

# 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

**Sam Goldstein, Cecil R. Reynolds**



## **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 An easy to understand detail by detail explanation of basic Mendelian Genetics Ideal for first and second year medical students pediatric and ob gyn clerks advanced undergraduates and genetic counseling graduate students Key concepts are illustrated with clinical examples frequently encountered in courses in genetics and medical genetics such as cystic fibrosis Down s Syndrome Neurofibromatosis familial breast cancer hereditary colon cancer Becker Muscular Dystrophy and sickle cell disease Features special sections on prenatal genetics and common and cancer mutations

**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 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

[The Importance of Genetic Literacy and Education in Medicine](#) Nina Pereza, Borut Peterlin, Saša Ostojić, 2022-05-24

**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

**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: 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

**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

Medical Genetics and Society Norio Fujiki, Victor Bulyzhenkov, Zbigniew Bańkowski, 1991 Medical Genetics Lynn B. Jorde, 2000 Spanish version also available ISBN 84 8174 423 9 **Genetics For Dummies** Rene Fester Kratz, Lisa Spock, 2023-12-27 Evolve your knowledge of the fast moving world of genetic research Genetics For Dummies shines a light on the fascinating field of genetics helping you gain a greater understanding of how genetics factors into everyday life Perfect as a supplement to a genetics course or as an intro for the curious this book is packed with easy to understand explanations of the key concepts including an overview of cell biology You'll also find tons of coverage of recent discoveries in the field plus info on how genetics can affect your health and wellbeing Whole genome sequencing genetic disease treatments exploring your ancestry non invasive prenatal testing it's all here in the friendly and relatable Dummies style you love Grasp the basics of cell biology and get a primer on the field of genetic research Discover what you can learn about yourself thanks to advances in genetic testing Learn how your genes influence your health and wellbeing today and as you age Follow along with your college level genetics course or refresh your knowledge with clear explanations of complex ideas Genetics For Dummies is great for students of the biological sciences and for the genetically curious everywhere

**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

**Genetics for the Health Sciences** Heather Skirton, Christine Patch, 2009 Based on their extensive experience of clinical work the authors emphasize the practical issues related to the healthcare of individuals and families Genetics for the Health Sciences takes an holistic approach from preconception to adulthood and addresses the false notion that clinical genetics is of relevance only to those who are planning a family The book enables nurses midwives genetic counselors and doctors to apply the general principles of genetics in their routine clinical practice As well as discussing the basic principles Genetics for the Health Sciences also describes the latest technologies and shows how these can be applied to clinical practice This is an essential text which helps all those in clinical healthcare understand the genetics they need in their professional roles It is also an ideal coursebook for students in the healthcare professions seeking an understanding of core genetic principles and how these are applied in practice

Reviews The text is a compact concise presentation of the basic concepts in genetics science and the impact of genetics across the lifespan The organization of the text increases its usefulness to clinicians as each specialty area could easily locate the information most pertinent to their work The text is well illustrated throughout again providing summarized information that is easily accessed An especially helpful feature of this text is the presence of multiple case studies in each chapter making this text particularly useful for teaching I think faculty teaching students in advanced practice nursing programs and allied health courses would find this text a succinct addition to their course However I could also see this text being useful in RN preparation curriculum It could easily be a text that would carry over for several specialty areas and provide additional material specific to genetics in each area

Genetics for Health Sciences A Handbook for Clinical Healthcare is a welcome addition to the rather sparse choices available for presenting genetics content in clinical practice curriculum

Rebekah Hamilton ISONG June 2010 Tremendous progress in recent years has shaped the field of medical genetics which continues to expand to involve almost every aspect of human health Hence it is necessary for every healthcare professional to acquire a basic understanding of this science This is clearly the objective of this well edited and structured book by Skirton and Patch in its second updated edition The authors considerable experience in genetics through direct clinical practice in addition to organisational and academic roles can be appreciated in the

practical aspect of their writing The book starts with useful definitions and alerts the reader to the wide applicability of genetics through clinical cases raising questions that are answered and discussed when relevant in later chapters Important aspects of genetic counselling from basic skills such as risk estimation using family trees to more complex ones related to effective ethical communication with the clients in the light of their needs and the different models of counselling are then discussed in two chapters However given their intrinsic function in medical genetics these issues are also efficiently tackled in the clinical cases outlined in each chapter The authors then describe important scientific concepts and techniques that are necessary for a good clinical understanding of genetics Although this section may seem short the additional resources provided at the end are very useful Likewise for all topics covered in the book the updated links provided serve as a handy catalogue for the health professional seeking extra information Next the concepts and techniques described in the first part of the book are applied directly to common genetic conditions and issues faced by different age groups from prenatal care to older adulthood in separate chapters This organisation puts into perspective the importance of genetics in different medical specialities and settings This book with its concise yet comprehensive explanation of a complex rapidly growing field should prove to be a valuable resource for a diverse audience of health professionals including midwives nurses and physicians who confront genetic issues during their daily practice without being specialists in medical genetics Nadine Taleb Journal of Medical Genetics

When first asked to write this book review I thought to myself how it would be important to give a rounded report on both the book s strengths and weaknesses After reading the book however I realised that it is difficult to find weaknesses The authors Heather Skirton and Christine Patch between them have over 30 years of experience in clinical genetics Their experience of explaining genetics in their professional roles shines through in the way they have written the book They make complex genetic principles interesting and understandable The book does not baffle or patronise The book is easily navigated It is broken down into clear chapters that are ordered in a pleasingly logical way The first chapter sets the scene by introducing the reader to important concepts related to genetic health care including issues such as ethical practice the different forms of genetic testing and the impact of genetic conditions on families The next chapters then discuss the family tree counselling issues genetic science and public health genetics The remaining chapters then explore the core topics relating to particular life stages from preconception to older adulthood It engages the reader from the start when it introduces seven core families that reappear throughout the book to highlight a number of key issues The text is peppered with clear illustrations useful step by step guides practical checklists and test yourself sections making the book lively and dynamic The test yourself sections make the book student friendly and would also make good teaching aids to lecturers and tutors At the start of the book there is a list of helpful websites and at the end of each chapter there is a list of resources for those who wish to extend their knowledge One of the main strengths of this book is its refreshingly practical approach All too often books can be written in a way in which it is difficult for the reader to transfer the knowledge that they have gained into

their day to day practice The introduction states that the aim of this book is to enable those in healthcare to update their knowledge on topics related to genetics and genomics that have an impact in their daily work and apply it usefully in patient care Given the fact that genetics has increasingly become a core component in a number of specialties one would think that meeting this aim for all would be a tall order This book however eloquently does so using case examples and key practice points boxes throughout to demonstrate the clinical application of the topic under discussion Another of the book s main strengths is the way in which it promotes individualised care by enabling the reader to consider the impact of genetic conditions from the patients point of view Genetics for the Health Sciences brings genetics into common healthcare settings It is highly recommended as an essential text for health care professionals in roles across all specialties It is also recommended to students lecturers social scientists to anyone who has an interest in genetics and wishes to extend their knowledge It is a joy to read and could be read from cover to cover Helen Thistlewood Medical Genetics Genetics is at the forefront of medicine and nurses are expected to have a basic understanding of the subject This handbook is well written and the authors do a good job of making this complex topic understandable The book lists prenatal childhood and adult genetic conditions that may involve testing and counselling Genetic services types of genetic testing and the impact of genetic conditions on families are discussed in depth and case studies highlight the emotional and psychological needs of patients and their families Guidelines for drawing family trees and practical examples of risk assessment are well explained The perspective of the patient regarding risk and lay knowledge is also considered All departments should have a copy of this book on their shelves Audrey Ardern Jones Nursing Standard

**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 **American Journal of Medical Genetics** ,1985

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