

Concept 12.3

Mendel's principles apply to humans.

Working With Human Pedigrees:

Pedigree - a family tree that records and traces the occurrence of a trait in a family.

A typical pedigree

1. Uses squares to represent males and circles to represent females.
2. The colored shapes represent individuals that show the trait.
3. Parents are connected by horizontal lines, with the children beneath them.
4. The genotype of most of the individuals in a pedigree can be determined by examining the pattern in which the trait occurs.

Disorders Inherited as Recessive Traits:

Most human genetic disorders are recessive.

Recessive disorders range in severity.

Albinism to Tay-Sachs disease (lethal)

Carrier - An individual who has one copy of the allele for a recessive disorder and does not exhibit symptoms.

Disorders Inherited as Dominant Traits:

A smaller number of human disorders are inherited as dominant traits.

e.g. - Extra fingers and toes

- Dwarfism (achondroplasia) All individuals with this disorder are heterozygous.

Inheriting two copies of the allele is fatal.

More than 99.9% of the population is homozygous for the normal, recessive allele.

Dominant alleles that are lethal are much less common than lethal recessive.

Sex-linked Disorders:

Sex-linked alleles are those located on one sex chromosome but not on the other.

In humans, most sex-linked alleles are located on the X chromosome.

A male therefore needs only one copy of a sex-linked recessive allele to exhibit the recessive trait.

Predicting and Treating Genetic Disorders:

Genetic counselor - a person trained to collect and analyze data about inheritance patterns and to explain the results and their significance.

Who needs to see a Genetic counselor?

- people who are considering having children, and one or both has a family history of a genetic disease.

What a Genetic counselor does.

- Examines the couple's family histories for any disorders that have Mendelian inheritance patterns.
- Interpret genetic tests performed on the parents to detect disorder alleles.
- Help couples determine the risk of passing on a disease trait to their child.