LESSON 7

GENETIC BASIS OF SEX. SEX-LINKED INHERITANCE

Sex-Determination System

Sex-determination system is a biological system which determines the development of sexual characteristics in an organism. Most sexual organisms have two sexes, male and female and in some cases hermaphrodites where both sexes are seen in place of one. Some organisms also have a single sex due the process of parthenogenesis where the female reproduce without fertilization. Sex determination is genetic in most of the cases. Male and female organisms possess different alleles or different genes that indicate the sexual morphology and characteristic of that organism. Chromosomal differences between the sexes are seen in the case of animals. Genetically determination of sexes is generally through chromosome combinations. The combinations may be XY, ZW, XO, ZO or haplodiploid conditions.

The X-Y system

The XX/XY sex-determination system (fig.1a) is the most familiar system of sex-determination, it is found in humans. In this system the females have two of the same type of sex chromosome (XX) and the males will have two different sex chromosomes (XY). The sex chromosomes are different in shape and size from the ***autosomes*** and are known as allosomes. Some species like humans have a specific gene present on the Y chromosome, the presence of this gene ***SRY*** determine the maleness of the progeny in some organisms like the fruit fly, the number of X chromosomes determine the sex of the offspring. If there are two X chromosomes it determines femaleness. As in fruit fly and some other species the number of X chromosomes determine the sex of the organism, these organisms are nonviable with an extra X chromosome. But the species which are dependent on the SRY gene can survive with the conditions like XXY and still be viable.

Sex-determination in the humans depends on the presence or absence of the SRY gene. The activation of the SRY promotes the cells to produce the *testosterone* hormone and *anti-mullerian hormone*, the secretion of these hormones turn the sex organs into male. The absence of the SRY gene becomes the females and the cells secretes estrogen, and drives the body to the female pathway.

In the mechanism of sex-determination that depends on the Y-chromosome the SRY is not the only gene that influences the sex of the organism. The SRY gene determines the male characteristics, multiple other genes are also required to develop testes.

The XY system of sex-determination is found in most other mammals.

The X-0 system

The XO system (fig.1b) of sex-determination is a variant of the XY system. In this system of sex determination the females have XX sex chromosome and the males have one X0 chromosome. In this type sex determination mechanism the sex of the organism is determined but the amount of genes expressed on the two chromosomes. This system of sex-determination is seen in insects like grasshoppers, crickets, cockroaches.

The Z-W system

The ZW sex-determination system (fig.1c) is a reverse system of the XY system. This type of system is found in birds, some reptiles and some insects and some other organisms. In this kind of sex-determination system the females have two different kinds of chromosomes (ZW) and males have two chromosomes of the same kind (ZZ). There are genes found on the W chromosomes for the expression of the female characteristics in birds. Not all the organisms following this type of system depend on the W chromosome for their sex. There are moths and butterflies that are ZW but there are females with ZO and ZZW. Another interesting mechanism of sex determination, known as the ***haplodiploid system*** (fig.1d).

Haplodiploid system

Haploidiploidy condition is found in *Hymenopteran* insects like ants and bees. Here the unfertilized eggs develop into males, which are haploid individuals. Diploid individuals are female but may be sterile males. Male insects here do not have any sons or fathers. If there is mating behavior of the queen bee with one drone, the daughters of the queen bee will have 3/4 of their genes unlike the 1/2 of the genes shared in the XY and ZW sex-determination systems. This kind of system is important for the development of eusociality as it increases the importance of kin selection. The Hymenopteran females can control the sex of the offspring by holding the received sperm in their spermatheca and they either release it into their oviduct or not. This creates more workers, and depends on the status of the colony.

Although sex in many species of animals is determined by chromosomes, other mechanisms are also known.

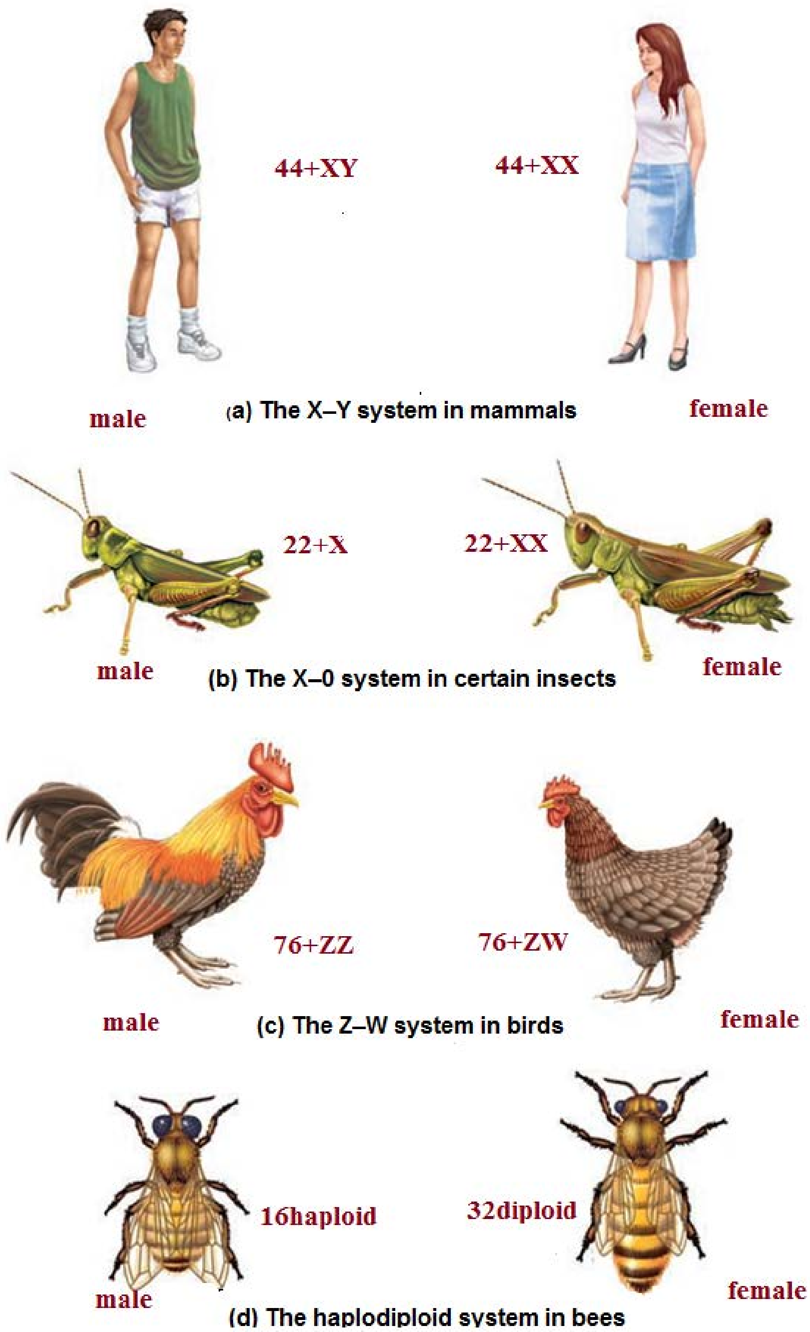

Fig. 1. Different mechanisms of sex determination in animals[1]

Temperature Dependent Sex-Determination

There are many other systems of sex-determination that exist other the chromosomal sex determination systems. In some species like the turtles, alligators and the tuatara the sex of the organisms is determined by the incubation temperature of the egg during the temperature sensitive period of the egg. For some species whose sex-determination is temperature dependent, sex determination is brought about by hotter temperature being one sex and cooler temperature being the other. In some species extreme temperature are in one sex and middle temperature are other sex. The temperature required for particular sexes are known as the female promoting temperature and the male promoting temperature. When the temperature is at the threshold during the temperature sensitive period, there is variation in between the sex ratio of the two sexes. In some species, the standards of the temperatures are based on the enzyme created. The species with temperature dependent sex determination do not have the SRY gene but they have some other genes whose expressions are not dependent on the temperature.

Sex Reversal

Sex reversal is a phenomenon where some organisms develop into sexes that are temperature specific and hatch with the opposite sex. This can be caused by pollution by humans like herbicides. This type of sex-determination system is environmental dependent. This is seen some species of snails, where the adults are male and they become female.

In tropical clown fish the dominant individuals group become the females while the other develop as males. In marine worm the larvae becomes males if they have physical contact with a female and they become females if they end on the bare sea floor.

Sex Linkage

Organism bears characters that are inheritable. The characters are represented by the genes present on the chromosomes. During the process of meiosis, all the genes present on any chromosomes segregate as a group and move together from generation to generation. The tendency of the genes on the same chromosome to stay together in hereditary transmission is known as ***linkage.*** These genes located on the same chromosomes are called ***linked genes***.

The genes that are located on the sex chromosomes are called as ***sex linked genes.*** The linkage of the sex genes or the sex linkage results in an inheritance pattern that is different from the inheritance patterns shown by the genes present on the autosomes. Sex-linked genes may be on the X chromosome which is known as the ***X linkage*** or the Y chromosome which is known as the ***Y linkage***.

In mammals the females are ***homogametic***, they have two X chromosomes (XX), while the males are ***heterogametic***, they have one X and one Y chromosomes (XY). ***Sex-linked traits*** are the characters that are determined by the genes present on the sex chromosomes.

X- Linked Inheritance

As the X chromosome is larger than the Y chromosome most of the genes that are sex-linked are present on the X chromosome. The X chromosomes also carries genes that determines the female characteristics and also non-sexual characters like the ability to see colors and the the blood clot mechanism.

X-Linked Dominant Inheritance

Relatively few genetic disorders in humans follow an X-linked dominant inheritance pattern. In most cases, males are more severely affected than females, probably because females carry an X chromosome with a normal copy of the gene.. In most of the X-linked dominant disorders (tabl.1), male embryos die at an early stage of development so that most individuals exhibiting the disorder are females. Also, due to their dominant nature and severity, persons with some of the disorders do not reproduce. Therefore, these dominant disorders, which include Rett syndrome and Aicardi syndrome, are not passed from parent to offspring. Instead, they are caused by new mutations that occur during gamete formation or early embryogenesis. For those X-linked dominant disorders in which the offspring can reproduce, the following pattern is often observed:

- ✓ Females are much more likely to exhibit the trait when it is lethal to males.
- ✓ Affected mothers have a 50% chance of passing the trait to daughters. Note: Affected mothers also have a 50% chance of passing the trait to sons, but for many of these disorders, affected sons are not observed because of lethality.

Table 1

Examples of Human Disorders Inherited in an X-linked Dominant Manner

Disorder	**Effects of Disease-Causing Allele**
Vitamin D-resistant rickets	Defects in bone mineralization at the sites of bone growth or remodeling, leading to bone deformity and stunted growth in children.
Incontinentia pigmenti	Characterized by morphological and pigmentation abnormalities in the skin, hair, teeth, and nails; fatal in males.

X-Linked Recessive Inheritance

Recessive X-linked inherited diseases (tabl.2) pose a special problem for males. Most X-linked genes lack a counterpart on the Y chromosome. Males are *hemizygous* — have a single copy — for these genes. Therefore, a female heterozygous for an X-linked recessive gene passes this trait on to 50% of her sons.

The pattern of X-linked recessive inheritance is revealed by the following observations:

- ✓ Males are much more likely to exhibit the trait.
- ✓ The mothers of affected males often have brothers or fathers who are affected with the same trait.
- ✓ The daughters of affected males produce, on average, 50% affected sons.

Table 2

Examples of Human Disorders Inherited in an X-linked Recessive Manner

Disorder	**Effects of Disease-Causing Allele**
Hemophilia	Defect in blood clotting.
Red-green color blindness	An inability or decreased ability to see color, or perceive color differences, under normal lighting conditions.

Y-Linked Inheritance (**Holandric inheritance**)

The Y linkage is known as ***holandric inheritance*** in mammals. The concepts of dominant and recessive do not apply to Y-linked traits, as only one allele (on the Y) is ever present in any one (male) individual.

Males with a single Y- or X-linked allele are described as ***hemizygotes***, because only one allele is present. The Y-linked trait is the mutation present on a gene of the Y chromosome. As the chromosome is small, and contains a few traits, the diseases linked with the Y chromosome are rare. Human males' posses only one Y chromosome, females do not have any Y chromosomes. The Y-linked traits are passed on from the father to son with no genetic recombination. The deletions in the Y chromosomes are the general cause of the male infertility. In humans, *hairy ears* appear to be inherited through the Y chromosome.

Sex-Influenced Traits

Sex influenced traits are the traits where the phenotypic expression is affected by the sex of the organism. The genes that are concerned with the sex-influenced traits may be present on any of the autosomes or on the homologous parts of the sex chromosomes. The expression of the alleles of the sex-influenced loci, either dominant or recessive is reversed in males and females, as the difference is provided by the internal environment of the sex chromosomes. Example for sex influenced trait is baldness in humans.

Sex-Limited Traits

Some genes in the autosomal chromosomes are expressed in one of the sexes. This happens either because of the differences in the internal environment of the hormones or due to the dissimilarities in the anatomy.

Examples of sex limited traits are: female sterility in Drosophila and polymorphic characters in insects in relation to mimicry.