos fully integrated on every page Linking pathological conditions to basic sciences throughout this award winning text helps you build a substantial clinical knowledge base while learning the structure and function of normal organs Provides cover to cover updates throughout reflecting current terminology basic science and clinical concepts Includes new material at the beginning of each chapter that assists you with the microscope as you first encounter the architectural complexity of

tissues or organs Includes Primers in most chapters that provide a practical self contained integration of histology cell biology and pathology helping you understand the relationship between basic and clinical sciences Contains Essential Concepts at the end of each chapter highlighting relevant issues perfect for studying and reviewing for in course and board examinations Uses a light green background throughout the text to identify essential concepts of histology a feature requested by both students and instructors to quickly locate which concepts are most important for beginning learners or when time is limited Identifies clinical terms throughout the text and lists all clinical boxes in the table of contents for quick reference Helps you understand the links between chapter concepts with concept mapping animations online providing a dynamic supplement to in class instruction

Manual de Genética Médica Fernando de Jesus Regateiro, 2003-12-01 Manual de Genética Médica inclui temas cujo conhecimento fundamental para sustentar um raciocínio em bases genéticas Cada tema congrega informa o fundamental para a percepção dos conceitos e a construção de conhecimento específico tendo como objectivo o desenvolvimento da capacidade crítica necessária para enfrentar as questões mais frequentes do mundo contemporâneo nesta área do saber e a necessidade de aprender ao longo da vida São temas deste livro história e desenvolvimento da genética bases celulares e moleculares da hereditariedade regulação da expressão genética diversidade humana mutações e reparação do DNA métodos de estudo do genoma humano história familiar heredograma tipos de hereditariedade Genética de populações ciclos de risco erros inatos do metabolismo Farmacogenética Ecogenética divisão celular cariótipo humano alterações cromossômicas numéricas e estruturais cromossomopatias Genética do desenvolvimento anomalias congénitas genes de regulação da proliferação celular apoptose senescência genes e cancro terapêutica genética aconselhamento genético genética Um extenso glossário foi também incluído

PDQ Medical Genetics Ronald G. Davidson, 2002 Description PDQ Medical Genetics challenges students to learn to think genetically to recognize genetic factors and to explain genetics to patients The author presents an approach to solving clinical problems with genetic components rather than on the presentation of facts that would soon fall out of date The text is problem oriented Each chapter opens with a case scenario designed to direct the student toward learning both the clinical and the basic biological science The featured scenarios also provide direction in the dealing with a patient's and family's personal difficulties by listing genetic disease support groups The informative text also emphasizes how to go about obtaining the latest information on such topics as molecular testing and imaging techniques Key Features Features topics not found in most genetics texts designed primarily for medical students such as complex disorders ambiguous genitalia pharmacogenetics and bone dysplasias Includes a dual platform mini CD ROM with the complete text and illustrations in fully searchable PDF files Incorporates references and links to carefully selected websites for additional readings and a bonus self assessment question and answer section for each chapter all included on the accompanying CD ROM This book will help students understand the nature of genetics and how its principles flow from those of evolution and natural selection It provides to the medical student

an approach to solving clinical problems and provides a conceptual framework within which to sort it all out Davidson approaches the subject of medical genetics as students will encounter it clinically The book is problem based and each chapter begins with a case scenario requiring a knowledge of genetics with which the practicing physician will have to come to grips fairly frequently with the emphasis on having the student recognize the applications of basic science data and solving the problem presented It *Emery and Rimoin's Essential Medical Genetics* David L. Rimoin, Reed E. Pyeritz, Bruce Korf, 2013-05-15 For decades Emery and Rimoin's Principles and Practice of Medical Genetics has provided the ultimate source for practicing clinicians to learn how the study of genetics can be integrated into practice Developed in parallel to the sixth edition and featuring 174 original contributions from the many authors of the full set this one volume work expertly condenses and synthesizes the most clinically relevant content for convenient desk reference Helping to bridge the gap between high level molecular genetics and individual application it follows the multi volume set in encompassing scientific fundamentals full spectrum discussion of major inherited disorders and actionable therapies Clinically oriented information is supported by concise descriptions of the principles of genetics research approaches and analytics to embrace the evolving population of students researchers and practitioners who are integrating their work to provide advanced diagnosis prevention and treatment of human disease This print volume is complemented and enhanced with online access to the complete text online only references and high quality illustrations on www.expertconsult.com Features 174 summarized contributions concisely discussing advances in cancer genetics genomic technologies and molecular genetics Contains hundreds of full colour illustrations supporting users with identification concept illustration and method processing Enhanced with full text online access high quality illustrations and online only references at www.expertconsult.com The Importance of Genetic Literacy and Education in Medicine Nina Pereza, Borut Peterlin, Saša Ostojić, 2022-05-24 **Genetics** Ruth M. DeBusk, 2003 Discover how the Human Genome Project will soon affect dietetic practice in this fascinating new reference Medical nutrition therapy nutrition and food service drug therapy gene therapy gene diagnostics and social and public nutrition policies will all feel the impact of this on going research Each chapter in the Genetic Connection begins to answer the question of how these advances will affect dietetics Prepare for the future with this exciting new title

Handbook of Clinical Adult Genetics and Genomics Shweta Dhar, Sandesh Sreenath Chakravarthy Nagamani, Tanya Eble, 2020-04-10 Handbook of Clinical Adult Genetics and Genomics A Practice Based Approach provides a thorough overview of genetic disorders that are commonly encountered in adult populations and supports the full translation of adult genetic and genomic modalities into clinical practice Expert chapter authors supplement foundational knowledge with case based strategies for the evaluation and management of genetic disorders in each organ system and specialty area Topics discussed include employing genetic testing technologies reporting test results genetic counseling for adult patients medical genetics referrals issues of complex inheritance gene therapy and diagnostic and treatment criteria for developmental

cardiovascular gastrointestinal neuropsychiatric pulmonary issues and much more Employs clinical case studies to demonstrate how to evaluate diagnosis and treat adult patients with genetic disorders Offers a practical framework for establishing an adult genetics clinic addressing infrastructure billing counseling and challenges unique to adult clinical genetics Features chapter contributions from authors at leading adult genetics institutions in the US and abroad

Techniques of Medical Genetics Dr. Ruchi Ratnesh, 2019-10-14 The knowledge of genetics and the bioinformatics has increased rapidly over the past few decades The application of this knowledge has become an integral part of patient care and the preventive thrust of modern medicine The aetiology of the majority of disorders has some genetic component The subject of genetics is gaining utmost importance in the medical science However most of the medical students find this subject difficult to understand and therefore neglect it The aim of this book is to present the text in such a way that it provides the clear and conceptual understanding of the subject A simple and concise account of different techniques of genetics will help students understand them better This book is not a comprehensive textbook of genetics Instead it takes a look at the basic concepts as well as interesting and clinically applicable aspects of genetics This book on techniques of genetics emphasize the central principles and how they apply to practice Photographs illustrations and tables containing patient family vignettes demonstrate clinical relevance and enhance visual impact of the material for easier and more effective learning and retention Includes access to the complete text and images online at student The latest knowledge and research on gene identification cancer genetics gene testing and gene therapy common disorders ethical and social issues and much more so you can keep up with current developments in genetics Bioinformatics NGS Data analysis Barcoding and Multiplexing Whole genome sequence analysis Gene bank Genomic browsers and Genomic databases are some of the interesting topics that has been provided for the students A knowledge of molecular genetics is essential to understand the latest developments in medicine An attempt has been made to outline the fundamentals of molecular genetics so that the reader gets familiar with the terminologies and techniques used in this field The students from various nonmedical background will also find this book useful in familiarizing them with the clinically applied aspects of genetics To make it easy text is supported with the flowchart

Integrating Evolutionary Biology Into Medical Education Jay Schulkin, Michael L. Power, Michael Power, 2019-12-12 This book builds a compelling case for integrating evolutionary biology into undergraduate and postgraduate medical education as well as its intrinsic value to medicine It achieves this within the broader context of medicine but through the focused lens of maternal and child health

The Encyclopaedia of Sports Medicine, Genetic and Molecular Aspects of Sports Performance Claude Bouchard, Eric P. Hoffman, 2011-08-02 This is the latest volume in the IOC Encyclopaedia of Sports Medicine series summarizing the evidence from all relevant sources on the genetic and molecular basis of sports and other human physical performance The initial chapters address the basic science of genomics and genetics and the regulation of gene expression Additional chapters provide authoritative information on the

genetics of complex performance phenotypes the contributions of small animal research family and twin studies and ethnic comparisons A final section addresses the issue of the contribution of specific genes and molecular markers as related to endurance strength and power and responsiveness to specific conditioning programs This latest volume in the Encyclopaedia of Sports Medicine Series from the Medical Commission of the International Olympic Committee is a must for sports and exercise scientists who require a thorough guide to the most cutting edge science in this expanding field

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