CHAPTER 3 – SEX LINKAGE

I. When Mendel's did his pea crosses, he found that reciprocal crosses yield identical results. Pollen from round seeded plant x egg from wrinkled seed pea = pollen from wrinkled seed plant x egg from round seed plant. However, other researchers discovered that the genetic ratios that they obtained were different depending upon with strain was male or female.

II. To understand how sex affects transmission of genes, we must first understand how sex is determined in animals.

A. Early observations

1. 1891 – H. Henking had observed in meiotic nucleus of males insects, 11 chromosome pairs and a single unpaired one. He called this the X body.
2. 1905 – Edmond Wilson noted that in another species of insect (Protenor), females have 6 chromosome pairs while males have 5 pairs plus an unpaired one.
3. 1905 - Nettie Stevens noted that male and female beatles and *Drosophila* have same number of chomosomes but one pair is heteromorphic in males

    a) She designated the member of the <u>heteromorphic pair</u> in males that did not have a counterpart in females "Y ". She designated the other member of the heteromorphic pair that did have a counterpart in females "X".

    b) Since it was seen that the Y was always in the male and the two X's always in the female, it was suggested that these were the determiners of sex.

    c) Found that the heteromorphic pair synapse and segregate as if homologs, so 2 kinds of sperm made - one with an X and one with a Y.

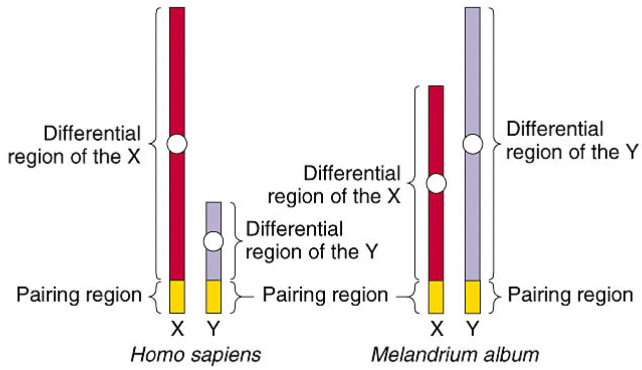
B. Humans and all other mammals show and X - Y sex determination pattern. The presence of the Y, which carries a gene that codes for TDF, testes determining factor, is important in determining maleness. In *Drosophila*, the ratio of X's to the other chromosomes that are not sex chromosomes determines maleness. For the female, there is a 1:1 ratio. That is there are 2 X's (this is designated as 1), 2 2's, 2 3's, and 2 4's. For males it is 1:2, 1 X: 2 2's:2 3's:2 4's. The presence of the Y doesn't matter as far as the phenotype of Male is concerned. It does matter as far as fertility. The Y carries a gene determines further spermatogenesis.

Table 3-1 Chromosomal Determination of Sex in *Drosophila* and Humans

	Sex chromosomes			
Species	XX	XY	XXY	XO
Drosophila	♀	♂	♀	♂
Humans	♀	♂	♂	♀

(from An Introduction to Genetic Analysis, 6th ed. By Griffiths et al. W. H. Freeman and Company)

C. X and Y have a pairing region and a differential region. The differential region carries genes that are on the X chromosome but not on the Y. Thus, genes occurring in the differential region of the X chromosome, whether recessive or dominant, will show the phenotype of the gene when it occurs in a male. These genes show an X-linkage pattern. These regions are called <u>hemizygous</u> since there is no possibility for being either homozygous or heterozygous in the heterogametic sex. In an X-Y system, this would be in the male.



(from An Introduction to Genetic Analysis, 6th ed. By Griffiths et al. W. H. Freeman and Company)

D. Heterogametic sex does not have to be male

<u>E. X-inactivation</u>

Early in development, one of the X chromosomes in each cell in females randomly becomes highly condensed and is visible as a dark staining spot called the <u>Barr body</u>. This chromosome is inactive and none of the genes on it are expressed. All of the subsequently derived cells in each cell lineage also have the same X chromosome inactivated. This results in a mosaic of cells with two different genotypes (relative to the X chromosome) in the female body.

<u>III. Historical examples of sex-linkage</u>

A. 1906 – L. Doncaster and G. H. Raynor crossed moths with two different wing colors - dark and light.

1. 1st cross

♀ light x ♂ dark -----------→ all dark

Therefore …. light is recessive

2. Reciprocal cross

♀ dark x ♂ light ---→ all ♀ light and all ♂ dark

B. W. Bateson crossed chickens with two different feather patterns – barred and nonbarred.

1. 1st cross

♀ nonbarred x ♂ barred --------→ all barred

Therefore …….. nonbarred is recessive

2. Reciprocal cross

♀ barred x ♂ nonbarred --------→ ♀nonbarred and ♂ barred

C. Explanations of the observed genetic ratios associated with sex were provided by Thomas Hunt Morgan in the early 1900's based on his work with *Drosophila*

1. Crosses

a) red ♀ x white ♂ -------→ F1 were all red

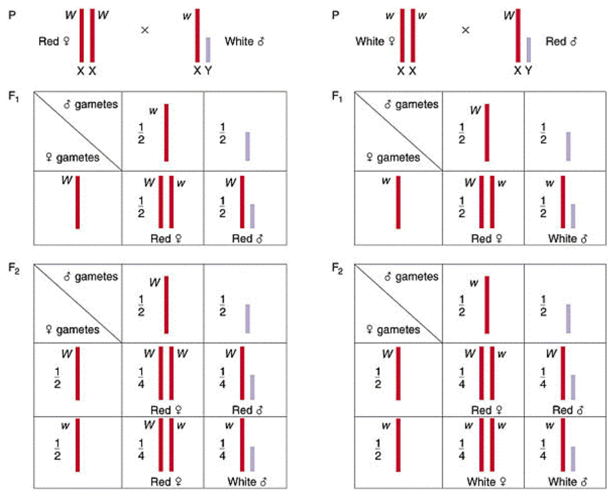
F1 cross ------→ 1/2 red ♀: 1/ 4 red ♂: 1/4 white ♂

b) white ♀ x ♂ red -------→ F1 were 1/2 red ♀ and 1/2 white ♂

F1 cross ------→ 1/4 red ♂:1/4 white ♂: 1/4 red ♀: 1/4 white ♀

2. Explanations: Morgan proposed that the gene encoding white eyes was located on the X chromosome. His results fit this hypothesis.

a)



*******It appears that the Y does not carry equivalent genes as on the X so that the recessive gene is expressed when in the male. However, to be expressed in the female, it must be homozygous.*******.

D. Importance of Morgan's work.
   1. Explained sex-linkage
   2. Basis for the proof that genes are transmitted on chromosomes. His student Calvin Bridges provided more definitive proof (talked about with Chapter 5) but Morgan's expts. described here laid the groundwork

E. Explanation of the Chicken crosses and Moth crosses

The chickens and the moths show a pattern that looks as if the male is the one that passes the recessive trait to the female. This is because the female contains the heteromorphic pair of sex chromosomes: ZW= female and ZZ=male.

Moths: ♂ Z^lZ^l (light) x ♀ Z^LW (dark) →
1/2 ♂ Z^LZ^l (dark) and 1/2 ♀ Z^lW (light)

Chickens: ♂ Z^bZ^b (nonbarred) x ♀ Z^BW (barred) →
1/2 ♂ Z^BZ^b (barred) and 1/2 ♀ Z^bW (nonbarred)

IV. Clues to Sex-linked genes:

A. General

1. For rare traits, crisscross inheritance (phenotypic traits controlled by genes on one sex chromosome are passed to all of the other sex)
2. A phenotype being expressed more often in one sex - the other sex not showing the phenotype.

B. X-Linkage

1. Genes are on the differential region of the X chromosome
2. Rare recessive alleles
   a) Many more males show the recessive phenotype. Females require both parents to pass on the recessive gene. Males require only mother to pass it down.
   b) For rare alleles, usually none of the offspring of an affected male will show it, but all daughters are carriers.
   c) For rare alleles, none of sons of affected father will show or carry the gene from him.
3. Rare Dominant alleles
   a) affected -males pass to all daughters, none of the sons.
   b) affected females pass to 1/2 daughters and 1/2 sons.

C. Y-linkage

1. Genes are on the differential region of the Y chromosome
2. Trait is inherited only by males.
3. In humans, the only proven example of Y-linkage is the *tdf* gene that confers maleness

D. X-and Y-linkage

1. Genes are on the pairing region of the sex chromosomes
2. Still are differences in the phenotypes of the sexes, but now the Y chromosome carries an allele that affects the genotype too.

V. Sex-limited inheritance

A. Expression of a phenotype is limited to one sex
B. Gene is NOT on a sex chromosome
C. Example – domestic fowl tail and neck plummage.
In the males, cock-feathering is more long and curved, while in the female, hen-feathering is more short and rounded.

Genotype	Phenotype in females	Phenotype in males
HH	Hen-feathered	Hen-feathered
Hh	Hen-feathered	Hen-feathered
hh	Hen-feathered	Cock-feathered

VI. Sex-influenced inheritance

A. Expression of a phenotype is influenced by the sex
B. Gene is NOT on a sex chromosome
C. Expression of dominance and recessiveness is reversed in males and females in the heterozygote
D. Example – male pattern baldness in humans

Genotype	Phenotype in females	Phenotype in males
BB	Bald	Bald
Bb	Not bald	Bald
bb	Not bald	Not bald