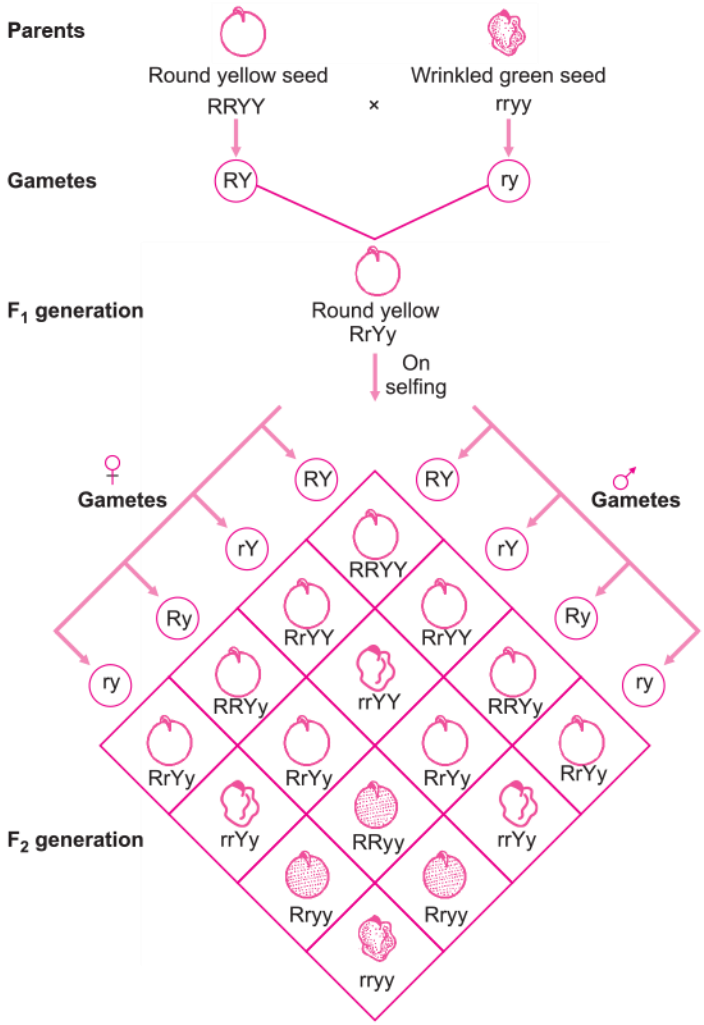
Long Answer Questions (PYQ)

[5 Marks]

Q.1. Work out a typical Mendelian dihybrid cross and state the law that he derived from it.

Ans.



Phenotypic ratio–	Round yellow			:	Round green			:	Wrinkled yellow			:	Wrinkled green			
		9		:		3		:		3		:		1		
Genotypic ratio–	1	:	2	:	2	:	4	:	1	:	2	:	1	:	2	: 1

Results of a dihybrid cross where the two parents differed in two pairs of contrasting traits: seed colour and seed shape For the dihybrid cross Mendel derived the law of Independent Assortment: It states that when two pairs of traits are combined in a hybrid, segregation of one pair of character is independent of the other pair of characters.

Q.2.

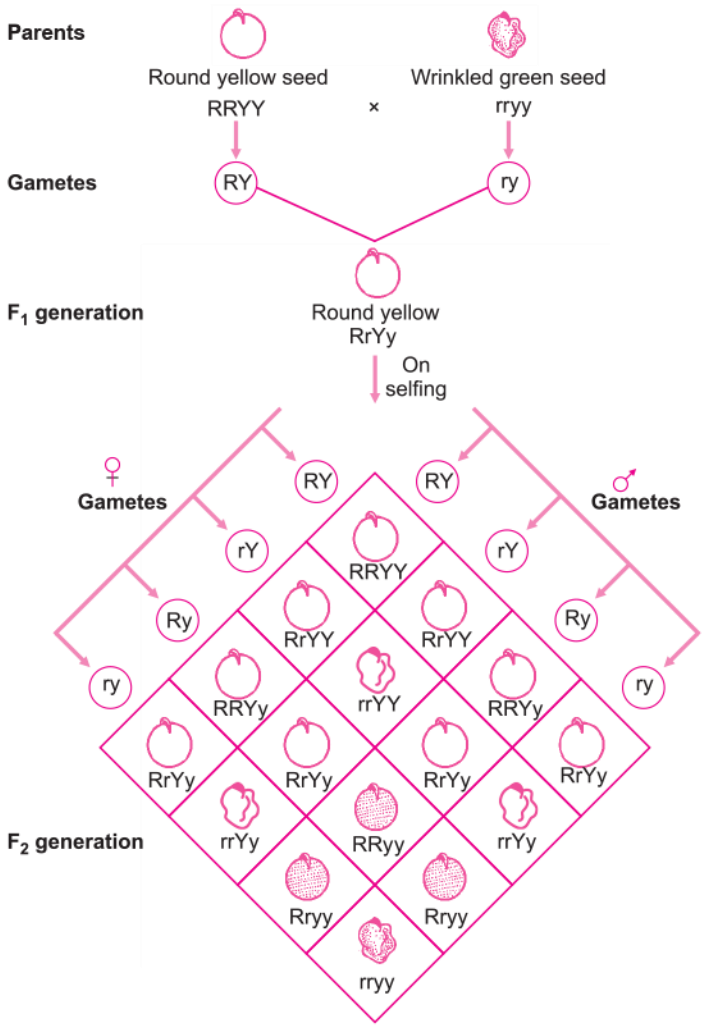
(a) State the law of independent assortment.

(b) Using Punnett square demonstrate the law of independent assortment in a dihybrid cross involving two heterozygous parents.

Ans.

a. According to this law the two factors of each character assort or separate out independent of the factors of other characters, at the time of gamete formation and get randomly rearranged in the offsprings, producing both parental and new combinations of characters.

b.



Phenotypic ratio–	Round yellow			:	Round green			:	Wrinkled yellow			:	Wrinkled green			
		9		:		3		:		3		:		1		
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Results of a dihybrid cross where the two parents differed in two pairs of contrasting traits: seed colour and seed shape

Q.3.

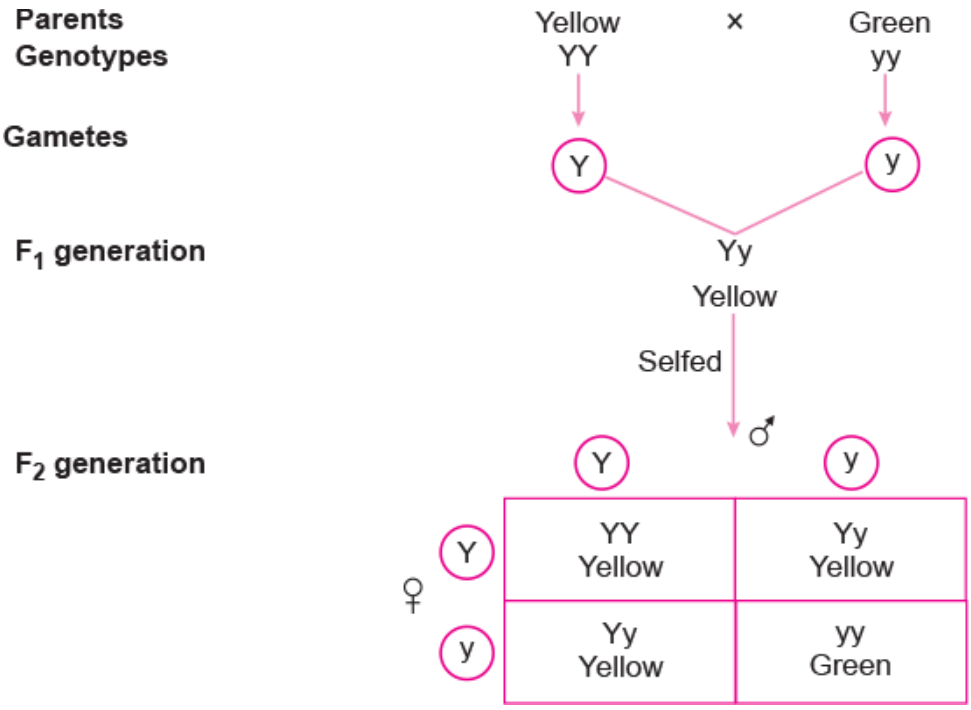
a. Explain a monohybrid cross taking seed coat colour as a trait in Pisum sativum. Work out the cross up to F2 generation.

b. State the laws of inheritance that can be derived from such a cross.

c. How is the phenotypic ratio of F2 generation different in a dihybrid cross?

Ans.

(a)



F2 Phenotypic ratio = 3 : 1; F2 Genotypic ratio = 1 : 2 : 1

(b)

* Law of Dominance: In a contrasting pair of factors, one member of the pair dominates (dominant) the other (recessive).

* Law of Segregation: Factors or allele of pair separate from each other such that gamete receives only one of the two factors.

(c) Phenotypic ratio of F2 in monohybrid cross is 3 : 1 whereas in a dihybrid cross the phenotypic ratio is 9 : 3 : 3 : 1.

Q.4.

(a) A true breeding homozygous pea plant with green pods and axial flowers as dominant characters, is crossed with a recessive homozygous pea plant with

yellow pods and terminal flowers. Work out the cross up to F2 generation giving the phenotypic ratios of F1 and F2 generation respectively.

(b) State the Mendelian principle which can be derived from such a cross and not from monohybrid cross.

Ans.

(a)

Parents — Green axial GGAA × Yellow terminal ggaa

Gametes — GA, Ga

F_1 generation — GgAa × GgAa—(Hybrid) green axial

Selfing

F_2 generation

	GA	Ga	gA	ga
GA	GGAA Green axial	GGAa Green axial	GgAA Green axial	GgAa Green axial
Ga	GGAa Green axial	GGaa Green terminal	GgAa Green axial	Ggaa Green terminal
gA	GgAA Green axial	GgAa Green axial	ggAA Yellow axial	ggAa Yellow axial
ga	GgAa Green axial	Ggaa Green terminal	ggAa Yellow axial	ggaa Yellow terminal

Phenotypic ratio

Green axial	:	Green terminal	:	Yellow axial	:	Yellow terminal
9	:	3	:	3	:	1

(b) From the above cross law of independent assortment can be derived which states that when two pairs of traits are combined in a hybrid, segregation of one pair of character is independent of the other pair of characters.

Q.5.

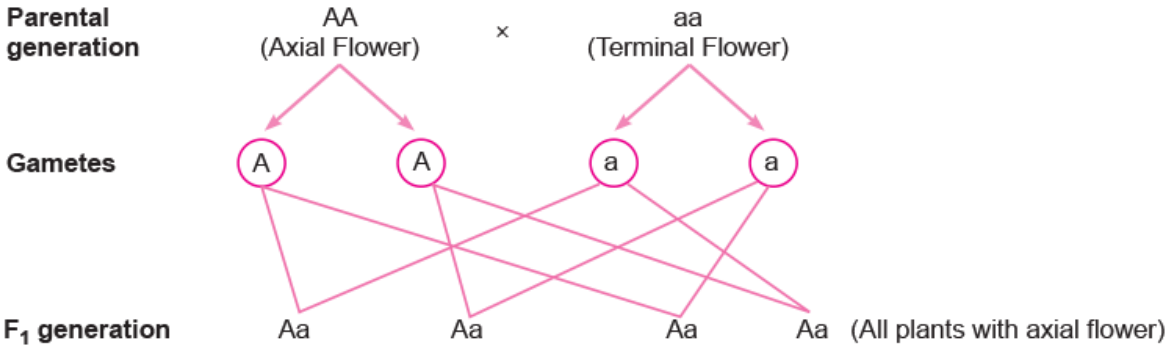
(a) A pea plant bearing axial flowers is crossed with a pea plant bearing terminal flowers. The cross is carried out to find the genotype of the pea plant bearing axial flowers. Work out the cross to show the conclusions you arrive at.

(b) State the Mendel's law of inheritance that is universally acceptable.

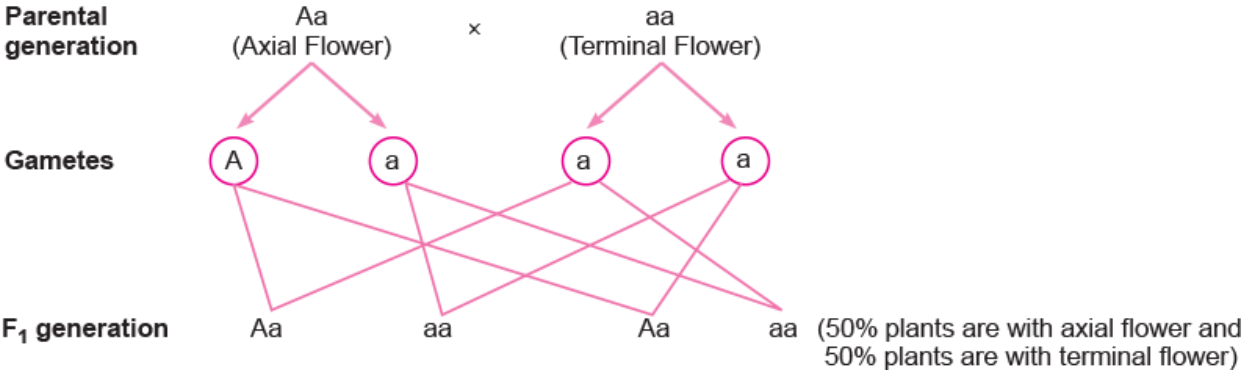
Ans.

(a) If the plant is homozygous for the dominant trait.

(*i*) If the plant is homozygous for the dominant trait.



(*ii*) If the plant is heterozygous for the dominant trait.



Conclusion: If all progeny show axial flowers (dominant) the plant is homozygous (AA), If 50% of progeny show axial flower (Dominant) and 50% terminal flower (Recessive) the plant is heterozygous.

(b) Law of Segregation is universally accepted. It states that allelic pair segregate (separates) during gamete formation.

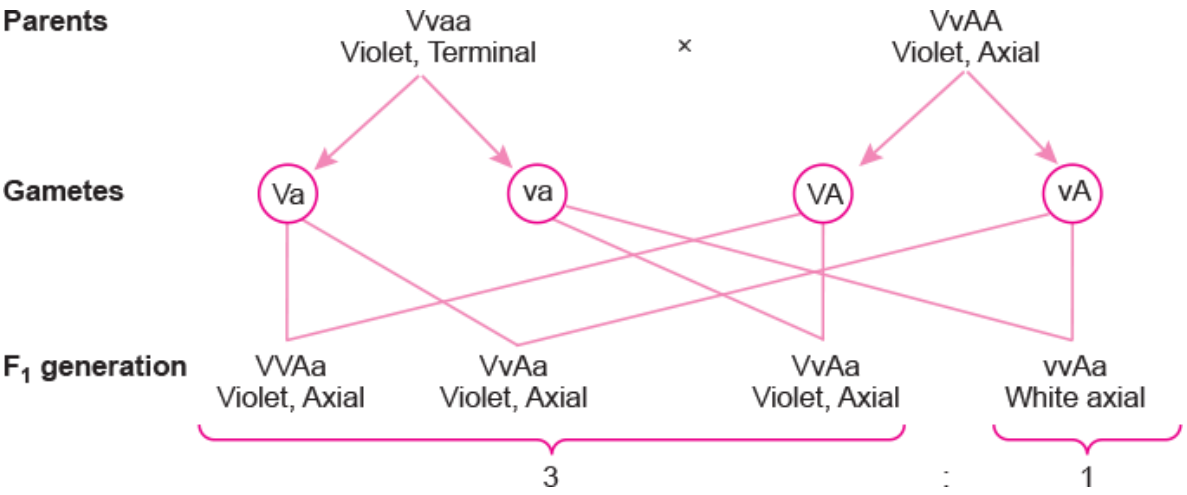
Q.6.

(a) A garden pea plant bearing terminal, violet flowers, when crossed with another pea plant bearing axial, violet flowers, produced axial, violet flower and axial, white flowers in the ratio of 3 : 1. Work out the cross showing the genotypes of the parent pea plants and their progeny.

(b) Name and state the law that can be derived from this cross and not from a monohybrid cross.

Ans.

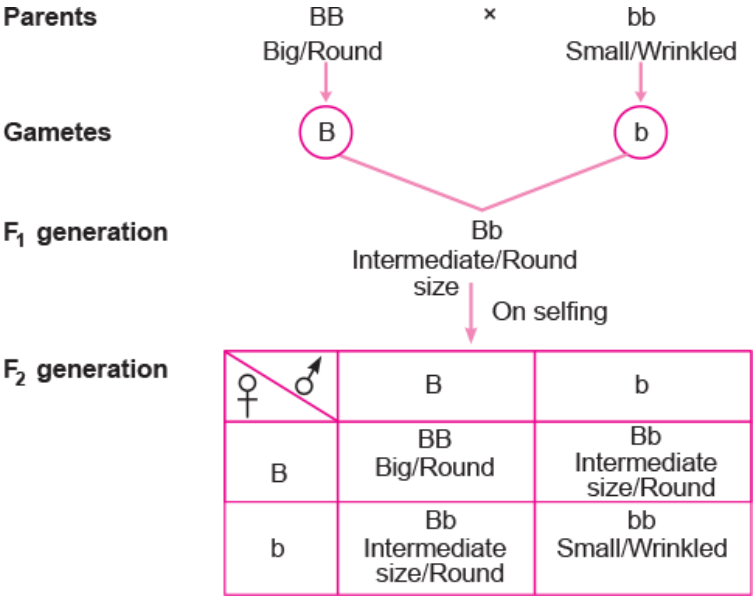
(a)



(b) Law of Independent Assortment: When two pairs of traits are combined in a hybrid, segregation of one pair of characters is independent of the other pair of character.

Q.7. What is the inheritance pattern observed in the size of starch grains and seed shape of Pisum sativum? Workout the monohybrid cross showing the above traits. How does this pattern of inheritance deviate from that of Mendelian law of dominance?

Ans. A single gene controls the size of the starch grains and seed shape of Pisum sativum.



♀ \ ♂	B	b
B	BB Big/Round	Bb Intermediate size/Round
b	Bb Intermediate size/Round	bb Small/Wrinkled

With respect to size of starch grains it shows 3 forms-big, Intermediate and small as in incomplete dominance but with respect to seed shape it follows Mendelian law of Dominance showing either round or wrinkled.

Q.8. You are given a red flower-bearing pea plant and a red flower-bearing snapdragon plant. How would you find the genotypes of these two plants with

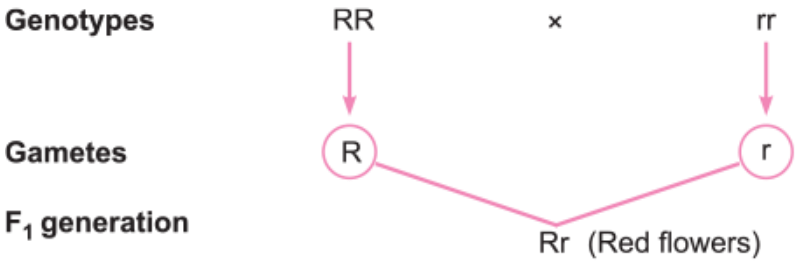
respect to the colour of the flower? Explain with the help of crosses. Comment upon the pattern of inheritance seen in these two plants.

Ans. A test cross is required to find out the genotype of both the plants.

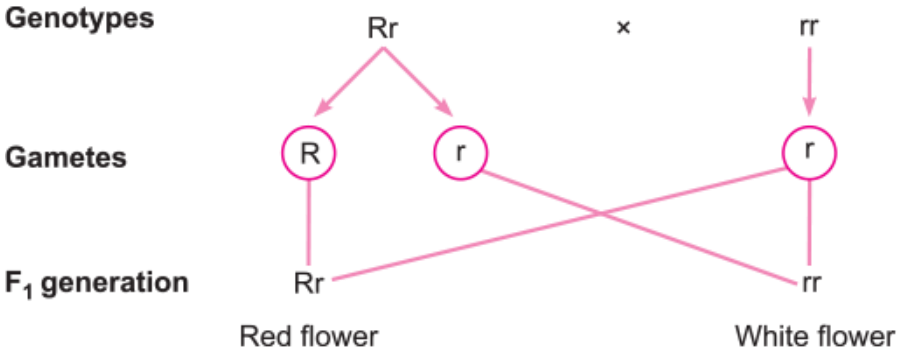
(a)

Garden pea

(Homozygous red)



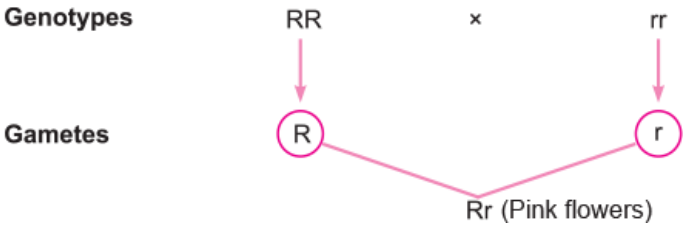
(Heterozygous red)



If the F_1 generation plants have all red flowers, the genotype of the parent plant will be homozygous dominant and if the F_1 generation plants have red and white flowers in the ratio of 1 : 1, then the genotype of the parent plant is heterozygous dominant. This inheritance follows the Mendelian law of dominance.

(b)

In snapdragon:



The parent plant will be homozygous for flower colour because a heterozygous plant will have pink flowers due to the phenomenon of incomplete inheritance.

Q.9. With the help of one example each, provide genetic explanation for the following observations:

Q. F_1-generation resembles both the parents.

Ans.

F_1 generation resembles both the parents: This happens in the case of co-dominance where both alleles express themselves fully in heterozygous condition. For example: different types of red blood cells determine ABO blood grouping in human beings.

Co-dominance

* The alleles which are able to express themselves independently, even when present together are called co-dominant alleles and this biological phenomenon is called co-dominance.For example, ABO blood grouping in humans.

* ABO blood groups are controlled by gene I. Gene I has three alleles IA, IB and IO/i.

* IA and IB produce RBC surface antigens sugar polymer A and B, respectively, whereas i does not produce any antigen.

* IA and IB are dominant over i hence IA and IB are dominant alleles and i is recessive allele as in IAi and IBi.

* When IA and IB are present together, both express equally and produce the surface antigens A and B, hence show co-dominance.

* Since humans are diploid, each person possesses any two of the three 'I' gene alleles, resulting into six different genotypic combinations and four phenotypic expressions.

Table showing the genetic basis of blood groups in human population

Allele fromParent 1	Allele fromParent 2	Genotype ofoffspring	Blood groups ofoffspring
I^A	I^A	$I^A I^A$	A
I^A	I^B	$I^A I^B$	AB
I^A	i	$I^A i$	A
I^B	I^B	$I^B I^B$	B
I^B	i	$I^B i$	B
i	i	$i\ i$	O

Q. F1-generation does not resemble either of the parents.

Ans.

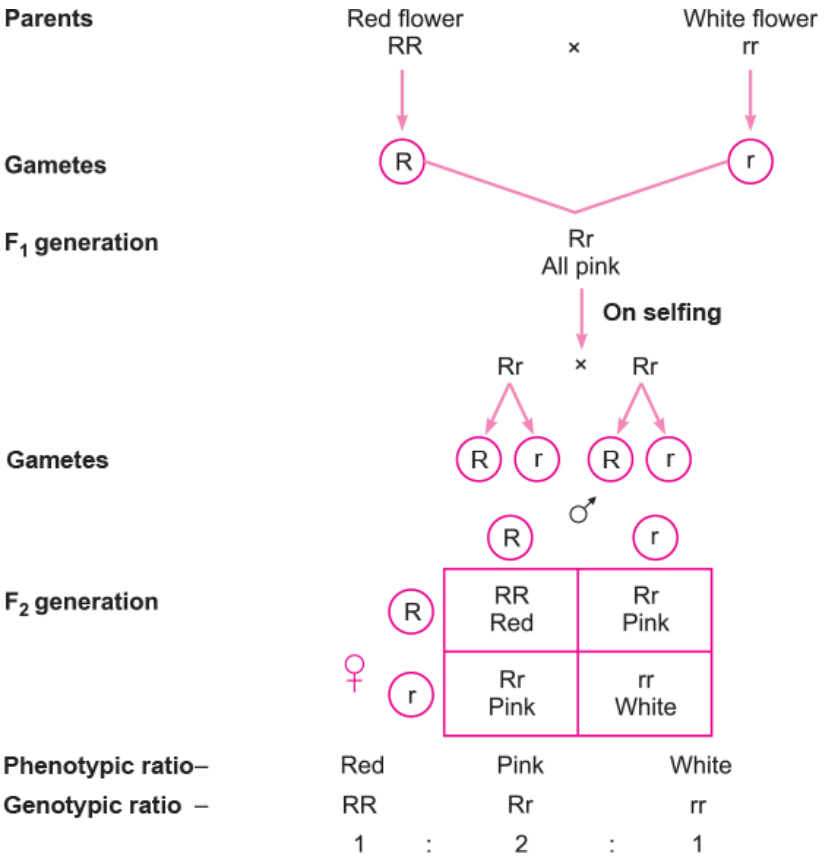
F_1 generation does not resemble either of the parents: In incomplete dominance, a heterozygous organism carrying two alleles wherein one is dominant and the other one is recessive, (e.g., Rr). Hence, the heterozygote (Rr) will have an intermediate phenotype and will not resemble any parent.

Incomplete Dominance

It is a phenomenon in which the F_1 hybrid exhibits characters intermediate of the parental genes.

Here, the phenotypic ratio deviates from the Mendel's monohybrid ratio.

It is seen in flower colours of Mirabilis jalapa (4 o' clock plant) and Antirrhinum majus (snapdragon), where red colour is due to gene RR, white colour is due to gene rr and pink colour is due to gene Rr.



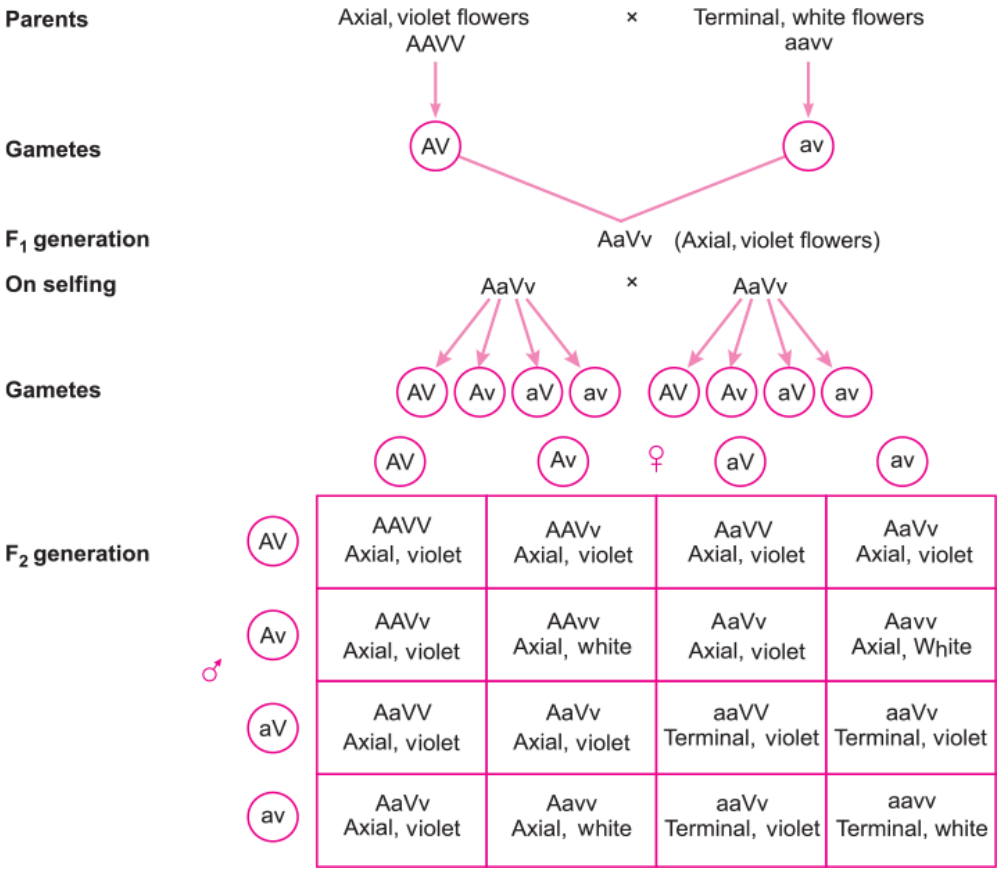
Q.10. A true breeding pea plant homozygous for axial violet flowers is crossed with another pea plant with terminal white flowers (aavv).

a. What would be the phenotype and genotype of F1 and F2 generations?

b. Give the phenotypic ratio of F2 generation.

c. List the Mendel's generalisations that can be derived from the above cross

Ans.



(a) Phenotype of F_1 generation—All axial, violet flowers.

Genotype of F_1 generation—AaVv.

(b) Phenotypic ratio of F_2 generation:

Axial violet flowers	:	Axial white flowers	:	Terminal violet flowers	:	Terminal white flowers
9		3		3		1

(c) Law of Independent Assortment: This law states that the different factors or allelomorphic pair in gametes and zygotes assort themselves and segregate independently of one another.

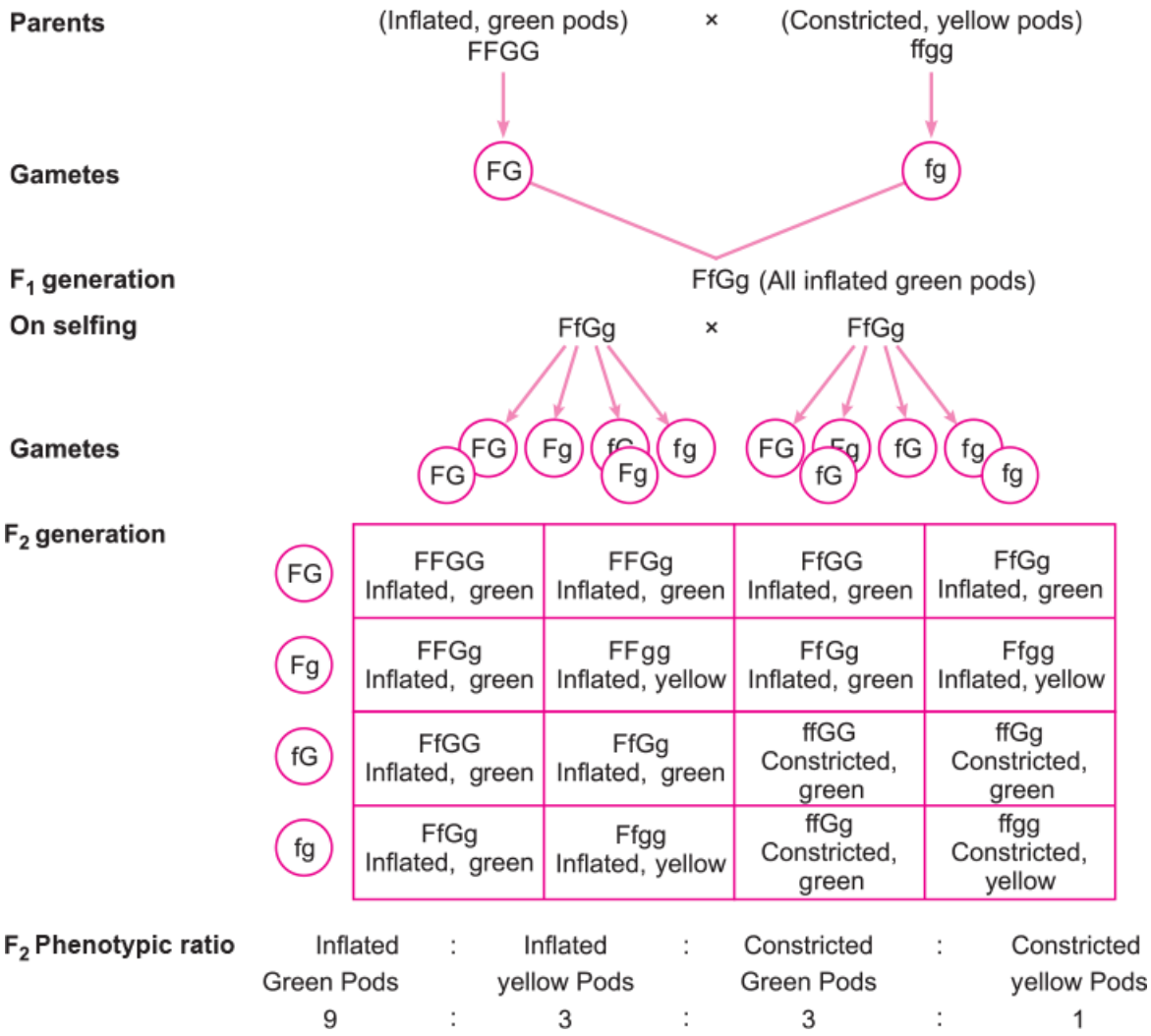
Q.11.

(a) A true breeding pea plant, homozygous for inflated green pods is crossed with another pea plant with constricted yellow pods (ffgg). What would be the phenotype and genotype of F1 and F2 generations? Give the phenotype ratio of generation.

(b) State the generalisation proposed by Mendel on the basis of the above-mentioned cross.

Ans.

(a)



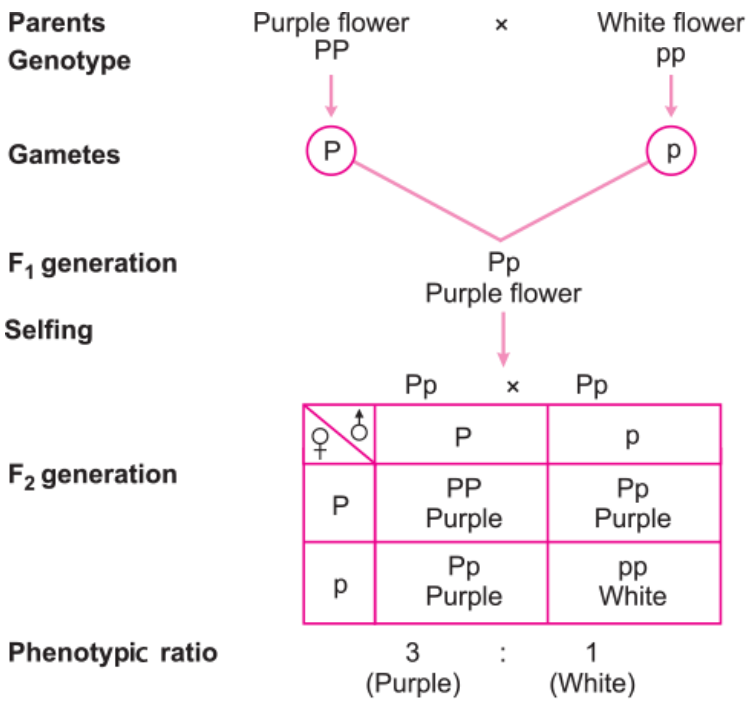
F_2 Phenotypic ratio

Inflated Green Pods	:	Inflated yellow Pods	:	Constricted Green Pods	:	Constricted yellow Pods
9	:	3	:	3	:	1

(b) Mendel's Law of Independent Assortment: This law states that the different factors or allelomorphic pair in gametes assort themselves and segregate independently of one another.

Q.12. Inheritance pattern of flower colour in garden pea plant and snapdragon differs. Why is this difference observed? Explain showing the crosses up to F_2 generation.

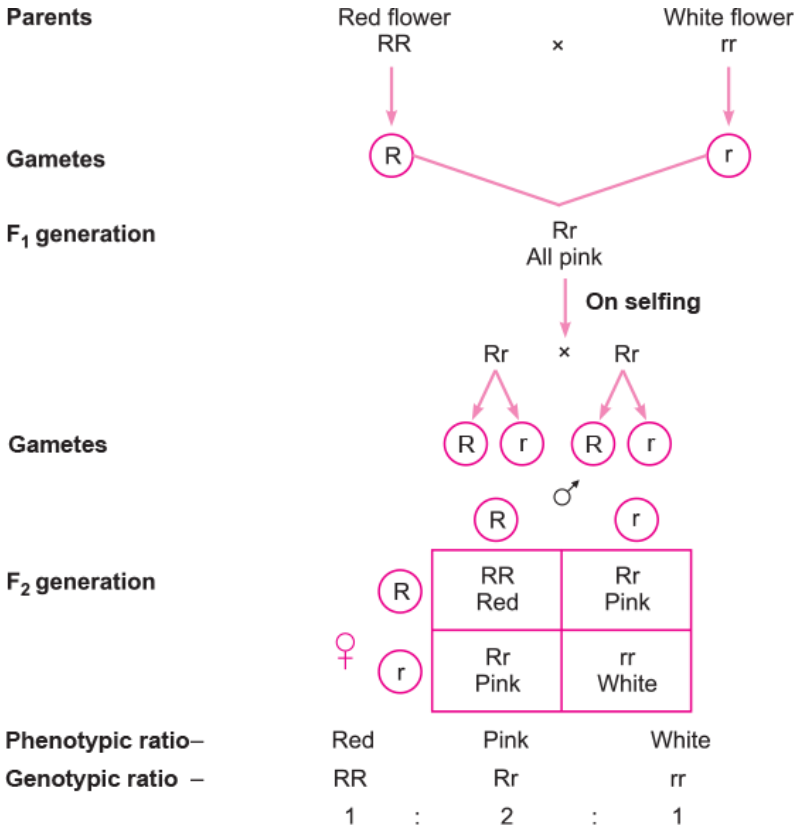
Ans. Inheritance pattern of flower colour in garden pea follows principle of dominance whereas inheritance in snapdragon shows incomplete dominance.



Phenotypic ratio—3 : 1

Genotypic ratio—1 : 2 : 1.

Inheritance of flower colour in snapdragon:



Phenotypic ratio—1 : 2 : 1

Genotypic ratio—1 : 2 : 1.

Q.13.

(a) Write the conclusions Mendel arrived at on dominance of traits on the basis of monohybrid crosses that he carried out in pea plants.

(b) Explain why a recessive allele is unable to express itself in a heterozygous state.

Ans.

(a) Mendel concluded that:

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

(ii) Factors occur in pair.

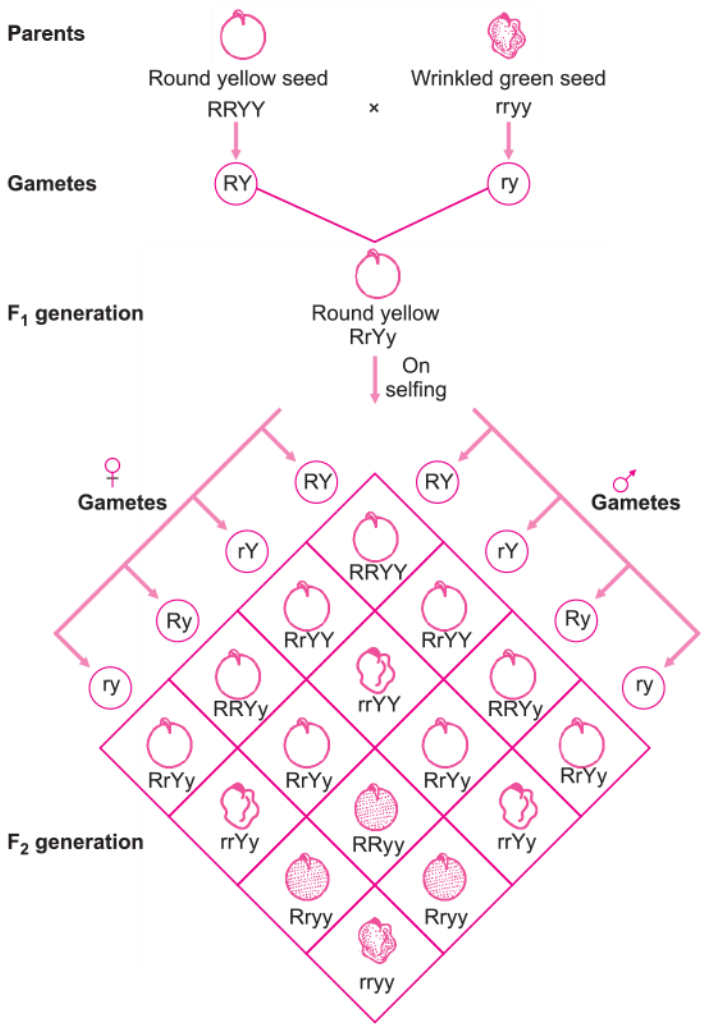
(iii) In a dissimilar pair of factors one member of the pair dominates/only one of the parental character is expressed in a monohybrid cross in the F1 and both are expressed in the F2.

(b) The alleles are present on homologous chromosomes. The recessive allele does not code for its product or codes for a defective product. The other allele remains normal and thus expresses itself.

Q.14. State and explain the "law of independent assortment" in a typical Mendelian dihybrid cross.

Ans. Law of independent assortment

According to this law the two factors of each character assort or separate out independent of the factors of other characters at the time of gamete formation and get randomly rearranged in the off springs producing both parental and new combinations of characters.



Phenotypic ratio–	Round yellow			:	Round green			:	Wrinkled yellow			:	Wrinkled green			
		9		:		3		:		3		:		1		
Genotypic ratio–	1	:	2	:	2	:	4	:	1	:	2	:	1	:	2	: 1

Results of a dihybrid cross where the two parents differed in two pairs of contrasting traits: seed colour and seed shape

Q.15. Let us assume in a given plant the genotype symbol "Y" stands for dominant yellow seed colour and "y" for recessive green seed colour; symbol "R" for round seed shape and "r" for wrinkled seeds. Two homozygous parents (plants) with genotypes "RRYY" and "rryy" are crossed and their F1-generation progeny is then selfed. What shall be the

a. Phenotype of F1-progeny

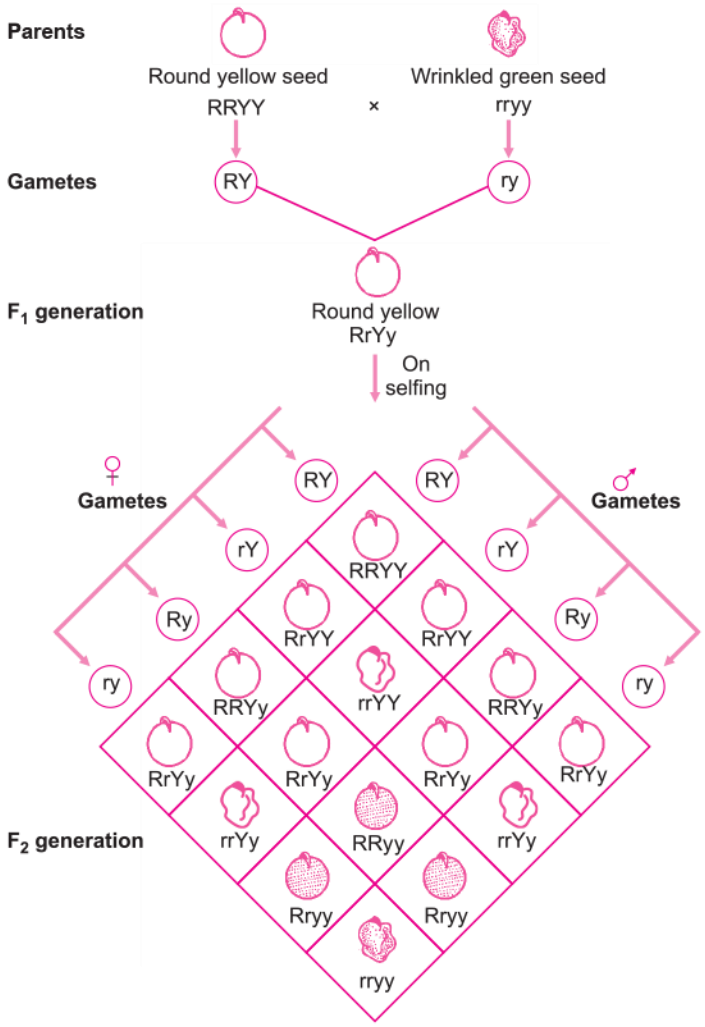
b. Genotype of F1-progeny

c. Gamete genotypes of F1-progeny

d. Phenotypic ratio of F2 population

e. Phenotypic ratio of yellow seed to green seed and round seed to wrinkled seed in F2 population.

Ans.



Phenotypic ratio–	Round yellow			:	Round green			:	Wrinkled yellow			:	Wrinkled green			
	9			:	3			:	3			:	1			
Genotypic ratio–	1	:	2	:	2	:	4	:	1	:	2	:	1	:	2	: 1

Results of a dihybrid cross where the two parents differed in two pairs of contrasting traits: seed colour and seed shape

a. Phenotype of F1-progeny: Round seeds that are yellow in colour

b. Genotype of F1-progeny: RrYy

c. Gamete genotypes of F1-progeny: RY, Ry, rY and ry

d. Phenotypic ratio of F2 population: 9 : 3 : 3 : 1.

Nine round-yellow seeds; three round-green seeds; three wrinkled-yellow seeds; one wrinkled-green seed.

e. Phenotypic ratio of yellow seed to green seed and round seed to wrinkled seed in F2 population:

Yellow seed to green seed = 3 : 1

Round seed to wrinkled seed = 3 : 1

Q.16.

a. Provide genetic explanation for the observation in which the flower colour in F_1 generation of snapdragon did not resemble either of the two parents. However, the parental characters reappeared when F_1 progenies were selfed.

b. State the three principles of Mendel's law of inheritance.

Ans.

(a) This is an exception to Mendel's principle of dominance and can be explained by the phenomenon of **'Incomplete dominance'**. It is a phenomenon where none of the two contrasting alleles or factors are dominant. The expression of the character in a hybrid or F1 individual is intermediate or a fine mixture of expression of the two factors (pink flowers in this case from two parents with red and white flowers). This may be considered as an example of quantitative inheritance where only a single gene pair is involved. F_2 phenotypic ratio is 1 : 2 : 1, similar to the genotypic ratio, in which the parental characters also reappear.

(b) Mendel's Laws of Inheritance

- Based on his hybridisation experiments, Mendel proposed the laws of inheritance.
- His theory was rediscovered by Hugo de Vries of Holland, Carl Correns of Germany and Eric von Tschermak of Austria in 1901.

(i)Law of dominance

- This law states that when two alternative forms of a trait or character (genes or alleles) are present in an organism, only one factor expresses itself in F_1 progeny and is called dominant while the other that remains masked is called recessive.

(ii) Law of segregation.

- This law states that the factors or alleles of a pair segregate from each other during gamete formation, such that a gamete receives only one of the two factors. They do not show any blending.

Q.17.

(a) Differentiate between dominance and co-dominance.

(b) Explain co-dominance taking an example of human blood groups in the population.

Ans.

(a) Dominance: It is a phenomenon in which when two contrasting alleles are present together, only one expresses itself and is called dominant whereas the other which does not express itself is called recessive.
Co-dominance: It is a phenomenon in which when two contrasting alleles are present together, both of the alleles express themselves.

(b) Co-dominance

- The alleles which are able to express themselves independently, even when present together are called co-dominant alleles and this biological phenomenon is called co-dominance.**For example,** ABO blood grouping in humans.
- ABO blood groups are controlled by gene *I*. Gene *I* has three alleles I^A, I^B and I^O/*i*.
- I^A and I^B produce RBC surface antigens sugar polymer A and B, respectively, whereas *i* does not produce any antigen.
- I^A and I^B are dominant over *i* hence I^A and *I*B are dominant alleles and *i* is recessive allele as in $I^A i$and I^Bi.
- When I^A and I^B are present together, both express equally and produce the surface antigens A and B, hence show co-dominance.
- Since humans are diploid, each person possesses any two of the three '*I*' gene alleles, resulting into six different genotypic combinations and four phenotypic expressions.**Table showing the genetic basis of blood groups in human population**

Allele fromParent 1	Allele fromParent 2	Genotype ofoffspring	Blood groups ofoffspring
I^A	I^A	$I^A I^A$	A
I^A	I^B	$I^A I^B$	AB
I^A	*i*	$I^A i$	A
I^B	I^B	$I^B I^B$	B
I^B	*i*	$I^B i$	B
i	*i*	*i i*	O

Q.18. Describe the mechanism of inheritance of the ABO system of blood group, highlighting the principle of genetics involved in it.

OR

Explain the genetic basis of blood grouping in human population.

Ans.

Co-dominance

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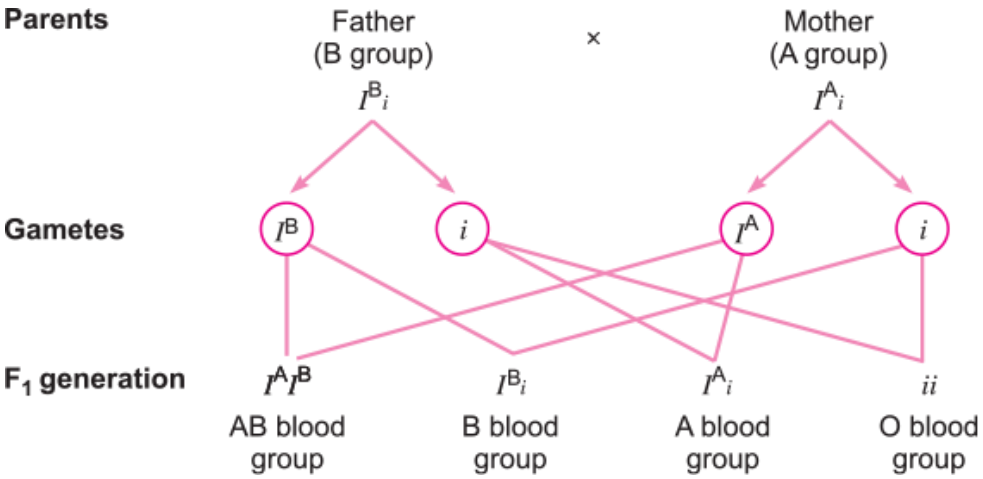
I^A	I^B	I^AI^B	AB
I^A	*i*	I^Ai	A
I^B	I^B	I^BI^B	B
I^B	*i*	I^Bi	B
i	*i*	*i i*	O

Q.19.

a. **Write the blood group of people with genotype I^AI^B. Give reasons in support of your answer.**
b. **In one family, the four children each have a different blood group. Their mother has blood group A and their father has blood group B. Work out a cross to explain how it is possible.**

Ans.

a. Blood group AB. Both the alleles I^A and I^B are co-dominant and express themselves completely.
b. A cross is carried out between heterozygous father (for blood group B) and heterozygous mother (of blood group A) to get four children with different blood groups.



All the four blood groups are controlled by three allelic genes I^A, I^B, i and thus it shows phenomena of multiple allelism. Both I^A and I^B are dominant over *i*. However, when together, both are dominant and show the phenomena of co-dominance forming the blood group AB. Six genotypes are possible with combination of these three alleles.

Q.20.

a. List the three different allelic forms of gene "I" in humans. Explain the different phenotypic expressions, controlled by these three forms.

b. A woman with blood group "A" marries a man with blood group "O". Discuss the possibilities of the inheritance of the blood groups in the following starting with "yes" or "no" for each:

i. They produce children with blood group "A" only.

ii. They produce children some with "O" blood group and some with "A" blood group.

Ans.

The three different allelic forms are: I^A, I^B, I^O/i.

Co-dominance

- The alleles which are able to express themselves independently, even when present together are called co-dominant alleles and this biological phenomenon is called co-dominance.**For example,** ABO blood grouping in humans.
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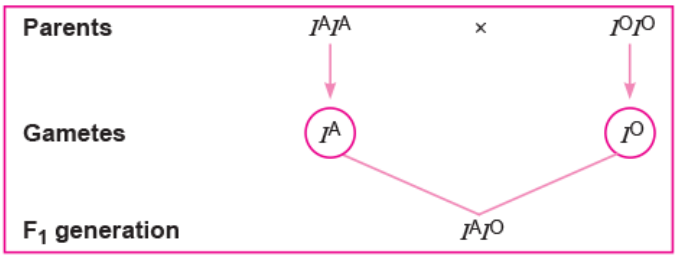
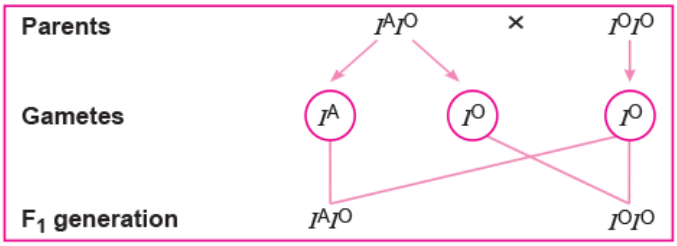
I^A	i	I^Ai	A
I^B	I^B	I^BI^B	B
I^B	i	I^Bi	B
i	i	$i\,i$	O

(b)

(*i*) Yes; when both the parent are homozygous.



(*ii*) Yes; when the woman is heterozygous.



Q.21.

a. How are Mendelian inheritance, polygenic inheritance and pleiotropy different from each other?
b. Explain polygenic inheritance pattern with the help of a suitable example.

Ans.

(a)

Mendelian Inheritance	Polygenic inheritance	Pleiotropy

One gene controls one trait/ character/phenotype	Two or more genes influence the expression of one trait/ character/phenotype	One genes controls the expression of more than one trait/character/phenotype

(b) Human height or skin colour are examples of polygenic inheritance. Height trait is controlled by at least three gene pairs. Additive effect allele contributes to the phenotypic expression of the trait. The dominant alleles more are, more pronounced is the phenotypic expression or more in other word. The recessive alleles less pronounced is the phenotypic expression.

Q.22.

i. How does a chromosomal disorder differ from a Mendelian disorder?
ii. Name any two chromosomal aberration associated disorders.
iii. List the characteristics of the disorders mentioned above that help in their diagnosis.

Ans.

i.

S. No.	Mendelian disorder	Chromosomal disorder
(*i*)	This disorder is mainly due to alteration or mutation in the single gene.	This disorder is caused due to absence or excess or abnormal arrangement of one or more chromosomes.
(*ii*)	This follows Mendel's principles of inheritance.	This does not follow Mendel's principles ofinheritance.
(*iii*)	This may be recessive or dominant in nature	This is always dominant in nature.
(*iv*)	For example, haemophilia, sickle-cell anaemia.	For example, Turner's syndrome.

ii. Two chromosomal aberration-associated disorders are Down's syndrome and Klinefelter's syndrome.

iii.

a. **Down's syndrome:** The individuals have overall masculine development but they express feminine development like development of east, *i.e.*, gynaecomastia. They are sterile.
b. **Klinefelter's syndrome:** The females are sterile as ovaries are rudimentary. Other secondary sexual characters are also lacking.

Q.23. Thalassemia and haemophilia are both Mendelian disorders related to blood. Write the symptoms of the diseases. Explain with the help of crosses the difference in the inheritance pattern of the two diseases.

OR

Why are thalassemia and hemophilia categorised as Mendelian disorders? Write the symptoms of these diseases. Explain their pattern of inheritance in humans. Write the genotypes of the normal parents producing a haemophilic son.

Ans. Both are caused due to alteration or mutation, in a single gene and follow Mendelian pattern of inheritance.

Symptoms:

Thalassemia: anaemia (caused due to defective/abnormal Hb)

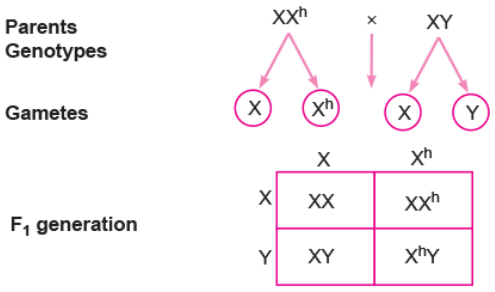
Haemophilia: non-stop bleeding even in minor injury.

Pattern of inheritance:

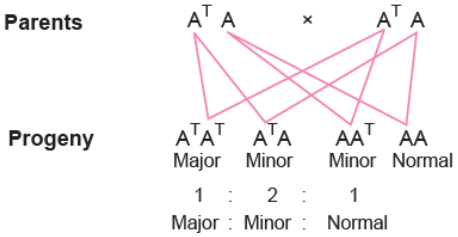
Thalassemia: autosomal recessive inheritance pattern inherited from eterozygous/parent carrier.

Haemophilia: X-linked recessive inheritance inherited from a haemophilic father/carrier mother (females are rarely haemophilic).

Cross for haemophilia:



Cross for thalassemia:



Thalassemia is an autosome-linked recessive blood disease. Its inheritance is like Mendelian inheritance pattern.

Q.24. Write the symptoms of haemophilia and sickle-cell anaemia in humans. Explain how the inheritance pattern of the two diseases differs from each other.

Ans. Symptoms of haemophilia: Patient continues to bleed through a minor cut as the patient does not possess natural phenomenon of blood clotting.

Symptoms of sickle-cell anaemia: Erythrocytes lose their circular shape and become sickleshaped. As a result, the cells cannot pass through narrow capillaries. Blood capillaries are clogged and thus affects blood supply to different organs.

S. No.	Haemophilia	Sickle-cell anaemia
(*i*)	It is a sex-linked recessive disorder.	It is an autosomal linked recessive trait.
(*ii*)	The gene for haemophilia is located onX-chromosome.	The disease is controlled by a single pair of allele Hb^A and Hb^S.
(*iii*)	More males suffer from haemophilia than females because in males single gene for the defect is able to express. Females suffer from this disease only in homozygous condition,*i.e.*, X^c X^c.	Only the homozygous individuals for Hb^S , *i.e.*, Hb^S Hb^Sshow the diseased phenotype.
(*iv*)	The defective alleles produce non-functional protein which later form a non-functional cascade of proteins involved in blood clotting.	Due to point mutation Glutamic acid (Glu) is replaced by Valine (Val) at sixth positions of beta globin chain of haemoglobin molecule.

Q.25.

a. Why are colour blindness and thalassemia categorised as Mendelian disorders? Write the symptoms of these diseases seen in people suffering from them.

b. About 8% of human male population suffers from colourblindness whereas only about 0.4% of human female population suffers from this disease. Write an explanation to show how it is possible.

Ans.

a. Both are caused due to mutation or alteration in a single gene, and follow Mendelian inheritance, therefore, they are called Mendelian disorders.
Symptoms of colour blindness: unable to discriminate between red and green colours.
Symptoms of thalassemia: formation of abnormal haemoglobin resulting in Anaemia.

b.

- **Thalassemia**

a. It is an autosome-linked recessive disease.
b. It occurs due to either mutation or deletion resulting in reduced rate of synthesis of one of globin chains of haemoglobin.
c. Anaemia is the characteristic of this disease.
d. Thalassemia is classified into two types:
    a. α-thalassemia—Production of α-globin chain is affected. It is controlled by the closely linked genes *HbA1* and *HbA2* on chromosome 16. It occurs due to mutation or deletion of one or more of the four genes.
    b. β-thalassemia—Production of β-globin chain is affected. It occurs due to mutation of one or both *HbB* genes on chromosome 11.

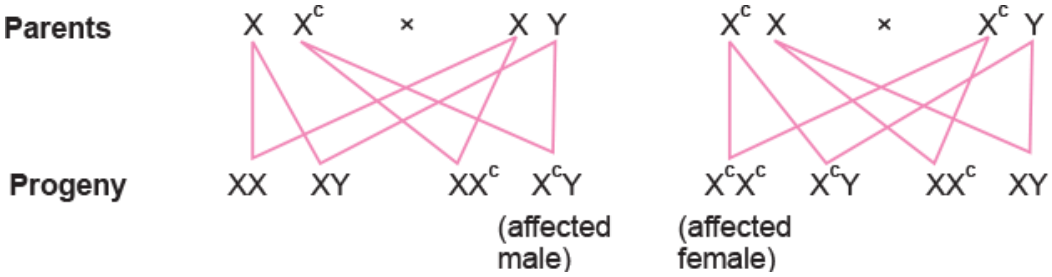
- **Colour blindness**
    a. It is a sex-linked recessive disorder.
    b. It results in defect in either red or/and green cone of eye, resulting in failure to discriminate between red and green colour.
    c. The gene for colour blindness is present on X chromosome.
    d. It is observed more in males (X^cY) because of presence of only one X chromosome as compared to two chromosomes of females.

Q.26.

a. State the cause and symptoms of colour-blindness in humans.
b. Statistical data has shown that 8% of the human males are colour-blind whereas only 0.4% of females are colour-blind. Explain giving reasons how is it so.

Ans.

a. Colour-blindness is a sex-linked recessive disorder.
   Its symptoms are failure to discriminate between red and green colour.
b. Since males have only one X chromosome gene for colour blindness, if present in any one parent will always be expressed, whereas in female it will be expressed only if it is present on both the X chromosome or when both parents are carrying gene for colour blindness.



Q.27. Write the type and location of the gene causing thalassemia in humans. State the cause and symptoms of the disease. How is sickle cell anaemia different from this disease?

Ans.

Sickle-cell anaemia

i. It is an autosome-linked recessive trait.
ii. The disease is controlled by a single pair of allele Hb^A and Hb^S.
iii. Only the homozygous individuals for Hb^S, *i.e.*, Hb^SHb^S show the diseased phenotype.
iv. The heterozygous individuals are carriers (Hb^AHb^S).
v. Due to point mutation, glutamic acid (Glu) is replaced by valine (Val) at the sixth position of β-globin chain of haemoglobin molecule.
vi. HbS behaves as normal haemoglobin except under oxygen stress where erythrocytes lose their circular shape and become sickle-shaped. As a result, the cells cannot pass through narrow capillaries. Blood capillaries are clogged and thus affect blood supply to different organs.

Thalassemia

i. It is an autosome-linked recessive disease.
ii. It occurs due to either mutation or deletion resulting in reduced rate of synthesis of one of globin chains of haemoglobin.
iii. Anaemia is the characteristic of this disease.
iv. Thalassemia is classified into two types:
- α-thalassemia—Production of α-globin chain is affected. It is controlled by the closely linked genes HbA1 and HbA2 on chromosome 16. It occurs due to mutation or deletion of one or more of the four genes.
- β-thalassemia—Production of β-globin chain is affected. It occurs due to mutation of one or both HbB genes on chromosome 11.

Q.28. Identify '*a*', '*b*', '*c*', '*d*', '*e*' and '*f*' in the table given below:

Ans.

S. No.	**Syndrome**	**Cause**	**Characteristics ofaffected individuals**	**SexMale/Female/Both**
1.	Down's	Trisomy of 21	'a' (*i*) (*ii*)	'*b*'
2.	'*c*'	XXY	Overall masculinedevelopment	'*d*'
3.	Turner's	45 with XO	'e' (*i*) (*ii*)	'*f*'

Ans.

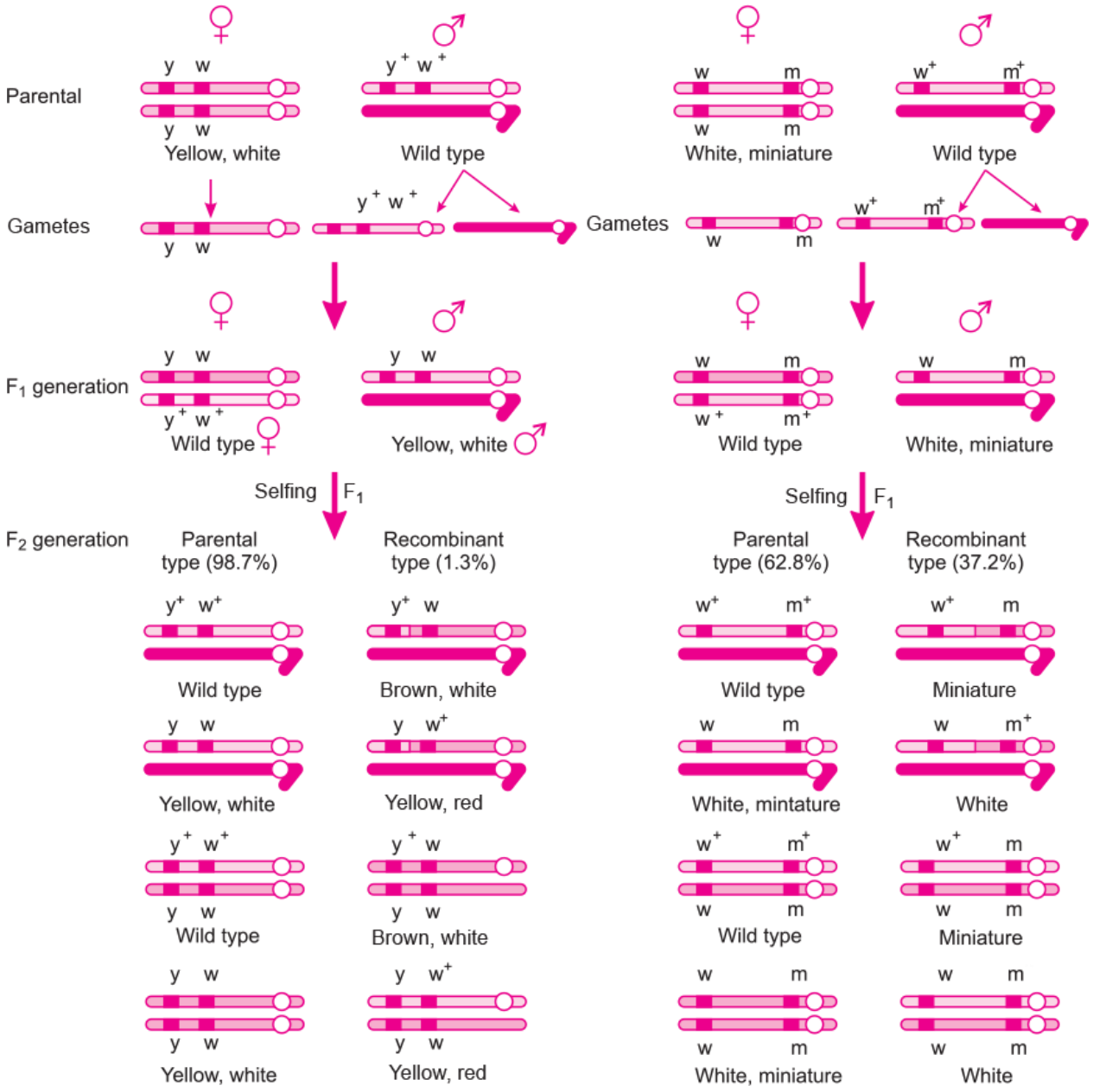
a. Short stature/small round head/furrowed tongue/partially open mouth/ mental development retarded.
b. Both.
c. Klinefelter's syndrome
d. Male
e.
    i. Sterile ovaries;
    ii. Lack of secondary sexual characters.
f. Female

Q.29. Describe the dihybrid cross carried on Drosophila melanogaster by Morgan and his group. How did they explain linkage, recombination and gene mapping on the basis of their observations?

Ans.

Linkage and Recombination

- **T. H. Morgan** carried out several dihybrid crosses in Drosophila to study the genes that are sex-linked. He observed that when the two genes in a dihybrid cross are located on the same chromosome, the proportion of parental gene combinations in the progeny was much higher than the non-parental or recombination of genes.
- Morgan and his group found that when genes are grouped on the same chromosome, some genes are tightly linked or associated and show little recombination.



- Results of two dihybrid crosses conducted by Morgan. Cross A shows crossing between genes y and w; Cross B shows crossing between genes w and m. Here dominant wild type alleles are represented with (+) sign in superscript.
- When the genes are loosely linked they show higher percentage of recombination.
- Morgan hybridised yellow bodied and white eyed females with brown bodied and red eyed males (wild type) (cross-A) and inter-crossed their F_1 progeny.
- Alfred Sturtevant determined that genes of *Drosophila* are arranged in a linear order. He measured the distance between genes and prepared **chromosome maps** with the position of genes on the chromosomes based on percentage of recombinants. These are also called **genetic maps**.

Q.30. Answer the following questions:

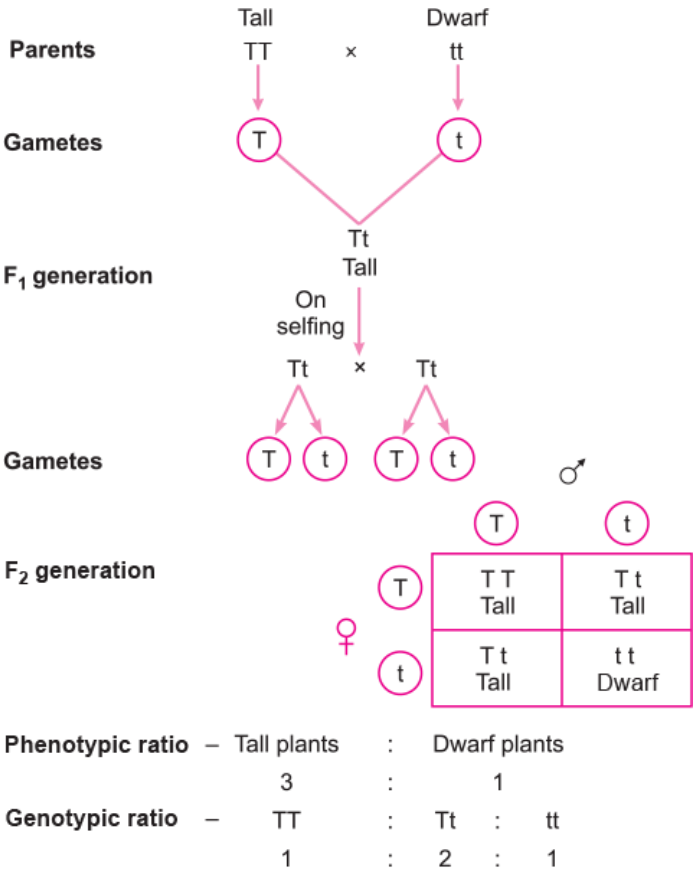
Q. State and explain the law of segregation as proposed by Mendel in a monohybrid cross.

Ans. Mendel's Observations

i. F_1 progenies always resembled one of the parents and trait of other parent was not seen.
ii. F_2 stage expressed both the parental traits in the proportion 3 : 1.
iii. The contrasting traits did not show any blending at either F_1 or F_2 stage.
iv. In dihybrid cross, he got identical results as in monohybrid cross.
v. He found that the phenotypes in F_2 generation appeared in the ratio 9 : 3 : 3 : 1.

Law of segregation.

- This law states that the factors or alleles of a pair segregate from each other during gamete formation, such that a gamete receives only one of the two factors. They do not show any blending.



Monohybrid cross of true-breeding pea plant

Q. Write the Mendelian F_2 phenotypic ratio in a dihybrid cross. State the law that he proposed on the basis of this ratio. How is this law different from the law of segregation?

Ans. The F_2 phenotypic ratio is 9:3:3:1. On the basis of this ratio Mendel proposed Law of Independent Assortment.

Law of independent assortment

- According to this law the two factors of each character assort or separate out independent of the factors of other characters at the time of gamete formation and get randomly rearranged in the offsprings producing both parental and new combinations of characters.

Q.31. Answer the following questions:

Q. State and explain the law of dominance as proposed by Mendel.

Ans. This law states that when two alternative forms of a trait or character (genes or alleles) are present in an organism, only one factor expresses itself in F_1 progeny and is called dominant while the other that remains masked is called recessive.
The characters are controlled by discrete units called factors. These factors occur in pairs.

Q. How would phenotypes of monohybrid F_1 and F_2 progeny showing incomplete dominance in Snapdragon and co-dominance in human blood group be different from Mendelian monohybrid F_1 and F_2 progeny? Explain.

Ans.

	Mendelian monohybrid cross	**Incomplete dominance**	**Co-dominance**
F_1	All members resemble the parent with dominant trait.	All members do not resemble either of the two parents but show an intermediate trait.	Blood groups of all members resemble combination of dominant traits of both the parents.
F_2	Both the parental traits reappear.	Both the parental traits and an intermediate trait appear.	Both the parental traits as well as the co-dominant trait appear.

Q.32. Answer the following questions:

Q. Explain Mendel's law of independent assortment by taking a suitable example.

Ans. According to this law, the two factors of each character assort or separate out independent of the factors of other characters at the time of gamete formation and get randomly rearranged in the offsprings producing both parental and new combinations of characters.

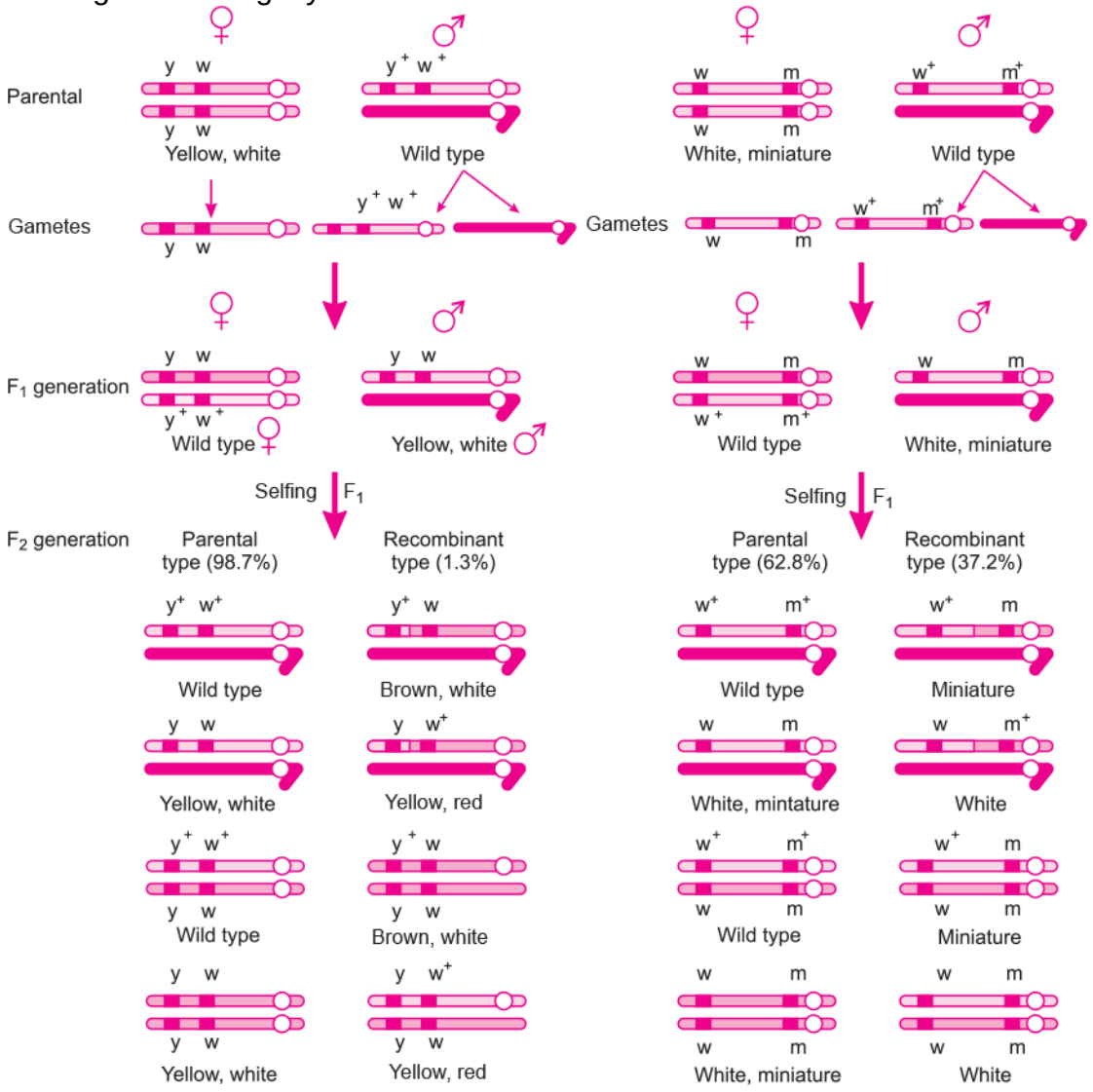
The Punnett square can be effectively used to understand the independent segregation of the two pairs of genes during meiosis and the production of eggs and pollen in the F_1 (RrYy) plant. Consider the segregation of one pair of genes R and r. Fifty per cent of the gametes have the gene R and the other 50 per cent have gene r. Now besides each gamete having either R or r, it should also have the allele Y or y. The important thing to remember here is that segregation of 50 per cent R and 50 per cent r is independent from the segregation of 50 per cent Y and 50 per cent y. Therefore, 50 per cent of the r bearing gamete has Y and the ther 50 per cent has y. Similarly, 50 per cent of the R bearing gamete has Y and the other 50 per cent has y.. Thus there are four genotypes of gametes (four types of pollen and four types of eggs). The four types are RY, Ry, rY and ry each with a frequency of 25 per cent of 1/4th of the total gametes produced.

Q. How did Morgan show the deviation in inheritance pattern in *Drosophila* with respect to this law?

Ans. Linkage and Recombination

- **T. H. Morgan** carried out several dihybrid crosses in Drosophila to study the genes that are sex-linked. He observed that when the two genes in a dihybrid cross are located on the same chromosome, the proportion of parental gene combinations in the progeny was much higher than the non-parental or recombination of genes.

- Morgan and his group found that when genes are grouped on the same chromosome, some genes are tightly linked or associated and show little recombination.



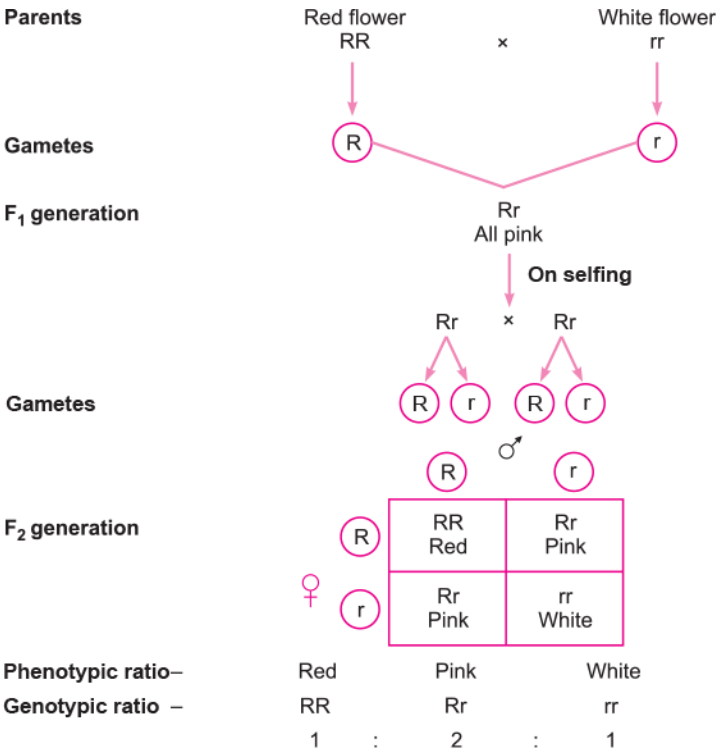
- Linkage: Results of two dihybrid crosses conducted by Morgan. Cross A shows crossing between genes y and w; Cross B shows crossing between genes w and m. Here dominant wild type alleles are represented with (+) sign in superscript.
- When the genes are loosely linked they show higher percentage of recombination.
- Morgan hybridised yellow bodied and white eyed females with brown bodied and red eyed males (wild type) (cross-A) and inter-crossed their F_1 progeny.
- Alfred Sturtevant determined that genes of *Drosophila* are arranged in a linear order. He measured the distance between genes and prepared **chromosome maps** with the position of genes on the chromosomes based on percentage of recombinants. These are also called **genetic maps**.

Q.33. Answer the following questions:

Q. During a cross involving true breeding red flowered and true breeding white flowered snapdragon plants, the F_1progeny did not show any of the parental traits, while they reappeared in F_2 progenies. Explain the mechanism using Punnett Square.

Ans. Incomplete Dominance

- It is a phenomenon in which the F_1 hybrid exhibits characters intermediate of the parental genes.
- Here, the phenotypic ratio deviates from the Mendel's monohybrid ratio.
- It is seen in flower colours of *Mirabilis jalapa* (4 o' clock plant) and *Antirrhinum majus* (snapdragon), where red colour is due to gene *RR*, white colour is due to gene *rr* and pink colour is due to gene *Rr*.



Monohybrid cross in snapdragon, where one allele is incompletely dominant over the other allele

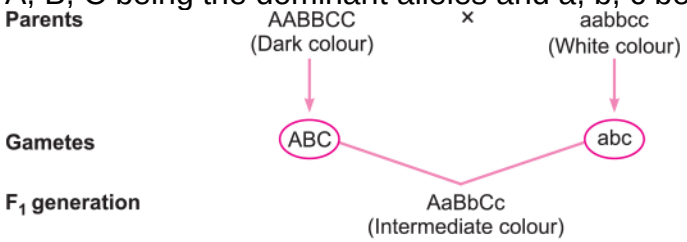
Q. Explain polygenic inheritance with the help of an example.

Ans.

Polygenic Inheritance

- It is a type of inheritance, in which traits are controlled by three or more genes. Such traits are called **polygenic traits**.

- The phenotype reflects contribution of each allele and is also influenced by the environment.
- For example, human skin colour. Suppose 3 genes A, B and C control skin colour with A, B, C being the dominant alleles and a, b, c being the recessive alleles. Then,



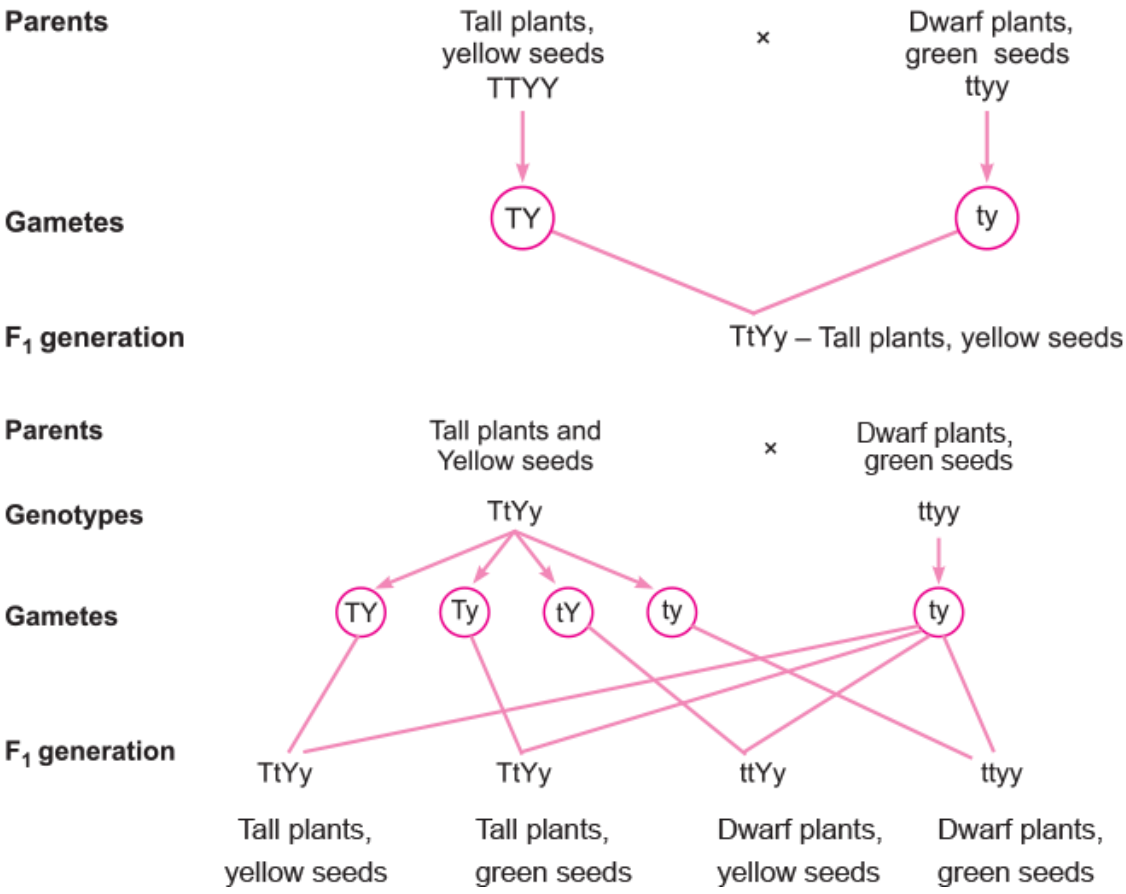
- The F_2 generation will have varied skin tones, with each type of allele in the genotype determining the darkness or lightness of the skin.

Q.34. Answer the following questions:

Q. You are given tall pea plants with yellow seeds whose genotypes are unknown. How would you find the genotype of these plants? Explain with the help of cross.

Ans.

Test cross will be performed to know the genotype of these plants.



If all the plants of F_1 generation are tall with yellow seeds, then the phenotype of the parent is homozygous dominant (case *i*). If the plants in F_1 generation are in the ratio of 1 : 1 : 1 : 1, then the parent plant is heterozygous dominant.

Q. Identify *a*, *b* and *c* in the table given below:

S. No.	Pattern of inheritance	Monohybrid F_1phenotypic expression
(*i*)	Co-dominance	*a*
(*ii*)	*b*	The progeny resembled only one of the parents
(*iii*)	Incomplete dominance	*c*

Ans.

a–Both the forms of a trait are equally expressed in F_1 generation.
b–Dominance.
c–Phenotypic expression of F_1 generation is somewhat intermediate between the two parental forms of a trait.

Q.35. Answer the following questions:

Q. Explain Polygenic inheritance and Multiple allelism with the help of suitable examples.

Ans. Co-dominance

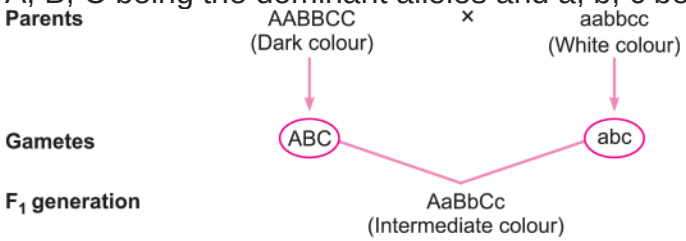
- The alleles which are able to express themselves independently, even when present together are called co-dominant alleles and this biological phenomenon is called co-dominance.**For example,** ABO blood grouping in humans.
- ABO blood groups are controlled by gene *I*. Gene *I* has three alleles I^A, I^B and I^O/i.
- I^A and I^B produce RBC surface antigens sugar polymer A and B, respectively, whereas *i* does not produce any antigen.
- I^A and I^B are dominant over *i* hence I^A and *I*B are dominant alleles and *i* is recessive allele as in I^Ai and I^Bi.
- When I^A and I^B are present together, both express equally and produce the surface antigens A and B, hence show co-dominance.
- Since humans are diploid, each person possesses any two of the three '*I*' gene alleles, resulting into six different genotypic combinations and four phenotypic expressions.**Table showing the genetic basis of blood groups in human population**

Allele fromParent 1	Allele fromParent 2	Genotype ofoffspring	Blood groups ofoffspring
I^A	I^A	I^AI^A	A
I^A	I^B	I^AI^B	AB
I^A	*i*	I^Ai	A
I^B	I^B	I^BI^B	B
I^B	*i*	I^Bi	B

i	*i*	*i i*	O

Polygenic Inheritance

- It is a type of inheritance, in which traits are controlled by three or more genes. Such traits are called **polygenic traits**.
- The phenotype reflects contribution of each allele and is also influenced by the environment.
- For example, human skin colour. Suppose 3 genes A, B and C control skin colour with A, B, C being the dominant alleles and a, b, c being the recessive alleles. Then,



- The F_2 generation will have varied skin tones, with each type of allele in the genotype determining the darkness or lightness of the skin.

Q. "Phenylketonuria is a good example that explains Pleiotropy." Justify.

Ans. In pleiotropy a single gene can exhibit multiple phenotypic expressions. In phenylketonuria single mutated gene express multiple phenotypic expression like mental retardation and reduction in hair and skin pigmentation.

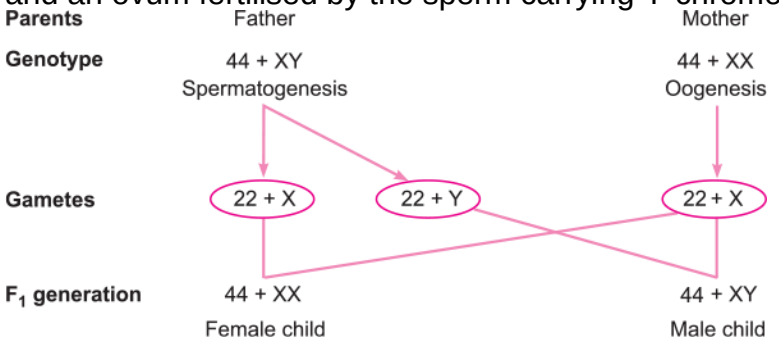
Q.36. Answer the following questions:

Q. Explain the mechanism of sex-determination in humans.

Ans. Sex Determination in Humans

- Humans show XY type of sex determining mechanism.
- Out of 23 pair of chromosomes, 22 are autosomes (same in both males and females).
- Females have a pair of X-chromosomes.
- Males have an X and a Y chromosome.
- During spermatogenesis males produce two types of gametes with equal probability – sperm carrying either X or Y chromosome.
- During oogenesis females produce only one types of gamete – having X chromosome.

- An ovum fertilised by the sperm carrying X-chromosome develops into a female (XX) and an ovum fertilised by the sperm carrying Y-chromosome develops into a male (XY).



Q. Differentiate between male heterogamety and female heterogamety with the help of an example of each.

Ans.

S. No.	Male heterogamety	Female heterogamety
(*i*)	Males produce two types of gametes.	Females produce two types of gametes.
(*ii*)	Example, male grasshopper produce gametes of two types—X and O.	Example, female birds produce gametes of two types—Z and W.

Q.37. Answer the following questions:

Q. Why is haemophilia generally observed in human males? Explain the conditions under which a human female can be haemophilic.

Ans. Haemophilia is caused due to the recessive gene on X chromosome. Y chromosome has no allele for this. If a male is X^hY, then he is haemophilic. If male inherits X^h from the mother, he will be haemophilic (with the genotype X^hY). If female inherits X^hX^h , one from the carrier mother and one from her haemophilic father, then she can be haemophilic.

Q. A pregnant human female was advised to undergo M.T.P. It was diagnosed by her doctor that the foetus she is carrying has developed from a zygote formed by an XX egg fertilised by Y-carrying sperms. Why was she advised to undergo M.T.P.?

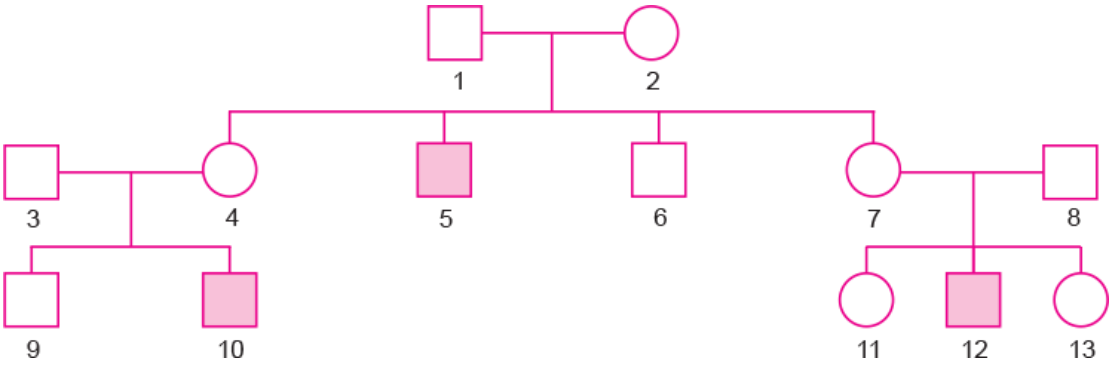
Ans. Embryo has (trisomy of sex chromosome) XXY karyotype or Klinefelter's syndrome.

She was advised to undergo MTP since the child will have the following problems:

i. male with feminine traits
ii. gynaecomastia
iii. underdeveloped testes
iv. sterile

Q.38. Answer the following questions:

Q. Haemophilia is a sex-linked recessive disease. Study the pedigree analysis given below showing the inheritance of the disease in a family and answer the questions that follow.



i. **Give the evidence from the above analysis which suggests that the disease is**
   1. **sex-linked and**
   2. **caused by a recessive allele.**
ii. **Write the possible genotypes of the individuals '2' and '5'.**

Ans.

i.
   1. In all the generations, only the males are affected with the disease.
   2. The parents of the affected individuals are not affected which implies that they are carriers and the gene is recessive.

ii. Possible genotype of '2': $X^d X$
Possible genotype of '5': $X^d Y$

Q. Why is thalassemia categorised as a Mendelian disorder? State the condition when an individual will suffer from the disease.

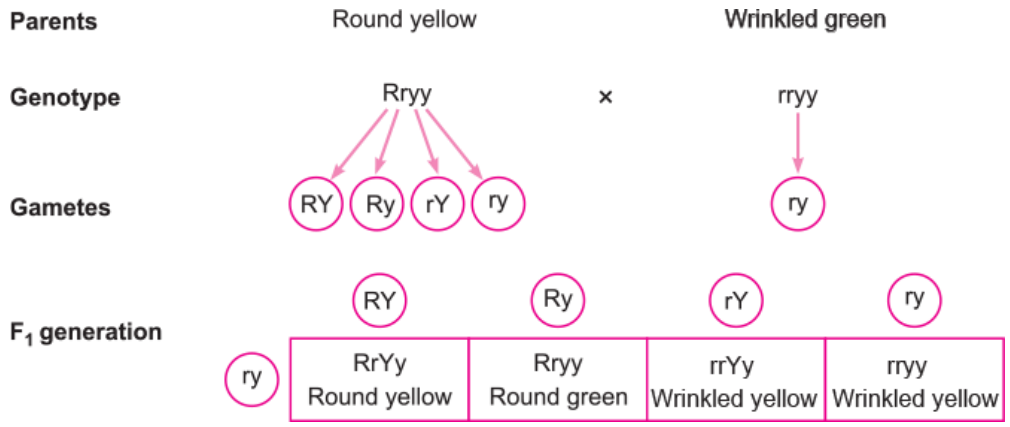
Ans. Thalassemia is categorised as Mendelian disorder because these are caused due to alteration or mutation in single gene and follow the Mendel's principles of inheritance. It occurs due to either mutation or deletion resulting in reduced rate of synthesis of one of globin chains of haemoglobin.

Q.39. A cross was carried out between a pea plant heterozygous for round and yellow seeds with a pea plant having wrinkled and green seeds.

a. **Show the cross in a Punnett square.**
b. **Write the phenotype of the progeny of this cross.**
c. **What is this cross known as? State the purpose of conducting such a cross.**

Ans.

(a)



(b) Both the phenotypic and genotypic ratio are same, i.e., 1: 1 : 1 : 1.

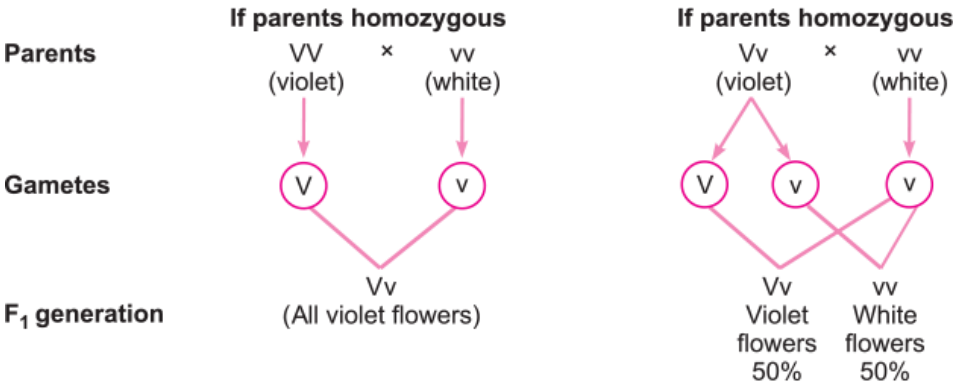
(c) This cross is known as test cross.

Q.40. A particular garden pea plant produces only violet flowers.

a. **Is it homozygous dominant for the trait or heterozygous?**
b. **How would you ensure its genotype? Explain with the help of crosses.**

Ans.

a. It could be homozygous dominant.
b. By performing test cross, genotype can be determined.



Long Answer Questions (OIQ)

[5 Marks]

Q.1. Describe the nature of inheritance of the ABO type of blood group in humans. In which ways does this inheritance differ from that of height of the plant in garden pea?

Ans. ABO blood group system in human:

Co-dominance

- The alleles which are able to express themselves independently, even when present together are called co-dominant alleles and this biological phenomenon is called co-dominance.**For example,** ABO blood grouping in humans.
- ABO blood groups are controlled by gene *I*. Gene *I* has three alleles I^A, I^B and I^O/i.
- I^A and I^B produce RBC surface antigens sugar polymer A and B, respectively, whereas *i* does not produce any antigen.
- I^A and I^B are dominant over *i* hence I^A and *I*B are dominant alleles and *i* is recessive allele as in I^Ai and I^Bi.
- When I^A and I^B are present together, both express equally and produce the surface antigens A and B, hence show co-dominance.
- Since humans are diploid, each person possesses any two of the three '*I*' gene alleles, resulting into six different genotypic combinations and four phenotypic expressions.**Table showing the genetic basis of blood groups in human population**

Allele fromParent 1	Allele fromParent 2	Genotype ofoffspring	Blood groups ofoffspring
I^A	I^A	I^AI^A	A
I^A	I^B	I^AI^B	AB
I^A	*i*	I^Ai	A
I^B	I^B	I^BI^B	B
I^B	*i*	I^Bi	B
i	*i*	*i i*	O

S. No.	Blood group in man	Height of plant in garden pea
(*i*)	The gene *I* responsible for the blood group exists in three allelic alternative forms I^A, $I^B$and *i*.	The gene for this trait exists in two allelic forms T and t.

(*ii*)	I^A and I^B are dominant over *i* and I^A and $I^B$are co-dominant.	Allele T is dominant over t.
(*iii*)	They exhibit four phenotypes with six possible genotypes.	They exhibit two phenotypes with three possible genotype
(*iv*)	Blood group in man exhibit phenomena of multiple allelism.	Height in garden pea plant do not exhibit multiple allelism.

Q.2. Explain the chromosomal theory of inheritance.

Ans. Chromosomal Theory of Inheritance

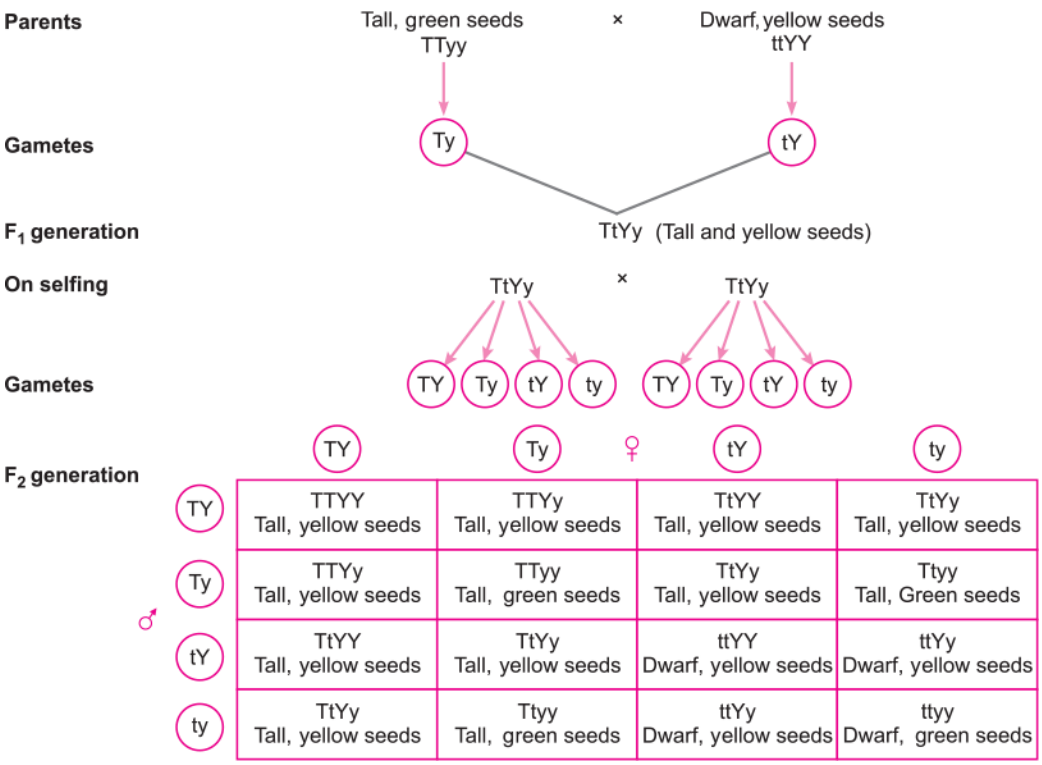
The chromosomal theory of inheritance was proposed independently by Walter Sutton and Theodore Boveri in 1902. According to this theory,

- **i.** Since the sperm and egg cells provide the only bridge from one generation to the other, all hereditary characters must be carried in them.
- **ii.** The hereditary factors are carried in the nucleus.
- **iii.** Like the Mendelian alleles, chromosomes are also found in pairs.
- **iv.** The sperm and egg having haploid sets of chromosomes fuse to re-establish the diploid state.
- **v.** The genes are located on the chromosomes in a linear order. As there are two chromosomes of each kind in somatic (diploid) cell there must be two genes of each kind, one in each of the two homologous chromosomes.
- **vi.** Homologous chromosomes synapse during meiosis and get separated to pass into different cells. This forms the basis for segregation and independent assortment. A gamete receives only one chromosome of each type and thus has only one gene for a trait. The paired condition is restored by fusion of gametes.

Q.3. A homozygous tall pea plant with green seeds is crossed with a dwarf pea plant with yellow seeds:

- **i. What would be the phenotype and genotype of F_1?**
- **ii. Work out the phenotypic ratio of F_2 generation with the help of a Punnett square.**

Ans.



i. Phenotype of F_1—Tall plants with yellow seeds.
Genotype of F_1—TtYy.

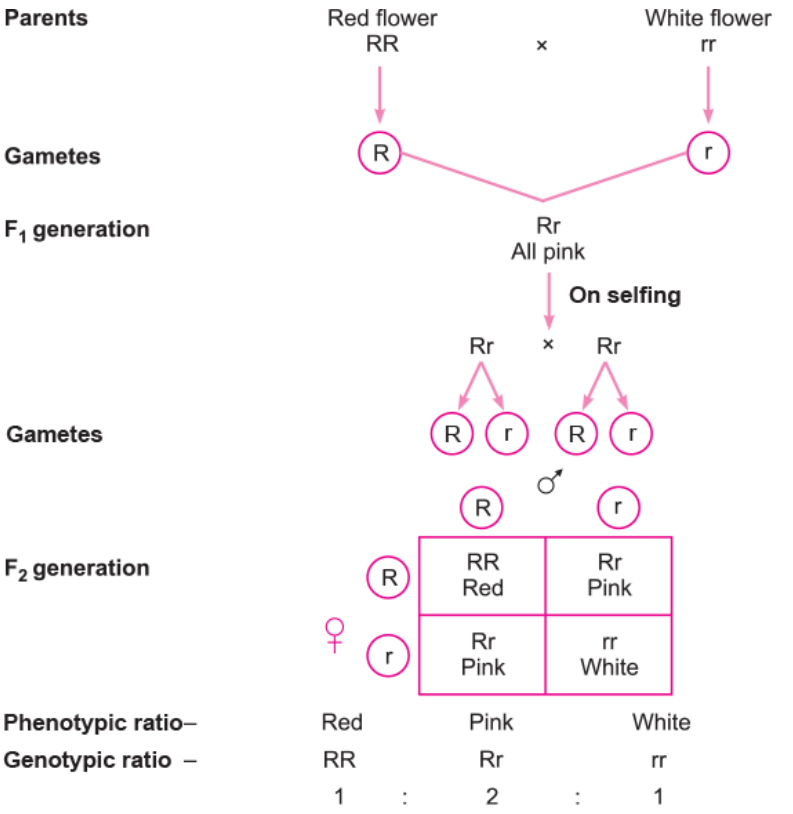
ii.

Phenotypic ratio of F_2 generation:

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

Q.4. In the case of snapdragon (*Antirrhinum majus*) a plant with red flowers was crossed with another plant with white flowers. Trace the inheritance of flower colour up to F_2 generation indicating the genotype and phenotype at each level. What special feature do you notice in the genotype and phenotype ratio in F_2 generation?

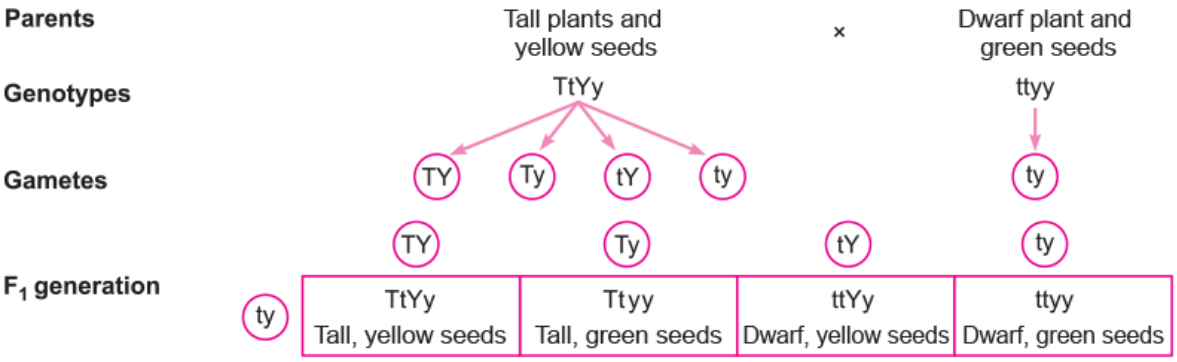
Ans.



Comment: This is a case of Mendelian deviation and that shows incomplete dominance as red and white both are not expressed but produce pink trait in F_1. Here, both the genotypic and phenotypic ratio are 1 : 2 : 1.

Q.5. A tall pea plant with yellow seeds (heterozygous for both the traits) is crossed with a dwarf pea plant with green seeds. Using a Punnett square work out the cross to show the phenotypes and the genotypes of F_1 generation.

Ans.



The phenotype and genotype of F_1 generation are

Tall, yellow seeds : Tall, green seeds : Dwarf, yellow seeds : Dwarf, green seeds

TtYy : Ttyy : ttYy : ttyy

1 : 1 : 1 : 1

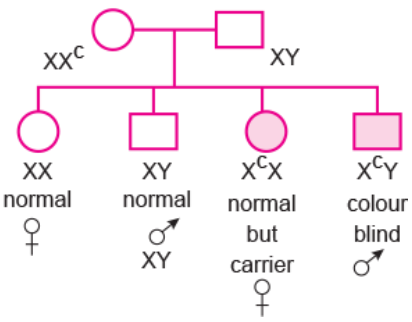
Q.6. A normal visioned woman, whose father is colour blind, marries a normal visioned man. What would be the probability of her (*a*) sons (*b*) daughters to be colour blind? Explain with the help of pedigree chart.

Ans.

The genotypes of parents are:

Father ⟶ XX^c as her father is colour blind.

Mother ⟶ XY



All daughters are normal visioned and 50% of sons are likely to be colour blind.

Q.7. Answer the following questions.

Q. Four children with four different blood groups are born to parents where the mother has blood group 'A' and the father has blood group 'B'. Work out the cross to show the genotypes of the parents and all four children.

Ans.

Parents $I^A I^O$ × $I^B I^O$

Blood group A B

Gametes I^A I^O I^B I^O

F_1 generation

♀ \ ♂	I^A	I^O
I^B	$I^A I^B$ Blood group AB	$I^B I^O$ Blood group B
I^O	$I^A I^O$ Blood group A	$I^O I^O$ Blood group O

Q. Explain the contribution of Alfred Sturtevant in 'Chromosome mapping'.

Ans. Alfred Sturtevant used the frequency of recombination between gene pairs on the same chromosome as a measure of the distance between genes and mapped their position on the chromosome.