**FOREST TREE IMPROVEMENT   (2+1)**

**Class:- M.Sc (Forestry) and Ph.D (Forestry) Previous year**

**Course Teacher :- Dr Kundan Singh**

## Theory

             History of genetics – Scope and significance of modern genetics – Mendel's laws of inheritance – Deviation from Mendel's laws – Multiple alleles – Pseudolalels – Pleiotropy – Lethal genes – Multiple factor inheritance – Linkage – Crossing over – Methods of estimating linkage – Sex linkage – Cytoplasmic inheritance – Mitochondrial genetics – Chloroplast genetics – Sterility and incompatibility mechanisms – Basis of sterility – Molecular nature of gene – Genetic codes – Protein synthesis - Cells and cell organelles – Structure of metaphase – Pachytene chromosomes – Karyotype and ideograms – Banding techniques.

             Application of genetics in tree improvement chromosome number and karyotype of gymnosperms, angiosperms, polyploidy in temperate and tropical forest tree species – Aneuploidy of conifers – Induction of chromosome variation in tree species cytogenetics of acacias, poplar, casuarina and eucalyptus.

**Practical**

             Probability and chi-square test – Working out problems relating to mono and dihybrid ratios – Modified dihybrid ratios – Epistasis – Supplementary, complementary, duplicate dominance, duplicate recessive and inhibitory interactions – Multiple alleles – Linkage, sex linked inheritance – Multiple factor inheritance – Description of mitotic and meiotic chromosomes from slides.

**Lecture Schedule**

 1.   History of genetic – Development on cell theory

2.    Scope and significance of modern genetics

3.    Pre-, Post-Mendelian and Mendelian periods

4.    Mendel's law of inheritance

5.    Chromosome theory of inheritance and parallelism

6.    Interaction of factors – Epistasis – Supplementary and inhibitory epistasis

7.    Duplicate, recessive and complementary and dominant epistasis

8.    Multiple alleles

9.    Deviation from Mendels laws – Inhibitory

10.  Duplicate dominant multiple alleles

11.  Pseudoalleles – Pleiotropy, modifying factors

12.  Expressively – Lethal gees

13.  Multiple factor inheritance – Multiple factor hypothesis

14.  Nature and effect of crossing over repulsion coupling phase, linkage groups

15.  Nature and effect of crossing over and methods of estimating linkage

16.  Genetic map, interference coefficient of coincidence

17.  Mid Semester Examination

18.  Sex linkage – Sex influenced characters – Sex determination

19.  Cytoplasmic inheritance – Mitochondrial genetics

20.  Chloroplast genetics

21.   Sterility and incompatibility mechanisms

22.   Genetic basis of sterility

23.   Nucleic acid – Chemical structure - Chemistry of chromosomes

24.   Replication – Transcription

25.   Molecular structure of gene-genetic code

26.   Protein synthesis and gene code concept

27.   Application of genetics in tree improvement

28.   Cell and cell organelles, structure of metaphase

29.   Pachytene chromosome – Karyotype and idiogram

30.   Structure of chromosomes – Banding techniques

31.   Extra chromosomal inheritance –Plastids – Mitochondria

32.   Cytogenetics of Eucalyptus, Casuarina, Acacia and Poplars

33.   Chromosome number and karyotype of gymnosperms and angiosperms

34.   Polyploidy and aneuploidy of conifer – Cytogenetics of hardwoods – Induction of chromosome variation in tree species.

### Practical Schedule

 1.     Probability and chi-square test

2.     Working out problems in inheritance – Monohybrid ratios

3.     Dihybrid ratios

4.     Trihybrid/Poly hybrid crosses

5.     Interaction of factors/genetics – Epistasis

6.     Supplementary interaction

7.     Complementary interaction

8.     Duplicate interaction

9.     Inhibitory dominance

10.   Duplicate dominance

11.   Multiple alleles

12.   Multiple factor inheritance

13.   Linkage

14.   Sex linked inheritance

15.   Preparation of fixatives and stains for cytological studies

16.   Mitotic and meiotic stages - Pollen morphology and pollen germination

17.   Final Practical Examination

**Assignment**

Problem solving exercise for each practical class relevant to tree genetics and cytogenetics

**References**

 1.    Agarwal, C. and Varma., 1978.  Genetics. Oxford IBH publication.  New Delhi. P 534.

2.     Daniel Sundara Raj, D., G. Thulasidas and M. Stephen Durairaj, 1997.  Introduction to Cytogenetics and Plant Breeding.  Popular Book, Depot, Chennai.  Pp 589.

3.      Eaver, R.F. and Hedrick, P.W., 1989.  Genetics.  WCB Publishers.  USA. P 649.

4.     Fins, L., Friedman, S.T., Brotschol, J.V., 1992.  Handbook of Quantitative Forest Genetics.  Kluwer Academic Dordrach, London.  Pp 403.

5.     Mandal, A.K. and Gibson, 1998.  Forest Genetics and Tree Breeding.  Pub. Kalyani, Dehra Dun.

6.      Namkoong, G., H.C. Kangand, J.S. Brouard, 1988.  Tree Breeding Principles and Strategies.  Springer – Verlag, XI New York.  P 384.

7.      Singh, B.D., 1990.  Fundamentals of Genetics.  Kalyani  Publishers, Ludiana.  Pp 859.

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10.   Zobel, B. and Talbert. R., 1984.  Applied Tree Improvement.  Pp 340.

**Websites**

 1.             courses.forestry.ubc.ce/genetics

2.             www.cabi.org/forestry

3.             www.café.edn

4.             www.kancdsoftware.ccw/genetics

**1. HISTORY OF GENETICS**

**Genetics**

Genetics is the science that deals with the principles that explains the similarity of the parents and their progeny and differences among the individual of the similar species.

It is the study of heredity and variation. Heredity transmission of characters from parents to offsprings.

**Cytogenetics**

The study of various aspects of chromosomes and their effects on the development of characters of organism.

**History:**

**Early concept:**

Among the biological sciences the science of genetics orginated 1900 with the rediscovery of Mendelian Principles. Though pre-historic plants and animal breeders employed hybridisations and selection they were not aware of principles of genetics.

Early works forwarded various speculation and theories explain the phenomenon of heridity. The ideas of early workers can be grouped into the following headings.

1. **Vapour and fluid theory:**

Early Greek Philosapers thought hereditary information of the parents exsist in the form of vapour and fluid. Pythagorus 500 B.C. proposed that the moist vapour from dry nurves and other parts of the body organs from the nerves form an embryo in the uterus of the female.

1. **Magnetic power (Harvey (1578 - 1657)**

He proposed the uterus had some magnetic power to consume an embryo.

1. **Pre-formation theory**

With discovery of Sperms and egg by 17th century biologist pre-formation theory was proposed. According to this new individuals of completely pre-formed in miniature size in gamates.

1. **Epigenetic theory ( Wolf) 1738**

He proposed that each egg had granules (01) globules which gradually develops into various organs of the embryo.

1. **Particulate - Maupertius (1698 - 1759)**

**Maupertius** proposed that both the parents produce the gametes. Egg and sperm united to produce embryo. Each organ of embryo is made of two parts of embryo one is come from male () another one female ().

1. **Theory of Lamarck (1744 - 1829)**

**Lamarck** proposed that environmental changes cause modification in organism and that such modification transmitted to subsequent generation. That is the character acquired in one generation to another generation. Acquire characters are inheritable.

e.g. Giraffie live in the interior part of the Africa. So it feed on the leaves of tall tree and to strain itself continuously to reach them. Such exercise caused the legs and necks to grow in length. The increased length was inherited by the progeny touched the legs and neck and over generation it will continue. This has evolved the present day 6 metre giraffie.

1. **Theory of pangenesis - Charles Darwin (1809 - 1882)**

**Darwin** proposed theory of natural selection. Many individuals of each species and there is always existence. If hereditary differences occurs with in a species of plants natural selection allowed only the fittest individuals of the species to survive and eliminating others. This is known as survival of fittest due to natural selection.

In 1868 Darwin published origin of species. He also proposed theory of Pangenesis . he called it hereditary particles. Germules that are produced every part of the body during the life time. The germules from all the organs are transported to the gonads. The gonads distributed gametes if male () gametes unit during fertilisation, germmules from the migrate in respective organ and determine the development of organ. Organ is modified some way. The germmules of the organ also modified accordingly which is transmitted to the offspring through gamets.

**Germplasm (Weismann) (1814 - 1914)**

Weismanns suggested the reduction in chromosome number takes places during formation of eggs / sperm and the original number resorted when eggs sperm fused. according to him acquired character not inherited. To prove this in mice to cut off tails 22 generation at last 1592. All of them having normal fail end. If the acquired characters are inherited the tails condition of mice produced by cuttings of their tails should be transmitted to offsprings. If the theory of pangenesis was correct the germmules for tail would be absent in the gamets of the tail less mice and their progency would be tails less.

Weismann proposed germplasm theory of heredity particles called IDS (Genes) situated on idants (chromosome) constitute the germplasm. The germplasm transferred to parents to offsprings and give rise body or some. The germplasm is independent of the body. Whatever happens to the body has not effect on the germplasm. This was prooved by ovary transplantation in guinea pigs. When albino guinea pigs are mated with alblono only albino progenies are produced.

Castle & Phillips (1909) removed the ovary of the albino guinea pigs and grafted in their place the ovary of guinea pig. The albino animal with the ovary of the black one when mated with the albino on the offsprings are black only. This proved the germplasm is not affected by body.

**Robert Hook (1665)**

He described the cell as empty vessel. He introduced term cell.

**Cameraious:**

He proved pollen is important for fertilizations. He is the first man to produce first artificial hybrid plant.

**Koelreutes**

He showed that F1 might resemble either male (or) female parents (or) combination of both.

Hereditary contribution of the two parents to their offspring was equal.

**Knight**

He obtained the dominant forms in F1 and segregation of various characters in F2.

**Gaertner**

F1 are uniform and their F2 produced considerable variation.

**Naudin**

Hybrids races and species of plants are often luxurient than either of the parents.

**Robert Brown:**

He described the cell nucleus in the flowering plants.

He observed random thermal motion of small particles known as Brownian movement.

**Schleiden and Schwann:**

They discovered the formation of nucleus in the cell and formulated cell theory, which says the cell is the smallest building element of a multicellar organism.

2. each cell has a specific work to complete.

3. the cell can only the produced from another by cell division.

**Strasberger**:

He described fertilization in Angio-sperms.

**Van Beneden - Mieosis:**

He showed number of chromosomes in the gametes, is half of the number of body cells. In fertilization the chromosome contribution of eggs and sperms to the zygote are numerically equal.

**Flemming - Mitosis**:

1. He proposed mitosis in cell.
2. He showed the chromosome spliited during nuclear division and the formation of daughter nuclei.
3. he also applied the name chromatin which is to the stainable position of the nucleus.

**3. PRE-, POST-MENDELIAN AND MENDELIAN PERIODS**

There are only a very few sciences for which a definite date of origin can be given. Genetics is one of those few, for although several investigators tried to tackle the problems of heredity since remote antiquity, no one succeeded until 1865 when Gregor Johann Mendel presented a paper embodying the results of his experiments on hybridization in peas. And in that paper, which was left neglected for thirty-five years and rediscovered in 1900, were contained the fundamentals of genetics.

The history of genetics can therefore be reviewed under three periods(1) Pre-Mendelian, (2) Mendelian, and (3) Post-Mendelian.

**PRE-MENDELIAN PERIOD**

Some of the scientists prior to Mendel tried to account for the differences existing between individuals and suggested theories to explain their inheritance. The most important theories are the following:

**Theory of Lamarck**

The French biologist Jean Baptiste de Lamarck (1744-1829) proposed the theory that environmental changes cause modifications in organisms and that such modifications are transmitted to subsequent generations. He believed that environment acts directly on plants and indirectly on higher animals.

Lamarck said that changes in environmental conditions create new needs in animals. Conscious efforts of the animals to adapt to the environment involves the use of certain organs, thereby causing them to become large, strong and well-developed. Other organs are not used and so become smaller, weaker and less well-developed. Such bodily changes are called acquired characters since an animal achieves them by its own exertions to adapt to the environment. Acquired characters, according to Lamarck, are then passed on to the offspring of the organism that acquired them, and new species originate by accumulation of these modifications.

The giraffe dwells in the interior arid parts of Africa where there is not much herbage. According to Lamarck, the giraffe was obliged to feed on the leaves of tall trees and to strain itself continuously to reach them. Such exercise caused the necks and legs to grow in length. The increased length was inherited by the progeny, which, in turn, stretched their necks and legs and transmitted their increased length to their own offspring. Thus has evolved the present day six-metre high giraffe.

Detailed studies have failed to show that acquired characters are inherited. Most biologists have therefore abandoned the theory of inheritance of acquired characters, otherwise known as Lamarckism.

**Darwin's Theory**

In 1858, Charles Darwin (1809-1881) and Wallace independently proposed the 'Theory of Natural Selection'. According to this theory, many more individuals of each species are born than can possibly survive and consequently there is always a struggle for existence. If hereditary differences occur within the wild species of plants, nature will eliminate some and select others.

Over-production, struggle for existence, hereditary variations and survival of the fittest are thus the important principles of the theory of natural selection.

Ten years after the publication of the Origin of Species (1859), Darwin adopted the doctrine of the inheritance of acquired characters but he proposed a new theory of how it happened. He modified the views of Spencer and proposed the 'Hypothesis of Pangenesis' (1868).

Darwin assumed that hereditary particles termed pangenes of gemmules, are produced by every part of the body during the life time of an organism and that, these assume the characters of the various parts of the body from which they were derived, together with whatever modifications the latter may have acquired. Eventually all the pangenes accumulate to form the germ cells which give rise to the new individual, thus ensuring the development of the parental characters and inheritance of acquired characters.

**Weismann's Germplasm Theory**

Weismann(1834-1914), a German zoologist, suggested in 1887 that a reduction in chromosome number took place during the formation of the egg and the sperm, and that the original number was restored when the egg and the sperm fused. In 1892, he suggested that the maternal and paternal chromosomes separated during the reduction division and that they recombined when the gametes united.

According to Weismann's Germplasm Theroy of Heredity', the hereditary particles called ids (what we now call as genes) situated on idants (what we now call chromosomes) constituted the germplasm. The germplasm is handed down from parent to offspring and it gives rise to the body or soma (somatoplasm) whose character it determines. The germplasm is independent of the body and whatever happens to this body has no effect on the germplasm which is contained within it.

According to Weismann, acquired characters cannot therefore be inherited. To prove this he cut off the tails of mice for twenty-two generations and found that the progeny consisting of 1,592 individuals had tails of normal length.

The independence of the germplam from the somatoplasm was shown by the ovary transplantation experiment in guinea pig. Ordinarily, when albino guinea pigs are mated with albinos, only albinos are produced. Castle and Phillips removed the ovaries of an albino guinea pig and grafted in their place the ovaries of a black guinea pig. The albino animal with the ovary of the black one was then mated with an albino. All the offspring were found to be black, thereby proving that the germplasm (i.e., the ovary from the black guinea pig) is not affected by the somatoplasm (i.e., the body of the albino).

**MENDELIAN PERIOD**

**Life of Mendel**

Gregor Johann Mendel was born in 1822 in Silesia, Austria. Mendel attended the elementary school of his village and subsequently the upper elementary school of a neighbouring town. Later he joined a high school at a town about 32 kilometres from his village. As his parents were not in a position to pay for the expenses connected with his studies , he earned by private teaching an amount sufficient for his living. In 1840, he completed his studies at the high school but due to financial difficulties he could not continue his studies for an year. Finally with the financial help from one of his sisters supplemented by the money earned by private teaching, he completed the two-year course at the Olmutz Philosophical Institute.

In 1843, Mendel entered the Augustinian monastery at Brunn because, in his own words, 'he felt compelled to enter a profession which freed him from the bitter worries about gaining a living'. He completed his theological studies in 1848 and was appointed as a substitute teacher in a high school. In 1850, he appeared for the examination for a teaching certificate but failed. The monastery sent him for studies at the University of Vienna from 1851 to 1853.

In 1854, he was appointed as a substitute teacher in the Brunn Modern School. In 1856, he appeared for the examination for the teaching certificate for the second time but failed again. Mendel's biographers believe that Mendel failed this time `because he showed unusual independence of thought and aroused the antipathy of the examiners by his defence of theories contrary to those of the examiners'. He continued as a substitute teacher for twelve more years.

Mendel began his experiments on hybridization in pea soon after his second failure in the examination for teachers in 1856 and continued them up to 1863. The experiments were carried on in the monastery gardens in a plot measuring 36 metres by 6 metres. He presented the results of his experiments before the Natural History Society of Brunn in February 1865. His paper was published in 1866 in the Annual Proceedings of the Society and distributed to libraries in Europe and America. No one who heard Mendel's paper or read it appreciated the importance of his work, for it lay neglected until 1900.

In 1868, Mendel was elected as the Albot (i.e., head of the monastery). He could not continue his hybridization experiments because his time and energy were consumed by a bitter dispute with the state over a law for taxation of the monasteries. He died in 1884, long before the world recognised the importance of his work.

**Rediscovery of Mendel's Work**

Mendel presented his paper in 1865 but the scientific world failed to recognise its importance for nearly thirty-five years. According to Iltis, Mendel's biographer, one of the reasons for the neglect by scientists was their intense interest in Drawin's Origin of Species published just a few years back. Moreover Mendel was not a biologist but only a monk. The union of botany with mathematics (statistics) must have also appeared strange to several readers of Mendel's paper. Above all, the importance of Mendel's findings was not recognised because contemporary scientists had very little knowledge of what went on inside the cell.

In the penultimate decade of the nineteenth century, rapid strides were made in Cytology. The German zoologist Oscar Hertwig (1876) showed that, in the sea urchin, fertilisation involved an union of the nuclei of one egg and one sperm. The German botanist Strasburger (1877) demonstrated the fusion of the egg nucleus and the sperm nucleus in the lily. In 1882, Flemming discovered mitosis (mitos=thread, because the chromosomes appear as threads in the early stages of cell division). He coined the name `chromatin' for the stainable portions of the nucleus. In 1888, Waldeyer introduced the name chromosome (chroma = colour; soma = body) for the deeply staining bodies in the cells. Weismann regarded the chromosomes as the material basis of heredity and developed the germplasm theory.

Biologists were stimulated to take up again the study of heredity and in the last decade of the nineteenth century, a number of them began an attack on the problem by crossing plants. Hugo de Vries in Holland, Carl Correns in Germany and Eric Tschermak in Austria independently began to hybridize plants and , unknown to one another , obtained results similar to those obtained by Mendel, though none of them had seen Mendel's paper before beginning his own experiments. Hugo de Vries published his results in March 1900, Carl Correns in April 1900, and Eric Tschermak in June 1900.

Genetics is thus one of the younger members of the family of biological sciences and can be considered to have been born during the year 1900.

**Reasons for Mendel's Success**

Mendel was able to discover the laws now bearing his name because of the following reasons:

He chose the garden pea, Pisum sativum as the material for his experiments on hybridization because he found that several pure-breeding varieties of peas were available, that they were normally self-fertilised, that the production of hybrids was not difficult and that the hybrids resulting from crossing two varieties were perfectly fertile.

Unlike, most of his predecessors, Mendel concentrated his attention on only one character at a time. Only when the behaviour of each trait e.g., form of seed-coat, was established, he began to study two traits together, e.g., form of seed-coat and colour of cotyledons.

Mendel kept accurate pedigree records of the members of successive generations.

He divided the offspring in each generation into classes according to their visible characteristics and recorded the exact numbers of each class.

Mendel proved that a single pollen grain fertilises a single egg of a plant. He obtained 18 well-developed seeds of Mirabilis jalapa (the four-o' clock plant) by fertilising 18 eggs with an equal number of pollen grains. He therefore concluded that heredity must depend on these individual germ cells. Since these gametes may be of different kinds, and since they unite at random, Mendel realized that large numbers of the progeny have to be studied before any conculsion on the principles of inheritance could be drawn. Mendel succeeded in discovering the laws of heredity mainly because he recognised that errors in sampling could be avoided by studying sufficiently large numbers of the progeny.

**Mendel's Principles of Heredity**

Mendel chose two plants differing in a pair of contrasting characters, eg., a plant with round seed-coat and another with wrinkled seed-coat, as parents for each of his experiments. He then confirmed that they bred true for several generations on self-fertilisation. He then crossed them and obtained the hybrid seeds. He found that the first generation (i.e., the first filial, or F1 and obtained as large a number as possible of a second generation, or F2 . He found that the F2 consisted of different kinds. He classified the F2 according to the characters exhibited and counted the number of each class.

In the light of present knowledge, Mendel's principles of heredity can be expressed as follows:

In sexual reproduction, the individual (or zygote) is formed by the fusion of two gametes, one (the egg) from the mother and the other (the sperm) from the father.

The hereditary particles are called genes (or factors) the female gamete contributes one of each kind of gene from the mother and the male gamete, one of each kind of gene from the father.

A zygote carries therefore every gene in duplicate. These genes however do not blend but preserve their individualities.

When this individual forms its own gametes, the maternal and paternal members of each pair of genes segregate and pass to different gametes.

Each gamete therefore has only one member of a pair of genes exiating in adult individuals.

The members of different maternal and paternal pairs of genes segregate independently and different gametes produced by the same individual may therefore contain different sets of genes.

These principles were summed up by Carl Correns, one of the rediscoveries of Mendel, in what are now known as Mendel's laws of heredity.

The first law is that hereditary factors (genes) are found in pairs in mature individuals. They do not blend but separate or segregate unchanged during the formation of gametes. The gametes therefore contain only one of a pair of factors responsible for each character. Even hybrids therefore gametes which are `pure'.

The second law is that the members of different pairs of factors responsible for different characters segregate and recombine independently in different gametes.

**De Vries' Mutation Theory**

Charles Darwin believed that evolution is due to natural selection of small hereditary variations occurring among individuals of any species. Bateson did not agree with Charles Darwin. He believed that evolution is due to large discontinuous variations. De Vries (1848-1935) introduced the term `mutation' for these large, discontinous changes in the genotype and proposed the `Mutation Theory', according to which sudden hereditary changes lead to evolution.

De Vries (1901) observed that the evening primrose Oenothera lamarckiana, a native of America, was growing wild in Holland. In a population of this weed, he observed some plants which differed in some characters from the typical Oenothera lamarckiana. Since it is a self -fertilised species, he felt that these variants have arisen suddenly rather than as hybrids. He transplanted them to his garden and studied them for several years. He observed that variations continued to arise spontaneously and that these variations were inherited. He called these drastic changed as mutations and maintained that mutations play an important role in the evolution of new species.

**POST-MENDELIAN PERIOD**

With the rediscovery of Mendel's work in 1900, plant and animal breeders, fascinated by Mendel's ratios, tried to find out the universality of the Mendelian theory in plants and animals. William Bateson (1902) was one among those who showed that several species of plants and animals follow the Mendelian laws of inheritance. And it was he who proposed the name `**Genetics**' for that branch of biology concerned with the elucidation of the phenomena of heredity and variation. Bateson coined the word `factor' for that which `determines' a character. He introduced the term `heterozygote' for a Mendelian hybrid and showed that the gametes of a hybrid are `pure'. Bateson and Punnett (1906) discovered that Mendelian ratios are modified by `interaction' of factors.

The Danish botanist Wilhelm Johannsen (1909) introduced the word `gene' for the materials present in the germ cells which determine the many properties of an organism. He also introduced the word `genotype' for the heredity of an individual plant of animal. And it was Johannsen who developed the concept of the `pure line'.

Populations were found to exhibit a continuous range of variations in respect of measurable characters and Francis Galton (1869) showed that this variation could be plotted on a graph as a `normal curve'. Nilsson Ehle (1908) published his multiple-factor hypothesis. Fisher (1918) developed the statistical methods which increased the efficiency of experiments. Kenneth Mather (1941) showed that the genetic components of continuous variation are genes whose individual effects are not easily distinguishable form environmental effects.

The American cytologist Wilson (1925) laid the foundations of the chromosome theory by stating that `chromatin' transmitted from generation to generations is responsible for heredity. Wilson's colleague Thomas Hunt Morgan (1910) found that some `factors' in the fruit fly Drosophila were not inherited at random, but in groups. Morgan and his associates, Muller, Sturtevant and Bridges, discovered that the number of such groups in Drosophila was four; corresponding to the number of chromosomes in the germ cells. Janssens (1909) discovered that, during meiosis, corresponding chromosomes from mother and father are held together at certain points called `chiasmata' and put forward the theory that exchange of segments between chromatids of maternal and paternal chromosomes took place at these points. All these discoveries led to an understanding of the chromosomes theory of heredity and the phenomena of linkage and crossing over.

The discovery by Painter (1939) of the usefulness in cytogenetic research of the gaint chromosomes in the salivary gland cell of Drosophila provided an additional avenue of approach to an understanding of the chromosome.

The discovery by Muller (1927) that X-rays could be used to induce mutations opened a new line of work.

The discovery by Blakeslee (1921) that colchicine could be used to double the number of chromosomes opened another new line of work.

The electron microscope with a resolving power of 0.001 micron (or 10 Angstroms) has opened up a new field to the cytologist. A number of cellular structures, that could not be seen with the unaided human eye which has a resolving power of only 100 microns or with the light microscope which has a resolving power of 0.2 micron, has been made visible by the electron microscope. The phase-contrast microscope is yet another new tool that has enabled the cytologist to study living or unstained cells.

Some knowledge of the molecular basis of heredity has been obtained by analysing the chromatin chemically. Though chromatin has been found to be composed of deoxyribose nucleic acid (DNA), ribose nucleic acid (RNA), and two forms of protins it has been shown that DNA is the hereditary material of the cell. Genes have the ability to reproduce their own specific duplicates by virtue of the specific forms of DNA possessed by them.

A knowledge of the structure of the DNA molecule is therefore of importance in understanding the physical and chemical basis of inheritance. Watson and Crick (1953) have proposed a double helical model of DNA consisting of four nitrogen bases, two purines (adenine and guanine) and two pyrimidines (thymine and cytosine) connected by hydrogen bonds and having deoxyribose sugar and phosphoric acid.

Kornberg and Ochoa shared the Nobel prize in 1959, and Wilkins, Watson and Crick shared the Nobel prize in 1962 for elucidation of the structure of the DNA.

The intimate relationship between the gene and the enzyme was established by Beadle and Tatum who, with Lederberg, received the Nobel prize in 1958.

The work of Horowitz, Benzer, Beadle, Tatum, Lederberg, Zinder and others on the bread mould Neurospora, the colon bacterium Escherichia coli, the typhoid bacterium Salmonella and bacteriophages has been responsible for the rapid progress made in recent years in the fields of physiological and biochemical genetics.

**4. MENDEL'S LAW OF INHERITANCE**

**1. Mendel's Law Of Independent Assortment**

As an individual possesses several characters, Mendel realised the need to find out how two or more pairs of contrasting characters would behave in relation to each other in their passage from generation to generation.

**Mendel's dihybrid cross**

As mentioned earlier, Mendel studied the inheritance of each of the seven pairs of contrasting characters selected by him in peas in separate crosses. For example, he crossed a variety of peas having round seeds with a variety having wrinkled seeds. The hybrid seeds from this cross were all round. He planted these hybrid round seeds and obtained an F2 generation consisting of round and wrinkled seeds in the proportion of 3:1. Similarly, he crossed a variety of peas having yellow cotyledons (the colour of which could be seen through the `transparent' or thin seed coats) with a variety having green cotyledons. The hybrid seeds resulting from this cross were all yellow. He planted these hybrid yellow seeds and obtained an F2 generation consisting of yellow and green seeds in the proportion of 3 yellow : 1 green.

Mendel then crossed a variety with round and yellow seeds with a variety with wrinkled and green seeds. The F1 hybrid seeds were all round and yellow. The plants raised therefrom yielded seeds of four sorts which frequently presented themselves in one pod. In all, 556 seeds were yielded by 15 plants and of these, there were:

|  |  |
| --- | --- |
| 315 | Round and yellow |
| 108 | Round and green |
| 101 | Wrinkled and yellow |
| 32 | Wrinkled and green |

Mendel observed that the segregation for form of seed and colour of cotyledons separately was in the ratio of 3 dominants : 1 recessive.

|  |  |  |  |
| --- | --- | --- | --- |
| Round seeds | (315 + 108) / 556 | = | 76.08% |
| Wrinkled seeds | (101 + 32) / 556 | = | 23.92% |
| Yellow seeds | (315 + 101) / 556 | = | 74.82% |
| Green seeds | (108 + 32) / 556 | = | 25.18% |

Three-fourths of the F2 seeds were, thus, round and one-fourth wrinkled. Three-fourths of the F2 seeds were yellow and one-fourth green.

Mendel observed that the segregation into three-fourths and one-fourth in each pair of contrasting characters was entirely independent of the similar segregation in the other pair.

|  |  |  |  |
| --- | --- | --- | --- |
| 423 | Round | Yellow  Green | 315  108 |
| 133 | Wrinkled | Yellow  Green | 101  32 |

Of the F2 seeds, three-fourths were round and of these, three-fourths in turn were yellow and one-fourth green. Of the F2 seeds, one-fourth were wrinkled and of these, three-fourths were yellow and one-fourth green.

In other words, three-fourths of three-fourths (i.e., 9/16) of the total number of the F2 seeds were round and yellow; one-fourth of three-fourths (i.e., 3/16) were round and green; three-fourths of one-fourth (i.e., 3/16) were wrinkled and yellow; and one-fourth of one-fourth (i.e., 1/16) were wrinkled and green.

Mendel concluded that the F2 of a cross involving two pairs of contrasting characters (i.e., a dihybrid cross) shows four kinds of individuals in the ratio of 9 : 3 : 3 : 1.

**Mendel's Second Law**

Mendel arrived at the following conclusion:

When an individual forms gametes, the members of a pair of alleles always segregate (i.e., separate) from each other but the members of different pairs of alleles assort independent of each other.

Mendel's Second Law of Inheritance or the Law of Independent Assortment can, therefore, be expressed as follows:

The segregation in one pair of alleles is independent of the segregation in any other pair of alleles.

In other words, when two or more independent characters are considered together, the factors responsible for them assort themselves freely and at random when gametes are formed.

**Dihybrid ratio**

Let the gene for round seeds be represented by **R** and the gene for yellow seeds by **Y**. The recessive alleles for wrinkled seeds and green seeds can therefore be represented by **r** and **y** respectively.

The round yellow-seeded parent is **RRYY** and forms gametes all of which are alike and have **RY.** The wrinkled green-seeded parent is **rryy** and forms gametes all of which are alike and have **ry**. the F1 hybrid seeds formed by the union of these gametes are therefore **RrYy** and all of them are round and yellow because **R** is dominant to **r** and **Y** is dominant to **y**.

When the plants raised from these hybrid seeds form ovules and pollen grains, the alleles **R** and **r** segregate so that two kinds of gametes, one with **R** and the other with **r** are produced in approximately equal numbers. Similarly the alleles **Y** and **y** segregate so that two kinds of gametes, one with **Y** and the other with **y** are produced in equal numbers. The separation in one pair of allelea is entirely unaffected by the separation in the other pair of alleles, but every gamete necessarily contains a gene for form of seed and a gene for colour of cotyledon. In any one gamete, it is purely a matter of chance whether the gene for round form is associated with that for yellow colour or with that for green colour. It is equally a matter of chance whether the gene for wrinkled form is associated with that for yellow colour or with that for green colour. Half of the gametes, thus, carry the gene for round and of these, half carry the gene for yellow and half carry the gene for green. The other half of the gametes carry the gene for wrinkled and of these, half carry the gene for yellow and half carry the gene for green. The F1 hybrids thus, produce four kinds of eggs, viz., **RY, rY** and **ry** in approximately equal numbers and four kinds of sperms, viz., **RY, Ry, rY** and **ry** in approximately equal numbers.

In the fertilisation, the gametes combine at random. All of the four kinds of male gametes have equal chances to fertilise any of the four kinds of female gametes. The F2 therefore consists of 16 possible zygotic combinations, as shown in the checkerboard, in approximately equal numbers.

It can be seen from the checkerboard that these zygotes are of nine genotypes. Their ratio is as follows:

|  |  |
| --- | --- |
| **Genotype** | **Ratio** |
| **RRYY** | 1 |
| **RRYy** | 2 |
| **RrYY** | 2 |
| **RrYy** | 4 |
| **Rryy** | 1 |
| **Rryy** | 2 |
| **RrYY** | 1 |
| **RrYy** | 2 |
| **Rryy** | 1 |

The genotypic ratio can be obtained without a checkerboard by combining the two monohybrid ratios, as follows:

The genotypic ratio in the F2 of a monohybrid **Rr** is 1 **RR** : 2 **Rr :** 1 **rr**. Similarly, the genotypic ratio in the F2 of the monohybrid **Yy**  is 1 **YY** : 2 **Yy** : 1 **yy**. Of the round seeds with genotype **RR**, one-fourth will have **YY,** one-half will have **Yy** and the remaining one-fourth will have **yy.** Of the round seeds with genotype **Rr,**  one-fourth will again have **YY,** one-half **Yy,** one-fourth **yy**. Of the wrinkled seeds, one-fourth will have **YY**, one-half **Yy** and one-fourth **yy**. The genotypic ratio in the F2 of a dihybrid can therefore be obtained by combining the two monohybrid ratios of which it is composed.

|  |  |  |  |
| --- | --- | --- | --- |
| **F2 ratio of Rr** | **F2 ratio of Yy** |  | **F2 ratio of RrYy** |
| **1 RR** | **1YY**  **2Yy**  **1 yy** | **=**  **=**  **=** | **1 RRYY**  **2 RRYy**  **1 Rryy** |
| **2 Rr** | **1YY**  **2Yy**  **1 yy** | **=**  **=**  **=** | **2 RrYY**  **4 RrYy**  **2 Rryy** |
| **1 rr** | **1Yy**  **2Yy**  **1 yy** | **=**  **=**  **=** | **1 rrYy**  **2 rrYy**  **1 rryy** |

Since **R** is dominant over  **r,** the genotype **Rr** has only round seeds. Similarly, since **Y** is dominant to **y**, the genotype **Yy** has only yellow seeds. It can be seen from the checkerboard that the phenotypic ratio is as follows:

**the dihybrid phenotypic ratio.**

|  |  |  |  |
| --- | --- | --- | --- |
| **Genotype** | **Ratio** | **Phenotype** | **Ratio** |
| **RRYY**  **RRYy**  **RrYY**  **RrYy** | 1  2  2  4 | Round yellow | 9 |
| **Rryy**  **Rryy** | 1  2 | Round green | 3 |
| **RrYY**  **RrYy** | 1  2 | Wrinkled yellow | 3 |
| **Rryy** | 1 | Wrinkled green | 1 |

The phenotypic ratio in the F2 of a dihybrid can also be determined by combining the two monohybrid ratios of 3 round: 1 wrinkled and 3 yellow : 1 green as follows:

Each character (i.e., phenotype) is represented by a single letter, for example, round by **R** and wrinkled by **r** in contrast to the genotype which is represented by two letters, for example, **RR, Rr** or **rr**.

|  |  |  |  |
| --- | --- | --- | --- |
| **F2 ratio of hybrid round** | **F2 ratio of hybrid yellow** |  | **F2 ratio of hybrid round and yellow** |
| 3 **R** | 3 **Y**  1 **y** | =  = | 9 **RY (**round yellow)  3 **Ry** (round green) |
| **1 r** | 3 **Y**  1 **y** | =  = | 3 **rY (**wrinkled yellow)  1 **ry** (wrinkled green) |

**Breeding behaviour of the F2**

That the F2 seeds are genotypically of nine different kinds can be shown from their breeding behaviour.

The round yellow peas are of four genotypes. Plants raised from seeds with **RRYY** on self-pollination produce only round and yellow seeds. Plants raised from **RRYy** produce seeds, all of which are round but three-fourths are yellow and one-fourth green. Plants raised from seeds with **RrYY** produce seeds, all of which are yellow but three-fourths are round and one-fourth wrinkled. Plants raised from **RrYy** produce seeds in the ratio of 9 round yellow : 3 round green : 3 wrinkled yellow : 1 wrinkled green.

The round green peas are of two genotypes. Plants raised from **Rryy** produce only green seeds but plants raised from **Rryy** produce seeds all of which are green but three-fourths are round and one-fourth wrinkled.

The wrinkled yellow peas are also of two genotypes. Plants raised from **rrYY** produce only wrinkled yellow seeds but plants raised from **rrYy** produce seeds all of which are wrinkled but three-fourths are yellow and one-fourth green.

There is only one genotype for wrinkled green peas, viz., **rryy** and this breeds true.

**Breeding behaviour of the F2 from a dihybrid cross in peas**

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Phenotype** | **Genotype** | **Numbers obtained by Mendel** | **Ratio** | **Breeding behaviour on self-fertilisation** |
| Round yellow | **RRYY**  **RRYy**  **RrYY**  **RrYy** | 38  65  60  138 | 1  2  2  4 | Breeds true  Segregates into3:1 for colour  Segregates into 3:1  For form of seed  Segregates into 9: 3 : 3 : 1 |
| Round green | **RRyy**  **Rryy** | 35  67 | 1  2 | Breeds true  Segregates into3:1 for form of seed |
| Wrinkled yellow | **RrYY**  **RrYy** | 28  68 | 1  2 | Breeds true  Segregates into3:1 for colour |
| Wrinkled green | **Rryy** | 30 | 1 | Breeds true |

**Reciprocal cross**

If, instead of a cross between a round yellow-seeded plant as female and a wrinkled green-seeded plant as male, a cross is made between a wrinkled green-seeded plant as female and a round yellow-seeded plant as male, the F1 is again round yellow and the F2 shows the same segregation into 9 round yellow : 3 round green : 3 wrinkled yellow : 1 wrinkled green.

The same results are also obtained even if the cross is between a plant with round green seeds and one with wrinkled yellow seeds.

**Test cross**

That the dihybrid forms four kinds of female gametes and four kinds of male gametes in equal numbers can be shown by crossing it with the double recessive.

From a cross between the dihybrid as female and the wrinkled green seeded plant as male, Mendel obtained 31 round yellow, 26 round green, 27 wrinkled yellow and 26 wrinkled green seeds. As the recessive plant produces only one kind of male gamete, **ry**, a 1 : 1 : 1 : 1 ratio is possible only if the dihybrid produces four kinds of female gametes, **RY, Ry, rY** and **ry,** in equal numbers.

From a back cross between the double recessive as the female and the dihybrid as the male, he obtained 24 round yellow, 25 round green, 22 wrinkled yellow and 27 wrinkled green seeds. The progeny is in the ratio of 1 round yellow : 1 round green : 1 wrinkled yellow : 1 wrinkled green, thereby showing that the dihybrid produces four types of gametes, **RY, Ry, rY**  and **ry** in equal numbers.

**Cytological interpretation of Mendel's Second Law**

The independent assortment of members of different pairs of alleles according to the Second Law of Mendel is possible only because the members of various homologous pairs of chromosomes are distributed to the gametes independent of each other.

Let us presume that the gene **R** responsible for the round seed coat in pea is situated at a particular locus in chromosome No.1 and the gene **Y** responsible for the yellow cotylendons is situated at a particular locus in chromosome No.2. The round yellow-seeded parent has a pair of No.1 chromosomes, each with a gene **R** and a pair of No.2 chromosomes, each with a gene **Y.** At meiosis one member of each pair of chromosomes enters each gamete. Each gamete therefore contains a single No.1 chromosome with **R** and a single No.2 chromosome with **Y.**

Let us presume that allele **r** responsible for the wrinkled seed coat in pea is situated at the same locus of chromosome No.1 as the allele **r**, and the allele **y** responsible for the green cotyledons is situated at the same locus of chromosome No.2, as the allele **Y.** The wrinkled green-seeded parent has a pair of No.1 chromosomes, each with an allele **r** and a pair of No.2 chromosomes, each with **y**. At meiosis, one member of each pair of chromosomes enters each gamete. Each gamete therefore contains a single No.1 chromosome with **r** and a single No.2 chromosome with **y**.

When pollen from the wrinkled green-seeded parent is dusted on the emasculated flowers of the round yellow-seeded parent, a female gamete with a single No.1 chromosome (with **R**) and a single No.2 chromosome (with **Y)**  is fertilised by a male gasmete with a single No.1 chromosome (with **r**) and a single No.2 chromosome (with **y**). The hybrid round yellow seeds have a pair of No.1 chromosomes, the maternal member with **R** and the paternal member with **r**, and a pair of No.2 chromosomes, the maternal member with **Y** and the paternal member with **y**.

Plants raised from the hybrid round yellow seeds form female and male gametes in their ovaries and anthers respectively. During meiosis in their flowers, the members of each homologous pair of chromosomes always separate from each other but the separation of the homologues of one pair is not at all affected by the separation of the homologues of the other pair. In any one gamete, it is a mater of chance whether a maternal chromosome 1 is associated with a maternal chromosome 2 or with a paternal chromosome 2. It is equally a matter of chance whether a gamete which has a paternal chromosome 1 also has a paternal chromosome 2 or a maternal chromosome 2.

Four types of gametes are, therefore, produced in equal numbers as follows:

1. with maternal chromosome 1 and maternal 2
2. with maternal chromosome 1 and paternal 2
3. with paternal chromosome 1 and maternal 2
4. with paternal chromosome 1 and paternal 2

Consequently, the gametes will have **RY, Ry, rY or ry**

During fertilisation, the gametes unite at random. The F2 therefore consists of 16 possible combination in equal numbers, as in Fig. 4-1

**Incomplete dominance in dihybrids**

The phenotypic ratio in the second generation from a cross involving two pairs of alleles is the same as the genotypic ratio if dominance is incomplete in both pairs of alleles.

For example, in snapdragon, when a plant with crimson flowers is crossed with a plant with white flowers, the F1 is pink and the F2 segregates into 1 crimson : 2 pink : 1 white. When a plant with broad leaves is crossed with a plant with narrow leaves, the F1 has medium-broad leaves and the F2 segregates into 1 broad : 2 medium : 1 narrow. When a plant with crimson flowers and broad leaves (**CCBB**) is crossed with one with white flowers and narrow leaves (**ccbb**), the F1 has pink flowers and medium broad leaves (**CcBb**).

The F2 segregates as follows:

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| 1 **CC** | 1 **BB**  2 **Bb**  **1 bb** | =  =  = | 1. **CCBB** 2. **CCBb**   1 **CCbb** | Crimson, broad  Crimson, medium  Crimson, narrow |
| 2 **Cc** | 1 **BB**  2 **Bb**  **1 bb** | =  =  = | 1. **CcBB**   **4 CcBb**  2 **CCbb** | Pink, broad  Pink, medium  Pink, narrow |
| 1 **cc** | 1 **BB**  2 **Bb**  **1 bb** | =  =  = | 1. **CcBB** 2. **CcBb**   1 **ccbb** | White, broad  White, medium  White, narrow |

**Trihybrid ratio**

Just as a monohybrid is a hybrid resulting from a cross between parents differing in a single gene and a dihybrid is a hybrid resulting from a cross between parents differing in two genes, a trihybrid is a hybrid resulting from a cross between parents differing in three genes.

A trihybrid behaves just like a dihybrid and when it forms gametes, the two members of each pair of alleles always separate from each other but the members of different pairs of alleles assort into the gametes independent of each other.

As an example, we can consider Mendel's cross between pea plant with round seeds, yellow cotyledons and grey-brown seed coats and one with wrinkled seeds, green cotylendons and white seed coats. All the hybrid seeds resulting from this cross are round, yellow and grey-brown.

An individual heterozygous for three independently assorting pairs of alleles produces eight types of gametes in equal numbers as follows:

|  |  |
| --- | --- |
| **RYB** | **RYB** |
| **Ryb** | **Ryb** |
| **RyB** | **RyB** |
| **Ryb** | **Ryb** |

Eight kinds of male gametes fertilising at random eight corrresponding kinds of female gametes produce an F2 consisting of 64 possible combinations composed of 27 different genotypes. Since **R** is dominant over **r, Y** over **y**, and **B** over **b**, the 27 different genotypes fall into eight visibly different types (i.e., phenotypes) as follows:

|  |  |  |  |
| --- | --- | --- | --- |
| 3  **R** | **3Y**  1 y | **3 B = 27 RYB**  **1 b = 9 Ryb**  **3 B = 9 RyB**  **1 b = 3 Ryb** | (round, yellow, brown)  (round, yellow, white)  (round, green, brown)  (round, green, white) |
| 1  **r** | **3Y**  1 y | **3 B = 9 rYB**  **1 b = 3 rYb**  **3 B = 3 ryB**  **1 b = 1 ryb** | (wrinkled, yellow, brown)  (wrinkled, yellow, white)  (wrinkled, green, brown)  (wrinkled, green, white) |

The progeny from a test cross between such a trihybrid and the triple recessive consists of eight phenotypes in equal proportions.

**Polyhybrid ratio**

Polyhybrid is a hybrid resulting from a cross between parents differing in several genes.

When two individuals heterozygous for one pair of alleles are crossed, two visibly different types (i.e., phenotypes) can be recognised in their offspring. When the individuals are dihybrids, the number of phenotypes potentially possible in their offspring is four and when the individuals are trihybrids, the number is eight. Most plants and animals, including man, are hetreozygous for several pairs of alleles and produce a very large number of visible different types of offspring. As man has 23 pairs of chromosomes, 23 pairs of genes can assort independent of each other and therefore, the number of phenotypes potentially possible in the offspring of two parents heterozygous for 23 pairs of alleles in 223 or 8,388,608.

The number of gametes produced by a hybrid, the number of phenotypes and genotypes in the F2 and the number of possible combination in the F2 are presented in table 4-3.

**Characteristics Of Hybrids**

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| No. of allelic pairs involved | No. of types of gametes produced by F1 | No. of phenotypes in F2 if dominance is complete | No. of genotypes in F2 | No. of possible combinations in F2 |
| 1 | 2 | 2 | 3 | 4 |
| 2 | 4 | 4 | 9 | 16 |
| 3 | 8 | 8 | 27 | 64 |
| 4 | 16 | 16 | 81 | 256 |
| N | 2n | 2n | 3n | 4n |

**Chi - Square Test**

The chi-square test is a test for significance. It establishes as to how closely the observed data fit the predicated ratio. It is made by ascertaining the probability that the deviation of the observed ratio from the predicated ratio is due to chance and not due to some other factors such as experimental condition, biased sampling or even a wrong hypothesis. It is based on the `null hypothesis' which assumes that the predicated value and the observed value are the same and the deviation observed is only due to chance.

If the probability (P) of the observed ratio is equal to or less than 5 in 100>0.05), the deviation between the expected and the observed ratio is considered significant and not simply attributable to chance. If the probability is one in 100 or less (P<0.5), the deviation is not considered statistically significant and can be expected on the basis of chance alone.

Chi-square will be zero if the observed and expected numbers in each class are the same. The value of chi-square will increase as the observed numbers deviate much from the expected. Conversely, chi-square value will decrease as the observed numbers approach the expected. The chi-square method is a biometrical too employed to find out whether the observed data fit to a hypothetical ratio.

The chi-square for goodness of fit of segregation ratios was first suggested by Harris(1912).

From Table 3-1 presenting the summary of results of Mendel's seven crosses, the F2 ratios varied from 2.82 : 1.00 to 3.15 : 1.00 but not exactly 3:1. Thus there are deviations from the expected ratio. Whether such deviations are statistically significant or not is tested by the chi-square(x2) test for goodness of fit. If the deviation is not significant, it indicates that the deviation is more due to causes other than heredity and the probability indicates that the observed and the expected ratios fit well (P<0.05).

The F2 population for round and wrinkled seeds from Mendel's experiment may be subjected to chi-square test:

|  |  |  |  |
| --- | --- | --- | --- |
| **Population** | **Round** | **Wrinkled** | **Total** |
| Observed number (o) | 5474 | 1850 | 7324 |
| Expected number (E) based on 3:1 ratio | 5493 | 1831 | 7324 |
| Deviation (d = o-E)  D2  D2 | -19  361  361 | 19  361  361 | 0 |
| E | 5493  = 0.0657  x2 = ∑[d2/E]  =0.2629 | 1831  = 0.1972 |  |

As there are two classes viz., round and wrinkled, the degree of freedom (df) is n-1 = 2-1 = 1.

Referring to the chi-square table (Fisher R.A. and F. Yates. Statistically Tables for Biological, Agricultural and Medical Research. Oliver and Boyd Ltd.,), for n = 1 and x2 =0.2629, P (probability) lies between 0.50 and 0.70. As P >0.05, the deviation is inferred to be non-significant and the goodness of fit for 3: 1ratio is proved.

Application of the chi-square test for the dihybrid ratio may be considered, taking the form of ripe seed (round or wrinkled) and colour of cotyledons (yellow or green) of Mendel's experiment together.

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **Population** | **Round**  **Yellow** | **Round**  **Green** | **Wrinkled**  **Yellow** | **Wrinkled**  **Green** | **Total** |
| Observed number (o) | 315 | 108 | 101 | 32 | 556 |
| Expected number (E) based on 9 : 3 : 3 : 1 ratio | 312.75 | 104.25 | 104.25 | 34.75 | 556 |
| Deviation (d=O-E) | 2.25 | 3.75 | -3.25 | -2.75 | 0 |
| D2/E | 5.0625/312.75  = 0.0162 | 14.0625/104.25  = 0.1349 | 10.5625/104.25  = 0.1013 | 7.5625  /34.75  = 0.2175 |  |
| X2 =∑ [d2/E] = 0.4699 |  |  |  |  |  |

As there are four classes, the degrees of freedom are 3. For n = 3 and x2 = 0.4699, P lies between 0.90 and 0.95. thus it is observed that the deviation of the expected is not significant from the observed and the ratio of 9 : 3 : 3: 1 fits well with the observed number of the four classes.

**Test of Independence or Contingency test**

In genetic studies, one set of observations taken under a particular condition may not be the same as those under a different condition. It is therefore required to find out whether the results are dependent (contingent upon) or independent of the conditions under which they are observed. This test is called a test of independence or contingency test.

If **A** and **a** are the phenotypes and the data on number of plants segregating for **A** and **a** in two experiments are considered, the x2 for each experiment is calculated. If the x2 value in each experiment is less than the x2 value for a probability level of 0.05, the hypothesis can be accepted that the observed results are independent of the experimental conditions.

If the calculated x2 value exceeds the x2 at P= 0.05, it indicates that the results depend upon the conditions in which the experiments are conducted.

To calculate a contingent x2, both the observed numbers and the marginal totals are used. If N is the total number of observations and n1, n2, n3, n4 are the individual numerical contributions, the x2 is calculated as follows.

|  |  |  |  |
| --- | --- | --- | --- |
| **Phenotype** | **A** | **A** | **Total** |
| Experiment  I  II | N1  N3 | N2  N4 | (n1 + n2)  (n3 + n4) |
| Total | (N1 + n3) | (n2 + n4) | (n1 + n2 + n3 + n4)= N |
| [ ( n1 n4 - n2 n3) - 1/2 N]2 N  x2 = \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_  (d.f. = 1) (n1 + n2) (n3 + n4) (n1 + n3) (n2 + n4) | | | |

If the calculated x2 value for d.f. = 1 is more than that for P = 0.05, then the contigency x2 test is satisfied for the independence of the experimental conditions.

**Test of Homogeneity**

When independent experiments are conducted to study the inheritance of a particular character, it may be necessary to pool the data of the different experiments if they are sufficiently uniform. To find out whether the separate samples are sufficiently uniform or not, the test of homogeneity is adopted.

For this, the x2 of each individual sample may be calculated based on the expected ratios. Then the individual x2 is summed up to get a total x2 which consists of the x2 contributed by the departure of the pooled data from the expected ratio and the x2 contributed by the differences between the individual samples. Then the x2 for the summed data of all the samples is calculated. For, this the degree of freedom is the total of the degree of freedom of each sample. If the d.f. of each sample is n-1=1 and if there are six samples, then d.f. is six. Then the x2 for the summed data is subtracted from the summed x2 to obtain the homogeneity x2. Similarly the number of d.f. for the summed data may be subtracted form the d.f. for the summed x2 data.

If the homogeneity x2 exceeds the x2 value for P =0.05, the hypothesis that the samples are homogeneous is rejected. If the homogeneity x2 is less than the chi-square value for P =0.05, it means that the samples are homogeneous and the data obtained from the different samples can be pooled together and the summed data tested for the expected ratio.

For example, if the summed total x2 of the six samples is 2.05 for 6 d.f. and the x2 for the summed data is 0.02 for 1 d.f., then the homogeneity x2 is 2.05 - 0.02 = 2.03 for 6 - 1 = 5 d.f.

|  |  |  |  |
| --- | --- | --- | --- |
| **Source** | **X2** | **d.f.** | **P value** |
| Total | 2.05 | 6 |  |
| Summed data | 0.02 | 1 | 0.80 - 0.90 |
| Homogeneity | 2.03 | 5 | 0.80 - 0.90 |

Chi-square value for 5 d.f. at P = 0.05 is 11.070 (according to Fisher's probability table). As the homogeneity x2 is only 2.03, i.e., less than 11.070, the samples are proved to be homogeneous and the data can be pooled.

**2. Mendel's Law Of Segregation**

So long as there are no observable differences between one parent and the other or between the parents and the offspring with reference to a particular character, it is not possible to find out whether the offspring has inherited the character from one parent or the other parent or both. Hybridisation between parents that differ in some way with references to a character is therefore the best method of studying the mode of inheritance of a character.

**Mendel's hybridisation experiments**

Mendel chose for his hybridisation experiments twenty-two varieties of pea (Pisum sativum) that differed from one another constantly in one or more characters.

The characters which were selected for his experiments are the following:

(1) Form of ripe seeds : round or wrinkled.

(2) Colour of cotyledons : yellow or green.

(3) Colour of seed-coat : grey-brown or white.

(4) Form of ripe pods : inflated or constricted.

(5) Colour of unripe pods : green or yellow.

(6) Position of flowers : axial, i.e., distributed along the

main stem, or, terminal, i.e., bunched at the top of the stem.

(7) Length of stem : tall or short.

Mendel confirmed that these varieties yielded `perfectly constant and similar offspring' when self-fertilised, i.e., they bred true.

Mendel crossed one variety with another and observed the appearance of the resulting hybrids in the first and succeeding generations. Thus, for instance, he crossed a variety of pea having round seeds with a variety having wrinkled seeds.

In the case of each of the seven crosses, he found that all the offspring (the first filial generation or F1) were uniform and resembled one of the parents so closely that the character of the other parent escaped observation completely. Those parental characters which appeared in the F1 were termed dominant and those parental characters which entirely disappeared in the F1 were termed recessive. Thus, for example, all of the hybrid seeds resulting from a cross between a round-seeded plant and a wrinkled seeded one were rounded.

Mendel self-fertilised the first-generation hybride and found that, unlike the F1 which was uniform, the second generation or the F2 consisted of two different kinds. In the F2' the recessive character reappeared, together with the dominant character. Three-quarters of the F2 showed the dominant character and one-quarter the recessive character. For example, from 253 plants raised from the hybrid seeds of a cross between the round-seeded variety and the wrinkled-seeded variety, Mendel obtained an F2 generation of 7,324 seeds. Among these seeds, 5,474 were round and 1,850 were wrinkled. The ratio 2.96 round: 1.00 wrinkled is very close to the ratio of 3 round: 1 wrinkled.

**Summary of results of Mendel's seven crosses**

|  |  |  |  |
| --- | --- | --- | --- |
| Parents | F2 | | |
|  | Dominant | Recessive | Ratio |
| Round X wrinkled seed | 5,474 | 1,850 | 2.96 : 1.00 |
| Yellow X green cotyledon | 6,022 | 2,001 | 3.01 : 1.00 |
| Coloured X white seed | 705 | 224 | 3.15 : 1.00 |
| Inflated X constricted pod | 882 | 299 | 2.95 : 1.00 |
| Green X yellow pod | 428 | 152 | 2.82 : 1.00 |
| Axial X terminal flower | 651 | 207 | 3.14 : 1.00 |
| Tall X dwarf plant | 787 | 277 | 2.84 : 1.00 |
| Total | 14,949 | 5,010 | 2.98 : 1.00 |

Mendel found that the F2 recessives, when self-fertilised bred true. One third of the F2 dominates bred true while the remaining two-thirds yielded offspring which displayed the dominant and recessive characters in the proportion of 3:1. There were, thus, three classes in the F2' viz., pure dominant, hybrid dominant and pure recessive in the porportion of 1:2:1. For example, among 565 plants raised by Mendel from the round seeds of the F2 generation, 193 yielded round seeds only, and 372 yielded round and wrinkled seeds in the proportion of 3:1. The ratio 1.00 pure round : 1.93 hybrid round is close to the ratio of 1 pure dominant : 2 hybrid dominant. All the plants raised by Mendel from the wrinkled seeds produced only wrinkled seeds.

**Gene**

Mendel postulated that `characters' are determined by `elements' found in the sex cells or gametes. The character and its determinant are thus different and Bateson coined the word `factor' for that which determines a character. The Danish geneticist Johannsen recognised that there is something in the gametes and in the fertilised egg that determines a character and he proposed the word `gene' for it. Gene can be defined as the hypothetical unit of inheritance located at afixed position (i.e., the locus) on a chromosome which by interaction with the other genes, the cytoplasm and the environment controls the development of a character.

**Allele**

Mendel recognised the presence of conxtant differentiating characters. These contrasting characters are attributed to the presence of allelomorphs (allelon = one another; morphus = form) or allelic genes or alleles. Allele is defined as one of a pair (or series) of forms of a fene situated at the same locus of homologous chromosomes.

**Gene symbols**

It is customary to symbolise genes by letters. The dominant gene is represented by a capital letter and its recessive allele by the corresponding small letter. Thus, the gene responsible for round seed coat in pea is represented as **R** and its allele responsible for wrinkled seed coat as **r**.

When several genes in the same organism have to be represented, various combinations of letters are used. Thus, in maize, the gene for striated leaves is represented as **sr,** silkless (i.e., female sterile) as **sk** , salmon coloured silk as **sm,**  and shrunken endospermm as **sh**.

**Homozygote and Heterozygote**

Mendel recognised that a gamete can possess only one of a pair of alleles, for example, either **R** or r and not both. An individual formed by the union of like gametes is said to be pure. The British geneticist Bateson introduced the term homozygote (home = same; zygos = yolk) for an organism in which the two genes at the same locus of homologous chromosomes are identical. The true-breeding round-seeded pea plant is formed by the union of an egg with **R** and a sperm, also with **R** and is represented as **RR**, and the true-breeding wrinkled-seeded plant formed by the union of an egg and a sperm, also with **r**, is represented as **rr**. An individual formed by the union of unlike gametes is said to be a hybrid. Bateson called this a heterozygote (hetero = different; zygos = yolk) because the two genes at the same locus of homologous chromosomes are not identical. The round seeds of the first hybrid generation (i.e., F1) formed by the union of a gamete with **R** and another gamete with **r** are therefore heterozygous and are represented as **Rr.**

**Phenotype and Genotype**

Johannsen clearly brought out the difference between the visible character and the invisible gene that is responsible for the character. He coined the word phenotype (pheno = appear) for the visible character of an individual and the word genotype for the heredity of a plant or animal. The genotype of an individual can be determined by observing its phenotype, but as two different genotypes may possess the same phenotype because of the phenomenon of dominance, it can be confirmed only by studying the ancestry or the progeny of the individual. Thus, for example, in pea, two different genotypes, **RR** and **Rr**, have the same phenotype, viz., round seeds, but whereas the former produces nothing but round seed, the latter produces round seeds and wrinkled seeds in the ratio of 3 round : 1 wrinkled.

**Mendel's First Law**

The data from the seven crosses led Mendel to suggest that the members of a pair of `differntiating elements' in a hybrid `mutually separate themselves' during the formation of gametes so that half of the gametes carry one `formative element element' and the other half carry the other element.

This conclusion is now known as Mendel's First Law of Segregation and can be expressed as follows:

When a pair of contrasting characters are broought together in a hybrid, the factors responsible for the characters do not blend or contaminate each other in the hybrid, but when gametes are formed, they segregate and pass into different gametes in a definite proportion.

**Monohybrid Ratio**

In Mendel's cross between the round-seeded and wrinkled seeded peas, both the parents belonged to true-breeding varieties. The true-breeding round-seeded parent **RR** forms gametes, all of which are alike and have **R** and the true-breeding wrinkled-seeded parent **rr** forms gametes, all of which are alike and have **r**. When pollen from the wrinkled seeded parent is dusted on the emasculated flowers of the round-seeded parent, an egg with **R** is fertilised by a sperm with **r**. The hybrid seeds are therefore **Rr** and all of them are round because **R** is dominant to **r**.

Though there is no visible indication of the presence of allele **r** in the F1, the alleles **R** and **r** do not blend or fuse with each other while they are together in the F1. The alleles **R** and **r** do not contaminate or affect each other even though all the hybrid seeds are only round.

When the hybrid seeds are sown and when these plants form eggs and sperms, the alleles **R** and **r** segregate so that two kinds of eggs, one with **R** and the other with **r**, and the two kinds of sperms, one with **R** and other with **r**, are produced in approximately equal numbers.

In fertilisation, the gametes combine at random (i.e., they unite freely in all possible combinations). The F2 therefore consists of four combinations, viz., **RR**, **Rr, rR** and **rr**  in approximately equal numbers. Seeds with **RR** have only the gene for round and are therefore round. Seeds with **Rr** have a gene for round and a gene for wrinkled form, but since **R** is dominant over its allele **r**, they are also round. Seeds with **rr** have only the gene for wrinkled and are therefore wrinkled.

The genotypic ratio in the F2 is therefore, 1 **RR** : 2**Rr** : 1**rr.** Since **R** is dominant over **r**, the genotypes **RR** and **Rr** have the same phenotype, viz., round, and the phenotypic (or visible) ratio is therefore, 3 round : 1 wrinkled.

As the round seeds of the F2 generation are genotypically of two kinds, about one-third being **RR** and about two-thirds being **Rr,** their breeding behaviour is also of two kinds. Plants raised from seeds with **RR**, on self-pollination, produce only round seeds, but plants raised from seeds with **Rr,** on self-pollination, produce round and wrinkled seeds in the ratio of 3 round : 1 wrinkled. As the wrinkled seeds are always homozygous for the recessive allele, all of them breed true. Plants raised from wrinkled seeds of the F2 generation, on self-pollination, therefore, produce only wrinkled seeds.

**Cytological interpretation of Mendel's First Law**

Allellegenes in an individual segregate according to Mendel's First Law only because they are carried on homologus chromosomes that sepearate at meiosis and go to different gametes.

Let us presume that the allele **R** responsible for the round seed coat in pea is situated at a particular locus in chromosome No.1. The true-breeding round-seeded parent has a pair of these chromosomes, and each of these will have a gene for round seed coat. At meiosis, the two homologous chromosomes separate and consequently the two allelles **R** and **R** will also separate. Each gamete has only one homologous chromosome and consequently will have only one **R**  allele. A homozygous round-seeded plant, thus, produces gametes, all of which are alike in having a single No.1 chromosome with the allele **R**.

Let us presume that the allele **r** responsible for the wrinkled seed coat in pea is situated at the same locus of chromosome No.1 as its allele **R**. The true-breeding wrinkled seeded parent has a pair of these chromosomes and each of these will have gene for wrinkled seed coat. At meiosis, the two homologus chromosomes separate and consequently the two genes **r** and **r** will separate. Each gamete has only one homologous chromosome and consequently will have only one **r** allele. A homozygous wrinkled seeded parent, thus, produces gametes, all of which are alike in having a single No.1 chromosome with the allele **r**.

When pollen from the homozygous wrinkled seeded parent is dusted on the emasculated flowers (i.e., flowers in which anthers have been removed) of the homozygous round-seeded parent, an egg with a single No.1 chromosome (with **R**) is fertilised by a sperm with a single No.1 chromosome (with **r**). The hybrid round seeds have a pair of No.1 chromosomes, one member of the pair with **R** and the other member of the pair with **r.**

Plants raised from the hybrid round seeds form eggs and sperms in their ovaries and anther respectively. During meiosis in their flowers, the two homologous chromosomes separate, go to opposite poles, and enter into different gametes. Consequently, the allelles **R** and **r** will also separate and go to different gametes. Each gamete has only one homologous chromosome and consequently will have only one allelle, either **R** or **r**, but never both.

Since the two homologous chromosomes enter two different gametes, half the gametes will have one No.1 chromosome and half the gametes will have the other No.1 chromosome. Consequently half the gametes will have **R** and half will have **r**. One half of the eggs will, therefore, have **R** and the other half will have **r**. Similarly, one half of the sperms will have **R** and the other half will have **r**.

During fertilisation, the union of like eggs and sperms occurs with the same frequency as the union of unlike eggs and sperms. The F2 therefore consists of four combinations is equal numbers: **RR, rR**  and **rr**.

**Purity of gametes**

The most important principle of Mendel's Law of Segregation is that even hybrid individuals produce gametes which are always pure. Individuals which are pure (homozygous) for one allele produce only one kind of gametes, but individuals which are hybrids (heterozygous) with reference to one pair of alleles produce two kinds of gametes, each of which has only one or the other of the two alleles. A gamete of even a hybrid is thus pure and has either the dominant allele or the recessive allele but never both.

That two kinds of gametes are formed by a hybrid in approximately equal numbers has been shown in several species. For, example, in maize, starchy varieties have endosperm and pollen grains that turn blue with iodine and waxy varieties have endosperm and pollen grains which do not turn blue with iodine but remain as red. The difference is due to a single pair of alleles of which the gene for starchy is dominant over its allele for waxy. The F1 generation from a cross between the starchy and the waxy varieties consists of starchy plants and the F2 shows a segregation of 3 starchy : 1 waxy. Demerec stained the pollen grains from such an F1 hybrid plant with iodine and found that 3,437 pollen grains stained blue and 3,482 stained red. It can thus be seen that the F1 which is heterozygous for starchy and waxy produces pollen grains of two kinds in approximately equal numbers, one kind with the allele for starchy and the other kind with the allele for waxy grains.

**Back cross and Test cross**

Back cross is a cross between a hybrid and either of its parents whereas test cross is a cross between hybrid and a recessive homozygote.

That individuals which are hybrid (heterozygous) for one pair of alleles produce two kinds of gametes in approximately equal numbers can also be shown by crossing the hybrid with its own recessive parent (i.e., back cross) or with any other recessive individual (i.e., test cross).

On crossing an F1 hybrid with the recessive parent, about half the offspring formed show the dominant character and the other half show the recessive character. For example, seeds obtained from a cross between a plant raised from the hybrid round seed, and the wrinkled-seeded parent are in the ratio of 1 round : 1 wrinkled.

As the recessive parent produces only one kind of gametes, all with the recessive allele, an 1 :1 ratio is possible only if the F1 hybrid produces two kinds of gametes, one kind with the dominant allele and the other kind with the recessive allele, in approximately equal numbers.

That an individual exhibiting the dominant character is a pure (homozygous) or a hybrid (heterozygous) one can be found out if it is test crossed with a recessive individual. If the individual is a pure dominant, all the offspring will exhibit the dominant character, e.g.,

|  |  |  |
| --- | --- | --- |
| Round pea  **RR** | X | Wrinkled pea  **Rr** |
|  | All round  **Rr** |  |

If the individual is a hybrid, the offspring will be in the ratio of 1 dominant : 1 recessive, e.g.,

|  |  |  |
| --- | --- | --- |
| Round pea  **Rr** | X | Wrinkled pea  **rr** |
| 1 round **Rr** | : | 1 wrinkled  **rr** |

**Reciprocal cross**

Reciprocal cross is a second cross involving the same characters as the first but with the sexes of the parents interchanged.

In crosses between two plants differing in a pair of contrasting characters, any one may be used as the female or male parent. Thus, a cross can be made between a round seeded pea plant as female and a wrinkled seeded one as male. A second cross between the wrinkled seeded plant as female and the round seeded one as male is called a reciprocal cross.

Whichever way cross is made, the results will be the same, in case nuclear genes determine the characters. However when hereditary factors in the cytoplasm also interact with nuclear genes, reciprocal differences have been observed.

In representing crosses, it is conventional to write the female parent first and the male parent second.

**Xenia**

Mendel crossed a true-breeding dwarf pea plant with a true breeding tall pea plant by dusting the pollen from the latter on the stigma of the emasculated flowers of the former. Seeds formed on the dwarf pea plant were the hybrid seeds and plants raised form these seeds showed the dominant character, i.e., tallness of plants.

Similarly Mendel crossed a true-breeding wrinkled-seeded pea plant with a true breeding round seeded plant by dusting pollen from the latter on the stigma of the emasculated flowers of the former. Seeds formed on the wrinkled-seeded parent were round. They were the hybrid seeds and they exhibited the dominant character. The round form of seed was attributed to xenia which is the effect of pollent on the embryonic and maternal tissue of a fruit.

Another example of xenia is given below. In maize, one gene produces purple-coloured seeds and its recessive allele produces colourless seeds. If the silks (i.e., styles) of plants producing colourless seeds are dusted with pollen from the purple seeded plants, the cobs of the former produce purple seeds. Xenia is due to a dominant gene that exerts its influence on the fruit (or seed) instead of on the parts of a plant.

**Incomplete dominance**

In several crosses dominance is incomplete and the hybrids resemble neither parent exactly but are more or less intermediate between the two.

In a cross between a black and a white Andalusian fowl, the F2 is neither black nor white but blue. The F1 hybrids are however uniform in this trait, all of them being only blue. When these are mated, the offspring are in the ratio of 1 black : 2 blue : 1 white.

The gene **B** produces black plumage and its allele **b** produces white plumage. Since neither allele is dominant, the heterozygote **Bb** is with blue plumage. A quarter of the F2 offspring are homozygous for black and another quarter homozygous for white. Half of the F2 offspring are heterozygous and are blue. They never breed true.

Several other cases of incomplete dominance can be cited. In Mirabilis, a red-flowered plant crossed with a white-flowered one gives F1 which are all pink in colour and a segregation in the F2 into 1 red : 2 pink : 1 white. In Antirrhinum (Snapdragon) a plant with crimson petals crossed with one with white petals gives first-generation hybrids that are all pink.

Whenever dominance is incomplete, the phenotypic ratio in the F2 is identical with the genotypic ratio of 1 : 2 : 1.

**Codominance**

Unlike incomplete dominance, where an intermediate expression between those of the parents is observed, there are cases where the heterozygote expresses the phenotype of both the parents mingled together, as neither of the alleles exhibit either the dominant or the recessive expression. Such a condition where both the otherwise dominant and recessive alleles are capable of expression equally in the heterozygote is called codominance. However as in incomplete dominance, the codominate heterozygote will have a distinct phenotype different from those of the parents.

The best example of codominance is found in the Short horn breed of cattle in which the heterozygote (**Ww**) is the highly prized `roan' animal whose coat consists of a mixture of red and white hairs equally distributed throughout the body, while its parents have red (**Ww**) and white (**ww**) hairs respectively. When two F1s with roan coat are crossed, the F2 population segregate as 1 red (**WW**) : 2 roan (**Ww**) : 1 white (**ww**).

The alleles **Lm** and **LN** in the MN blood group series of human beings are codominant which could be identified by the agglutination test based an the antigen - antibody relationship as detailed below:

|  |  |  |  |
| --- | --- | --- | --- |
| **Reaction to** | | | |
| **Genotype** | **Antiserum - M** | **Antiserum - N** | **Phenotype** |
| LM LM | + | - | M |
| LN LN | - | + | N |
| LM LN | + | + | MN |

Codominance is also referred to as mosaic dominance as the nondominance relationship of the two alleles exhibits as a mosaic of both their expressions.

5. **CHROMOSOME THEORY OF INHERITANCE AND PARALLELISM**

Mendel's Laws of Inheritance assume that the hereditary materials are particles called genes found in the cells of all living organisms. Genes have neither been seen nor analysed chemically but it is estimated that the diameter of one gene, assuming it to be a spherical particle, is something like 6 millimicrons (0.000006 millimetre) and its molecular weight is round about 1,00,000. Genes are thus fundamental units of life, just like atoms which are the ultimate units of matter.

Mendel established the existence of genes without knowing anything about chromsomes, in fact, several years before chromsomes had been named or described in detail. His conclusions on the existence of genes were based on his breeding experiments.

During the thirty-four years from 1866, when Mendel's paper was published to 1900, when his work was rediscovered, chromosomes were described and their behaviour was studied with great care and enthusiam. The regualr and pracise longitudinal division of the chromosomes into two identical halves and the distribution of the two halves to the two daughter cells by mitosis,the neat separation of chromosomes and the reduction in the number of chromosomes from the diploid (2n) to the haploid state (n) during the formation of gametes by meiosis and the restoration of the diploid number of chromosomes in the zygote by fertilisation showed that the chromosomes are of great importance to the ell. Even before Mendel's work was rediscovered, and the existence of Mendelian units known, it was realised that chromosomes are intimately concerned with inheritance.

With the rediscovery of Mendel's work, the question naturally arose as to what the relationship between the genes and the chromosomes is. **Genetics** which is the study of heredity thus joined hands with **Cytology** which is the study of cells, and within a short time, these two independent subjects of investigation fused to form a hybrid subject called **Cytogenetics**.

**Hypothesis of Sutton and Boveri**

The hypothesis that the Mendelian genes must be carried on the chromosomes was put forth simultaneously, but independently, in 1902 by Sutton, an American biologist, and Boveri, a German cytologist. They assigned genes to chromosomes because the behaviour of chromosomes at meiosis and fertilisation resembled in a very striking way the behaviour of genes as inferred from breeding experiments.

The chromosomes maintain their individual identity, just as do genes. In favourable materials, each pair of chromosomes can be seen to be different form every other pair. Similarly genes have an individuality as can be inferred from the specific effects produced by each gene.

Chromosomes are found in pairs, each member of which has been derived from one of the two parents. The facts of inheritance can be satisfactorily explained only on the assumption that genes also occur in pairs, one member of each pair being contributed by one parent and the other by the other parent.

At meiosis, the members of a pair of chromosomes separate and go to different gametes. Thus, for example, of the first pair of chromosomes in an individual, the maternal member will enter one gamete and the paternal member will enter naother gamete. No gamete will contain the maternal and paternal members of one pair of chromosomes. Half the number of gametes will therefore contain maternal chromosome No.1 and the other half the number of gametes will contain paternal chromosome No.1. That the gametes contain only half the number of chromosomes found in the mother cells can be verified by actual counts of chromosomes. According to the Law of Segregation, each gene separates from its allele, the two members of a pair always entering different gametes. For example, of the genes **R** for round seed and **r** for wrinkled seed in pea, **R** enters one gamete and **r** enters another gamete. No gamete will contain **R** and **r** together. Half the number of gametes will therefore contain **R** and the other half the number of gametes will contain **r**. Both chromosomes and genes thus undergo segregation and in respect of both, each gamete contains only one member of the pair.

The segregation in one pair of chromosomes has been observed to be independent o f the segregation in another pair of chromosomes. Thus, of a second pair of chromosomes, the maternal member will enter one gamete and the paternal member will enter another gamete. Half the umber of gametes will therefore contain maternal chromosome No.2 and the other half the number of gametes will contain paternal chromosome No.2. In any one gamete, however, it is a matter of chance whether a maternal chromosome 1 is associated with a maternal chromosome 2 or with a paternal chromosome 2. It is equally a matter of chance whether a gamete which has a paternal chromosome 1 also has a paternal chromosome 2 or a maternal chromosome 2. The gametes may therefore contain the following combinations of chromosomes: (1) maternal 1 and maternal 2, (2) maternal 1 and paternal 2, (3) paternal 1 and maternal 2 and (4) paternal 1 and paternal 2.

According to the Law of Independent Assortment, the segregation in one pair of allelic genes is independent of the segregation in another pair of alleles. Thus, of a second pair of alleles in pea, **Y** for yellow cotyledons and **y** for green cotyledons, **Y** will enter one gamete and **y** will enter another gamete. Half the number of gametes will contain **Y** and the other half the number will contain **y**. It is however, a matter of chance whether a gamete which has **R** also has **Y** or has **y**. It is equally a matter of chance whether the gene for **r** is associated with **Y** with **y**. The gametes may therefore contain the following combinations of genes: **R** and **Y**; **R** and **y**; **r** and **Y; r** and **y**.

Thus the members of different pairs of chromosomes assort into the gamete independently of each other. Likewise, the members of different pairs of alleles assort into the gametes independently of each other. Each gamete contains only one member of each pair of chromosomes and each gamete likewise contains only one member of each pair of allelic genes.

**Proof for the theory of Sutton and Boveri**

The first definite suggestion that a chromosome determines a character came from McClung, an American zoologist, when he discovered that the male grosshoppers differ from the females in the absence of one chromosome. The female has an even number of chromosomes, all the chromosomes being in pairs. All the eggs produced by the XX female are alike in having a single X chromosome each. The male, however, has an odd number of chromosomes, one of the chromosomes being always without a partner. Two types of sperms are produced in equal numbers by the XO male, one type with the odd X chromosome and the other without it. Since the eggs are all alike and the two kinds of sperms are equal in number, the ratio of 1 female : 1 male observed in the offspring is possible only if eggs fertilised by sperms with the X chromosomes develop into female and those fertilised by sperms without the X chromosome develop into males. That the X chromosome determines sex is seen from the fact that the two types of sperms differ only in that, one type has a X chromosome while the other lacks it.

Although Sutton postulated that a random distribution of chromosomes occurred in meiosis such that the resultant haploid gametes received all possible combinations of maternal and paternal chromosomes, the proof for the independent assortment of different pairs of chromosomes was first furnished only if 1913 by Carothers. Ordinarily it is not possible to distinguish the maternal and paternal homologous chromosomes under the microscopic because they are identical in their cytologically visible features. But, in a grasshopper, in which the females are of the XX type and the males are of the XO type, Carothers found a heteromorphic pair of chromosome, the members of which regularly synapse and segregate from each other at meiosis but which differ visibly in size. She proved that there is a random distribution of the X chromosome and the members of the heteromorphic pair of chromosomes by demonstrating that four classes of a sperms as shown in Fig.10-1 are produced in equal numbers.

**Morgan's proof for the chromosome theory**

The discovery of sex-linked genes by Morgan in 1910 furnished another proof for the chromosomal theory of inheritance. He showed that the transmission of the recessive gene for white colour of the eye in Drosophila melanogaster depends upon the sex which carries the gene initially.

In a cross between a red-eyed female and a white-eyed male, the F1 flies of both sexes are red-eyed. Of the F2 offspring, all the females are red-eyed, whereas half the males are red-eyed and the other half are white-eyed. The F2 shows a segregation of 3 red : 1 white, but strangely enough, the white-eyed flies are always male.

In the reciprocal cross between a white-eyed female and a red-eyed male, the F1 females are red-eyed. In the F2 generation, one half of the females and males are red-eyed and the other half white eyed.

The different results from the reciprocal crosses can be explained only on the assumption that the gene for colour of the eyes is located on the X chromosome. Morgan thus showed that the distinctive pattern of inheritance of sex-linked genes parallels the transmission of the X chromosome.

**Bridges' proof for the chromosome theory**

The discovery of non-disjunction of X chromosomes in Drosophila by Bridges, a student of Morgan, furnished yet another proof for the chromosome theory of heredity.

Bridges made a cross between a white-eyed female and red-eyed male. He found that in a F1 population that should have contained only red-eyed females and white-eyed males, there appeared some exceptional white-eyed females and red-eyed males. He examined the chromosomes of the exceptional white-eyed daughters and the red-eyed sons under the microscope and found that the white-eyed daughters had not only two X chromosomes but also a Y chromosome and that the red-eyed sons had only one X-chromosome and no Y chromosome.

Bridges explained the results as follows: In some females, the two X chromosomes fail to disjoin at meiosis and this primary non-disjunction leads to the production of eggs either with the two X chromosomes or with no X chromosome. Fertilised by normal sperms, four types of zygote are produced as follows:

|  |  |  |
| --- | --- | --- |
| **Egg** | **Sperm** | **Zygote** |
| XX | X | XXX (usually die) |
| XX | Y | XXY (white-eyed female) |
| No X | X | X (red-eyed male) |
| No X | Y | Y (always die) |

Individuals with three X chromosomes (superfemales) usually die. The few that survive are so different form other flies that they are easily identified.

Flies with two X chromosomes and Y chromosome are like normal females in appearance and are fertile. They are white-eyed because they have inherited both their X chromosomes from their white-eyed mothers.

Flies with one X chromosomes and no Y chromosome are like normal males in appearance although they are sterile due to the absence of the Y chromosome. They are red-eyed because they have inherited their single X chromosome from their red-eyed fathers.

Individuals lacking an X chromosome die before hatching from the egg.

The work of Bridges thus proved beyond doubt that the gene for colour of the eye in Drosophila is located on the X chromosome.

**Morgan's Theory**

The work of Morgan and Bridges firmly established the fact that specific genes are borne on specific chromosomes. Study of linkage and crossing over in Drosophila melanogaster by Morgan, Sturtevant, Muller and Bridges threw more light on the genes on the one hand and the chromosomes on the other.

From the co-ordinated genetic and cytological studies on Drosophila, Morgan postulated that genes are arranged in a linear order along the length of the chromosome, each gene having a fixed place on the chromosome, and its allele a corresponding position on the homologous chromosome. He also put forward the hypothesis that the degree of linkage depends upon the distance between the linked genes in the chromosomes. This led to a new field dealing with mapping of chromosomes.

**CELL THEORY**

The theory, that the cell is the basic unit of life, and all plants and animals are composed of one or more cells, was enunciated in 1833 by two German scientists, Schledian and Schwann. That cells arise only from pre-existing cells in an equally important generalization made by another German scientist, Virchow in his `Theory of Cell Lineage', proposed in 1858.

Every plant or animal starts its life only as a single cell. This gives rise to two new cells by division. Each of these again divides into two and the process is repeated. In multicellular organisms, a number of cells is thus formed and the appearance of the mature organism depends upon the arrangement of these cells. Growth in multicellular forms thus depends upon cell division accompanied generally by cellular enlargement and differentiation. In unicellular organisms, the division of cells is a process of asexual reproduction. It leads to an increase in the total number of individual. In sexual reproduction, two cells unite to give rise to a new individual. Life is thus an uninterrupted succession of cells and what is inherited must therefore be contained in cells.

The first cell of a new individual arising from sexual reproduction is formed by the union of the egg nucleus from the female and the sperm nucleus from the male. The physical links between the parents and the offsprings are thus the nuclei of the egg and the sperm, and the hereditary material passed on from one generation to another must therefore be contained in those nuclei. The nuclei are thus the carries of heredity.

**THE CELL**

The cell consists typically of a denser body, the nucleus, enclosed in a membrane, the nuclear membrane, and a less dense semi-viscous cytoplasm surrounding the nucleus. The cell is enclosed in a semipermeable membrane, the cell membrane (or the plasma membrance), through which metabolic exchanges occur. A cell wall, often made up of cellulose, is found in addition, in plant cells.

In the cytoplasm there are a variety of structures concerned with the metabolic and synthetic activities of the cell. Animal cells contain golgi bodies and centrosomes. Plant cells contain plastids of various kinds, the most important of which are the chloroplasts which are centres of photosynthetic activity. Other particles found in the cytoplasm are the mitrochondria which are centres of enzymatric activity and microsomes which are centres of protein synthesis.

The nucleus is the most conspicuous body in stained cells. It is bounded on the outside by a nucllear membrane and is filled with a non-staining, colourless fluid called the nuclear sap or karyolymph. Lying in the karyolymph are one or more rounded bodies, the nuceoli. The main mass of the nucleus in a cell not undergoing division appears as a fine network of the threads constituting the chromatin reticulum. Chromatin is the hereditary material of the cell and it becomes distinguishable as a definite number of individual chromosomes during the cell division.

The chromosome is a thread-like body composed of two longitudinal halves called chromatids. The chromosome has a nonstaining constriction, the centromere, also called kinetochore (or the primary constriction). The centromere does not usually occupy a terminal position on the chromosomes. The portion of the chromosome on either side of the centromere is callec an arm. The centromere is an indispensable portion of the chromosome, for without it, the chromosome cannot orient itself on the spindle during nuclear division.

The shape of a chromosome is largely determined by its centromere. If the centromere is located near the end of a chromosome, the chromosome is rod-shaped (telocentric or acrocentric) If it is located at a small distance from one end of a chromosome , the chromosome is J-shaped (submetacentric). If the centromere is located at the middle of a chromosome, the chromosome is V-shaped (metacentric).

The chromosome consists of an outer membrane called pellicle, an amorphous matrix and two highly coiled thin filaments called chromonemata. These filaments are coiled in a spiral manner and have a series of bead-like swellings called chromomeres along their length.

Chromosome consists of euchromatic and heterochromatic regions. The euchromatin which makes up most of the genome is visible only in the mitotic cell. It has special affinity to basic stains and is genetically active. Heterochromatin is in a condensed state and is darkly stainable by many standard dyes used to make chromosome visible. It is genetically inert.

In the somatic cells (or the vegetative cells) of most organisms that reproduce sexually, chromosomes are found in pairs, of which one member has come form the mother and the other from the father. The chromosomes of each pair (called homologous pair) are mophologically identical but not necessarily genetically identical. The number of chromosomes in the somatic cells is called as the zygotic or somatic number and is represented as 2n. This number varies widely in both plants and animals but is constant for each species.

The functional units of heredity are called genes. Though genes are too small to be seen with even the present electron microscope, there is evidence to show that they are arranged in a linear order on the chromosomes and that each gene is specific in its action.

**Parallelism of the chromosome and genes.**

1. Chromosomes occur in pairs one received from male () parent other received from female () parent.

Gene also exsist in pairs (allele) one from another .

1. Each pair of chromosome differ from other pairs. Likewise each gene has individuality and produce specific effect.
2. At meiosis the member of one pair of chromosomes separate and go into different gametes. According to law of segregation each gene separate from its allele and each one enter into different gamete.

The segregation of one pair of chromosome has been observed is independent of the segregation in another pair chromosome. According to law of independent assortment, segregation of one pair of allele to another pair of alleles is independent.

# 6. Interaction of Genes

When expression of one gene depends on the expression or absence of another gene in an individual, it is called gene interaction

The interaction of genes at different loci that affects the same character is called Epiatasis. The term **Epistasis** was first used by Bateson in 1909 to describe two different genes that affects the same character, one of which masks the expression of the other gene.

The gene that masks the expression of another gene is called **Epistatic gene** and the gene whose expression is masked is termed **Hypostatic gene.**

**Epistasis** is also referred to as **intergenic or interallelic gene interaction**

### Characteristics of gene interaction

1. **Number of genes**

The epistatic gene interaction always involves two or more genes

1. **Affects the same character**

The epistatic gene interaction always affect the expression of one and the same

character of an individual

1. **Expression**

The phenotypic expression of one gene usually depends on the presence or   
 absence of epistatic gene

1. **Modification of the dihybrid segregation ratio**

Epistasis leads to modification of dihybrid or trihybrid ratio in F2

1. **Genetic control**

The epistatic gene interaction is usually governed by dominant gene. But now   
 recessive epistasis are also reported.

## Types of Epistasis

The various types of epistatic gene interaction include

**1. Dominant epistasis or Simple epistasis (12:3:1)**

In Dominant Epistasis, the dominant allele at one locus mask the expression of both dominant and recessive alleles at another locus resulting in 12 : 3 :1 ratio

**2. Recessive epistasis or Supplementary gene action (9:3:4)**

In Recessive Epistasis, the recessive alleles at one locus mask the expression of both dominant and recessive alleles at another locus resulting in 9 :3 : 4 ratio

**3. Dominant and recessive epistasis or Inhibitory gene action (13:3)**

In this type of Epistasis, the dominant and recessive alleles at one locus mask the expression of both dominant and recessive alleles at another locus resulting in 13 : 3 ratio

**4. Duplicate recessive epistasis or Complementary gene action (9:7)**

In this epistasis, the recessive alleles at either of the two loci mask the expression of dominant alleles at the two loci, resulting in 9 : 7 ratio

**5. Duplicate dominant epistasis or Duplicate gene action (15:1)**

In this epistasis, the dominant alleles at either of the two loci mask the expression of recessive alleles at the two loci, resulting in 15 : 1 ratio

**6. Duplicate genes with cumulatie effect or polymeric gene action (9: 6 : 1)**

In this type of epistasis, two dominant alleles have similar effect when they are separate but produced enhanced effect when they are together, resulting in 9 : 6 : 1

## Difference between Dominance and Epistasis

|  |  |  |
| --- | --- | --- |
| **Sl.No** | **Dominance** | **Epistasis** |
| 1. | Interaction of two alleles of the same gene, thus involving single locus | Interaction of two or more genes, thus involving two or more loci |
| 2. | Always refers to Heterozygotes therefore, it is not fixable | Refers to homozygotes and heterozygotes therefore, it is fixable in homozygotes |
| 3 | Dominance is of three types viz., complete, incomplete and overdominance | Epistasis is of several types viz., dominance, duplicate and recessive |
| 4. | Partial dominance alters the normal segregation ratio of 3:1 into 1:2:1 | It modifies the normal dihybrid ratio in F2 |
| 5. | It is known as intragenic or intralocus gene interaction | It is known as intergenic or interallelic or interlocus gene interaction |
| 6. | Recessive genes can express only in homozygous condition | Recessive genes can also exhibit masking effect |

**Non - allelic interaction of gene:**

The alleles of single gene located in different loci interact with one another for the expression of single character.

**Dominance:**

One allele mask the effect of another allele of the same gene.

**Epistasis**

One gene mask the effect of anther gene of the another locus

Wgandho - Rose

Brahmas - Pea

Leghorn - Single

R > S Rose x Pea

P > S RRpp ↓ rrPP

F1 Walnut

F2 9/16 walnut R - P-

3/16 rose R - PP

3/16 pea rrP-

1/16 single rrpp

Some times two independently segregating genes which influence a particular character produce new characters by interaction.

e.g. Bateson and Punnet (1907) in their experiment on comb shape of the fowls found that two gene influence the same character. Breed Wandatle - Rose comb Brahmas - Pea, Leghorn - Single comb. All of them pure breed. It was proved rose is dominant over single, pea - dominant with single. When Rose x Pea comb all F1 are Walnut and in F2 9/16 walnut, 3/16 rose, 3/16 pea, 1/16 single. It is understand double dominant (9/16) walnut (R - P-) double recessive rrpp (1/16). It differs from normal dihybrid ratio. F1 resembles either of the parents

1. New character appeared in F1
2. Another new character appeared in F2

3. Walnut is due to two independently segregating dominant genes and single comb due to two recessive gene.

**Non allelic and epitasis**

Interaction of non alleles is explained by the internal factors such as hormones & enzymes. Enzymes involved stepwise conversion of one substance to another. Then more than one gene involved in stepwise conversion they form the bio-synthetic pathway. G1+, G2+, G3+ produce functional enzyme. That involving step wise conversion and produce the end product 'C' for a particular phenotype. Mutants g1, g2, g3 produce defective enzymes either of mutant genes will prevent end product 'C' and cause metabolic blacks. A gene which prevents the expression of other non-allelic gene is called **epistatic gene**. The gene which is being masked is called **hypostatic gene**.

Due to non allelic interaction the typical dihybrid ratio 9 : 3 : 3 : 1 is modified into various types of interaction of which three types of interaction with three phenotype, 3 types of interaction with 2 type of phenotype.

**7. DUPLICATE, RECESSIVE AND COMPLEMENTARY AND DOMINANT EPISTASIS**

**Dominant epistatis**

Dominant gene (A) at one locus mask the effect of other gene (B-) at other locus. The gene B can express only when the epistatic gene is in the recessive condition (aaB-). When both of them in recessive condition (aabb) produce another phenotype.

e.g. bulb colour in onion has three colours white, red, yellow. When pure white crossed with yellow, F1 white, F2 there are 12 white 3 red 1 yellow. Here the allele I in dominant condition mask the expression of another gene R which is non its allele. The hypostatic gene (R, r) can produce its effect only when the epistatic gene is in epistatic condition.

**Recessive epistasis (supplementary gene action: 9:3:4 )**

**In** flax blue, lilac (light blue) and white

white x lilac

llQQ ↓ LLqq

F1  blue LlQq

F2 - 9/16 blue, 3/16 lilac, 4/16 white

L - Q- L - qq llqq

LlQ -

The recessive allele at one locus (aa) mask the action of another gene (B - bb), the alleles of B locus can express themselves only when the epistatic gene has dominant alleles (A -).

e.g. flower colour flax, has blue, lilac, white, when pure breeding white crossed with pure lilac, the F1 blue and then F2 9/16 blue, 3/16 lilac, 4/16 white. Lilac is basic colour L - gene Q - in the presence of L - interacts and intensifies the lilac colour to blue. Two dominant gene alleles required for blue. In the absence of basic colour (ll) the intensifying gene (Q -) has no effect. Therefore llQ - Q is white in other words Q - has no action of its won but supplements the action of another dominant gene. Here the lilac colour intensity by the supplementary gene (Q-) into blue.

**Duplicate gene with cumulative effect: (Additive)**

[Mutually supplementary]

Barly grain colour : lilac, purple, white

light purple x light purple

P1 P1 p2 p2

**Duplicate gene / cumulative / additive / mutually :**

Dominant gene at either locus (but not together) produce one A - bb, aaB = 6. Dominant allele with both locus have cumulative effect and have different phenotype (A - B-) both of them recessive (aabb) produced another phenotype e.g. barly grain colour light purple crossed with white, F1 - light purple. In the same way another purple crossed with white it shows a same result. When these light purple varities are crossed the F1 was dark purple and in F2 9/16 showed dark purple 6/16 light purple 1/16 white.

Light purple is due to a dominant gene P1 (or) another dominant gene P2. If these two dominant genes P1, P2 present together the effect is cumulative the colour is dark purple. Here both the non allelic genes mutually supplementary each other. Both the genes in recessive condition the grain colour is white.

**Duplicate gene ( 15 : 1)**

Shepherd's purse - fruit shape - triangular x round

CCDD ↓ ccdd

F1 triangular

CcDd

F2 triangular - C - D - 9

C - dd - 3

Cc D - - 3

Dominant allele at both loci alone and together produce the same phenotype with out any cumulative effect, different genes are identical but are situated in different chromosomes.

There are two different fruit shape traingular x round. and F1 - triangular and F2 15 triangular and 1 round.

**Duplicate recessive ( 9 : 7 ) [complementary recessive]**

Sweet pea - flower colour white

White x white

Ccee x ccEE

↓

purple

CcEe

C - chromogen (colour base) 9/16 purple C - E - =9

↓ ← E 7 white C - ee - = 3

purple ccE - =3 = 7

Ccee =1

Here the recessive allele at either locus intensifies with the expression of dominant alleles at other locus the recessive alleles in homozygous condition in either locus allele (or) together ( A - bb, aaB - , aa bb). The dominant alleles have no action of their own. When alone but when brought together (A - B-) complement each other and produce new phenotype.

e.g Sweet pea flower colour

when two pure breeding white crossed F1 purple colour emerged. The purple colour in F1 was due to two domiant genes each one from one parent. The gene 'C' produces chromogen (colour base) from which purple colour is produced by the action of an enzyme which is produced by another dominant gene (E) cc-absence of chromogen. So the gene E in dominant condition (E-) produces an enzymes which converts chromogen into purple colour product

**8. DEVIATION FROM MENDELS LAW – INHIBITORY GENE ACTION**

**Dominant and recessive (13 : 3) inhibitary action**

sorghum node colour

|  |  |
| --- | --- |
| Purple x green | Purple x green |
| PP x pp  ↓ | PPii x IIpp  ↓ |
| F1 Pp  Purple | Green  ↓ |
| F2 3 purple  1 green | 13/16 green, 3 purple  I - P - =9  I - pp =3 = 13  .ii pp =1  3/16 iiP- |

Some times dominant alleles of one locus (A-) and recessive alleles of another locus (bb) produce the same phenotype (A - B -, A - bb, aabb), aaP - produce another phenotype.

e.g. Node colour in sorghum generally when purple x green F1 is purple and in F2 3 purple 1 green indicating purple is dominant over green. In another cross when purple x green mated, the F1 was green and in F2 13 green, 3 purple. The gene for purple node unable to express due to the presence of another gene called inhibitant gene (I).this gene is capable of inhibiting the production of purple colour with (P), I inhibits the expression of in otherwards P is unable to express when I is in dominant condition. P can produce purple colour when the inhibitary gene is in recessive gene (ii). When the gene 'P' is homozygous recessive condition (pp) Colour is green the inhibitary gene as no phenotype effect on his own but prevent the expression of non-allelic dominant gene.

Non epistatis 9 : 3 : 3 :1

Non-allelic On gene dominant. 12 : 3 : 1 13:3

Epistatis Recessive. 9 : 6 : 4

Duplicate domi cumulative 9:6

2 gene without ---

cumulative 15:1

recessive 9 : 7

**10. MULTIPLE ALLELES**

* Allele is the short term for allelomorph (*allelon* – of one another and *morphus*-form)
* The term allelomorph was coined by Bateson
* Allele is the alternative form of a gene
* Alleles are of two types viz., either dominant or recessive and wild and mutant type
* Mendel observed only two forms of a gene for all the seven characters he studied in garden pea. Later, it was observed that in some cases, a gene had more than two allelic forms.
* More than two alleles at the same locus gives rise to a multiple allelic series. Therefore, the existence of more than two alleles at the same locus of a homologous chromosome is referred to as multiple alleles
* Presence of multiple alleles adds variability for a character
* The number of possible genotypes in a series of multiple alleles can be calculated by using the formula ½ {n x 1 (n + 1)} where, n is the number of identified alleles in that group. For example, if there is 4 alleles in the multiple allelic series, then, ½ (4 x 5)=10 genotypes are possible

**Main features of alleles**

1. Alleles are alternative forms of a gene. They occupy the same locus on a particular chromosome
2. Alleles govern the same character of an individual
3. A haploid cell has single copy of an allele, in diploid two alleles and in polyploids ,more than two alleles for a character
4. an individual may have identical alleles at the corresponding locus of homologous chromosomes (homozygous) or two different alleles (heterozygous)
5. The alleles may be dominant and recessive types or wild and mutant types

**Main features of multiple alleles**

1. Multiple alleles always belong to the same locus and one allele is present at a locus at a time in a chromosome
2. Multiple alleles always control the same character of an individual. However, the expression of the character will differ depending on the allele present.
3. There is no crossing over in a Multiple allelic series. When two alleles are involved in a cross, the same two alleles are recovered in the F2 or test cross progeny. This is based on the classical concept of the gene, according to which crossing over takes place between gene and not within a gene.
4. In a series of Multiple alleles, wild type is always dominant. Rest of the alleles in the series may exhibit dominance or intermediate phenotypic expression when two alleles are involved in a cross.
5. The cross between two mutant alleles will always produce mutant phenotype (intermediate). Such cross will never produce wild phenotype. In other words, Multiple alleles do not show complementation (Complementation refers to appearance of wild phenotype when two mutants are crossed)

**Examples of Multiple Alleles**

Several cases of multiple alleles are known to occur in both plants and animals. Some well known examples for expression of multiple alleles include,

1. Fur color in Rabbits
2. Wing type in Drosophila
3. Eye colour in Drosophila
4. Self incompatability alleles in Plants
5. ABO Blood group in man
6. **Fur colour in Rabbits**

It is a well known example for multiple alleles

In rabbits, the fur colour is of four types, agouti, chinchilla, Himalayan and albino

1. **Agouti:** 
   * This has full colour, known as wild type
   * This colour is dominant over all the remaining colours
   * Produces agouti colour in F1 and 3:1 in F2 when crossed with any of the other three colours in rabbits.
   * This colour is represented by **C**
2. **Chinchilla:**

* This is lighter than Agouti.
* This colour is dominant over Himalayan and albino
* Produces chinchilla in F1 and 3:1 ratio in F2 when crossed with either Himalayan or albino.
* This colour is represented by **cch**

1. **Himalayan:**

* The main body is white while the tips of ear, feet and tail and snout are coloured
* This colour is dominant over albino
* Produces 3:1 ratio in f2 when crossed with albino.
* This is represented by **ch**

1. **Albino:**

* This has pure white fur colour and is recessive to all other types.
* This is represented by **c**

Thus the fur colour in rabbits can be represented in the order of dominance as,

**Agouti Chinchilla Himalayan Albino**

**(C) ( cch) (ch) (c)**

|  |  |  |
| --- | --- | --- |
| **Cross between** | **Expression in F1** | **Segregation in F2** |
| Agouti x chinchilla | Agouti | 3 Agouti : 1chinchilla |
| Agouti x Himalayan | Agouti | 3 Agouti : 1himalayan |
| Agouti x albino | Agouti | 3 Agouti : 1albino |
| Chinchilla x Himalayan | Chinchilla | 3 Chinchilla : 1Himalayan |
| Chinchilla x albino | Chinchilla | 3 Chinchilla : 1 albino |
| Himalayan x albino | Himalayan | 3 Himalayan : 1albino |

Thus variation in fur colour in rabbits is due to multiple alleles of a single gene **c.**

**Test for Allelism**

Two types of tests are used for allelism Recombination Test and Complementation Test

1. **Recombination Test**

* It was believed that recombination can occur between two genes, but not within a gene.
* Thus, in a cross between two mutants, **m1m1 x m2m2**
* if wild type appears in a test cross or in F2,  then m1 and m2 are considered as non allelic because production of wild type is not possible without recombination.
* If no wild type appears in test cross or F2, then m1 and m2 are considered as non allelic forms.
* Since intragenic recombination has been reported in many organisms, this concept is no more valid.

1. **Complementation Test**
   * Alleles may be arranged in two ways, **Cis position and trans position**
   * When two wild alleles are located in one chromosome and their mutant alleles in homologous chromosomes (++/ab), it is known as **Cis arrangement**
   * **Thus, in Cis position alleles are linked in coupling phase.**
   * When one wild and one mutant type alleles are located in each homologous chromosome (+a/+b), it is known as **trans position or repulsion phase** of alleles.
   * Complementation test is used to determine whether two mutant alleles belong to same gene or two different genes.
   * Complementation refers to appearance of wild phenotype when two mutants are crossed..
   * If there is complementation, the mutants are located in different genes, otherwise they are located in the same gene.

**11. PSEUDOALLELES, PLEIOTROPUAND MODIFYING FACTORS**

The designation of a gene is only a matter of convenience and it indicates only the prominent effect produced by the gene. It does not give a complete picture of the effects produced by the gene. A single gene may sometimes affect more than one characteristic of the organism.

**Pleiotropism**

In cotton, the Punjab hairy lintless gene, lic produces seeds which are without lint. This gene also causes incomplete laciniation of the leaf, reduction in the number and length of internodes and reduction in boll size and fertility.

When a gene causes changes in two or more parts or characters that are not obviously related, the gene is called a pleiotropic gene.

Multiple or manifold phenotypic expression of a single gene is called pleiotropism.

The recessive gene for vestigial wing in its homozygous condition (**II**) also affects by bringing about modification in the hind wings or balancers (halters), certain bristles, structure of the reproductive organs besides lowering egg production and reducing longevity in Drosophila.

The erectoides locus (ert) in barley causes short stature, reduced internodal length and dense ears.

In plants a gene may produce a red pigment in several organs such as flowers, stems and leaves but still it is not quite correct to say that the gene is pleiotropic because the gene has only one general effect, the production of pigment.

**Pseudoalleles**

Trans arrangement

Cis arrangement

Pseudoalleles are non alleles so closely linked as often inherited as one gene, but shown to be separate by cross over studies.

One of the first demonstration of pseudoallelic condition was that of star-asteroid analysed by Lewis in 1951. He found a recessive mutation in Drosophila producing a small rough eye when homozygous. It was at locus 1.3 in the second chromosomes. This was also the identical location of gene star, a dominant mutation also affecting the morphology of the eye. The eye was rough and had a slight gleam, hence the name star, with gene symbol **S**. In crosses among these flies, recombination between star and asteriod occurred at a low frequency of one in five thousand.

The apricot and eiosin which were considered to be two alleles of a multiple allelic series affecting eye colour in Drosophila were laster proved to be two separate alleles located very close together on the same chromosome and found to be pseudoalleles in which crossing over occurred at a rate of 10-4 or one in 10,000.

Pseudoallelic effects are found in Drosophila, corn, cotton, Aspergillus, Neurospora, bacteria and in viruses.

**Isoalleles**

Usually wild type allele (represented as +) is dominant over its recessive allele. In some natural populations, different wild type alleles affecting the same character were found and these wild type alleles had similar allelic dominance or they may differ in their degree of expression that could be detected in special combinations. Such alleles are called isoalleles.

Timofeev - Ressovsky and Muller found that the wild type Drosophila from different natural populations had different dominant (red eye) alleles as judged by their stability or by their different effects in combinations. Stern found three different wild type alleles of another Drosophila mutant, cubitus interruptus, which showed different degrees of dominance over the same mutant allele. He called such alleles as isoalleles because they were alike in their homozygous effect and their differences appeared only in special combinations.

**Phenocopy**

We know that phenotype denotes the external expression of a genotype in a particular environment. Normally the phenotypic expression such as the shape of seed (round or wrinkled), colour of flower (red or white) etc. may not vary in different environments due to their adaptedness which is the result of natural selection of the genotype for over a long period of time in the course of evolution.

However differences in the phenotypic expression of a particular genotype have been observed and certain of such altered phenotypes are similar to some of the naturally occurring phenotypes. They are called phenocopies.

Generally the body colour of the fruit fly, Drosophila melanogaster is light brown. A naturally occurring variant resulting out of mutation, a sudden discrete and hereditary variation with yellow body colour was found by Morgan in 1910. These yellow fruit flies on mating with other yellow flies produced yellow offspring only, denoting that yellow body colour is hereditary.

Rappoport in 1939 found that when the larvae of the normal brown bodied fruit flies were reared on food with silver salts, the emerging adults had yellow body. They were genotypically brown, but phenotypically yellow because of the changed environment, the environment in this case being changed food constituent. These phenotypically yellow, but genotypically brown flies were phenocopies of the naturally occurring yellow bodied mutants.

These phenocopies when their larvae are fed with food without silver salts produce only brown bodied adults as their genotype is that of brown body. Thus a phenocopy can last only for that generation in which the environment that induces the change is present.

The phenomenon of phenocopy is taken advantage of in correcting certain hereditary diseases in human beings such as diabetes, myopia etc.

**Modifying genes**

A quantitative character is determined by a very large number of genes whose individual effects, though small, are almost equal to one another. There are, however, instances where a certain character is determined fundamentally by one gene but is modified slightly by other genes. The main or major gene gives expression to a charcter and the minor or modifying genes slightly alter the degree of expression of this character. A modifying gene is one that alters the expression of a major gene but has no effect on the allele of the major gene. The modifiers have very similar but individually small effects and are usually present in such large numbers that they cannot be individually identified.

In the Guernsey breed of dairy cattle, the `solid' colour (fawn i.e., light yellowish brown) of the coat is due to dominant gene **S** and the `spotted' coat (white spotting) is due to its recessive allele **s**. A number of these modifying genes influences the intensity of spotting. If a large number of these modifying genes is present in animals with **ss**, the animals are highly `spotted'. If only a small number of these modifying genes is present in animals with **ss,** they are medium `spotted'. If the modifying genes are absent, animals with **ss** have only few `spots'. These modifying genes have no effect in the presence of the gene for `solid' colour and animals with **SS** or **Ss** have solid-coloured coats irrespective of the number of modifyig genes present.

In Gossypium barbadense the presence of petal spot is due to a gene **S** and the absence of petal spot is due to its recessive allele **s**. A number of modifying genes increases the intensity of colour in the presence of the gene **S**.

**12. EXPRESSIVITY AND LETHAL GENES**

**Penetrance and Expressivity**

The ability of a gene or gene combination to be expressed phenotypically to any degree is called penetrance.

The gene form of ripe seeds in garden pea produces round seeds if homozygous dominant (**RR**) or heterozygous (**Rr**) and wrinkled seeds if homozygous recessive (**rr**). Genes which always produce the expected phenotype are said to possess complete penetrance.

In a few instances, some of the genes fail to produce complete phenotypic expression in the homozygous and heterozygous conditions and such incompletely penetrant genes may find expression in some individuals but not in others.

In the case of polydactyly in man where one extra finger is present in the palm or foot, the heterozygous condition (**Pp**) brings forth polydactyly in some and normal condition in others. Huntington's Chorea, a human syndrome and taste sensitivity for Phenyl thio-carbomide (PTC) in human beings are some of the examples for incomplete penetrance of a gene.

The degree of effect produced by a penetrant genotype is called expressivity. The expressivity or the phenotype effect may be fairly constant atleast in the normal environment. But it is influenced by change in environmental conditions.

Change of temperature, nutrition, hormone deficiency etc., influence the expressivity of the curly wing in *Drosophila*.

**Lethal genes**

When seeds from self-pollinated maize plants are sown, sometimes green seedlings and white seedlings (i.e., albinos) emerge in a ratio of approximately 3 green : 1 white. The albinos die within a few says after germination and only green plants remain. One-third of these green plants, on selfing, produce offspring, all of which are green but two-thirds of them on selfing produce offspring, some of which are green and others are white.

This peculiar behaviour can be explained as follows: Maize being a highly cross-pollinated crop is likely to be heterozygous. If the gene for chlorophyll is represented as **W** and its recessive allele for albinism as **w**, the plant is likely to have the genotype **Ww**. When this is selfed, homozygous recessives appear and these die.

|  |  |  |  |
| --- | --- | --- | --- |
| Green plants **Ww** | | | |
| Selfed | | | |
| **WW** | **Ww** | **Ww** | **Ww** |
| Green | Green | Green | White (die) |
| Selfed | Selfed | Selfed |  |
| Green only | Green and white | Green and white |  |

Lethal genes are genes which in the homozygous state have such a marked deleterious effect that such homozygous organisms are inviable.

Seedlings of genotype **ww** always die after a short time in the field because they have no chlorophyll which is absolutely essential for plants.

Albinos are with genotype **ww** and die out in the seedling stage itself but still the recessive allele for albinism is carried in green plants with genotype **Ww**. Whenever the heterozygous green plants are selfed the albinos appear again.

An example of lethal genes which cause death of the embryos in a very early stage can be cited in mice.

Yellow mice do not breed true. When mated among themselves, they produce yellow and black individuals. Yellow mice may therefore be considered as heterozygous.

Black mice, when inbred, produce only black progeny. Black may therefore be considered as recessive to yellow.

When yellow mice are mated with black, the progeny consists of 1 yellow : 1 black, confirming that all the yellow mice are heterozygous.

Mating between two yellow mice is therefore expected to produce a progeny of 3 yellow : 1 black, but actually the progeny consists of 2 yellow : 1 black.

The progeny of a cross between two yellow mice should consist of 1 homozygous yellow, 2 heterozygous yellow and 1 black but only the two latter classes are born in the proportion of 2 yellow: 1 black.

Operations on pregnant mice have disclosed that approxiamtely one-fourth of the embryos die very early and disintegrate in the uterus. The observation that the actual numbers of individuals born from mating between yellow mice are less than the number of individuals born from mating between yellow and black by about one-fourth confirms the hypothesis.

The homozygous yellow is therefore assumed to be inviable, i.e., the homozygous yellow has a lethal effect on the embryo.

The gene for yellow **Yv** has a lethal effect in addition to a dominant effect on colour whereas its allele for black **Yv** has a dominant viability effect in addition to a recessive effect on colour.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
|  | Yellow | X | Yellow |  |
|  | **Yvyv** |  | **Yvyv** |  |
| **YvYv**  (die) | **Yvyv**  yellow |  | **Yvyv**  Yellow | **yvyv**  black |

Lethality is thus a condition in which death of a certain genotype occur prematurely.

Dominant lethal allele kills the carrier individual both in the homozygous and / or heterozygous conditions. Individuals with dominant lethal alleles die before they can produce their progeny. Therefore such an allele is removed from the population in the same generation in which it arose.

Recessive lethal allele kills the carrier individual only in the homozygous condition. It may have no obvious phenotypic effect in the heterozygote or may exhibit a distinct phenotype when in heterozygous condition.

Complete lethal genes cause death of the zygote, developing embryo, fully developed embryo or the fully developed organism before reproductive stage as in the case of coat colour in mice, seedling colour (albino) in maize, snapdragon and many other plants, human syndromes such as Sickle cell anaemia, congenital ichthyosis and amaurotic idiocy.

In many cases, lethal genes become operative at the time when the organism becomes sexually mature. Such lethal genes which handicap but do not destroy their possessor are called sublethal or semilethal or subvital genes. Human syndromes such as retinoblastoma, epiloia, huntington's chorea etc. are sublethal in effect.

According to the stage at which lethal effect is expressed. Lethality may be zygotic, gametic or gmetophytic.

**13.**  **MULTIPLE FACTOR INHERITANCE**

The inheritance of many of the differentiating characters of plants and animals, for example, yellow and green colour of cotyledons in pea, brown and white colour of grains in Sorghum, can be studied easily because the individuals can be separated into sharply distinct classes by mere observation without resorting to any scale of measurements. Colour of cotyledons in peas, colour of grains inSorghum, etc., are examples of qualitative characters that show discontinuous variation and are governed by one or two major genes or oligogenes. An understanding of the inheritance of characters like length of ear in corn, yield of grain in rice, yield of milk in dairy cattle is not so easy because the individuals show subtle differences and can be separated into classes only after making measurements. These characters are examples of quantitative characters that show more or less continuous variation and are governed by a large number of genes called multiple genes or multiple factors or polymeric or polygenes.

**Nilsson-Ehle's studies on kernel colour in wheat**

The Swedish geneticist Nilsson-Ehle (1908) effected crosses between different true breeding strains of wheat with red kernels and those with white kernels. In some crosses of red with white, a ratio of 3 red : 1 white was found among the F2, indicating a single gene difference. Careful examination however revealed that the red colour of the F1 was not as intense as the red colour of the parent and that in the F2, some red grains were as dark as those of the parent and others only as dark as those of the F1.

In some other crosses, a ratio of 15 red : 1 white was found in the F2, indicating that there are two pairs of genes for red colour and that either or both of these can produce red kernels. Careful examination revealed that all the red kernels were not of the same intensity of colour. It was possible to separate the F2 into the following classes:

|  |  |
| --- | --- |
| Dark red | 1 |
| Medium dark red | 4 |
| Medium red | 6 |
| Light red | 4 |
| White | 1 |

It is evident that red colour is due to two pairs of alleles. Each gene is capable of producing red colour. Each is incompletely dominant over white and is cumulative in its effect. The intensity of the red colour depends upon the number of colour producing genes present. Dark red is due to the presence of four contributing genes for red, medium dark red to three contributing genes, medium red to two contributing genes and light red to one contributing gene.

The genotypes of the F2 together with their phenotypes are given below:

Table 7-1. The F2 ratio in wheat

|  |  |  |
| --- | --- | --- |
| Genotype | Genotypic ratio | Phenotype |
| **R**1 **R1 R** 2 **R**2 | 1 | Dark red |
| **R**1 **R1 R** 2 **r**2 | 2 | Medium dark red |
| **R**1 **r1 R** 2 **R**2 | 2 | Medium dark red |
| **R**1 **r1 R** 2 **r**2 | 4 | Medium red |
| **R**1 **R1 r** 2 **r**2 | 1 | Medium red |
| **R**1 **r1 R** 2 **R**2 | 1 | Medium red |
| **R**1 **r1 r** 2 **r**2 | 2 | Light red |
| **R**1 **r1 R** 2 **r**2 | 2 | Light red |
| **R**1 **r1 r** 2 **r**2 | 1 | White |

In still other crosses, Nilsson-Ehle found a ratio of 63 red : 1 white in the F2, a segregation which suggested that three independent pairs of alleles were involved. If the red parent is represented by **R**1 **R1 R** 2 **R**2 **R**3 **R3**  and the white by **r**1 **r1 r** 2 **r**2 **r**3 **r3,** the F1, which was essentially uniform but intermediate between the parents in colour, can be represented by **R**1 **r1 R** 2 **r**2 **R**3 **r3**. In the F3 there was a marked increase in the range of colour types. About 1 in 64 of the F2 was with very deep red kernels and has 6 contributing genes for red ; 6 with deep red kernels have 5 contributing genes; 15 with dark red kernels have 4 contributing genes; 20 with medium dark red kernels have 3 contributing genes; 15 with medium red kernels have 2 contributing genes; 6 with light red kernels have 1 contributing gene and 1 in 64 was with white kernels and has no contributing genes for the red colour. It was difficult to distinguish these differences in colour as there was a more or less continuous variation among the F2.

From these studies, Nilsson-Ehle proposed the multiple factor hypothesis for the inheritance of quantitative characters. This assumes that there is a series of independent genes for a given quantitative trait. Dominance is usually incomplete but these genes are cumulative or additive in their effect. Each gene adds something to the strength of expression of the character whereas its allele does not possess any effect. The F1 is essentially uniform but intermediate between the two parents. The F2 shows considerable variability but is intermediate between the two parents, the F2 mean value being approximately equal to the parental mean and also, the F1 mean.

**Hypothetical example of quantitative inheritance**

Let us suppose that one true-breeding tall plant with a height of 200 cm. has the genotype **T**1 **T1 T**2 **T**2 and a true-breeding short plant with a height of 100 cm. has the genotype **t**1 **t1 t** 2 **t**2. Let us also suppose that the environment is so uniform that it is not responsible for variation in height. Let us further suppose that except for the difference in the two loci (i.e.,**T**1/**t**1 and **T**2/**t**2), the two plants have the same genotype which is responsible for a plant height 100 cm. The difference in height of 100 cm. between the two plants is due to the four duplicate, cumulative, incompletely dominant genes designated by capital letters, **T**1, **T1, T** 2and **T**2 (called contributing or active genes), each gene adding 25 cm to the height of the plant. The alleles designated by small letters **t**1, **t1 t**2 and **t**2 (called neutral or inert alleles), do not in any manner influence the height of the plant.

The F1 hybrid would be **T**1 **t1 T** 2 **t**2. As the two contributing genes **T**1  and **T2** adds 25 cm. each to the residual heredity of 100 cm, the F1 would be 150 cm. high, exactly intermediate between the parents. The F2 would segregate for plant height and hence would exhibit considerable variability in height.

**The F2 from a cross between plants differing in height**

|  |  |  |
| --- | --- | --- |
| Genotype | Genotypic ratio | Phenotype |
| **T**1 **T1 T**2 **T**2 | 1 | 200 cm. |
| **T**1 **T1 T**2 **t**2 | 2 | 175 cm. |
| **T**1 **t1 T**2 **T**2 | 2 | 175 cm. |
| **T**1 **t1 T**2 **t**2 | 4 | 150 cm. |
| **T**1 **T1 t**2 **t**2 | 1 | 150 cm. |
| **T**1 **t1 T**2 **T**2 | 1 | 150 cm. |
| **T**1 **t1 t**2 **t**2 | 2 | 125 cm. |
| **T**1 **t1 T**2 **t**2 | 2 | 125 cm. |
| **T**1 **t1 t**2 **t**2 | 1 | 100 cm. |

The frequency of each class in the F2 would be as follows:

**Frequency distribution in the F2**

|  |  |  |
| --- | --- | --- |
| Plant height | Frequency | No. of active alleles |
| 200 cm. | 1 | 4 |
| 175 cm. | 4 | 3 |
| 150 cm. | 6 | 2 |
| 125 cm. | 4 | 1 |
| 100 cm. | 1 | 0 |

The mean height of the F2 plants would be 150 cm., which is equal to the parental mean and also, the F1 mean.

If instead of four contributing genes, a very large number of genes, each with a very small individual influence, are assumed to be responsible for plant height, the expected hereditary difference between two successive classes in the F2 is likely to be smaller than even the difference normally due to environment. Where these class differences are very small, the variation in the F2 population would appear to be continuous and would be typical of quantitative inheritance.

The features of inheritance of quantitative characters are the following:

The individuals of each homozygous parental line have the same genotype and therefore the two lines to which the parents belong would show very little variability within themselves. The phenotypic differences between individuals within a parental line are only due to environment.

All the individuals of the F1 have the same genotype and, therefore, the F1 as a whole, would show very little variability. The F1 would, hoever, be intermediate between the parents, the mean of the F1 being equal to the mean of the two parental values. The phenotypic differences between individuals in the F1 population are only due to environment.

The F2 would exhibit considerable variability. Some of the F2 values would overlap with the values of one parent, some other F2 values would overlap with the values of the other parent and a large number of the F2 values would be intermediate between the values of the two parents. This variation in the F2 is more or less continuous and is largely due to differences in genotype between individuals of the F2. The F2 mean would however be equal to the F1 mean and also, the parental mean.

Quantitative characters are controlled by the joint action of a very large number of multiple genes which cannot be distinguished from one another because their individual effects on the phenotype are insignificant in comparison with the fluctuations due to the environment. Multiple genes are usually incompletely dominant duplicate genes with cumulative effects.

**Studies on ear length in corn**

Emerson and East (1913) crossed a long eared sweet corn plant from a line having a mean ear length of 16.80 cm. with a short-eared popcorn plant from a line having a mean ear length of 6.63 cm. Within each parental line there was some variability in ear length. As each parental line was homozygous for genes affecting ear length, this variability could only be due to environment.

The F1 mean was 12.12 cm. which was intermediate between the two parental lines. The F1 was however, uniform and the small variation around the mean exhibited by the F1 individuals could be attributed only to the environment as all the F1 plants were typically alike.

In the F2 there were plants with ears as long as the longer parent and as short as the shorter parent. The number of these extreme types was small, a relatively large number of the F2 being intermediate in their ear length.

The increase in variability in the F2 was due to genetic segregation and recombination and it was not because the F2 was more susceptible to environmental influence than the parental lines or the F1.

The mean length of ear of the F2 was 12.89 cm. This is almost equal to the F1 mean of 12.12 cm. and was approximately intermediate between the mean of the long-eared parent (16.80 cm.) and the mean of the short-eared parent (6.63 cm).

The facts that the F1 was uniform but intermediate between the parents and that the F2 exhibited continuous variation but was equal in its mean length of ear to the F1 mean and the parental mean suggest that length of ear is a quantitative character governed by a number of incompletely dominant genes which have effects similar to one another and which supplement each other.