SEX DETERMINATION AND SEX LINKAGE

Objectives:

- To analyze the causes of sex determination in various organisms

Additional Reading:

Read on the following;

H. Henking

C. E. McClung

E. B. Wilson

Note: They all contributed to investigations relating chromosomes to sex determination.

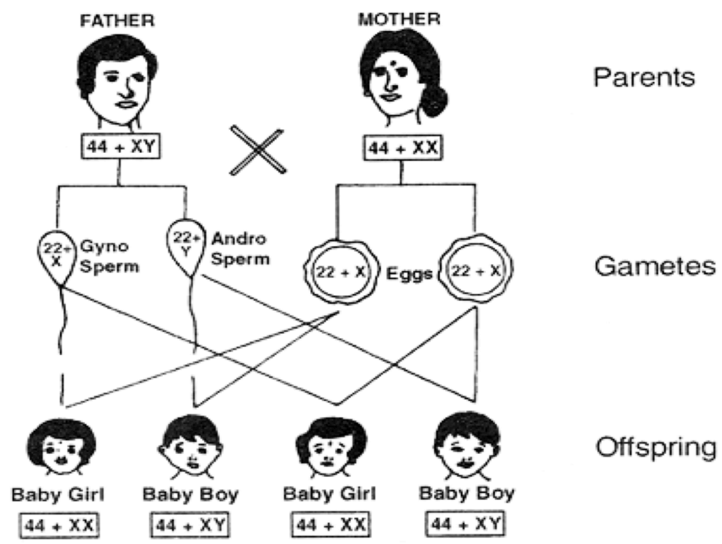
Sex Determination

- Most mechanisms for the determination of sex are under genetic (and hormonal) control.
- Basically, four types of chromosomal sex determining mechanisms exist. These are the XY, ZW, XO and compound chromosomal mechanisms.
- In humans, and most other mammals, there are two different sex chromosomes (i.e., X and Y chromosomes); **heteromorphic**

- The presence of the Y chromosome determines maleness.
- Normal males have 22 pairs of autosomes and an X and a Y chromosome.
- Since the male produces two kinds of gametes (X and Y) as far as the sex chromosomes are concerned, he is said to be the ***heterogametic*** sex.

- Females also have 22 pairs of autosomes, but have two X chromosomes.
- The female, producing only one kind of gamete, is the ***homogametic*** sex.

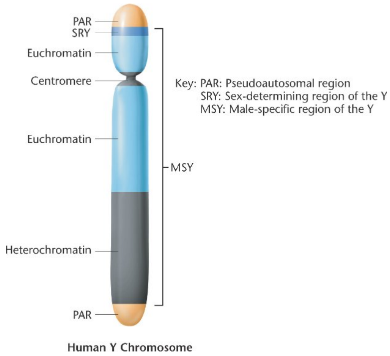
XY method of sex determination



- In both *Drosophila* and humans, normal females and males have an XX and an XY sex chromosome composition respectively **but** one should not assume that, in both species, the genes for females are on the X chromosomes and those for males are on the Y chromosomes.
- The Y chromosome plays no significant role in sex determination in *Drosophila*. Rather, it contains genes active in forming sperm in adults.

The SRY (Sex-determining region Y) gene

- A gene called SRY has been identified on the short arm of the Y chromosome.
- It encodes a gene product called ***testis-determining factor*** (TDF).
- SRY is highly conserved in mammals.
- The gene in combination with several other genes, encodes a DNA-binding protein that activates the expression of testicular development and fertility.

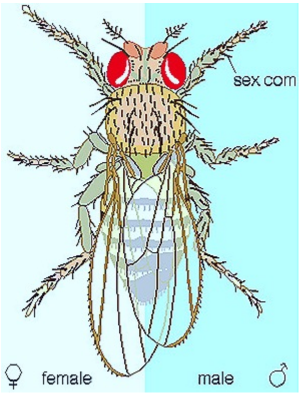


Heterogametic females (ZZ-ZW)

- This method of sex determination is found in many species, including most birds, butterflies, moths, and some fish.
- The mode is basically identical to the XX-XY mechanism.
- Females are heterogametic (designated as ZW) whilst males are homogametic (ZZ).

The Balance Concept of Sex Determination in *Drosophila* (Genic Balance)

- Investigations by C. B. Bridges showed that sex determination in *Drosophila* was more complicated.
- **He observed that, female determiners were located on the X chromosomes, and male determiners on the autosomes.**



- Specifically, the male-determining genes were shown to be located on the three autosomal chromosomes of *Drosophila.*
- The ***genic balance theory*** of sex determination explains the mechanics of sex determination in *Drosophila melanogaster.*

- It was deduced from comparisons that, one (X) and two sets (haploid sets) of autosomes (A) produced a normal male.
- Normal males had a ratio of **0.5** (1 : 2). This combination of one X and two As resulted in a normal diploid male.

- The combination of two X chromosomes, and two sets of autosomes (2X:2A) produced a normal diploid female. The ratio is **1.0**

- Several abnormal combinations have confirmed Bridges hypothesis. E.g., an individual with 2X : 3A has a ratio of 2:3 or 0.67.
- This is in between the ratios for normal maleness and femaleness.
- This kind of fly, called **intersex**, is sterile and has sexual characteristics intermediate between the male and female.
- Ratios above 1.0 produce sterile **metafemales** (or **superfemales**) whilst ratios below 0.5 produce sterile **metamales**.

Table 17.2. **Different doses of X-chromosomes and autosome sets and their effect on sex determination in *Drosophila*.**

Ploidy level	Number of X chromosomes	Sets of autosomes (A)	X/A ratio	Sex
Diploid	3	2	1.50	Superfemale
Triploid	4	3	1.33	
Haploid	1	1	1.00	Female
Diploid	2	2		
Triploid	3	3		
Tetraploid	4	4		
Triploid	2	3	0.67	Intersex
Tetraploid	3	4	0.75	
Diploid	1	2	0.50	Male
Tetraploid	2	4	0.50	
Triploid	1	3	0.33	Supermale

- The ratio determines sex by activating sex-specific gene expression of several genes.

(Read on the following genes):

sex-lethal (***Sxl***), **transformer** (***tra***) and **doublesex** (***dsx***)

Haplodiploidy and Sex Determination in *Hymenoptera*

- More involved sex determination mechanisms have been described in the insect order *Hymenoptera* e.g., ants, wasps, and bees.
- In several species, males develop **parthenogenetically** (from unfertilized egg) and have a haploid chromosome number (16 in the drone honeybee).

- The queen and the workers, on the other hand arise from fertilized eggs, and carry the diploid chromosome number of 32.

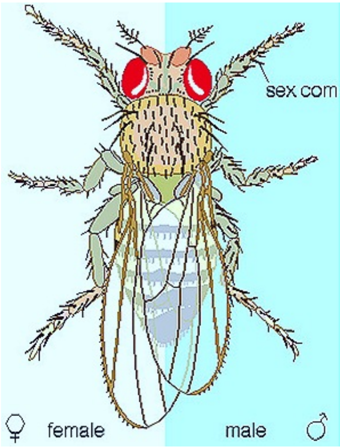
- Another example is found in the wasp *Bracon hebetor*. Females are diploid with 20 chromosomes, and males are haploid with 10 chromosomes.

- Females originate from fertilized eggs and males from unfertilized eggs.
- Because normal males are haploid and normal females are diploid, this mechanism of sex determination is often referred to as **haplodiploidy**.

Mosaics and Gynandromorphs

- Abnormal chromosomal behavior in insects can result in the formation of **gynandromorphs** or **sexual mosaics** in which some parts of the animals are female and other parts are male.

Read more on it.



Environmental Factors and Sex Determination

- Sex determination in some lower animals is non-genetic and depends on factors in the external environment.
- In these cases, males and females have similar genotypes, but stimuli from environmental sources can initiate development toward one sex or the other.

- In some reptiles, the temperature at which the fertilized eggs are incubated prior to hatching, determines the sex of the offspring.
- In the turtle, *Chrysema picta*, high incubational temperatures result in mostly female progeny, whereas in the lizard *Agama agama*, high incubation temperature result in mostly male offspring.

- **All the above examples illustrate the diversity of sex determination mechanisms that exist in nature.**

Sex Linkage or X-linked

Or

Morgan's Discovery of Sex Linkage in *Drosophila*

- The first extensive experimental evidence for sex linkage in a particular species came in 1900 by T. H. Morgan of a white-eyed mutant in *Drosophila*.

- His work showed the presence of a white-eyed (mutant) in a culture of red-eyed (wild type) flies.

- The white-eyed male was mated with red-eyed female.
- All the F_1 generation exhibited red-eye, as expected for flies heterozygous for a recessive white-eye allele.
- The F_2 generation included both red and white-eyed flies in the proportion of about 3:1

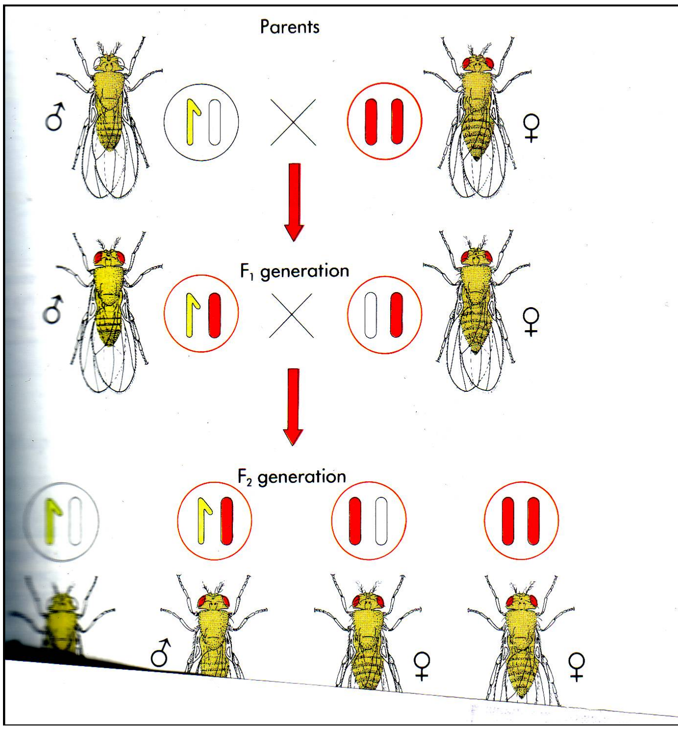


FIGURE 12-18
Morgan's experiment demonstrating the chromosomal basis of sex linkage in *Drosophila*. The white-eyed mutant male fly was crossed to a normal female. The F_1 generation flies all exhibited red eyes, as expected for flies heterozygous for a recessive white-eye allele. In the F_2 generation, all the white-eyed F_2-generation flies were male.

- However, all the white-eyed flies in the F_2 generation were males.
- About half of the F_2 males had white eyes and half had red eyes.
- All females had red eyes.

- ***In sex-linked genes, reciprocal crosses do not show similar results as shown above.***

- In the above experiment, the recessive allele was expressed **only** in males.
- Morgan arrived at an explanation by associating this “gene” with the **X chromosomes**.

- He was able to correlate these observations with a difference found in chromosome composition between male and female *Drosophila*.
- Females possess two rod-shaped homologues called the X chromosomes designated XX whilst males possess a single X chromosome and a J-shaped chromosome, designated as XY.

- Knowing that the white-eye trait is recessive to the red-eye trait, we can now see that his result was a natural consequence of the Mendelian assortment of chromosomes.

Take note:

- In normal diploid organisms with sex determining mechanisms like that of humans or *Drosophila*, a trait governed by a sex-linked **recessive** gene usually manifests itself in the following manner:

  (i) it is usually found more frequently in the male than in the female of the species.

(ii) it fails to appear in females unless it also appeared in the paternal parent.

On the other hand, a trait governed by a sex-linked **dominant** gene usually manifests itself by:

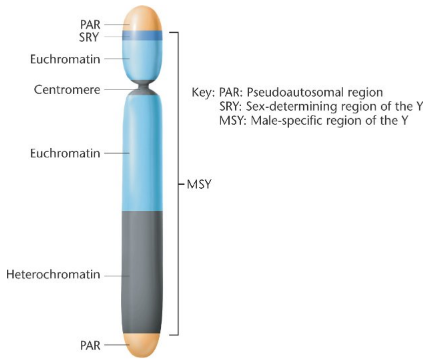
(i) being found more frequently in the female than in the male of the species.

(ii) being found in all female offspring of a male which shows the trait.

(iii) failing to be transmitted to any son from mother which did not exhibit the trait itself.

VARIATIONS OF SEX LINKAGE

- The sex chromosomes (X and Y) are of unequal size, shape and/or staining qualities.
- The fact that they pair during meiosis is indication that they contain at least some homologous segments.



Human Y Chromosome

- Genes on the homologous segments are said to be **incompletely sex-linked** or **partially sex-linked** and may recombine by crossing over in both sexes just as it happens on homologous autosomes.

- Genes on the nonhomologous segments of the X chromosome are said to be **completely sex-linked** and exhibit the peculiar mode of inheritance described above.

- In humans, a few genes are known to reside in the nonhomologous portion of the Y chromosome.
- In such cases, the trait would be expressed only in males and would always be transmitted from father to son.
- Such completely Y-linked genes are called **holandric genes**.