

THE BASICS OF GENETICS



WHAT IS GENETICS?

Genetics is the study of genes and inheritance. Genetics influences how we look and why certain traits and diseases run in families. The study of genetics allows us to understand how our genes are involved in the development of disease. To understand genetics, you need to know a bit about DNA, genes and chromosomes.

DNA (deoxyribonucleic acid) is a long molecule with a twisting ladder-like structure, called a double helix, that carries the instructions for making the human body. DNA is made of a chain of molecules called bases. There are four types of bases represented by the letters A, T, C and G and the order in which they occur is called the DNA sequence.

Genes are sections of DNA with a unique code (specific base sequence) and most genes contain the instructions to produce proteins in our bodies which carry out specific functions in the cell.



Genes are packaged inside **chromosomes**, which are found inside the nucleus of our cells.



HUMAN CELLS CONTAIN 46 CHROMOSOMES CONSISTING OF TWO SETS OF 23

We inherit one copy of all genes from each parent, **22 chromosomes** called autosomes that are the same in males and females and a **single sex chromosome, X or Y**. Following fertilization, we end up with **46 chromosomes** with two copies of each gene found on autosomes and two sex chromosomes. Females are XX and males are XY.



Sex chromosome X inherited from his mother



Sex chromosome X inherited from her mother

Sex chromosome Y inherited from his father



Sex chromosome X inherited from her father



Inheritance is the passing down of genetic material (DNA) from parent to child and is responsible for some of the traits that make us who we are, like hair color or eye color.

HUMAN GENOME

All the genetic material within our cells makes up the **human genome**. Each chromosome contains many genes and the human genome is estimated to contain **20,000 to 25,000 genes**.



GENETIC VARIATION

Overall, our DNA sequences are very similar. However, there are small differences between the DNA sequences of individual genomes and this is called **genetic variation**. These variations make us **unique**.



Genetic variations can be caused by differences in a single base (**single nucleotide polymorphisms**), a common cause of natural variations in the DNA sequence

OR

by **mutations** which alter the sequence of larger sections of DNA. Mutations within genes are also known as **variants**. Variants can lead to changes in genes that affect the production or function of proteins. In turn this can have a negative impact on your body and cause disease.



Basic Concepts In Medical Genetics

Rosina Ehmman



Basic Concepts In Medical Genetics:

Basic Concepts in Medical Genetics Marshall Horwitz, Mary Beth Dinulos, Kris Carroll, 2000 **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 **Basic Concepts in Medical Genetics** Marshall Horwitz, Mary Beth Dinulos, 2000 The main focus of this text is on medical genetics with a general approach to explanations It offers key concepts by the way of example to common problems and illustrations of concepts through working through some of the classic and prototypical problems that students encounter **Emery and Rimoin's Principles and Practice of Medical Genetics and Genomics** Reed E. Pyeritz, Bruce R. Korf, Wayne W. Grody, 2024-10-23 Selected for 2025 Doody's Core Titles with Essential Purchase designation in Clinical Genetics For decades Emery and Rimoin's Principles and Practice of Medical Genetics and Genomics has served as the ultimate resource for clinicians integrating genetics and genomics into medical practice With detailed coverage in contributions from more than 250 of the world's most trusted authorities in medical genetics and a series of 11 volumes available for individual sale the Seventh Edition of this classic reference includes the latest information on seminal topics such as prenatal diagnosis genome sequencing public health genetics genetic counseling and management and treatment strategies to complete its coverage of this growing field for students health providers and researchers involved in the care of patients with genetic conditions and increasingly all areas of health and disease This comprehensive yet practical resource emphasizes theory and research fundamentals related to the applications of medical genetics and genomics across the full spectrum of inherited disorders and applications to medicine more broadly In this volume leading physicians and researchers thoroughly examine medical genetics and genomics as applied to developmental disorders as well as genetic conditions that affect hearing and vision Here genetic researchers students and health professionals will find new and fully revised chapters on human developmental genetics disorders affecting craniofacial development chromosomal abnormalities including aneuploidies and structural abnormalities hereditary hearing impairment and various genetic conditions of the eye With regular advances in genomic technologies propelling precision

medicine into the clinic Emery and Rimoin's Principles and Practice of Medical Genetics and Genomics Seventh Edition bridges the gap between high level molecular genetics and practical application and serves as an invaluable clinical tool for health professionals and researchers Thoroughly introduces genetic researchers students and healthcare professionals to the principles of human developmental genetics Examines a wide range of developmental disorders including craniofacial development as well as disorders affecting hearing and vision Includes color images supporting identification concept illustration and method processing Features contributions by leading international researchers and practitioners of medical genetics

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 E-Book Lynn B. Jorde, John C. Carey, Michael J. Bamshad, 2009-10-06 Medical Genetics is the clearest and most concise text on the subject providing state of the art coverage of clinically relevant molecular genetics Lynn B Jorde PhD John C Carey MD and Michael J Bamshad MD integrate recent developments with clinical practice and emphasize the central principles of genetics and their clinical applications Now in full color this edition provides you with the stunning visual clarity so important in this field Get the very latest on hot topics like gene identification cancer genetics gene testing and gene therapy common diseases ethical and social issues personalized medicine and much more This is an indispensable resource that should be on every reading list This title includes additional digital media when purchased in print format For this digital book edition media content is not included Features mini summaries study questions suggested reading and a detailed glossary to supplement and reinforce what you learn from the text Demonstrates clinical relevance through over 230 photographs illustrations and tables along with boxes containing patient family vignettes Enhances the visual impact of the material with full color illustrations throughout the text for easier and more effective learning and retention Presents a new chapter on genomics and personalized medicine for the latest on these hot topics Provides you with 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 Includes study questions at the end of every chapter so you can test yourself and retain the material Features additional clinically commentary boxes throughout the text to show the relevance of genetics to everyday patient problems to prepare you for problem based integrated courses

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

Emery and Rimoin's Principles and Practice of Medical Genetics and Genomics Reed E. Pyeritz, Bruce R. Korf, Wayne W. Grody, 2021-11-02 Selected for Doody's Core Titles 2024 in Clinical Genetics Emery and Rimoin's Principles and Practice of Medical Genetics and Genomics Perinatal and Reproductive Genetics Seventh Edition includes the latest information on seminal topics such as prenatal diagnosis genome and exome sequencing public health genetics genetic counseling and management and treatment strategies in this growing field The book is ideal for medical students residents physicians and researchers involved in the care of patients with genetic conditions This comprehensive yet practical resource emphasizes theory and research fundamentals related to applications of medical genetics across the full spectrum of inherited disorders and applications to medicine more broadly Chapters from leading international researchers and clinicians focus on topics ranging from single gene testing to whole genome sequencing whole exome sequencing gene therapy genome editing approaches FDA regulations on genomic testing and therapeutics and ethical aspects of employing genomic technologies Fully revised and up to date this new edition introduces genetic researchers students and healthcare professionals to genomic technologies testing and therapeutic applications Examines key topics and developing methods within genomic testing and therapeutics including single gene testing whole genome and whole exome sequencing gene therapy and genome editing variant Interpretation and classification and ethical aspects of applying genomic technologies Includes color images that support the identification concept illustration and method of processing Features contributions by leading international researchers and practitioners of medical genetics Provides a robust companion website that offers further teaching tools and links to outside resources and articles to stay up to date on the latest developments in the field

The Importance of Genetic Literacy

and Education in Medicine Nina Pereza, Borut Peterlin, Saša Ostojić, 2022-05-24 *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

Medical Genetics and Genomics Lynn B. Jorde, John C. Carey, Michael J. Bamshad, 2026-02 Known for its up to date coverage of both basic science and clinical application Medical Genetics and Genomics 7th Edition has been fully revised to reflect recent advances in the genetics of common diseases as well as current progress in genetic testing and gene therapy 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 materia Discusses topical issues such as polygenic scores and their potential uses in the diagnosis and treatment of diseases such as diabetes cancer and heart disease along with the latest advancements in sequencing technologies and their role in genetic testing and diagnosis Provides an up to date discourse on the modes and applications of genetic testing Contains useful concept summaries more than 230 photographs illustrations and tables as well as patient family vignettes that present valuable perspectives on disease and treatment Features a larger quantity of 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 Uses examples of diseases to illustrate important concepts demonstrating their relevance to medicine Study questions for self assessment are provided within the print book as well as 200 additional USMLE style questions online An eBook version is included with purchase The eBook allows you to access all of the text and figures with the ability to search customize your content make notes and highlights and have content read aloud *Medical Genetics and Genomics - E-BOOK* Lynn B. Jorde, John C. Carey, Michael J. Bamshad, 2026-03-15 Known for its up to date coverage of both basic science and clinical application Medical Genetics and Genomics 7th Edition has been fully revised to reflect recent advances in the genetics of common diseases as well as current progress in genetic testing and gene therapy 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 materia Discusses topical issues such as polygenic scores

and their potential uses in the diagnosis and treatment of diseases such as diabetes cancer and heart disease along with the latest advancements in sequencing technologies and their role in genetic testing and diagnosis Provides an up to date discourse on the modes and applications of genetic testing Contains useful concept summaries more than 230 photographs illustrations and tables as well as patient family vignettes that present valuable perspectives on disease and treatment Features a larger quantity of 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 Uses examples of diseases to illustrate important concepts demonstrating their relevance to medicine Study questions for self assessment are provided within the print book as well as 200 additional USMLE style questions online

Understanding Genetics Genetic Alliance, New York-Mid-Atlantic Consortium for Genetic and Newborn Screening Services, 2009 The purpose of this manual is to provide an educational genetics resource for individuals families and health professionals in the New York Mid Atlantic region and increase awareness of specialty care in genetics The manual begins with a basic introduction to genetics concepts followed by a description of the different types and applications of genetic tests It also provides information about diagnosis of genetic disease family history newborn screening and genetic counseling Resources are included to assist in patient care patient and professional education and identification of specialty genetics services within the New York Mid Atlantic region At the end of each section a list of references is provided for additional information Appendices can be copied for reference and offered to patients These take home resources are critical to helping both providers and patients understand some of the basic concepts and applications of genetics and genomics

Medical Genetics and Society Norio Fujiki, Victor Bulyzhenkov, Zbigniew Bańkowski, 1991

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 **Medical Genetics** Lynn B. Jorde,1999
The emphasis of this book is on those aspects of medical genetics most useful in a modern clinical practice Clinical aspects of molecular genetics research have been incorporated throughout the spectrum of genetically determined diseases As such the book covers not only chromosomal and single gene disorders but also the more challenging multifactorial diseases

American Journal of Medical Genetics ,1985 **Mutating Concepts, Evolving Disciplines: Genetics, Medicine, and Society** L.S. Parker,Rachel A. Ankeny,2012-12-06 Advances in genetics such as the Human Genome Project s successful mapping of the human genome and the discovery of ever more sites of disease related mutations invite re examination of basic concepts underlying our fundamental social practices and institutions Having children assigning responsibility identifying causes using social and scientific resources to improve human well being among other concepts will never be the same Our concepts of moral and legal responsibility cause and effect disease prevention health disability enhancement personal identity and reproductive autonomy and responsibility are all subtly changing in response to developments in genetics Biology law medicine and other disciplines are also evolving in response to mutating concepts in genetics itself for example dominance causation behavior gene expression and gene The selections in this volume employ philosophical and historical perspectives to shed light on classic social ethical and philosophical issues raised with renewed urgency against the backdrop of the mapping of the human genome **Progress in Medical Genetics** Arthur Gerald Steinberg,1985

Genetics and Genomics in Medicine Tom Strachan,Anneke Lucassen,2022-10-31 The second edition of this textbook written for undergraduate students graduate students and medical researchers Genetics and Genomics in Medicine explains the science behind the uses of genetics and genomics in medicine today and how it is being applied Maintaining the features that made the first edition so popular this second edition has been thoroughly updated in line with the latest developments in the field DNA technologies are explained with emphasis on the modern techniques that are revolutionizing the use of genetic information in medicine and indicating the role of genetics in common diseases Epigenetics and non coding RNA are covered in depth as are genetic approaches to treatment and prevention including pharmacogenomics genetic testing and personalized medicine A dedicated chapter charts the latest insights into the molecular basis of cancers cancer genomics and novel approaches to cancer detection Coverage of genetic testing at the level of genes chromosomes and genomes has been significantly expanded and updated Extra prominence has been given to additional genomic analyses ethical aspects and novel therapeutic approaches Various case studies illustrate selected clinical applications Key Features Comprehensive and integrated account of how genetics and genomics affect the entire spectrum of human health and disease Exquisite artwork illuminates the key concepts and mechanisms Summary points at the end of each chapter help to consolidate learning For each chapter an abundance of further reading to help provide the reader with direction for further study Inclusive online question bank to test understanding Standard boxes summarizing certain key principles in genetics Clinical boxes

summarizing selected case studies pathogenesis mechanisms or novel therapies for selected diseases This book is equally suited for newcomers to the field as well as for engineers and scientists that have basic knowledge in this field but are interested in obtaining more information about specific future applications

Unveiling the Magic of Words: A Report on "**Basic Concepts In Medical Genetics**"

In a global defined by information and interconnectivity, the enchanting power of words has acquired unparalleled significance. Their power to kindle emotions, provoke contemplation, and ignite transformative change is actually awe-inspiring. Enter the realm of "**Basic Concepts In Medical Genetics**," a mesmerizing literary masterpiece penned with a distinguished author, guiding readers on a profound journey to unravel the secrets and potential hidden within every word. In this critique, we shall delve into the book's central themes, examine its distinctive writing style, and assess its profound affect on the souls of its readers.

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