CHAPTER **05**

Heredity and Variation

'Like begets like' is an important and universal phenomenon of life. It means that living beings produce offsprings of their own kind. The branch of science which deals with the study of hereditary traits and variations is termed as **genetics**.

Inheritance is the process by which characters of traits are passed from parent to their progeny, *i.e.* from one generation to the next. It is the basis of heredity. **Gene** is the unit of inheritance.

Variation is the degree by which progeny differs from their parents.

1. Genetic Terminology

A. **Gene** or **Factor** is a structure located on chromosomes which transfers a hereditary character from one generation to another generation. It is the unit of DNA.

B. The alternative forms of genes for a single trait or character are called **alleles**, *e.g.* colour of flower (red and white),stem height (tall and dwarf).

C. **Homozygous** A diploid condition, in which both alleles are same, *e.g.* TT, tt.

D. **Heterozygous** A diploid condition, in which both alleles are different, *e.g.* Tt.

E. **Monohybrid Cross** A cross in which only one pair of character is taken.

F. **Dihybrid Cross** A cross in which two pairs of characters are taken.

G. **Reciprocal Cross** A cross where sexes of parents are reversed.

H. **Back Cross** A cross between homozygous dominant parent and heterozygous F_1-hybrid is known as back cross.

I. **Test Cross** A cross between homozygous recessive parent and heterozygous F_1-hybrid is known as **test cross**. By this cross, it can be identified that a plant is homozygous or heterozygous.

2. Mendel's Experiments

Mendel conducted his experiments on garden pea for seven years to investigate inheritance pattern. He had applied statistical analysis and mathematical logic to his experiments.

The following contrasting traits were studied by Mendel in pea.

Character	Dominant	Recessive
Stem height	Tall	Dwarf
Flower position	Axial	Terminal
Pod colour	Green	Yellow
Pod shape	Inflated	Constricted
Seed shape	Round	Wrinkled
Flower colour	Purple	White
Seed coat colour	Yellow	Green

3. Laws of Mendel

On the basis of his observations, Mendel proposed three general rules to consolidate his understanding of inheritance in monohybrid cross.

These laws are given below

A. **Law of Dominance** It states following rules

(i) Characters are controlled by discrete units called **factors**.

(ii) Factors occur in pairs.

(iii) In a pair of dissimilar factors, one member of the pair is dominant while the other is recessive.

B. **Law of Segregation** It states that the parents contain two alleles during gamete formation. The factors or alleles of a pair segregate from each other, such that a gamete receives only one of the two factors. The following table shows the phenotypic and genotypic

ratio(s) of a monohybrid cross for better understanding of the above two laws

Generations/Ratio	F_1	F_2
Phenotypic	1	3 : 1
Genotypic	1	1 : 2 : 1

C. **Law of Independent Assortment** It states that when two pairs of traits are combined in a hybrid, segregation of one pair of characters is independent of the other pair of characters at the time of gamete formation. It also gets randomly rearranged in the offspring producing both parental and new combinations of characters. The following table explains the law of independent assortment on the basis of ratio(s) obtained through dihybrid cross.

Generations/Ratio	F_1	F_2
Phenotypic	1	9 : 3 : 3 : 1
Genotypic	1	1 : 2 : 2 : 4 : 1 : 2 : 1 : 2 : 1

4. Deviations from Mendel's Laws

All patterns of inheritance could not be explained on the basis of Mendel's principles which are as follows

(i) **Incomplete Dominance** It is a phenomenon in which phenotype of the F_1-hybrid offspring does not resemble any of the parent but is an intermediate between the expression of two alleles in their homozygous state. Both phenotypic and genotypic ratios is 1 : 2 : 1, *e.g.* flower colour in *Mirabilis jalapa.*

(ii) **Codominance** It is a phenomenon in which two alleles express themselves independently when present together in an organism, *e.g*. ABO blood group in humans.

(iii) **Multiple Alleles** More than two alternative forms (allele) of a gene occupying the same locus on a chromosome in a population are known as multiple alleles, *e.g*. ABO blood grouping.

Human Blood Groups, their Genotype and Transfusion

Blood groups	Antigen in RBC	Antibody in plasma	Can give blood to	Can take blood from	Gene type
O (universal donor)	None	a,b	O, A, B, AB	O	$I^\circ I^\circ$
A	A	b	A, AB	O, A	$I^A I^A$ or $I^A I^\circ$
B	B	a	B, AB	O, B	$I^B I^B$ or $I^B I^\circ$
AB (universal receiver)	A or B	None	AB	O, A, B, AB	$I^A I^B$

(iv) **Polygenic Inheritance** The inheritance of trait controlled by multiple genes where each gene contributes a little to the phenotype, *e.g.* human skin colour.

(v) **Pleiotropy** It is the phenomenon in which a single gene may produce multiple or more than one phenotypic effects, *e.g.* sickle-cell anaemia, white eye in *Drosophila*, Phenylketonuria (PKU).

5. Chromosomal Theory of Inheritance

It was proposed by **Boveri** and **Sutton** in 1902. It is the fundamental unifying theory of genetics which identifies chromosomes as the carriers of genetic material.

6. Crossing Over

The exchange of genetic material between non-sister chromatids of homologous chromosomes is known as crossing over.

Linkage and Recombination The physical association of two genes is called **linkage** while the generation of non-parental gene combinations is called **recombination**.

7. Sex-Determination

The establishment of sex through differential development in an individual at the time of zygote formation, is called sex-determination. There are following types of sex-determination mechanisms observed in various organisms

Male Heterogamety

- **XX-XO** type is seen in insects like grasshopper, etc. It is an example of male heterogamety where males have only one X-chromosome (called XO), whereas females have two X-chromosomes (XX).
- **XX-XY** type is seen in insects like *Drosophila melanogaster* and humans. Males have XY type chromosomes while females have XX type (homomorphic) chromosomes.

Female Heterogamety

- **ZZ-ZW** type is seen in birds, fowls and fishes. It is an example of female heterogamety because female produces two different types of gametes.
- **ZZ-ZO** type female is heterogametic (ZO) and male is homogametic (ZZ), *e.g.* in butterflies and moths.

Haplo-Diploidy is a sex-determination mechanism in which males develop from unfertilised eggs and are haploid. Females develop from fertilised eggs and are diploid. It is common in insects like honeybee, wasps, etc.

Note
- On the basis of type of allosomes present in the gamete, the parents can be of two types
  (i) Homogametic produces similar gametes.
  (ii) Heterogametic produces different gametes.

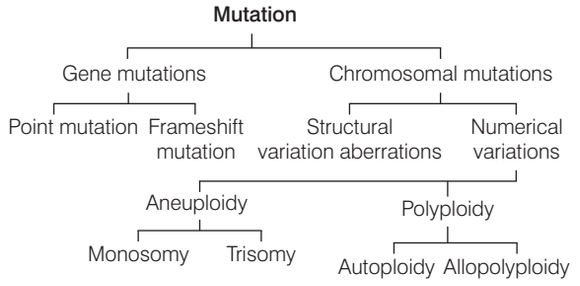
8. Sex-Determination in Human

Males have two types of gametes, *i.e.* X and Y, while females have only one type of gamete, *i.e.* XX. If an ovum gets fertilised with a sperm carrying X-chromosome, the zygote develops into a female (XX).

If an ovum fertilises with a sperm carrying Y-chromosome then it will develop into a male (XY). Hence, the type of sperm, which fertilises the ovum determines the sex of a child.

9. Mutation

It is a sudden, stable and inheritable change in genetic material or DNA sequences of an organism. The organism which undergoes mutation is called **mutant**. The factors, *i.e*. chemical and physical which induce mutations are called **mutagens**, *e.g.* UV-radiations, etc. The mutations are of following types



- **Point mutation** arises due to change in single base pair of DNA.
- **Frameshift mutation** refers to deletions and insertions of base pairs in DNA.
- The loss or gain of a segment of DNA, results in **structural alteration in chromosomes** because genes are located on the chromosomes.
- When the members of a homologous pair of chromosomes fail to segregate during meiosis, **aneuploidy** occurs, *i.e.* loss or gain of one or more chromosomes. Due to this, there may be **monosomy**, *i.e.* lack of one chromosome of normal complement or trisomy, *i.e.* three instead of the normal two chromosomes.
- **Polyploidy** occurs when there is failure of cytokinesis after telophase stage of cell division. It results in an increase in a whole set of chromosomes in an organism.

10. Pedigree Analysis

The analysis of traits in several generations of a human family in the form of a family tree or a diagram is known as pedigree analysis.

11. Genetic Disorders

The disorders or illness which are caused by one or more abnormalities in autosomes or sex chromosomes of a person are called **autosomal disorders** and **sex-linked disorders**, respectively. It can be divided into the following

A. **Mendelian Disorders** These are determined by alteration or mutation in a single gene. These disorders can be recessive or dominant.
Some diseases are described below

(i) **Colour Blindness** Person suffering from this disorder is not able to distinguish between red and green colour. Gene controlling colour blindness is located on X-chromosome and is recessive.

(ii) **Haemophilia** (bleeder's disease) Person suffering from this disorder is not able to form blood clots when hurt or wounded. The continuous bleeding causes death of the person. This disorder is caused due to X-linked recessive gene.

(iii) **Sickle-cell Anaemia** It is an autosomal linked recessive trait that can be transmitted from parents to the offsprings, when both the partners are carrier for the gene (heterozygous). In this, the substitution of valine by glutamic acid changes the shape of the RBCs to a sickle like structure.

(iv) **Phenylketonuria** (PKU) is an inborn error metabolism, which is inherited as an autosomal recessive trait. The affected individual lacks the enzyme that converts phenylalanine to tyrosine. Its gene is associated with the 12th chromosome.

(v) **Thalassemia** is an autosomal recessive disease, which occurs due to either mutation or deletion of genes. It results in reduced rate of synthesis of one of the globin chains (α or β) of haemoglobin.

B. **Chromosomal Disorders** These are caused by the absence, excess or abnormal arrangement of one or more chromosomes. Some of the disorders are described below

(i) **Down's Syndrome** or **Mongolism** It is caused by the presence of an extra copy of chromosome number 21 in autosomes, *i.e.* trisomy. An individual suffering from Down's syndrome is short statured, have furrowed tongue, slow motor and mental development.

(ii) **Turner's Syndrome** It is caused by the absence of one X-chromosome in females. The females have 45 (44+X) chromosomes. Females are sterile with less developed uterus, short height and webbed neck are the symptoms.

(iii) **Klinefelter's Syndrome** It is due to the presence of an extra copy of X-chromosome in males like 44+XXY, 44+XXXY, etc. The affected individuals are phenotypically males. The symptoms include underdeveloped testes, presence of breast-like structures, little growth of hair on face and body, sterility (no sperm formation), mental retardedness, etc.

Practice Questions

1. Genetics is the branch of biology which deals with
(a) variation (b) inheritance
(c) Both (a) and (b) (d) study of characters

2. The tendency of offspring to differ from their parents is called
(a) variation (b) heredity
(c) inheritance (d) resemblance

3. Mendel investigated characters in garden pea plant manifested in two traits which were
(a) similar (b) non-zygote
(c) identical (d) opposite

4. A true breeding line is characterised by the presence of
(a) stable trait inheritance due to the continuous self-pollination
(b) varying traits in different generations due to the cross pollination
(c) single trait in all generations due to allogamy
(d) varying trait inheritance in a single generation due to geitonogamy

5. Out of 7 contrasting trait pairs selected by Mendel, how many traits were dominant and recessive?
(a) 7 and 7 (b) 8 and 6
(c) 6 and 8 (d) 5 and 9

6. Which is correct about traits choosen by Mendel for his experiment on pea plant?
(a) Terminal pod was dominant
(b) Constricted pod was dominant
(c) Green coloured pod was dominant
(d) Tall plants were recessive

7. The first hybrid progeny obtained by Mendel were called
(a) F_1-progeny (b) F_0-progeny
(c) F_2-progeny (d) F_3-progeny

8. F_1-progeny of a cross between pure tall and dwarf plant is always
(a) tall
(b) short
(c) intermediate
(d) None of these

9. The Mendel crossed true breeding tall and dwarf plant varieties in his experiment. Tallness was the dominant character and dwarfness was recessive. The recessive character appeared in
(a) F_1 (b) F_2
(c) F_3 (d) F_2 and F_3

10. How did Mendel obtained recessive (dwarf) character in F_2-generation?
(a) By self-pollinating F_1 (b) By self-pollinating F_2
(c) By cross-pollinating F_1 (d) By cross-pollinating F_2

11. Mendel crossed tall and dwarf plants. In F_2-generation both the tall and dwarf plants were produced. This shows
(a) blending of characters
(b) atavism
(c) non-blending of characters
(d) intermediate characters

12. During his experiments, Mendel used the term factor for
(a) genes (b) traits (c) characters (d) qualities

13. Choose the incorrect match.
(a) Phenotype – Physical appearance of an organism
(b) Genotype – Expressed genes
(c) Homozygous – Identical alleles of a gene present at the same locus
(d) Heterozygous – Genes of an allelic pair are not same

14. The phenotypic ratio of a monohybrid cross in F_2-generation is
(a) 3 : 1 (b) 1 : 2 : 1
(c) 2 : 1 : 1 (d) 9 : 3 : 3 : 1

15. The genotypic ratio of a monohybrid cross in F_2-generation is
(a) 3 : 1 (b) 1 : 2 : 1
(c) 2 : 1 : 1 (d) 9 : 3 : 3 : 1

16. Graphical representation to calculate the probability of all possible genotype of an offspring in genetic cross is called
(a) Bunett square (b) Morgan square
(c) Punnett square (d) Mendel square

17. When alleles of two contrasting characters are present together and one of the character expresses itself during the cross while the other remains hidden gives the
(a) law of purity of gametes
(b) law of segregation
(c) law of dominance
(d) law of independent assortment

18. 3:1 ratio in F_2-generation is explained by
(a) law of partial dominance
(b) law of dominance
(c) law of incomplete dominance
(d) law of purity of gametes

19. The law of dominance is applicable in inheritance of
(a) seed colour in pea
(b) flower colour in *Mirabilis jalapa*
(c) starch grain size in pea
(d) roan coat colour in cattles

20. The law based on fact that the characters do not show any blending and both the characters are recovered as such in F_2-generation although one character was absent in F_1-progeny, is
(a) law of purity of gametes
(b) law of independent assortment
(c) law of incomplete dominance
(d) law of dominance

21. Theoretically in incomplete dominance one allele functions as normal, while another allele may function as
(a) normal allele
(b) non-functional allele
(c) normal but less efficient allele
(d) All of the above

22. Incomplete dominance is similar to codominance in having identical
(a) phenotypic ratio
(b) genotypic ratio
(c) Both (a) and (b)
(d) None of the above

23. The recessive trait in case of incomplete dominance is seen due to the
(a) non-functional enzyme produced by modified gene
(b) absence of any enzyme that may otherwise be produced by modified gene
(c) normal or less efficient enzyme produced by recessive allele
(d) Both (a) and (b)

24. In human blood group inheritance
(a) I^A and I^B are codominant
(b) I^A and I^B are dominant over i
(c) I^A is dominant over I^B
(d) Both (a) and (b)

25. If two persons with 'AB' blood group marry and have sufficiently large number of children, these children could be classified as 'A' blood group : 'AB' blood group : 'B' blood group in 1 : 2 : 1 ratio. Modern technique of protein electrophoresis reveals presence of both 'A' and 'B' type proteins in 'AB' blood group individuals. This is an example of
(a) codominance
(b) incomplete dominance
(c) partial dominance
(d) complete dominance

26. Which of the following option (s) is/are correct for starch synthesis in pea seeds controlled by single gene which has two allelic forms B and b?
(a) BB is round seed with large starch synthesis
(b) bb is wrinkled seed with large starch synthesis
(c) Bb is round seed with less starch synthesis
(d) All of the above

27. The types of gametes formed by the genotype RrYy are
(a) RY, Ry, rY, ry (b) RY, Ry, ry, ry
(c) Ry, Ry, Yy, ry (d) Rr, RR, Yy, YY

28. In Mendel's experiments with garden pea, round seed shape (RR) was dominant over wrinkled seeds (rr) and yellow colour (YY) was dominant over green colour (yy). What are the expected phenotypes in the F_1-generation of the cross RRYY× rryy?
(a) Only round seeds with yellow cotyledons
(b) Only wrinkled seeds with yellow cotyledons
(c) Only wrinkled seeds with green cotyledons
(d) Round seeds with yellow cotyledons and wrinkled seeds with yellow cotyledons

29. In cross between pure breeding pea plants having yellow round (YYRR) and green wrinkled (yyrr) seeds, find out the total seeds (plants) having yellow colour in F_2-generation.
(a) 12 (b) 10 (c) 14 (d) 11

30. In a cross between plants having yellow round (YYRR) and green wrinkled (yyrr) seeds, what will be the ratio between seeds having yellow and green seed colour?
(a) 3 : 2 (b) 3 : 1 (c) 9 : 7 (d) 7 : 9

31. Mendel's result on inheritance of characters were rediscovered by
(a) de Vries (b) Correns
(c) von Tschermak (d) All of these

32. The chromosomes as well as genes occur in pair and the two alleles of a gene pair are located on
(a) homologous chromosomes
(b) non-homologous chromosomes
(c) single chromosome
(d) All of the above

33. Who proposed the chromosomal theory of inheritance?
(a) Sutton and Mendel (b) Boveri and Morgan
(c) Morgan and Mendel (d) Sutton and Boveri

34. In Morgan's experiment, white and yellow genes were linked tightly, while white and miniature wing were loosely linked. The per cent recombination shown by these genes were
(a) 50% each
(b) 72% and 8.3%, respectively
(c) 0.3% and 53%, respectively
(d) 1.3% and 37.2%, respectively

35. Polygenic traits are controlled by

(a) one gene (b) two genes
(c) three or more genes (d) mutant genes

36. Phenylketonuria in human

(a) manifests through phenotypic expressions
(b) is characterised by mental retardation
(c) leads to hair reduction and skin pigmentation
(d) All of the above

37. Which one of the following pairs is wrongly matched?

(a) XO type of sex-determination – Grasshopper
(b) ABO blood grouping – Codominance
(c) Starch synthesis in pea – Multiple allele
(d) TH Morgan – Linkage

38. Choose the incorrect pair with respect to sex determination in different organisms.

(a) Grasshopper = XO type
(b) Birds = ZZ-ZW type
(c) *Drosophila* = XX-XO type
(d) Human = XX-XY type

39. A human male contains the karyotype of …*A*… and a human female has …*B*… chromosomes.

	A	B		A	B
(a)	44 + XX	44 + XY	(b)	44 + XY	44 + XX
(c)	44 + XO	44 + XX	(d)	44 + XX	44 + XO

40. If there are four different types of nitrogenous bases (A, T, G and C) then how many different types of transitions and transversion are possible?

(a) Transition = 8, Transversion = 4
(b) Transition = 4, Transversion = 4
(c) Transition = 8, Transversion = 4
(d) Transition = 4, Transversion = 8

41. Sickle-cell anaemia is a classical example of

(a) frame-shift mutation
(b) point mutation
(c) Both (a) and (b)
(d) None of the above

42. Colour blindness in humans

(a) results in defect in either red or green cone of eyes
(b) is caused due to the mutation in gene found on X-chromosome
(c) affects males more frequently than females
(d) All of the above

43. In haemophilia, the affected protein is a part of a cascade of protein which is involved in the

(a) formation of RBCs
(b) formation of WBCs and platelets
(c) coagulation of blood
(d) anticoagulation

44. In sickle-cell anaemia,

(a) Both parents are heterozygous carriers, but are unaffected
(b) Single pair of allele controls the disease
(c) Only Hb^sHb^s show diseased phenotype
(d) All of the above

45. Thalassemia in humans

(a) is an autosome linked recessive blood disorder
(b) can transmit from parents to offspring when both parents are unaffected carriers (heterozygous)
(c) caused due to the mutation or deletion of one of the α or β-globin chain
(d) All of the above

ANSWERS

1.	(c)	**2.**	(a)	**3.**	(d)	**4.**	(a)	**5.**	(a)	**6.**	(b)	**7.**	(a)	**8.**	(a)	**9.**	(b)	**10.**	(a)
11.	(c)	**12.**	(a)	**13.**	(b)	**14.**	(a)	**15.**	(b)	**16.**	(c)	**17.**	(c)	**18.**	(b)	**19.**	(a)	**20.**	(a)
21.	(d)	**22.**	(c)	**23.**	(d)	**24.**	(d)	**25.**	(a)	**26.**	(a)	**27.**	(a)	**28.**	(a)	**29.**	(a)	**30.**	(a)
31.	(d)	**32.**	(a)	**33.**	(d)	**34.**	(d)	**35.**	(c)	**36.**	(d)	**37.**	(c)	**38.**	(c)	**39.**	(b)	**40.**	(d)
41.	(b)	**42.**	(d)	**43.**	(c)	**44.**	(d)	**45.**	(d)										

Hints & Explanations

11. (*c*) In Mendel's experiment dominant and recessive traits were expressed or appeared separately. This shows that there was no mixing of characters, thus non-blending of character in F_2-generations is observed.

13. (*b*) Option (b) is incorrect and can be corrected as
The genetic composition of an organism, with respect to one or more characters whether the gene is expressed or not is called genotype.

18. (*b*) 3:1 ratio in F_2-generation can be explained by the law of dominance. It states that only dominant allele shows its effect even in the heterozygous condition and masks the effect of recessive allele. In the given ratio, 3 represents the dominant phenotype, while 1 represents the recessive phenotype.

20. (*a*) Law of purity of gametes states that the characters which were hidden or masked in F_1 progeny get recovered in the progeny of F_2-generation.

21. (*d*) In incomplete dominance, the two genes of an allelomorphic pair are not related as dominant or recessive but each of them express themselves partially. In this phenomenon, one allele may function normally and the other may either function normally, may be non-functional or may perform normally, but with less efficiency. This occurs due to some changes in the allele which modifies the information present in it. Thus, option (d) is correct.

22. (*c*) Both codominance and incomplete dominance give an identical genotypic and phenotypic ratio of 1 : 2 : 1. Codominance and incomplete dominance differ in the fact that in codominance both the alleles are dominant and express themselves at the same time. But in incomplete dominance, the two alleles are neither dominant nor recessive to each other.

25. (*a*) AB blood group is characterised by the presence of both antigen A and B, i.e. I^A and I^B over the surface of RBCs.

Phenotype AB
↓
Genotype I^AI^B
↓
Antigen A + Antigen B

Here, both alleles are able to express themselves forming antigens A and B. This is called as codominance, a phenomenon in which both the alleles in a heterozygote are expressed.

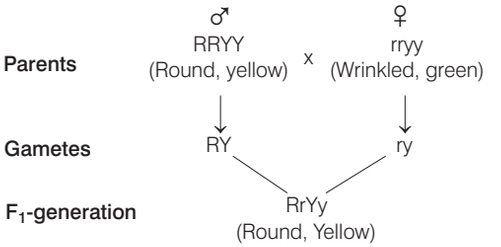
26. (*a*) Option (a) is correct. This can be explained as occasionally a single gene product may produce more than one effect. Like in starch synthesis in pea seeds controlled by one gene having two alleles (B and b). Thus, the phenotype and genotype expressed are

- BB–round seed, large starch synthesis.
- bb–wrinkled seed, less starch synthesis.
- Bb–round seed, intermediate starch synthesis.

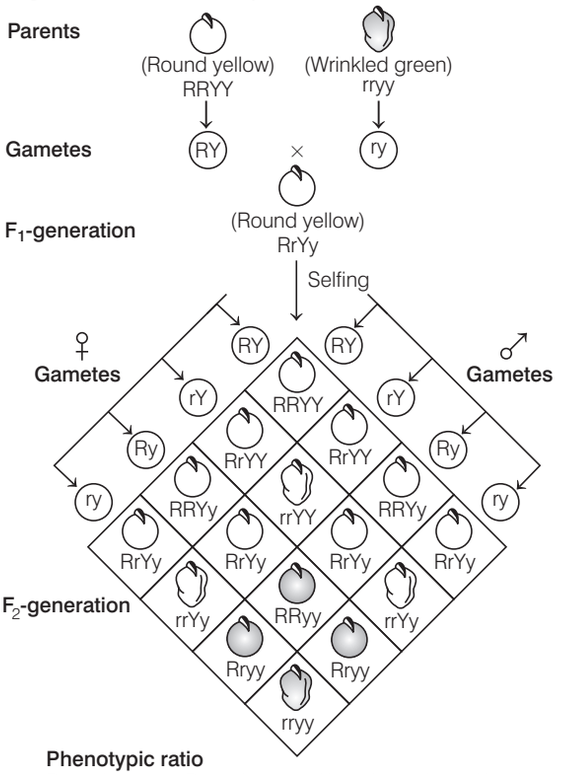
Thus, if we take size as a phenotype, allele B shows complete dominance over 'b', but if we take starch synthesis as a phenotype, allele B and b shows incomplete dominance.

27. (*a*) The formula to determine the number of gametes is $2^n = 2^{(2)} = 4$
Thus, RrYy would produce 4 gametes of the types RY, Ry, rY, ry.

28. (*a*) When plants bearing round yellow (RRYY) seeds are crossed with plants bearing and wrinkled green (rryy) seeds, all the plants in F_1-generation will have yellow round seeds. It can be depicted by the cross given below



29. (*a*) A cross between pure yellow round seeded plants and pure green wrinkled seeded plants is shown below



Phenotypic ratio
Round yellow : Round green : Wrinkled yellow : Wrinkled green
9 : 3 : 3 : 1

Genotypic ratio 1 (RRYY) : 2 (RRYy) : 2 (RrYY) : 4 (RrYy) : 1 (RRyy) 2 (Rryy) : 1 (rrYY) : 2 (rrYy) : 1(rryy)

31. (*d*) In 1900, three scientists (de Vries, Correns and von Tschermak) independently rediscovered Mendel's results on the inheritance of characters. Also, by this time due to advancements in microscopy that were taking place, scientists

were able to carefully observe cell division.

This led to the discovery of structures in the nucleus that appeared to double and divide just before each cell division.

33. (*d*) Sutton and Boveri proposed the chromosomal theory of inheritance. In 1902, the chromosomal movement during meiosis was worked out. Walter Sutton and Theodore Boveri stated that pairing and separation of a pair of chromosomes would lead to the segregation of a pair of factors they carried. They united the knowledge of chromosomal segregation with Mendelian principles and called it chromosomal theory of inheritance.

37. (*c*) In the given pairs, option (c) is wrongly matched. It can be corrected as Starch synthesis in pea is an example of pleiotropy.

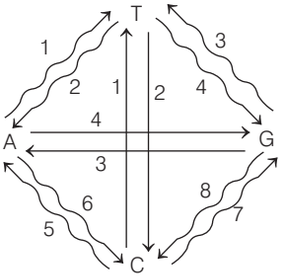
Rest of the pairs are correctly matched.

38. (*c*) Option (c) contains the incorrect pair. It can be corrected as In *Drosophila*, XX-XY type of sex-determination is seen, i.e. same as humans.

Rest of the pairs are correct.

40. (*d*) Transition occurs when a purine base (A or G) is replaced by another purine base. Transversion occurs when a purine base is substituted by a pyrimidine base or *vice-versa*.

Thus, the number of possible transitions and transversions among the four bases (A, T, G, C) are 4 and 8, respectivley and can be represented by the figure given below



(Transition → and Transversion⇝)

43. (*c*) Haemophilia is a sex-linked recessive disease, which shows its transmission from unaffected carrier female to some of the male progeny has been widely studied. In this disease, a single protein that is a part of the cascade of proteins involved in the clotting of blood is affected. Due to this, in an affected individual, a simple cut will result in non-stop bleeding.

45. (*d*) Thalassemia is also an autosome linked recessive blood disease transmitted from parents to the offspring when both the partners are unaffected carrier for the gene (or heterozygous). The defect could be due to either mutation or deletion which ultimately results in reduced rate of synthesis of one of the globin chains (α and β-chains) that make up haemoglobin. This causes the formation of abnormal haemoglobin molecules resulting into anaemia which is characteristic of the disease.