

Study Guide**CHAPTER 11****Section 1: Basic Patterns of Human Inheritance**

In your textbook, read about basic patterns of human inheritance.

Use the terms below to complete the passage. These terms may be used more than once.

albinism
heterozygous

alleles
homozygous

cystic fibrosis
pedigree

dominant
recessive

A (1) **PEDIGREE** shows the inheritance of a particular trait over several generations. An organism with two of the same (2) **ALLELES** for a particular trait is said to be (3) **HOMOZYGOUS** For that trait. An organism with two different (4) **ALLELES** for a particular trait is heterozygous for that trait. When alleles are present in the (5) **HETEROZYGOUS** state, the (6) **DOMINANT** _____ trait will be observable. An individual who is heterozygous for a (7) **RECESSIVE** disorder is called a carrier. Examples of recessive genetic disorders in humans are (8) **CYSTIC FIBROSIS** and (9) **ALBINISM**.

In your textbook, read about recessive and dominant genetic disorders.

Complete the table by writing the disease name for each description.

albinism
galactosemia

achondroplasia
Huntington's disease

cystic fibrosis
Tay-Sachs disease

Disease	Description
10. ALBINISM	caused by altered genes; results in lack of skin pigment
11. GALACTOSEMIA	recessive genetic disorder; characterized by body's inability to tolerate galactose
12. TAY-SACHS DISEASE	recessive genetic disorder; gene found on chromosome 15; characterized by lack of enzyme that breaks down fatty acids
13. CYSTIC FIBROSIS	recessive genetic disorder; affects mucus-producing glands, digestive enzymes, sweat glands
14. HUNTINGTON'S DISEASE	dominant genetic disorder; affects the nervous system
15. ACHONDROPLASIA	autosomal dominant genetic condition; affects height and body size

Inheritance Patterns And Human Genetics Study Guide

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common and uncommon A complete understanding remains elusive however geneticists are still refining theories about what causes chromosomes to mutate and genetic diseases remain difficult to diagnose and challenging to treat This fascinating reference explores the scientific and human aspects of this complex field of science Encyclopedia of Human Genetics and Disease features nearly 400 entries including well known genetic diseases rare and lesser known genetic diseases and the genetic factors that may contribute to common diseases and health conditions such as breast cancer and obesity The author presents in depth discussions of concepts essential to understanding genetic disease in 18 entries that provide background on key topics such as Genetics 101 the genome and the foundations of genetics genetic counseling and newborn screening Each of the 355 disorders profiled provides the history of the condition its prevalence causes treatment if any and further reading Interesting sidebars and compelling photos that help inform content accompany many entries

Life Histories of Genetic Disease Andrew J. Hogan, 2021-04-27 A richly detailed history that uncovers the challenges and limitations of our increasing reliance on genetic data in medical decision making Shobita Parthasarathy author of Building Genetic Medicine Medical geneticists began mapping the chromosomal infrastructure piece by piece in the 1970s by focusing on what was known about individual genetic disorders Five decades later their infrastructure had become an edifice for prevention allowing expectant parents to test prenatally for hundreds of disease specific mutations using powerful genetic testing platforms In this book Andrew J Hogan explores how various diseases were made genetic after 1960 with the long term aim of treating and curing them using gene therapy In the process he explains these disorders were located in the human genome and became targets for prenatal prevention while the ongoing promise of gene therapy remained on the distant horizon In narrating the history of research that contributed to diagnostic genetic medicine Hogan describes the expanding scope of prenatal diagnosis and prevention He draws on case studies of Prader Willi fragile X DiGeorge and velo cardio facial syndromes to illustrate that almost all testing in medical genetics is inseparable from the larger and increasingly big data oriented aims of biomedical research Hogan also reveals how contemporary genetic testing infrastructure reflects an intense collaboration among cytogeneticists molecular biologists and doctors specializing in human malformation Hogan critiques the modern ideology of genetic prevention which suggests all pregnancies are at risk for genetic disease and should be subject to extensive genomic screening He examines the dilemmas and ethics of the use of prenatal diagnostic information in an era when medical geneticists and biotechnology companies offer whole genome prenatal screening essentially searching for any disease causing mutation Hogan s analysis is animated by ongoing scientific and scholarly debates about the extent to which the preventive focus in contemporary medical genetics resembles the aims of earlier eugenicists Written for historians sociologists and anthropologists of science and medicine as well as bioethics scholars physicians geneticists and families affected by genetic conditions Life Histories of Genetic Disease is a profound exploration of the scientific culture surrounding malformation and mutation

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