KRISHNAGAR ACADEMY

BIOLOGY-

PRINCIPLE OF INHERITANCE &VARIATION

Class -12

Genetics is the branch of biology that deals with the study of heredity and variations (Gk. *genesis*-descent). It deals with the mechanism of transmission of characters (inheritance) as well as variation of characters from parents to offspring. It is concerned with the principles that govern origin or birth of a living being. The term '**genetics**' was given by **William Bateson** in the year 1906.

HEREDITY AND VARIATION

Heredity (heirship or inheritance) is the transmission of genetically based characters from parents to their offsprings. Inheritance is the process by which characters or traits are passed on from parent to progeny, generation after generation. It is the basis of heredity. '**Like begets like**' means the young ones look like their parents. Cat produces cat and not dog. An elephant gives birth only to a baby elephant and not some other animal. A mango seed germinates into a mango tree and not any other plant. Humans give birth to only humans and not to apes. Even the curd bacteria which undergoes hundred of generations each day, continue to produce same type of bacteria and not of any different type. All organisms whether animals, plants or microorganisms, produce their own kind through reproduction. But the offspring are never identical to their parents. Some difference, howsoever small it may be is found in them. This is called **variation** - the degree by which progeny differ from their parients. All human beings share many main characters among themselves, yet they look different in several features like eye colour, hair shape, tongue rolling, skin colour, type of ear lobe, body's height, etc. The individual members of a family too show differences in body features. The same is true for animals too. Dogs, cat, cow or buffalo each one of them have so many breeds and even in the same breed, there are minor differences and variations among individuals.

Early humans knew that causes of variation was hidden in sexual reproduction. They successfully bred domesticated varieties from wild plants and animals through selective crossing and artificial selection. For example chicken is the domesticated form of ancestral wild fowl and the Indian cow (sahiwal of Punjab) is the domesticated form of ancestral wild cow. However, though our ancestors knew about the inheritance of characters and variation but they had very little idea about the scientific basis of these phenomena.

SOME BASIC TERMS USED IN INHERITANCE STUDIES

1. Gene or **Factor.** In modern sense an **inherited factor** that determines a biological character of an organism is called **gene**. This is functional unit of hereditary material. Chemically gene is a segment of DNA and equivalent to factor of Mendel.

2. Allelomorphs or alleles, homozygous and heterozygous. Alleles, the abbreviated form of terms allelomorphs (meaning one form or the other) indicates alternative forms of the same gene. Each character has two determiners called **factors**. If the factors represent the extremes or alternatives of the character, they are called alleles or allelomorphic pair.

For example, in pure tall or pure dwarf plants same allele is duplicated (TT and tt), while in hybrid tall both the alleles are present (Tt). An organism, having two identical alleles is known as **homozygous**. An individual with two different alleles (Tt) will be called **heterozygous**. So, homozygous refers to a pair of same, while heterozygous refers to a pair of different units or elements or alleles. In this case, we mean a pair of same genes and a pair of different genes, thus genes and alleles are interchangeable. However, the term gene can be used for any factor, the term allele is mentioned with reference to another allele. T and t are alleles while selfed or inbred the homozygous organisms will be true breeding.

3. Gene locus. It is the portion or region on chromosome representing a single gene. The alleles of a gene are present on the same gene locus on the homologous chromosomes.

4. Homozygous. The organism in which both the genes of a character are identical is said to be homozygous or genetical pure for that character. It gives rise to offspring having the same character on self breeding e.g., TT (**Homozygous dominant**) or tt (**Homozygous recessive**).

5. Heterozygous. The organism in which both the genes of a character are unlike is said to be heterozygous or **hybrid**. Such organisms do not breed true on self fertilization e.g., Tt.

6. Genotype and Phenotype. The **genotype** is the genetic constitution of an organism. TT, Tt and tt are the genotypes of the organism with reference to these particular pairs of alleles. **Phenotype** expresses the characters of individuals like form, sex, colour and behaviour etc. Phenotype can be read out from individuals by direct observation. On the other hand, genotype can be ascertained from ancestory or progeny of individuals. Phenotype of an organism is the expression or observable structural and functional traits produced due to interaction of genes and environment.

7. Pure Line. Generations of homozygous individuals which produce offsprings of only one type i.e., they breed true for their phenotype and genotype. For example, tall pea plants when produce only tall plants generation after generation on being self pollinated or cross pollinated among themselves. These form a pure line of tall pea plants for 3 or 4 generations.

8. Monohybrid, dihybrid and polyhybrid. When only one allelic pair is considered in cross breeding, it is called **monohybrid cross**. For example, inheritance of tall and dwarf characters is monohybrid cross. Similarly when two allelic pairs are used for crossing, it is called **dihybrid cross**. For example, inheritance of yellow and round seed characters and green wrinkled characters is a dihybrid cross. Involvement of more than two allelic pairs in a cross is called **polyhybrid cross**.

9. Reciprocal cross. The reciprocal crosses involve two crosses concerning the same characteristics, but with reversed sexes. For example, if in first cross A is as the female parent and B as the male parent than in the second or reciprocal cross A will be used as male parent and B as female parent.

10. Genome. Total set of genes (DNA instructions) in the haploid dose of chromosomes and inherited as unit from parents to offspring is called genome.

11. Gene pool. All the genotypes of all organisms in a population form the gene pool.

THE INTERPRETATION OF MENDEL'S RESULTS

We have now laid most of the basis for interpreting Mendel's results. The following principles of inheritance were given by Mendel :

(a) Principle of dominance

(b) Principle of segregation or Purity of gametes

(c) Principle of independent assortment

MENDEL'S EXPERIMENTS

Mendel carried out his experiments on the common garden pea, *Pisum sativum* for seven years (1856-1863), in his monastery garden. He procured seeds of 34 different varieties of peas from the local seedsman and grew them in the garden. He chose garden pea as plant material for his experiments due to the following reasons.

(1) Peas were available in many pure breeding varieties with observable alternative forms for a trait or characteristics.

(2) Pea flowers are bisexual. The structure and shape of flower was suitable for cross pollination.

(3) Peas are self-pollinating as such the possibility of the introduction of outside genetical influences was eliminated. Self fertilization could easily be controlled.

(4) Hybrids resulting from crossing two varieties were perfectly fertile.

(5) Pea plants produce new generation in a reasonable short time and a single plant produces numerous seeds.

(6) Presence of more number of contrasting characters.

Mendel's Experimental Technique Involved Four Steps

(*i*) Selection of parents

(*ii*) Hybridisation

(*iii*) Self-pollination of hybrids

(*iv*) Calculations

Mendel's Monohybrid Cross

- This was a cross dealing with a single pair of contrasting characters.
- Mendel cross-pollinated a pure tall plant with a pure dwarf plant of pea.
- They were called the **parental generation (P)**.
- He used the seeds to generate the first hybrid or first filial generation (F_1).
- The F_1 plants were self-pollinated (selfing).
- The seeds were collected to generate the second filial generation (F_2).
- In the F_1 generation, all plants were found to be tall.

- The F_2 generation had both tall and dwarf plants in the ratio of 3 : 1.
- The trait that appeared in the F_1 hybrid was dominant.

Parents (P): Tall plant (TT) × Dwarf plant (tt)

Gametes: T   t

F_1 generation: Tt (Tall)

Selfing: Tt × Tt

Gametes: T, t   T, t

F_2 generation:

♀ \ ♂	T	t
T	TT (tall)	Tt (tall)
t	Tt (tall)	tt (dwarf)

Tall plants : 3
Dwarf plants : 1
Ratio : 3 : 1 (phenotypic)
1 : 2 : 1 (genotypic)

Conclusions Drawn from Monohybrid Cross

- Based on his observations and conclusions drawn from monohybrid cross, Mendel formulated three laws:
  - (i) *Law of Paired Factors:* This law states that various hereditary characters are controlled by factors and there are two factors for each character.
  - (ii) *Law of Dominance:* This law states that one factor in a pair may express itself and prevent the expression of the other. The factor that expresses itself is known as **dominant** and that which is unexpressed as **recessive**.
  - (iii) *Law of Segregation:* Factors of each character segregate during gamete formation so that each gamete receives only one factor for each character.

Reciprocal Cross: It is a cross involving two groups of individuals where half function as males and half as females and vice versa.

Punnett Square: Punnett square is a graph or a table that has been created to yield a diagram that allows an easy representation of hybridisation data. It is a checkerboard or square, divided into smaller squares where all types of gametes are shown. Each box in the square represents an offspring.

Back Cross

- It is a genetic cross between a hybrid organism and one of the original parental type.
- Results of the back cross depend on the parental type.

  Example: Cross between heterozygous F_1 and homozygous dominant parent.

Parents: BB (Black) × bb (White)

↓

F_1: Bb (Black, heterozygous)

Gametes: Bb (B)(b) × BB (B)(B) (Back cross)

↓

F_2:

♀ \ ♂	B	b
B	BB (Black)	Bb (Black)
B	BB (Black)	Bb (Black)

Phenotype: All black

Genotype:

(*a*) Homozygous black: 50%

(*b*) Heterozygous black: 50%

Test Cross

- It is a special type of back cross which is made between individuals with a dominant phenotype and its homozygous recessive parent to know whether the individual is homozygous or heterozygous.
- If the individual is homozygous dominant, the offspring of the test cross will be 100 per cent dominant.
- If the individual is heterozygous, the offspring will be 50 per cent dominant and 50 per cent recessive.

Example:

Test Cross 1

T T (Tall) × t t (Dwarf)

↓

Gametes: (T)(T) × (t)(t)

↓

♀ \ ♂	T	T
t	Tt (Tall)	Tt (Tall)
t	Tt (Tall)	Tt (Tall)

All tall

Test Cross 2

T t (Tall) × t t (Dwarf)

↓

Gametes: (T)(t) × (t)(t)

↓

♀ \ ♂	T	t
t	Tt (Tall)	tt (Dwarf)
t	Tt (Tall)	tt (Dwarf)

50% tall; 50% dwarf

Mendel's Dihybrid Cross

- A breeding experiment dealing with two characters at the same time is called a dihybrid cross.
- Mendel used two pairs of contrasting characters—he crossed a pea plant having round seeds and yellow cotyledons with a pea plant having wrinkled seeds and green cotyledons.
- All plants in the F_1 generation had round seeds and yellow cotyledons.
- F_1 plants on selfing, produced four types of plants in the F_2 generation in the ratio of 9 : 3 : 3 : 1. These were:
  - Round seeds/yellow cotyledons : 9
  - Round seeds/green cotyledons : 3

- Wrinkled seeds/yellow cotyledons : 3
- Wrinkled seeds/green cotyledons : 1

Parents: Round/Yellow × Green/Wrinkled

RRYY rryy

Gametes: RY ry

↓

F_1 generation: Rr Yy (Round/Yellow)

↓

Selfing

Rr Yy × Rr Yy

Gametes: RY Ry rY ry (for both parents)

↓

♀ \ ♂	RY	Ry	rY	ry
RY	RR YY Round/Yellow	RR Yy Round/Yellow	Rr YY Round/Yellow	Rr Yy Round/Yellow
Ry	RR Yy Round/Yellow	RR yy Round/Green	Rr Yy Round/Yellow	Rr yy Round/Green
rY	Rr YY Round/Yellow	Rr Yy Round/Yellow	rrYY Wrinkled/Yellow	rr Yy Wrinkled/Yellow
ry	Rr Yy Round/Yellow	Rr yy Round/Green	rr Yy Wrinkled/Yellow	rr yy Green/Wrinkled

Law of Independent Assortment

- This law was derived by Mendel after his observation from the dihybrid cross.
- The law states that the alleles of different characters located in different pairs of homologous chromosomes are independent of each other in their segregation during gamete formation and in coming together in the offspring by fertilisation.

Summary of Mendel's Hypothesis

(*i*) Each trait of an organism is controlled by a pair of alleles [Principle of Paired Factors].

(*ii*) In an organism having two unlike alleles for a given trait, one (*i.e.*, the dominant allele) may express and the other (*i.e.*, the recessive allele) may remain unexpressed [Principle of Dominance].

(*iii*) The two alleles of a trait separate during gametogenesis, so that each gamete receives only one allele of each trait [Principle of Segregation].

(*iv*) During gamete formation, the allele pairs of different traits segregate independently of each other [Principle of Independent Assortment].

(*v*) Each allele is transmitted from generation to generation as a discrete unchanging unit.

(*vi*) Each organism inherits one allele for each trait from each parent.

Reasons for Mendel's Success

The following are the main reasons for the success of Mendel's experiments.

(1) His choice of pea plants for his breeding experiments was excellent.

(2) He kept complete records of every cross and applied statistical methods and laws of probability for computing his results. He thus could know the pedigree of the progenies.

(3) He took one or two traits at one time for his experiments. His predecessors usually studied many traits simultaneously that caused confusion and made matters complicated.

(4) His experiments had a large sampling size, which gave greater credibility to the data that he collected.

(5) He was fortunate enough that the characters he chose for his experiments did not show linkage, incomplete dominance, gene interaction, etc.

(6) He took utmost care to check contamination from foreign pollen at the time of hybridization.

(7) He prevented any chances of self pollination by emasculation (removal of anthers) and bagging (covering stigma by a small bag).

(8) He started his experiments with plants having pure line characters.

Practical Applications of Mendel's Laws

The fact that many hereditary traits show dominance and recessiveness is of considerable practical importance. A knowledge of the basic mendelian principles gives us an idea about the new combinations that would appear in the progeny of hybrids and enable us to predict their frequency. This information is of great importance both for plant and animal breeders. New types of plants with new combinations of useful characters can be produced by hybridization.

. Exceptions to the Principles of Mendel

Incomplete Dominance

- It is a phenomenon of neither of the two alleles of a gene being dominant over each other, so that when both of them are present together, a new phenotype is formed.
- This phenotype is intermediate between the independent expression of the two alleles.
- This phenomenon is found in both plants and animals.
- The inheritance of flower colour in snapdragon (*Antirrhinum majus*) and 4 o'clock plant (*Mirabilis jalapa*) are common examples.
- When a cross was made between red-flowered plant and white-flowered plant, the F_1 hybrid was pink.
- When F_1 hybrids were selfed, the F_2 generation consisted of red, pink and white-coloured flower plants.

Parents: RR (Red flower) × rr (White flower)

Gametes: R r

↓

F_1 generation: Rr (Pink flowers)

↓

Selfing

Rr × Rr

Gametes: R r R r

↓

♀ \ ♂	R	r
R	RR (Red)	Rr (Pink)
r	Rr (Pink)	rr (White)

- The phenotypic and genotypic ratios are same, *i.e.*, 1 : 2 : 1 or 1 (red) : 2 (pink) : 1 (white).

l. Co-dominance and Multiple Allelism

- In a heterozygous individual, when both the alleles express their traits independently without showing the dominant-recessive relationship, they are called **co-dominant alleles.**
- The phenomenon of the independent expression of two contrasting alleles in a heterozygous individual is called **co-dominance.**
- More than two alternative forms (alleles) of a gene in a population, occupying the same locus on a chromosome or its homologue are known as **multiple alleles.**
- A well-known example of these phenomena is the inheritance of ABO blood groups in man.
- The gene for blood groups exists in three allelic forms, I^A, I^B and i.
- Any individual carries two of these three alleles.

- The allele I^A produces the glycoprotein A (antigen) found on the surface of RBC.
- I^B produces glycoprotein or antigen B.
- i does not produce any glycoprotein.
- Therefore, people with blood group A have antigen A; blood group B have antigen B; blood group AB have both antigens A and B; blood group O have no antigen.
- Both I^A and I^B are dominant over i but not over each other.
- When both I^A and I^B are present in a person, both are able to express themselves forming antigens A and B (co-dominance).
- The blood plasma of group A individuals has antibody *b* (or Anti A).
- Group B has antibody *a* (or Anti B).
- Group AB has no antibody.
- Group O has both antibody *a* and *b*.
- The following chart shows the blood groups, their possible genotypes, antigens and antibodies.

Blood Group	*Genotype*	*Antigen*	*Antibody*
A	$I^A I^A/I^Ai$	A	*b* (Anti A)
B	$I^B I^B/I^Bi$	B	*a* (Anti B)
AB	$I^A I^B$	A&B	Nil
O	ii	Nil	*a* and *b*

- The inheritance of blood group character follows the Mendelian pattern of inheritance.

Pleiotropy

- The ability of a gene to have multiple phenotypic effects because it influences a number of characters simultaneously is called **pleiotropy**.
- All traits may not be equally influenced.
- The most evidently expressed trait is called **major effect** and the less evidently expressed trait is called **minor effect**.

Examples:

(*i*) The gene for starch synthesis in pea can produce more than one effect. It has alleles B and *b*.
- In BB genotypes, large starch grains are produced and after maturation, the seeds are round.
- In bb genotypes, smaller starch grains are produced and the mature seeds are wrinkled.
- In Bb (heterozygous) genotypes, seeds are round but of an intermediate size.

(*ii*) Phenylketonuria in human beings.

PEDIGREE ANALYSIS

While plants can be crossed at will, humans cannot be. Thus the application of Mendel's laws for a study of problems of heredity in humans is a very difficult task. It is due to the fact that in human beings controlled crosses cannot be made, they have a long generation time of nearly 20 years and often produce a small progeny. Therefore human geneticists often scrutinise family histories (pedigree) in the hope that informative matings might have occurred by chance. This is called **pedigree analysis**. For such an analysis information about the family's history for a particular trait is collected. Then the expression of the trait is assembled into the family tree using standard symbols (*Fig. 14*). For instance, many human diseases or defects (e.g., albino, Tay-Sachs disease, cystic fibrosis, phenylketonuria, etc.) are governed by recessive genes. The following generalizations can be made for their pedigree analysis.

(1) A recessive character may appear in the progeny of both unaffected parents.

(2) Two affected parents cannot have an unaffected child.

(3) Often, these characters appear among children born from consanguineous marriage (e.g., first cousin marriage), which enhance the chances of mating among two heterozygotes for the same recessive trait.

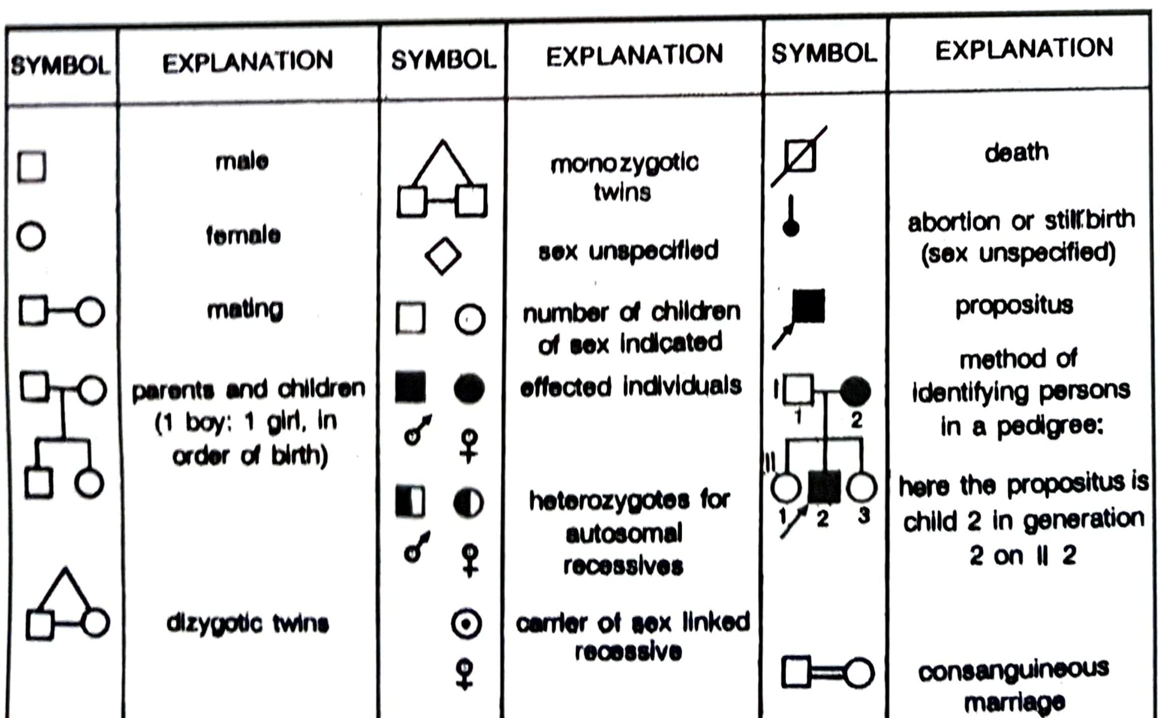

Fig. 14. Symbols used in human pedigree analysis.

A typical pedigree for a rare recessive condition is shown in figure 15A.

Sometimes the exceptional character may be governed by dominant alleles (e.g., Huntington's chorea, brachydactyly). Such a condition is characterised as follows.

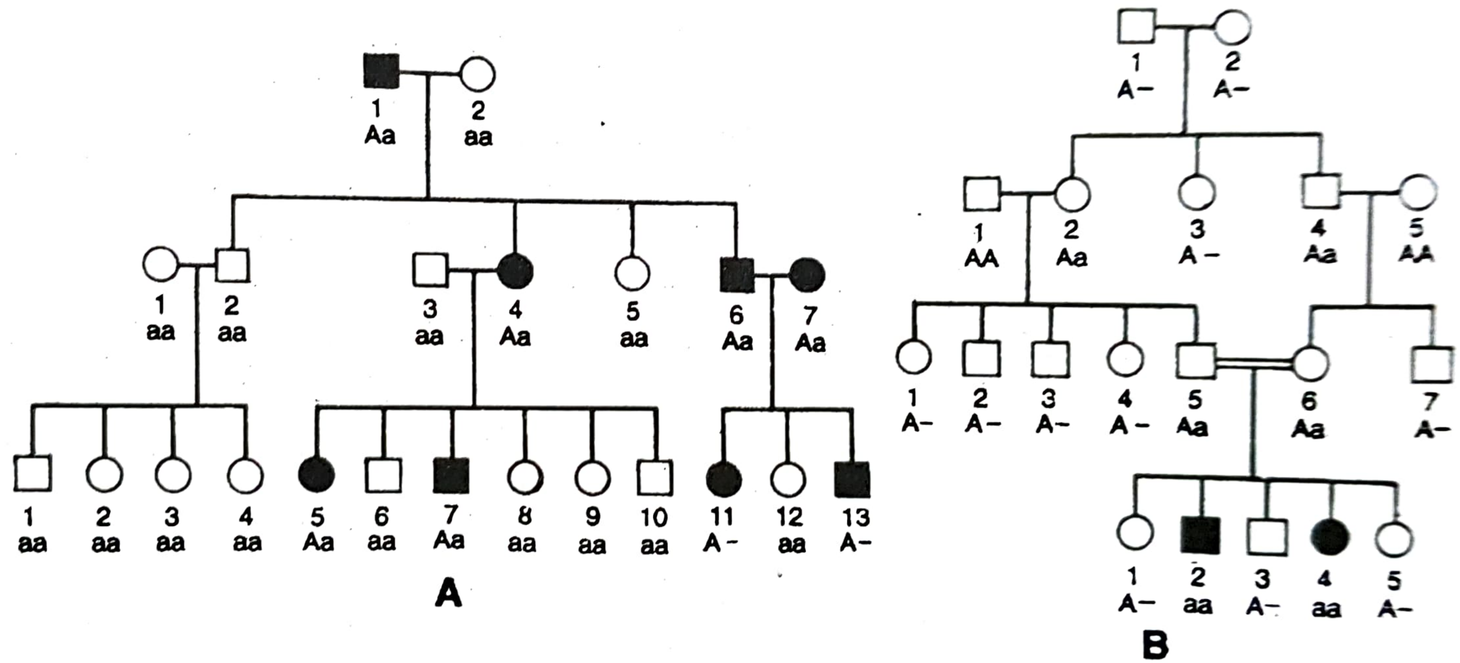

Fig. 15 A-B. Pedigree analysis : A. a pedigree involving an exceptional phenotype controlled by recessive allele 'a'; B. a pedigree involving a rare phenotype controlled by a dominant allele 'A'.

(1) It occurs in every generation.

(2) Unaffected parent can never transmit the condition to the offspring.

(3) Two affected parents may have unaffected children.

(4) It is passed on, on an average, to one half of the children of an affected individual.

A typical pedigree for a rare dominant condition is shown in figure 15 B.

Significance of pedigree analysis. Pedigree analysis helps in the identification of genes and gives information about the mode of its inheritance. It also helps genetic counsellors to advice couples worried of the possibility of having genetically defective children, when such a defect runs in their family.

Q. 1. How garden pea (*Pisum sativum*) proved to be a suitable plant for Mendel's work?

Q. 2. Define the following terms :

(a) Homozygous (b) Heterozygous

(c) Genotype (d) Phenotype

(e) Dominant (f) Recessive

(g) Allele (h) Back cross

Q. 3. Discuss Mendel's laws of inheritance. Which one of these laws you consider the most important and why?

Q. 4. Why was Gregor Johann Mendel crowned with success, whereas his predecessors failed to discover the basic principles of inheritance?

Q. 5. A heterozygous individual plant has a genotype AB/ab. List the type of gametes that will be produced with and without crossing over between the two genes. Explain your answer.

Q. 6. In human beings blue eye colour is recessive to brown eye colour. A brown-eyed man has a blue-eyed mother.

(a) What is the genotype of the man and his mother?

(b) What are the possible genotypes of his father?

(c) If the man marries a blue-eyed woman, what are the possible genotypes of their offspring?

Q. 7. How did Mendel's procedure differ from that of his predecessors? What mechanisms did the use of set aside any personal beliefs he may have had?

Q. 8. How did the monohybrid crosses performed 2by Mendel refute the blending concept of inheritance?

Q. 9. Using Mendel's monohybrid cross as an example, trace his reasoning to arrive at the law of segregation.

1. Write a note on term 'Multiple alleles'. **(2014)**
2. Explain pleiotropy with reference to phenylketonuria. **(2017)**
3. Explain the process of sex determination in honey bees. **(2016)**
4. Define complete linkage. Give an example of a cross showing complete linkage. **(2016)**
5. If the mother is a carrier of colour blindness and the father is normal, show the possible genotype and phenotype of the offspring of the next generation, with the help of a Punnett square. **(2018)**

Q. 6. A homozygous pea plant with round seed coat and yellow cotyledons is crossed with another homozygous pea plant having wrinkled seed coat and green cotyledons.
   (i) Give the types of gametes produced by plants of F1-generation.
   (ii) Give the dihybrid phenotypic ratio with the corresponding phenotypes.
   (iii) State the Mendel's principle involved in this cross. **(2019)**