

Concept and Definition of Genetics

The word “GENETICS” is derived from ‘genesis’ meaning creation, to become, or the way in which something is formed; and it was coined by Batson in 1906. Genetics may be defined as the science of heredity and variation which deals with resemblances and differences among individuals related by descent. Heredity means the similarity which progeny shows to its parents, and provides for the organic continuity between generations while variation deals with the differences and similarities exhibited by individuals in the progeny. In other words heredity constitutes a way of biological preservation whereby parental characteristics continue from generation to generation. The resemblance expressed by new born places a confidence in the heredity of that family. Wheat grains will produce wheat seedlings and puppies grow into dogs and never into cats. The general growth and development pattern of the off-spring in the Living organisms, resembles that of their parents so closely that the inheritance of characters becomes a matter of fact. Heredity is, therefore, the genetic continuity between the parents and the offsprings, and thus preserves a race by developing progeny in the parental image.

Variation

There are billions of peoples living on this earth; no two of them are exactly alike. Members of the same family and, in some instances, even the identical twins are different from one another for many characters such as complexion, body size, intelligence, voice etc. They show differences although they share common parentage. This is deviation from the heredity and is called variation. The variation present in the various characteristics of the members of the same race or a group of closely related individuals may arise due to change in the gene structure, new combination of genetic factors or change in the environment. Heredity and variation are common attributes of all biological organisms, and the science of genetics deals with the study of these two phenomena.

Heredity and environment

Heredity-the force of preservation of the ancestral characters generation after generation-determines fundamental biological patterns and the ultimate development of a character is the

product of its heredity and environment. Some of the traits are influenced more by the environment than others. A change in environment, therefore, may change the expression of a trait but such a change will only persist for the life-time of that individual and should not be expected to be transmitted to its off-spring.

Environment is of two types, i.e. internal and external. The internal environment includes the cell contents within which lies the hereditary material (chromosomes), while the external environment comprises extracellular conditions, e.g. soil, climate and similar other factors. As we have said before, environment exerts vital influence, on the development of a character. For instance, if the whole body hair of a Himalayan rabbit (white body except the tips of feet, nose and ears) are removed and new hair allowed growing under cool temperature, it will be black. On the other hand, if the hair from the black part (i.e. ear tips) is removed and that part is kept warm, the new hair that grows will be white. This shows that hereditary factors responsible for the production of black pigments in the Himalayan rabbit produce their effect only in cool temperatures.

To cite another instance, there is a maize variety Sun red, in which the outer husk of the cob develops red color in the presence of sunlight. The portion of husk emerging at night or in the dark is green and the one growing in the sunlight is red.

HISTORY OF GENETICS

Genetics has been developing as a vigorous science since the beginning of the twentieth century. We find in the literature on the history of biology that speculations on the nature of heredity are as old as the history of mankind. An old Babylonian tablet dating back to 6,000 BC shows the pedigree of a large number of successive generations of horses. Stone carvings of the same period illustrate cross pollination of the date palm. The early Chinese are known to have made considerable improvements in the varieties of rice. It is believed that most of the common domestic animals and plants were brought in the service of man before the beginning of the recorded history.

Sexual reproduction and hybridization

Camerarius (1764), a German Professor, was the first to describe sexual reproduction in plants. He is also credited with the production of the first hybrid plant artificially. Later, Kolreuter performed experiments on artificial hybridization on tobacco varieties and studied their hybrids. Schleiden and Schwann (1838) first recognized the cell as a unit of structure and function and propounded the theory that the new cells arise only from the pre-existing cells. Strasburger and Hertwig (1875-1885) found that only a single sperm is required to fertilize an egg. They suspected that the nucleus of the cell was the basis of heredity and that male and female gametes took equal part in the transmission of heredity.

Before Mendel, various scientists tried to solve the mystery of heredity; views of some of them are summarized below:

Preformation and epigenesis

In 1679, Swammerdam claimed that he was able to see in the human sperm under his crude microscope a miniature of man which he named 'homunculus'. He believed that man's body was performed in the sperm and that during the embryonic stage, only the growth of different organs to a full-size baby occurred. Bonnet who believed more in egg cell than sperm differed with this theory only slightly. He advanced his own theory called "egg encapsulation or box theory", which held that the female contains all the germ cells of her immediate and remote progeny, and that Mother Eve in her ovary had the germ cells of all the men to be born. With some improvement in the microscope, Wolf (1733-1794) and Von Baer (1792-1876) studied the structure of sex cells carefully and discovered nothing like homunculus or the little man but observed that cells were composed of structure less fluid. They replaced the idea of preformation with epigenesis. According to this idea, the process of development involved both male and female sex cells and that a vital force *was* responsible for the formation of organs in the embryonic stages.

Inheritance of acquired characters

The Greek philosophers thought that 'be inherited characters of the individuals were acquired through direct contact with the environment. This idea first materialized into a theory advanced by a French biologist, Lamarck (1744-1829) who emphasized an animal's desire to determine its need. The desire in turn, would determine the use or disuse of the body parts and this selective use and disuse would bring about modification of that organ. The modification so induced in the parents by the environment was believed to be imprinted upon the germinal material (egg and sperm) and was thus transmitted to the offspring. According to Lamarck, variations are induced in the organism upon germinal material. To support this idea he cited instances of the long neck of giraffes to secure food from tall trees, absence of eyes of fish living in dark caves under the sea, webbed feet of frogs, etc. The present knowledge of the germinal material and the developmental processes in plants and animals, however, do not support Lamarckism.

Darwin's hypothesis of Pangenesis

In the mid-nineteenth century Charles Darwin tried to figure out the pathway of heredity in a very interesting manner. He proposed that each body cell produced its rudimentary copy, which he called pangene or a gemmule. These gemmules were supposed to be delivered in the animal's blood stream ultimately to reach the germinal tissues (ovary and testicle), where the gemmules obtained from all body cells form gametes. A gamete so synthesized had all the cells of an organism in rudimentary form which would develop into a full-fledged new individual; thus heredity was supposed to be due to the gemmules representing blueprints of different body organs. This hypothesis was rejected by Galton (1822-1911) during the lifetime of Darwin. He transfused blood of a black rabbit into the body of a white rabbit and expected a progeny of black and white from it, in accordance with Darwin's theory. But he did not obtain the expected results.

Germplasm theory

Weismann, a German biologist (1834-1914), conducted his famous experiment on mice to show the difference in body tissue and germplasm. He cut the tails of mice for 22 generations

and observed that the progeny in the 23rd generation still had normal tails. He believed that the reproductive tissue (germplasm) is quite separate and distinct from the body tissue (somatoplasm). He emphasized the remarkable stability of germplasm which is transmitted unchanged from generation to generation. Somatoplasm is a product of germplasm and, therefore, any change induced by the environment on the body cannot be transmitted to the germinal tissue. Acquired characters are not inherited because germplasm is the only channel through which heredity is passed to the offspring. Of all the theories discussed heretofore, this theory comes closest to the Mendelian concept of heredity.

PROBLEMS

1. Define heredity and explain its basis in detail.
2. List some human characters which are easily influenced by the environment and those which are not.
3. Make a list of some of the characters of domestic animals which you think are controlled by heredity.
4. How will it help the geneticist to improve a race, assuming that the acquired characters are (a) inherited (b) not inherited?
5. What were the factual bases of the theories of inheritance of acquired characters, epigenesis, preformation, pangenesis and continuity of germplasm?
6. A boy refuses to marry a girl whose father, grandfather and great grandfather all died of tuberculosis. Comment on his thinking.
7. If a character is transmitted only through mother, which part of the female gamete (cytoplasm, nucleus) do you think will carry the factor responsible for this inheritance?

The Cell, Cell Division and Cell Reproduction

The Cell

The cell may be defined as a piece of nucleated cytoplasm surrounded by a cell wall (in plants) or a membrane (in animals). A group of cells called a tissue. The theory was first formulated by Schneider and Schwann in 1838. According to their theory, the cell is the structural unit of living organisms and that new cells arise from the pre-existing cells.

A cell is composed of living and non-living parts; the various parts of a typical cell are briefly described as follows

(a) Non-living: Protective membrane or cell wall and vacuole.

(b) Living: Protoplast structurally differentiated into nucleus or nucleoplasm and cytosome or the cytoplasm.

(a) Non-living Parts

(i) cell wall: Non-living ,a protective structure deposited by the cytoplasm surrounding the living material, permeable or semi-permeable,

(ii) Vacuole: Present mostly in plant cells and less so in animal cells, frequently serves as a store for non-living substances such as, minerals starch, food particles and other by-products of metabolic activity of the protoplasm; such substances are called inclusions.

(b) Living Parts

(i) Nucleus: It is the most conspicuous, and essential part of the cell, globular in structure, centrally located in a young cell, but in older plant cells it usually attains a peripheral position, it has its own organization as given below: \

(1) Chromatin: Deeply staining appear threadlike during interphase but during cell division appears organized as chromosome which are the carriers of hereditary information.

(2) Karyolymph: Except chromatin, the rest of the nucleus is filled with nuclear sap or karyolymph. It is a Stain-resistant material.

(3) Nucleolus (Pl. nucleoli): Darkly-staining, round body, one or more found in a nucleus, visible in interphase and early prophase of the cell division, supposed to act as organizer for nuclear material.

(4) Chromosome: The chromosomes known as the nuclear bodies have their own special organization; they are capable of self-reproduction during the cell division and maintain their morphophysiological properties during the life cycle of an organism. The chromosomes, their behaviour, equational distribution at mitosis and reduction in their number during meiosis were adequately understood before the discovery of Mendel's work. Chromosome is a Latin word meaning "the coloured body" which in the natural stage is colourless but can take colour, when stained with some specific dyes (Carmine, basic fuchsin etc.) particularly during the process of cell division. They lie within the nuclei of the living cell except for a short time during cell division when the nuclear wall disappears. The chromosome number remains constant from species to species; the body cell maintains the diploid number ($2n$) while the sex cells have haploid number (n) where the number is reduced to one half in the process of gamete formation. The chromosomes also change in shape and form during the various phases of cell division. Such changes are cyclic and at the end of each cell division the original form of the chromosome is restored. Each chromosome can be distinctly recognized due to its definite form and morphology. Salivary gland chromosomes of the fruit fly, by virtue of their large size, are called giant chromosome

(ii) Cytoplasm: Bounded by plasma membrane from outside and similarly by membranes of nucleus and vacuoles from inside, chemically complex, storehouse for various enzymes and cannot live long without nucleus. Plasmagones and cytogenes are believed to be equivalent to the nuclear genes; these are self-duplicating,

(iii) Mitochondria: Very small structures, visible by special techniques, mostly round or rod-like in shape: they lie free in the cytoplasm but usually aggregate near the nucleus and increase in number by simple division.

(iv) Golgi bodies: Present only in the animal cells and in the cells of certain lower plants, absent in higher plant cells, appear as a continuous network of strands; they are conspicuously present in secretory cells.

(v) Plastids: Three main types are recognized:

(1) Chloroplasts: develop green colouring pigments.

(2) Leucoplasts: colourless in plant parts not exposed to light.

(3) Chromoplasts: develop yellow and orange pigments. They increase in number by simple division.

(vi) Centrosome: Present in many animals and lower plant cells, lie near nuclear membrane, darkly-staining, granule present in the centre, gives out asters during cell division, divide into two during cell division and assume polar positions.

Cell Division

Two types of cell divisions occur in the life cycle of a living organism and they are,

(i) Mitosis (ii) Meiosis.

1. Mitosis:

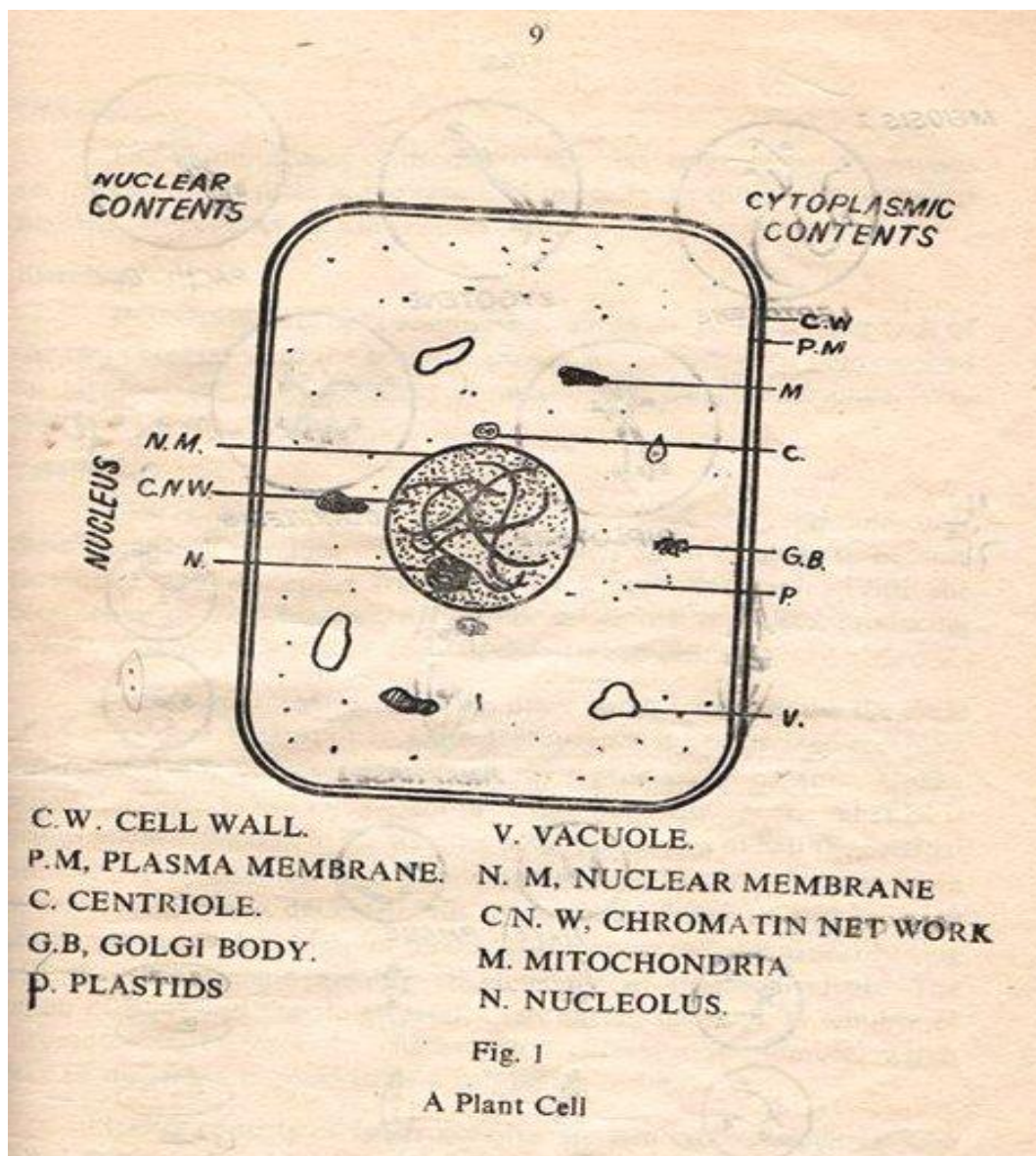
Throughout the development of an individual from the zygotic stage up to its maturity, the individual grows by *mitosis*, which may be defined simply as an equational cell division. When a cell divides by mitosis into two daughter cells, the chromosomal components equally distribute to the two resulting daughter cells. This equational distribution is achieved through four phases of mitosis, namely, (1) Prophase, (2) Metaphase, (3) Anaphase and (4) Telophase which are briefly described below:

Prophase

The chromosomes shorten in length and get thicker, each chromosome appears double stranded as if split longitudinally into two chromatids held by the centromere; the nucleolus evident in the beginning disappears during late prophase.

Metaphase

The chromosomes shortened to the maximum, orient themselves on the equatorial plate in the centre of the cell; the spindle fibers appear and the centromeres get attached to them



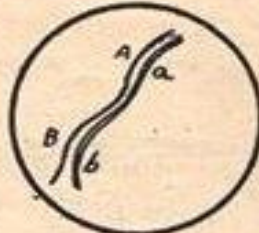
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FIG. 2

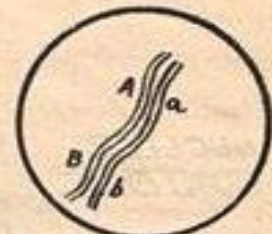
MEIOSIS I



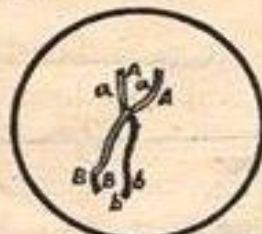
LEPTOTENE



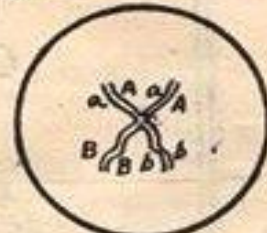
ZYGOTENE



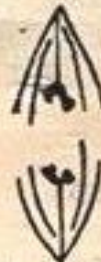
PACHYTENE



DILOTENE



DIAKINESIS



ANAPHASE I



TELOPHASE I

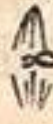
MEIOSIS II



INTER PHASE



PROPHASE II



METAPHASE II



ANAPHASE II



TELOPHASE II

Meiotic Divisions

Anaphase

The centromeres split and move towards the poles, pulling each of the two chromatids to the opposite pole. As a result, each pole receives an identical group of daughter chromosomes the chromosomes have now divided equationally.

Telophase

Each chromatid attains the status of an individual chromosome, the nuclear wall develops. The chromosomes again elongate to become threadlike and disappear from view to enter interphase. With the completion of the telophase, the mitotic division is complete, producing a new cell which is a precise copy of the original cell.

2. Meiosis:

Meiosis, on the other hand, occurs in the life cycle of an individual, a plant or an animal, only when it has attained complete differentiation and development of its reproductive organs. Unlike mitosis, meiosis is not equational in nature but reductional. That is, it of chromosomes to half the original number. For instance, if the dividing cell has 20 chromosomes as in maize, the resulting daughter cells at the end of meiosis will have 10 chromosomes each. Meiosis results in the germ cells (gametes) having half the chromosome number characteristic of that individual. The union of male and female gametes, each having the haploid number of chromosomes, restore the characteristic chromosome number, which may be diploid, tetraploid or of a still higher order.

Meiosis consists of two successive division cycles, which may be called Division I and Division II. Division I has a relatively prolonged prophase followed by the same phases as described for mitosis. At the end of Division I, the number of chromosomes in the two resulting daughter nuclei is halved. These two daughter nuclei undergo Division II, Whichin its mechanism and operation, is like mitosis, and further divides these nuclei into two cells each, and this time the number of chromosomes is not halved but remains the same as at Division I. Meiosis in its various phases may be briefly described below:

A. Meiosis I (Division 1)

1. Prophase:

Sub-divided into five or more phases.

(a) Leptotene. Cell size (meiocyte) relatively larger than Surrounding somatic cells; chromosomes long thread like and slender, beadlike structures (chromosomes) on the chromosomes are identified

(b) Zygotene. Homologous chromosome synapse lengthwise each chromosomes into two chromatids and the tetrad condition develops.

(c) Pachytene. Chromosomes shorten and thicken but are still threadlike; homologous closely attached to one another, already synthesized chromatids start separating longitudinally, relational coiling occurs, nucleolus clearly seen.

d) Diplotene. The four chromatids tetrad open out, start repelling each other but remain connected at certain points called chiasmata singular (chiasma), chromatids exchange corresponding portions by cross-over, chromosomes further shorten due to tightening of coils.

(e) Diakinesis. The chromosomes further shorten to about one tenth of their length in the leptotene stage, deeply stained pairs may assume typical configuration, nucleolus generally disappears.

2. Metaphase:

Nuclear membrane disappears and spindle fibres appear, chromosomes orient themselves on to the equatorial plate and centromeres lie along the a of the spindle.

3. Anaphase:

Chiasmata between the paired chromosomes are released and each dyad (the two sister chromatids all original or comprising a crossover) migrates towards any of the two poles;

chromatids are held on together by undivided centromere. Reduction of chromosomal complement is completed.

4. Telophase:

Chromosomes arrive at the poles, two new daughter nuclei with haploid number of chromosomes are formed; is the reverse of prophase; reduction is completed quantitatively, but not necessarily qualitatively.

The two newly formed nuclei enter interphase, rest for a while in some cases before second meiotic division starts. The four stages of the second part of meiosis (Meiosis II) are completed in quick succession.

B. Meiosis II (Equational Division)

1. Prophase:

The chromosomes regain threadlike structure, which thicken and shorten as the stage advances; chromatids of the dyads are attached to the centromere, each chromatid may be qualitatively different from its mate depending upon whether or not crossing has occurred.

2. Metaphase:

Chromosomes move on to equatorial plate, spindle fibers appear and are attached to the centromeres.

3. Anaphase:

Centromere divides, chromatids separate, each becoming an independent chromosome, segregating to the two poles.

4. Telophase:

Chromosomes reach the poles; lose their identity, nuclear membrane reappears (reverse of prophase). The end product in plants is a four-celled structure, (quarter) each individual cell results into gametes as explained below:

Reproduction:

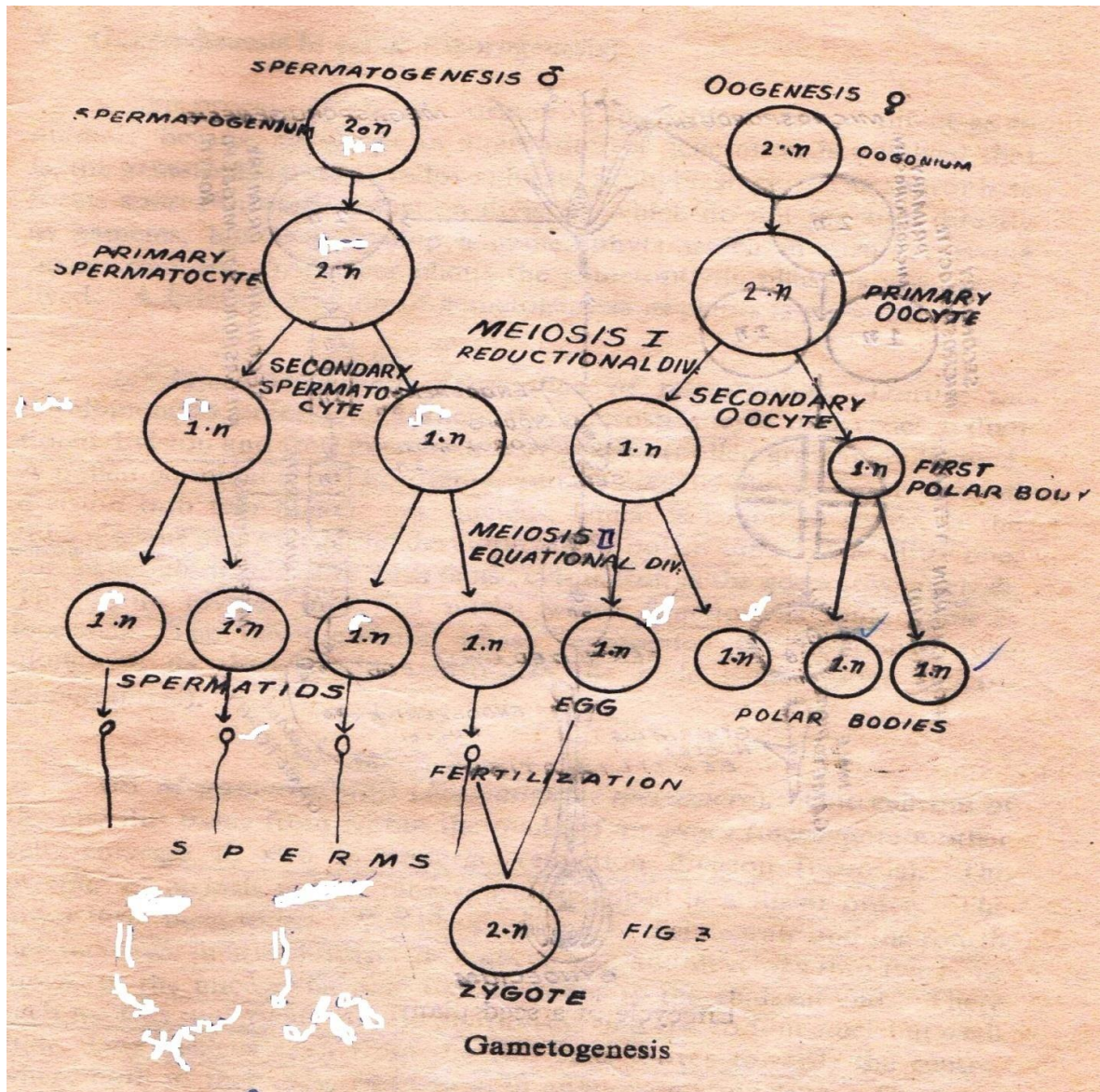
The sex cells thus formed will become reproductive cells after certain modifications. In animals they are directly transformed into functional gametes, whereas in plants they go through a gametophytic phase which then forms the true gametes. The cells in which meiosis is initiated are called primary meiocytes, and the cells resulting from the Division I of meiosis are the secondary meiocytes. If a primary meiocyte produces spores (in plants) it is called sporocyte, i.e., primary microsporocyte and primary megasporocyte and the phenomenon is known as sporogenesis. If a primary meiocyte produces sperms or eggs (animals) it is called primary spermatocyte and primary oocyte, respectively, and the phenomenon is termed gametogenesis. These processes of gamete formation are now described below:

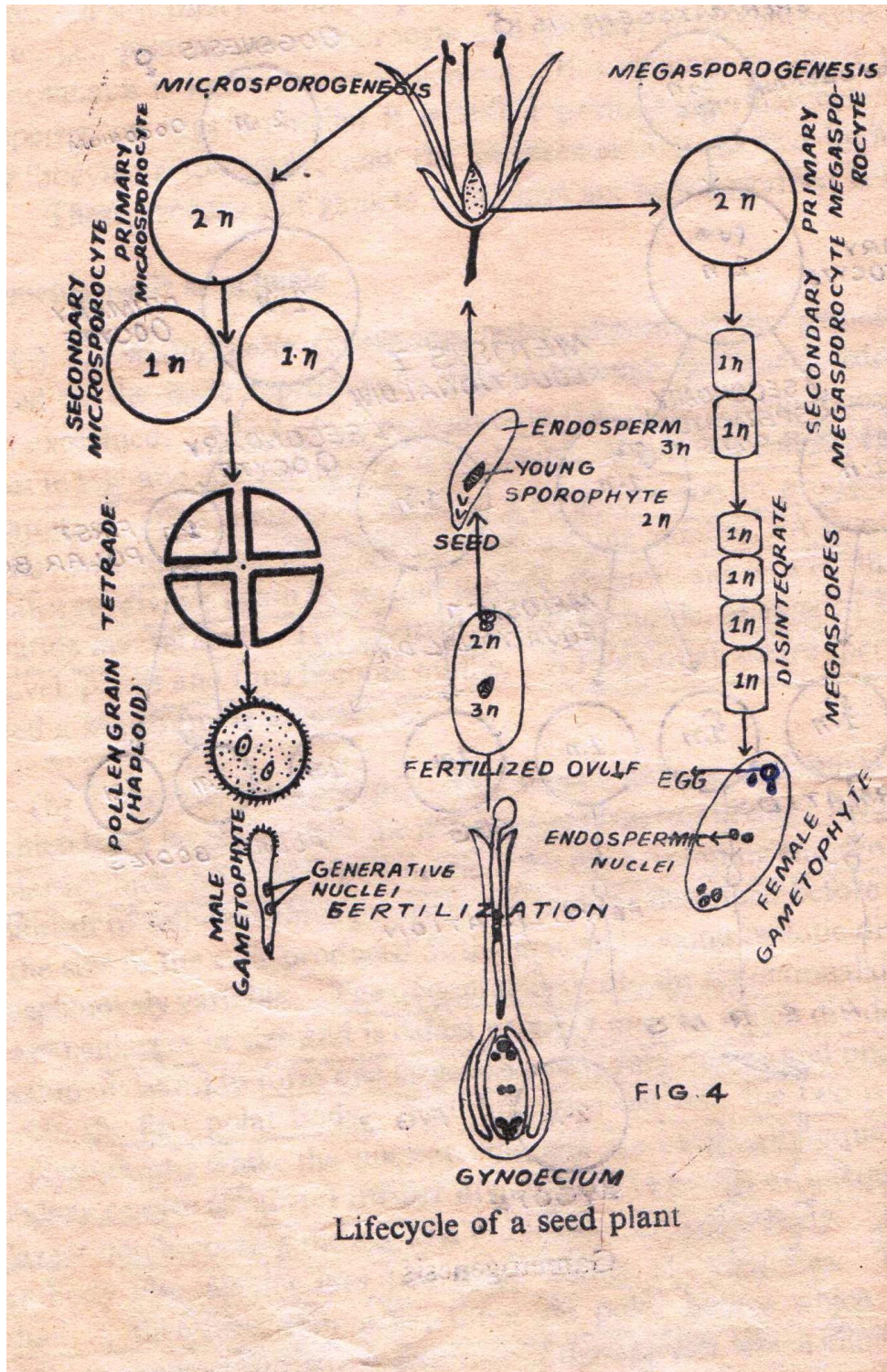
1. Gametogenesis in animals

(a) Spermatogenesis (Spermiogenesis). Sperm (male gamete) is produced in the male reproductive Organs: through meiotic division as already explained. A cell of the reproductive tissue, the spermatogonium, enlarges in size and therein meiosis is initiated. The cell is called spermatocyte. This undergoes the first meiotic division to produce two secondary spermatocytes with half the chromosome number and as a result, four spermatids are formed. The spermatids are immotile, and develop tails and thus become motile. A functional, active spermatid is called a sperm (male gamete)

(b) Oogenesis (Ovogenesis). The female gamete in animals is the egg which has a large size than the male gamete (sperm) because its mechanism of cell division is precisely the same as described before except that the size of the cells produced at the first and second meiotic divisions is conspicuously variable. The oogonium prior to the reductional division (meiosis) enlarges in size and is called primary oocyte, which divides (first reduction division) to form one large secondary oocyte and one small size cell the first polar body. The difference between the two is in size only (cytoplasm) while the nuclear contents are essentially equal. The secondary oocyte (ovocyte) further divides into two cells of unequal size. The largest one becoming the egg and the smaller the polar body also divides into two polar bodies of equal size. The end product is therefore four cells three are polar body which are not sexually functional and

the fourth one fourth one is the egg cell which functions as the female gamete without any further changes.





2. Gametogenesis in plants (Sporogenesis)

The process of gamete formation in plants is not as direct as animals, because in plants an alternation of generation is involved that is, the gametophytic stage follows the sporophytic stage. The sporophyte forms spores through reduction division, which do not function directly as gametes, but first develop a gametophyte which in turn forms the sexual gametes. In higher plants the gametophytic phase is very short-lived. A description of gametogenesis is given below:

(a) Microsporogenesis (Formation of microspore). During the developmental process of anthers the microsporocyte undergoes reductional division and four microspores or pollens (pollen grains) are formed. Pollen germinates on the stigma, and its nucleus divides by equational division into two nuclei; one nucleus, which is round in shape is the tube nucleus and guides the development of pollen tube. The other one somewhat elongated, and with dense cytoplasm, is the generative nucleus. This again divides into two nuclei which function as the male gametes (sperms). The development phase represented by the pollen tube along with its three nuclei constitutes the male gametophyte (micro gametophyte).

(b) Megasporogenesis (Formation of megaspore). One cell out of the nucleus tissue from within the ovule of an ovary (megaspore mother cell) enlarges in size to undergo reduction division (meiosis). The product of meiosis is four megaspores arranged in a linear order. The lower three megaspores which are small in size disintegrate and the fourth one increases in size to form embryo sac, and develops into two; one moves to the micropylar end and other to the chalazal end. There each nucleus undergoes mitotic divisions giving rise to four nuclei at each end. One nucleus each from the two ends moves towards the centre, where both of them fuse to form a $2n$ endosperm nucleus. The three nuclei on the chalazal end, called the antipodals develop cell wall around them and in most plant species degenerate after fertilization. At the micropylar pole, the large-size central nucleus is the “egg” and the two lateral ones are the synergids. The egg functions as female gamete.

Fertilization

The pollen-tube pierces through the style of the gynaecium and enters the ovule through the micropylar end. The pollen-tube ruptures and the two male gametes are released; one fertilizes the egg cell to form $2n$ embryo or the zygote and the second male gamete fuses with the endospermic nucleus. The $3n$ tissue forms the endosperm which serves as reserve food material. Since for the developing embryo the two male gametes released by the pollen tube are simultaneously used in fertilization within the ovule, this process is called double fertilization.

PROBLEMS

1. Which part of the cell do you think is more important from genetic viewpoint and why?
2. Do you think that different plant organs have different chromosome number?
3. The somatic ($2n$) chromosome number of maize is 20. What is its gametic number? How many chromosomes are there in the pollen grain, the egg, the endosperm, the root tip, aleuronic layer and testa?
4. What will happen to the chromosome number and the resulting gametes, if the second division fails to occur during the meiotic process in a common wheat plant?
5. How does the process of gamete formation in animals differ from that in plants?
6. In what way does the division of cytoplasm differ from that of the nucleoplasm in a plant cell undergoing meiosis?
7. The ($2n$) chromosome number in man is 46. What is the chromosome number in (a) primary spermatocyte (b) secondary spermatocyte (c) spermatid (d) sperm (e) polar body (f) red blood cells?
8. In a normal process of spermatogenesis, how many sperms, secondary spermatocytes and spermatids will be formed from 100 primary spermatocytes?
9. A mature pea pod contains six viable seeds. What could be the number of pollens used during fertilization to produce these 6 viable seeds?

10. How does reduction division (meiosis) differ from somatic cell division (mitosis) in plants?

11. Why is the prophase longer in meiosis than mitosis?

12. Plants and other higher living organisms grow, develop reproduce by cell divisions. Of the two types of cell divisions, i.e., meiosis and mitosis, could it be that one is more important than other?

Mendelian Inheritance

We have seen in our discussion of gametogenesis (sporogenesis) that the gametes or the spores produced by the maturation process essentially have the haploid chromosomes number. While the male gamete (sperm) is mainly composed of chromosomes, the female gamete, (egg) possesses cytoplasmic material in addition to the chromosomes. The union of these two gametes initiates a new individual, whose subsequent growth is guided by the genetic information contained in the chromosomes in the form of genes. A diploid $2n$ organism has a double dose of a gene whereas a haploid (n) has a single dose. It is the genes arranged in a linear fashion on the chromosomes which are passed along with the chromosomes from generation to generation and determine the limits of growth of an individual. The chromosomes thus being the carriers of genes are

Brief life sketch of Mendel

Gregor Mendel (1822-1884), who is called the father of genetics, was born in a poor farmer family of Austria. After completing education up to secondary school, he joined a monastery during 1843, and on his 25th birthday he became a monk. Mendel conducted hybridization work on various plants, i.e., snapdragon, pumpkin, flax, bean, pea, plum, pear, maize, etc. He was sent to Vienna University during 1851 to study science and mathematics. On his return to Brunn (1854), in addition to his duties as priest, he was appointed a substitute teacher in a school. He collected garden pea seed from the market during 1857 and started regular hybridization work and was thus able to present his results before the History Society of Brunn in 1865. The results were published in 1866 and were sent to various countries. His work remained unnoticed for a period of 34 years. In 1900 three biologists, Correns, deVries, and Tschermak read Mendel's forgotten paper and also obtained the similar results from their own experiments.

Mendel's choice of pea plant was not a mere chance, but was based on these facts: (i) many pure-breeding pea varieties were available

(ii) floral structure and mode of pollination in pea ensures self-pollination and (iii) hybrids are fertile. He finally selected the following seven easily recognizable contrasting character:

No.	Character	Dominant
1.	Tall versus dwarf vine	Tall
2.	Green versus yellow color of unripe pods	Green
3.	Inflated versus Constricted pods	Inflated
4.	Axillary versus terminal flowers	Axillary (Axial)
5.	Yellow versus green endosperm (Cotyledon)	Yellow
6.	Round versus wrinkled seed surface	Round
7.	White versus grey seed coat	Grey

Mendel made several crosses of pea plants having contrasting characters to find out how the hereditary elements behaved while passing from parents to offspring. We shall consider here only one or two of the several experiments that he performed.

For instance, Mendel crossed a tall pea plant with a dwarf one and observed that all the resulting hybrid plants (F_1) were tall. The F_1 plants were self-fertilized to produce F_2 seed which was planted to study the behavior of the F_2 generation. Mendel noted that of all the F_2 plants, $3/4$ plants were tall and $1/4$ plants were dwarf.

It may be pointed out here that the outward expression of a character later came to be referred to as “phenotype” and the complex of genetic factors controlling this expression as “genotype”. We may now write the above cross genotypically using symbols as follows.

Parents x (Tall) TT, tt (Dwarf)

Gametes T, T : t, t,

Combination of (female)

and male gametes produces F_1 Tt (Tall)

F₁ generation.

On selfing, two types of gametes (pollens and ovules) are possible from Tt plant and they are T and t. The random union of the male T and t with the female T and t will produce the F₂ generation as follows:

Male gametes	T,	t	
Female gametes	T,	t	

	TT,	Tt	
		Tt	tt

Genotypes in F ₂	ITT	2Tt	1tt
Phenotypes being	3 Tall:		1 Dwarf

The same combinations be obtained by the checker-board can method as:

Male gametes			
	$\frac{1}{2}$ T	$\frac{1}{2}$ t	
Female gametes	$\frac{1}{2}$ T	$\frac{1}{2}$ TT	$\frac{1}{2}$ Tt
	$\frac{1}{2}$ t	$\frac{1}{2}$ Tt	$\frac{1}{4}$ tt

F₂ phenotypic ratio: 3 tall : 1 dwarf or $\frac{3}{4}$ tall : $\frac{1}{4}$ dwarf

F₂ genotypic ratio: 1 tall (TT): 2 tall (Tt) : 1 dwarf (tt)

or $\frac{1}{4}$ (TT) : $\frac{1}{2}$ (Tt) : $\frac{1}{4}$ (tt)

When Mendel selfed these F₂ plants, he observed that the dwarf plants, which were $\frac{1}{4}$ of the total F₂ population, produced all dwarfs, while out of the $\frac{3}{4}$ tall plants, produced all tall and $\frac{2}{3}$ produced both tall and dwarf plants: that is precisely what was expected from the genotypes contained in the F₂ population above.

Law of Segregation

All other 6 characters behaved in the similar manner in crosses. From this, Mendel formulated his first law of inheritance: the law of Segregation or Mendel's first law of inheritance. It states that heredity characters are determined by some particular factors (genes) these factors occur in pairs and at the time of gamete formation segregate at random so that only member of a gene pair is transmitted to a particular gametes

Dominance

The other important phenomenon which Mendel discovered was that of dominance. He noticed that the hybrids (F₁'s) between tall and dwarf were all tall like the tall parent, although it also inherited the dwarf factor. The factor which dominated the effect of the other allele in the F₁ was designated by Mendel as dominant and the alternative factor that remained latent in F₁ as recessive. He showed the dominant characters but the use of capital letters and the recessive by small letters while explaining his results.

Note: In crosses, it is not essential for one character to behave always as dominant, since situations midway between the two parents are also obtained in F₁.

Mendel's law of segregation and its explanation

Mendel's first law of inheritance is the law of segregation, which explains the behaviour of alleles of a gene pair in a diploid organism at gamete formation. According to this law, the two alleles which may be identical or dissimilar i.e. AA, aa or Aa segregate by going to different poles in meiotic divisions and thus end up into different gametes. Note that segregation of alleles occurs during the meiotic divisions.

Consider a cross between two true-breeding plants. One of them is tall and the other dwarf. Because both of them are diploid, each has a pair of alleles. The tall one has both the

alleles, T, T and the dwarf one has both its alleles, t, t. The gametes which they will produce will have one allele only as the number is reduced to one half. We shall get a gamete of T constitution from one parent and a gamete of t constitution from the other. They reunite and their reunion is called fertilization. The new plant produced by the union of T and t will be Tt. This means that the new hybrid plant has two alleles but they are not alike, as one of them is T (for tallness) and the other is t (for dwarfness). If the hybrid plant is tall, we will be able to say that the character tallness controlled by T is dominant over the corresponding contrasting character, dwarfness, controlled by t. Further, when this hybrid plant Tt matures and forms gametes, the two alleles T and t, carried on the two homologous chromosomes, cannot stay together, because the homologous chromosomes must go to the opposite poles in meiotic division and so will the two contrasting alleles. This going of the two alleles to the opposite poles in meiosis provides the mechanism for segregation of the alleles of the same gene pairs. Consequently, two types of gametes, T and t are formed by the hybrid plant in its male and female reproductive organs. Their reunion will produce these off springs 1 TT: 2 Tt: 1 tt, as explained earlier.

From this discussion, it is apparent that the two alleles remain together in a diploid individual and segregate when that individual forms gametes and again come together in various combinations when male and female gametes unite to produce new individuals.

Mendel's second law of inheritance or Mendel's law of independent assortment

We may now consider two characters together as to their behaviour in inheritance. For instance, Mendel crossed a tall plant having axillary flowers with a dwarf plant having terminal flower. Character dominance is indicated on page 18. As before, T and t represent tallness and dwarfness, respectively, and let A and a represent axillary and terminal flower positions, respectively.

We may write the cross genotypically as follows:

Parents TTAA (tall axillary), ttaa (dwarf terminal)

Gametes TA ta

F1 TtAa (tall with axillary flowers)

The F1 plants will produce these four types of gametes TA, Ta, tA, ta, in equal proportion. The four types of gametes will be male as well as female and their random union will produce 16 combinations in the next generation (F₂).

Female gametes 1/4 TA + 1/4 Ta + 1/4 tA + 1/4 ta.

Male gametes 1/4 TA + 1/4 Ta + 1/4 tA + 1/4 ta

1/16 TTAA + 1/16 TTAa + 1/16 TtAA + 1/16 TtAa

1/16 TTAa + 1/16 TTaa + 1/16 TtAa + 1/16 Ttaa

1/16 TtAA + 1/16 TtAa + 1/16 ttAA + 1/16 ttAa

1/16 TtAa + 1/16 Ttaa + 1/16 ttAa + 1/16 ttaa

These sixteen combinations for the dihybrid cross can also be calculated by the checker-board method:

F1		TtAa			
Gametes		TA, Ta, tA, ta			
		Male gametes			
		TA	Ta	tA	ta
Female Gametes	TA	TTAA	TTAa	TtAA	TtAa
	Ta	TTAa	TTaa	TtAa	Ttaa
	tA	TtAA	TtAa	ttAA	ttAa
	ta	TtAa	Ttaa	ttAa	ttaa

Mendel noted that all F₁'s were tall with axillary flowers where from he confirmed that tall was dominant over dwarf and axillary flower over terminal flower and that out of these 16 combinations of the F₂, 9 were all with axillary flowers, 3 were tall with terminal flowers, 3 were

dwarf with axillary flowers and only one was dwarf with terminal flowers. it may be seen that, as the 9 tall, axillary-flowered plants have at least one T and one A, these show both the dominant characters, 3 have T but no A they show one dominant character, the other 3 have A but no T, and they show the other dominant character, one has neither T nor A but is *ttaa* and is double recessive.

Explanation. It will be noticed that when the hybrid plant, *TtAa*, matures, it forms four types of gametes (pollens as well as ovules). T and t are the two members of an allelic pair and gametes are formed as a result of meiotic divisions whereby the two alleles must segregate. In the case of the hybrid plant *TtAa*, T and t must segregate, i.e they must not be present in the same gamete and. similarly, A and a must segregate and go to different gametes. But while the two members of a given allelic pair must segregate they do so independently of the members that is, if T goes to one pole, either A or a of the allelic pair *Aa*, can go with T to the same pole. So the resulting gamete (pollen or ovule) can have T with A (TA) or T with a (Ta). Similarly, the gamete produced at the other pole can have t with A (tA) or t with a (ta).

Accordingly, this plant, *TtAa*, must produce four types of gametes, i.e., (TA), (Ta), (tA) and (ta) in equal proportion. This is actually what the plant does.

The production of these four types of gametes in equal numbers is possible only if the two gene pairs, *Tt* and *Aa* assort independently of each other. This phenomenon is called “independent assortment” of characters, the Second Mendelian Law. If the four types of gametes, male as well as female, unite at random in the process of fertilization, 16 F_2 combinations as shown above should be possible.

Back-cross and Test cross

When F_1 hybrid is crossed with one of its parents, it is called a backcross; when the cross involves the recessive parents it is called as test cross. We may remember that the F_1 hybrid between Mendel's tall and dwarf pea plants were all tall, which may be crossed with dwarf recessive (*tt*) parent to form a test cross. The hybrid tall parent (*Tt*) would produce equal number of T and t gametes and the dwarf being homozygous should form only one type of gametes, i.e., t. Half the ovules of the genotype, T when fertilized by t pollen will produce *Tt*

plants and the other half ovules of t genotype fertilized by t pollen produce tt plants. A definite ratio of $1/2$ tall and $1/2$ dwarf plants would thus be obtained. The $1:1$ ratio based on theoretical calculations was confirmed by Mendel through such experiments. Mendel clearly demonstrated that the characters are determined by particulate factors which do not blend with or contaminate one another during the course of their transmission from generation to generation.

Similarly, results from a dihybrid situation were also tested by Mendel. He crossed yellow round with green wrinkled plants to be symbolized genotypically as $YYRR$ & $yyrr$, respectively. The F_1 hybrid would be $YyRr$ (yellow-round) which when backcrossed to double recessive $yyrr$ parent will result in four phenotypes in equal number, i.e., Yellow- round $YyRr$; yellow wrinkled $Yyrr$; green round $yyRr$; and green wrinkled $yyrr$ (Dihybrid test cross ratio $1:1:1:1$).

This proved that:

1. The factors responsible for character expression are particulate in nature.
2. Segregation of factors takes place which shows that they do not blend with one another.
3. Grouping or the assortment of the factors is random during the process of gamete formation.
4. $1/4: 1/4 : 1/4 : 1/4$ test cross ratio proves that all types of gametes were produced in equal number and that they had an equal chance for union amongst themselves.

Blending theory

Before Mendel's work on hybridization was duly recognized, the "Blending theory" enjoyed popular support as the basis of inheritance of characters in living organisms. According to this theory, heredity determinants were fluid like which would always produce a blended expression of the characters of the two mating individuals. For instance, offspring must all be grey, which cannot be separated again in black and white in the progeny.

Particulate theory

Mendel contradicted the above idea by showing from his experimental evidence that the determiners of heredity are individual particles, which, in various combinations, can produce numerous shades of expression of a character and that these determinants are separable unaltered at the time the individual forms gamete. This theory is called the particulate theory of inheritance and is now accepted as valid.

Resume of Mendel's Experiments on Pea

Mendel made experiments on the pea using seven different contrasting characters. He selected pea plant as his experimental material because of the following advantages that the pea plant offered him:

1. Crossing was easy.
2. The characters were easily distinguishable.
3. The hybrids were fertile and easily mutually crossable
4. Due to special floral structure, contamination from foreign source was avoided.
5. Their culture was easy.

From his extensive data Mendel developed the following formula, which illustrate how fast the hybrid nature of a population disappears.

Segregating generations	Genotype			Ratios		
	AA	Aa	aa	AA	Aa	aa
1.	1	2	1	1:	2:	1
2.	6	4	6	3:	2:	3
3	28	8	28	7:	2:	7
4.	120	16	120	15	2:	15
n.	---	---	---	$2^n - 1:$	2:	$2^n - 1$

(The Formula $2^n - 1:2:2^n - 1$)

Conclusions:

1. Characters are controlled by hereditary determiners (Genes).
2. These determiners (genes) can be dominant or recessive.
3. The genes segregate at the time of gamete formation (they do not get blended).
4. All possible combinations of genes occur in the gametes (Independent assortment).
5. The male gametes combine at random to produce off springs showing all possible combinations of character.

Reasons for Mendel's Success

1. His choice of the pea plant was excellent: the plant was self- pollinated and, therefore, highly homozygous, was easy to culture and to cross; contamination of the crosses from the foreign pollen was avoidable due to its special floral structure.
2. Mendel started with simple experiments involving only one character difference and then proceeded with more complicated ones involving more than one character, making careful analysis of his data and testing his theoretical expectations.
3. He made use of mathematics to explain the biological behaviour of the factors determining character expression.
4. He was lucky that the characters he studied were not linked, otherwise he might have had difficulty in explaining the character variation not conforming with his hypotheses, as he had no knowledge of genes being located on the chromosomes.

Reasons for slow acceptance of Mendel's work

1. Some well-known scientists would have no confidence in Mendel's interpretation of the basis of inheritance, since they did not think Mendel was qualified enough to do that.
2. The results of some of the other scientists (Nageli's work on *hieracium*) were not in conformity with those of Mendel.

3. Mendel used mathematics to explain his results, whereas other scientists did not use this type of approach in the study of biological material.
4. Mendel was a kind of genius and a little ahead of his time. His contemporaries were ill equipped to understand him.

Intermediate dominance

In our previous discussion on Mendelian inheritance, we observed that in the F_1 , one form of the character was dominant over the other. During later investigations, this generalization did not hold good showing expression intermediate between the two parents were observed. Where dominance exists, a single dose of the dominant gene produce same effect as a double dose, but in case the dominance is incomplete the two alleles in the hybrid conditions (F_1) interact expression more or less intermediate between the two origin the character. In a cross between red-flowered (AA) and white (aa) snapdragons, the flower colour of F_1 (Aa) is pink, i.e., i between red and white. This seems a sort of blending between the original colours. When the F_2 was grown, segregation occurred in the usual proportion of 1/4 red AA; 1/2 pink Aa; 1/4 white a actually a modification of the monohybrid phenotypic ratio (shows that gene (A) alone is not sufficient to produce red fib but the intensity of the red colour is reduced to pink when allelic constitution is (Aa). The segregation of the original colours showed that the heredity material did not blend in F_1 . A backcross between the red and pink Aa produced 1/2 red and 1/2 pink and the back cross white (aa) and pink (Aa) will produce 1/2 pink and 1/2 white. that the characters are controlled by the particulate substances which segregate from one another uncontaminated.

Take another example: Some varieties of snapdragons leaves; others have narrow leaves. In a cross between broad (BB) and narrow (bb) leaved varieties the F_1 (Bb) has intermediate leave segregation of 1 broad (BB): 2 intermediate (Bb) and 1 narrow plants i obtained. When flower colour and leaf shape together i.e., the variety with red flower and broad leaves crossed with the white flower and the narrow leaf variety aabb; the F_1 had pink flowers and intermediate leaves. The F_2 segregated into 9 distinct phenotypes as follows:

1 AABB red broad :2 AaBB pink broad :2 AABa red intermediate: 4 AaBb pink intermediate : 1 AAbb red narrow : 2 Aabb pink narrow aaBB white broad : 2 aaBb white

intermediate : 1 aabb white i Each phenotype corresponds to a genotype. Here 9:3:3:1 are modified to 1:2:1:2:4:2:1:2:1.

Physical basis of Mendelian inheritance

Mendel had no knowledge of the existence of chromosome rediscovery of Mendel's work, attributing inheritance of characters particulate factors (now called genes) inspired scientists to explain nature of these factors. They argued that in order to be heredity determiners these particulate factors must possess the following attributes:

1. They should be continuous from one cell division to another.
2. They should be present in pairs in diploid organism separate out into different gametes at the time of gamete formation (segregation).
3. Each should have its own individuality and differ qualitatively from the other.
4. They should be capable of random reunion at the fertilization process.

In 1902 Sutton and Boveri put forth the chromosomes theory of heredity which correctly answered the above questions. According to Sutton-Boveri hypothesis, genes (determiners) are present on the same in a linear order. The chromosomes exist in pairs in the organism ($2n$) and in the haploid organism the chromosome are reduced to one half the original number (n) (Mendel's law of segregation). Members of homologous pairs of chromosomes separate or segregate each other during meiosis their diploid number is restored at fertilization. Two chromosomes carrying different genes assort independently original mates (homologues) depending upon the way they reorient themselves on the metaphase or equatorial plate (Mendel's independent assortment). It is just a chance that the paternal and the maternal sets may go in original combination or may form new combinations. It will be noted that the behaviour of the chromosomes conforms that of the Mendelian factors (genes) and are, therefore, the physical basis of heredity. There is a close parallelism between the two things and the chromosomes. To emphasize this fact once again repeated that:

1. Genes exist in pairs, because chromosomes, which carry them exist in pairs.
2. Genes segregate at meiosis, because of the fact that chromosomes segregate.
3. Genes assort at random, because of the fact that chromosomes assort in this fashion.
4. Genes recombine during fertilization process combinations because chromosomes do

so

Some useful formulae

1. Number of kinds of secondary meiocytes $=2^n$
 2. Number of kinds of gametes $=2^n$
 3. Number of kinds of genotypes $=3^n$
 4. Number of kinds of phenotypes
 - (i) When dominance is incomplete $=3^n$
 - (ii) When dominance is complete $=2^n$
 - 5 Number of kinds of homozygotes $=2^n$
 6. Number of kinds of heterozygotes $=3^n - 2^n$
 7. Number of kinds of gametic combinations 4^n
- where n-Number of heterozygous gene pairs.

PROBLEMS

1. What are the two most important laws of genetics that you have studied so far? Who formulated them and succeed in enunciating these laws while others more qualified than him did not?
2. Biologists of Mendel's time did not appreciate his work because he was a mere priest. Do you think they were right in snubbing him like that?

3. Suppose that Mendel's work was not to be discovered. Do you think somebody, else in that case might have p these laws round the turn of the century?
4. The F₁ plants between two different homozygous varieties always uniform. Will you regard these F₁'s as homozygous?
5. In linseed, purple flower colour (P) is dominant flower (p). Write a cross between two such homozygous parents. What will be the F₁ and F₂ phenotypes? Backcross the above F₁ to either parent. What phenotypes will you get and in what proportion?
6. Mendel performed two separate experiments to laws. Was it not possible for him to explain both from one experiment only?
7. In tomato, the fruit skin may be hairy or smooth between the two smooth and hairy varieties pr smooth fruits. Intercrossing of the F₁ plants produced 174 hairy and 520 smooth textured fruits. How is skin texture inherited?
8. What types will be produced if F₁ plants in the problem were crossed with smooth parent, hairy parent?
9. In summer squashes, white fruit is due to dominant and colored fruit to its recessive allele (w). Disk-shaped is due to a dominant gene (D) and roundish fruit to allele (d). Make a cross between homozygous varieties having white, disk fruit and the other coloured. Raise F₂ generation. How many different genotypes and phenotypes do the squash plants have with respective and fruit shape? What will be the proportion of double recessive and double dominant, plants in F₂?
10. In pea, green cotyledon colour (YY) is dominant colour (yy) and red flower colour (RR) is dominant (rr). Make out the genotypes and phenotype following cross combinations:

- | | | |
|----------|---|------|
| (a) RRYy | x | rryy |
| (b) RRyy | x | rryy |
| (c) RRYy | x | rrYY |
| (d) RRyy | x | RRYY |

- (e) RrYy x rryy
- (f) RrYY x rryy
- (g) RrYy x RRYY
- (h) Rryy x rryy

11. In cattle, the F₁ offspring between red and white are roan (intermediate) and absence of horns (polled) is dominant horned character. A horned red bull is mated polled cow (homozygous for horn character). The offspring are mated to raise the F₂ generation. Figure out and phenotypes of the expected second generation individuals.

Inheritance of Simple Mendelian characters in Man and Lethal Genes

The laws of segregation and independent assortment which Mendel formulated on the basis of his work on pea are valid not only for peas and other plants but also for animals and human beings. It is not possible to use humans as a breeding material, so it takes longer observations on the expression of human characters to verify that human heredity follows the same principles as other living beings. We shall discuss heredity of a few human characters here:

1. Inheritance of albinism

An albino individual is one which completely lacks pigment all over his body. This condition behaves like a simple Mendelian character and may be recessive while non-albino is dominant and can be homozygous and heterozygous, i.e., AA or Aa, if a denotes the gene for albinism.

2. Inheritance of taste blindness

Phenyl-thio-carbamid (PTC) has an intensely bitter taste, but to some individuals this substance has no taste. It was found that 70 percent of the American white people are “Tasters” and the remaining 30 percent “Non-tasters”. The ability to taste is a simple Mendelian trait and is governed by a single dominant gene, say T gene. The genotypes of the “Tasters” can be homozygous (TT) or heterozygous (Tt) and of the “Non-tasters” (tt).

3. Inheritance of eye colour

The eye colour (iris of eye) has three division: (i) blue (ii) brown (iii) black. The inheritance of this character is simple in most people but it may be more complicated in others. In the case of simple inheritance, the blue eye behaves as recessive to the dark eye (usually brown). Other intermediate shades of eye colour, green, greenish gray, hazel, etc are also found and it is difficult to explain the heredity of eye colour as a simple qualitative character.

4. Inheritance of MN blood group

Using immune sera against the blood of different persons beings can be classified into 3 blood groups called M.N. and MN, respectively. The red blood cells of persons with M blood type carry an antigen called M antigen and these cells can be agglutinated with a Similarly, N blood type can be agglutinated by anti-N serum MN blood type can be agglutinated by both anti-M and anti-N sera.

The inheritance of these blood types is simple Mendel N blood types are due to the presence of M and N genes dominant. The heterozygous (MN) will have MN-type, MM-type will have M-type and NN will have N-type of blood.

Phenotype and phenocopy etc.

In our previous discussion on genes and characters, we frequently used the two terms genotype and phenotype. As define earlier genotype is the sum total of genetic factors which an organism inherits from its parents for a certain character; individuals of the same genotype breed alike, and they produce similar type of gametes. Phenotype describe the observed character of an individual without any reference to its genetic constitution. Members with the same phenotype may not breed alike, because they may have different genotypes. In Mendalian inheritance in a cross of tall (TT) and dwarf pea plants (tt) the F_1 is tall genotype is different from TT genotype but both have the same phenotype as both are equally tall.

Phenocopy

Another term is Phenocopy, which should not be phenotype. Sometimes it is possible to bring about phenotypic changes by changing the environments. These apparently seem similar to those caused by genes as a result of mutation. The normal body colour of *Drosophila* fruit fly is light brown. Due to a mutation, flies with yellow body colour are also produced. If the normal pupae (brown) are fed on silver nitrate salt, their colour yellow. This silver nitrate induced yellow is genotypically the same as the normal type, but their phenotypes are different. The silver yellow types are the phenocopies of the true mutated yellow types will produce only normal types (brown) when interbred, whereas mutated yellow flies will always produce yellow progeny.

Lethal Genes

The expression of all minor and major traits in living organisms is controlled by genes. Genes which control the development (traits, such as the development of the brain or any other major character are more important for a normal function and are regarded as major genes. On the other hand, genes which control less ir traits, e.g., production of hair on the second segment of fingers minor genes. The absence of, or change in, minor genes may not adversely affect the vitality of the individual as do the major genes.

When a major gene is missing or is replaced by a mutated may cause the death of the individual. The genes which exert such a drastic effect on the organism as to kill it are called d lethal genes. In some cases bases, the effect of these genes is dominant and or immediate, homozygous individual for it dies. in the early embryonic stages after birth, while in the other cases, the effect may be delayed to permit organism to live for some time it is not necessary that the lethal always cause death; it may handicap the carrier individual in some important physiological activity. If this handicap could be overcome artificially, such individuals can successfully perform their life functions. Albino seedlings (lacking chlorophyll) in maize and barley are mutant lethal gene, and are incapable of synthesizing their own food. If fed on suitable nutrient media, they can grow normally and set seed. Similarly, certain hereditary human diseases are controlled by medicines now days.

Let us take another instance. In poultry, gene ‘C’ is responsible for the development of short crooked legs (creepers). This crooked abnormality puts the creeper birds at a disadvantage to compete the normal birds. When two creepers are inter-crossed, a ratio of 2 creepers and 1 normal is obtained instead of the usual 3:1 explained below:

Parents	creeper	x	creeper
Gametes	Cc		Cc
	C, c		C, c
		C	c

C	CC	Cc
c	Cc	cc

Genotypes	1CC:	2Cc:	1 cc
Phenotypes	dies	creeper	normal

When the creepers are back-crossed to the normal bird test cross ratio of 1 creeper and 1 normal is obtained. From these results two conclusions emerge: firstly, the creepers are heterozygous gene 'C', i.e. they are Cc and secondly, the missing class is the dominant homozygous class CC. Study on the embryology of the creeper normal chicken has shown that the mutant gene C cause retardation of growth, and the leg abnormality is most conspicuous. Dominant homozygous zygotes die during the first week of incubation.

When crossed to a normal, a creeper will produce half normal and half creepers as follows:

Parents	Cc	cc
Gametes	Cc, c	c
	C	c
c	Cc	cc

Genotypes	1 Cc	:	1 cc
Phenotypes	Creeper		Normal

Recessive lethal genes are carried in a heterozygous condition and show their lethal effect when a chance mating between such individuals occur to produce homozygous recessive

individual. In maize other crop plants, albino mutation is mostly lethal, since it checks the chlorophyll development, without which the plant cannot perform its photosynthetic activities.

A normal green maize plant = AA

Mutation for albinism occurs in one allele = Aa

(phenotypically not distinguishable from a normal green plant)

Carrier for albinism when crossed will produce normal plants in the 3:1 ratio as follows:

Parents Aa x Aa

Gametes A, a A, a

	A	a
A	AA	Aa
a	Aa	aa

Genotypes 1 AA : 2 Aa : 1 aa

Phenotypes Normal Normal Albino

PROBLEMS

1. A woman and her husband have normal skin pigment children, a girl and a boy are albino. What are the albino children and of the parents?

2. Both the albino persons from the above problem mated with pigmented people and produced children as follows:

(a) All the children of the albino woman were normal

(b) Half the children of the albino man were albino pigmented.

What are the genotypes of the normal persons?

3. A taster woman both of whose parents were t-----Non-taster man and they had four children, two tasters, and two non-tasters, Work out the genotypes of the family.

4. A brown-eyed man marries a woman whose eyes are also brown. Can they expect a blue-eyed baby? If so, illustrate you giving genotype of each.

5. What is a lethal gene? Give an example to show how a modifies the F_2 phenotype ratio of 3:1.

6. How can a lethal gene stay in a population'? Do you this gene is always dominant or recessive?

EPISTASIS

The Expression and Interaction of Genes

We have already discussed the Mendelian laws of (i) segregation and (ii) independent or random assortment of genes in the previous chapter. To properly understand the basis of these two laws, we can acquire a clear concept of the process of meiosis. Reduction of chromosome number is also of the genes carried on them, takes place during meiosis. Segregation of genes means the separation of the allele's gene pair through the separation of the homologous chromosomes, while independent assortment of genes refers to segregation of the member one pair of genes regardless of the members of the other gene pairs.

The phenotypic F_2 ratio is 3:1, when the individuals crossed in only one gene pair (monohybrid) and one of the two character forms dominant. The F_2 phenotypic ratio is 9:3:3:1, when the difference between the two crossing individuals is in the nature of two gene (dihybrid) and dominance is involved in both the characters. phenotypic ratio of 9:3:3:1 implies that a total of 16 possible combination resulting from the random union of 4 types of male and female gametes produce only four phenotypes; 9 of them show the double dominance phenotype (A-B-), 3 show one dominant and one recessive character (A-bb); 3 show the other dominant character and the recessive (aaB-) whereas only one has a phenotype representing both the recessive characters (aabb). It must be borne in mind that 9:3:3:1 ratio is dependent the following facts:

- (i) The difference between the two parents is of two independent gene pairs which determine mostly two different characters
- (ii) Dominance is involved in both the characters.
- (iii) The assortment of the genes controlling the two characters independent of each other.
- (iv) The union of the gametes is random.

If these conditions get disturbed, the usual F_2 phenotypic ratio 9:3:3:1 will also be upset. For instance, if the two gene pairs involved a dihybrid cross control expression of only one

character instead of the two gene pairs must be present to produce the given phenotype absence of the one or the other results in the non-expression phenotype and the 9:3:3:1 ratio will change.

So far, we have emphasized that a character is controlled pair of allele, and the crosses involving contrasting character segregation into definite Mendelian ratios. It has been, however, observed that in some cases more than one gene pair may be essential expression of a particular character and the same is also true inversely a single pair may affect more than one character. In the development of a character, there may be many genes involved, interacting in a manner. The phenomenon of interaction of non-allelic genes modifies dihybrid Mendelian ratio. In the usual dihybrid ratio of 9:3:3:1, classes may add up together on the basis of their phenotypic similarity but the number of total combinations remains the same, i.e., 1 change occurs only in the number of the phenotypic classes.

I. Modification of the 9:3:3:1 ratio by the lack of dominance

In snapdragons, a pair of alleles controls the expression colours i.e. RR produces red colour, Rr produces pink and rr white. Similarly, there is another pair of alleles affecting leaf shape that BB produces broad leaves, Bb intermediate and bb narrow. When RRBB is crossed with rrrb, the F₁ will consist of pink intermediate leaved plants and in the F₂ will be obtained this ratio i.e., 1:2:4:1:2:1. This is a modification of 9:3:3:1 ratio dominance is absent.

II. Modification of the 9:3:3:1 by epistasis

Sometimes, it so happens that the genes which are non-allelic, but affect the same characters of an organism, show interaction in same way as the effect of the recessive genes is masked by its dominant allele. When a non-allelic gene exerts a dominant influence over another, the dominating gene is said to be epistatic and the gene whose effect is repressed is regarded as hypostatic. This phenomenon is called epistatic. It may be noted that dominance involves allelic genes, while epistasis involves non-allelic genes. The interaction may take the form complementary gene action, supplementary gene action or an inhibiting action on the part of one of the genes. Several modifications 9:3:3:1 ratio representing the various types of gene interaction: briefly discussed here under:

(1) 9:3:4 ratio (recessive epistasis). To produce a black rat dominant gene must be present R.C; cc produces albino and is epistatic to R.

Parents	(Black) RRCC	x	rrcc (albino)
Gametes	(RC)		(rc)
F ₁	RrCc (All black)		
	RrCc x RrCc (Cross F ₁ amongst themselves)		
F ₂	9 R-C-	black	
	3 Rcc	albino	
	3 rrC-	cream	
	1 rrcc	albino	
	16		

The ratio is 9:3:4 and is a case of recessive epistasis.

(2) 12:3:1 ratio (Dominant epistasis). In summer squash to produce yellow coloured fruit, Y gene is essential. In some white varieties W gene is epistatic to Y gene. The epistatic W gene does not allow the hypostatic gene Y to produce yellow colour. Yellow colour appears when W is absent and Y gene alone is present. Both white yellow fruit colours are dominant over green.

Parents	(White) WWYY	x	wwyy (green)
Gamete	WY		wy
F ₁	WwYy		All white
F ₂	9 W-Y-		White
	3 W-yy		White
	3 ww Y-		Yellow
	1 ww yy		Green
	16		

The F₂ ratio is modified to 12:3:1 due to dominant epistasis.

(3) 9:7 ratio (duplicate recessive epistasis). In daisy, there are two yellow flowered varieties which are genetically different; the gene producing yellow color e.g., P in one is

different from the gene producing yellow color R in the other. The recessive alleles are epistatic. When both the independent dominant genes are brought together by crossing they produce purple color:

Parents	(Yellow) PPrr	x	ppRR (Yellow)
Gametes	(Pr)		(pR)
F ₁	PpRr (Purple)		
	PpRr x PpRr		
F ₂	9 P-R-	Purple	
	3 P-rr	Yellow	
	3 ppR-	Yellow	
	1 ppr	Yellow	
	16		

The F₂ ratio is modified to 9:7 and the genes involved here are complementary. Genes which are similar in phenotypic effect when present in different individuals, but produce a different expression y interaction on coming together are called ‘complementary genes’.

(4) 15:1 ratio (duplicate dominant epistasis). When two factors affect the same charact and the dominant allele of each acts as epistatic. This case may be illustrated by an example from the poultry.

Parents	(Feathered shanks) FFSS	x	ffss (Unfeathered)
Gamete	FS		fs
F ₁	FfSs	Feathered shanks	
F ₂	9 F-S-	Feathered shanks	
	3 F-ss	Feathered shanks	
	3 ff S-	Feathered shanks	
	1 ffss	Unfeathered shanks	
	16		

The F₂ ratio is modified to 15:1. Two identical pairs of genes with the same expression present in one individual are called duplicate genes.

(5) 13:3 ratio (dominant and recessive epistasis). An inhibiting gene is generally assumed to have no effect of its own but only inhibits function of another gene. In poultry, White Leghorn birds are genetically coloured birds but they are unable to develop colour because along the colour gene C they also possess an inhibitor gene I.

The white plumage in White Plymouth Rocks is recessive coloured plumage.

Parents	(White Leghorn) IICC	x	iicc (White Plymouth Rocks)
Gametes	IC		ic
F ₁		IiCc	White
F ₂	9 I-C-		White
	3 I-cc		White
	3 iiC-		Coloured
	1 iicc		White
	16		

I gene is epistatic (inhibitor) to C gene which develops colour where I is absent. The F₂ ratio is changed to 13:3.

(6) 9:6:1 ratio (factor interaction). In summer squash A and B genes are essential to the production of disc-shaped fruit. When A gene is separately present in different individuals each produces round-shaped fruit. Their recessive alleles a and b interact to develop elong fruit shape.

Parents	(Round fruit) AAbb	x	aaBB	(Round fruit)
Gametes	Ab		aB	
F ₁	AaBb			Disc-shaped fruit
	9 A-B-			Disc
	3 A-bb			Round
	3 aaB-			
	1 aabb			Elongated
	16			

III. 9:3:3:1 ratio resulting from factor interaction

The walnut comb shape in poultry birds results from the interaction between the two independent dominant genes R and P, which present in different individuals, produce rose and pea combs, respectively

Parents	Rose	RRpp	x	rrPP	(Pea)
Gametes		Rp		rP	
F ₁		RrPp	Walnut (interaction between dominant alleles)		
F ₂		9 R-P-			Walnut
		3R-pp			Rose
		3 rrP-			Pea
		1 rrpp	Single (Interaction between recessive alleles)		
		16			

Two things may be noted: (i) the F₁ resembles neither parent nor (ii) 1/16 I, birds have a new comb shape (single) not present parents and the F₁ These are the products of factor interaction.

PROBLEMS

Note: In poultry, gene R alone produces rose comb, while responsible for the pea comb. R and P together produce walnut comb and their recessive alleles pprr single comb.

1. What phenotypic ratios will be obtained from the following?

- (a) PPRR x rrpp
- (b) RrPp x Rrpp
- (c) RrPp x RrPP
- (d) RrPp x rrPP
- (e) RrPp x rrPp
- (f) RrPp x RRPP
- (g) RrPp x RRPP

2. Determine the genotypes of the parents in the following four problems:

- (i) A walnut rooster crossed with a pea-combed hen produced half of the chickens walnut and half with pea combs.
- (ii) Both the male and the female had walnut combs and their progeny segregated into 618 walnut and 2/8 rose.
- (iii) A walnut-combed male crossed with a pea-combed hen produced these off springs: 3/8 walnut, 3/8 pea, 1/8 rose and 1/8 single.
- (iv) A rose comb crossed with a pea comb produced half walnut and half rose.

3. Work out the test cross ratios for various modifications of dihybrid ratios you have studied.

Note: In the pea plant C and P genes together produce purple flower colour while individually each produces white flower.

4. What will be the flower colour of the plants result from following crosses?

- | | | |
|----------|---|------|
| (a) CcPP | x | ccpp |
| (b) CcPP | x | ccPP |
| (c) CcPP | x | ccPP |
| (e) CcPP | x | CcPP |
| (f) CcPP | x | ccpp |

5. Determine the genotypes of the parents in the following three problems:

- (i) A horticulturist made a cross between a purple and a white plant to obtain 3/8 purple and 5/8 white plants.
- (ii) In another cross between purple and white the segregation observed was 1/2 purple and 1/2 white.
- (iii) Two purple flowered plants were crossed and the progeny into 6/8 purple and 2/8 white.

6. Why did Mendel not mention factor interaction in his dihybrid pea crosses?
7. What is the difference between epistasis and dominance your answer with a suitable example.
8. What are the differences between Mendel's dihybrid ratio and the 9:3:3:1 ratio obtained from a cross between the rose combed and the pea combed birds?

Multiple Alleles

We have been talking a great deal about “alleles” and by now clear understanding of the meaning of the gene and its alleles must have been acquired. A gene can exist in several alternative forms. These alternative forms are called alleles. Alleles occur in pairs, and are located on chromosomes. Although, in the diploid individuals, only two alleles are present, it does not mean that there are only these two alleles for a particular character. There can be more than two alleles of a gene; as many as 20 or even more in some cases are reported in the literature. These sets of alleles are called “Multiple alleles”.

Every new allele arises as a result of gene change (mutation) somewhere in the ancestral line. It will be interesting to note that if there have been no mutated genes, it would have been rather impossible to determine the genetic basis of inheritance of characters. Members of an allelic series affect almost the same trait. The criteria for the identity of a multi allelic series are (i) the heterozygote should phenotypically resemble either of the parents or be intermediate between the two parents and (ii) alleles must segregate out into separate gametes producing 3:1 or 1:2 phenotypic ratio in the F_2 generation.

Members of the same multiple allelic series are conventionally designated by the same basic letter symbol and each allele being identified by a specific superscript or subscript. We shall now consider a few cases where more than two alleles have been reported:

(i) Rabbits have several alleles for their coat colour. The coat colour may be:

1. Full colour (grey or black body) (CC).
2. Chinchilla (traces of yellow lacking) $c^{ch} c^{ch}$
3. Himalayan (only extremities are coloured and the rest of the body white) ($c^h c^h$)
4. Albino (no colouring at all)

The existence of these four alleles for coat colour in rabbits can be confirmed by making crosses between any two of them and obtaining typical monohybrid ratio. This is how they behave in such crosses:

Cross I

	CC x cc	(albino)
F ₁	Cc--	(all coloured)
F ₂	3 C-	(coloured)
	1 cc	(albino)

Cross II

	(coloured) CC x c ^h c ^h	(Himalayan)
F ₁	Cc ^h	(all coloured)
F ₂	3 C-	(coloured)
	1 c ^h C ^h	(Himalayan)

Cross III

	(Himalayan) CC X c ^h c ^h	(albino)
F ₁	c ^h c	(all Hamalyan)
F ₂	3 c ^h -	(Himalyan)
	1 cc	(albino)

From these data it is clear that three different form in rabbit are determined by the three alleles of the gene ft (coat colour). Full colour is dominant over Himalayan and albino; Himalayan is dominant over albino. Crosses of these individuals with the ones having chinchilla is recessive to full colour have shown that chinchilla is recessive to full colour, but dominant over the other two, i.e, Himalayan and albino. The four rabbit colours for their phenotype and shown as under:

Phenotype	Genotype
Full colour	CC, Cc ^{ch} , Cc ^{he} , Cc
Chinchilla	c ^{ch} c ^{ch} , c ^{ch} c ^h , c ^{ch} c
Himalayan	c ^h C ^h , c ^h c
Albino	cc

Similarly, in the drosophila, the colour of eye which is a sex-linked character is controlled by many alleles. Quite a large number of shad of the eye colour may be observed, each dependent upon a separate allele. There may have been reported more alleles for the eye

colour in drosophila. Red eye colour is the wild form, and is dominant over all the other alleles, while different combinations among the mutated forms show intermediate dominance.

Phenotype	Genotype
Red	WW
White	w w
Wine	w ^w w ^w
Coral	w ^{co} w ^{co}
Blood	w ^{bl} w ^{bl}
Eosin	w ^e w ^e
Cherry	W ^{Ch} W ^{Ch}
Apricot	w ^a w ^a
Buff	W ^{bf} W ^{bf}
Tinged	w ^t w ^t
Ivory	w ^I w ^I
Pearl	W ^P W ^P
Honey	w ^h w ^h

Blood groups in man

When a foreign substance called antigen (protein) is introduced to the body, it stimulates a reaction resulting in the formation of antibodies in the blood serum. Antibodies are a type of protein, and when proteins react with the antigen and destroy it. As a result of this reaction blood cells agglutinate (clump) and thus block blood circulation in blood vessels of the recipient, causing death.

In man, blood groups are controlled by multiple alleles. There are four kinds of blood groups according to the type of antigen present in group A, group B, group AB and group O. Persons having blood A will have antigen A in his cells, but no antibody against this antigen. Similarly, persons with blood group B will have antigen B and no antibody against B. When an antigen is not present in the blood cells, the corresponding antibody is present.

The following shows these relationships:

Group	Genotype	<u>Blood contains</u>		<u>Reaction with antigen</u>	
		Antigens	Antibodies	Anti. A	Anti.B
A	$I^A I^A, I^A i$	A	Anti.* B	Positive	Negative
B	$I^B I^B, I^B i$	B	Anti. A	Negative	Positive
AB	$I^A I^B$	AB	None	Positive	Positive
O	ii	None	Anti. A Anti B	Negative	Negative

*Antibodies

It is noted that the four blood groups in man are due to combination of three multiple alleles and are inherited in a Mendelian fashion. Each antigen is due to a dominant gene and a blood group may be homozygous or heterozygous. The heterozygote will produce two types of antigens. The gene responsible for A type of blood group is symbolized as I^A and for O blood group as i the former is dominant over the latter, An A type individual may be homozygous ($I^A I^A$) or heterozygous ($I^A i$). Similarly, gene I^B for blood group B is dominant over i gene and the individuals of the group B may be $I^B i$ or $I^B I^B$. A B blood group type has only one genotype, $I^A I^B$, they are co-dominant and both express themselves in the same individual. O type blood group persons always have homozygous recessive genotype (ii).

Transfusion of blood between two persons is possible only if they have compatible blood groups. The parenthood of babies may also be established by the study of the blood groups, in certain situations.

Rhesus alleles in man

In addition to A and B antigens, there are numerous other antigens, like M, N and Rh antigens. The Rh antigens like those of M-N series are different from the A-B antigens in that normal antibodies against them are not present in the human serum, but when Rh antigens are introduced into the blood, they provoke the formation of corresponding antibodies. When Rh-positive blood (containing Rh antigens) is introduced into a person whose blood lacks the Rh antigen (Rh-negative), no harmful reaction will occur, but if another transfusion takes place at a later time, the antibodies which started forming at the first transfusion, might have grown so strong as to agglutinate the transfused blood cells resulting in death in some cases. Similarly, marriage between an Rh positive man and an Rh- negative woman will be undesirable, as after the second or third child, further conceptions will result in abortions for the reason explained above.

Compound loci (Pseudo-alleles)

In some organisms, certain areas on some chromosomes were previously considered to be single loci which carry only a single gene, but later, such genes were found to subdivide by crossover. The subdivisions were at first called false alleles or pseudo-alleles now they are considered sites of compound loci. These genetic units or subunits, affecting the same character lie very close together on the same chromosome. Due to this closeness they tend to remain together but occasionally they crossover and produce a combination effect. Compound loci have been discovered in drosophila (lozenge and normal eye, white and apricot eye colour) and in maize (pigment production and nutritive content of seed).

Isoalleles

As a matter of fact, it should not be assumed that the dominant or wild type allele is always and everywhere the same. For example flower colour allele in the pea may not be the same in all the varieties, but we know that it shows identical dominance over white. This type of genes has been worked out in drosophila where the red eye colour different degree of dominance over the white eye colour. Such genes are called isoalleles because they are alike in their homozygous ef differences appear only in special combinations.

PROBLEMS

1. As many as 13 alleles for eye colour in drosophila have b How many of them may be present together in one individual? Will they all be present on the same locus?
2. In rabbits, full colour, chinchilla, Himalayan and albino allele for full colour and show dominance in the same order
 - (i) From a cross of two full coloured rabbits if 1/4 Four Himalayans was obtained.' What could be the genetic constitutions of the parents?
 - (ii) From another cross between chinchilla and Himalayans ratio of 1/4 albino, 1/4 Himalayan and 1/2 obtained. What is the genetic constitution of the parents?
3. What will be the phenotypes of the off springs from the following crosses?
 - (a) $Cc^{ch} \times c^{ch} c^{ch}$
 - (b) $Cc^h \times cc$
 - (c) $c^{ch} c^h \times c^h c$
 - (d) $c^{ch} c^h \times cc$
4. A woman belongs to blood groups A and her husband to B. Their children have all the four blood groups (A, B, AB and O). What are the genotypes of the parents?
5. A woman of blood group A gives birth to four children. One baby belongs to the group O and the rest of the children have all A group What can be the genotypes of the woman and her husband?
6. A boy whose paternity is disputed has blood group O. He claims to be the rightful heir to his father's property. His father belongs to AB group. If you are the judge, how will you decide, his claim?

Sex-Linked Inheritance

So far we have considered inheritance of such characters as were assumed to be controlled by one or more genes located on the chromosomes other than sex chromosomes. The chromosomes other than sex chromosomes are called autosomes. In humans, there are 46 chromosomes and of these, 44 are autosomes and 2 sex or X and Y chromosomes. Similarly, in other living organisms there are generally 2 sex chromosomes and the rest, the autosomes. In some insects, you might come across more or less than 2 sex chromosomes.

In human beings, there are two sex chromosomes and called X and Y chromosomes. Any variation in their number produces an abnormal individual. A human embryo having XY chromosomes will develop into a boy and the one with XX chromosomes will develop into a girl. It will be seen that the woman will produce only one type of eggs, i.e. (X) (X), whereas the man will form two types of sperms carrying X and the other Y. The Y sperm will produce a boy fertilizes an egg, i.e. the genotype will be XY and the X produce a girl when it fertilizes the egg, i.e. the genotype will be XX.

In *Drosophila*, as well as in some other insects the same mechanism for sex determination operates as in humans, i.e., XY will be male and XX female. But in birds, the situation is just the reverse for instance, a cock, will be ZZ. To keep this mechanism from that found in man and other animals, use of different preferred. One can use Z and W to show the sex constitution. A male bird will then be designated as ZZ and the female bird as ZW.

In man, the male sex develops when the two sex chromosomes are dissimilar, i.e., XY, while in birds, the male sex develops when chromosomes are different i.e., ZZ. The sex which depends upon the presence of two different sex chromosomes is called "heterozygous or heterogametic sex" while the other type will be "homozygous homogametic". In man, the male sex is heterogametic whereas in birds the female sex is heterogametic.

We encounter still a different mechanism in some insects. For example, the bug will have all the 14 chromosomes while the male have only 13 chromosomes. The male will, therefore, produce two types of sperms, one with 6 chromosomes and the other with 7 chromosomes. The 6

chromosome sperm will produce a male and the 7 chromosome sperm will produce a female bug or uniting with a 7 chromosome egg.

Sex-Linked Characters in Man

Colour blindness in man is an instance of sex-linked characters, i.e the gene or genes developing colour blindness are on the two sex chromosomes. Of the two sex chromosomes in man (XY), Y is, by and large, empty of genes. Only the X chromosome contains the genes which are called sex-linked genes. The character, colour blindness is recessive; it will develop a female only when there are two recessive alleles, one on each X chromosome. So, for the colour blindness to develop in male only one allele on the X chromosome, will be needed because the other is Y, which is empty. A female of $X^b X^b$ constitution will be colour blind. A male $X^b Y$ Constitution will be colour blind. The gene B is dominant and will not produce colour blindness, as is shown below;

Parents	Normal vision female $X^B X^B$	colour blindness $X^b y$ male
Gametes	X^b	X^B, y
F ₁	$X^B X^{*b}$	$X^B y$

Neither sex will show colour blindness.

Parents Female normal vision $X^B X^b$ x $X^B y$ male normal vision

		Female gametes	
		X^B	X^b
Male gametes	X^b	$X^B X^b$ female	$X^b X^b$ female
	Y	$X^B y$ Male	$X^b *y$ Male

shows the pathway of criss-cross inheritance; male transmits its sex-linked character (colour blindness) through his daughter to the grandson.

Similarly, hemophilia a sex linked disease in man and it as a recessive character. The same explanation as for colour blindness holds good for this character.

Sex-Linked Genes in poultry

As we have said before in poultry (birds) the female is the gametic sex contrary to man where male is the heterogametic sex. Barred plumage in poultry is a sex linked character and is dominant over plumage. The allele for barred may be designated as B and its allele as b for black.

B-will be female and barred, Bb will be male and barred, bb will be male and black

Parents	Female barred	$Z^B W$	x	$Z^b Z^b$	Male non-barred
Gametes		Z^B, W		Z^b	
F_1 's		$Z^B Z^b$		$Z^b W$	
		(Male barred)		(Female non-barred)	

Cross the above parents.

Female gametes

Z^b W

Male gametes	Z^B	$Z^B Z^b$ Male	$Z^b Z^b$ Male
	Z^b	$Z^B Z^b$ Female	$Z^b W$ Female

1/2 male: 1/2 female. Both males and females half barred and half non-barred.

Non-Disjunction of X-Chromosomes in Drosophila

In drosophila, colour of the eye is sex-linked. Red eye colour is dominant over white eye colour. If W stands for red colour and w for white, then $X^W X^W$ will be red eyed female. $X^w X^w$ will also be red-eyed female, $X^w X^w$ will be white-eyed ale. $X^W Y$ will be red-eyed male and $X^w Y$ will be a white eyed male.

If white-eyed drosophila females $X^w X^w$ are crossed with red-eyed males $X^W Y$ all the females obtained should b red-eyed and all the males white-eyed. This virtually happens in almost all the cases. But some exceptions are also detected, i.e., one fly out of 2000-3000 F_1 offsprings is a red-eyed male and a white-eyed female. Bridges, a student of Morgan, made these experiments in 16 and showed that the exceptional behaviour mentioned above is due to the non-disjunction of the two chromosomes at the time of gamete formation (meiosis). This would result in the production of diploid gametes (XX) instead of haploid (X). The diploid gamete (XX) having both the recessive alleles for white eye colour can produce white-eyed female on fertilizing with a normal gamete bearing Y the so-called empty chromosome. Their genotypes will be XXY. Similarly, on account of non-disjunction, one of the two types of female gametes (eggs) will have no X chromosome. A sperm having X^w may unite with an egg having no sex chromosome and will produce a male with red eyes.

Primary Non-Disjunction

	White eye female	Red ey male
Parents	$X^w X^w$	$X^w y$

Eggs due to non-disjunction

 X^w $X^w X^w$ X^w

$X^w X^w$ Red female	$X^w X^w X^w$ Super female usually dies	X^{w-} Red male sterile
$X^w Y$ White male	$X^w X^w Y$ Exceptional white female	$Y-$ dies

Exceptional white-eyed female ($X^w X^w Y$) and red eyed male X^{w-} arise because the x chromosomes sometimes do not disjoin during egg formation.

Secondary Non-disjunction

White eyed

Red eyed exceptional female

Parents

 $X^w X^w Y$ X $X^w Y$

Female gametes

Attached X chromosome in drosophila

Another exceptional behaviour of the sex-linked inheritance was discovered by Morgan which varied from the normal criss-cross inheritance and the non-disjunction pattern already described. For instance, an female, when crossed to a normal (brownish grey) male, produced only yellow daughters and normal sons, whereas, in normal sex inheritance, all the female should have been brown and males yellow. Morgan explained that the x chromosomes, at the time of gametes formation, remained attached together and thus produced two types of eggs namely, one with attached XX chromosomes and the other without X chromosome. A cross between yellow coloured female and a normal male fly produces the exceptional XXY female.

This yellow female XXY when crossed to normal male will produce two types of gametes XX and Y; the two attached XX either go to polar body or remain in the egg.

	Exceptional yellow female		normal red male				
Parents	$X^r X^r Y$	x	$X^R Y$				
	Female gametes						
				$X^r X^r$			
Y	<table border="1"> <tr> <td>$X^r X^r X^R$ Usually dies</td> <td>$X^R Y$ Red male</td> </tr> <tr> <td>$X^r X^r Y$ White female</td> <td>YY dies</td> </tr> </table>	$X^r X^r X^R$ Usually dies	$X^R Y$ Red male	$X^r X^r Y$ White female	YY dies		
$X^r X^r X^R$ Usually dies	$X^R Y$ Red male						
$X^r X^r Y$ White female	YY dies						
	X^R						
				Y			

The following inferences can be drawn:

- I. Zygotes of the constitution $XX X$ usually die but sometimes may survive and are called super females.
- II. Zygotes having YY but no X usually dies
- III. Yellow exceptional females with two attached XX at chromosome survive and then, besides the above types, we have normal red males produced.

sex-linked inheritance and work on non-disjunction provided a brilliant proof for the chromosome theory of heredity.

PROBLEMS

1. Why is there a greater proportion of colour-blind boys than girls in human population.
2. In drosophila, vestigial gene is present on chromosome No. 2 (autosome). Will you call it a sex-linked character?

3. A colour-blind man marries a normal vision woman whose father was also colour-blind. What type of vision expected in their children?
4. A man and a woman, both of normal vision marry produce two daughters having normal vision, and (one normal and the other colour blind). Both the daughters marry normal vision men. All children from one normal girl are normal while 50% boys from the other are colour-blind. What are the genotypes of the parents?
5. In human beings, hemophilia is a recessive sex-linked character. explain the phenomenon of crisscross inheritance of the character?
6. If the mother is colour-blind and the father of normal vision, what will be the vision of their boys and girls?

Inheritance of Quantitative Factors or Multiple-Factor or Multigenic Inheritance

In the previous sections, we have discussed genetic laws governing inheritance of qualitative characters. Such characters are not continuous in their expression and are classifiable into different groups. For instance, in pea, on which Mendel performed his classic experiments, all the 7 characters were qualitative and displayed discontinuous variation. The flower position was either terminal or axillary; the seed either round or wrinkled, the pea plant was either tall or dwarf and so on and so forth. Such characters are called qualitative, discontinuous, or Mendelian.

On the other hand, we come across several other characters in plants and other living organisms, whose expression is continuous, i.e., they cannot be divided into distinct classes; there are no breaks in their beings behaves as a continuous character. Similarly, body weight, plant yield, mental faculties and several other characters are continuous characters. In these cases, one cannot distinguish and count different forms of a character merely by looking at them, as is possible in qualitative characters. But, measurements in the appropriate scale are made in order to study the mode of their expression and inheritance. Since the study of these characters involves their measurements, they are called quantitative or metric characters

Genetic studies on quantitative or metric characters have demonstrated that the inheritance of these characters is not as simple as that of qualitative or Mendelian characters, but it is rather quite complex and their inheritance is termed multiple-factor or multigenic inheritance as contrasted with the simple inheritance of a qualitative character where only one, two, three or four genes may be involved.

Although inheritance of quantitative characters is multigenic i.e involving many gene more or less equal in effect, yet the same Mendelian laws are applicable in. this type of inheritance, as in simple inheritance. These genes segregate and assort independently (subject to linkag because of the operation of many genes, the number, of phenotypes very large and since the genes have equal effect and the environment effect is overwhelming they produce a continous expression of character which is not classifiable into distinct groups.

The individual effect of such genes is relatively small and highly susceptible to environmental conditions. In animals, milk yield is a quantitative character. A cow, for instance,

may have the potentiality (genotype) of yielding 40 lbs. of milk daily if feed on reduced in quality or quantity; the milk yield will suffer a corresponding reduction. The reduction in ration has not actually affected the genotype of the animal but has changed phenotype (milk yield). It is evident that a variation in the environment affects the expression of the quantitative characters, but similar changes are not so conspicuously expressed for qualitative characters. Another feature of the quantitatively controlled character is that the number of genes involved is usually large and there is a correspondingly large number of genotypes and phenotypes in the segregating generations.

Multiple factors or multiple genes or polygene should not be confused with multiple alleles. Multiple alleles (skin colour in rabbits) are members of an allelic series and in a diploid organism. Whereas several multiple genes, effecting a particular trait are present together in a diploid organism and variation in the number of contributing genes (plus, major) brings about a change in the phenotype.

Pleiotropy

Sometimes a gene may have more than one effect, i.e. influences more than one character simultaneously. It has long been blue-eyed white cats are always deaf; blue eye gene also causes deafness. In wheat the presence of awns usually has a pleiotropic effect on grain yield. Awned wheat is better yielders than those without awns.

Kernel Colour in Wheat

The first case of quantitative inheritance was studied by Nilsson-Ehle (1910-13) about kernel colour of wheat. He crossed varieties differing for red and white kernel colour, and observed that the F₁ grains were intermediate in colour between the two parents. The F₂ raised from the selfed seed of F₁ produced a continuous variation ranging from red to white which was grouped arbitrarily into 5 classes. 1/16 of the total F₂ plants resembled red parent in grain colour and another 1/16 the white parent and the rest 14/16 plants were midway between the two parent for colour expression. Of these 14/16 plants, 6/16 were like the F₁ 4/16 were lighter than the red parent and darker than F₁, and 4/16 were intermediate between F₁ and the white parent. This pattern of segregation showed that two gene pairs controlled the inheritance of kernel colour and

that they were cumulative (additive) in their effect. In another study the same author discovered that three gene pairs instead of two, controlled kernel colour difference.

Skin colour in man

In man, skin colour is due to the presence of melanin, which is genetically controlled. It is believed that several genes may be involved. A negro has the maximum amount of melanin pigment while a white possesses none or a very small amount of it. In other words, negroes possess all the colour genes and the whites recessive alleles. From marriage of a negro and a white all the children are mulattoes (intermediate in skin colour). When the mulattoes marry together, their children (F_2) show all grades of skin colour between black and white. The melanin production is, however, greatly influenced by exposure to sunlight. The more the sunlight the more melanin and vice versa.

Plant height in wheat

We shall now consider inheritance of another quantitative character plant height in wheat. It has been determined that plant height in wheat is controlled by several major and minor genes. Let us suppose that 5 gene pairs control inheritance of plant height, dominance exists at each locus and each dominant allele contributes two units to the expression plant height. Consider this cross between a tall and a dwarf parent dwarf parent measures 40 inches and the tall parent 60 inches in height.

Parents	AABBCCDDEE	x	aabbcc
	60"		40"
F1	AaBbCcDdEe (50 inches tall)		

To obtain frequency of various classes in the F_2 , expands binomial $(A + a)$ is helpful, where A is the dominant allele recessive.

Frequency	Genotype	Phenotype
I	A^{10}	60" tall
10	$A^9 a^1$	58" tall

45	$A^8 a^2$	56" tall
120	$A^7 a^3$	54" tall
210	$A^6 a^4$	52" tall
252	$A^5 a^5$	50" tall
210	$A^4 a^6$	48" tall
120	$A^1 a^7$	46" tall
45	$A^2 a^8$	44" tall
10	$A^1 a^9$	42" tall
1	a^{10}	40" tall

It will be observed that in the F_2 of this cross, one plant out of 1024 plants one is 60" tall, 10 plants are 58" tall; 45 plants are 56" tall and so on down to one plant being 40" tall which does not have any dominant gene.

A large variety of characters behaves quantitatively and of their inheritance may be attempted as shown above. Other methods are also available for this purpose.

PROBLEMS

1. What do know about a qualitative character? Make out important difference between them. List at least five characters of common plants and domestic animals that behave either qualitatively or quantitatively.
2. It is frequently observed that the seed size within the same crop variety and even on the same plants shows considerable variation. Explain.
3. Assume that skin colour in humans is controlled by two additive gene pairs. The genotype of a negro may be written as AABB and that of a white aabb. What will be the skin colour of children from a marriage between a negro and a white and between the two F_1 's of this mating?
4. If two mulattoes from the above problem marry each a white and a black person what will be the skin colour of the children from each marriage'?

5. Suppose per-plant seed yield of a certain homozygous wheat variety is 3 grams and that of another 9 grams and suppose further that the difference in grain yield between the two plants is due to 3 gene pairs acting cumulatively. What will be the grain yield of F_1 and F_2 plants?

6. Assume that the difference in the height of the two rice varieties, measuring 18" and 34" tall, is due to four pairs of additive genes. Make a cross between 18" tall (aa bb cc dd) and 36" tall (AA BB CC DD) plants. What will be the phenotypes of the F_1 and F_2 plants resulting from this cross. Also figure out the contribution of each plus gene.

7. Three gene pairs with equal effect are supposed to control fruit weight in the squash plant. Fruit in one variety weights 5 lbs. (AABBCC) and in the other 2 lbs. (aabbCC). F_1 and F_2 generations are raised from the cross between these two varieties.

What will be:

- (i) The effect of each contributing gene.
- (ii) The fruit weight of F_1 and F_2 plants.
- (iii) The number of genotypes and phenotypes in F_2 .
- (iv) The proportion of plants carrying 4 lbs and 3 lbs fruits.

8. If the plants (AaBbCc) with 3 lbs fruit from the preceding intercrossed, how will fruit weight segregate in the progeny?

9. Two maize varieties having yellow and white kernels were crossed. In the F_2 progeny, 1/64 plants had yellow kernel like yellow and 1/64 were white like white parent, but 62/64 were between the two parents. What are the genotypes of the parents and also calculate the number of genes involved?

Linkage and Crossing over

The two most important genetic laws that Mendel formulated as a result of his studies on the pea plant are (i) Law of segregation and (ii) Law of Independent Assortment. According to the law of segregation, two homologous chromosomes, one each contributed by the two parents segregate in the maturation process and are included into different gametes. Segregation of homologous chromosomes brings about segregation of the two alleles of the allelic pair. The Law of Independent Assortment states that, while segregation of the members of the pair of homologous chromosomes must take place, each of these two homologous chromosomes can form all possible combinations with the rest of the chromosomes at the time of gamete formation. That is to say the chromosomes assort independently of one another, have no preferential association and are included into the gametes in haploid number.

What is Linkage? Genes are on the chromosomes. There are thousands of genes determining expression of all the characters of a plant or any living body. All these genes have got to be located on the limited number of chromosomes that a plant may have consequently several genes must be located on individual chromosome the same are therefore linked by virtue of their being present on the same chromosome or to enter the gametes in parental combinations.

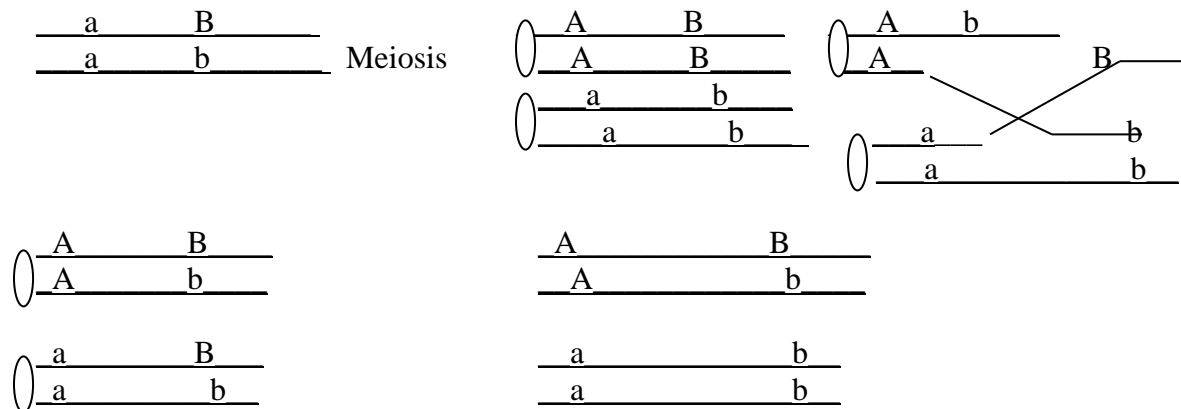
What is crossing over? At the time of gamete formation (gametogenesis) genes on a particular chromosome will be carried together wherever that chromosome goes. But, this does not happen all the time and all of the genes on a chromosome do not go together due to the breakage of chromosomes and the resultant exchange of the homologous part of the chromosome during synapses. Crossing over may be defined as the tendency of genes to enter the gametes in combinations other than parental.

The exchange or crossing over in the homologous chromosomes occurs in almost all animals and plants with a few exceptions. In male *Drosophila* and female silk moth crossing-over does not occur and linkage is complete.

The crossing-over occurs at the four-strand stage during prophase of meiosis. At the point of chiasma (cross-shaped configuration) the broken ends of the non sister chromatids reunite and

thereby the exchange of genes takes place. These chromatids separate at the time of second meiotic division and the cross-over chromatids separate the parental combinations and enter into different gametes

A crossing-over in an organism heterozygous for two gene pairs is shown diagrammatically



The cross-over gametes will be less frequent because:

1. Only 1/2 of the chromatids take part in the crossing-over
2. All the chiasmata do not represent a crossing-over.

Factors effecting the crossing-over

1. Environments: High temperature usually increases the rate crossing over.
2. Age: Aged individuals may show more crossing-over.
3. Distance from the centromere: The Genes in the immediate vicinity of the centromere do not show crossing-over
4. Distance between the genes: The closer the distance between the genes, the stronger the linkage and less the crossing-over.

Linkage group

A linkage represents all the genes situated on a chromosome. There are as many linkage groups as the number of haploid chromosome in a particular organism.

Linkage studies in the pea

Bateson and Punnett (1906) discovered the linkage of some characters in sweet peas. They, however, could not present a satisfactory explanation of this phenomenon as they did not believe that genes are on the chromosomes. They crossed two varieties of the pea, i.e. one with a purple flower colour and long pollen (PPLL) and the other with red flower and round pollen (ppll). In an earlier experiment they had noted the flower colour and the pollen size were controlled by different genes. F_1 was purple and long (PpLl), while the F_2 , did not conform to the law of independent assortment as the ratio of 9:3:3:1 was not obtained. Parental types were more frequent and the two classes (recombination's) occurred less frequently. The two gene combinations PL and pl stayed together more frequently came from the parent, and their separation occurred less frequently to produce new combination.

From these results, they developed the theory of coupling repulsion. As understood today, when the two dominant characters for that matter, the two dominant genes are on the same chromosome their corresponding recessive alleles on the other homologue, the phenomenon is called coupling and when the two dominant genes are located on two different chromosomes, the phenomenon is termed repulsion. Suppose A and B are linked, then a cross between AABB x aabb will show the coupling and a cross between AAbb x aaBB will show the repulsion phase.

Linkage studies in drosophila

Morgan (1910) came across a similar situation in his studies on drosophila. He was able to explain the tendency of the two characters to remain together in crosses by demonstrating that these characters controlled by the genes on the same chromosome and thus replaced of coupling and repulsion with that of linkage and crossing over.

Detection of linkage

Detection of linkage is possible when two or more genes considered simultaneously. In the event of the two genes being located on two different chromosomes, independent assortment of these genes take place and a plant of AaBb genotype will form four types of AB, Ab, aB and ab in equal numbers. If the two genes A are linked, i.e., they are on the same chromosome, the AaBb plant form the same four types of gametes, because of the crossing-over, but however, the

two linked genes will tend to remain together and be in the same gamete, and consequently, AB and ab gametes will be numerous than the Ab and aB gametes.

Let us now consider a cross that Hutchison made in maize one variety whose seed was coloured and normally filled (coloured and the other variety having colourless and shrunken seed (c shrunken). The colour gene was dominant over the colorless full endosperm was dominant over the shrunken. Using genetic formula we can describe his results as follows:

	Coloured full	x	Colourless shrunken
Parents	CCFF (CF/CF)		ccff (cf/cf)
F ₁	CcFf (CF/cf) (coloured, full)		

He test-crossed CcFf with ccff and got the following results:

- (1) CcFf (CF/cf) = 4032 coloured full
- (2) Ccff (Cf/cf) = 149 coloured shrunken
- (3) ccFf (cf/cF) = 152 colourless full
- (4) ccff (cf/cf) = 4035 colourless shrunken

It will be observed that parental combinations in the progeny of this test cross are far in excess of the new combinations, i.e. the coloured full and the colourless, shrunken being the parental combination, are far more numerous. This proves that, of the four types of the gametes formed by Ff (F₁), the parental gametes CF and cf are very many compared to a few recombination's, Cf, cF. If assortment were independent then all of these four gametes would have been equally frequent. This reveals that two genes C and F are on the same chromosome (they are linked). such a test cross where parental combinations occur in the progeny more frequently than the new combinations, it is indicative of linkage.

What is linkage calculated?

1. Testcross method: The most convenient and efficient method determine strength of linkage is to make a test cross (cross F₁ with double recessive). The cross will yield four phenotypes if dominance is involved. If there is no linkage, all the four phenotypes i.e., the parental as well as the new combinations will be equal in number. If the two genes linked the four types will be unequal, the parental ones being more frequent.

If the number of parental combinations and of new combinations separately added the percentage of each can be worked out. For instance, in the above testcross of Hutchison, the total parental combinations are $4032+4035=8067$, and the total of new combinations is $149+152=301$. The percentage of parental combinations will represent strength of linkage and the percentage of new or recombination will indicate the cross-over value or recombination value.

$$\text{Recombination or cross-over value} = 301/8368 \times 100 = 3.6 \%$$

$$\text{Linkage strength} = 8067/8368 \times 100 = 94.4 \%$$

If one value is known, the other is obtained by subtracting it from one hundred. The two genes are 3.6 units apart on the chromosomes.

2. F₂ analysis to calculate the crossing-over value of the two linked genes. F₂ data from dihybrid individuals can also be subjected to the analysis to determine the strength of linkage or the crossing-over value. The key to F₂ analysis is to look for the double recessive individuals in the F₂ population, count them and compute their relative frequency (fraction). It will be realized that a double recessive male and female gametes. So the frequency of the double recessive individuals is equal to the product of the frequencies of the male equally frequent in population, the frequency (fraction) of the double recessive gamete (male or female) can be obtained by taking the square root of the frequency (fraction) of the double recessive individuals. The fraction of the double recessive gametes thus obtained is also the fraction of the double double recessive gamete thus obtained is also the fraction of the dominant alleles are carried on the other homologous chromosomes. When the frequency of ab and AB gametes are known, the frequencies of their recombination or cross-over types can be calculated by subtracting the sum of AB and ab from one. The following example, where A&B are linked, may be considered

Parents	=	AB/AB	x	ab/ab
F ₁	=	AB/ab		
F ₂	=	A-B- = 160		
		A- bb = 40		
		aaB- = 40		
		aabb = 60		

if the entire F₂ individuals are added up, the frequency (fraction) of aabb, the double recessive individuals can be calculated

$$\text{Frequency of aabb} = 60/300$$

Frequency of ab gametes; their

$$\text{Sum} = 0.44 + 0.44 = 0.88$$

The frequency of Ab and aB gametes will be $1 - 0.88 = 0.12$. Since Ab and aB are the cross-over types, their frequency in the gametic population is the cross-over value of the A and B genes. So the cross-over value = 0.12 or 12 % and linkage strength = 0.88 or 88 %

PROBLEMS

1. Could you name the scientist who first discovered linkage; how did he explain it?
2. How is gamete formation affected in a dihybrid organism where two genes are linked, compared to when the two genes are independently inherited? Illustrate your answer by cytological diagrams.
3. Suppose that the two linked genes B and C show 14 percent crossing over. In what proportion will be F₁ resulting from a cross x produce the parental and recombination gametes?
4. If the F₁ from the preceding problem is test crossed with the double recessive parent, what will be the genotypes of the backcross progeny? Also figure out the proportion of parental classes.
5. A maize variety with coloured full grains was crossed with colourless, shrunken grains. The F₁ was test crossed with the double recessive parent. The test cross progeny was classified as follows:

Coloured full — 453

Colourless full — 48

Coloured shrunken — 52

Colourless shrunken — 447

Write the genotype of each class and work out the strength of linkage and cross-over value.

6. A tomato variety having tall vine and round fruit was crossed with another with dwarf vine and pear fruit. The F_1 plants were all tall and bore round fruit and were test crossed to raise progeny.

Tall round	=1509
Dwarf pear	=1441
Tall pear	=556
Dwarf round	= 496

Do you think that the genes for tall vine and fruit shape are linked? If so, how strongly?

7. Now make a cross between homozygous tall pear and varieties as obtained in the preceding problem. Test with double recessive. What will be the genotypes, phenotypes their proportion in the testcross progeny; calculate the linkage and the cross-over value.

8. What do you mean by a linkage group? How many linkage groups will there be in maize whose (2n) chromosome number

9. Consider the following cross in pea and calculate the value:

Parents	Coloured terminal <u>CT</u> X Colourless axial
	CF
F_1	Coloured terminal <u>CT</u>
	ct
F_2	C--T-- =424
	ccT-- = 99
	C--tt =102
	cctt =91

10. Comment upon the following statements.:

- Linkage violates both the Mendelian laws.
- Mendel was lucky not to run into linkage in his experiments.
- Linkage can be detected in homozygotes.
- Mendel had a very small chance to discover linkage.

The Modern Concept of Gene

According to the classical concept of gene which primarily developed from the Mendelian analysis of genetic data, a gene is an indivisible entity and at once a unit of function, recombination, and mutation. This notion implies that a given segment of the chromosome responsible for a particular genetic activity would, as a whole, be the basis of various genetic functions, like segregation, recombination and mutation.

More sophisticated analysis of these genetic phenomena on a molecular level in recent years has provided a new, more comprehensive understanding of the gene. As a result, the old, classical concept of gene changed drastically. In the modern enlightened sense, the gene is no longer to be considered a unit of function, recombination and mutation but, on the other hand, these phenomena are now known to occur at different molecular levels. For instance, a unit of recombination and or mutation is not a unit of function. These three units are not of the same dimensions. The unit of function is much larger than the units of recombination and mutation, the latter two units (recombination, mutation) being enclosed within the former (function). Since these units have different entities, they are named differently. The unit of function is called “cistron”, the unit of recombination “recon”, and the unit of mutation “muton”.

Structure of Gene

Chemically speaking, the genetic material present in the chromosomes is DNA (Deoxyribonucleic acid) and a gene is represented by a given sequence of the nucleotides, of which the DNA is composed. There are four kinds of nucleotides in the DNA corresponding to the four nitrogenous bases namely (i) adenine, (ii) guanine (purines), (iii) cytosine and (iv) thymine (pyrimidines). Each of these four bases along with pentose sugar and inorganic phosphate makes a nucleotide. These nucleotides are the building blocks of the DNA molecule which is a double-stranded, helical structure with enormous molecular weight. The DNA molecule provides the basis of life and is rightly recognized as the master molecule.

As shown from the DNA structure illustrated, the two strands are intertwined around each other in opposite directions. Between two strands, adenine pairs with thymine and guanine with cytosine are held in position by hydrogen bonding. The two strands open permit DNA replication. According to more recent studies, the replication can take place with unopened strands. Anyway, the process and mechanism of replication are quite intricate. DNA not only replicates itself but also directs the synthesis of RNA (ribonucleic acid).



DNA, RNA is a single-stranded molecule consisting of the same nucleotides except thymine which is substituted by uracil. It acts as a messenger between DNA and the cytoplasm factory for protein synthesis.

Protein Synthesis

As explained above, a certain sequence of the nucleotides equivalent to a particular gene. This sequence (gene) is transcribed into RNA and carried to the cytoplasm to direct the synthesis of a special portion of protein. A protein molecule consists of twenty different amino acids arranged in a particular sequence which is determined by sequence of the nucleotides in the DNA. In other words the amino specificity is the function of the nucleotide specificity.

Important steps in protein synthesis are mentioned briefly. The genetic information contained in the DNA for the synthesis of a protein is copied out in the form of RNA, which carries to the cytoplasm is called messenger RNA. Several molecules of the messenger RNA take part in carrying all the information needed to construct certain portion of protein. The messenger RNA arranges itself in proper position at the ribosome, the site of protein synthesis in cytoplasm. Also present in the cytoplasm are different amino acids 20 different transfer RNA corresponding to each amino acid. A particular transfer RNA will form a complex with a particular amino acid. Before this complex is formed, the amino acid is activated by (adenosine triphosphate) the energy-rich compound. This activated complex moves to the ribosome and the amino acid is thus inserted in the protein chain at the proper place with the help of the messenger RNA. Likewise, other, amino acids that are needed in the synthesis of the protein are brought to the ribosome and the desired molecule of protein obtained.

The protein thus synthesized under the direction of DNA produce a given phenotypic effect. It should be clearly understood that a phenotypic expression is the direct outcome of the DNA activity and any change in the protein structure shall have no effect on the DNA; that is why acquired characters cannot be inherited.

PROBLEMS

1. How would you define a gene on a molecular basis and in classical sense?
2. What is DNA and its chemical composition? How does it form RNA?
3. How would you explain the relationship between DNA and protein synthesis?
4. Could you explain in terms of DNA-protein relationship acquired characters are not inherited?

Chromosomal Aberrations

Chromosomal contents (number and structure) of an individual, a crop variety or a species remain constant from generation to generation. The constancy of the chromosomal contents, ensures the constancy of the individual with all its characteristics. If a viable change occurs in the number or structure of the chromosomes of an individual, the properties of the individuals change accordingly and the changed properties are also expressed in the off-springs of the individual faithfully over the generations.

Such a change can occur in the following three ways:

1. Recombination of the existing chromosomal material through crosses.
2. Change in the number of chromosomes.
3. Change in the chemical or physical composition chromosomes.

1. Change by recombination

Much has already been said in the previous sections about how individuals change genetically through recombination of chromosomes and, for that matter, of the genes carried on them. The genetic changes caused by this process are expressed phenotypically in the next generation, the changed phenotypes can further change by going through further combinations

2. Change in chromosome number

There is a basic chromosome number within a genus or a species and additions to, or subtraction from, that basic number through evolution produce new individuals or species with modified character. This phenomenon of change in chromosome number is termed polyploidy. If the changed number is an exact multiple of the basic chromosome number, the polyploidy is called Euploidy and if it is not an exact multiple, the polyploidy is called aneuploidy. Take, for instance, wheat. The basic chromosome number in wheat genus (*Triticum*) is 7, which may be designated as X. Its exact multiple numbers will be 14(2x), 21(3x), 28(4x), 35(5x), and 42(6x). The species with 14 chromosomes will be diploid; with 21 chromosomes triploid; with 28 chromosomes tetraploid; with 35 chromosomes, pentaploid; with 42 chromosomes hexaploid,

while the basic number of 7 constitutes a monoploid. Our common bread wheat grown in this region is *Triticum aestivum* which is a hexaploid.

It may be noticed that in euploidy, the basic set of chromosomes is added or taken out, as a whole.

Euploid type of polyploidy is further divided into two classes:

i) Allopolyploidy. Different sets of chromosomes are from different sources through crossing followed by doubling. Species hybrids are usually sterile, and fertility may be restored if the chromosome number of such hybrids is doubled. Such chromosome doubling provides suitable mates (homologues) to synapse during cell division. Take the example of *Raphanobrassica*, a polyploid developed through the crossing of two different species, cabbage (AA) and radish (BB). The hybrid (AB) between these species was found to be sterile. On doubling its chromosome number, it produced viable gametes, because each duplicated genome provided proper homologues for due synapses. It is illustrated as under:

Parents:	Cabbage	X	Radish
	AA (2n 18)		BB (2n 18)
Gametes	A, (n, 9)		B, (n, 9)
F1	AB (2n, 18)		
Chromosome doubling AA BB (2n, 36) Raphanobrassica			

Our common bread wheat is an allopolyploid, because its three different chromosome sets (genomes) have been contributed by three different species.

(ii) Autopolyploidy. All the sets of the chromosomes (genomes) present in a polyploid belong to the same individual, and no prior crossing is involved. In other words, one genome is repeated many times. Autopolyploids are usually sterile, because more than one homologue are present during meiosis and irregular pairing of chromosomes results in the production of unbalanced gametes which are usually sterile. The autopolyploids may originate as a result of (1) doubling of the chromosome number in the somatic tissue, that takes part in gamete formation (2) failure of reductional division resulting in reduced gametes.

Triploid. These have three sets of chromosomes and may be produced when an unreduced ($2n$) gamete is fertilized by a normal (n) gamete. They may also arise from a tetraploid crossed with a diploid. The triploids are usually unstable and sterile.

Tetraploid. These have four sets of chromosomes. Frequently, their origin is through doubling of diploid which may be allopolyploid or autopolyploid. Irregularity at the time of mitosis causes doubling of chromosomes. Any agency that does not allow the spindles to develop properly in cell division will obstruct the division of duplicated chromosomes which may then be enclosed in the same nuclear membrane producing a tetraploid.

In aneuploidy, the chromosome number is modified by adding or subtracting only one, two, three, etc. chromosomes and not the entire set. If from an amphidiploid ($2n$), one chromosome is missing, it is called monosomic ($2n-1$), and similarly with the addition of one extra chromosome resembling any other existing chromosomes, the individual is called trisomic ($2n + 1$); ($2n + 2$) will be tetrasomic and ($2n-2$) nullisomics

The aneuploids have a practical significance in identifying genes on different chromosome. The trisomic individuals (one chromosome is repeated three times), produce different phenotypes than the normal and so do the monosomes. The deficient or the additional chromosome can be identified cytologically and the changed phenotypic effect may be attributed to genes present on those chromosomes. It is rather difficult to establish monosomic lines in organisms which are not polyploids. Nullisomics are also usually inviable.

3. Changes in chromosome structure

Physical and chemical changes in the chromosomes of an individual can occur in several ways. We may first discuss physical ones.

(1) Deficiency. When chromosome somehow loses a portion of itself, deficiency occurs. A break may occur at the end of the chromosome, which will produce a terminal deficiency. Two breaks may delete a middle portion of the chromosome, which will be an intercalary deficiency. When a normal homologue pairs with its deficient mate, it shows a buckling effect as follows:

A B C D E F

Normal Chromosome

A B C D

Terminal deficiency

A B E F

Intercalary deficiency

ii) Duplication. A chromosome gains a portion of a homologous chromosome and is thus doubled for that chromosome material. The duplication provides a means to study the effect of some genes in single, double or more doses.

A B C D E FA B C D E FA B C D E F E F

Duplicated chromosome

Break occurs in one chromosome

(iii) Translocation. When two non-homologous chromosomes exchange their corresponding portions, they are said to have undergone translocation; the translocation will be reciprocal. In the non-reciprocal translocation corresponding portions are not exchanged. Translocation usually do not involve any Loss or addition of the chromosome material but only represent the engineering of new chromosomes and alter the linkage groups, besides inducing partial. Sterility in the gametes.

A B C DW X Y ZNon-homologous
chromosomesA B C DW X Y ZBreak occurs in
corresponding portionA B Y ZW X C DReciprocal
translocationA B C DW X Y ZBreak occurs in one
chromosomeA B C D Z

Non-reciprocal translocation

(iv) Inversion. Certain segment of a chromosome may break at two places and if the breaking points are close to each other due to the formation of a loop, the broken ends may reunite on the wrong ends and produce an altered gene order.

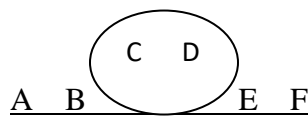
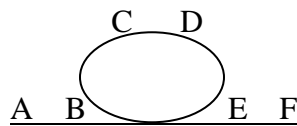
A B C D E F

Normal chromosome

Formation
of loopA B D C E F

Inverted chromosome

When the inverted chromosome pairs with a normal chromosome, forms a loop, as shown in the following diagram:

A B C D E FA B C D E FInversion has occurred
in one homologuePairing between inverted and normal
homologous pair of chromosome

Chemical change in the chromosome

Structural change does not obviously alter its physical shape and form, but definitely imparts new qualities to the genetic matter contained therein. A gene which is located on the chromosome, is a chemical entity and its three important components are (i) Nitrogenous bases, which are of 4 kinds (ii) Pentose sugar and (iii) Phosphoric acid. These three chemicals combine to form 4 nucleotides according to the 4 nitrogenous bases. In the chromosome, these nucleotides are arranged in different orders giving rise to various genes. If this arrangement is disturbed by changing the order through some means, the gene is changed and a new phenotype results. Such changes occur very rarely in nature, but can be induced by artificial means and this process is called mutation.

Mutation is permanent in nature but a gene can back-mutate restoring the original genotype and the phenotype.

Mutation can be induced by the following agencies:

- (i) Ultraviolet light
- (ii) Ionizing radiations (X-ray, Gamma rays) etc.
- (iii) Chemical substance, i.e. Colchicines, acenaphthene, Sodium azide, mustard gas, E.M.S. etc.
- (iv) Temperature

Agents which cause mutation are called mutagens and the new forms produced are mutants. The process of inducing mutation is called mutagenesis. The use of mutants in plant breeding may be called Mutation breeding.

PROBLEMS

1. Name different forms of polyploidy and discuss each in brief, giving suitable examples.
2. Which form of ploidy (allo or auto) do you think is more common in plants and why?
3. A & B represent the two basic genomes. What type of polyploidy do the following individuals represent?
 - (i) A A A A
 - (ii) A A B B
 - (iii) A A B B B B
 - (iv) A A B
4. The basic chromosome number of a species is 5. Calculate the number of chromosomes of the individuals belonging to diploid, triploid, tetraploid, pentaploid and hexaploid series.
5. How many types of physical changes can occur in the chromosome structure. Why are inviable gametes produced by most of such individuals as have undergone such changes?
6. Discuss various mutagens. How does a point mutation occur?

Application of Genetics in Crop Improvement

Philosophy

Throughout the discussion of different genetic phenomena in the previous chapters, it was quite apparent that an essential feature of biological populations is the variability expressed in growth and development and of their characteristics. As a matter of fact has been stated above, the science of genetics attempts to explain the basis of the variability and similarity in the expression of a character. One may look around and be struck by an amazing degree of diversity present in living beings and yet they are alike in several ways. Consider, for instance, human skin colour. Some people are white, some are dark, and a great many possess varying shades of colour between the two extremes, dark and white. Similarly, people differ in body weight, height, mental capacity, eye color, etc.

In plants, too, there may be seen a wealth of variability. Within a crop species some plants are taller, some produce more tillers, some are better yielders and some are better resistant to various diseases and other adversities. Unless an effort is made to produce and maintain a perfectly pure crop, it may ordinarily be a mixture of variable plants. You will realize that a self-pollinated crop is relatively less variable than a cross-pollinated crop.

The causes of variability are to be found in the genetic structure of a population and the environments under which the various members of the population are living. That is to say, the causes of variability are (i) genetic and (ii) environmental. Genetic causes may largely consist of mutation and recombination of the genetic material (genes). A mutation, represents a change in the chromosome number or structure, and may occur spontaneously or be induced artificially by different means, e.g. irradiation and chemicals. Recombination does not, however, involve a structural or a numerical change but is simply a recombining of the genetic material through independent assortment following hybridization.

All such changes are permanent and therefore, heritable. On the other hand, changes induced in the phenotypic expression of a character by variation in the environments, may at best last during the lifetime of the individual and are not transmissible to the next generations. Breeders are naturally not interested in non-heritable changes, because they seek such changes

which have their basis in the genetic material and stay permanently. Creation of genetic changes, their identification and ingenious manipulation to evolve new crop varieties, constitute the field of plant breeding.

Methods of Plant Breeding

As it has been pointed out before, the basis of crop improvement is the existence of genetic variability in plant material. A breeder exploits it in several different ways to create new crop varieties which are capable of out-performing their ancestors in the expression of various economic characters, e.g. crop yield; quality, resistance to diseases, insect pests etc. The Success of a breeder's effort has a direct relationship with the amount of genetic variability present for a particular character. The greater variability, the easier the method available for breeding and more rapid its progress. The choice of a breeding method is therefore, dictated, on the one hand by the magnitude of genetic variability and, on the other hand, by the genetic system of the plant. Some of the important fundamental methods of plant breeding are discussed below briefly./

A. Self -pollinated crops

1. Single Plant Selection

In self-pollinated crops, when it can be used, single-plant selection is the simplest of all the methods of crop improvement. Single plants, having the desired characters, are selected and their progenies raised separately to test the performance of single plants against a standard variety. Plants found performing better are selected, their seed multiplied and after appropriate yield trials, the best single plants recognized as new varieties. It may sometime be considered profitable to bulk all of them to be used as a multilineal variety. But on the other hand, when a variety descends from a single plant, it is called a pure line and the method of its production is called pure-line method of selection. The pure-line theory was developed by Johanson, a Danish breeder. According to it, desirable plants are selected, their progeny raised the following year, only Superior progenies harvested and their progenies are then tested for yield. The best ones are released.

2. Mass selection

This method may be practiced both in self and cross-pollinated crops. A number of plants, having desirable characters, are selected from un-improved crop material; their seed is bulked without testing the performance of their progeny and thus all the selections are mass-planted for early evaluation or for further mass selection. The objective is to finally make a collection of as many plants as may possess the desired characters and to use them together as a new variety. It may be pointed out that in cross-pollinated crops, mass selection of plants is required to sustain a maximum level of reproduction and vigor of the crop.

3. Hybridization

If the plant material intended to be improved for certain characters does not show enough variability or if the desired combinations of characters are not available, then one may resort to hybridization of the individual plants to produce new combinations on which individual or mass selection may effectively operate. Hybridization is an effective means of recombining genetic material. Suppose there are some good characters possessed by one variety and some other good characters possessed by another variety. The purpose of breeding may be to get together the good characters of these two varieties in one single variety. This can be achieved by making a cross between the two varieties and then selecting from the segregating progeny of the cross in F_2 or a later generation. The method of selection may be single-plant or bulk selection, depending upon the type of material and the breeding objective. By suitable genetic experiments it may first be determined as to how many gene pairs control the expression of the characters under study. Once obtained, this information should be of considerable help to the breeder in regulating the size of segregating populations so that he must be able to pick up the desired segregates.

(1) Pedigree Method

According to this method, superior segregates are selected in successive segregating generations, maintaining a complete record of the parent-progeny performance. The selection is made in the F_2 generation and the progenies of each selection are planted in succeeding generations until the desired level of genetic purity is reached. Then follow the testing. During yield tests observations are made on such characters as height, straw stiffness, winter hardiness,

maturity, disease resistance, quality, etc. The strains scoring highest among all are finally selected as varieties.

(ii) Bulk Method

This method differs from the pedigree method in that the selection is not made in F_2 but the material is grown in a bulk plot from the F_2 to about F_6 generations, followed by head selection in F_6 . Thus selection is delayed until F_6 generation. By that time sizeable homozygosity is reached. This material is space-planted and selection is made of the desired plants from the population. These selections treated as families are evaluated in the same way as in the pedigree method, and best progenies are released as varieties.

(iii) Backcross Method

Suppose, again there is a commercial crop variety which has all the good characters except its susceptibility to a certain hazard, as for instance, a fungal or bacterial disease. The breeder would like to add disease resistance to this otherwise a very fine variety, maintaining all its good attributes. The way open to him to achieve this object is to find another variety which carries disease resistance and not necessarily other desired characters and to hybridize it with the commercial variety. The hybrid between the two may develop resistance if disease resistance is a dominant character, otherwise the hybrid may either be selfed or back-crossed to the resistant parent followed by selection of the recessive resistant plants. The resistant plants are then repeatedly back-crossed to the commercial variety and selfed as the case may be for a number of generations and selections made until all the original good characters of the commercial variety are restored to it along with the ability to resist disease. The last progeny, if heterozygous for disease resistance, should be selfed to obtain homozygous resistant plants.

B. Cross-Pollinated Crops

As you might have noted, the breeding methods employed in the self-pollinated crops aim at evolving varieties that are homozygous or near homozygous. The idea is to assemble the desired genes in a variety, in a homozygous condition. On the other hand, in cross-pollinated crops, the ultimate aim of breeding is to develop a variety in a heterozygous condition.

The cross-pollinated crops as they have evolved, maintain their natural vigour, only when heterozygous. If such a cross-pollinated population is selfed to make it homozygous, it will suffer depression in yield, which is called inbreeding depression. Maize, an important cereal belongs to the cross-pollinated group, and suffers inbreeding depression on selfing. The philosophy in maize breeding therefore is to maintain heterozygosity to keep up hybrid vigour. So all the breeding methods used in maize aim at the evolution of hybrid (heterozygous) populations. For instance a new maize variety may be a single cross, a double cross, a three-way cross or a synthetic, evolved by combining suitable inbred lines.

A single cross involves two inbred lines, i.e., (A x B) while a double cross is a cross between two single crosses i.e., (A x B) (C x D) and is not so uniform as a single cross. A three-way cross is obtained by crossing an inbred line with a single cross and a synthetic variety is evolved by crossing several tested inbred lines.

PROBLEMS

1. Define plant breeding and explain its genetic basis.
2. Name some of the self-pollinated and cross-pollinated crops, commonly cultivated in the country. Explain that the breeding philosophy for both is basically the same.
3. Describe briefly the salient features of important breeding methods like introduction, single plant selection, pure line selection and mass selection.
4. Explain how pedigree method differs from the bulk method of breeding.
5. What is the fundamental difference between wheat breeding, and maize breeding.

Glossary of Some Genetic Terms

1. *Acquired characters*. A change or modification impressed on an organism by the environmental influences during the course of development e.g., development of muscles by exercise, acquiring skills by practice.
2. *Allelomorph or Allele*. Alternative form of a gene: a member of a pair of genes occurring on the same locus in homologous chromosome and for this reason inherited in alternative pairs entering into different gametes. A gene can exist in several alternative forms, each of which is an allele. For instance, a gene controlling expressions of colour can have a number of alleles, each one determining a different shade of that colour.
3. *Backcross*. The cross of the F₁ with one of its parents. The cross of the hybrid with the recessive parent is called test cross.
4. *Bivalent*. A pair of synapsed or paired homologous chromosome during meiosis.
5. *Centromere*. Spindle fibers attachment region on the chromosome some, which controls chromosomal movements during cell division.
6. *Character*. Characteristic or Trait. The Mendelian characters in genetics means the end product of development process which is under the control of the interaction of genes and the environments
7. *Chromatid*. The longitudinal half of the replicated chromosome; appears during prophase and later becomes daughter chromosomes on the completion of the cell division.
8. *Chromatin*. The part of the nucleus, which forms the chromosomal net-work; is readily stained by specific dyes that is why the name “chromatin”.
9. *Chromosomes*. Microscopically small, threadlike, darkly staining bodies; visible in the nucleus at the time of the cell division; the number in any species constant; carry genes arranged in a linear order.
10. *Cistron*. According to the modern concept, a unit of function.

11. *Determiner* (Gene). An element carried on the chromosome which controls the development and expression of Mendelian character.
12. *Dihybrid*. An individual which is heterozygous with respect to two pairs of genes (AaBb).
13. *DNA*. Deoxyribonucleic acid, the chemical basis of heredity.
14. *Diploid*. Organisms having two sets of chromosomes. The body cells of higher plants and animals are diploid for their chromosome number (2n), whereas their gametes are haploid (n).
15. *Dominant and Recessive Alleles*. In a diploid individual, each character is represented by a pair of alleles. The two alleles may be alike or different. In the latter case, one of the two different alleles may be powerful enough to hide the effect of the other. In that case, the former is called the dominant and the weaker one, the recessive allele.
16. *Dyad*. A replicated chromosome during meiotic division composed of two chromatids, also applied to daughter nuclei formed as a result of meiotic division.
17. *F₁*. The first filial generation resulting from a cross. If the parents entering the cross are homozygous, the F₁ individual will also be genetically uniform.
18. *F₂*. The second filial generation produced by crossing or self pollinating the F₁ individuals; the F₂ generation is the segregating generation in which the number of different segregates obtainable depends upon the number of heterozygous gene pairs involved and also the size of the population.
19. *Gamete*. A mature germ cell with haploid chromosome number which produces the zygote with diploid chromosome number on fertilization.
20. *Gametophyte*. The particular phase in the life-cycle of plants which produces gametes; it carries (n) chromosome number.
21. *Genotype*. The sum-total of the genes affecting the expression of a character.
22. *Gene*. The hereditary unit carried on the chromosome; it controls the development of a character.