

## Chapter 1: Human Genome

### 1.1. INTRODUCING GENES AND GENOME

- **Genetics** – study of heredity, traits and their variations.
  - study of how traits are transmitted.
- **Heredity** – transmission of traits and biological information.
- **Genes** – basic unit of heredity.
  - instruct cell's production of proteins.
  - consist of long DNA.
  - provide-encoding genes are scattered among 3.2 billion DNA bases in each set of chromosomes.
- **Phenotype** – control the characteristics of every individual.
- **DNA (Deoxyribonucleic Acid)** – transmits information in sequence of nitrogenous bases.
- **Genome** – make up of complete set of genetic instructions characteristic of each an organism.
  - all of our cells contain two sets copies of genome.
- **Genomics** – scientific field that analyzes genomes.
  - reveals how closely related are different species.

### 1.2 LEVELS OF GENETICS AND GENOMICS

- **DNA (Deoxyribonucleic Acid)**
  - Shape: double helix
  - Consist of: alternating sugar and phosphates
  - Function: provide information to manufacture specific proteins



- 4 types of nitrogenous bases:
  - Adenine (A) and Thymine (T)
  - Cytosine (C) and Guanine (G)
  - Each set of these complementary bases is a code for a particular amino acid.
- **Amino Acid** – building blocks of protein.
  - align and link like snap beads forming protein.

### 1.3. THREE PROCESSES OF ACCESSING GENETIC INFORMATION

- **DNA replication** – (1) chains of double helix untwist and separate.
  - (2) each full helix is a new partner chain from two DNA bases.
- **Transcription** – (3) copies the sequence of one strand of DNA molecule into mRNA.
- **Translation** – (4) each 3 DNA bases is a new attract another type of RNA, that functions as a connector, bringing in a particular amino acid.
  - (5) Amino acids align and link to form protein.

### 1.2 THE HUMAN GENOME

- **20,125 protein-encoding genes** comprise the genome.
  - Accounts: 1.5 percent of human genome, 8.5 percent of known genetic diseases.
- **Alloles ("gene variants")** – some proteins – encoding genes may vary in DNA base sequence from parent to parent that is passed to the next generation.
- **Mutations ("changed gene")** – variation or change in genes, causing a disease.

### 1.1. MUTATION DISEASES

- **Cystic Fibrosis** – inherited disease
  - Change of a "C" to "T" inserts amino acid rather than the amino acid glycine which results to protein becoming abnormal.
  - Cystic Fibrosis Transmembrane

# Chapter 1the Human Genome Continued

**Andreas D. Baxevanis, Gary D.  
Bader, David S. Wishart**



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**Bioinformatics** Andreas D. Baxevanis, Gary D. Bader, David S. Wishart, 2020-05-12 Praise for the third edition of Bioinformatics This book is a gem to read and use in practice Briefings in Bioinformatics This volume has a distinctive special value as it offers an unrivalled level of details and unique expert insights from the leading computational biologists including the very creators of popular bioinformatics tools ChemBioChem A valuable survey of this fascinating field I found it to be the most useful book on bioinformatics that I have seen and recommend it very highly American Society for Microbiology News This should be on the bookshelf of every molecular biologist The Quarterly Review of Biolog The field of bioinformatics is advancing at a remarkable rate With the development of new analytical techniques that make use of the latest advances in machine learning and data science today s biologists are gaining fantastic new insights into the natural world s most complex systems These rapidly progressing innovations can however be difficult to keep pace with The expanded fourth edition of the best selling Bioinformatics aims to remedy this by providing students and professionals alike with a comprehensive survey of the current field Revised to reflect recent advances in computational biology it offers practical instruction on the gathering analysis and interpretation of data as well as explanations of the most powerful algorithms presently used for biological discovery Bioinformatics Fourth Edition offers the most readable up to date and thorough introduction to the field for biologists at all levels covering both key concepts that have stood the test of time and the new and important developments driving this fast moving discipline forwards This new edition features New chapters on metabolomics population genetics metagenomics and microbial community analysis and translational bioinformatics A thorough treatment of statistical methods as applied to biological data Special topic boxes and appendices highlighting experimental strategies and advanced concepts Annotated reference lists comprehensive lists of relevant web resources and an extensive glossary of commonly used terms in bioinformatics genomics and proteomics Bioinformatics is an indispensable companion for researchers instructors and students of all levels in molecular biology and computational biology as well as investigators involved in genomics clinical research proteomics and related fields *Immunology & Serology in Laboratory Medicine - E-Book* Mary Louise Turgeon, 2020-12-16 Building on a solid foundation of knowledge and skills this classic text from trusted author Mary Louise Turgeon clearly explains everything from basic immunologic mechanisms and serologic concepts to the theory behind procedures performed in the lab This go to resource prepares you for everything from mastering automated techniques to understanding immunoassay instrumentation and disorders of infectious and immunologic origin Packed with learning objectives review questions step by step procedures and case studies this text is the key to your success in today s modern laboratory environment Procedural protocols help you transition from immunology theory to practical aspects of the clinical lab Case studies allow you to apply your knowledge to real world situations and strengthen your critical thinking skills Updated illustrations photographs and summary tables visually clarify key concepts and information Full color presentation

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**Core Laboratory Technologies in Clinical Immunology E-Book** Robert R. Rich, 2018-10-24 Edited by clinical immunology expert Dr Robert R Rich this concise focused title covers today s most important technologies used in the diagnosis and evaluation of immunologic disease Core Laboratory Technologies in Clinical Immunology is ideal for immunology researchers and scientists as well as immunologists and others interested in the principles and uses of current lab technologies in immunology Focuses on how today s technologies relate to the diagnosis of disease including state of the art technologies that are significantly impacting cancer therapy research Covers flow cytometry assessment of functional immune responses in lymphocytes assessment of neutrophil function molecular methods and more Provides information of special interest to researchers and scientists who are directly involved in the rapidly changing world of clinical immunology as well as immunologists oncologists and medical technology and biomedical engineers Consolidates today s available information and guidance into a single convenient resource

**Harper's Illustrated Biochemistry** Mr. Rohit Manglik, 2024-03-02 EduGorilla Publication is a trusted name in the education sector committed to empowering learners with high quality study materials and resources Specializing in competitive exams and academic support EduGorilla provides comprehensive and well structured content tailored to meet the needs of students across various streams and levels

Improving Global Health Barry B. Hughes, Randall Kuhn, Cecilia Mosca Peterson, Dale S. Rothman, Jose Roberto Solorzano, 2015-11-17 Improving Global Health is the third in a series of volumes Patterns of Potential Human Progress that uses the International Futures IFs simulation model to explore prospects for human development how development appears to be unfolding globally and locally how we would like it to evolve and how better to assure that we move it in desired directions Earlier volumes addressed the reduction of global poverty and the advance of global education Volume 3 sets out to tell a story of possible futures for the health of peoples across the world Questions the volume addresses include What health outcomes might we expect given current patterns of human development What opportunities exist for intervention and the achievement of alternate health futures How might improved health futures affect broader economic social and political

prospects of countries regions and the world

**Biology: How Life Works (Volume 1)** James Morris,2013-01-04

Ordinarily textbooks are developed by first writing chapters then making decisions about art and images and finally once the book is complete assembling a test bank and ancillary media This process dramatically limits the integration across resources and reduces art media and assessments to ancillary material rather than essential resources for student learning Biology How Life Works is the first project to develop three pillars the text the visual program and the assessment at the same time All three pillars were developed in parallel to make sure that each idea is addressed in the most appropriate medium and to ensure authentic integration These three pillars are all tied to the same set of core concepts share a common language and use the same visual palette In this way the text visual program and assessments are integral parts of student learning rather than just accessories to the text

Lippincott Williams and Wilkins' Administrative Medical Assisting Laura Southard Durham,2008 Lippincott Williams Wilkins Administrative Medical Assisting Second Edition teaches students the theory and skills to become effective medical office assistants The text and ancillary resources address all the required administrative competencies for CAAHEP and ABHES program accreditation The book includes critical thinking questions and is written for maximum readability with a full color layout over 100 illustrations and boxes to highlight key points A bound in CD ROM and a companion Website include CMA RMA exam preparation questions an English to Spanish audio glossary a clinical simulation administrative skill video clips competency evaluation forms and worksheets for practice A Skills DVD with demonstrations of the most important medical assisting skills is available separately An Instructor s Resource CD ROM and online instructor resources will be available gratis upon adoption of the text

**Law, Medicine and Ethics** Patricia A. King,Judith Areen,Lawrence Ogalthorpe Gostin,2006 This reader is a spin off publication from Law Science and Medicine Third Edition It offers an interdisciplinary perspective on ethical and moral questions raised by medicine and introduces undergraduates to the manner in which law and policy shape society s views on birth death human experimentation reproduction use of genetic information and beyond

*Medical Genetics* G. Bradley Schaefer,James N. Thompson,2013-11-22 A complete introductory text on how to integrate basic genetic principles into the practice of clinical medicine Medical Genetics is the first text to focus on the everyday application of genetic assessment and its diagnostic therapeutic and preventive implications in clinical practice It is intended to be a text that you can use throughout medical school and refer back to when questions arise during residency and eventually practice Medical Genetics is written as a narrative where each chapter builds upon the foundation laid by previous ones Chapters can also be used as stand alone learning aids for specific topics Taken as a whole this timely book delivers a complete overview of genetics in medicine You will find in depth expert coverage of such key topics as The structure and function of genes Cytogenetics Mendelian inheritance Mutations Genetic testing and screening Genetic therapies Disorders of organelles Key genetic diseases disorders and syndromes Each chapter of Medical Genetics is logically organized into three sections Background and

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*Harper's Illustrated Biochemistry, Thirty-Second Edition* Peter J. Kennelly, Kathleen M. Botham, Owen McGuinness, Victor W. Rodwell, P. Anthony Weil, 2022-08-30 Gain a thorough understanding of the principles of biochemistry as they relate to clinical medicine A Doody's Core Title for 2024 2023 The Thirty Second Edition of Harper's Illustrated Biochemistry combines top quality full color illustrations with authoritative integrated coverage of biochemical disease and clinical information Featuring numerous medically relevant examples this respected text presents a clear succinct review of the fundamentals that every student must understand in order to succeed in medical school All 58 chapters help you understand the medical relevance of biochemistry Full color presentation with 600 illustrations Chapters have been updated to reflect the latest information Case studies emphasize the clinical relevance of biochemistry Review questions follow each of the 11 sections Boxed objectives define the goals of each chapter Tables encapsulate important information Each chapter contains a section on biomedical importance and a summary of the topics covered Applauded by medical students for its current and engaging style Harper's Illustrated Biochemistry is an essential for USMLE review and the single best reference for learning the clinical relevance of any biochemistry topic

**Genetics** Lori B. Andrews, Maxwell J. Mehlman, Mark A. Rothstein, 2002

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**Modeling in Higher Organisms** Lawrence Sean Hon, 2005 It is hoped that these efforts using large scale datasets and  
efficient algorithms will allow further advances in understanding mammalian transcriptional regulation at the sequence level



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