

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

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Table of Contents Basic Concepts In Medical Genetics

1. Understanding the eBook Basic Concepts In Medical Genetics
 - The Rise of Digital Reading Basic Concepts In Medical Genetics
 - Advantages of eBooks Over Traditional Books
2. Identifying Basic Concepts In Medical Genetics
 - 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 Basic Concepts In Medical Genetics
 - User-Friendly Interface
4. Exploring eBook Recommendations from Basic Concepts In Medical Genetics
 - Personalized Recommendations
 - Basic Concepts In Medical Genetics User Reviews and Ratings
 - Basic Concepts In Medical Genetics and Bestseller Lists

5. Accessing Basic Concepts In Medical Genetics Free and Paid eBooks
 - Basic Concepts In Medical Genetics Public Domain eBooks
 - Basic Concepts In Medical Genetics eBook Subscription Services
 - Basic Concepts In Medical Genetics Budget-Friendly Options
6. Navigating Basic Concepts In Medical Genetics eBook Formats
 - ePub, PDF, MOBI, and More
 - Basic Concepts In Medical Genetics Compatibility with Devices
 - Basic Concepts In Medical Genetics Enhanced eBook Features
7. Enhancing Your Reading Experience
 - Adjustable Fonts and Text Sizes of Basic Concepts In Medical Genetics
 - Highlighting and Note-Taking Basic Concepts In Medical Genetics
 - Interactive Elements Basic Concepts In Medical Genetics
8. Staying Engaged with Basic Concepts In Medical Genetics
 - Joining Online Reading Communities
 - Participating in Virtual Book Clubs
 - Following Authors and Publishers Basic Concepts In Medical Genetics
9. Balancing eBooks and Physical Books Basic Concepts In Medical Genetics
 - Benefits of a Digital Library
 - Creating a Diverse Reading Collection Basic Concepts In Medical Genetics
10. Overcoming Reading Challenges
 - Dealing with Digital Eye Strain
 - Minimizing Distractions
 - Managing Screen Time
11. Cultivating a Reading Routine Basic Concepts In Medical Genetics
 - Setting Reading Goals Basic Concepts In Medical Genetics
 - Carving Out Dedicated Reading Time
12. Sourcing Reliable Information of Basic Concepts In Medical Genetics
 - Fact-Checking eBook Content of Basic Concepts In Medical Genetics
 - 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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