Sex determination and sex-linked inheritance
Reading Lewis Chapter 6, pp 113-132; Cartoon Guide pp 84-96

Sex determination
Male and females differ genetically by 1 chromosome pair. Human females have two X chromosomes and human males have one X and one Y chromosome. The other 22 pairs are identical.

Figure 6.6 Inheritance of a Y chromosome with a functional SRY gene determines maleness.

The X chromosome has several genes. The Y chromosome has fewer genes.

There are three classes of genes on the Y
• Genes shared with X chromosome define the pseudoautosomal regions (PAR)
• Genes similar to X chromosome genes are X-Y homologs
• Genes unique to the Y including SRY gene
Y-linked traits
- very rare
- transmitted male to male
- no females affected
most known Y-linked diseases are associated with infertility - not transmitted

A male will inherit only one allele of a gene carried on the X chromosome because he only inherits one X chromosome from Mom - that means the human male is **hemizygous** for X-linked traits - because he has only one set of genes from his Mom. Traits carried on the X are called **X-linked** and Y are called **Y-linked**.

Human male inherits an X-linked trait from his Mom only. He inherits the Y from his Dad.

**Criteria for X-linked recessive traits.** Expression varies between the sexes.
1. always expressed in the male
2. expressed in the female who has two recessive alleles
3. passed from heterozygous or homozygous recessive mother to son
4. an affected female progeny must have an affected Dad and an affected or heterozygous Mom

**Examples of a few X-linked recessive traits:**
- color blindness
- ichthyosis

*Figure 6.7 An X-linked recessive trait - Ichthyosis - the grandfather (I-1) and grandson (III-1) were affected in this family. Mother (II-1) is an carrier.*

- hemophilia A - blood clotting disorder - Royal European families
Is baldness an X-linked genetic trait? Some folks say yes - The Cartoon Guide to Genetics says the BALD gene is on the X chromosome. But Ricki Lewis, the author of Human Genetics, 7th edition textbook says pattern baldness is an autosomal trait that is affected by the levels of hormones in the body, this type of trait is called **SEX-INFLUENCED**. In this case, it appears that the level of a male hormone can affect the severity of the baldness.

Why are women normally unafflicted by these X-linked recessive diseases? Because they carry a second X chromosome that typically carries a WT copy of the defective gene.

**X-linked dominant traits**
- expressed in female even when the mutant allele is present only in one copy
- typically much more severe in males
- expressed in males who carry the mutant X-linked allele.
- A heterozygous female has a 1 in 2 chance of passing it to her offspring.
- **incontinentia pigmenti** - very severe disease - males with the condition do not survive to be born while the females have only a 75% chance of survival.
- **congenital generalized hypertrichosis** - Fig 6.10