

10. Sex Determination in Animals

The development of a zygote into male or female is called **sex determination**.

Sex is a character. It has two alternatives, namely **maleness** and **femaleness**. The male produces the sperm and the female produces the egg.

Sex is determined by the following factors:

1. Chromosomes
2. Environment
3. Hormone
4. Metabolism
5. Parasites.

Sex Determination in Man

The development of an organism into male or female is called **sex determination**.

Sex is a character. It has two alternatives, namely **maleness** and **femaleness**. The male produces the sperm and the female produces the egg.

In man, sex is determined by three factors, namely-

1. Chromosomes
2. Barr body
3. Hormones

1. Sex Determination by Chromosomes

The determination of sex by chromosomes is called **chromosomal theory** of sex determination. It was proposed by **Mc Clung**.

The female has two X chromosomes and the male has one X chromosome and one Y chromosome.

The female is **homogametic** and it produces only one type of eggs all carrying one X chromosome.

The male is **heterogametic** and it produces two types of gametes; one type of sperm carries one X chromosome and the other type of sperm carries one Y chromosome. This phenomenon is called **heterogamesis**.

In human beings, the sex is determined by the sperms. When a sperm carrying X chromosome fuses with the egg, the resulting baby is female. When a sperm carrying Y chromosome fuses with the egg, the resulting baby is male. Thus, **father** determines the sex of a baby.

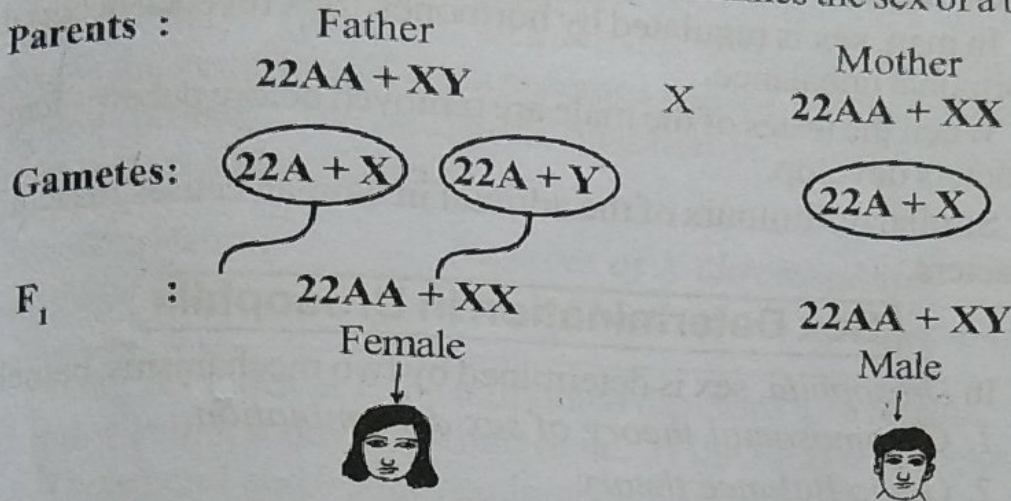


Fig.10.1: Sex determination in Man.

2. Sex Determination by Barr body

The darkly stained inactivated X chromosome attached to the inner surface of nuclear membrane is called **Barr body**. It was discovered by **Barr**. It is also called **sex-chromatin**.

The Barr body is the **inactivated X chromosome**.

The sex is identified by the presence or absence of Barr body.

The male has no Barr body in the nucleus.

The female has one Barr body.

The triploid female has two Barr bodies.

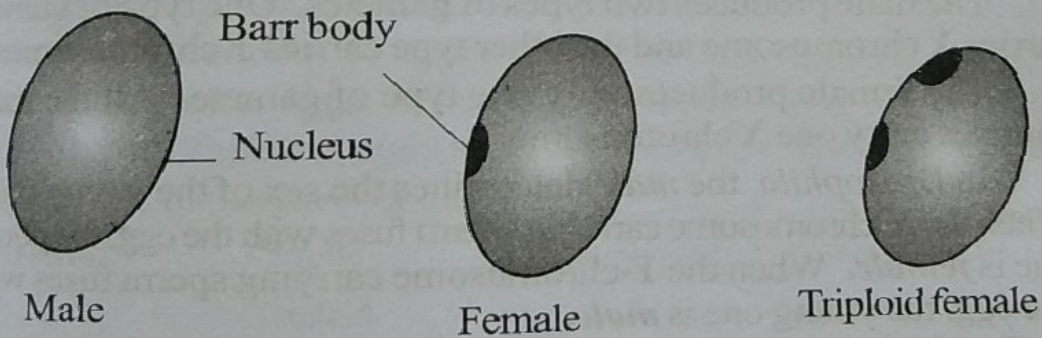


Fig.10.2 : Sex determination by Barr bodies.

The number of Barr bodies is always one less than the expected number of X chromosomes. The normal females have 2X chromosomes. So the number of Barr body is $2 - 1 = 1$.

The normal males have one X chromosome. So there is no Barr body ($1-1=0$). The triploid female has 3X chromosomes. So it has 2 Barr bodies ($3-1$).

3. Sex Determination by Hormones

In man, sex is regulated by hormones. Sex reversal occurs due to hormonal unbalance.

When the testes of the male are removed before puberty, female characters develop.

Similarly, tumours of the adrenal in woman causes masculine characters.

Sex Determination in *Drosophila*

In *Drosophila*, sex is determined by two mechanisms, namely

1. Chromosomal theory of sex determination

2. Genic Balance theory

1. Chromosomal Theory of Sex Determination

According to chromosomal theory, sex chromosomes determine the sex of *Drosophila*. The female has XX chromosomes and it is **homogametic**. The male has XY chromosomes and it is **heterogametic**.

Drosophila has 4 pairs of chromosomes. Of these 3 pairs are **autosomes** and one pair is **allosomes** (sex chromosomes).

The male has 3 pairs of autosomes and one pair of sex chromosomes. The sex chromosomes are **XY**.

The female has 3 pairs of autosomes and one pair of sex chromosomes. The sex chromosomes are **XX**.

The male produces two types of gametes. One type of gamete carries X chromosome and the other type carries Y chromosomes.

The female produces only one type of gamete. All the male gametes carry one X chromosome.

In *Drosophila*, the **male** determines the sex of the young ones. When the X-chromosome carrying sperm fuses with the egg, the young one is **female**. When the Y-chromosome carrying sperm fuses with the egg, the young one is **male**.

2. Genic Balance Theory

This theory was formulated by **Bridges**. According to this theory, sex is determined by the relative number of X chromosomes and autosomes. It is actually the ratio between the X chromosomes and autosomes determines the sex.

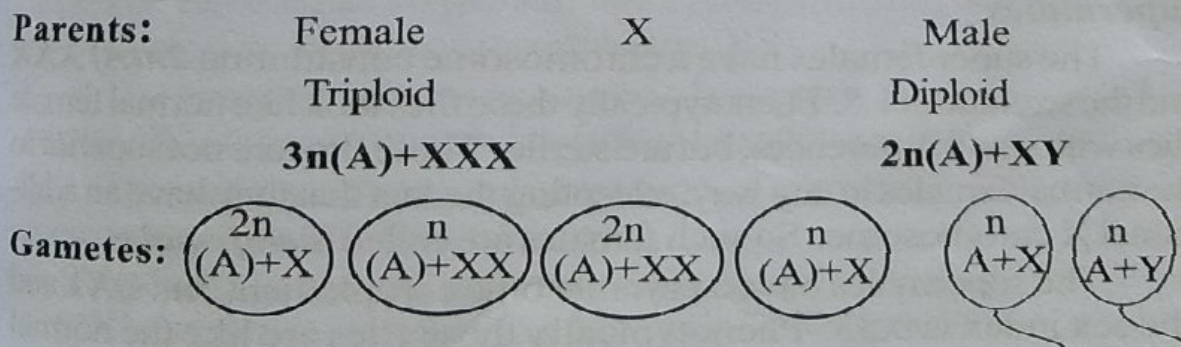
The X chromosomes carry female stimulating genes and the autosomes (A) seem to carry the male stimulating genes. There is no sex influencing genes in Y chromosomes. Haploid sets of autosomes are represented as $n(A)$ and diploid sets of autosomes are represented as $2n(A)$. The sex of an animal is determined by the ratio between the number of X chromosomes and the number of haploid sets of autosomes. The ratio is the quantitative balance between X chromosomes and autosomes.

$$\text{Sex determining ratio (Sex index)} = \frac{\text{Number of X chromosomes}}{\text{Number of haploid sets of autosomes}}$$

If the sex index is 1, the individual develops into female. If the sex index is 0.5, it develops into male. If the ratio is intermediate (0.67) between 1 and 0.5 the resulting individual is an *intersex*. If the ratio is above 1 (1.5), the sex is super female and if the ratio is below 0.5 (0.3), the sex is supermale.

In male *Drosophila*, there are 2 sets of autosomes $2n(A)$ and one X chromosome. Hence the ratio is $X/2n(A) + 1/2 = 0.5$. In female, there are two sets of autosomes $2n(A)$ and two X chromosomes. Hence the ratio is $2/2 = 1$.

Bridges also explained the formation of supersexes and intersexes in *Drosophila*. He found some *Drosophila* females with triploid sets of chromosomes $3n(A)XXX$. These triploid females are much like the normal diploid ones in appearance and are fertile. Bridges crossed this triploid female with normal diploid male. The diploid normal male produces two types of sperms. The triploid female produces four types of eggs. When the four types of eggs are fertilized by two types of sperms, eight sexually distinct kinds of offspring are produced as in the checker board.



Gametes	$2nA+X$	$nA+XX$	$2nA+XX$	$nA+X$
n $A+X$	$3nA+XX$ $\frac{X}{A} = \frac{2}{3} = 0.67$ Inter sex	$2nA+XXX$ $\frac{X}{A} = \frac{3}{2} = 1.5$ Super female	$3nA+XXX$ $\frac{X}{A} = \frac{3}{3} = 1.0$ Trip. female	$2nA+XX$ $\frac{X}{A} = \frac{2}{2} = 1.0$ Female
n $A+Y$	$3nA+XY$ $\frac{X}{A} = \frac{1}{3} = 0.33$ Supermale	$2nA+XXY$ $\frac{X}{A} = \frac{2}{2} = 1.0$ Exceptional female	$3nA+XXY$ $\frac{X}{A} = \frac{2}{3} = 0.67$ Inter sex	$2nA+XY$ $\frac{X}{A} = \frac{1}{2} = 0.5$ Male

Fig. 10.3 : Development of intersexes and supersexes in *Drosophila*.

Table 10.1: Sexes and sex index of *Drosophila*.

No.	Nature of the sex	Number of chromosomes	Number of sets of Autosomes	Sex Index Ratio X/A
1.	Superfemale	3X	2n(A)	$3/2 = 1.5$
2.	Triploid female	3X	3n(A)	$3/3 = 1$
3.	Diploid female	2X	2n(A)	$2/2 = 1$
4.	Intersex	2X	3n(A)	$2/3 = 0.67$
5.	Intersex	2XY	3n(A)	$2/3 = 0.67$
6.	Normal male	X	2n(A)	$1/2 = 0.5$
7.	Super male	X	3n(A)	$1/3 = 0.33$
8.	Exceptional female	2XY	2n(A)	$2/2 = 1$

Supersexes

Supersexes are of two types; they are *superfemales* and *supermales*.

The super females have a chromosome constitution $2n(A)XXX$ and the sex ratio is 1.5. Phenotypically these flies look like normal female flies with small differences, but are sterile. These flies are not superior to the normal females in any way, excepting the fact that they have an additional X chromosome. So such females are called *metafemales*.

The supermales have a chromosome complement $3n(A)XY$ and the sex index is 0.33. Phenotypically these flies are like the normal

male with slight changes and also are sterile. These are called *supermales*, just because these have an overdose set of autosomes than the normal males. So such males are called as *metamales*.

Intersex

The cross between diploid male and a triploid female *Drosophila* produces some flies that are intermediate between normal males and females. Their chromosome constitution is either $3n(A)+XX$ or $3n(A)+XXY$ and the sex index is 0.67. Such flies are called *intersexes*. The intersexes are sterile individuals intermediate between females and males. Their reproductive organs are intermediate between testis and ovary and have a mixture of other male and female sex characters.

Sex Determination in Fowl

In fowl, sex is determined by ZW chromosomes. The *male* is *homogametic* and the *female* is *heterogametic*.

The chromosome number of fowl is 17 pairs. The male has two similar sex chromosomes, namely ZZ instead of XX. The female has two dissimilar sex chromosomes, namely ZW. Here Y is named as W.

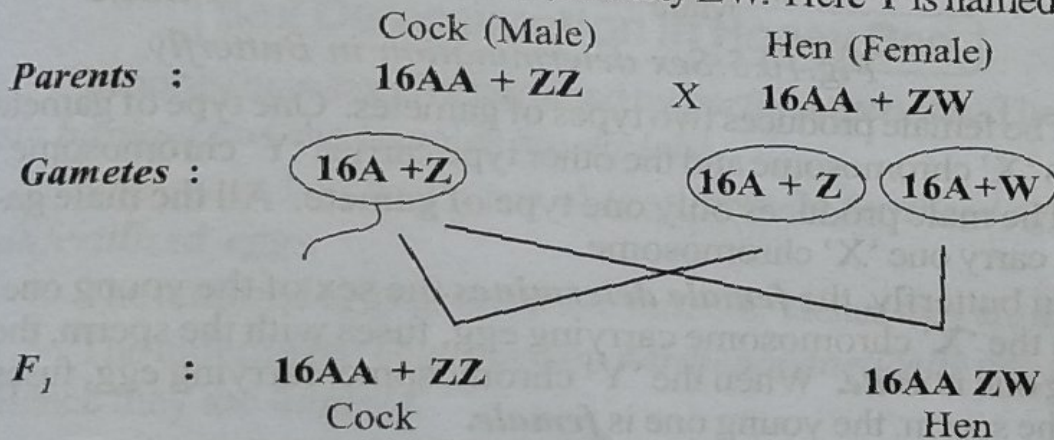


Fig.10.4: Sex determination in fowl.

The male has 16 pairs of autosomes and one pair of sex chromosomes. The sex chromosomes are 'XX'.

The female has 16 pairs of autosomes and one pair of sex chromosomes. The sex chromosomes are 'XY'.

The female produces two types of gametes. One type of gamete carries 'X' chromosome and the other type carries 'Y' chromosome.

The male produces only one type of gamete. All the male gametes carry one 'X' chromosome.

In fowl, the *female determines* the sex of the young ones. When the 'X' chromosome-carrying egg, fuses with the sperm, the young

mating within a race normal flies appear and intersexes do not appear because of balance between factors of maleness and femaleness. But when mating occurs between different races intersexes appear along with normal flies.

Any zygote with a weak cytoplasmic factor and a strong male X chromosome factor will be female intersex. Any zygote with a strong female factor and a weak male determining factor will be a male intersex. If an European female (weak) is crossed with a Japanese male (strong), the males, are normal. But the females become intersex because the female determining factors are inherited from weak female. In the same way, if a European male (weak) is crossed with a Japanese female (strong), the female offspring are normal while a male develops into intersex.

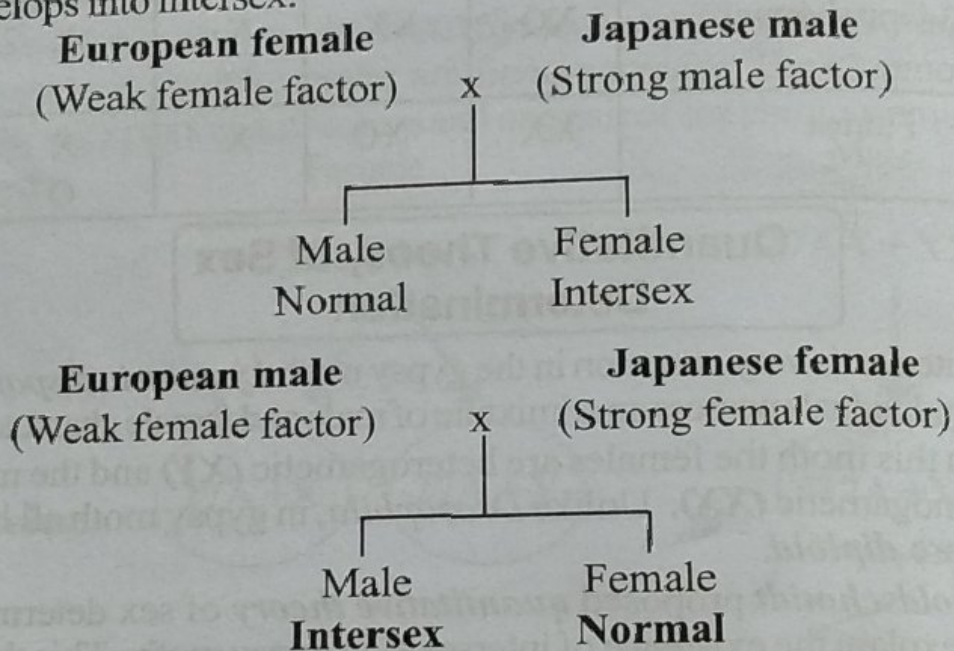


Fig.10.9: Crosses between different races of gypsy moths.

Gynandromorphs

Gynandromorphs are individuals which show male characters on some parts of the body and female characters on other parts of the body. They are also called **gynanders**. The gynandromorphs are **sterile**.

Gynanders are rare. They occur in *Drosophila*, butter flies, beetles, wasps, bees, silk worms, etc.

There are three types of gynanders. They are the following:

1. Bilateral Gynanders: These have male traits on one lateral side of the body and female traits on the other lateral side. Eg. *Drosophila*.

2. Anteroposterior-gynanders: They have features of one sex on the anterior half of the body and those of the other sex on the posterior half of the body. Eg. *Beetles*.

3. Sex pie balds: These are gynanders having a mixture of male and female tissues in the body.

Gynandromorphism is produced in two ways.

One type is produced by the loss of one X chromosome in a blastomere. Another type is produced by a binucleate egg.

1. Loss of X Chromosomes

A gynander begins its development with $2n(A) + XX$ chromosomes. But in the course of cell division, an X gets lost from one of the products of cell division. So one daughter cell possesses $2n(A) + XX$ chromosome and other $2n(A) + X$. In case, this should happen during first zygotic division two blastomeres with unequal number of X chromosomes are formed. The blastomere with $2n(A) + XX$ chromosomes develops into female half, while the second blastomere with $2n(A) + X$ chromosomes produces male half.

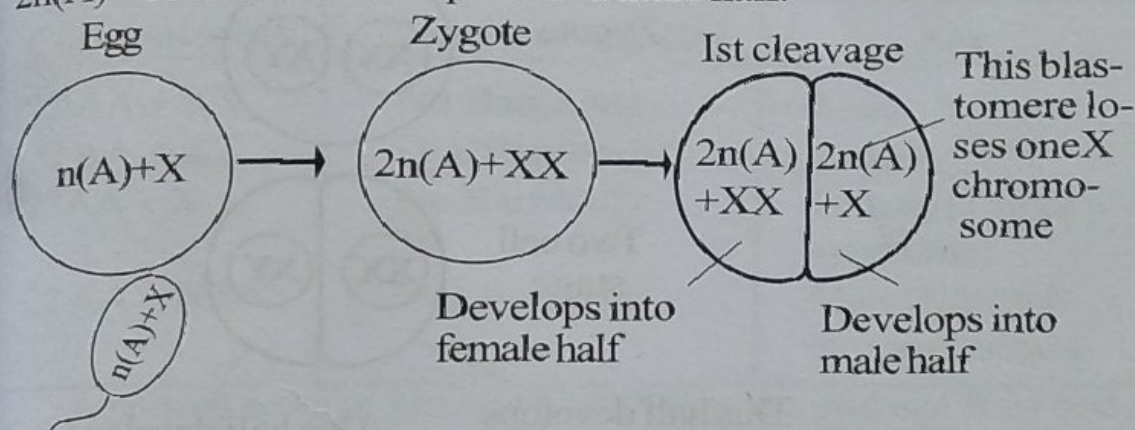


Fig.10.10: Development of gynanders in *Drosophila* owing to the loss of one X chromosome.

2. Binucleated Eggs

This mechanism was explained by **Goldschmidt** in silkworm moths. In silk worm, females are **XY** and males are **XX**. During oogenesis, X and Y chromosomes normally separate, one passing into egg and the other into the polar body.

Sometimes, both the nuclei are present in the egg and a binucleate egg (**XX** and **XY**) is produced. The binucleate egg may be fertilized by two sperms, each fertilizing one egg nucleus. After fertilization and cleavage, one blastomere (**XX**) develops into male parts and the other blastomere (**XY**) develops into female parts.

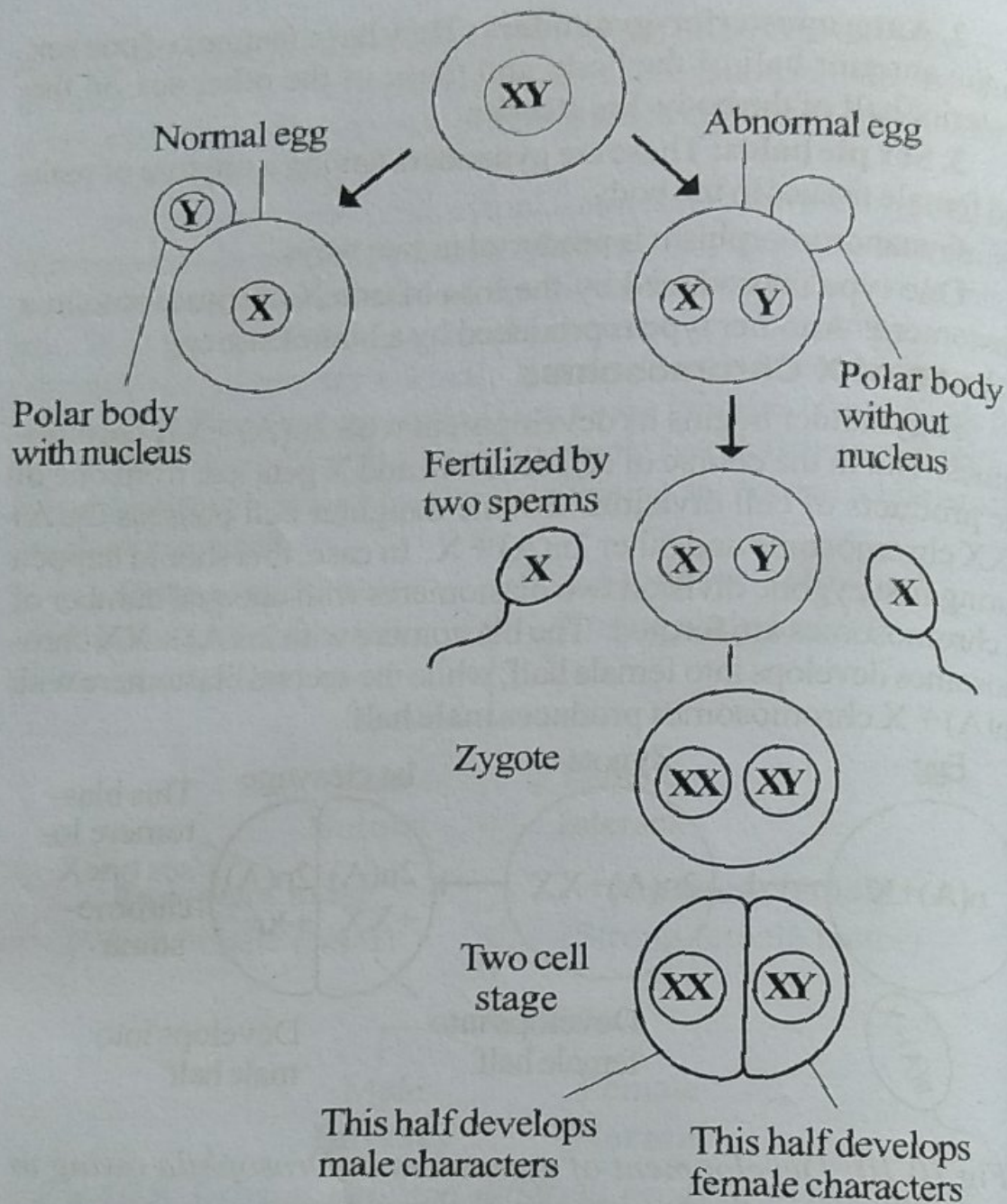


Fig.10.11: Formation of gynanders in silkworm.

Barr Body or Sex Chromatin

A darkly staining chromatin body present in the nucleus is called **Barr body**. It is also called **sex chromatin**.

It was discovered by **Barr** 1949. The Barr body is attached to the inner surface of the nuclear membrane. It contains a large amount of **heterochromatin** and lesser amount of **euchromatin**. Hence it contains large amount of RNA and lesser amount of DNA.

It is nothing but the **inactivated X chromosome**.

Origin of Polyploidy

The polyploid condition arises owing to the abnormalities in cell division. During gametes formation, the homologous chromosomes may not separate completely; hence one cell is formed with the diploid number of chromosomes and the other cell without any chromosome. During fertilization, such a gamete with two sets of chromosomes pair with a gamete having haploid (N) number of chromosomes resulting individual is a **Tetraploid** (4N). When both diploid gametes belong to the same species, the resulting tetraploid is called **autotetraploid**. When the two diploid gametes belong to two different species the resulting tetraploid is called **allotetraploid**.

II. Gene Mutations

The change in the base sequence of genes is called **gene mutation**. Mutation produces an altered gene. The organism carrying the altered gene is called a **mutant**. The organism carrying the normal (unaltered) gene is called **wild type**.

The process of producing mutation is called **mutagenesis**. Mutation is classified into the following types:

1. Spontaneous Mutation

The mutation occurring naturally is called **spontaneous mutation**. It is due to normal cellular operations or due to random interactions with the environment.

2. Induced Mutation

Artificially produced mutations are called **induced mutations**. They are caused by certain factors called **mutagens**. Mutagens may be physical or chemical factors. Eg. X-rays, nitrous acid, etc.

3. Point Mutation

When a single base pair is altered, the mutation is called **point mutation**.

A point mutation is classified into three types, namely

- | | |
|----------------------|-------------------|
| a. Base substitution | c. Base deletion |
| b. Base insertion | d. Base inversion |

a. Base Substitution: In base substitution, a base is replaced by another base.

b. Base Insertion: In base insertion, a new base is inserted.

c. Base Deletion: In base deletion, a base is missing.

d. Base Inversion: In base inversion, the base sequence is reversed.

Normal gene

A T G A C T G

Base substitution

A T G T C T G

Base insertion

A T G G A C T G

Base deletion

A T G C T G

Base inversion

A T C A G T G

Fig.22.9: Point mutations.

4. Missense Mutation

Sometimes in a polypeptide chain, an amino acid is substituted by another amino acid. This is due to substitution of three bases (a codon) in the DNA. This mutation is called *missense mutation*.

5. Temperature Sensitive Mutation

In some cases, substitution of bases produces a protein that is active at one temperature (typically 30° C) and inactive at a higher temperature (usually 40 - 42° C). This mutation is called *temperature sensitive mutation* or *Ts mutation*.

6. Non-sense Mutation

Sometimes mutation produces a base sequence that does not code for any amino acid (non-sense codon). In such cases, termination of the synthesis of protein occurs at this point. This mutation is called *non-sense mutation* or *chain termination mutation*.

There are three kinds of non-sense mutations. They are *amber*, *ochre* and *opal*.

7. Silent Mutation

Substitution of 3 bases in the DNA may result in the substitution of a new amino acid in the polypeptide chain. When the substituted amino acid is closely related to the original amino acid, mutation has no detectable effect on the phenotype of the cell. This mutation is called *silent mutation*.

8. Leaky Mutation

When the substitution of an amino acid results in the reduction in the activity of the protein or enzyme, the mutation is called

leaky mutation. For example, a bacterium carrying a leaky mutation in the enzyme will grow very slowly, when the enzyme controls the synthesis of an essential substance.

9. Transition

Transition is a point mutation where one purine base is substituted by another purine or one pyrimidine base is substituted by another pyrimidine. Eg. *G-C pair is exchanged with an A-T pair or vice versa.*

10. Transversion

Transversion is a point mutation where a purine is replaced by a pyrimidine or vice versa. Eg. *An A-T pair is replaced by a T-A or C-G pair.*

11. Base-analogue Mutation

Certain chemicals are similar to the bases of DNA. These chemicals are called **base-analogues**. The base-analogue has the ability to pair with a base of the DNA causing an alteration in the gene. *The mutation caused by the pairing of a base-analogue with a base of the DNA is called base-analogue mutation.*

The base **5-bromouracil (BU)** is an analogue of thymine. Hence BU functions like thymine and can easily pair with adenine causing base-analogue mutation.

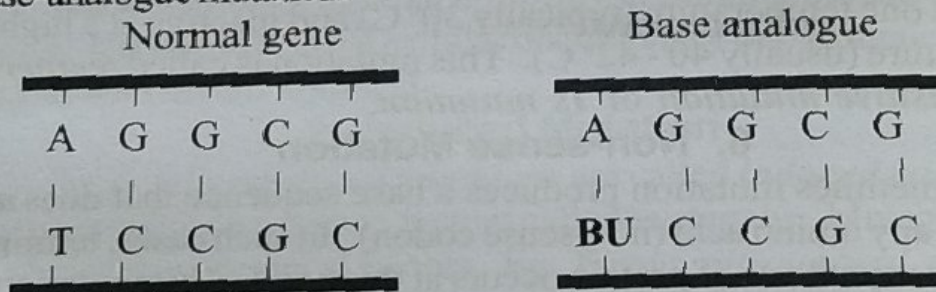


Fig.22.10: Base-analogue mutation by BU.

12. Frameshift Mutation

A mutation that inserts or deletes a single base will change the reading frame for the entire subsequent sequence. A change of reading frame is called frameshift mutation.

When the genetic code is read in non-overlapping triplets, there are three possible ways of translating a nucleotide sequence into protein, depending on the starting point. These are called **reading frames**. For example, the following sequence has three reading frames:

DNA sequence

ACGACGACGACGACGACGACG

Reading frames

ACG ACG ACG ACG ACG ACG ACG
 CGA CGA CGA CGA CGA CGA CGA
 GAC GAC GAC GAC GAC GAC GAC

Fig.22.11: Three possible reading frames of a DNA sequence.

As the sequence of the new reading frame is completely different from the old one, the entire amino acid sequence of the protein is altered beyond the site of mutation. Thus the function of the protein is altered.

Tyr	Glu	Tyr	Gly	Ile
TAC	GAA	TCG	GGT	ATT
ATG	CTT	AGC	CCA	TAA



TAC	GAGA	TCG	GGT	ATT
ATG	CTCT	AGC	CCA	TAA
Tyr	Glu	Ile	Gly	Tyr

Fig.22.12: Frame shift mutation. The addition of a new base results in the change in the sequence of amino acids.

The base shift mutation is induced by acridine compounds that bind to DNA and distort the structure of the double helix causing additional bases to be incorporated or omitted during replication.

13. Back Mutation

The regaining of normal (wild) gene by mutation is called **back mutation** or **reverse mutation** or **reversion**. A reverse mutant is called a **revertant**. As a result of back mutation, the original wild type phenotype is regained.

Hot Spot

Hot spot is a site of DNA at which the frequency of mutation is very high. Hot spots are not frequent. Some hot spots yield large deletions rather than point mutations.

The mutation frequency can be very high at a Me C site. Hence Me C site is a hot spot.

The chromosome with the inverted segment produces an inversion loop.

Inversion prevents crossing over.

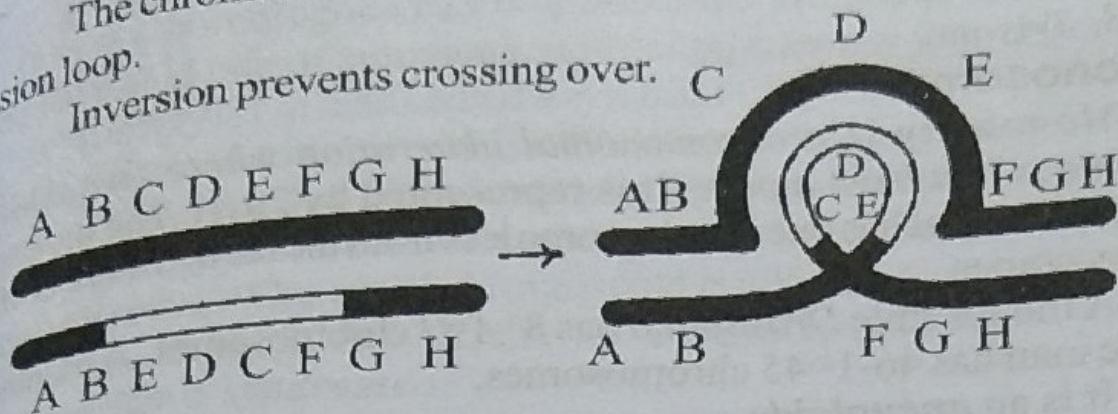


Fig.22.5: Inversion loop.

It brings about position effect.

Inversion produces variation and speciation.

4. Translocation

Translocation is a chromosomal aberration where non-homologous chromosomes exchange segments.

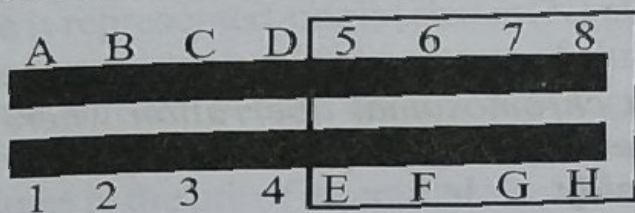


Fig.22.6: Translocation.

Translocation produces a **cross-shaped** structure during pairing.

Translocation causes **position effect**.

Translocation alters the **linkage groups**.

2. Change in the Number of Chromosomes

Change in the number of chromosomes is called **ploidy**.

Ploidy may be due to a loss or gain of a chromosome of a set or changes in the number of chromosome sets. Based on this, there are two kinds of ploidy, namely

1. Aneuploidy
2. Euploidy

1. Aneuploidy

Aneuploidy is a chromosomal aberration where there is a gain or loss of one or more chromosomes in a set. Aneuploidy is caused by **non-disjunction** of chromosomes. It is of three types, namely

1. Monosomy
2. Nullisomy
3. Trisomy

1. Monosomy

Monosomy is a chromosomal aberration where one chromosome is lost from a pair. It is represented by $2n-1$. The monosomic individual has one chromosome less from the normal number of chromosomes.

A monosomic *Drosophila* has $8-1=7$ chromosomes. A monosomic man has $46-1=45$ chromosomes.

It is an **aneuploidy**.

It is produced in woman when an egg without an X chromosome fuses with a sperm containing, an X chromosome. It causes a syndrome called **Turner's syndrome**.

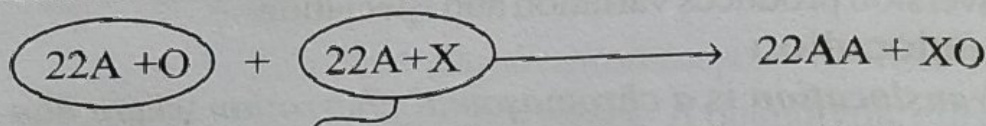


Fig.22.7: Monosomy with the loss of one X chromosome. Turner's syndrome.

2. Nullisomy

Nullisomy is a chromosomal aberration where both chromosomes of a pair are lost. It is represented by $2n-2$.

It is an **aneuploidy**.

A nullisomy is produced by the fusion of gametes having one chromosome less.

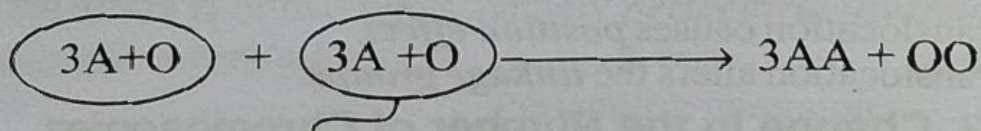


Fig.22.8: Nullisomy with the loss of a pair of chromosomes.

Nullisomic individuals cannot survive.

3. Trisomy

Trisomy is a chromosomal aberration where one chromosome is added to a pair. It is represented by $2n+1$. A trisomic individual has an additional chromosome from the normal number.

Thus a trisomic *Drosophila* has $8+1=9$ chromosomes. A trisomic man has $46+1=47$ chromosomes.

It is an **aneuploidy**.

Trisomy is caused by **non-disjunction**.

There are two types of trisomy, namely **trisomy of autosomes** and **trisomy of sex chromosomes**.

Trisomy of autosome is due to the addition of one chromosome to any one homologous pair of autosome. When a chromosome is added to 21st pair of autosome, it is called *trisomy-21*.

Trisomy-21 in man causes a syndrome called *Down's syndrome* (Mongolism). A trisomic man has 47 chromosomes instead of 46. They are mentally retarded. They have broad face and flat stubby nose.

Trisomy of sex chromosome is due to the addition of one sex chromosome. When an X chromosome is added to a man, he has 47 chromosomes, 22AA+XXY. It causes a syndrome called *Klinefelter's syndrome*.

2. Euploidy

Euploidy is a chromosomal aberration involving change in the number of chromosome sets. It is of two types, namely

1. Haploidy
2. Polyploidy

1. Haploidy or Monoploidy

The basic set of chromosome in any species is haploid; each chromosome is represented singly; that is (N) number.

The gametes carry haploid number of chromosomes. During fertilization the parental chromosomes unite together by the fusion of gametes forming diploid number (2N) of chromosomes.

Sometimes in the life of an animal a set of chromosomes will be lost and this leads to haploidy. So some characters which are present in any parent, will be lost from the resulting individual.

2. Polyploidy

Polyploidy is the condition in which an organism contains more than the usual two sets of chromosomes. Such animals are said to be *polyploid*.

Polyploid organisms may have three, four or more sets of chromosomes and they are called *triploids* (3N); *tetraploids* (4N); *pentaploids* (5N); *hexaploids* (6N) *hectaploids* (7N); *octoploids* (8N); *nanaploids* (9N); *decaploids* (10N) and so on.

Polyploidy may be *autopolyploidy* or *allopolyploidy*. In autopolyploidy, the chromosome sets are derived from the same species so no addition of new genes occurs but in allopolyploidy the chromosome sets are derived from distinct species it involves the addition of new genes hence much variations occur in organisms. These variations are inherited by the offspring which seem to be different from their parents.

Origin of Polyploidy

The polyploid condition arises owing to the abnormalities in cell division. During gametes formation, the homologous chromosomes may not separate completely; hence one cell is formed with the diploid number of chromosomes and the other cell without any chromosome. During fertilization, such a gamete with two sets of chromosomes pair with a gamete having haploid (N) number of chromosome resulting individual is a **Tetraploid** (4N). When both diploid gametes belong to the same species, the resulting tetraploid is called **autotetraploid**. When the two diploid gametes belong to two different species the resulting tetraploid is called **allotetraploid**.

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3. Point Mutation

When a single base pair is altered, the mutation is called **point mutation**.

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d. Base Inversion: In base inversion, the base sequence is reversed.

23. Chromosomal Aberrations (Chromosome Abnormalities) (Genome Mutation)

Chromosome abnormalities are changes resulting in a visible alteration of the chromosomes. Most chromosomal aberrations are produced by *misrepair* of broken chromosomes, by *improper recombination* or by malsegregation of chromosomes during mitosis or meiosis.

Types of Chromosomal Abnormalities

A chromosomal abnormality present in all cells of the body is called *constitutional abnormality*. If it is present in only certain cells or tissues it is called *somatic* or *acquired abnormality*. Constitutional abnormalities are the result of an *abnormal sperm* or *egg* or maybe *abnormal fertilization* or an *abnormal event in the early embryo*.

An individual with a somatic *abnormality* is a *mosaic* containing cells with two different chromosome constitutions, with both cell types deriving from the same zygote.

Chromosomal abnormalities, whether constitutional or somatic, mostly fall into two categories: *numerical abnormalities* and *structural abnormalities*.

Occasionally, abnormalities have been identified in which chromosomes have the correct number and structure, but represent unequal contributions from the two parents.

Numerical Chromosomal Abnormalities

Three classes of numerical chromosomal abnormalities can be distinguished. They are *polyploidy*, *aneuploidy* and *mixoploidy*.

Polyploidy

In human pregnancies, about 3% are *triploids*. The most usual cause is two sperm fertilizing a single egg (*dispermy*); sometimes the cause is a *diploid gamete*. Triploids very seldom survive to term and the condition is not compatible with life.

Chromosomal Abnormalities

