CLASS NOTES FOR SEMESTER –IV STUDENTS, Date- 6.4.2020

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B.Sc (HONOURS) MICROBIOLOGY (CBCS STRUCTURE)
C-8: MICROBIAL GENETICS (THEORY)
SEMESTER –IV

TOTAL HOURS: 60 **CREDITS: 4**

Unit 1 Mendelian Principles **No. of Hours: 12**

Mendel's Laws: Dominance, segregation, independent assortment, deviation from Mendelian inheritance, Rediscovery of Mendel's principles, Chromosome theory of inheritance: Allele, multiple alleles, pseudoallele, complementation tests, Extensions of Mendelian genetics: Allelic interactions, concept of dominance, recessiveness, Incomplete dominance and co-dominance, Multiple alleles, Epistasis, penetrance and expressivity.

Q. Write the Mendel's Postulates and describe.

Ans. Mendel's First Three Postulates

Gregor Johann Mendel, who in 1866 put forward the major postulates of transmission genetics as a result of experiments with the garden pea and thereby laid the foundation of modern genetics,. Using the consistent pattern of results in the monohybrid crosses, Mendel derived the following three postulates, or principles, of inheritance.

1. UNIT FACTORS IN PAIRS

Genetic characters are controlled by unit factors existing in pairs in individual organisms.

In the monohybrid cross involving tall and dwarf stems, a specific unit factor exists for each trait. Each diploid individual receives one factor from each parent. Because the factors occur in pairs, three combinations are possible: two factors for tall stems, two factors for dwarf stems, or one of each factor. Every individual possesses one of these three combinations, which determines stem height.

2. DOMINANCE/RECESSIVENESS

When two unlike unit factors responsible for a single character are present in a single individual, one unit factor is dominant to the other, which is said to be recessive.

In each monohybrid cross, the trait expressed in the F1 generation is controlled by the dominant unit factor. The trait not expressed is controlled by the recessive unit factor. The terms *dominant* and *recessive* are also used to designate traits. In this case, tall stems are said to be dominant over recessive dwarf stems.

3. SEGREGATION

During the formation of gametes, the paired unit factors separate, or segregate, randomly so that each gamete receives one or the other with equal likelihood.

If an individual contains a pair of like unit factors (e.g., both specific for tall), then all its gametes receive one of that same kind of unit factor (in this case, tall). If an individual contains unlike unit factors (e.g., one for tall and one for dwarf), then each gamete has a 50 percent probability of receiving either the tall or the dwarf unit factor.

Q. Briefly describe the Rediscovery of Mendelís work

Mendel's conclusions were largely ignored. Although they were not completely unknown to biologists of the time, they were not seen as generally applicable, even by Mendel himself, who thought they only applied to certain categories of species or traits. A major block to understanding their significance was the importance attached by 19 th century biologists to the apparent blending of inherited traits in the overall appearance of the progeny, now known to be due to multigene interactions, in contrast to the organ-specific binary characters studied by Mendel. In 1900, however, his work was "re-discovered" by three European scientists, Hugo de Vries, Carl Correns, and Erich von Tschermak.

The exact nature of the "re-discovery" has been somewhat debated: De Vries published first on the subject, mentioning Mendel in a footnote, while Correns pointed out Mendel's priority after having read De Vries's paper and realizing that he himself did not have priority. De Vries may not have acknowledged truthfully how much of his knowledge of the laws came from his own work, or came only after reading Mendel's paper. Later scholars have accused Von Tschermak of not truly understanding the results at all. Regardless, the "re-discovery" made Mendelism an

important but controversial theory. Its most vigorous promoter in Europe was William Bateson, who coined the term "genetics", "gene", and "allele" to describe many of its tenets. The model of heredity was highly contested by other biologists because it implied that heredity was discontinuous, in opposition to the apparently continuous variation observable for many traits. Many biologists also dismissed the theory because they were not sure it would apply to all species, and there seemed to be very few true Mendelian characters in nature. However, later work by biologists and statisticians such as R.A. Fisher showed that if multiple Mendelian factors were involved in the expression of an individual trait, they could produce the diverse results observed. Thomas Hunt Morgan and his assistants later integrated the theoretical model of Mendel with the chromosome theory of inheritance, in which the chromosomes of cells were thought to hold the actual hereditary material, and create what is now known as classical genetics, which was extremely successful and cemented Mendel's place in history.

Mendel's Laws of Inheritance

Mendel postulated three laws, which are now called after his name as Mendel's laws of heredity. These are:

1. Law of dominance and recessive
2. Law of segregation
3. Law of independent assortment

1. Law of Dominance

Definition: When two homozygous individuals with one or more sets of contrasting characters are crossed, the characters that appear in the F_1 hybrids are dominant characters and those do not appear in F_1 are recessive characters.

Explanation: The dominance and recessive of genes can be explained on the basis of enzymatic functions of genes. The dominant genes - are capable of synthesizing active polypeptides or proteins that form functional enzymes, whereas the recessive genes (mutant genes) code for incomplete or non-functional polypeptides. Therefore, the dominant genes produce a specific phenotype while the recessive genes fail to do so. In the heterozygous condition also the dominant gene is able to express itself, so that the heterozygous and homozygous individuals have similar phenotype.

Critical appreciation of Law of Dominance

Scientists conducted cross-breeding experiments to find out the applicability of law of dominance. The experiments were conducted by Correns on peas and maize, Tschermak on peas, by De Vries on maize etc., by Bateson and his collaborators on a variety of organisms, by Davenport on poultry, by Furst on rabbits, by Toyama on silk moth and by many others. These scientists observed that a large number of characters in various organisms are related as dominant and recessive.

Importance of law of dominance

The phenomenon of dominance is of practical importance as the harmful recessive characters are masked by the normal dominant characters in the hybrids. In Human beings a form of idiocy, diabetes, haemophilia etc. are recessive characters. A person hybrid for all these characteristics appears perfectly normal. Thus harmful recessive genes can exist for several generations without expressing themselves. **Exceptions to Law of Dominance is the Incomplete Dominance.** After Mendel several cases were recorded by scientists, where F_1 hybrids exhibited a blending of characters of two parents. These hybrids were found to be midway between the two parents. This is known as incomplete dominance or blending inheritance. It means that two genes of the allelomorphic pair are not related as dominant and recessive, but each of them expresses itself partially. As for example, in four-o'clock plant, *Mirabilis jalapa,* when plants with red flowers (RR) are crossed with plants having white flowers (rr), the hybrid F_1 plants (Rr) bear pink flowers. When these F_1 plants with pink flowers are self-pollinated they develop red (RR), pink (Rr) and white (IT) flowered plants in the ratio of 1 : 2 : 1 (F_2 generation).

2. Law of Segregation (Purity of Gametes)

Explanation - The law of segregation states that when a pair of contrasting factors or genes or allelomorphs are brought together in a heterozygote (hybrid) the two members of the allelic pair remain together without being contaminated and when gametes are formed from the hybrid, the two separate out from each other and only one enters each gamete.

Example - Pure tall plants are homozygous and, therefore/possess genes (factors) TT; similarly dwarf possess genes tt. The tallness and dwarfness are two independent but contrasting factors or determiners. Pure tall plants produce gametes all of which possess gene T and dwarf plants t type of gametes. During cross fertilization gametes with T and t unite to produce hybrids of F_1 generation. These hybrids possess genotype Tt. It means F_1 plants, though tall phenotypically, possess one gene for tallness and one gene for dwarfness. Apparently, the tall and dwarf characters appear to have become contaminated developing only tall character. But at the time of gamete formation, the genes T (for tallness) and t (for dwarfness) separate and are passed on to separate gametes. As a result, two types of gametes are produced from the heterozygote in equal numerosity. 50% of the gametes possess gene T and other 50% possess gene t. Therefore, these gametes are either pure for tallness or for dwarfness. (This is why the law of segregation is also described as Law of purity of gametes). Gametes unite at random and when gametes are numerous all possible combinations can occur, with the result that tall and dwarf appear in the ratio of 3 :1.

3. Law of Independent Assortment

Definition: The inheritance of more than one pair of characters (two pairs or more) is studied simultaneously, the factors or genes for each pair of characters assort out independently of the other pairs. Mendel formulated this law from the results of a dihybrid cross.

Explanation: The cross was made between plants having yellow and round cotyledons and plants having green and wrinkled cotyledons.

Q. Give an example of Mendel's Laws

These postulates provide a suitable explanation for the results of the monohybrid crosses. Let's use the tall/dwarf cross to illustrate. Mendel reasoned that P1 tall plants contained identical paired unit factors, as did the P1 dwarf plants. The gametes of tall plants all receive one tall unit factor as a result of **segregation**. Similarly, the gametes of dwarf plants all receive one dwarf unit factor. Following fertilization, all F1 plants receive one unit factor from each parent—a tall factor from one and a dwarf factor from the other—reestablishing the paired relationship, but because tall is dominant to dwarf, all F1 plants are tall. When F1 plants form gametes, the postulate of segregation demands that each gamete randomly receives either the tall *or* dwarf unit factor. Following random fertilization events during F1 selfing, four F2 combinations will result with equal frequency:

1. tall/tall

2. tall/dwarf

3. dwarf/tall

4. dwarf/dwarf

Combinations (1) and (4) will clearly result in tall and dwarf plants, respectively. According to the postulate of dominance/ recessiveness, combinations (2) and (3) will both yield tall plants. Therefore, the F2 is predicted to consist of 3/4 tall and 1/4 dwarf, or a ratio of 3:1. This is approximately what Mendel observed in his cross between tall and dwarf plants.

Q. What is Phenotype and Genotype?

Phenotype

A **phenotype** may be any measurable characteristic or distinctive trait possessed by an organism. The trait may be visible 10 the eye, such as the color of a flower or the texture of hair, or it may require special tests for its identification, as In the determination of the respiratory quotient or the serological test for blood type. The phenotype is the result of gene products brought to expression in a given environment.

Example 2.1, Rabbits, of the Himalayan breed in the usual range of environments develop black pigment at the tips of the nose. tail. feet, and cars. If raised at very high temperatures, an all

white rabbit is produced. The gene for Himalayan color pattern specifics a temperature sensitive enzyme that is inactivated at high temperature, resulting in a Joss of pigmental ton.

Genotype

All of the genes possessed by an individual constitute its **genotype.**

Q. **Define Dominant and Recessive Alleles. Carriers. Codominant Alleles and Lethal Alleles Or deviation from Mendelian inheritance**

Dominant and Recessive Alleles

Whenever one of a pair of alleles can come to phenotypic expression only in a homozygous genotype, we call that allele a recessive factor. The allele that can phenotypically express itself in the heterozygote as well as in the homozygote is called a **dominant** factor.

Carriers

Recessive alleles (such as the one for albinism) are often deleterious to those who possess them in duplicate (homnzygous recessive genotype). A heterozygote may appear just as normal as the homozygous dominant genotype. A heterozygous individual who possesses a deleterious recessive allele hidden from phenotypic expression by the dominant normal allele is called a **carrier.** Most of the deleterious alleles harbored by a population are found in carrier individuals.

Codominant Alleles (Deviation)

Alleles that lack dominant and recessive relationships may be called incompletely dominant, partially dominant, semidominam or codominanl. This means that each allele is capable of some degree of expression when in the heterozygous condition. Hence the heterozygous genotype gives rise to a phenotype distinctly different from either of the homozygous genotypes. Usually the heterozygous phenotype resulting from codominance is intermediate in character between those produced by the homozygous genotypes; hence the erroneous concept of "blending."The phenotype may appear lobe a "blend" in heterozygotes, but the alleles maintain their individual identities and will segregate from each other in the formation of gametes.

Example : The alleles governing the M-N blood group system in humans are codominanis and may be represented by the symbols L^M and L^N. the base letter (£.) being assigned in honor of its discoverers (Landstciner and Lcvine). Two antiscra (anti-M and anti-N) arc used to distinguish

three genotypes and their corresponding phenotypes (blood groups). Agglutination is represented by + and nonagglutinution by -.

Genotype	Reaction with:		Blood Group (Phenotype)
	Anti-M	Antli-N	
L^ML^M	+	_	M
L^ML^N	+	+	MN
L^NL^N	_	+	N

Incomplete dominance(Deviation)

Mendel always observed complete dominance of one allele over the other for all the seven characters, which he studied, in garden pea. Later on cases of incomplete dominance were reported. For example, in four ëoí clock plant (Mirabilis jalapa) there are two types of flower viz., red and white. A cross between red and white flowered plants produced plants with intermediate flower colour i.e. pink colour in F1 and a modified ratio of 1 red: 2 pink: 1 White in F2.

Lethal Alleles (Deviation)

The phenotypic manifestation of some genes is the death of the individual in either the prenatal or postnatal period prior to maturity. Such factors are called lethal genes. A fully dominant lethal allele (i.e., one that kills in both the homozygous and heterozygous conditions) occasionally arises by mutation from a normal allele. Individuals with a dominant lethal die before they can leave progeny. Therefore the mutant dominant lethal is removed from the population in the same generation in which it arose. Lethals that kill only when homozygous may be of two kinds: (I) one that has no obvious phenotypic effect in heterozygotes. and (2) one that exhibits a distinctive phenotype when heterozygous.

Example: The amount of chlorophyll in snapdragons *(Antirrhinum)* is controlled by a pair of codominant alleles, one of which exhibits a lethal effect when homozygous, and a distinctive color phenotype when heterozygous.

Genotype	Phenotype
C^1C^1	Green (normal)
C^1C^2	Pale green
C^2C^2	White (lethal)

Penetrance and Expressivity

Differences in environmental conditions or in genetic backgrounds may cause individuals that are genetically identical at a particular locus to exhibit different phenotypes. The percentage of individuals with a particular gene combination that exhibits the corresponding character to any degree represents the penetrance of the trait.

Example: In some families, extra fingers and/or toes (polydactyly) in humans is thought IO be produced by a dominant gene *{P).* The normal condition with five digits on each limb is produced by the recessive genotype (pp). Some individuals of genotype ***Pp*** are not polydactylous, and therefore the gene has a penetrance of less than 100%.

A trail, although penetrant, may be quite variable in its expression. The degree of effect produced **By a penetrant genotype is termed expressivity.**

Example: The polydactylous condition may be penetrant in the left hand (6 fingers) and not in the right (5 fingers), or it may be penetrant in the feel and not in the hands.

A recessive lethal gene that lacks complete penetrance and expressivity will kill less than 100% of the homozygotes before sexual maturity. The terms semilethal or subvital apply to such genes. The effects that various kinds of lethals have on the reproduction of the next generation form a broad speclrum from complete lethality to sterility in completely viable genotypes. Problems in this book, however, will consider only those lethals that become completely penetrant, usually during the embryonic stage. Genes other than lethals will likewise be assumed completely penetrant.

Multiple Alleles

The genetic systems proposed thus far have been limited lo a single pair of alleles. The maximum number of alleles at a gene locus that any individual possesses is 2, with 1 on each of the homologous chromosomes. But since a gene can be changed to alternative forms by the process of mutation, a large number of alleles is theoretically possible in a population of individuals. Whenever more than two alleles are identified at a gene locus, we have a multiple allelic series.

Q. **What do you mean by Testcross and Back Cross?**

Testcross

If a homozygous dominant genotype has the same phenotype as the heterozygous genotype, a **Testcross** is required to distinguish between them. The testcross parent is always homozygous recessive for **all** of the genes under consideration. The purpose of a testcross is to discover how many different kinds of gametes are being produced by the individual whose genotype is in question. A homozygous dominant individual will produce only one kind of gamete; a **monohybrid** individual (heterozygous at one locus) produces two kinds of gametes with equal frequency.

Because some alleles are dominant over others, the phenotype of an organism does not always reflect its genotype. A recessive phenotype (yellow) is only expressed with the organism is homozygous recessive (gg). A pea plant with green pods may be either homozygous dominant (GG) or heterozygous (Gg). So testcross is more important to determine whether an organism with a dominant phenotype (e.g. green pod color) is homozygous dominant or heterozygous.

The breeding of an organism of unknown genotype with a homozygous recessive. If all the progeny of the testcross have green pods, then the green pod parent was probably homozygous dominant since a GG x gg cross produces Gg progeny. If the progeny of the testcross contains both green and yellow phenotypes, then the green pod parent was heterozygous since a Gg x gg cross produces Gg and gg progeny in a 1:1 ratio. The testcross was devised by Mendel and is still an important tool in genetic studies.

Backcross

If the F, progeny are mated back to one of their parents (or to individuals with a genotype identical to that of their parents) the mating is termed backcross. Sometimes "backcross" is used synonymously with "testcross" in genetic literature.

Backcrossing is a crossing of a hybrid with one of its parents or an individual genetically similar to its parent, in order to achieve offspring with a genetic identity which is closer to that of the parent.

Q. **What is epistatic? Explain the epistatic with example.**

Originally a gene or locus that suppressed or masked the action of a gene at another locus was termed **epistatic.** The gene or locus suppressed was hypostatic. Later it was found that both loci could be mutually epistatic to one another. Now the term "epistasis" has come to be synonymous with almost any type of gene interaction. Dominance involves gene suppression, or the masking effect that one allele has upon the expression of another allele at the same locus. Epistasis involves gene suppression, or the masking effect that one gene locus has upon the expression of another. The classical phenotypic ratio of 9 : 3 : 3 : It was observed in the progeny of dihybrid parents becomes modified by epistasis into ratios that are various combinations of the 9 : 3 :3: 1 groupings.

Example: A particularly illuminating example of gene interaction occurs in white clover. Some strains have a high cyanide content; others have a low cyanide content. Crosses between two strains with low cyanide have produced an F, with a high concentration of cyanide in their leaves. The F? shows a ratio of 9 high cyanide: 7 low cyanide. Cyanide is known to be produced from the substrate cyanogenic glucoside by enzymatic catalysis. One strain of clover has the enzyme but not the substrate. The other strain makes substrate but unable convert it to cyanide.

Referance:

1. *William S. Klug, Concept of Genetics*, Tenth **edition,**
2. *Schavm's outline of* **theory and problems, genetics, third edition, william d. stansfield, Ph.D.** *Emeritus Professor of Biological Sciences California Polytechnic State University at San Luis Obispo.*