4 CHAPTER

Principles of Inheritance and Variation

Level - 1 CORE SUBJECTIVE QUESTIONS

MULTIPLE CHOICE QUESTIONS (MCQs) (1 Mark)

1. Option (C) is correct
Explanation:

Suresh	Rajesh
Sickle Cell Anaemia – Autosomal linked Recessive trait	Thalassemia – Autosomal Recessive blood disorder

2. Option (A) is correct
Explanation: The sex-determination in honey bee is haplo-diploid type. It is done on the basis of the number of sets of chromosomes one individual receives. An offspring formed by the fertilisation of a sperm and an egg results into a female (2n). An unfertilised egg develops as a male (n) by the process of parthenogenesis.

3. Option (B) is correct
Explanation: 44 + XXY refers to Klinefelter syndrome, which occurs in males. These individuals have an extra X chromosome and show male development with some feminine characteristics (like gynecomastia and less body hair), but they do not exhibit overall feminine development.

4. Option (C) is correct
Explanation: The correct matching pair are:
1. Mendelian monohybrid - (iii) 3:1 (F_2)
2. Mendelian dihybrid - (iv) 9:3:3:1 (F_2)
3. Incomplete dominance - (i) 1:2:1 (F_2)
4. Test cross - (ii) 1:1

5. Option (A) is correct
Explanation: Colour blindness is a sex linked recessive trait vision and the father is colour blind means the woman must be a carries (X^NX^C) because she got one X chromosome with the colour blind gene (X^C) from her father and a normal one (X^N) from her mother. She marries a man with normal vision – his genotype is X^NY.

	X^N (man)	Y (man)
X^N (woman)	X^NX^N	X^NY
X^C (woman)	X^NX^C	X^CY

Possible progeny
X^NX^N – normal daughter
X^NX^C – carrier daughter
X^NY – normal son
X^CY – colour blind son
So, out of 4 possibilities – 1 is colour blind child (X^CY) and 3 children are without colour blindness (though one daughter is a carrier.)
Percentage chance of their progeny being colour blind = 25%.

6. Option (D) is correct
Explanation: In a pedigree chart, this is typically represented by a double line between the individuals who are mating, indicating that they are biologically related.

7. Option (C) is correct
Explanation: In this cross between a heterozygous tall pea plant (Tt) and a homozygous dwarf pea plant (tt), the expected phenotypic ratio follows a 1:1 ratio, as it is a test cross. Half of the offspring will be tall (Tt) and the other half will be dwarf (tt).

Phenotype of Parents ⟶ Heterozygous Tall × Dwarf
Genotype ⟶ Tt tt
Gametes ⟶ (T) (t) × (t)

F_1 generation

♀ \ ♂	T	t
t	Tt Heterozygous tall	tt Homozygous dwarf

Thus, among 60 individuals, 30 will be tall, and 30 will be dwarf.

8. Option (C) is correct
Explanation: In Snapdragon plants, flower colour exhibits incomplete dominance, where the red (R) and white (r) alleles combine to form an intermediate phenotype (pink). A pink-flowered plant (Rr) crossed with a white-flowered plant (rr) would result in the following possible offspring: 50% Rr (pink) and 50% rr (white).

Rr × rr Parents

♀ \ ♂	r	r
R	Rr Pink	Rr Pink
r	rr White	rr White

F_1 generation

9. Option (A) is correct
Explanation: Klinefelter's syndrome occurs in males with an extra X chromosome (45 + XXY). Individuals typically have a tall stature, feminised characteristics (such as gynecomastia, less body and facial hair).

10. Option (B) is correct
Explanation: In female heterogamety, females have two different sex chromosomes (e.g., ZW), while males have two of the same kind (e.g., ZZ). Hen (birds) exhibit this system, where the female is ZW and the male is ZZ.

11. Option (C) is correct
Explanation: Parthenogenesis is a form of asexual reproduction where an egg develops into an individual without fertilisation. In honey bees, this process leads to the development of drones, which are the male bees. Drones are haploid, meaning they have only one set of chromosomes, which they inherit solely from the mother.

12. Option (D) is correct
Explanation: Non-disjunction refers to the failure of homologous chromosomes or sister chromatids to separate properly during cell division. In humans, if non-disjunction occurs with the 21st pair of chromosomes, it results in Down's Syndrome (trisomy 21), where individuals have three copies of chromosome 21 instead of the usual two.

13. Option (B) is correct.
Explanation: Sickle cell anemia is an autosomal recessive disease. This genetic disorder is caused by a mutation in the Hb^A gene on chromosome 11, leading to the production of abnormal haemoglobin (haemoglobin S), which causes red blood cells to become rigid and sickle-shaped.

14. Option (C) is correct.
Explanation: ABO blood group is an example of (c) co-dominance and multiple allelism. In this system, there are three alleles: I^A, I^B, and i. Individuals can have any combination of these alleles, leading to four possible blood types (A, B, AB, and O). The alleles I^A and I^B are co-dominant, meaning both can be expressed simultaneously in individuals with type AB blood, while the i allele is recessive.

15. Option (B) is correct
Explanation: The production of recombinants in future generations is primarily driven by two processes: independent assortment during meiosis I and crossing over of bivalents.
Independent assortment during meiosis I occurs when homologous chromosomes are segregated into gametes, leading to various combinations of alleles and contributing to genetic variation among offspring. Crossing over of bivalents takes place during prophase I of meiosis, where homologous chromosomes exchange segments of genetic material. This recombination creates new allele combinations, resulting in the production of recombinants.

16. Option (B) is correct
Explanation: The correct order of the genes R, S, and T on the chromosome can be determined from their recombinant percentages:
- The recombinant frequency between S and T is the smallest (15%), indicating they are closest together.
- The next smallest frequency is between R and S (20%).
- The largest frequency is between R and T (35%), suggesting R is further away from T.
Thus, the predicted order is R-S-T.

17. Option (B) is correct
Explanation: In grasshoppers, the male has an XO system, where XO indicates a male, while females are XX.

18. Option (C) is correct
Explanation: Consanguineous mating refers to the breeding or reproduction between individuals who are closely related, such as between relatives like cousins, uncles and nieces, or aunts and nephews. In pedigree analysis, it is represented with a double line connecting the individuals.

19. Option (C) is correct
Explanation: A Punnett square is a graphical tool used in genetics to predict the potential genotypes of offspring from a genetic cross between two parents. It visually represents the different combinations of alleles that can result from the mating of two individuals, helping to calculate the probability of various genotypes and phenotypes among the offspring.

ASSERTION-REASON QUESTIONS (1 Mark)

1. Option (A) is correct
Explanation: Linked genes tend to be inherited together because they are located close to each other on the same chromosome. Therefore, they do not exhibit the typical 9:3:3:1 phenotypic ratio seen in dihybrid crosses of unlinked genes. Linked genes are inherited together and do not assort independently during gamete formation.

2. Option (A) is correct
Explanation: In birds, including pigeons, the females are heterogametic, meaning they have two different sex chromosomes (ZW), while the males are homogametic, having two of the same sex chromosomes (ZZ).

3. Option (A) is correct
Explanation: In the context of polygenic inheritance, the genotype with all dominant alleles is typically associated with the phenotype exhibiting the most extreme trait expression—in this case, the darkest skin color. Polygenic traits are influenced by multiple genes, and the phenotype results from the cumulative effect of all contributing alleles. Each dominant allele contributes to a darker skin pigmentation, while recessive alleles contribute less or have no effect.

4. Option (D) is correct
Explanation: Thalassemia is primarily associated with defects in haemoglobin production, not myoglobin. α-Thalassemia is caused by mutations in the HBA1 and HBA2 genes, which are located on chromosome 16 and are responsible for the production of the alpha globin chains of haemoglobin.

5. Option (C) is correct
Explanation: In a test cross, the organism with the dominant phenotype (which may be either homozygous dominant or heterozygous) is crossed with a homozygous recessive parent. The purpose of the test cross is to reveal the genotype of the dominant phenotype individual based on the resulting offspring.

VERY SHORT ANSWER TYPE QUESTIONS (2 Marks)

1.

Haemophilia	Sickle cell anaemia
(i) Sex linked/X-linked recessive disorder	Autosomal recessive disorder.
(ii) More males than females are affected.	Affects both males and females equally.
(iii) Carrier/unaffected female transmits the disease to some of her male offspring.	When both the parents are carriers, the disease is transmitted to the offspring.

(Any two)

2. Case I -Homozygous dominant

GG × gg

Green Pods — Yellow Pods

	G	G
g	Gg	Gg
g	Gg	Gg

Phenotype: All green coloured pod

Case II -Heterozygous dominant

Gg × gg

Green Pods — Yellow Pods

	G	g
g	Gg	Gg
g	Gg	Gg

Phenotype: 50% green & 50% yellow colour pod

3. (i) HBA1, HBA2 genes located on chromosome 16.

(ii)

α-Thalassemia	Sickle cell anaemia
Synthesis of too less haemoglobin.	Incorrectly functioning globing.

SHORT ANSWER TYPE QUESTIONS (3 Marks)

1. No,
Son inherited disease from the mother and daughter inherited disease from both mother and father.

Mother X^CX (Normal (Carrier)) × Father X^CY (Colour blind)

	♀ X^C	♀ X
♂ X^C	X^CX^C Colour blind daughter	X^CX Carrier daughter
♂ Y	X^CY Colour blind son	XY Normal son

2. (i) Sickle cell Anemia

Cause : Substitution of glutamic acid by valine at sixth position of beta globin chain of haemoglobin molecule or due to single base substitution at the sixth codon of the beta globin gene from GAG to GUG.

Symptoms: Change in shape of RBC from biconcave disc to elongated sickle like structure causes anemia due to which oxygen carrying capacity of the RBCs decreases/tiredness/ breathlessness.

(ii) Autosomal recessive genetic disorder. Controlled by pair of allele Hb^AHb^S.

$Hb^A\ Hb^S$ (carrier male) × $Hb^A\ Hb^S$ (carrier female)

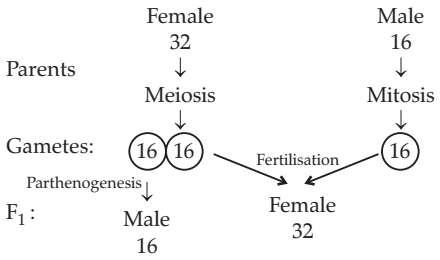
	Hb^A	Hb^S
Hb^A	Hb^AHb^A Normal	Hb^AHb^S Carrier
Hb^S	Hb^AHb^S Carrier	Hb^SHb^S Affected

3. (i) Karyotype - 44 + XXY/47-XXY/AA+XXY,
Genetic disorder - Klinefelter's syndrome

(ii) Gynaecomastia/development of breast, Sterile individuals, overall masculine development (any two symptoms)

(iii) Failure of segregation of chromatids or chromosomes/non-disjunction of chromatids or chromosomes during cell division or gametogenesis.

4. Honey bee shows mechanism of haplo-diploid pattern of sex-determination, female (queen or worker) develops from fertilised egg, so are diploid, males (drones) develop from unfertilised egg by parthenogenesis, so are haploid, Females are diploid having 32 chromosomes and the males are haploid having 16 chromosomes. This is why this pattern is called haplo-diploid sex determination.



5. In a dihybrid cross the F_2 phenotypic ration deviated very significantly from 9:3:3:1, the proportion of the parental gene combination was much higher than the non-parental type, the parental combinations were due to linkage of genes (physical association) and the non-parental types were due to distant genes (recombination/ crossing over).

6. Human blood group inheritance is regulated by the gene 'I' which is present in more than two allelic forms – I^A, I^B, i/ Hence, human blood group inheritance is controlled by more than two alleles that is called multiple allelism.
When I^A and I^B are present together in blood group AB they both express their own types of allele, because both the dominant alleles hence show co-dominance.

7. **(i)** Carrier daughters- 50% of the daughters or 25% of the total offsprings.
Sufferer sons-50% of the sons or 25% of the total offsprings.
(ii) Carrier daughters- 25% of the total offsprings.
Sufferer daughters- 25% of the total offsprings
Normal sons - 25% of the total offsprings
Sufferer sons- 25% of the total offsprings.
(iii) Sex linked recessive disease/ criss -cross pattern of inheritance.

8. **(i)** Offspring 1
Blood Group – A/B, Genotype – I^Ai/I^Bi.
(ii) Offspring 2
Blood Group –A, Genotype – I^Ai or Blood Group –O, genotype – ii
Offspring 3
Blood Group –A, Genotype – I^Ai , $I^A I^A$
Blood Group- AB, Genotype- $I^A I^B$

9. **(i)** Recessive trait. Both the parents in generation I do not express the trait yet it appears in the progeny.
(ii) Autosomal trait. Both male and females have equal chances of getting the trait.
(iii) Child '1' :Aa/AA , Child '3' : Aa

10. **(i)** Recessive.
Individuals 1 and 2 don't have disease but their offspring individual 3 shows the disease.
(ii) Individual 3 is homozygous.
(iii) Individual 7/8.
(iv) Individual 2/5.
(v) Autosomal disorder.

11. **(i)**

Gene	Identification	Contrasting forms
y	Yellow body	Brown body
w	White eye	Red eye
m	Miniature wings	Normal wings

(ii) Cross A- 1.3%
Cross B- 37.2%
(iii) The genes yellow and white are very tightly linked and showed very low recombination frequency (1.3%), while white and miniature wing are very loosely linked and showed higher recombination frequency (37.2%).

LONG ANSWER TYPE QUESTIONS (5 Marks)

1. Tall Pea plant with violet flowers can have 4 types of genotypes:
TTVV, TtVV, TtVv, TTVv
Case I- TTVV × TTVV
↓
Genotype TTVV
Phenotype All will be tall and violet
Case II-
TtVV × TtVV
↓

Gametes	TV	tV
TV	TTVV Tall Violet	TrVV Tall Violet
tV	TtVV Tall Violet	ttVV Dwarf Violet

Phenotypic ratio Tall Violet : Dwarf Violet
3 : 1
Genotypic ratio TTVV : TtVV : ttVV
1 : 2 : 1
Case III-
TtVv × TtVv
↓

Gametes	TV	Tv	tV	tv
TV	TTVV Tall violet	TTVv Tall Violet	TtVV Tall Violet	TtVv Tall Violet
Tv	TTVv Tall Violet	TTvv Tall white	TtVv Tall Violet	Ttvv Tall white
tV	TtVV Tall violet	TtVv Tall violet	ttVV dwarf violet	ttVv dwarf Violet
Tv	TtVv Tall violet	Ttvv Tall white	ttVv dwarf violet	ttvv dwarf white

Phenotypic ratio - Tall Violet : Dwarf violet: Tall white : dwarf white
9 : 3 : 3 : 1
Genotypic ratio-
TTVV : TTVv : TtVV : TtVv : TTvv : ttVV : ttVv : Ttvv : ttvv
1 : 2 : 2 : 4 : 1 : 1 : 2 : 2 : 1

Case IV-

TTVv X TTVv

↓

Gametes	TV	Tv
TV	TTVV Tall Violet	TTVv Tall Violet
Tv	TTVv Tall Violet	TTvv Tall White

phenotypic ratio- Tall violet : Tall white

3 : 1

Genotypic ratio- TTVV : TTVv : TTvv

1 : 2 : 1

2. The four patterns of inheritance that deviate considerably from the pattern of inheritance as explained by Mendel are:

(i) Incomplete dominance

(ii) Codominance;

(iii) Polygenic Inheritance;

(vi) linkage

(i) **Incomplete Dominance:** In incomplete donuinance, the heterozygous phenotype is an intermediate blend of the two parental traits rather than showing complete dominance.

Example: Flower colour in *Mirabilis jalapa* (4 O'clock plant). When a red flowered plant (RR) in crossed with a white flowered plant (rr), the F, generation has pink flowers (Rr), showing an intermediate trait. The F_2 generation shows a 1 : 2 : 1 ratio of red : pink : white flowers.

(ii) **Codominance:** In codominance, both alleles in a heterozygous organism are expressed equally, without blending.

Example: AB Blood group in humans. The I^A and I^B alleles in the ABO blood group system are codominant. A person with genotype I^AI^B has blood group AB, where both A and B antigens are expressed equally.

(iii) **Polygenic inheritance:** It occurs when a trait is controlled by multiple genes, leading to continuous variation.

Example: Human skin colour. Human skin colour is determined by multiple genes, each with multiple alleles more donuinant alleles result in darker skin, while more recessive alleles result in lighter skin.

3. (i)

	Pea plant (Violet/ White)	Snapdragon plant (Red/White)
(1)	F_1-All violet (100%)	F_1- All pink flowers (100%)
(2)	F_2 Phenotype - 3 : 1 Violet : white Genotye 1 : 2 : 1 VV : Vv : vv	F_2 Phenotype - 1 : 2 : 1 Red : pink : white, Genotype - 1 : 2 : 1 RR : Rr : rr
(3)	Conclusion : Inheritance of flower colours in pea plant shows violet colour gene is comletely dominant over white colour gene (recessive)/shows dominance.	In snapdragon red colour gene shows incomplete dominance over white colour gene in heterozygous state/ shows incomplete dominance.

(ii) (1) It shows multiple allelism with alleles I^A, I^B, i.

(2) I^A, I^B genes show co-dominance in blood group AB,

(3) I^A and I^B shows complete dominance over i.

4. (i) 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.

(ii) Parents phenotype - Axial violet flower × Axial violet flower

Parents Genotype - AaVv × AaVv

♀/♂	AV	Av	aV	av
AV	AAVV Axial Violet	AAVv Axial Violet	AaVV Axial Violet	AaVv Axial Violet
Av	AAVv Axial Violet	AAvv Axial white	AaVv Axial Violet	Aavv Axial White
aV	AaVV Axial Violet	AaVv Axial Violet	aaVV Terminal Violet	aavv Terminal Violet
av	AaVv Axial Violet	Aavv Axial White	aaVv Terminal Violet	aaVv Terminal White

(Ratio and phenotypes)

Axial violet – 9

Axial white – 3

Terminal violet – 3

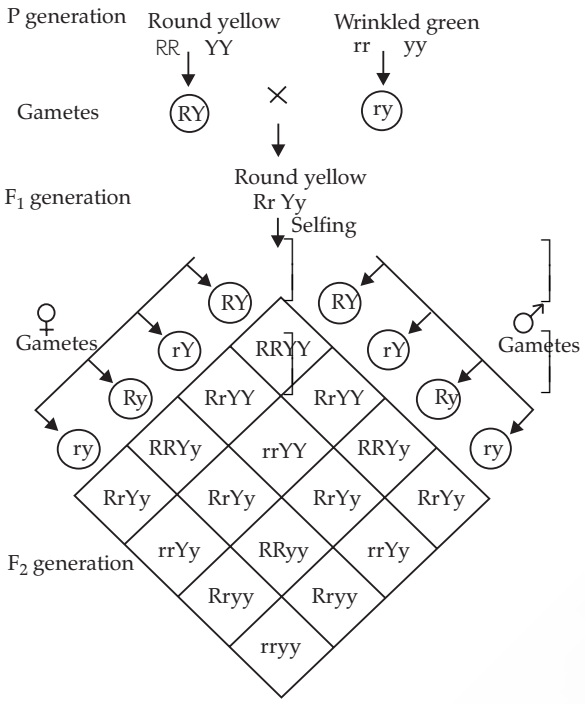
Terminal white – 1

Non-parental recombination in the F_2 progeny shows independent assortment of characters.

5. The plasma membrane of the red blood cell has sugar polymers that protrude from its surface and the kind of sugar is controlled by the gene I present in blood.

- The gene (I) has three alleles I^A, I^B and i
- The alleles I^A and I^B produce a slightly different form of the sugar.
- I^A and I^B are completely dominant over i.
- When I^A and I^B are present as in AB blood group they both express their own type of sugars and show co-dominance.

6. (i)



Phenotypic ratio : round yellow : round green : wrinkled yellow : wrinkled green

9 : 3 : 3 : 1

(ii) The law 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.

7. (i) Phenotype – Tall plant with Yellow seeds
Genotype – TtYy

(ii) **Law of Dominance:** Out of two contrasting traits only one trait will appear in F_1 generation and is called dominant trait while the one which remain unexpressed is called recessive trait.

Law of segregation: In a hybrid union trait simply remain together and segregate at the time of gamete formation.

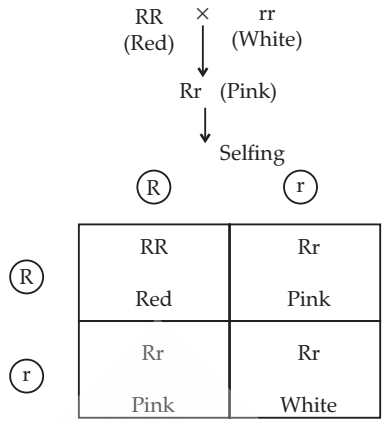
(iii) Phenotypic ratio of F_2

Tall : Tall : Dwarf : Dwarf
yellow : green : yellow : green
9 : 3 : 3 : 1

(iv) Male gametes of F_1-TY, Ty, tY, ty
Female gametes of F_1- TY,Ty,tY,ty

8. In cases where the F_1 progeny shows an intermediate phenotype that does not resemble either parent, it is due to Incomplete Dominance. In this phenomenon, neither allele is completely dominant over the other, resulting in a blending or intermediate expression of the trait.

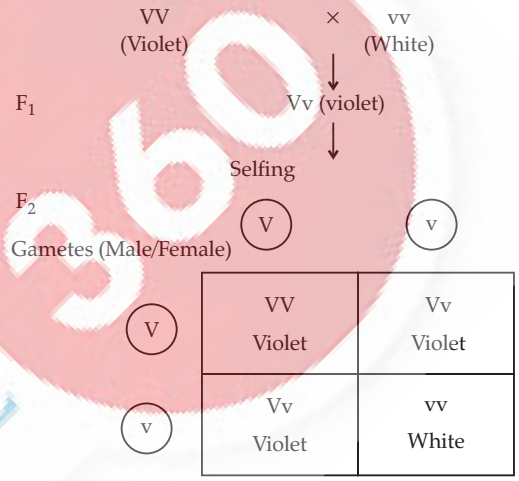
Example: Snapdragon flower / Dog flower / *Antirrhinum*



Phenotype - Red : Pink : White
1 : 2 : 1

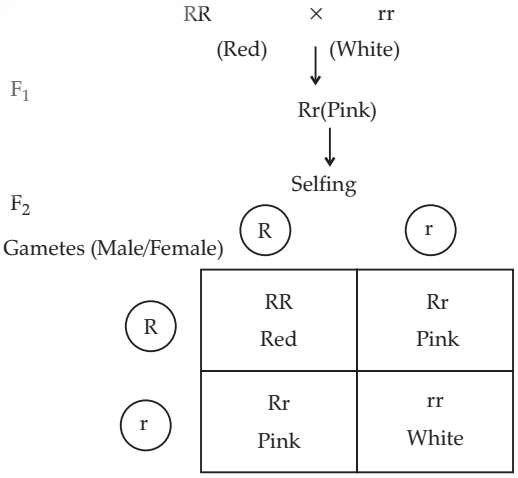
Genotype - RR : Rr : rr
1 : 2 : 1

9. (i) • In case of Pea plant



Phenotype: Violet : White
3 : 1

(ii) In case of Snapdragon flower / Dog flower / *Antirrhinum*



Phenotype - Red : Pink : White
1 : 2 : 1

One Allele is incompletely dominant over the other allele, i.e., Incomplete dominance.

Level - 2 ADVANCED COMPETENCY FOCUSED QUESTIONS

MULTIPLE CHOICE QUESTIONS (MCQs) (1 Mark)

1. Option (B) is correct
   Explanation: Thalassemia is an autosomal recessive disorder. For a child to be born with thalassemia, they must inherit two copies of the recessive allele (i.e., be homozygous recessive: tt). This is only possible if both parents are carriers (heterozygous: Tt)
2. Option (C) is correct
   Explanation: The ABO blood group system is determined by multiple alleles:
   I^A (for A blood group)
   I^B (for B blood group)
   i (for O blood group)
   Blood group O means the genotype is ii (homozygous recessive). So, for a child to have blood group O, both parents must carry the 'i' allele.
3. Option (A) is correct
   Explanation: In Mendel's dihybrid cross, he studied two traits:
   Seed shape: Round (R) is dominant over Wrinkled (r)
   Seed color: Yellow (Y) is dominant over Green (y)
   The cross:
   RRYY × rryy (Round Yellow × Wrinkled Green)
   → F_1 genotype: RrYy (all round yellow)
   F_2 Generation (RrYy × RrYy):
   Using the dihybrid Punnett square, the phenotypic ratio is:

Phenotype	Fraction
Round Yellow	$\frac{9}{16}$
Round Green	$\frac{3}{16}$
Wrinkled Yellow	$\frac{3}{16}$
Wrinkled Green	$\frac{1}{16}$

   So, the proportion of Round and Yellow seeds is $\frac{9}{16}$.
4. Option (C) is correct
   Explanation: X-linked recessive traits are more common in males because males have only one X chromosome (XY), so one copy of the mutant gene causes the condition. Females have two X chromosomes (XX), so both copies must be mutant to express the trait—making it rarer in females.
   The trait is not passed from father to son, because fathers pass their Y chromosome to sons (not X). If a trait is X-linked, a father cannot pass his X-linked gene to his son.
5. Option (C) is correct
   Explanation: Recessive traits are only expressed when an individual inherits two copies of the mutant allele (one from each parent). If both parents are heterozygous carriers, they may appear normal but can pass on the recessive allele to their children. The trait may skip one or more generations if no child inherits two recessive alleles. Siblings can express the trait if both inherit the recessive allele.
6. Option (C) is correct
   Explanation: Multiple allelism refers to the presence of more than two alleles for a gene in the population. However, each individual can have only two alleles (one from each parent).
   The ABO blood group system in humans is determined by a gene with three alleles:
   I_A (for A antigen)
   I_B (for B antigen)
   i (for O, no antigen)
   These alleles can combine to produce four blood types: A, B, AB, and O.This is a classic example of multiple alleles and co-dominance (e.g., I^AI^B gives AB blood type).
7. Option (C) is correct
   Explanation: Hemophilia is an X-linked recessive disorder. This means that the gene causing hemophilia is on the X chromosome. Males (XY) only need one affected X to have the disease. Females (XX) need two affected Xs to have the disease (but one makes them carriers).
8. Option (B) is correct
   Explanation: Gregor Mendel's work on pea plants laid the foundation for modern genetics. However, his work was largely ignored during his lifetime (mid-1800s) and was only rediscovered around 1900 by scientists like Hugo de Vries, Carl Correns, and Erich von Tschermak. Mendel used a quantitative and mathematical approach, applying statistics to biology—an approach that was ahead of its time. The scientific community of that era was not ready to accept mathematical explanations in biology. Also, cell biology and chromosomal theory were not developed enough then to relate his findings to chromosomes and meiosis.

ASSERTION-REASON QUESTIONS (1 Mark)

1. Option (A) is correct
   Explanation: Assertion is true. In a monohybrid cross, the F_2 generation shows a phenotypic ratio of 3 : 1 — 3 individuals show the dominant trait, and 1 shows the recessive trait.
   Reason is also true. Law of Segregation (Mendel's First Law) states that the two alleles of a gene separate during gamete formation, ensuring each gamete receives only one allele. This separation and recombination during fertilisation explain the 3 : 1 phenotypic ratio.

2. Option (A) is correct
Explanation: Assertion is correct. Colour blindness is a sex-linked recessive disorder that is more common in males than females.
Reason is also true. Males have only one X chromosome (XY), so a single recessive allele for colour blindness on the X chromosome will express the trait (no second X to mask it).
In contrast, females (XX) would need two copies of the recessive allele to be affected.

3. Option (A) is correct
Explanation: Assertion is true. Down's syndrome is a genetic disorder caused by the presence of an extra copy of chromosome 21, i.e., trisomy 21. Individuals have 47 chromosomes instead of the normal 46.
Reason is also true. The cause of this extra chromosome is non-disjunction during meiosis, which means the chromosomes fail to separate properly during gamete formation. This results in a gamete carrying two copies of chromosome 21, leading to trisomy after fertilisation.

4. Option (C) is correct
Explanation: Assertion is true. A person with AB blood group has both A and B antigens on the surface of red blood cells. This is only possible when their genotype is I^AI^B, meaning they inherit the I^A allele from one parent and the I^B allele from the other.
Reason is false. The alleles I^A and I^B do not show incomplete dominance. They show co-dominance, because both alleles are expressed equally in the phenotype (i.e., both A and B antigens are produced).

VERY SHORT ANSWER TYPE QUESTIONS (2 Marks)

1. **(i)**

Monohybrid cross	Test cross
(i) It takes place between two parents of any genotype.	One parent is necessarily homozygous recessive for a trait.
(ii) Monohybrid cross between known parents is done to determine the pattern of inheritance of one single gene.	Test cross is done to determine the unknown genotype of one individual / parent of the cross.

(Any one)

(ii) All heterozygous dominant, if the unknown genotype is homozygous dominant.
1:1 (heterozygous dominant: homozygous recessive) if the unknown genotype is heterozygous dominant.

2. **(i)** Since colour blindness is an X-linked recessive disorder, a colour blind male will share the normal Y chromosome and the colour blind mother will share a chromosome with the colour blindness trait to the child, making him a colour blind.
(ii) A colour blind male will transfer the normal Y chromosome and the mother with normal vision will transfer chromosome with the normal vision trait to the child so the child will not be a carrier too.

3. **Two differences**
(1) Klinefelter's syndrome is caused due to the presence of an additional copy of the X chromosome whereas Turner's syndrome is caused due to the absence of an X chromosome.
(2) Klinefelter's syndrome occurs in both genders whereas Turner's syndrome is found in females.
Two similarities:
(1) Both are chromosomal disorders related to the sex chromosomes.
(2) In both disorders, the individuals are sterile.

4. **(1)** May or may not have an impact on reproductive features.
(2) Short stature with a small round head.

5. X-linked recessive disorder
The affected mother has all affected male children but female children are not affected.

SHORT ANSWER TYPE QUESTIONS (3 Marks)

1. (i) Haemophilia is an X-linked recessive disorder. Let X^H = normal allele
Let X^h = haemophilia allele
Since the woman has a family history but is not affected, she is likely a carrier (X^HX^h).
The man is normal (X^HY).
Punnett square:

	X^H (father)	Y (father)
X^H (mother)	X^HX^H	X^HY
X^h (mother)	X^HX^h	X^hY (haemophilic son)

Probability of son being haemophilic (X^hY) = 1 out of 2 = 50%

(ii) Genetic counselling can:
(1) Assess the risk of transmitting haemophilia to children.
(2) Explain inheritance patterns and probability of occurrence.
(3) Guide couples on prenatal diagnostic options (like amniocentesis).
(4) Offer advice on reproductive choices and psychological support.

(iii) Females have two X chromosomes. They must inherit two copies of the defective gene (X^hX^h) to be affected. Usually, carriers (X^HX^h) are not affected due to the presence of one normal allele. Males (XY) need only one defective allele (X^hY) to be affected. Hence, haemophilia is rare in females but common in males.

2. (i) Thalassemia is an autosomal recessive genetic disorder. It occurs when a child inherits two defective alleles (one from each parent) for the gene controlling haemoglobin production. If a

person inherits only one defective allele, they are a carrier. It does not show symptoms but can pass it on.

(ii) If both parents are carriers, there is a 25% chance with each pregnancy that the child will inherit both defective genes and suffer from Thalassemia major. Testing helps assess the risk of recurrence in the next child and the carrier status of each parent. It enables informed decision-making and early medical guidance.

(iii) Thalassemia can be prevented using:

(1) Genetic counselling – to understand the risks and options.

(2) Carrier screening – of both parents before conception.

(3) Prenatal diagnosis – using techniques like: Chorionic villus sampling (CVS) and Amniocentesis. These detect whether the fetus is affected.

(4) Assisted Reproductive Technologies (ART) – such as IVF with Preimplantation Genetic Diagnosis (PGD) to ensure only healthy embryos are implanted.

3. (i) Down's Syndrome is caused by a genetic abnormality called trisomy 21. It occurs when an individual has three copies of chromosome 21 instead of the normal two. This results from non-disjunction during meiosis, where chromosomes fail to separate properly in the egg or sperm.

(ii) As maternal age increases, especially after 35 years, the chances of non-disjunction events also increase. Older eggs are more prone to errors during cell division, making women over 40 more likely to have a child with chromosomal abnormalities, such as Down's Syndrome.

(iii) The two characteristic symptoms of Down's Syndrome are:

(1) Distinct facial features – such as a flat facial profile, upward slanting eyes, and a small nose.

(2) Intellectual disability – ranging from mild to moderate developmental delays in mental and motor skills.

4. (i) A person with blood group O has the genotype ii. This means they carry two recessive alleles for the ABO blood group gene.

(ii) No, a person with O blood group cannot donate to an AB person in terms of plasma compatibility, though they can donate red blood cells. Blood group O is a universal donor for red blood cells because it has no A or B antigens on the surface of RBCs, which prevents agglutination. However, the plasma of O group has both anti-A and anti-B antibodies, which may react with the recipient's blood if whole blood is transfused. So, for red blood cell transfusion only, a person with O blood can donate to AB, but for whole blood, it is not safe.

(iii) The ABO blood group system is an example of multiple allelism and co-dominance.

Multiple allelism: There are three alleles – I^A, I^B, and i.

Co-dominance: Alleles I^A and I^B are co-dominant (e.g., AB blood group expresses both). Recessiveness: Allele i is recessive to both I^A and I^B.

5 (i) Sons inherit their X chromosome from the mother and Y from the father. The mother is X^NX^C, so there's a 50% chance she passes the X^C to the son. The probability that their son will be colour blind is 50%.

(ii) Daughters inherit one X from each parent. The father provides X^N, and the mother can give either X^N or X^C. So, there's a 50% chance the daughter will be X^NX^C (carrier).

(iii) Colour blindness is an X-linked recessive disorder. It only expresses in females when they have two copies of the defective gene (X^CX^C). A single normal allele (X^N) is enough to mask the defect in females, making them carriers, not affected.

CASE BASED QUESTIONS (4 Mark)

1. (i) Option (D) is correct

Explanation: Where a cross is performed between a homozygous dominant male (AABB) and a homozygous recessive female (aabb), the offspring (F_1 generation) will all have the genotype AaBb. Since A & B are dominant, the phenotype of the F_1 offspring will be determined by number of dominant allele.

(ii) Option (B) is correct

Explanation: In wheat, the colour of the kernel is controlled with the help of two genes, the red colour kernel in wheat is dominant than the white colour.

(iii) Option (C) is correct

Explanation: When red and white color kernels of wheat were crossed, the F_1 generation was observed to be red and even the combination of both red and white pairs. When the cross was done in between F_1 generation, the F_2 progeny had 5 different types of phenotypic ratio and it was found to be 1 : 4 : 6 : 4 : 1.

Parents: AABB (Red) x aabb (White)

Gametes: AB, ab

F_1 generation: AaBa

Gamete: AB, Ab, aB, ab

	AB	Ab	aB	ab
AB	AABB (dark red)	AABb (Medium red)	AaBB (Medium red)	AaBb (Intermediate red)
Ab	AABb (Medium red)	AAbb (Intermediate red)	AaBb (Intermediate red)	Aabb (Light red)
aB	AaBB (Medium red)	AaBb (Intermediate red)	aaBB (Intermediate red)	aaBb (Light red)
ab	AaBb (Intermediate red)	Aabb (Light red)	aaBb (Light red)	aabb (white)

(iv) Option (B) is correct

Explanation: The alleles for kernel color in wheat primarily exhibit polygenic inheritance. This means that multiple genes contribute to the variation in kernel color, resulting in a range of phenotypes.

2. (i) Sex linked disorder

More males are affected in the family as males have only one X chromosome which if affected expresses.

(ii) Recessive disorder.

(iii) C -XX^c; D- XX^c; H- XX^c

(iv) (a) Probability 0%

OR

(b) Probability-50%

3. (i) Gain or loss of chromosome due to failure of segregation of chromatids during cell division cycle is known as aneuploidy.

(ii) 22 + XY, 22 + 0

(iii) (a) **Disorder:** Klinefelter's syndrome

Symptoms: Gynaecomastia/ feminine development, sterile individual, tall stature, overall masculine development.

OR

(b) Down's Syndrome.

Symptoms: Short statured with small round head, furrowed tongue, partially open mouth, broad palm with palm crease, physical/psychomotor/mental retardation, flat back of head, loops on finger tips, congenital heart disease, big and wrinkled tongue, broad flat face. (Any two)

LONG ANSWER TYPE QUESTIONS (5 Marks)

1. (i) Genetic counselling is a communication process where trained professionals provide information, support, and guidance to individuals or families who have, or may be at risk for, genetic disorders.

Benefits:

(1) Helps couples understand genetic risks and chances of passing on a disorder to offspring.

(2) Provides information on genetic testing options (e.g., carrier testing, prenatal testing).

(3) Aids in decision-making regarding reproduction and family planning.

(4) Offers emotional support to cope with the implications of genetic conditions.

(ii) In autosomal recessive inheritance, a person must inherit two copies of a mutated gene (one from each parent) to express the disorder. If both parents are carriers (heterozygous), they do not show symptoms but can pass the gene to their children.

Thalassemia caused by a mutation in genes controlling haemoglobin production. If a child inherits the defective gene from both parents, they develop thalassemia major. If only one copy is inherited, the child becomes a carrier (thalassemia minor).

(iii) Pedigree analysis is a chart showing the occurrence of a trait across several generations in a family. It helps:

(1) Determine inheritance patterns (dominant, recessive, X-linked, etc.).

(2) Identify carriers and individuals at risk.

(3) Assist in predicting genetic outcomes for future offspring.

2. (i) Sickle cell anaemia is an autosomal recessive genetic disorder caused by a mutation in the HBB gene on chromosome 11. This gene codes for the β-globin chain of haemoglobin. A point mutation replaces adenine (A) with thymine (T), changing the codon GAG (glutamic acid) to GTG (valine). The mutant allele is denoted as Hb^S, while the normal allele is Hb^A.

(ii) Both parents are heterozygous carriers (Hb^AHb^S), meaning they carry one normal and one defective allele. Since sickle cell anaemia is recessive, the presence of one normal allele prevents the expression of the disease. Thus, carriers are phenotypically normal but can pass the defective gene to their children.

(iii) In sickle cell anaemia, the mutated β-globin chain causes haemoglobin molecules to polymerise under low oxygen conditions. This leads to the formation of rigid, sickle-shaped RBCs instead of the normal round, flexible shape. These abnormal RBCs:

(1) Have reduced oxygen-carrying capacity, leading to fatigue and weakness.

(2) Tend to block capillaries, causing pain, organ damage, and anaemia.

(3) Have a shorter lifespan, leading to chronic anaemia.

(iv) Heterozygous individuals (Hb^AHb^S) are resistant to malaria caused by *Plasmodium falciparum*. The parasite cannot survive well in the sickle-shaped red blood cells, offering a survival advantage in regions where malaria is common.

3. (i) The chromosomal abnormality responsible for Down's Syndrome is trisomy of chromosome 21 — i.e., the presence of an extra copy of chromosome 21.

(ii) Down's Syndrome occurs due to non-disjunction during gamete formation. In this process, chromosome 21 fails to separate properly during meiosis. This results in a gamete with two copies of chromosome 21. After fertilisation, the zygote ends up with three copies (trisomy 21) instead of the normal two.

(iii) Common physical and mental features of Down's Syndrome include:
- **(1)** Flat facial profile and upward slanting eyes
- **(2)** Mental retardation or delayed cognitive development
- **(3)** Poor muscle tone and short stature

(iv) The risk of non-disjunction increases with maternal age, especially after 35 years. This is because older eggs are more prone to meiotic errors, increasing the chances of trisomy.

4. (i) ABO blood group is controlled by a single gene with three alleles, I^A, I^B, and i
I^A and I^B are co-dominant, and both are dominant over i.

(ii) The ABO blood group exhibits:
- **(1)** Multiple allelism (presence of more than two alleles — I^A, I^B, i)
- **(2)** Co-dominance (I^A and I^B both expressed in AB blood group)
- **(3)** Simple dominance (I^A and I^B are dominant over i)

(iii) Individuals with O blood group have genotype ii, and their RBCs have no A or B antigens. So, they can donate blood to all other blood groups (A, B, AB, O) without causing an immune response. However, they can only receive blood from O group.