Single Trait Inheritance in Humans

(Mendelian Inheritance)

- Most Mendelian conditions affect 1 in 10,000 or fewer individuals.
- Examples of some Mendelian disorders in humans:
  - Cystic fibrosis
  - Maple syrup urine disease
  - Phenylketonuria (PKU)
  - Sickle cell anemia
  - Tay sachs disease
  - Huntington’s disease
  - Familial hypercholesterolemia
  - Familial Hypertrophic Cardiomyopathy
  - Polydactyly
  - Achondroplasia

Modes of Inheritance

- Rules that explain the common patterns that inherited characteristics follow as they are passed through families.
- Autosomal Dominant and Autosomal Recessive are the two modes of inheritance directly derived from Mendel's laws.
- An **autosome** is a non-sex chromosome (ex. Chromosomes #1-22 in humans)

Autosomal Dominant Inheritance

- Can appear in either sex
- If a child has the trait, at least one parent must also have it
- Does NOT skip a generation
- If one parent has an autosomal dominant trait and the other does not, each offspring has a 50% chance of inheriting the disorder.

Autosomal Recessive Inheritance

- Can also appear in either sex.
- Homozygous recessive genotype (ex. aa)
- Heterozygotes are known as “carriers” because the mutant allele is masked by a dominant allele
- Blood relatives who have children together have a higher risk of having children with an autosomal recessive condition (consanguinity)
- When both parents are carriers, offspring has a 25% chance of inheriting the disorder.

- cracking the code
- PBS NOVA

- Tay Sachs- 16:15
- Cystic Fibrosis- 58:28