Human Inheritance

Big Picture

The human genome is passed on from generation to generation, from parents to offspring. Human inheritance is governed by Mendelian inheritance and non-Mendelian inheritance patterns. Although many beneficial and necessary genes are passed on, so are mutated genes that cause genetic disorders, such sickle-cell anemia. One goal of biotechnology is to use genetic material itself to treat these disorders, such as in gene therapy.

Key Terms

Gene: Segment of DNA that codes for a single protein or RNA. Controls what characteristics are expressed.

Trait: A variation of a characteristic. For example, hair is a characteristic and brown hair is a trait.

Allele: Variant of a specific gene. Codes for different traits.

X-Linked Trait: Coded for by a gene on an X sex chromosome.

Pedigree: Charts familial relationships and the inheritance of a specific trait.

Genetic Disorder: A genetic disorder may be caused by a mutation at the gene level or by an unusual number of chromosomes.

Single-Gene Disorder: Disorder caused by the mutation of a single gene.

Chromosomal Disorder: Disorder caused by an unusual number of chromosomes.

Nondisjunction: Failure of replicated chromosomes to separate.

Mendelian Inheritance

The Mendelian pattern of inheritance assumes that **traits** are controlled by only one **gene** and that each gene has only two **alleles**.

Examples of traits that exhibit the Mendelian pattern of inheritance:

- Autosomal trait: Coded for by a gene on an autosome.
- Sex-linked trait: Coded for by a gene on a sex chromosome (X or Y).
  - **X-linked traits** are more prevalent in men than in women. Men will express any X-linked recessive trait they inherit because they only have one X chromosome.
- Sex chromosome inheritance: The sex determinant chromosome always comes from the father. The mother's gamete always donates the X chromosome. The father's gamete donates another X chromosome if the child is a girl and a Y chromosome if the child is a boy.

Diseases caused by Mendelian traits, or traits that exhibit the Mendelian pattern of inheritance, include sickle-cell anemia and cystic fibrosis.

Pedigrees

Pedigrees are useful for examining how traits are passed from generation to generation. The example below shows how an autosomal recessive trait is passed through a family.

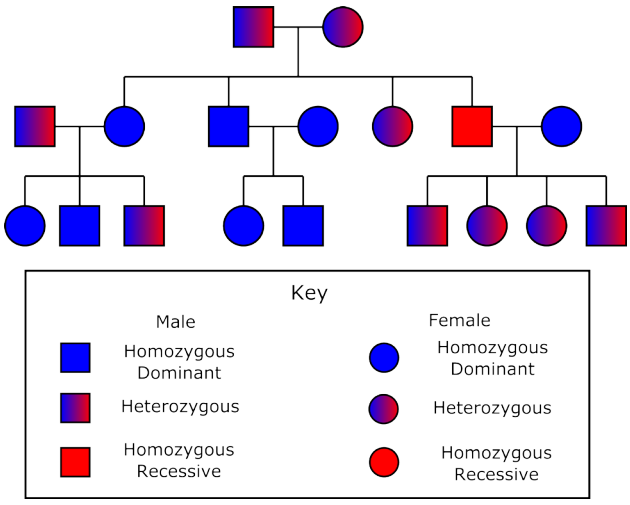


Image Credit: CK-12 Foundation, CC-BY-NC-SA 3.0

Notes

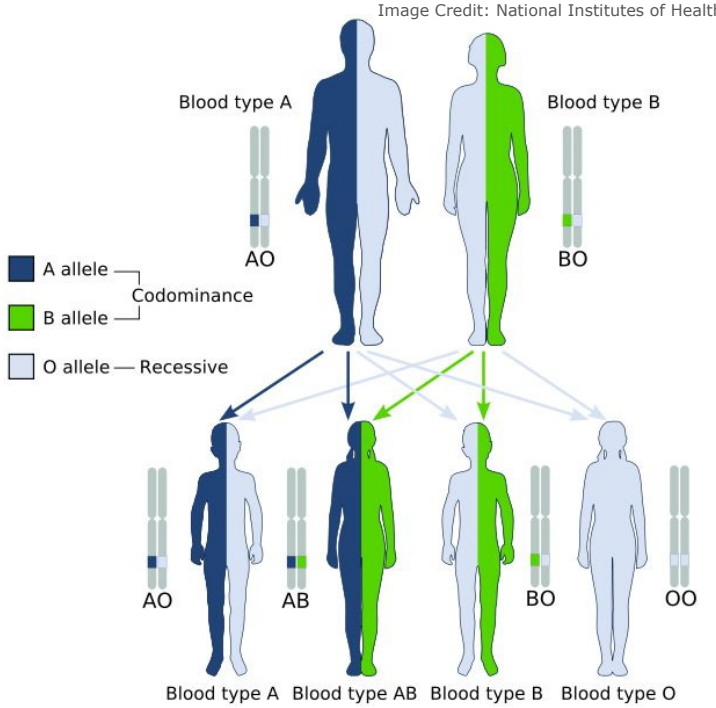
Human Inheritance cont.

Non-Mendelian Inheritance

Non-Mendelian inheritance is a more complex pattern of inheritance, involving multiple genes, multiple alleles, and multiple traits.

- Multiple allele trait: Traits coded by genes that have more than two alleles. For example, the gene that codes for blood type has three alleles.

Image Credit: National Institutes of Health, Public Domain



- Polygenic trait: Trait coded by multiple genes. For example, height and skin color are coded by genes that together have an additive effect.
- Pleiotropy: When one gene affects multiple traits.
- Epistasis: When the expression of one gene is determined by another gene. For example, in some mice, one gene controls fur color and another gene determines whether or not pigment is deposited.

Genetic Disorders

Genetic disorders can be the result of mutations at the gene level or by unusual number of chromosomes.

Single Gene Disorder

Below are some various classifications of **single gene disorders**:

- Autosomal dominant: An individual only needs to receive one copy of the mutated gene to express the disorder.
- Autosomal recessive: An individual needs to receive two copies of the mutated gene to express the disorder. An individual is a carrier if there is only one copy of the mutated gene.
- X-linked dominant: An individual, regardless of sex, needs only to receive one copy of the mutated gene to express the disorder.
- X-linked recessive: A man only needs to receive one copy of mutated gene, while a woman needs to receive two to express the disorder.
- Y-linked: A man needs only to received one copy of the mutated gene to express the disorder.

Chromosomal Disorder

Chromosomal disorders are caused by problems at the chromosome level.

- **Nondisjunction** is one cause of chromosomal disorder, causing some gametes to have extra chromosomes and some to have too few.
  - For example, Down syndrome is caused by the presence of three copies of chromosome 21.

Diagnosis and Treatment

One way parents who have recessive genetic disorders check for the presence of the disorder in their fetus is by prenatal testing.

- One form of prenatal testing is amniocentesis, which removes a sample of the amniotic fluid surrounding the fetus.

One possible way to treat genetic disorders is by gene therapy. Gene therapy uses a vector, usually a virus, to insert or express a functional gene in target cells. The gene will counter the harmful effects of the mutated gene(s).