Sex-Linked Traits, Karyotyping & Pedigrees
**Karyotype** - a photograph of chromosomes grouped in order in pairs
Human Karyotypes

- Two of the 46 human chromosomes = sex chromosomes
  - Determine an individual's sex (male/female)
    - Females have two copies of an X chromosome.
    - Males have one X and one Y chromosome.

- The remaining 44 chromosomes = autosomes

Female genotype: _______
Male genotype: _______

Hint: What is the sex of this individual?
Karyotyping Lab
Pedigree Charts

pedigree - a chart that shows the relationships within a family. (how one trait is passed from one generation to the next)

Genetic counselors analyze pedigree charts to infer the genotypes of family members.
Pedigrees

A horizontal line connecting a male and a female represents a marriage.

A circle represents a female.

A square represents a male.

A vertical line and a bracket connect the parents to their children.

A shaded circle or square indicates that a person expresses the trait.

A circle or square that is not shaded indicates that a person does not express the trait.
Pedigree Practice!
Pedigree Worksheet - practice problems
Sex-Linked Traits

How is sex determined?

All human egg cells carry a single X chromosome (23,X).

Half of all sperm cells carry an X chromosome (23,X) and half carry a Y chromosome (23,Y).

About half of the zygotes will be 46,XX (female) and half will be 46,XY (male).

Who determines sex, the male or female?
Sex-Linked Traits

• Traits that are determined by genes on the sex chromosomes (100+ disorders linked to X chromosome)

• Carrier – female that carries a gene for a disorder but does not exhibit the disorder
  - Her XX genotype may have one X that is normal and the other X<sup>c</sup> that carries the gene for the disorder
    • Example: color blindness – recessive trait - an individual cannot distinguish between 2 colors.
      - Carrier genotype = XX<sup>c</sup>
      - Disorder genotype = X<sup>c</sup>X<sup>c</sup>

• Easy for males to get this - they only have 1 X chromosome
The Y chromosome is much smaller than the X chromosome and appears to contain only a few genes.
Sex-Linked Genes

Why are sex-linked disorders more common in males than in females?

Females = XX so both must be recessive
Males = XY so if the X has recessive allele, then the disorder will show in the phenotype.
Sex-Linked Genes

Colorblindness

Three human genes associated with color vision are located on the X chromosome.

In males, a defective version of any one of these genes produces colorblindness.
Possible Inheritance of Colorblindness Allele

- **Father (normal vision):** $X^C_Y$
  - **Daughter (normal vision):** $X^C_C$
  - **Son (normal vision):** $X^C_Y$
- **Mother (carrier):** $X^C_X^c$
  - **Daughter (carrier):** $X^C_X^c$
  - **Son (colorblind):** $X^C_C$

Legend:
- Blue square: Colorblind
- White circle: Normal vision
Ishihara Color Blindness Test

Red-Green Color Blind

25

45

29

56
Ishihara Color Blindness Test

- Normal color vision:

- Red/Green color blindness:
Sex-Linked Genes

Hemophilia

The X chromosome also carries genes that help control blood clotting. A recessive allele in either of these two genes may produce hemophilia. In hemophilia, a protein necessary for normal blood clotting is missing. Hemophiliacs can bleed to death from cuts and may suffer internal bleeding if bruised.
Sex-Linked Genes

Duchenne Muscular Dystrophy

Duchenne muscular dystrophy is a sex-linked disorder that results in the weakening and loss of skeletal muscle. It is caused by a defective version of the gene that codes for a muscle protein.
nondisjunction - (“not coming apart”) a common error in meiosis occurs when homologous chromosomes fail to separate
Chromosomal Disorders

Down Syndrome

If two copies of an autosomal chromosome fail to separate during meiosis, an individual may be born with three copies of a chromosome.

Down syndrome involves three copies of chromosome 21.
Chromosomal Disorders

Down syndrome produces mild to severe mental retardation.

characterized by:
• increased susceptibility to many diseases
• higher frequency of some birth defects
Chromosomal Disorders

Sex Chromosome Disorders

In females, nondisjunction can lead to Turner’s syndrome.

A female with Turner’s syndrome usually inherits only one X chromosome (karyotype 45,X). Women with Turner’s syndrome are sterile.

In males, nondisjunction causes Klinefelter’s syndrome (karyotype 47,XXY).

The extra X chromosome interferes with meiosis and usually prevents these individuals from reproducing.
Sex-Linked Practice

1. Hemophilia is a sex linked recessive trait that keeps blood from clotting properly. If a woman who has normal blood-clotting but is a carrier for hemophilia marries a man who is a hemophiliac, what are the chances their daughters will be hemophiliacs like their father?

A. Parental genotypes:
   mom = ______
   dad = ______
B. Punnett Square

   ______
   ______
   ______
   ______
2. Red/Green colorblindness is a sex linked trait. If a man with normal color vision marries a colorblind woman, will all of the offspring be colorblind? If not, who won't be colorblind (sons or daughters)?

A. Parental genotypes:
   - mom = _________
   - dad = _________

B. Punnett Square
Autosomal Disorders

Recessive Alleles

Many disorders are caused by autosomal recessive alleles.

<table>
<thead>
<tr>
<th>Disorder</th>
<th>Major Symptoms</th>
</tr>
</thead>
<tbody>
<tr>
<td>Albinism</td>
<td>Lack of pigment in skin, hair, and eyes</td>
</tr>
<tr>
<td>Cystic fibrosis</td>
<td>Excess mucus in lungs, digestive tract, liver; increased susceptibility to infections</td>
</tr>
<tr>
<td>Galactosemia</td>
<td>Accumulation of galactose (a sugar) in tissues; mental retardation; eye and liver damage</td>
</tr>
<tr>
<td>Phenylketonuria (PKU)</td>
<td>Accumulation of phenylalanine in tissues; lack of normal skin pigment; mental retardation</td>
</tr>
<tr>
<td>Tay-Sachs disease</td>
<td>Lipid accumulation in brain cells; mental deficiency; blindness; death in early childhood</td>
</tr>
</tbody>
</table>
Autosomal Disorders

Dominant Alleles

Two examples of genetic disorders caused by autosomal dominant alleles are achondroplasia and Huntington disease.

<table>
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<tr>
<th>Disorder</th>
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</thead>
<tbody>
<tr>
<td>Achondroplasia</td>
<td>Dwarfism (one form)</td>
</tr>
<tr>
<td>Huntington disease</td>
<td>Mental deterioration and uncontrollable movements; symptoms usually appear in middle age</td>
</tr>
<tr>
<td>Hypercholesterolemia</td>
<td>Excess cholesterol in blood; heart disease</td>
</tr>
</tbody>
</table>
Autosomal Disorders

Codominant Alleles

Sickle cell disease is a serious disorder caused by a codominant allele.

Sickle cell is found in about 1 out of 500 African Americans.

<table>
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<th>Disorders Caused by Codominant Alleles</th>
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<td>Disorder</td>
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<tr>
<td>Sickle cell disease</td>
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Mutations

How do small changes in DNA cause genetic disorders?

In both cystic fibrosis and sickle cell disease, a small change in the DNA of a single gene affects the structure of a protein, causing a serious genetic disorder.
Cystic Fibrosis

- caused by recessive allele

- symptoms: a thick, heavy mucus that clogs the lungs and breathing passageways

The cells in the person’s airways are unable to transport chloride ions. As a result, the airways become clogged with a thick mucus.
Sickle Cell Disease

- a common genetic disorder found in African Americans

- characterized by the bent and twisted shape of the red blood cells

- caused by mutation which makes it difficult for hemoglobin to carry oxygen in the blood
Sickle Cell Anemia

People who are heterozygous for the sickle cell allele are generally healthy and they are resistant to malaria.

Malaria and the Sickle Cell Allele

Regions where malaria is common

Regions where the sickle cell allele is common