

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") – alteration or change in genes, causing a disease.

1.3. INHERITED 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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Ogalthorpe Gostin**



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Table of Contents Chapter 1the Human Genome Inquiry Activity

1. Understanding the eBook Chapter 1the Human Genome Inquiry Activity
 - The Rise of Digital Reading Chapter 1the Human Genome Inquiry Activity
 - Advantages of eBooks Over Traditional Books
2. Identifying Chapter 1the Human Genome Inquiry Activity
 - Exploring Different Genres
 - Considering Fiction vs. Non-Fiction
 - Determining Your Reading Goals
3. Choosing the Right eBook Platform
 - Popular eBook Platforms
 - Features to Look for in an Chapter 1the Human Genome Inquiry Activity
 - User-Friendly Interface
4. Exploring eBook Recommendations from Chapter 1the Human Genome Inquiry Activity
 - Personalized Recommendations
 - Chapter 1the Human Genome Inquiry Activity User Reviews and Ratings
 - Chapter 1the Human Genome Inquiry Activity and Bestseller Lists
5. Accessing Chapter 1the Human Genome Inquiry Activity Free and Paid eBooks
 - Chapter 1the Human Genome Inquiry Activity Public Domain eBooks
 - Chapter 1the Human Genome Inquiry Activity eBook Subscription Services
 - Chapter 1the Human Genome Inquiry Activity Budget-Friendly Options
6. Navigating Chapter 1the Human Genome Inquiry Activity eBook Formats

- ePub, PDF, MOBI, and More
- Chapter 1the Human Genome Inquiry Activity Compatibility with Devices
- Chapter 1the Human Genome Inquiry Activity Enhanced eBook Features
- 7. Enhancing Your Reading Experience
 - Adjustable Fonts and Text Sizes of Chapter 1the Human Genome Inquiry Activity
 - Highlighting and Note-Taking Chapter 1the Human Genome Inquiry Activity
 - Interactive Elements Chapter 1the Human Genome Inquiry Activity
- 8. Staying Engaged with Chapter 1the Human Genome Inquiry Activity
 - Joining Online Reading Communities
 - Participating in Virtual Book Clubs
 - Following Authors and Publishers Chapter 1the Human Genome Inquiry Activity
- 9. Balancing eBooks and Physical Books Chapter 1the Human Genome Inquiry Activity
 - Benefits of a Digital Library
 - Creating a Diverse Reading Collection Chapter 1the Human Genome Inquiry Activity
- 10. Overcoming Reading Challenges
 - Dealing with Digital Eye Strain
 - Minimizing Distractions
 - Managing Screen Time
- 11. Cultivating a Reading Routine Chapter 1the Human Genome Inquiry Activity
 - Setting Reading Goals Chapter 1the Human Genome Inquiry Activity
 - Carving Out Dedicated Reading Time
- 12. Sourcing Reliable Information of Chapter 1the Human Genome Inquiry Activity
 - Fact-Checking eBook Content of Chapter 1the Human Genome Inquiry Activity
 - Distinguishing Credible Sources
- 13. Promoting Lifelong Learning
 - Utilizing eBooks for Skill Development
 - Exploring Educational eBooks
- 14. Embracing eBook Trends
 - Integration of Multimedia Elements
 - Interactive and Gamified eBooks

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