CHROMOSOME THEORY IN SEX DETERMINATION

UNIT-IV

Topics:

- Chromosome theory in sex determination.
- Genic balance theory of sex determination, X/A ratio in Drosophila
- Chromosomal mechanisms of sex determination in mammals, Grasshoppers, birds and Man
- Variation in chromosomal number; haploidy, diploidy, polyploidy, aneuploidy.

Chromosome theory in sex determination

A sex-determination system is a biological system that determines the development of sexual characteristics in an organism.

- **The chromosome theory of sex determination proposed by McClung.**
- **According to this theory, chromosomes play a major role in determination of sex.**
- **In diploid organisms, a pair of chromosomes determines the sex of individual.**
- **They are called the sex chromosomes or allosomes or heterosomes.**

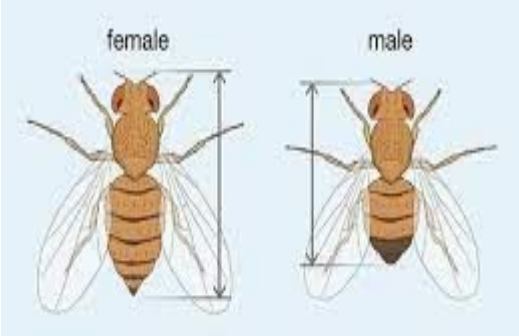
Autosomes	Allosomes (Sex Chromosomes)
Chromosomes which are not connected with sex determination.	Chromosomes which are connected with sex determination.
Similar in males and females.	Dissimilar in males (XY) and female (XX).
Generally 44 or 22 pairs in human beings.	Generally 2 or one pair in human beings.

Genic balance theory of sex determination

The theory of genic balance given by Calvin Bridges (1926) states that instead of XY chromosomes, sex is determined by the genic balance or ratio between X-chromosomes and autosome genomes.
It is applicable to Drosophila melanogaster.

Sex Determination in Drosophila

- In Drosophila, Y chromosome is not involved in the process of sex determination.
- Instead, each fly's sex is determined by a balance between genes on the autosomes and genes on the X chromosome. i.e
- The value of the ratio X/A is considered for the fly to be female or male.
- This type of sex determination is called the **genic balance system**
- The X chromosome contains genes with female producing effects, whereas the Autosomes contain genes with male-producing effects.



X/A ratio in Drosophila

- Consequently, Drosophila fly's sex is determined by the X:A ratio,
  The number of X chromosomes divided by the number ofhaploid sets of autosomal chromosomes.

$$\text{Sex Index} = \frac{\text{No. of X chromosomes}}{\text{Autosomal sets}} = \frac{X}{A}$$

X/A Ratio	Sex of that individual
Greater than 1	Super females/Meta females
Equal to 1	Normal Females
Between 1 & 0.5	Intersex
Equal to 0.5	Normal males
Less than 0.5	Super males/Meta males

Chromosome Constitution	X/A ratio	Sex Index
AA + XXX	3/2 = 1.50	Super ♀
AA + XX	2/2 = 1.00	Normal ♀
AAA + XXY	2/3 = 0.67	Intersex
AA + XY	1/2 = 0.50	Normal ♂ (Fertile)
AA + XO	1/2 = 0.50	♂ (Sterile)
AAA + XY	1/3 = 0.33	Super ♂

Sex Determination in Drosophila

Sex-Chromosome Complement	Haploid Sets of Autosomes	X:A Ratio	Sexual Phenotype
XX	AA	1.0	Female
XY	AA	0.5	Male
XO	AA	0.5	Male
XXY	AA	1.0	Female
XXX	AA	1.5	Metafemale
XXXY	AA	1.5	Metafemale
XX	AAA	0.67	Intersex
XO	AAA	0.33	Metamale
XXXX	AAA	1.3	Metafemale

An X:A ratio of 1.0 produces a female fly; X:A ratio of 0.5 produces a male.

- If the X:A ratio is less than 0.5, a male phenotype is produced, but the fly is weak and sterile such flies are sometimes called metamales.
- An X:A ratio between 1.0 and 0.50 produces an intersex fly, with a mixture of male and female characteristics.
- If the X:A ratio is greater than 1.0, a female phenotype is produced, but these flies (called metafemales) have serious developmental problems and many never emerge from the pupal case.

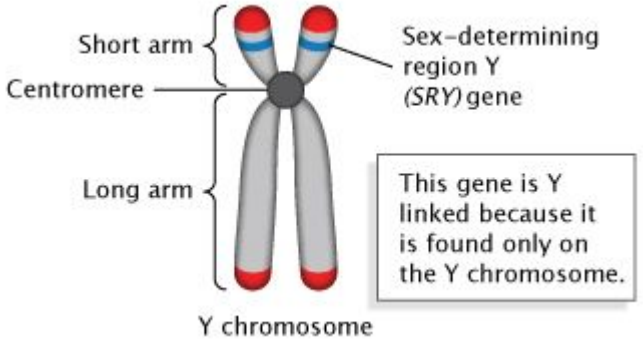
Chromosomal mechanisms of sex determination in mammals

In mammals, primary sex is determined by chromosomes known as the **X and Y** and is not usually influenced by the environment.
In most cases,

the **female is XX** and
the **male is XY**.

Every individual must have at least one X chromosome. Since the female is XX, each of her eggs has a single X chromosome.

- Maleness in mammals is genetically determined by the Y chromosome. On the Y chromosome SRY is known as the mammalian male-determining gene. Both placental mammals (Eutheria) and marsupial mammals (Metatheria) have SRY genes.

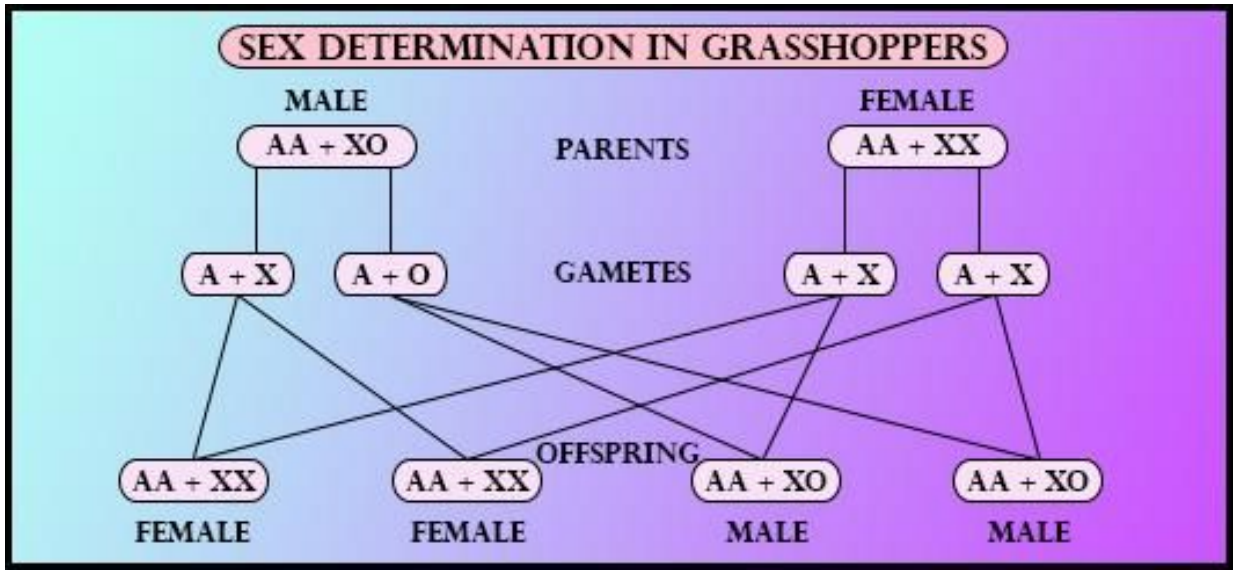


Sex determination in Grasshoppers

The sex determining mechanism in grasshopper is **XX-XO type**.
(e.g. crickets, grasshoppers, and some other insects)

- The males have only one X chromosome besides the autosomes whereas female has two X chromosomes.

- **The female is XX** and is the **homogametic sex**.
- **The male is XO** and the heterogametic sex but only has one sex chromosome.

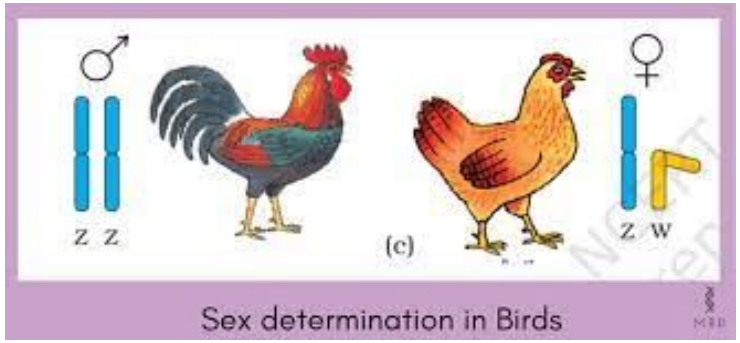


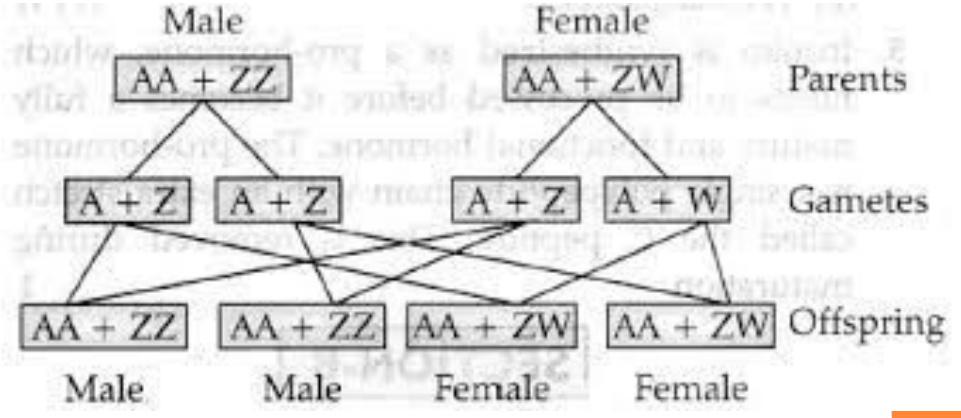
Sex determination in bird

In birds, the sex determination is by Z and W sex chromosomes.
The sex determining mechanism in bird is **ZZ-ZW type**.

- Female birds are heterogametic, that is they contain ZW type of chromosomes.
- Male birds are homogametic, they contain ZZ type of chromosomes.

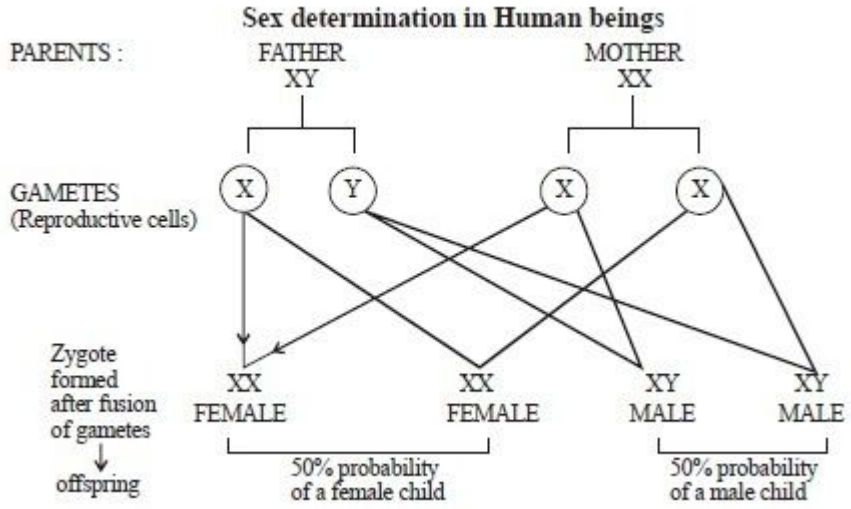
The **W chromosome contains the female determining properties.**

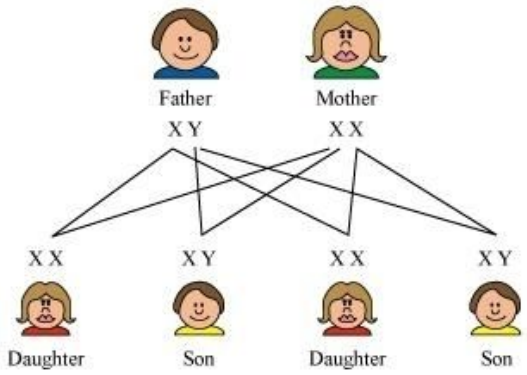




Sex determination in man

- The sex determination in humans is based on the **XX and XY chromosomes**.
- In males, the XY chromosomes are present. During the gamete formation, 50% of the sperms receive the X-chromosomes and 50% of the sperms receive the Y-chromosomes. Hence, males are heterogametic.
- **Both males and females carry two sets of sex chromosome.**
    - **Male has one X and one Y (XY) sex chromosome in which both are active**
    - **Female has both X (XX) sex chromosome in which one is active.**
- **The XY sex-determination system is found in humans, mammals, in some insects, and in few plant species.**





Variation in Chromosome Number

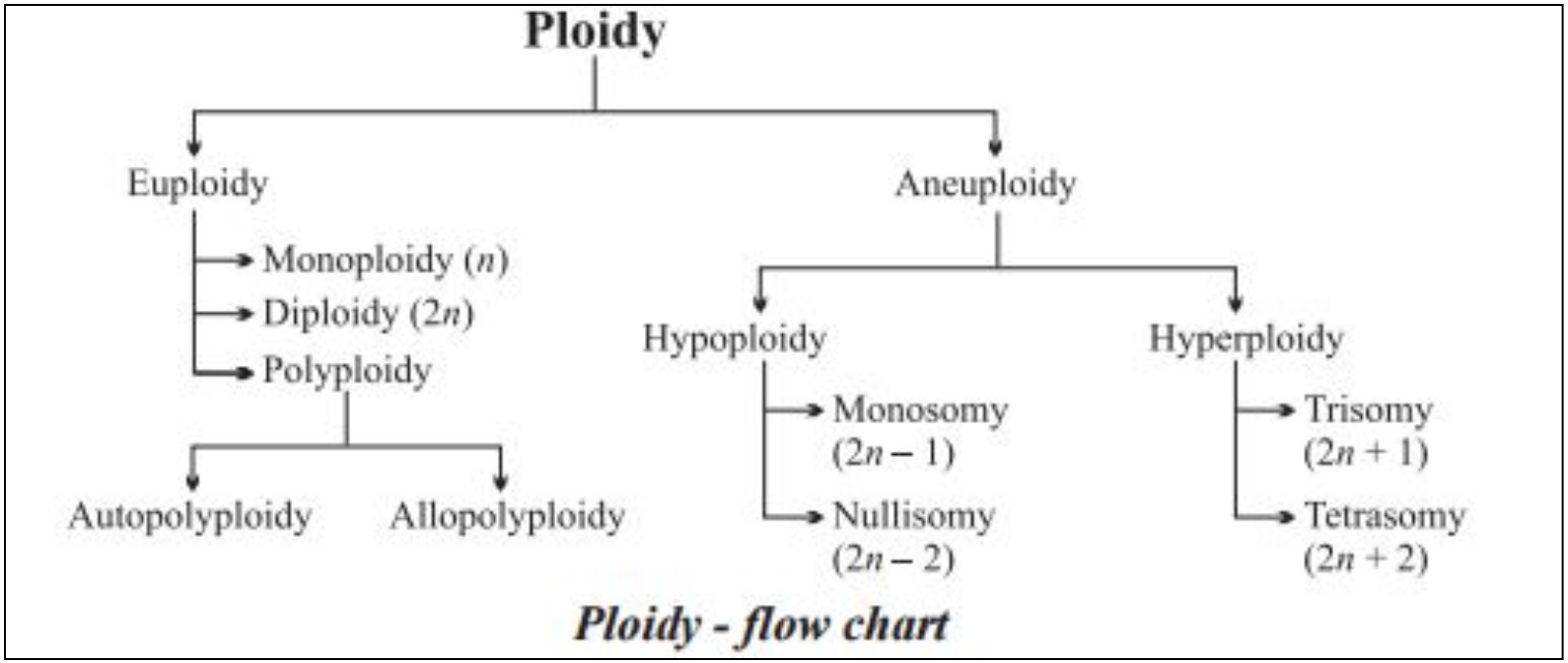
- The variation in chromosome number due to addition or loss of one or more chromosomes.

Chromosome numbers can vary in two main ways

- **Aneuploidy**
  - Variation in the number of particular chromosomes within a set
  - Regarded as abnormal
  - Examples: **trisomy** ($2n+1$), **monosomy** ($2n-1$)

- **Euploidy**
  - Variation in the number of complete sets of chromosomes
  - Occur occasionally in animals and frequently in plants
  - Examples: **triploid** ($3n$), **tetraploid** ($4n$)

Variation in Chromosome Number classification



Ploidy - flow chart

Term	Explanation
Aneuploidy	$2n \pm x$ chromosomes
Monosomy	$2n - 1$
Trisomy	$2n + 1$
Tetrasomy, pentasomy, etc.	$2n + 2$, $2n + 3$, etc.
Euploidy	Multiples of n
Diploidy	$2n$
Polyploidy	$3n$, $4n$, $5n$, . . .
Triploidy	$3n$
Tetraploidy, pentaploidy, etc.	$4n$, $5n$, etc.
Autopolyploidy	Multiples of the same genome
Allopolyploidy (Amphidiploidy)	Multiples of different genomes

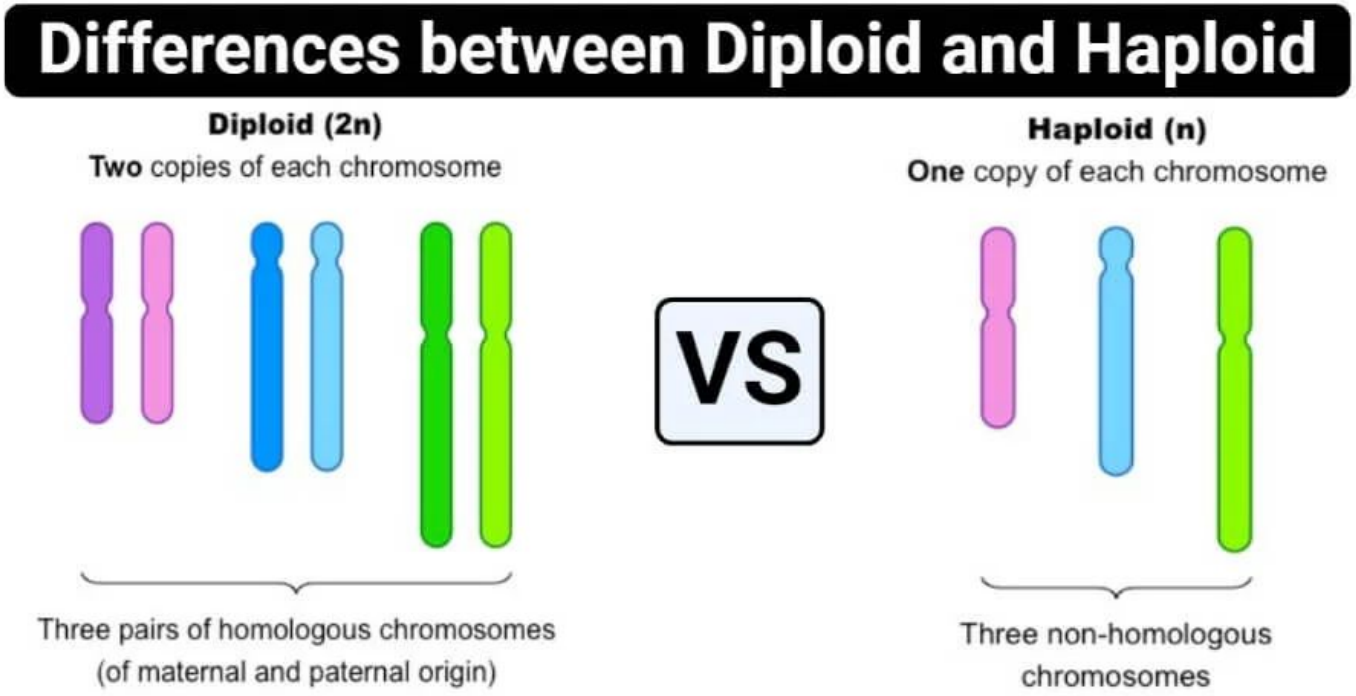
Haploid and Diploid

Haploid refers to the presence of a single set of chromosomes in an organism's cells.

- Sexually reproducing organisms are diploid (having two sets of chromosomes, one from each parent). In humans, only the egg and sperm cells are haploid.

Diploid is a term that refers to the presence of two complete sets of chromosomes in an organism's cells, with each parent contributing a chromosome to each pair.

- Humans are diploid, and most of the body's cells contain 23 chromosomes pairs.

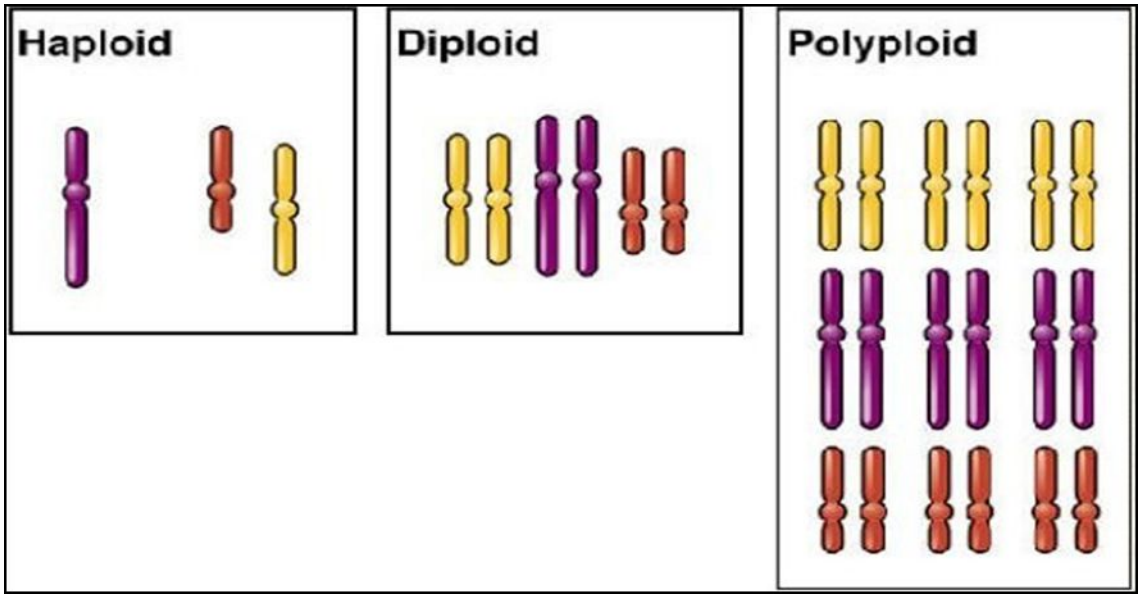


Polyploidy and Aneuploidy

- **Polyploidy** is the heritable condition of possessing more than two complete sets of chromosomes.
  - Polyploids are common among **plants**, as well as among certain groups of **fish** and **amphibians**. For instance, some salamanders, frogs, and leeches are polyploids.

- **Aneuploidy** is usually defined as the condition which is characterised by having an abnormal number of chromosomes in a haploid set.
  - Aneuploidy is a type of chromosomal aberration, where there is one extra chromosome or one missing chromosome.

ANEUPLOIDY	**POLYPLOIDY**
• results in the gain or lose of achromosome	• results in an increase in a whole set of chromosomes
• often seen in humans	**• often seen in plants**
• Down's syndrome and Turner's syndrome	**• It may be diploid (2 sets of chromosomes), triploid or tetraploid.**



Thank You!