

Chapter 7 Extending Mendelian Genetics

Section 7.1 Chromosomes and Phenotypes

- **The chromosomes on which genes are located can affect the expression of traits.**
- I. Two copies of each *autosomal gene* affect phenotype
 - * Mendel studied autosomal gene traits, like flower color, seed shape, and plant height.
- II. Mendel's *rules of inheritance* apply to autosomal genetic disorders.
 1. Disorders caused by *dominant alleles are uncommon*

 2. Most mutations are *recessive*.
 - A heterozygous individual *would not express the condition*.
 - A *heterozygote* for a recessive disorder *is a carrier*.
 - It is unlikely that a person, who is heterozygous for a recessive mutation, will *mate with someone who is heterozygous* for the same mutation.
 - Therefore, it is *less likely for the most* recessive mutations to show up in an individual.

III. Simple Recessive Heredity

A. Mutations are Changes in Genes

1. Mutation = *a change in a gene, caused by damage or an error in copying.*

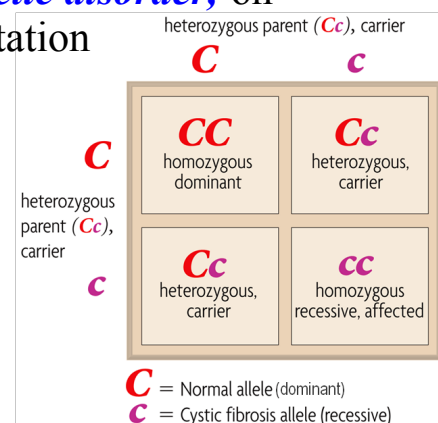
- Mutations are a source of *variation* a species needs in order to adapt to changing conditions and thus evolve over time.

B. Genetic Disorders

- Genetic disorders = *the harmful effects that some mutations produce.*

1. Cystic Fibrosis

- Cystic fibrosis is a *recessive genetic disorder*, on chromosome #7, caused by a mutation in a gene that codes for a **protein responsible for transporting chloride ions.**

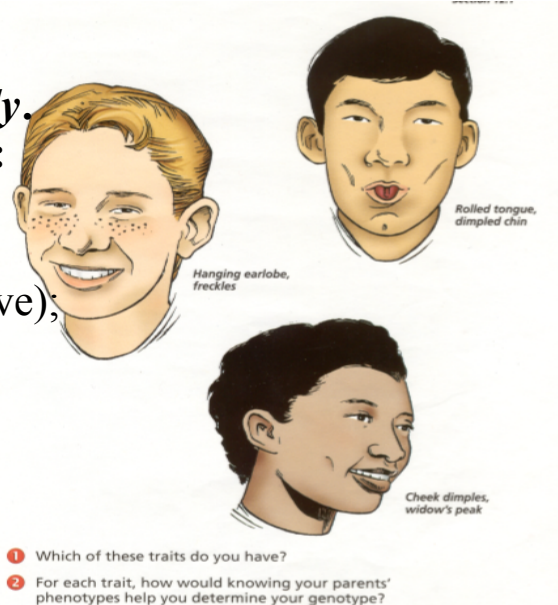


IV. Simple Dominant Heredity

1. Most dominant traits not *deadly*.

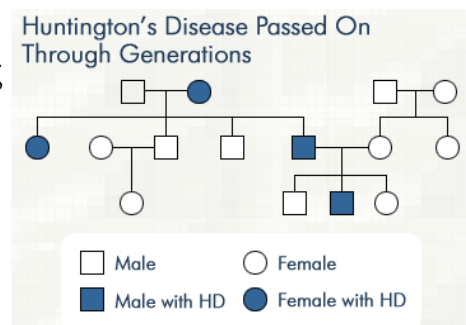
A. *Simple dominant* traits include:

- Cleft chin; Widow's peak;
- Hitchhiker's thumb;
- Almond shaped eyes (round recessive);
- Thick lips (thin recessive)
- Earlobe type:
- free hanging - dominant;
- attached earlobes - recessive



B. Huntington's Disease

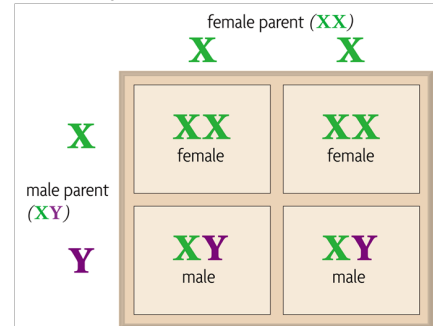
- Is a *lethal* genetic disorder caused by a rare dominant allele. Result in *breakdown certain areas of the brain*. No effective treatment known.
- Onset of disorder doesn't occur until individual affected is in their *30s to 50s*, so an individual could have children unknowing they had disorder.
- There is a test to determine carriers; however, some individuals choose not to know.



V. Males and females can differ in sex-linked traits

A. Genes on *sex chromosomes are called sex-linked genes*.

- *Y* chromosome genes in mammals are responsible for male characteristics.
- *X* chromosome genes in mammals affect many traits.



B. Some Traits/Disorders are Sex Linked

-*Sex linked* = Characteristics carried on the X-sex chromosome.

1. In males, the part that is *missing* is what balances a recessive gene

a. All of a male's sex-linked genes are *expressed*.

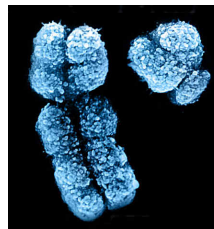
Males have no second copies of sex-linked genes.

EXAMPLES: red-green color-blindness, hemophilia, baldness, speech disorders

b. Because a female has 2 X chromosomes *she must have 2 recessive alleles to show the trait/disorder*.

Symbols used for Sex Linked Crosses

Female = X^cX^c



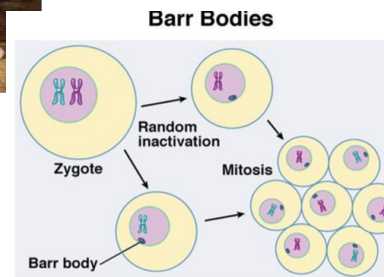
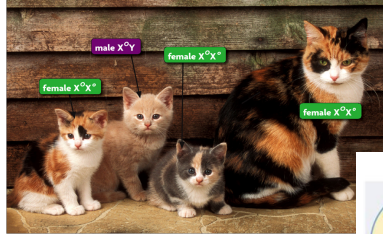
Male = X^cY

2. Female mammals have an XX genotype.
 - a. Expression of sex-linked genes is similar to *autosomal genes* in females.
 - b. In mammals, female "*X-inactivation*" may occur, in which a gene is randomly "turned off" in one of the X chromosomes.

EX: Calico cat coloring

ADD TO NOTES:

A Barr body (named after discoverer Murray **Barr**) *is the inactive X chromosome in a female somatic cell.*



Section 7.2 When Heredity Follows Different Rules

A. Complex Patterns of Inheritance

- Patterns of inheritance by simple dominant and recessive alleles are explained by Mendelian rules. However, some are more complex than simple rules.

1. **Two traits appear to blend in *incomplete dominance*.**
Incomplete dominance = when an individual has an *intermediate form*, or a *blending of two factors*, which are displayed by the two parents.



Green beta



Steel blue







Royal Blue

2. Two traits are fully displayed in codominance.

Codominance = *when both genes in a heterozygote are fully expressed.*

- Codominant alleles are *neither dominant nor recessive*.
- The *AB blood types* result from codominant alleles.
- Blood types are also an example *multiple alleles*, since there are three alleles available for the two allele expression.

PHENOTYPE (BLOOD TYPE)		GENOTYPES
A	antigen A 	$I^A I^A$ or $I^A i$
B	 antigen B	$I^B I^B$ or $I^B i$
AB	both antigens 	$I^A I^B$
O	no antigens 	ii

3. Some traits are controlled by many genes.

-Polygenic = *a trait that is controlled by more than one pair of genes.*

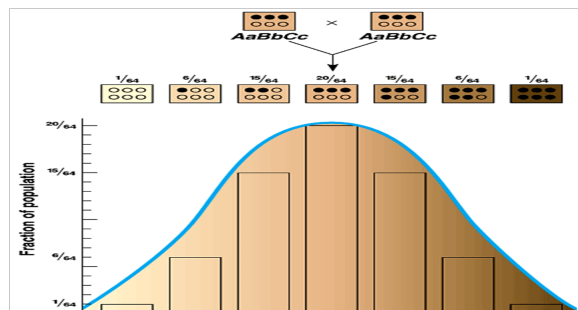
The genes may be scattered along the same chromosome or located on different chromosomes.

*For example: human height, weight, body build, hair, and skin color.



Order of dominance:
brown > green > blue.

GENE NAME	DOMINANT ALLELE	RECESSIVE ALLELE
BEY1	brown	blue
BEY2	brown	blue
GEY	green	blue



4. An *epistatic* gene can interfere with other genes.

- Certain genes determine whether or not a trait will be expressed.

-The system of genes that determines skin color in mammals, for example, is independent of the gene responsible for albinism (lack of pigment).

-This gene is an *epistatic* gene.

When the albino condition occurs, the *genes that determine skin color are present but not expressed*.

**5. Phenotype is a combination of genes and environment.**

1. Gene expression can be affected by *external* environmental conditions.

An individual's phenotype often depends on surrounding environments.

*For example, during the *warm* temperatures, sea turtles eggs will develop into *female* eggs and in *cooler* temperatures they develop into *male* eggs.

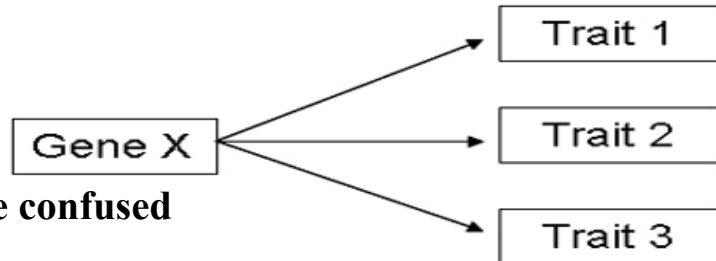
2. *Internal* environment conditions include differences in *hormones and nutrition*.

EX: Twins during fetal development.

When one gene affects many traits -

-*Pleiotropy* occurs when *one gene influences multiple traits*.

Example: Phenylketonuria (PKU), which is a human disease that affects multiple systems but is caused by one gene defect. If left untreated, the **build up of phenylalanine in the central nervous system it leads to mental retardation, eczema, and pigment defects that make affected individuals lighter skinned.**



Pleiotropy should not be confused with polygenic traits,

in which multiple genes converge to result in a single phenotype.