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PRINCIPLES OF INHERITANCE AND VARIATION

Topics in this chapter :

- **MENDEL'S LAW OF INHERITANCE**
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  - Deviations from Mendelism
  - Rediscovery of Mendel's Law
  - Chromosomal Theory of Inheritance
  - Linkage & Recombination.

- **Sex-Determination & Genetic Disorders**
  - Mechanisms of Sex-Determination
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  - Pedigree Analysis
  - Genetic Disorder.

PRINCIPLES OF INHERITANCE AND VARIATION

INTRODUCTION

- The study of heredity, or how the characteristics of living things are transmitted from one generation to the next is called genetics.
- Heredity is the process of transmission of traits from parents to their offspring's either via asexual reproduction or sexual reproduction is called Heredity.
- Individual of same species have some differences is known as Variations.
- Autosome is a chromosome that is not an allosome (a sex chromosome)
- It helps to determine sex, In human only 1 pair of sex chromosomes exists is termed as sex Chromosomes.
- Genes are found on structures called chromosomes, long pieces of DNA wound up around protein.
- Alleles is one of the possible forms of a gene. Most genes have two alleles, a dominant allele and a

recessive allele.

- Homologous chromosomes :- Pair of chromosomes containing maternal and paternal chromatid joined to together at the centromere

- Heterologous chromosomes

- Differ in shape, size or function.
- Do not belong to the same pairs.

GREGOR MENDEL

- Gregor mendel (Father of genetics) start studying inheritance in peas.
- Performed experiments with pea plants for 7 years (1856 - 1863)
- Proposed law of inheritance
- Statistical analysis and mathematical logic used.
- He chose pea plant (pisum sativum)
- He proposed the law of inheritance in living organism.

Why mendel selected pea plant for his experiment ?

- Pea plant was easy to cultivate. It grew well in his garden
- Its flowers were hermaphrodite
- It is self pollinated in nature.
- Cross pollination is easy to be done artificially.
- It Completes its lifecycle in one season.
- Pea had many sharply distinct. its each trait had two clear cut alternative varieties.

Seven Different Traits

He traits that Mendel studied are listed below.

- Form of ripe seed (R) – Smooth or wrinkled
- Color of seed albumen (Y) – yellow or green
- Color of flower (P) – Purple or white
- Form of ripe pods (I) – Inflated or Constricted
- Color of unripe pods (G) – green or yellow
- Position of flowers (A) – axial or terminal
- Length of stem (T) – tall or dwarf.

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MENDEL'S OBSERVATION

- Cross pollination between tall & dwarf pea plant.
- In F_1 Generation all the off springs were tall.
- Phenotypic ratio of pea plant is tall.
- Genotype of offsprings was different.

REASON FOR MENDEL'S SUCCESS

- Mendel applied statistical method and mathematical logic for analyzing his results.
- He kept accurate records of his experiment
- Mendel experimented on a number of plants for the same trait and obtained hundreds of offspring.
- He tried to formulate theoretical explanation for the observed result.

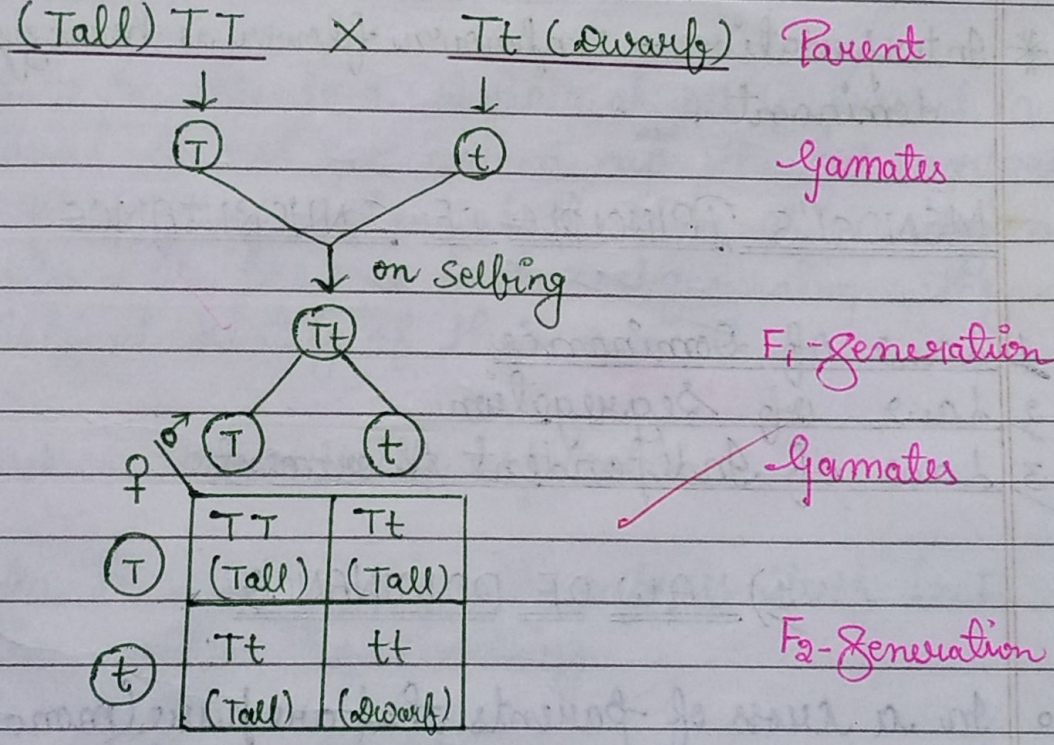
PUNNETT SQUARE

- It was developed by British geneticist Reginald Punnett.
- A punnett square is a graphical representation

of the possible genotypes of an offspring arising from a particular cross or breeding event.

- Some terms are often used in the study of genetics and these are particularly useful in understanding the function of Punnett squares. Among these is a term 'allele' and is used to denote a variant of a gene.

- for e.g., a pea plant can have red or white flowers and the gene variant coding for each of these is called an allele.



Phenotypic Ratio — Tall Palants : Dwarf plant
3 : 1

Genotypic Ratio — TT : Tt : tt
1 : 2 : 1

Fig - A Punnett square used to understand a typical mono hybrid cross conducted by Mendel b/w tall & dwarf plant (True-breedi

TEST CROSS

- It is the cross that is made to know whether a trait is homozygous (TT) or heterozygous (Tt) dominant by its cross with recessive trait.
- In this cross, the organism with unknown dominant genotype is crossed with recessive parent.
- This cross find out the homozygous and heterozygous nature of the genotype.

* Interpretation → unknown flower is homozygous dominant.

MENDEL'S PRINCIPLE OF INHERITANCE

1. Law of Dominance
2. Law of Segregation
3. Law of Independent Assortment

1) LAW OF DOMINANCE

- In a cross of parents that are pure (monohybrid) for contrasting trait, only one form of the trait will appear in the next generation. Offspring that are hybrid for a trait will have only the dominant trait in the Phenotype.
- Offspring that are hybrid for a trait will have only dominant trait in the Phenotype.

- so, if there exists two contrasting traits, one of the traits will always suppress the other, thereby expressing itself, T suppresses, t, thus making the offspring plant tall.
- Such a trait is known as a dominating trait. The suppressed trait is known as Recessive trait.
- Also, the recessive trait freely express itself in the absence of the dominant state. And this is what Mendel's law of dominance is all about.

2) LAW OF SEGREGATION

- According to this law. A pair of allele present in an individual do not get mixed up. They get segregate or regret from each other at the time of gametogenesis and express its presence in the next generation. They still have distinct identity of their own.
- This law is universelly applicable.
- fo ex- In Tt allele, Both will express ilself in F_2 generation when F_1 is selfing.

3) LAW OF INDEPENDENT-ASSORTMENT

- This law states that when two pairs of traits are combined in a hybrid, segregation of one pair of character is independent of the other pair of characters at the time of gamete formation.

Round yellow
RRYy

Wrinkled Green
rrYy

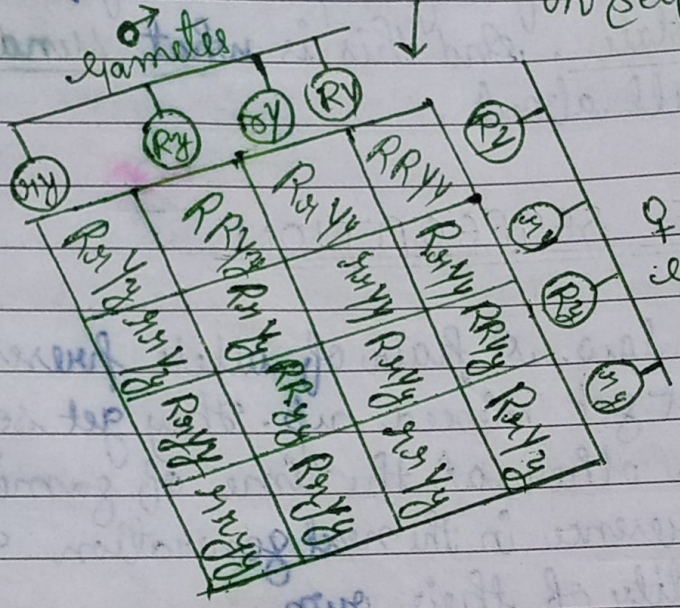
Parents

(RY) × (rY)

Gametes

RrYy
Round yellow
on selfing

F_1 - Generation



F_2 - Generation

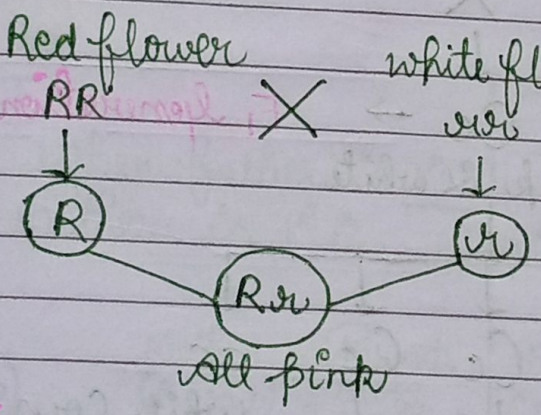
Phenotypic Ratio = 9:3:3:1
Genotypic = 1:2:2:4:1:2:1:2:1

Fig - Result of dihybrid cross where the two parents differed in two pairs of contrasting traits seed colour and seed shape.

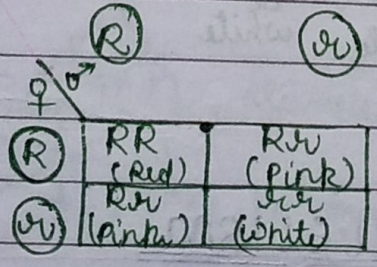
EXCEPTIONS TO PRINCIPLE OF DOMINANCE AND PRINCIPLE OF PAIRED FACTORS

INCOMPLETE DOMINANCE

- A few cases were observed where F_1 is intermediate of both Dominant and recessive Phenotype.
- In incomplete dominance non of the allele whether it is dominant or recessive is able to express itself completely when present in heterozygous condition.
- For e.g - Flowers in Snapdragon (dog flower) or Mirabilis jalapa (4 o'clock plant) where red colour is due to gene RR white colour is due to gene rr and pink colour is due to gene Rr.



Rr X Rr — on Selfing



F2 - Generation

Phenotypic Ratio Red : Pink : white
(RR) : (Rr) : (rr)

Genotypic Ratio 1 : 2 : 1

CO-DOMINANCE

- In co-dominance both the alleles of heterozygous condition are able to express itself independently.
- There is no dominant and recessive relationship between both the allele in heterozygous condition.

e.g

Black bull CC × white cow CC — Parent

↓

Cc → F_1 generation

Black & white

↓

CC (Black Bull) | Cc (Black & white) | Cc (Black & white) | cc (white cow) → F_2 gener...

MULTIPLE ALLELISM

- Mendel proposed that each gene has two

contrasting forms i.e, Alleles, But there are some genes which are having more than two alternative forms.

- Presence of more than two alleles for a gene is known as multiple allelism and each are able to express itself.

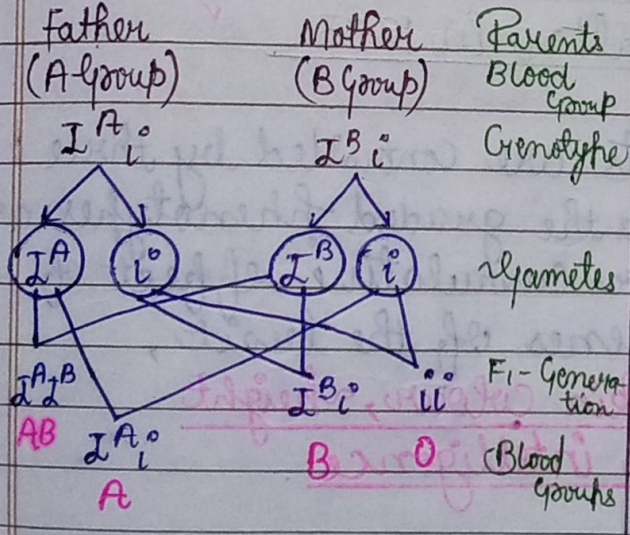
- Example – Different types of red blood cells that determine ABO blood grouping in human beings. ABO blood grouping in human beings. ABO Bloods groups are controlling by gene I.

The gene I has three Alleles.
i.e I^A, I^B, i^o

EXAMPLE

CASE I

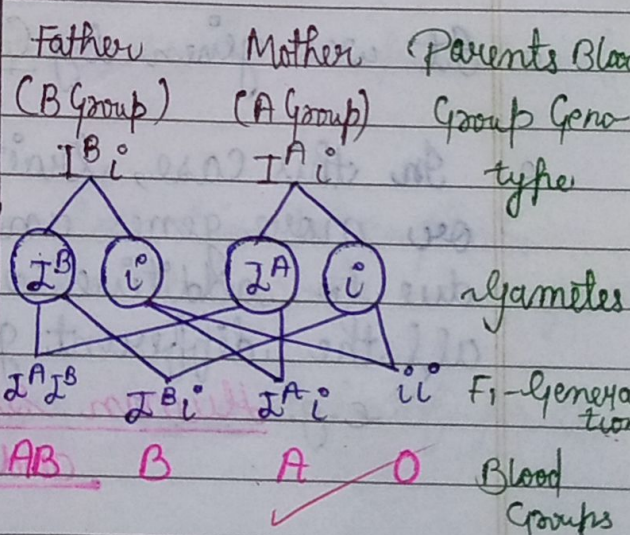
When father is $I^A i$ and mother is $I^B i$.



* Offsprings will have the above possible blood Groups.
i.e. AB, A, B and O

CASE II

When father is $I^B i$ and mother is $I^A i$



Offsprings will have the above possible blood Groups also.
i.e. AB, B, A and O

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PLEIOTROPY

* It is the phenomenon in which a single gene product may produce multiple or more than one phenotypic effect.

- A pleiotropic gene often has a more evident effect on one trait called the major effect.
- and less evident effects on other traits called Secondary effects.

No. of related changes are caused by a pleiotropic gene, they are called Syndrome.

e.g. of pleiotropy are- Phenylketonuria (PKU)
Drosophila

POLYGENIC INHERITANCE

- It was given by Galton in 1833.
- In this case, traits are controlled by three or more genes and the graded phenotypes are due to additive or cumulative effect of all the different genes of the trait,
  e.g. Human skin colour, height and intelligence.

- Polygene is a gene where one dominant allele controls only a unit or partial quantitative expression of a trait.

- Skin colour in humans is caused by a pigment called melanin.

✱ COMPLEMENTARY GENES

These are two pairs of non-allelic dominant gene which interact to produce only one phenotypic, dominant trait but neither of them produces the trait in absence of other.

REDISCOVERY OF MENDEL'S LAWS

Mendel published his work on inheritance of characters in 1865. It remained unrecognised for several reasons till 1900. Some of them are follows.

i) Communication was difficult, so his work could not be widely publicised.

ii) His approach of using mathematics to explain biological phenomenon was new and unacceptable.

iii) He could not provide any physical proof for the existence of factors.

In 1900, de Vries, Correns ~~Von~~ and Von Tschermark rediscovered Mendel's results.

independently.
Due to microscopy, they carefully observed cell division. This led to discovery of chromosomes.

CHROMOSOMAL THEORY OF INHERITANCE

It was proposed by Walter Sutton and Theodore Boveri in 1902.
According to this theory.

i) All hereditary characters must be with sperms and egg cells as they provide bridge from one Generation to the other.

ii) The hereditary factors must be carried by the nuclear material.

iii) Chromosomes are also found in pairs.

iv) The two alleles of a gene pair are located on homologous site on the homologous chromosomes.

v) The sperms & eggs have haploid sets of chromosomes, which fuse to re-establish the diploid state.

vi) The genes are carried on the chromosomes.

vii) Homologous chromosomes Synapse during meiosis and get separated to pass into different cells.

this is the basis for segregation & independent assortment.

Experimental verification of Chromosomal Theory of Inheritance

It was done by Thomas Hunt Morgan & his colleagues.

He observed that the two genes under consideration in his experiments, did not segregate independently as in the case of characters studied by Mendel.

* Morgan Selected fruit-fly, Drosophila Melanogaster for his experiment because.

i. They could be grown on simple artificial medium in the laboratory.

ii. Their life cycle is only about two weeks.

iii. A single mating could produce a large number of flies.

iv. It has 4 pairs of chromosomes which differ in size.

v. There was a clear differentiation of the sexes.

vi. It has many type of hereditary Variation which can be easily seen through low power microscope.

LINKAGE AND RECOMBINATION

Mendel's Law of independent assortment was not taking place in this case. From this Morgan got the idea of linkage.

They attributed due to physical association of the two genes and coined the term linkage to describe physical association of gene on a chromosome & the term 'recombination' to describe the generation of non-parental gene combination.

Linkage is a phenomenon of genic inheritance in which genes of a particular chromosome show their tendency to inherit together.

If genes were grouped on the same chromosome some genes were tightly linked. i.e.. linkage is stronger b/w two genes, if the frequency of recombination is low whereas frequency of recombination is higher if genes are loosly linked i.e. linkage is weak b/w two genes.

LINKAGE GROUPS

All the genes linked together in a single chromosome constitute a linkage group. The number of linkage group in an organism is equal to their haploid number

of chromosomes.

Chromosomes Maps or Linkage Maps.

Alfred Sturtevant (Morgan's Student) used the frequency of recombination b/w gene pairs on the same chromosome as a measure of the distance b/w genes & 'mapped' their position on the chromosome.

Genetic maps are now used as a starting point in the sequencing whole genomes as done in case of human genome sequencing project.

Sex-Determination & Genetic Disorders

MECHANISMS OF SEX-DETERMINATION

The establishment of sex through differential development in an individual at the time of zygote formation is called Sex-determination.

Henking in 1891 could trace a specific nuclear structure all through spermatogenesis in few insects. He named this structure as X-body. Scientists further explained that X-body was a chromosome and called as X-chromosome.

The chromosomal theory of sex-determination was worked out by EB Wilson and Stevens (1902-1905). They named XY chromosomes as allosome or Sex chromosomes.

✱ And other chromosomes which have no relation with sex & contain genes, which determine the somatic characters as Autosomes (A.A)

i Homogametic, i.e, Producing similar gametes.

ii Heterogametic, i.e, Producing different gametes.

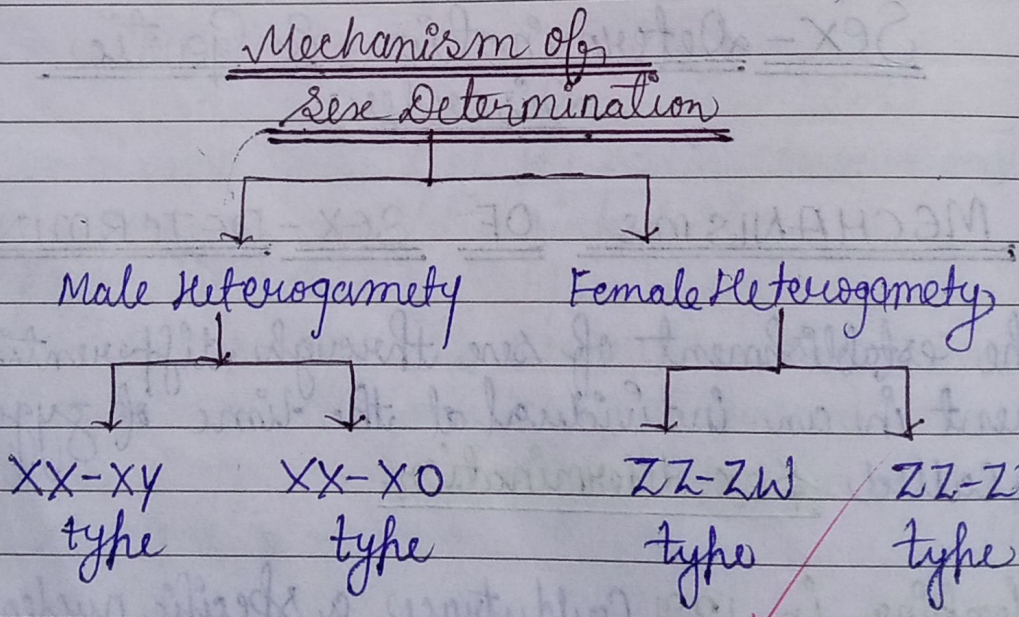


Fig- Mechanism of Sex determination

Male Heterogametey

Two different types of gametes.

i. either with or without X-chromosome

ii. Some gamete with X-chromosome & Some without Y-chromosome.

I XX-XY type (insects like Drosophila)

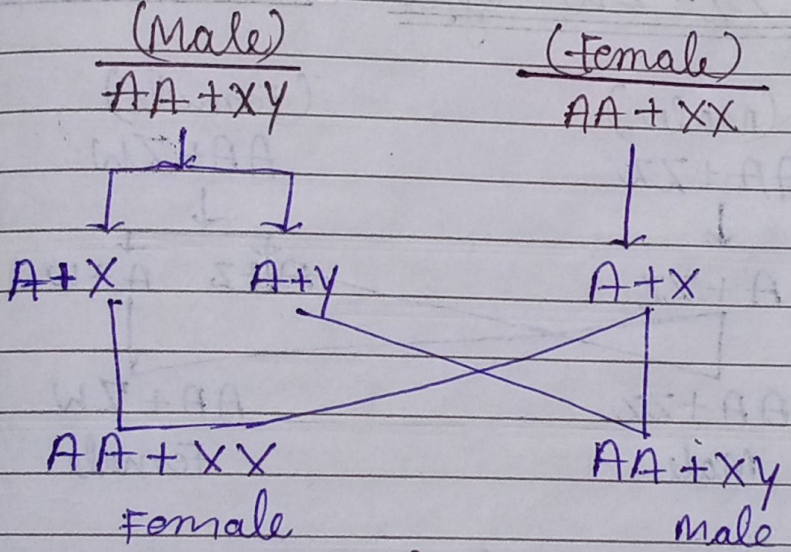
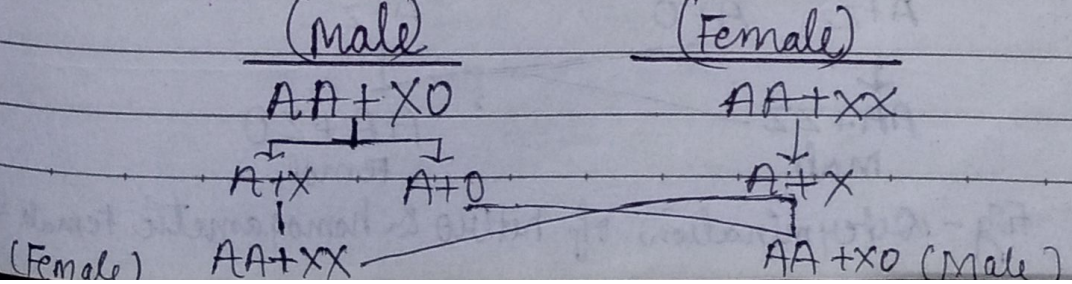


Fig- determination of Sex in Drosophila

The presence of y-chromosomes determines the maleness

II XX-XO Type (insects like grasshopper)



Eggs fertilised by sperms having an X-chromosome become females & those fertilised by sperms which do not possess X-chromosome become males.

FEMALE HETEROGAMETY

In this case, the total number of chromosomes are same in both males & females. But two different types of gametes in term of sex-chromosomes are produced by females

I ZZ-ZW Type (seen in birds, fowls & fish)

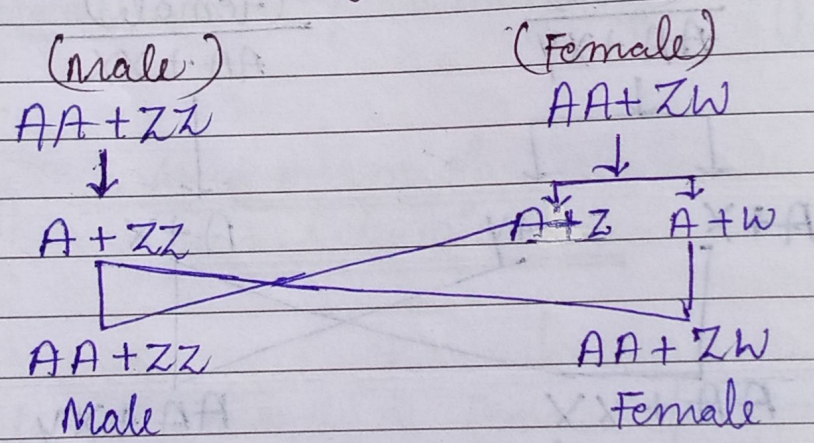


Fig- determination of sex in birds

II ZZ-ZO Type

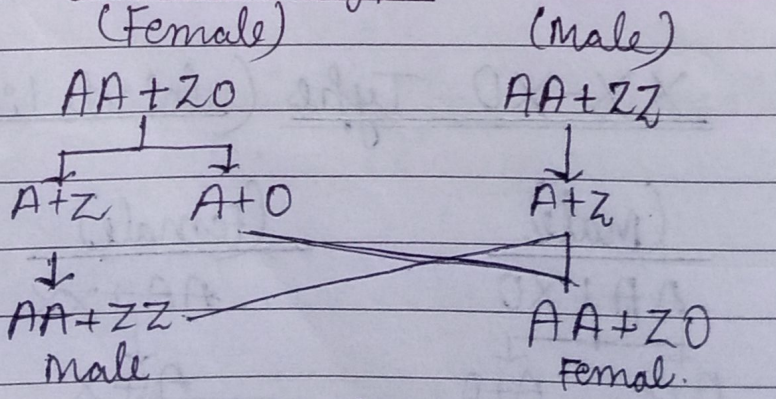
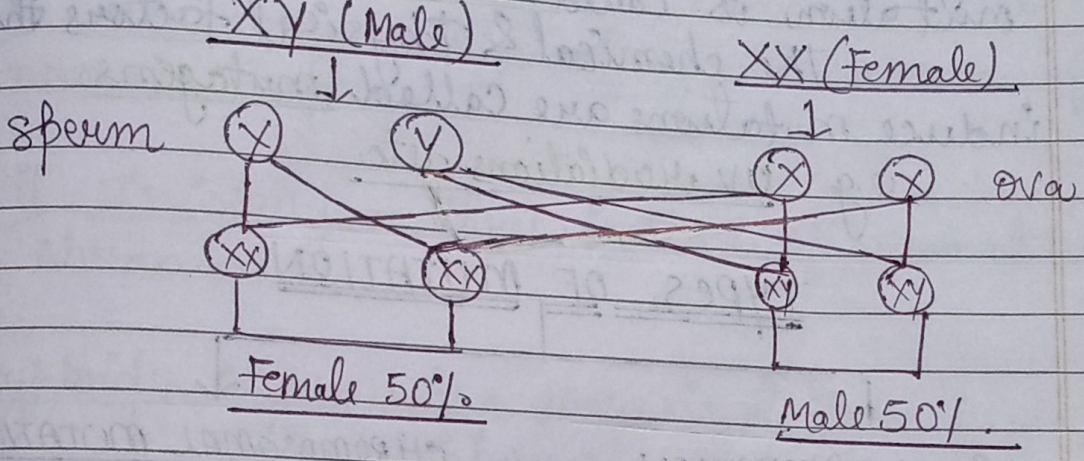


Fig- Determination of hetero & homogametic female &

SEX-DETERMINATION IN HUMANS

As it has been already mentioned that the sex determining mechanism in case of humans is XX-XY type.



There are 50% chances of having either a male or female in each pregnancy.

Sex-Determination in Honeybee (Haploid-Diploid Method)

In Insects like honeybees and ants, sex chromosomes are not differentiated & sex is determined on the basis of Ploidy of the individual.

In honeybees, drones are males & are haploid ($n=16$). The females are diploid ($2n=32$).

Therefore in them sex-determination is referred as haploid-diploid method.

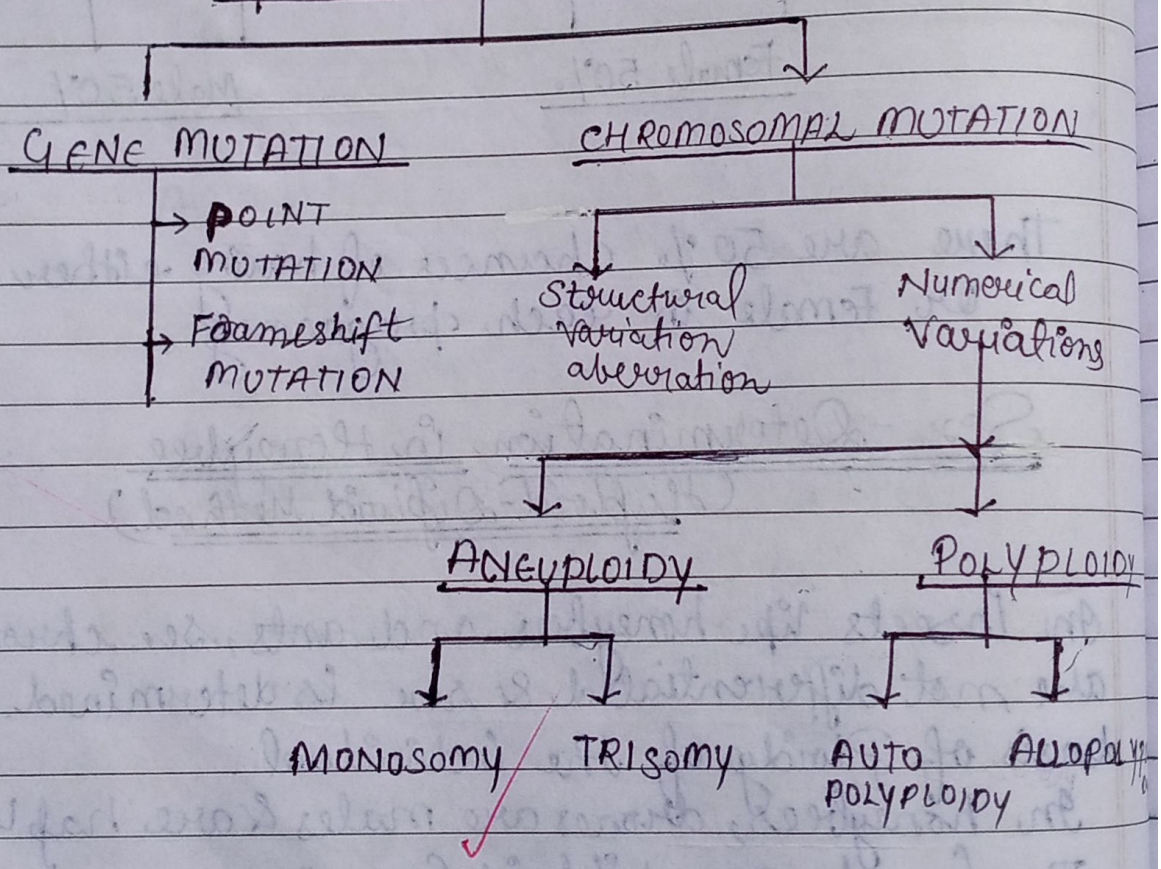
MUTATION

It is a sudden, stable and inheritable change in genetic material or DNA sequences of an organism. The organism, which undergoes mutation is called mutant.

The chemical & physical factors that induce mutations are called mutagens,

e.g. UV radiations etc.

TYPES OF MUTATION



- Mutation occuring due to change in a single base pair of DNA. this is called Point mutation.

- Deletions & insertions of base pair of DNA Cause Frameshift mutation
- when number of a homologous pair of chromosomes fail to segregate during meiosis, Aneuploidy occurs.

  i. Monosomy - lack of one chromosome of normal complement.

  ii Trisomy - three instead of normal two chromosomes.

- Polyploidy occurs when there is failure of cytokinesis after telophase stage of cell division resulting in an increase in a whole set of chromosomes in an organism.

  i Autopolyploids are polyploids with multiple chromosome sets derived from a single species.

  ii Allopolyploids is where chromosomes are derived from different species, i.e, result of multiplying the chromosome number & forming a hybrid.

PEDIGREE ANALYSIS

The analysis of trait in several generation of a human family in the form of a family or diagram is called Pedigree Analysis.

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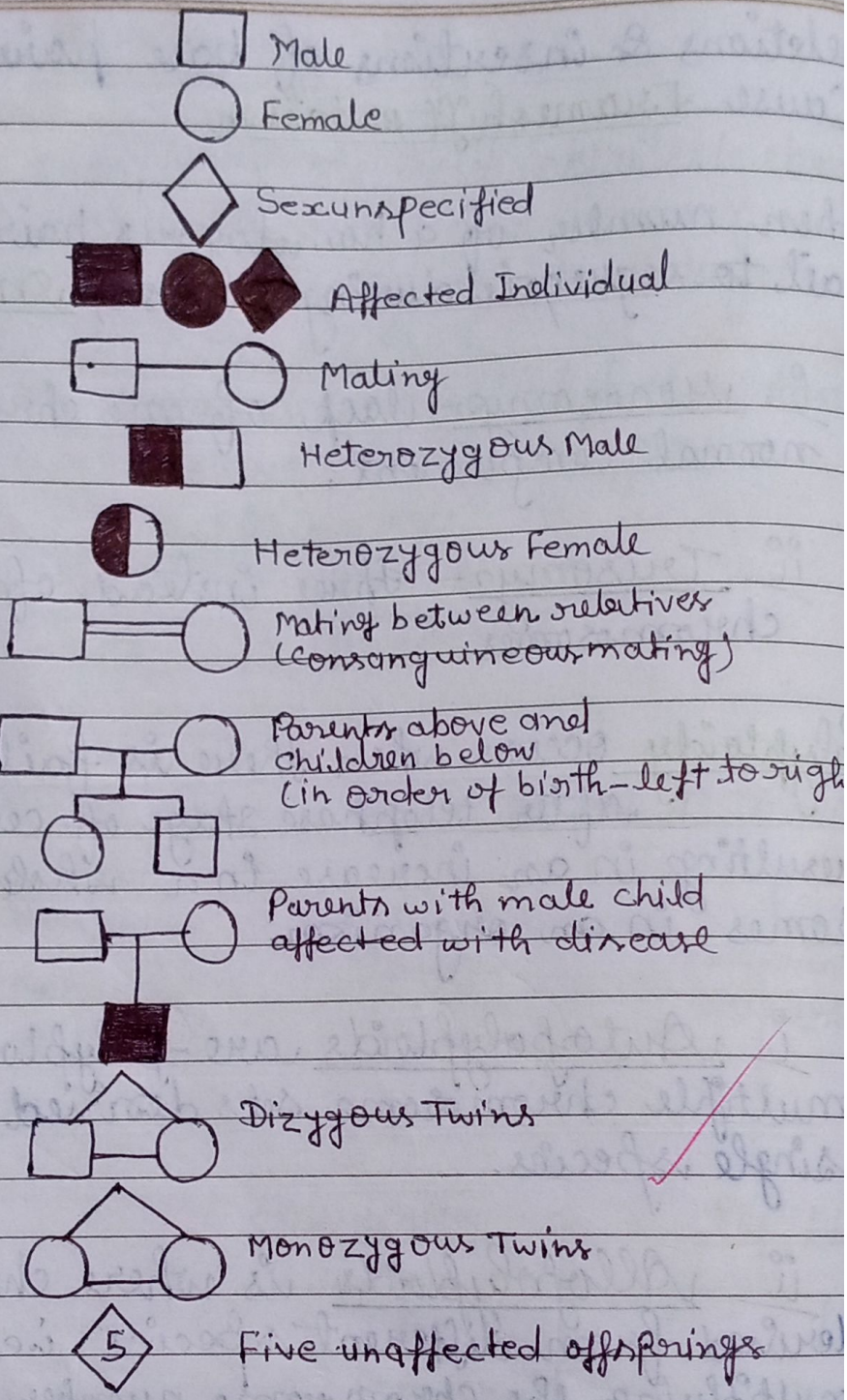


Fig - Symbols used in the human Pedigree analysis

GENETIC DISORDERS

These are disorders or illness caused by one

or more abnormalities in the autosomes or sex chromosomes of the person. Thus referred to as autosomal disorders. or

Genetic Disorders

- Mendelian Disorders — e.g. Colour blindness, Thalassemia etc.
- Chromosomal disorders — e.g. Turner's Syndrome, Down's Syndrome etc.

2. **Mendelian Disorders**

These are mainly determined by alteration or mutation in a single gene. These are transmitted to next generation according to the principle of inheritance.

These can be dominant or recessive as follows

i Autosomal dominant, e.g. Huntington disease

ii Autosomal recessive, e.g. thalassemia

iii Sex-linked dominant, e.g. defective tooth enamel

iv Sex-linked recessive, e.g colour blindness.

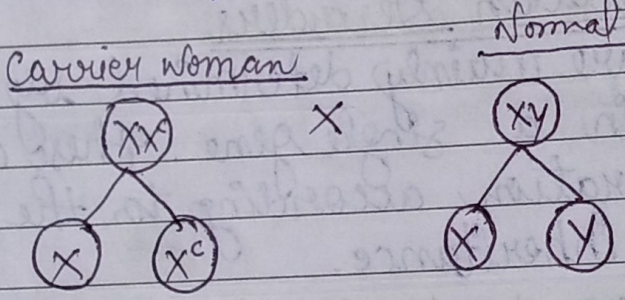
Haemophilia

It is a sex-linked recessive disease, which is

transmitted from an unaffected carrier female to some of the male offsprings. Due to this, patient continues bleeding even to a minor injury because of defective blood coagulation, thus also called bleeder's disease.

Colour Blindness

It is a sex-linked recessive disorder, which results in defects in either red or green cone of eye. It does not mean not seeing any colour at all, infact it leads to the failure in discrimination b/w red & green colour.



♀ \ ♂	X	Y
X	XX (Normal girl)	XY (Normal boy)
X^c	XX^c (carrier girl)	X^cY (Colourblind boy)

Sickle-Cell Anaemia

It is an autosomal linked recessive trait that can be transmitted from parent to the

offsprings, when both the partners carrier for the gene.

This disease is controlled by single pair of allele Hb^A and Hb^S.

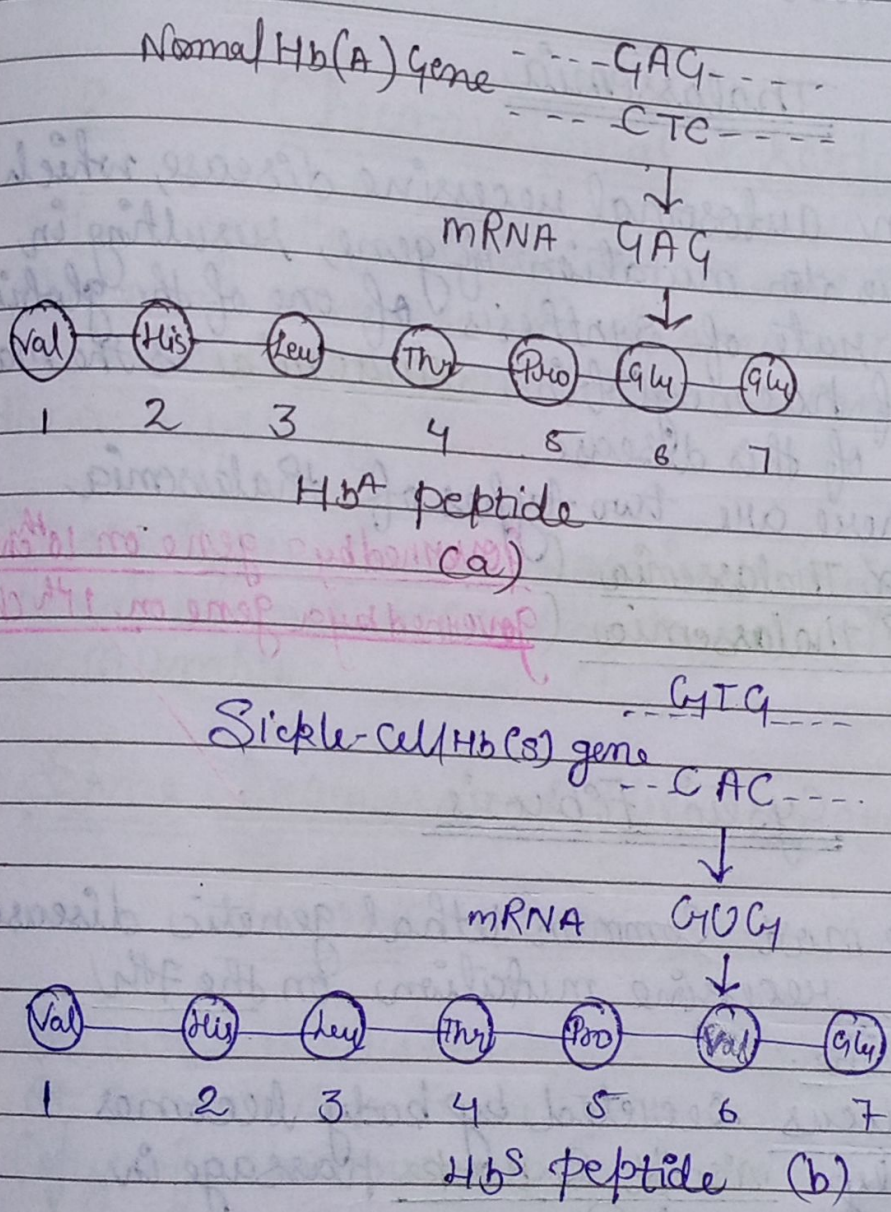


Fig - The amino acid composition of the relevant portion of β-chain of haemoglobin:
(a) From a normal individual;
(b) From an individual with sickle-cell anaemia.

It is caused by the substitution of Glutamic acid (Glu) by Valine (Val) at the Sixth position of the β globin chain of the haemoglobin molecule.

Thalassemia

It is an autosomal recessive disease, which occurs due to mutation of gene, resulting in reduced rate of synthesis of one of the globin chains of haemoglobin. Anaemia is the main feature of this disease.

There are two types of thalassemia

i. α thalassemia (governed by a gene on 16th chromosome)
ii. β thalassemia (governed by a gene on 11th chromosome)

Cystic Fibrosis

It is the most common lethal genetic disease due to a recessive mutation on the 7th chromosome.

The mucus secreted by body becomes abnormally viscid & blocks passage in lungs, liver and Pancreas.

Phenylketonuria (PKU)

It is an inborn error of metabolism.
Its gene is associated to 12th chromosome

It is a rare disease in which individual lack an enzyme called Phenylalanine hydroxylase, which is needed to breakdown an essential amino acid Phenylalanine into tyrosine in liver.

2- Chromosomal Disorders

These are caused by the absence, excess or abnormal arrangement of one or more chromosomal. The chromosomal disorders can be studied by the analysis of Karyotypes.

✓ Karyotypes is an organised profile of a person's chromosomes according to their shape, size & number.

Some chromosomal disorders are discussed below

Down's Syndrome

It was described by J Langdon Down in 1866
It occurs due to the presence of an additional copy of chromosome in humans.
It is also seen in chimpanzees & other related primates.

Symptoms

i. Individuals are short statured with small, round head & furrowed tongue & partially open

mouth.

ii) Palm is broad with characteristic palm crease.

iii) Slow mental, Physical & Psychomotor development.

Turner's Syndrome

It is a disorder which is caused due to the absence of one of the X-Chromosome i.e. 45 with XO.

Symptoms

i) Affected females are sterile as ovaries are rudimentary.

ii) Lack of secondary sexual characters & poor breast's development.

iii) Short stature, small uterus, puffy fingers & webbed neck.

Klinefelter's Syndrome

It occurs due to the presence of an additional copy of X- Chromosome resulting in the Karyotype of 47, XXY in males or 47, XXX in females.

TE - The extra inactive X-chromosome
karyotype of Klinefelter Syndrome is cal
Barr body.
Thus, XXY individual have single Barr
whereas XXX individual have two Barr

Symptoms

ver beard growth and feminine pitch
ice.

uch nuclear individuals are sterile

dividuals have masculine development b
eminine characters like development of