

REVISION NOTES (XII)

ZOOLOGY



PRINCIPLES OF INHERITANCE AND VARIATION

Genetics: deals with the inheritance, as well as the variation of characters from parents to offsprings.

Inheritance: is the process by which characters are passed on from parent to progeny.

Variation: is the degree by which progeny differ from their parents.

MENDEL'S LAWS OF INHERITANCE:

Gregor Mendel. Conducted hybridization experiments on garden peas for seven years (1856 – 1863) and proposed laws of inheritance. Mendel conducted artificial pollination/cross pollination experiments using several true-breeding pea lines.

Characters of Pea plants selected by Mendel

S.No	Characters	Contrasting Traits
1.	Stem height	Tall/dwarf
2.	Flower colour	Violet/white
3.	Flower position	Axial/terminal
4.	Pod shape	Inflated/constricted
5	Pod colour	Green/yellow
6.	Seed shape	Round/wrinkled
7.	Seed colour	Yellow/green

INHERITANCE OF ONE GENE:

Mendel crossed tall and dwarf pea plants to study the inheritance of one gene. He collected the seeds produced as a result of this cross and grew them to generate plants of the first hybrid generation. This generation is called filial progeny or the F1.

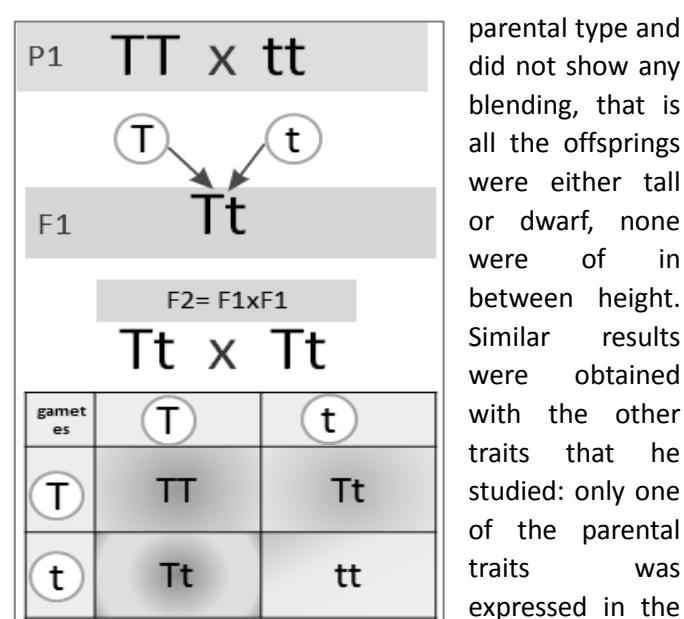
Mendel observed that all the F1 progeny plants were tall, like one of its parents; none were dwarf.

He made similar observations for the other pairs of traits – he found that the F1 always resembled either

one of the parents, and that the trait of the other parent was not seen in them.

Mendel then self-pollinated the tall F1 plants and to his surprise found that in the F2 generation some of the offsprings were dwarf; the character that was not seen in the F1 generation was now expressed. The proportion of plants that were dwarf was 1/4th of the F2 plants while 3/4th of the F2 plants were tall.

The tall and dwarf traits were identical to their parental type and did not show any blending, that is all the offsprings were either tall or dwarf, none were in between height.



Similar results were obtained with the other traits that he studied: only one of the parental traits was expressed in the F1 generation while at the F2 stage both the traits were expressed in the proportion of 3:1.

Mendel's proposition:

Mendel proposed that something was being stably passed down, unchanged, from parent to offspring through the gametes, over successive generations. He called these things as 'factors'. Now a day we call them as genes.

***Term gene was given by Johannsen.

Gene is therefore are the units of inheritance.

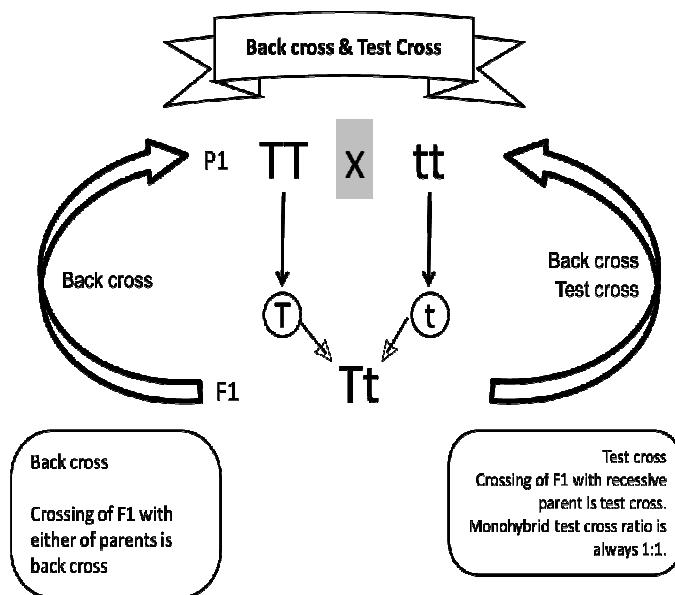
- Genes which codes of a pair of contrasting traits are known as alleles, i. e. they are alternate forms of the same gene.

Test cross:

When F₁ hybrid is crossed back with the recessive parent, it is known as test cross.

It is used to know the genotype of the given plant/animal.

Test cross ratio is 1:1

**Law of Dominance:**

Characters are controlled by discrete units called factors. Factors occur in pairs.

In a dissimilar pair of factors one member of the pair dominates (dominant) the other (recessive).

Law of Segregation:

During gamete formation; the factors or alleles of a pair segregate or separate from each other such that a gamete receives only one of the two factors.

Incomplete dominance:

When hybrid phenotype does not resemble either of the two parents and was in between the two, the condition is called incomplete dominance.

Inheritance of flower color in the dog flower (snapdragon or Antirrhinum sp.) is a good example of incomplete dominance.

In incomplete dominance the phenotypic and genotypic ratio are same ie. 1:2:1.

Co – dominance:

F₁ generation resembles both parents side by side is called (co-dominance).

Eg. AB blood group in man. Here the two dominant genes (gene A and gene B) expresses together.

Multiple Alleles:

Condition in which more than a pair of allele (genes) control a single character.

Example of ABO blood grouping in man

There are more than two i. e. three allele, control the same character (Blood group)

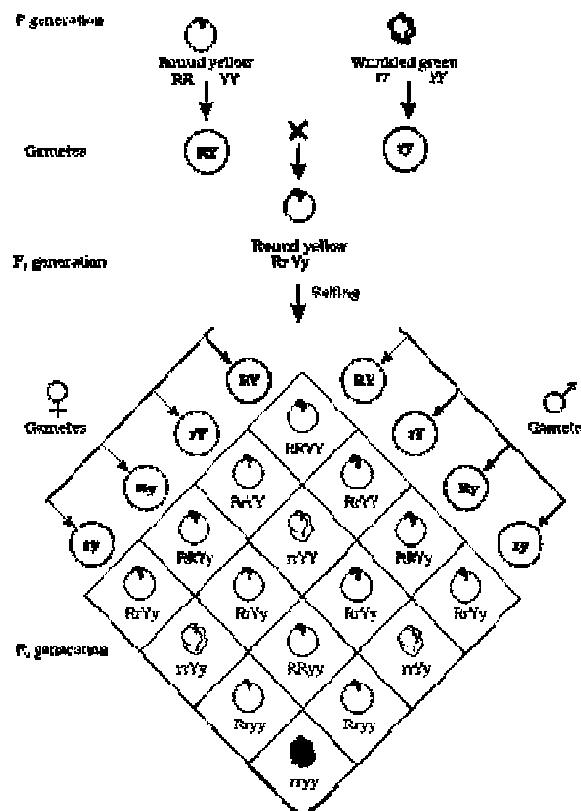
They are IA, IB and i. Each person possesses any two of the three I gene alleles.

I A and I B are completely dominant over i.

When I A, and I B present together they both express as in AB blood group

INHERITANCE OF TWO GENES:

Dihybrid Cross Ratio is 9:3:3:1

**Law of independent Assortment:**

When two characters (dihybrid) are combined in a hybrid, segregation of one pair of traits is independent of the other pair of traits.

Reasons for unrecognition of Mendel's concepts

1. Communication was not easy in those days and his work could not be widely publicized.
2. His concept of genes (or factors, in Mendel's word) as stable and discrete units that controlled the expression of traits and of the pair of alleles which did not 'blend' with each other, was not accepted by his contemporaries as an explanation for the apparently continuous variation seen in nature.
3. Mendel's approach of using mathematics to explain biological phenomena was totally new and unacceptable to many of the biologists of his time.
4. He could not provide any physical proof for the existence of factors.

Rediscovery of Mendel's result:

In 1900 three scientists (deVries, Correns and von Tschermak) independently rediscovered Mendel's result on the inheritance of character.

CHROMOSOMAL THEORY OF INHERITANCE:

Proposed by Walter Sutton and Theodore Bovery in 1902.

1. Genes present in chromosomes are the vehicles of inheritance
2. Both chromosome and genes are present in pairs in diploid cells.
3. Homologous chromosomes separate during gamete formation (meiosis)
4. Fertilization restores the chromosome number to diploid condition.
5. Chromosomes segregate and assort independently.

***Experimental verification of chromosomal theory of inheritance by Thomas Hunt Morgan and his colleagues. Morgan worked with tiny fruit flies, *Drosophila melanogaster*.

Drosophila is an excellent genetic material -Reasons

- Suitable for genetic studies.
- Grown on simple synthetic medium in the laboratory.
- They complete their life cycle in about two weeks.

- A single mating could produce a large number of progeny flies.
- Clear differentiation of male and female flies
- Have many types of hereditary variations that can be seen with low power microscopes.

Morgan's experiments

Morgan hybridized yellow bodied, white eyed females to brown-bodied, red eyed male and intercrossed their F1 progeny. He observed that the two genes did not segregate independently of each other and the F2 ratio deviated very significantly from 9:3:3:1 ratio (expected when the two genes are independent).

When two genes in a dihybrid cross were situated on the same chromosome, the proportion of parental gene combinations was much higher than the non-parental type. Morgan found that this due to the physical association or linkage of the two genes and coined the term linkage.

Linkage: physical association of genes on a chromosome.

Due to linkage genes fail to segregate, and thus the linked genes inherit together throughout generations.

Recombination: the generation of non-parental gene combinations.

Morgan found that even when genes were grouped on the same chromosome, some genes were very tightly linked (showed very low recombination) while others were loosely linked (showed higher recombination).

The genes white and yellow were very tightly linked and showed 1. 3 percent recombination.

The genes white eye and miniature wing showed 37. 2 percent recombination, hence loosely linked.

Alfred Sturtevant used the frequency of recombination between gene pairs on the same chromosome as a measure of the distance between genes and 'mapped' their position on the chromosome.

SEX DETERMINATION:

***Henking (1891) discovered by X-chromosome.

XX-XO type.

Eg. Sex-determination of grass hopper:

All egg bears one 'X' chromosome along with autosomes. Some sperms (50%) bear's one 'X' chromosome and 50% do not. Egg fertilized with sperm (with 'X' chromosome) became female (AA+XX).

Egg fertilized with sperm (without 'X' chromosome) became male (AA + X0)

XX-XY type

Eg. Sex determination in some insects and mammals
Both male and female has same number of chromosomes.
Female have autosomes and a pair of X chromosomes. (AA+ XX)
Male have autosomes and one large 'X' chromosome and one very small 'Y'-chromosome. (AA+XY)
This is called male heterogamety and female homogamety.

ZZ-ZW type

Eg. Sex determination in birds:
Female birds have two different sex chromosomes designated as Z and W.
Male birds have two similar sex chromosomes and called ZZ.
Such type of sex determination is called female heterogamety and male homogamety.

Autosomal Dominant:

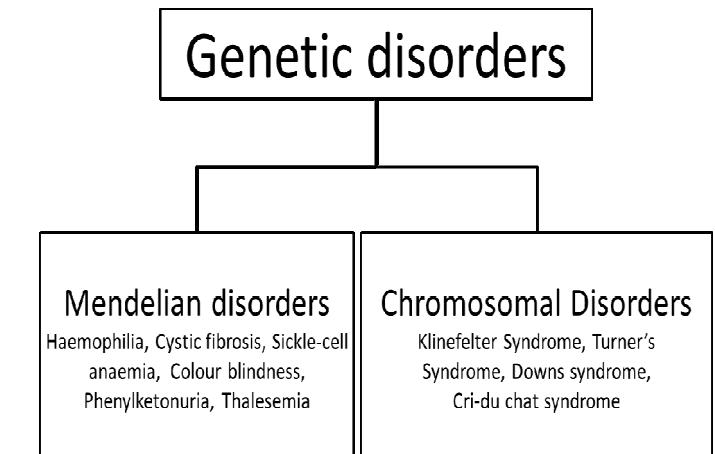
E.g. polydactyly, tongue rolling ability etc

Autosomal recessive:

E.g. - Albinism, sickle cell anaemia

GENETIC DISORDERS

- Genetic disorders grouped into two categories

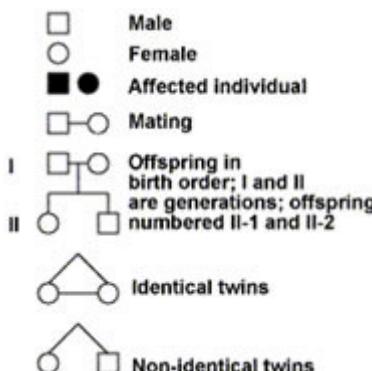


MUTATION

Mutations are sudden heritable genetic changes. It results in alteration of DNA sequences and consequently results in changes in the genotype and phenotype of an organism. Loss (deletion) or gain (insertion/duplication) of a segment of DNA results in alteration in chromosomes. Alteration in chromosomes results in abnormalities or aberration.

- Chromosomal aberrations are commonly observed in cancerous cells.
- Mutations also arise due to change in a single base pair of DNA. This is known as point mutation. E.g. sickle cell anemia.
- Deletion and insertions of base pairs of DNA causes frame shift mutations.

Pedigree Analysis:



Analysis of traits in several of generations of a family is called the pedigree analysis.

In the pedigree analysis the inheritance of a particular trait is represented in the family tree over generations.

Mendelian disorders & Chromosomal disorder

Mendelian disorders are mainly determined by alteration or mutation in the single gene.

Transmission occurs in Mendelian patterns. Transmission can be predicted. Can be expressed in pedigree analysis.

E.g. Haemophilia, colorblindness, Cystic fibrosis, Sickle cell anemia, Phenylketonuria, Thalassemia etc.

Mendelian Disorders

Hemophilia:

In this disease a single protein (enzyme) that is needed for clotting of blood is not produced. Due to this in an affected individual a simple cut will result in non-stop bleeding.

- Sex linked recessive disease.
- The diseases transmitted from unaffected carrier female to some of the male progeny.
 - Female becoming hemophilic is extremely rare because mother of such a female at least carrier and the father should be hemophilic.
 - Affected transmits the disease only to the son not to the daughter.
 - Daughter can receive the disease from both mother and father.

Sickle cell anaemia:

- Genetic disorder due to mutation in DNA (point Mutation) characterised by sickling of RBC. Shape of the RBC changes from circular to sickle shape.
- The defect is caused due to substitution of Glutamic acid (Glu) by Valine (Val) at the sixth position of the beta globin chain of the haemoglobin molecule.
- Substitution of amino acid takes place (single base substitution) at the sixth codon of the beta globin gene from GAG to GUG.
- The mutant haemoglobin molecule undergoes polymerization under low oxygen tension causing the change in the shape of the RBC from biconcave disc to elongated sickle like structure.
- This is an autosome linked recessive trait.
- Transmitted from parents to the offspring when both the parents are carrier for the gene (heterozygous).
- This disease is controlled by single pair of allele, Hb^A, and Hb^S.
- There are three possible genotypes (HbAHbA, Hb^AHb^S, and Hb^SHb^S).
- Only homozygous individuals for HbS (HbSHbS) show the diseased phenotype.
- Heterozygous (Hb^AHb^S) individuals appear apparently unaffected but they are carrier of the disease.

Phenylketonuria:

- Autosomal recessive trait.
- Inborn error of metabolism.
- The affected individual lack one enzyme called phenyl alanine hydroxylase that converts the amino acid phenyl alanine to tyrosine. In the absence of the enzyme phenyl alanine accumulated and converted into phenylpyruvic acid and other derivatives. Accumulation of these in brain results in mental retardation. These derivatives excreted through kidney.

Chromosomal disorders:

Caused due to abnormal increase or decrease of one or more chromosome.

Failure of segregation of chromatids during cell division cycle results in the gain or loss of chromosome(s), called Aneuploidy.

Down syndrome:

- Caused due to presence of an additional copy of the chromosome number 21 (trisomy of 21).
- This disorder was first described by Langdon Down (1866).
- Short stature with small round head.
- Furrowed tongue
- Partially opened mouth
- Palm is broad with characteristic palm crease.
- Physical, psychomotor and mental development is retarded.

Klinefelter's syndrome:

- Caused due to the presence of an additional copy of X-chromosome resulting into a karyotype of 47, (44+XXY).
- Overall masculine development.
- Also develop feminine character (development of breast i. e. Gynaecomastia)
- Individuals are sterile.

Turner's syndrome:

- Caused due to the absence of one of the X-chromosomes i. e. 45 (44 + X0).
- Such females are sterile as ovaries are rudimentary.
- Lack of other secondary sexual characters.



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