

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.
- **Genetics** – 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 half builds a new partner chain from free DNA bases.
- **Transcription** – (3) copies the sequence of one strand of DNA molecule into mRNA.
- **Translation** – (4) each 3 RNA bases in a row 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 in protein becoming abnormal
 - Cystic Fibrosis Transmembrane

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Functional Analysis of the Human Genome Farzin Farzaneh, David N. Cooper, 1995 An excellent review of the relationship between structure and function in the human genome and a detailed description of some of the important methodologies for unravelling the function of genes and genomic structures

Bioinformatics and the Cell Xuhua Xia, 2018-07-05 This second edition integrates the more technical and mathematical aspects of bioinformatics with concrete examples of their application to current research problems in molecular cellular and evolutionary biology This broad unified approach is made possible in large part by the very wide scope of Dr Xia s own research experience The integration of genomics proteomics and transcriptomics into a single volume makes this book required reading for anyone entering the new and emerging fields of Systems Biology and Evolutionary Bioinformatics

The Human Disease Sabrina Sholts, 2024-04-09 How the very fact of being human makes us vulnerable to pandemics and gives us the power to save ourselves The COVID 19 pandemic won t be our last because what makes us vulnerable to pandemics also makes us human That is the uncomfortable but all too timely message of The Human Disease which travels through history and around the globe to examine how and why pandemics are an inescapable threat of our own making Drawing on dozens of disciplines from medicine epidemiology and microbiology to anthropology sociology ecology and neuroscience as well as a unique expertise in public education about pandemic risks biological anthropologist Sabrina Sholts identifies the human traits and tendencies that double as pandemic liabilities from the anatomy that defines us to the misperceptions that divide us Weaving together a wealth of personal experiences scientific findings and historical stories Sholts brings dramatic and much needed clarity to one of the most profound challenges we face as a species Though the COVID 19 pandemic looms large in Sholts s account it is in fact just one of the many infectious disease events explored in The Human Disease With its expansive evolutionary perspective the book explains how humanity will continue to face new pandemics because humans cause them by the ways that we are and the things that we do By recognizing our risks Sholts suggests we can take actions to reduce them When the next pandemic happens and how bad it becomes are largely within our highly capable human hands and will be determined by what we do with our extraordinary human brains

Targeted Genome Engineering via CRISPR/Cas9 in Plants Alvina Gul, 2024-07-18 Targeted Genome Engineering via CRISPR Cas9 in Plants provides in depth insights into the use of the emerging CRISPR Cas9 technology for precise genome editing This technology has revolutionized plant science research particularly for crop improvement owing to its simplicity and efficiency The book provides a wide range of CRISPR Cas9 gene editing techniques for a variety of plants Chapters include the latest applications of CRISPR Cas9 system in connection with abiotic stresses biotic stresses biofortification yield improvement disease modelling and prognosis and molecular diagnosis Targeted Genome Engineering via CRISPR Cas9 in Plants also evaluates various regulatory and ethical aspects that must be considered when implementing the CRISPR Cas9 approach This book is a valuable resource for professionals and researchers

as it provides effective CRISPR Cas9 based strategies for sustainable agriculture and treatment of various diseases Explains basic mechanism and implementation of CRISPR Cas9 technology in a wide range of plants Provides practical guidance on the applications of CRISPR Cas9 in different scientific disciplines of plant science Discusses the risks and challenges of genome engineering *Cell and Gene Therapies for Neurologic Diseases*, 2024-10-02 Cell and Gene Therapies for Neurologic Diseases Volume 205 comprehensively covers the scientific background translational efforts clinical developments and registered biologics that have entered into clinical practice Coverage includes types of therapies available and in development and best practice uses for a variety of neurological disorders including Parkinson s Huntington s ALS stroke spinal cord RP demyelination and epilepsy As the emergence of gene and cellular therapeutics has changed the clinical landscape for a variety of disorders and is now ready to do so for neurological diseases these therapeutic modalities currently complement and may in time supplant small molecule drugs Summarizes advances in cell and gene therapy for neurological diseases Describes the therapies available and in development Includes surgical ethical and manufacturing considerations Identifies best practices for specific neurological diseases Covers Huntington s Parkinson s ALS Stroke Demyelination epilepsy and more **Plant Cell Biology** Randy O. Wayne, 2024-11-15 Plant Cell Biology From Astronomy to Zoology Third Edition connects the fundamentals of plant anatomy plant physiology plant growth and development plant taxonomy plant biochemistry plant molecular biology and plant cell biology It covers all aspects of plant cell biology without emphasizing any one plant organelle molecule or technique Although most examples are biased towards plants basic similarities between all living eukaryotic cells animal and plant are recognized and used to best illustrate cell processes This is a must have reference for scientists with a background in plant anatomy plant physiology plant growth and development plant taxonomy and more Includes chapter on using mutants and genetic approaches to plant cell biology research and a chapter on omic technologies Explains the physiological underpinnings of biological processes to bring original insights relating to plants Includes examples throughout from physics chemistry geology and biology to bring understanding on plant cell development growth chemistry and diseases Provides the essential tools for students to be able to evaluate and assess the mechanisms involved in cell growth chromosome motion membrane trafficking and energy exchange **Incapacitating Biochemical Weapons** Alan M. Pearson, Marie Isabelle Chevrier, Mark Wheelis, 2007 Incapacitating Biochemical Weapons comprehensively examines the scientific military legal political and social issues raised by the pursuit of such weapons issues that urgently require the attention of scientists military and civilian practitioners and policy makers and broader civil society *Instructors Guide to Text and Media: Igenetics* Rebecca V. Ferrell, 2001-10-18 *Genetics of Hematological Disorders* Christos S. Bartsocas, Dimitris Loukopoulos, 1992 Examines recent advances in the genetics of haematologic disorders The papers upon which the book is based were presented at an international seminar They deal with haemoglobinopathies and the thalassemias the erythrocyte enzymopathies haemophilias and blood malignancies The Usage of Ochre at the Verge

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