

Review of Genetics

Genes

Segments of DNA that code for a specific trait.
Variations of a gene called alleles.

Classification of genes:

Dominant vs. Recessive

Homozygous vs. Heterozygous (diploid cond.)

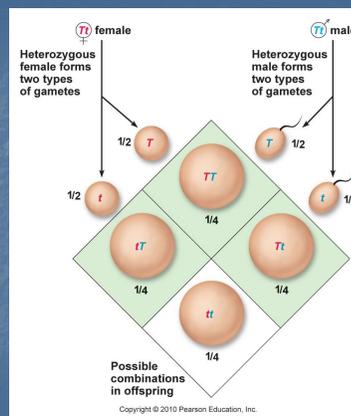
Punnett Square Example

The characteristic of freckles is considered to be dominant and the symbol is "F."

A couple are wondering if their children will have freckles. The man has freckles but his mother did not have them.

The woman in this couple has no freckles.

What percentage of the children will have freckles?



Incidence of Genetic Diseases

There are approximately
1300 recessive genetic diseases

BUT

There are also ~ 1300 dominant genetic diseases.

Examples: Huntington's, dwarfism,
Syndactyly (webbed digits)

Tests for Identifying Carriers

Exist for the following diseases:

- > Tay-Sachs
- > Sickle Cell
- > Cystic Fibrosis

Sex Linked Genes

Genes that are on one sex chromosome but not on the other.

The Y chromosome carries about 78 genes (2003).

The X chromosome carries about 1000+ genes.

Code for traits important in both sexes e.g. color vision, blood clotting and structural proteins.

Sex linked Punnett Square

Color blindness is an X-linked trait.

XC represents a normal allele.

"Xc" represents the recessive X linked allele associated with color-blindness

A couple are planning a family and the father has normal color vision but the mother is heterozygous for color-blindness (i.e. she is a carrier).

What percentage of the boys from this union will be expected to be color blind?

Methods used in Genetic Counseling

Punnett Squares

Carrier recognition:

- Pedigrees
- Blood typing

Fetal testing/Karyotyping

- > Amniocentesis
- > Chorionic villi sampling (can be done earlier)

