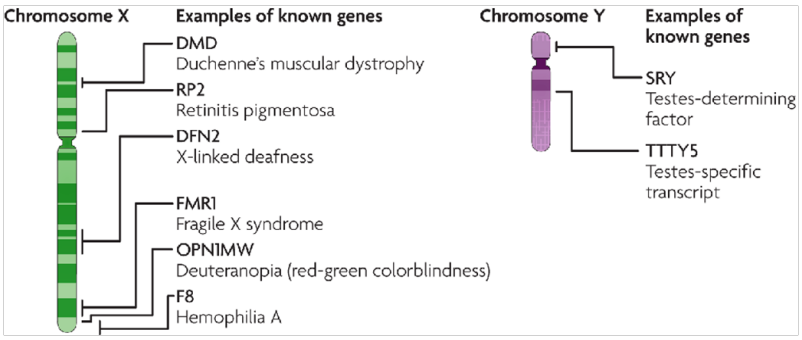
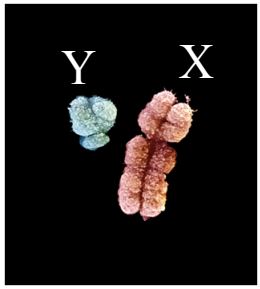
Section 7.4 Human Genetics & Pedigrees

I. Human genetics follows the patterns seen in other organisms.

- The ***basic principles*** of genetics are the same in all sexually reproducing organisms.
- Inheritance of many human traits is ***complex***.
- ***Single-gene traits*** are important in understanding human genetics.

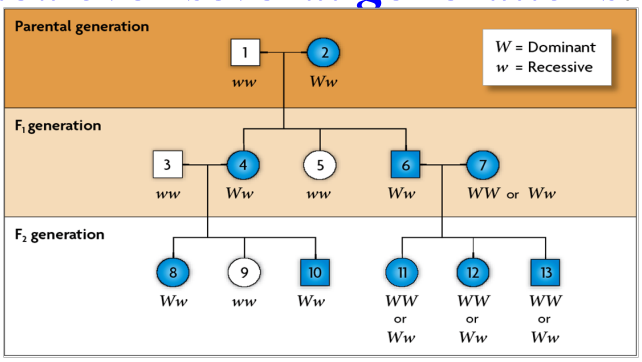
- **Females can carry *sex-linked* genetic disorders.**
  - Males (***XY***) ***express*** all of their sex linked genes.
  - ***Expression*** of the disorder depends on which parent carries the allele and the sex of the child.



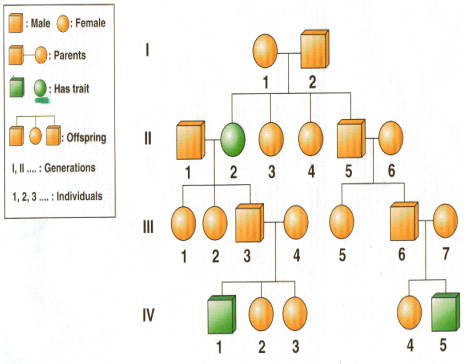


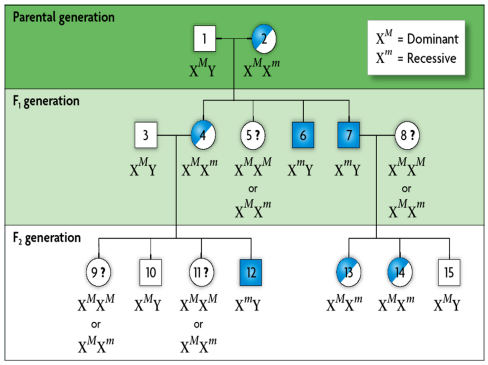
II. Pedigrees illustrate inheritance.

A. Pedigree = graphic representation of genetic inheritance. A diagram made up of a set of symbols that identify male and female relationships and ***individuals affected by the trait being studied over several generations.***



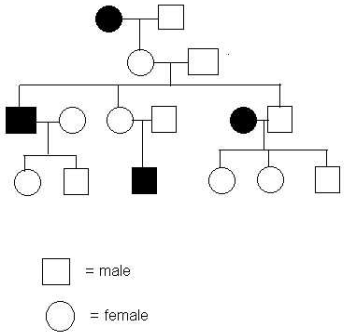
- **A pedigree is a chart for tracing genes in a family.**
  - ***Phenotypes*** are used to infer genotypes on a pedigree.
  - ***Autosomal*** genes show different patterns on a pedigree than ***sex-linked*** genes.
  - If the phenotype is ***more common in males***, the gene is likely sex-linked.





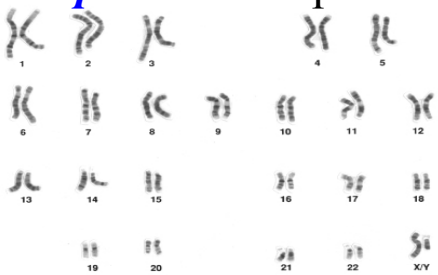
B. Analyzing a pedigree.

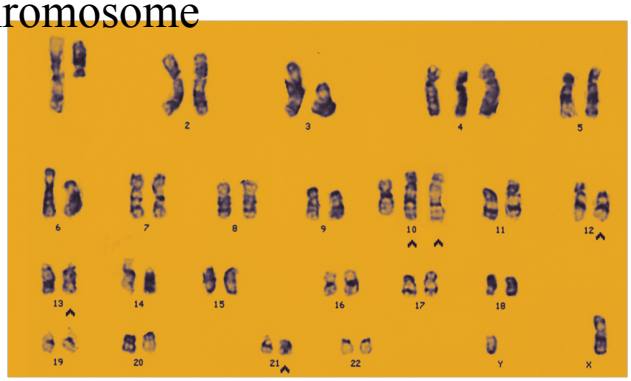
1. Physicians may ***recommend genetic counseling*** to family members. In some cases, a genetic counselor can prepare a family pedigree.



III. Karyotypes can show changes in chromosomes.

- ***deletion*** of part of a chromosome or loss of a chromosome
  - large changes in chromosomes
- extra chromosomes (***trisomy***)or loss of a chromosome (***monosomy***)
- ***duplication*** of part of a chromosome



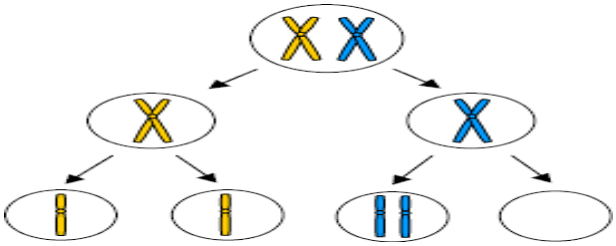


- Prenatal Diagnosis - Ways to detect ***genetic disorders*** during pregnancy. Often used if after pedigree analyzed a suspected disorder runs in a family or because of older age of female.
  - Types of prenatal diagnosis-
    An ***amniocentesis*****,** when done a ***karyotype*** is prepared and studied to see if all is normal in the embryo
  - **Chorionic villi sampling** ***(CVS)*** is an alternative to the amniocentesis.

IV. Results of Karyotypes -

- ***Nondisjunction*** **-** a type of mutation that involves whole chromosomes. When ***homologs fail to separate during meiosis*****.**

Depending on the gamete fertilized there are ***more or less*** the normal number of chromosomes present in an organisms cell.



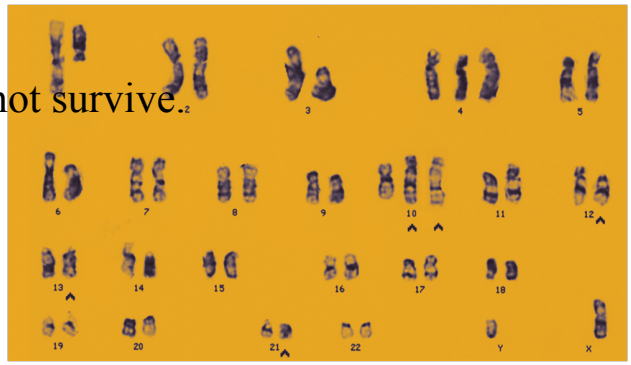
Trisomy = a condition when a gamete with an extra chromosome is fertilized by a normal gamete. The zygote will have an extra chromosome (47, not 46).

Ex: Down Syndrome, Edwards Syndrome, etc.

Monosomy = condition when a gamete with a missing chromosome fuses with a normal gamete. The zygote lacks a chromosome (45, not 46).

Ex: Turner Syndrome

Most **monosomy zygotes** do not survive.



***Polyploidy**

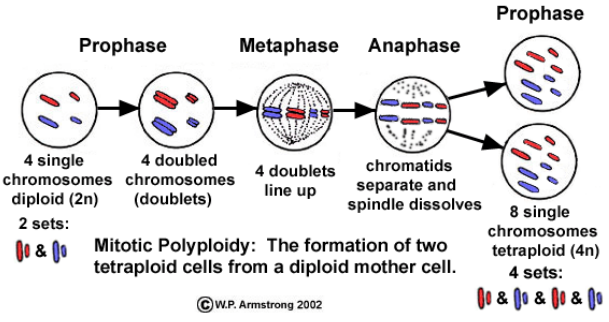
-Polyploids = organisms with more then the usual number of chromosomes.

-Polyploidy is ***rare in animals***, and almost always causes ***death***.

-It is ***common in plants***; often the flowers and fruits of these plants are ***larger*** than normal and the plants ***healthier***.

-Many polyploid plants are of great commercial value. Plant breeders inject chemicals to force ***nondisjunction*** to occur.

Ex: sterile banana plant (triploid), wheat (hexaploid), apples (triploid).



V. Complex Inheritance of HumanTraits

1. Co-dominance - Sickle Cell anemia results from faulty hemoglobin.

- It is a genetic disorder, on chromosome #11, caused by a ***mutated gene that codes from hemoglobin,*** a protein red blood cells that transports ***oxygen*** throughout the body. When the faulty hemoglobin is inside a red blood cell, it gives the cell a deformed, "***sickle***" shape appearance.
- The sickle shaped cells get caught in small blood vessels and restrict blood flow to tissues. They are also ***fragile and break down easily.*** This leaves the person with fewer red blood cells that the average person, a condition called ***anemia.***
- Symptoms include fatigue, headaches, muscle cramps, and sometimes kidney and heart failure.

2. Hemophilia results from a faulty blood-clotting protein.

- It is a ***recessive genetic disorder*** **caused by a mutation on the X chromosome (*sex-linked*).** The mutation causes the body to build a defective form of a protein needed to clot blood. Long-term sufferers are susceptible to serious arthritis, arising from bleeding in joint cavities, and other disorders. Treatments include, such as injections of the correct form of the faulty protein.

-Since hemophilia is a **recessive sex-linked trait .** It is easier for a male to inherit this disorder than a female.

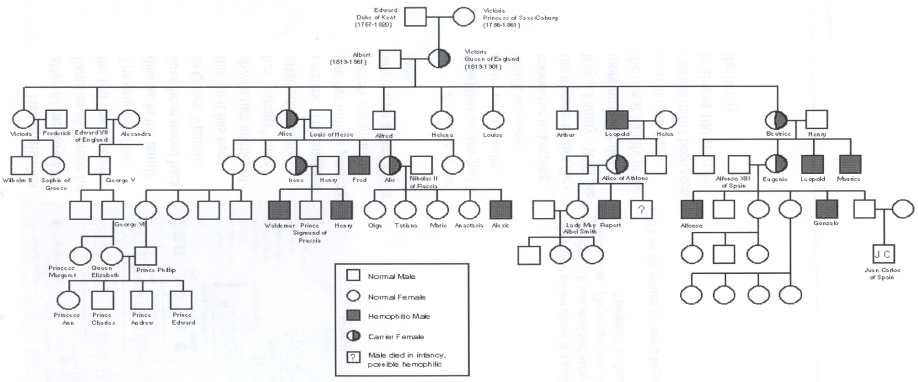
H = normal h = hemophiliac

X^HX^H = normal female X^HY = normal male

X^HX^h = normal female X^hY= hemophiliac male

X^hX^h = hemophiliac female

The European Royal Family



Hemophilia has affected the Royal Families of Europe for the last few centuries. The origins of the disease in the family have been traced back to Queen Victoria, though it has been shown that neither of her "parents" were affected or were carriers. This suggests that her mother possibly had an extramarital affair with a hemophilic man.

It used to be the trend that royalty had to marry other royalty, which was very conducive to keeping the disease within the Royal Family. With the more relaxed rules of the Royal Family in the current time, the disease hasn't been seen in almost 100 years in the family.

3. Down Syndrome

- The features that characterize Down Syndrome include extra folds in the upper eyelids, a broad and somewhat flattened nose, short stature and ***varying degrees of intellectual disabilities*.**
- This disorder occurs in about 1 out of every 1,000 births. This syndrome is much more common among children born t***older mothers*** **(over 45 years).**
- An individual receives a***third copy of chromosome #21*** b/c the chromosomes are **not properly separating during meiosis *(nondisjunction)*.**

4. Abnormal number of sex chromosomes

Many abnormalities occur with the absence or extra sex chromosome.

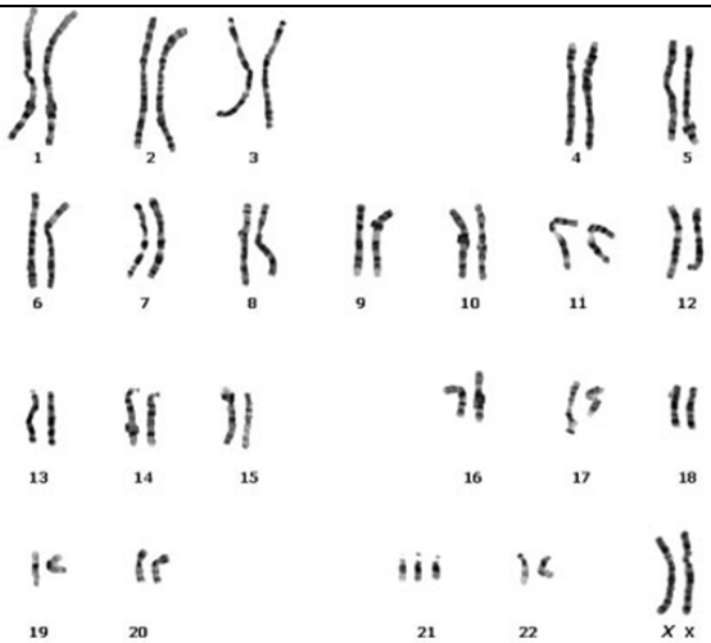
Turner's Syndrome = ***XO*** **- female**

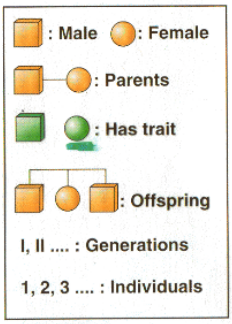
Klinefelter's Syndrome= ***XXY*** **- male**

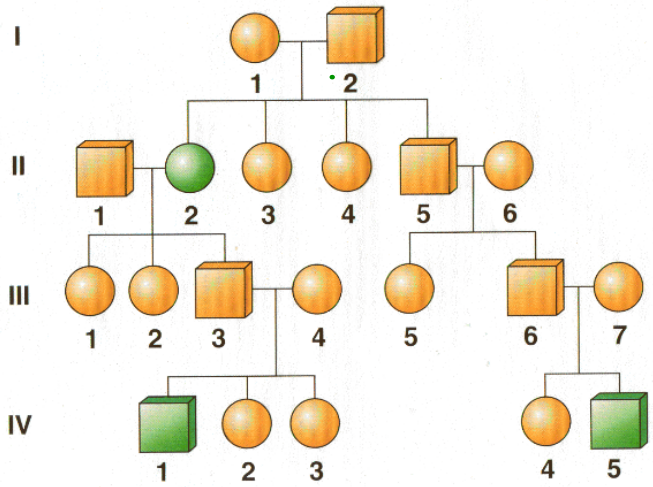
Jacob's Syndrome =***XYY*** **- male**

Triple XXX Syndrome**- female**

-Most lead productive lives, however, may be ***sterile*** or have varying ***degrees of intellectual disabilities***.



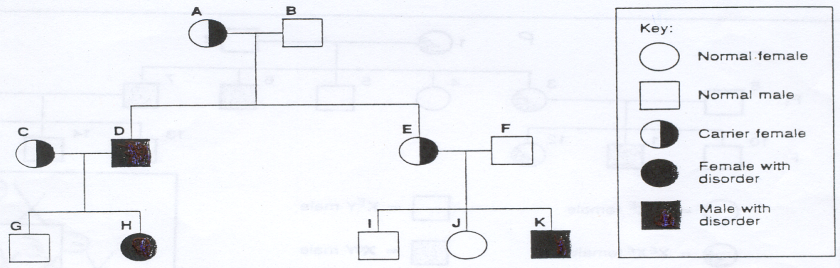




X

Applied Genetics

A. Duchenne muscular dystrophy is a deadly disorder in which the muscles grow progressively weaker. The disease is caused by a recessive gene on the X chromosome. The pedigree chart below illustrates the inheritance of this gene. Use the chart to answer the questions that follow.



1. Is Duchenne muscular dystrophy more likely to occur in males or in females? Explain your answer.

2. Individual H is a female with this disorder. Explain how she inherited this disease.

3. Individual K has this disorder, yet his father did not. Explain how this is genetically possible.

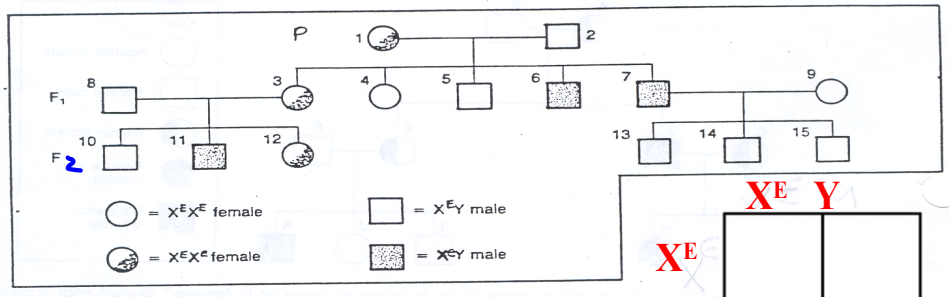
4. Individual G does not have the disease, yet his mother was a carrier and his father had the disease. Explain how this is possible.

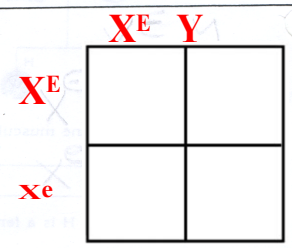
5. Why is the genotype of the father unimportant when investigating sex-linked traits inherited by male offspring?

Sex-Linked Inheritance

Sex-linked traits are characteristics carried on the X chromosome. Because human females are XX and males are XY, certain recessive sex-linked traits are expressed more commonly in males than in females. A female would need to receive the recessive allele on both of her X chromosomes to show the trait. But a male who receives the recessive allele on his X chromosome will show the trait, because his Y chromosome cannot mask the expression of the recessive allele.

- The pedigree below shows the inheritance of the recessive sex-linked trait of red-green color blindness through three generations. It depicts a cross between a female carrier ($X^E X^e$) and a normal male ($X^E Y$). (e is the recessive allele for color blindness.) Use this information to answer the questions below.





1. Complete the Punnett square at the right, which depicts the cross in the P generation.
2. What is the probability of the P female passing the allele for color blindness to her offspring? _____
3. In the above pedigree, how many female children in the F_1 generation received the allele for color blindness? _____ How many male F_1 children received the allele? _____ . How many F_1 children have normal color vision? _____
4. What are the genotypes of the F_2 offspring of the cross between 3 and 8? _____ _____ Which of those offspring are color blind and why? _____ _____
5. If 7 and 9 have a female offspring, what will be her genotype? _____