Principles of Inheritance and Variation

INTRODUCTION

- The branch of biology that deals with the inheritance of character from parents to offspring is called as genetics. The word genetics was coined by William Bateson.
- The word 'heredity' is taken from *hereditus* meaning heirship. It deals with the inheritance of similar characters from parents.
- The question of how offsprings resemble their parents in some characters leads to the detailed study of the organisms at the genetic level.
- Aristotle pointed out that the concept proposed by Pythagoroas was not correct. He said that only the sperms do not carry the information to the offsprings but the male and female pass on information to the offsprings.

MENDEL LAW OF INHERITANCE

- Gregor Johann Mendel was an Austrian Monk. He conducted many experiments with garden pea plants and is considered as the 'Father of Genetics'.
- He conducted a large number of hybridization experiments on Pea plants and arrived at some conclusions.
- Initially, his work did not get recognition because of the following reasons-
- He applied mathematical knowledge to biology.
- He talked about certain factors that people did not believe in.
- During that time scientists were more interested in the work of Darwin that they could relate with.
- In 1900, his results were independently rediscovered by three biologists namely, Hugo De Vries, Carl Correns and Enrich Von Tschermak.
- A few years later, W. Bateson and others confirmed Mendel's work and found that the same laws were applied to animals also.
- Mendel selected pea plant as his experimental material because-
- Was easily available, flowers were easy to handle.

Definition

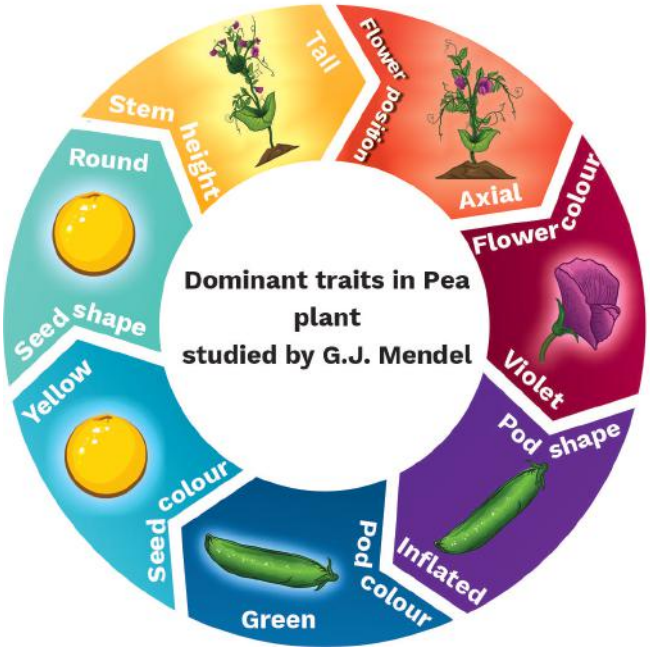
Heredity: The process by which the characters are transferred from the parents to the offspring .

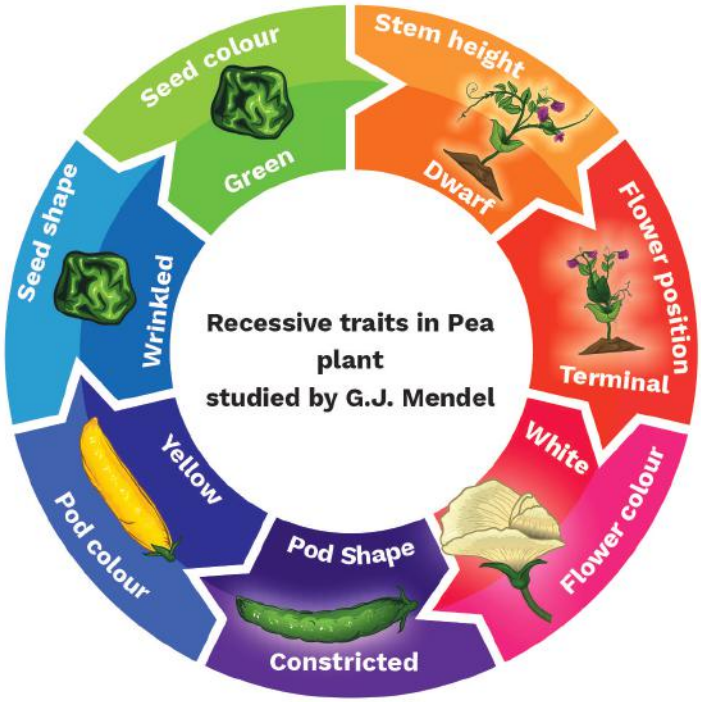
Gregor Johann Mendel
"Father of Genetic"

Rack Your Brain

What is the imporatnce of a short life period for the oragnism choosen as an experimental material by a geneticist for conduction of studies on pattern of inheritance?

- Flowers of pea plants remain closed till pollination.
- It showed seven pairs of contrasting characters.





Previous Year's Question

How many pairs of contrasting characters in pea plants were studied by Mendel in his experiments?
(1) Eight
(2) Seven
(3) Five
(4) Six

Definition

Allele: A pair of contrasting genes of a trait present on the same loci of a homologous chromosome is called an allele.

Previous Year's Question

In his classic experiments on pea plants, Mendel did not use
(1) seed shape
(2) flower position
(3) seed colour
(4) pod length

- Self and cross-pollination both were possible in the plant.
- It had a short life cycle.
- Pure line varieties were present.

INHERITANCE OF ONE GENE

- Mendel took tall and dwarf plants and made them cross and the seeds available after the cross were planted.
- All the seeds that were planted grew into tall plants. These plants were called the F_1 generation or first filial generation.
- He took plants from the F_1 generation and made them to self-pollinate. The seeds obtained from them were again planted.
- The seeds gave rise to a new generation known as the F_2 generation i.e., second filial generation.
- Some of the plants of F_2 generation were tall, some were dwarf. The 1/4th of the plants were tall, 3/4th of them were tall. Only one parental trait was expressed in F_1 generation and both the traits expressed in F_2 generation.
- If we use alphabets for each, then 'T' is the allele that controls tall trait and 't' is the allele that controls dwarf trait. Gene for homozygous tall can be represented as 'TT' while dwarf can be represented as 'tt'.
- The F_1 generation that was obtained was heterozygous (Tt). These plants were phenotypically tall like the parent with gene TT. From this he concluded that T is the dominant allele which expresses itself irrespective of the second allele present.
- In F_2 generation, tall plants with genotype TT and Tt appeared while dwarf plants with tt gene appeared. He concluded that 't' is the recessive allele and it can only express itself in the presence of the same recessive allele.

Definition

Homozygous: The organism having two identical allele for a particular gene expressing a character.For example,TT, tt

Rack Your Brain

At which stage of meiosis does the segregation of Mendelian factors can be seen under observation?

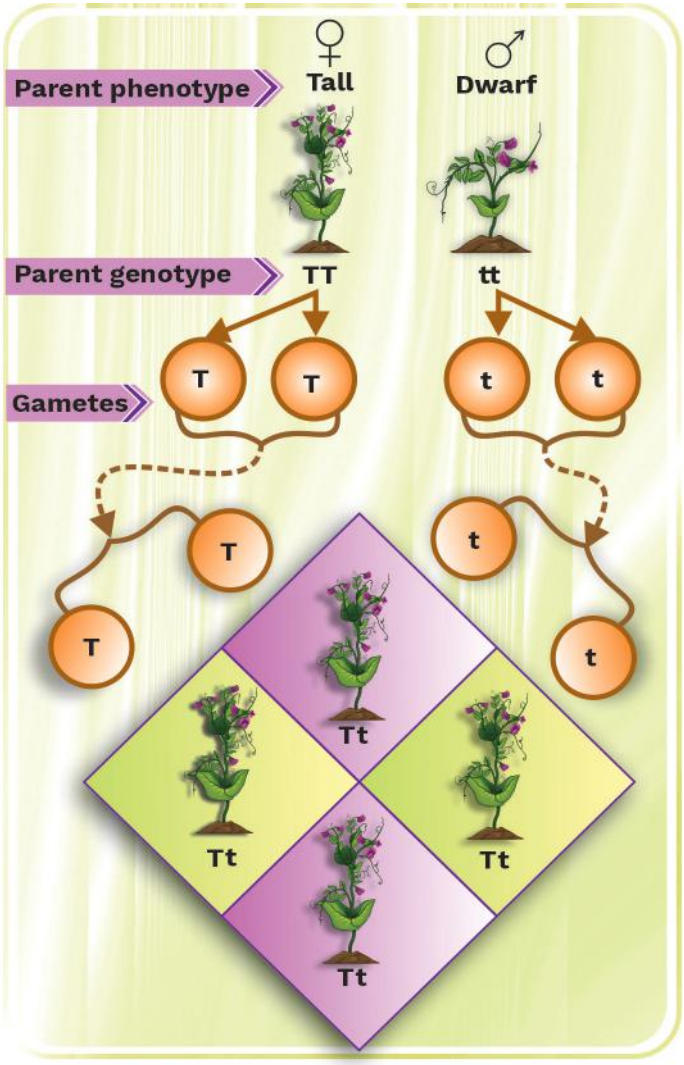
Previous Year's Question

In organism with two identical alleles is
(1) dominant
(2) hybrid
(3) heterozygous
(4) homozygous

Definition

Heterozygous: The individual having two dissimilar allele of a geneexpressing a character. For example Tt,

- Mathematically he calculated that in F_2 generation ¼ tall plants were with genotype TT while 1/2 were Tt and ¼ were tt. But Tt and TT were tall, so phenotypically ¾ of the plants were tall while the only ¼ of them were phenotypically and genotypically dwarf.
- The phenotypic ratio was calculated to be ¾ of tall : 1/4 of dwarf i.e., 3 : 4.
- The genotypic ratio was calculated to be ¼ : 1/2 : 1/4 (TT : Tt : tt), i.e 1 : 2 : 1.
- Mendel concluded all this with the help of Punnett square. The cross explained is a monohybrid cross as inheritance of only one trait is considered at one time only.



Rack Your Brain

In sexually reproducing organisms what is the contribution of the parents to their offspring ½ of their genes or ¼ of their genes?

Definition

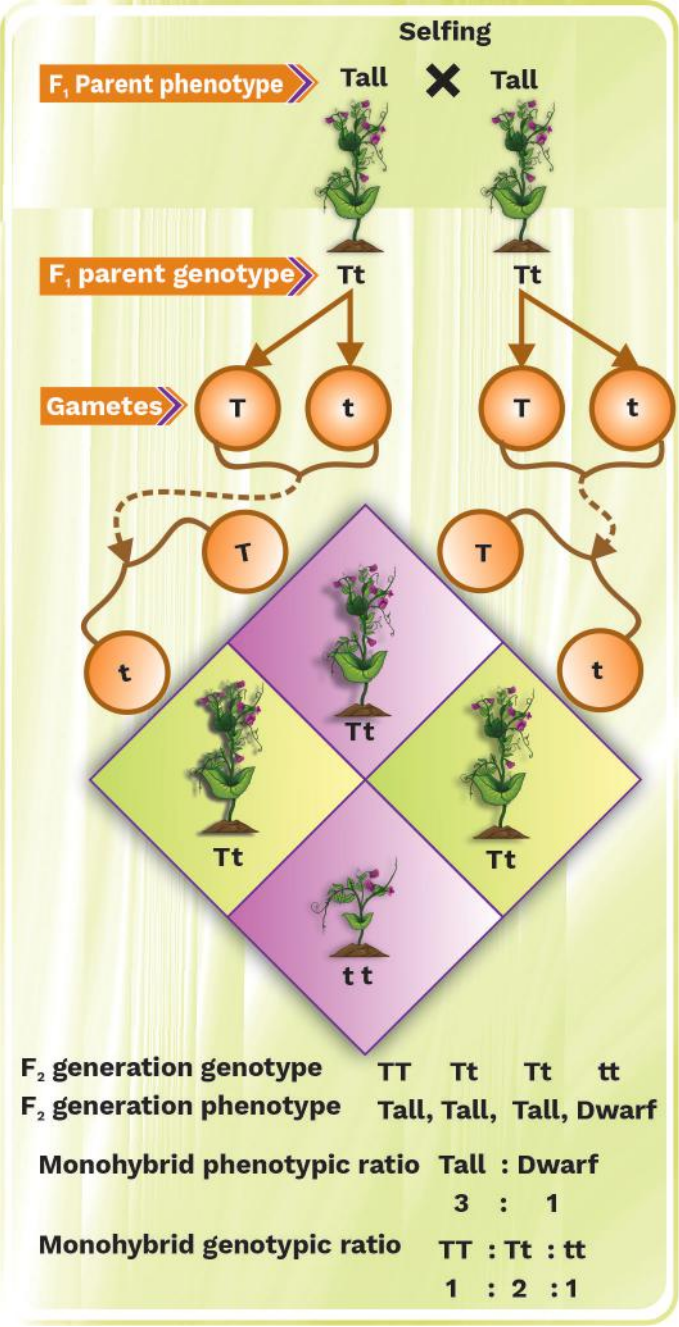
Phenotype: The individual having two dissimilar allele of a geneexpressing a character. For example Tt,

Previous Year's Question

How many different types of genetically different gametes will be produced by a heterozygous plant having the genotype AABbCc?
(1) Six
(2) Nine
(3) Two
(4) Four

Definition

Genotype: The genetic constitution of the phenotype of an organism.For example,TT and Tt are the genotype for the character height.



Mendel's Findings

- F_1 resembles only one parent.
- F_2 generation obtained by self-pollination-Dominant and recessive forms appear in a 3 : 1 ratio.(phenotypically)

Previous Year's Question

In hybridization, Tt × tt gives rise to the progeny of ratio
(1) 2 : 1
(2) 1 : 2 : 1
(3) 1 : 1
(4) 1 : 2

Rack Your Brain

Which type of the type of allele expresses itself only in homozygous state in an organism?

Previous Year's Question

The production of gametes by the parents, formation of zygotes, the F_1 and F_2 plants can be understood from a diagram called
(1) net square
(2) bullet square
(3) punch square
(4) punnett square

- F_2 generation obtained after self-pollination gave genotypic ratio as 1 : 2 : 1 ratio of pure and hybrid forms.

Interpretation of Mendel's Results

- Mendel said that factors are transmitted from parents to offspring. Each factor contains information about the form of a trait.
- **The Principle Law of Dominance:** If a cross is conducted between two characters of parents then the character that appears in F_1 generation is called dominant and the suppressed one which appears in F_2 generation is called recessive.

Law of Segregation

- It states that the alleles remain together but segregate at the time of gamete formation.
- The alleles T and t segregate at the time of gamete formation and when the gametes fused the recessive trait appeared in F_2 generation even when it was absent in F_1 generation.

Reasons for Mendel Success

- Mendel organised his experiments methodically, obtained observations systematically and analysed the obtained data mathematically.
- He selected garden pea plant which is bisexual and self-pollinated them.
- He obtained pure breeds of each character.
- He studied the offspring of not only the first generation but also the second generation.
- Mendel studied inheritance of seven different pairs of contrasting characters in garden pea.
- He counted the offsprings after each cross and then analysed the results mathematically.
- He organized his data in such a way that his results could be evaluated simply and objectively.

INHERITANCE OF TWO GENES

- Mendel studied the inheritance of two traits at a time. Such a cross where inheritance of two traits

Definition

Law of Dominance: Discrete units called factors occur in pairs and control characters.
In dissimilar pair of factors one member of the pair dominates and thus is the dominant factor while the the other is the recessive factor.

Previous Year's Question

Which one of the following cannot be explained on the basis of Mendel's law of dominance?

(1) The discrete unit controlling a particular character is called a factor.
(2) Out of one pair of factors one is dominant, and the other recessive.
(3) Alleles do not show any blending and both the characters recover as such in F_2 generation.
(4) Factors occur in pairs.

Definition

Law of Segregation: It states that the alleles remain together but segregate at the time of the gamete formation.

is studied simultaneously is known as dyhybrid cross.

- He crossed plants having Round and Yellow seeds with plants having Wrinkled and green seeds.
- The plant with Round and yellow seed was represented by RRYY and the one with Wrinkled and green seed was represented by rryy.
- The F_1 generation consisted of round and yellow seeded plants.
- F_1 generation when self-pollinated lead to the formation F_2 generation which consisted of Round yellow, round green and wrinkled yellow and wrinkled green seeded plants in the ratio 9:3:3:1.
- Consider the segregation of one pair of genes RRYY rryy. Fifty per cent of the gametes have the gene R and the other 50 per cent have r gene. Besides this, allele Y and y also need to segregate.
- Fifty percent of the gametes have Y gene while the other fifty percent have y gene. The segregation R and r is independent of Y an y gene.
- 50 per cent of the r genes bearing gametes have Y gene and the other 50 per cent genes have y gene and 50 per cent gene of the R bearing gametes have Y gene and the other 50 per cent have y gene.
- There are four genotypes of gametes-RY, Ry, rY and ry each with a frequency of 25 per cent or 1/4th of the total gametes produced.

Interpretation of Mendel's Results

- **The Principle Law of Segregation or Law of Independent Assortment:** It states that when two or more characters are crossed or combined in a hybrid, the alleles segregate independently of each other during gamete formation.

Previous Year's Question

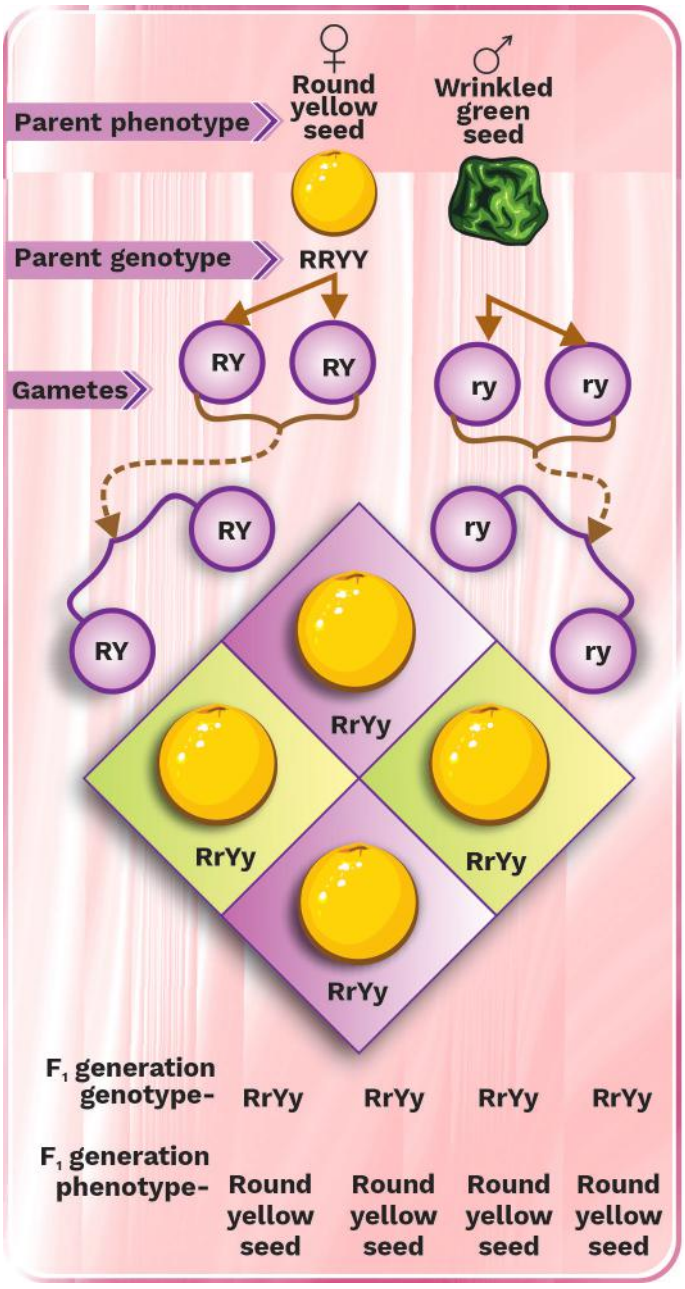
Cross between AaBB and aaBB will form
(1) 1AaBB : 1aaBB
(2) all AaBB
(3) 3AaBB : 1aaBB
(4) 1AaBB : 3aaBB

Definition

Law of Segregation: When inheritance of two or more characters is tsudied simultaneously, the alleles segregate independent of each other.

Previous Year's Question

Phenotype of an organism is the result of
(1) genotype and environment interactions
(2) mutations and linkages
(3) cytoplasmic effects and nutrition
(4) environmental changes and sexual dimorphism.



Previous Year's Question

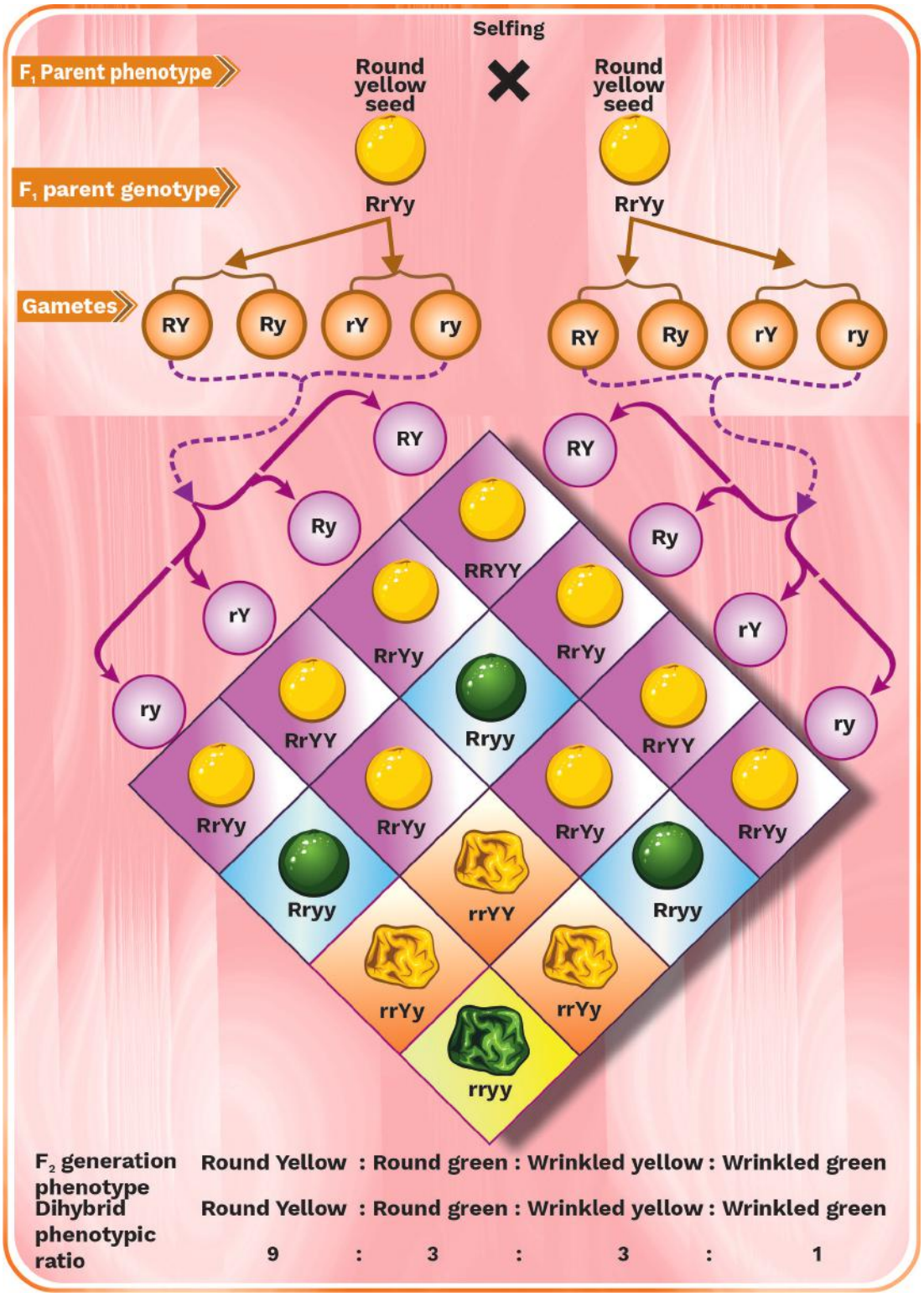
In Mendel's experiments with garden pea, round seed shape (RR) was dominant over wrinkled seeds (rr), yellow cotyledon (YY) was dominant over green cotyledon (yy). What are the expected phenotypes in the F_2 generation of the cross RRYY × rryy?

(1) Round seeds with yellow cotyledons, and wrinkled seeds with yellow cotyledons
(2) Only round seeds with green cotyledons
(3) Only wrinkled seeds with yellow cotyledons
(4) Only wrinkled seeds with green cotyledons

Previous Year's Question

In a cross between AABB × aabb, the ratio of F_2 genotypes between AABB, AaBB, Aabb and aabb would be

(1) 9 : 3 : 3 : 1
(2) 2 : 1 : 1 : 2
(3) 1 : 2 : 2 : 1
(4) 7 : 5 : 3 : 1


Selfing
F1 Parent phenotype
Round yellow seed
Round yellow seed
F1 parent genotype
RrYy
RrYy
Gametes
RY
Ry
rY
ry
RY
Ry
rY
ry
RY
RY
Ry
Ry
rY
rY
ry
ry
RRYY
RrYy
RrYy
RrYy
Rryy
RrYY
RrYY
RrYy
RrYy
RrYy
RrYy
Rryy
Rryy
rrYY
rrYy
rrYy
rryy
F2 generation phenotype
Round Yellow : Round green : Wrinkled yellow : Wrinkled green
Dihybrid phenotypic ratio
Round Yellow : Round green : Wrinkled yellow : Wrinkled green
9 : 3 : 3 : 1

TEST CROSS AND BACK CROSS

- In genetic it is important to find out the genotype of the parent involved in the cross.
- This can be don by back cross or test cross.
- A cross where F_1 progeny is crossed with either of the parents is called back cross.
- F_1 X TT (dominant parent)
- F_1 X tt (recessive parent)
- A cross where the F_1 progeny is crossed with recessive parent (homozygous) to test the homozygosity or heterozygosity of the parent is called test cross.
- The progeny in F_2 will be produced in the ratio of 1 : 1 (i.e., 50%).
- Test cross is used for determining whether the parent plants in the cross are pure for the traits under study.
- In genetic it is important to find out the genotype of the parent involved in the cross.
- This can be don by back cross or test cross.
- Example-
  - If we want to determine the genotype of a violet flower pea plant then it can be crossed with the dominat parent or the recessive parent

Previous Year's Question

A common test to find the genotype of a hybrid is by

(1) crossing of one F_2 progeny with female parent

(2) studying the sexual behaviour of F_1 progenies

(3) crossing of one F_1 progeny with male parent

(4) crossing of one F_2 progeny with male parent

Rack Your Brain

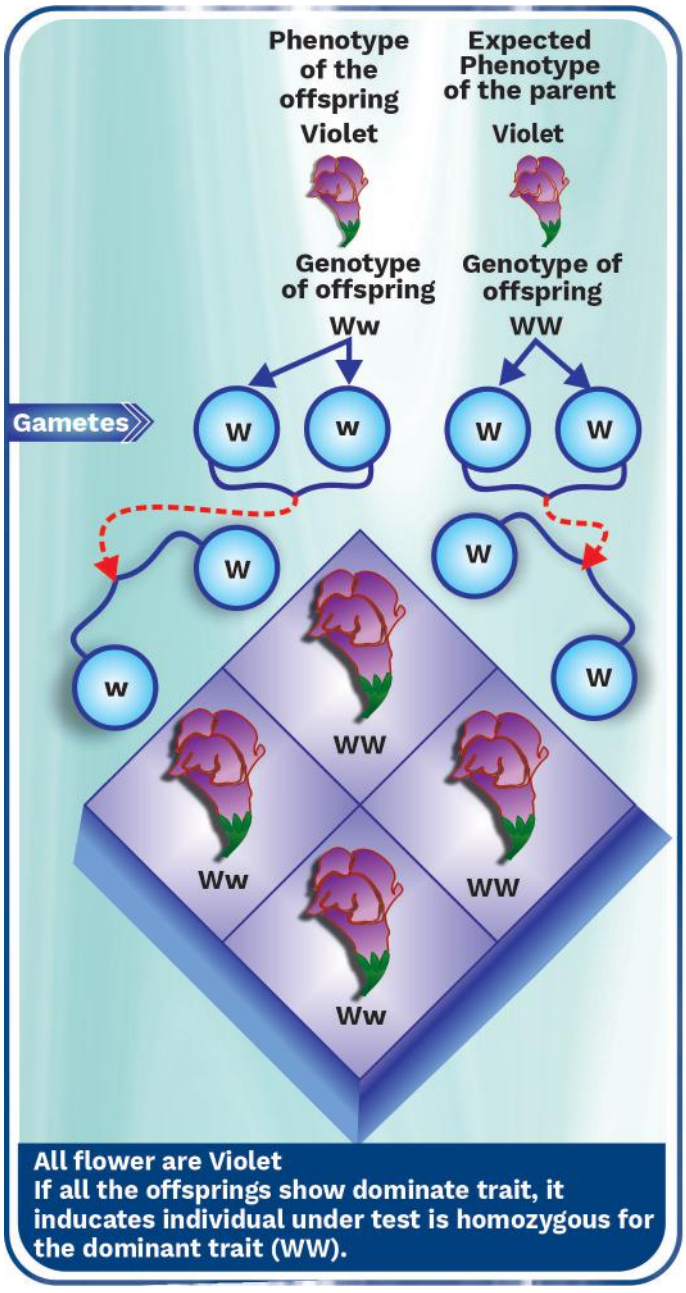
Give the genotypes of the parent pea plant in a test cross ,when all the progeny were bearing violet flowers

Previous Year's Question

Which is functional unit of inheritance?

(1) Cistrone (2) Intron

(3) Chromosome (4) Gene



Previous Year's Question

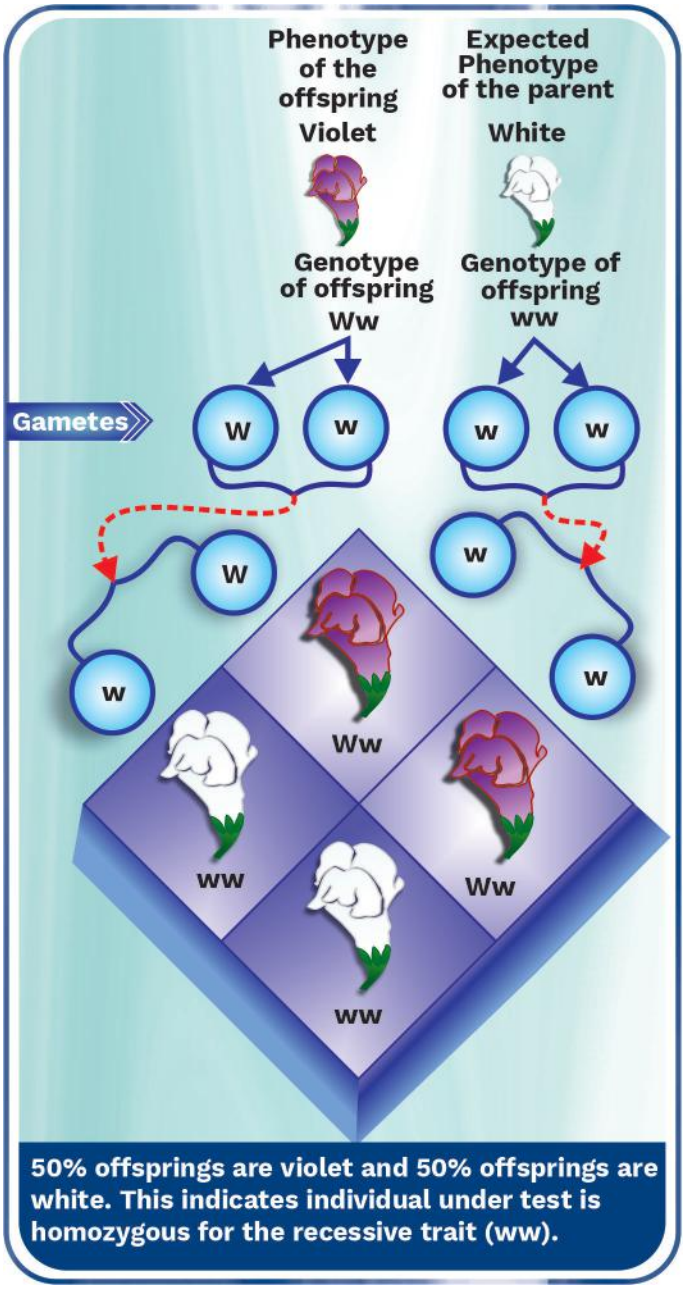
A test cross is carried out to
(1) determine the genotype of a plant at F_2
(2) predict whether two traits are linked
(3) assess the number of alleles of a gene
(4) determine whether two species or varieties will breed successfully

Rack Your Brain

Explain the reason why a tall pea plant when selfed produces one-fourth of its progeny as dwarf.

Previous Year's Question

A cross in which an organism showing a
dominant phenotype is crossed with the recessive
parent in order to know its genotype is called
(1) monohybrid cross
(2) back cross
(3) test cross
(4) dihybrid cross



EXCEPTION TO MENDELIAN PRINCIPLES

- Gene Interaction can be of two types-
- Intragenic Interaction
- Intergenic Interaction
- **Intragenic Interaction-**Two alleles of a gene present on the same locus of the homologous

Previous Year's Question

Complete haploid set of chromosomes a species is
(1) genome (2) genotype
(3) genetic code (4) allele

Rack Your Brain

Name the test that helps to study the genotype and the zygosity of the parents involved in a cross.

Previous Year's Question

PYQ-In a test cross involving F 1 dihybrid flies, more parentaltype offspring were produced than the recombinanttype offspring. This indicates
(1) the two genes are linked and present on the same chromosome
(2) both of the characters are controlled by more than one gene
(c) the two genes are located on two different chromosomes
(d) chromosomes failed to separate during meiosis.

chromosome produce modified phenotype.

- It is of the following type-
  - Incomplete dominance
  - Multiple Allelism
  - Coodominace
- **Intergenic Interaction-**The genes are present on different chromosomes and produce a changed phenotypic effect.
- It includes-
  - Epistatic gene
  - Dupliacte gene
  - Complementary gene
  - Supplementary gene

INCOMPLETE DOMINANCE

- The inheritance in which the alleles occupying the same position on homologous chromosome express themselves partially when present together in a hybrid is known as incomplete dominance.
- The F_1 individuals have the characters intermediates to the characters of the parents in this type of inheritance.
- For example- In Snapdragon (*Antirrhinum majus*) a cross between red-flowered plant and a white-flowered plant produces pink-flowered plants in F_1 generation.
- On selfing these F_1 plants give red, pink and white-flowered plants in F_2 generation in the ratio of 1 : 2 : 1
- 'R'(red) is incompletely dominant over 'r' (white) ratio of 1 : 2 : 1.
- In this case, the genotypic ratio is the same i.e., 1 : 2 : 1.

Previous Year's Question

F_1 hybrid is intermediate between the two parents.This phenomenon is
(1) dominance
(2) blending inheritance
(3) codominance
(4) incomplete dominance

Previous Year's Question

A monohybrid cross between two plants.one having 24 cm long internodes and other having 12 cm internodes and the other having 12 cm internodes produced F_1 hybrids all having 18 cm long internodes.This is a case of
(1) Multiple allelism
(2) Complete dominance
(3) Recessive dominance
(4) Incomplete dominance

Previous Year's Question

I:2:1 phenotypicand genotypic ratio is found in
(1) pseudoalleles
(2) mulyiple alleles
(3) complementary genes
(4) incomplete dominance

Explaination of Dominance

- A pair of alleles is present in a gene and in a diploid organism during gamete formation the two alleles segregate out.
- The two alleles present may be identical(homozygous) or not identical (hetezygous).
- Let us consider that the alleles produce an enzyme needed for the transformation of the substarte S.
- Mutation may occur and one of the allele may get modified and the muatated allele may produce the following -
  - the normal or less efficient enzyme
  - a non-functional enzyme
  - no enzyme at all
- In case where the normal or less efficient enzyme is produced,the modified allele produces the same effect as the unmodified allele and thus transform the substrate S.
- In the case when the allele produces a non functional enzyme or no enzyme,the substrate will not be transformed and hence the phenotype will be affected.
- The unmodified allele is the dominant allele while the modified allele is the recessive allele.
- Thus the the recessive trait is due to non-functional enzyme or because no enzyme is produced.

Rack Your Brain

Which idea is depicted by a cross in which the F_1 generation resembles both the parents?

Previous Year's Question

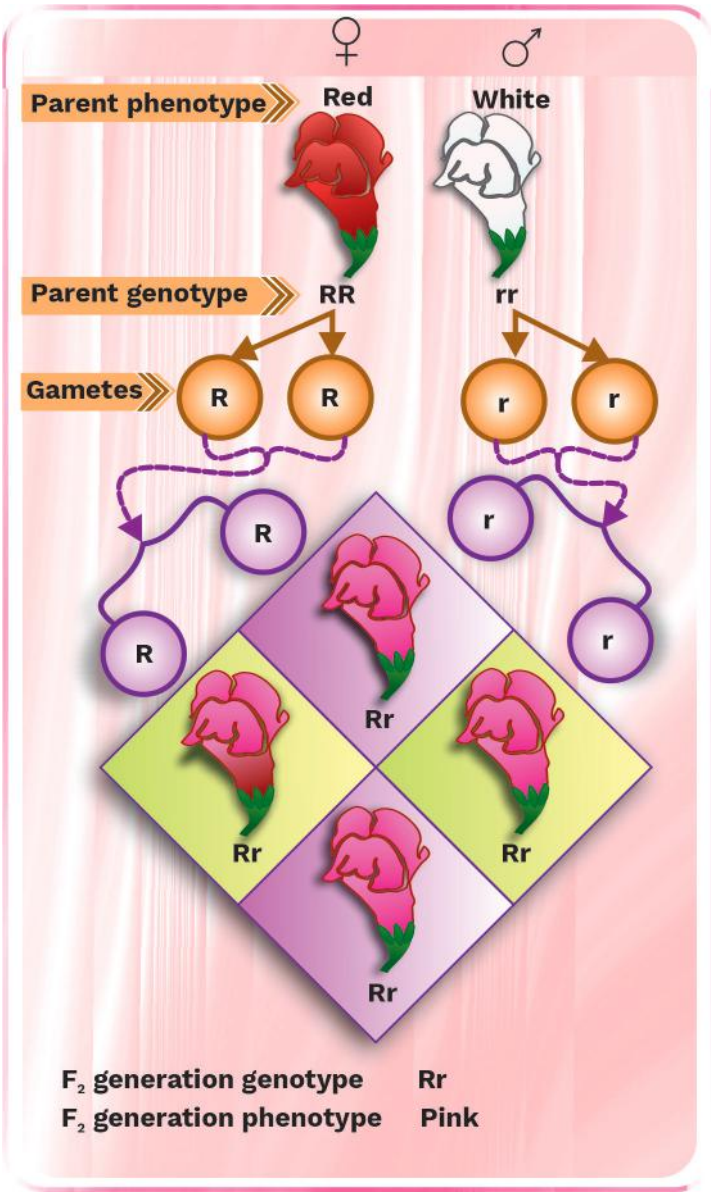
"Gametes are never hybrids" This is the satement of law of
(1) dominance
(2) segregation
(3) random fertilisation
(4) independent assortment

Previous Year's Question

The genes controlling the seven pea characters studied by Mendel are now known to be located on how many different chromosomes?
(1) 4 (2) 7
(3) 5 (4) 6

Rack Your Brain

Explain why the pink colour of flowers in Snapdragon is not termed as blending.



Previous Year's Question

Number of Barr bodies in human female is

(1) 1 (2) 2

(3) 3 (4) 4

Previous Year's Question

A child of O-group has B-group father. The genotype of father will be

(1) I^OI^O (2) I^BI^B

(3) I^AI^B (4) I^BI^O

Previous Year's Question

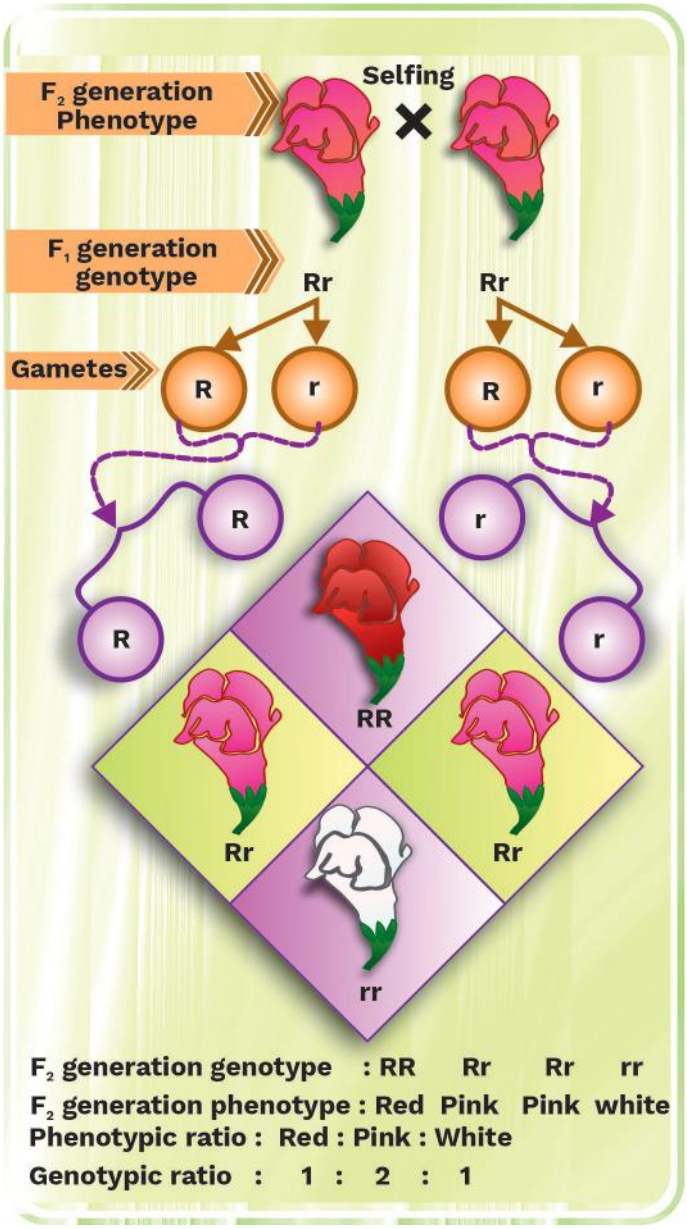
When F_1 progeny of Snapdragon plant is selfed,theexpected phenotypic and genotypic ratios res[ectively are

(1) 3:1 and 2:1:2

(2) 1:2:1 and 3:1

(3) 1:1 and 1:1:1

(4) 1:2:1 and 1:2:1



MULTIPLE ALLELE AND CODOMINANCE

- A set of more than two genes present on the same loci controlling the same characteristic is known as multiple allelic genes. Example-ABO blood group type in human beings .'ABO blood groups are controlled by the gene I.

Previous Year's Question

Lack of independent assortment between two gene A and B would be due to
(1) crossing over
(2) linkage
(3) repulsion
(4) recombination

Previous Year's Question

ABO blood group system is due to
(1) multifactor inheritance
(2) incomplete dominance
(3) multiple allelism
(4) epistasis

Previous Year's Question

What would be the colour of the flowers of F_1 progeny as a result of cross between homozygous red and homozygous white flower Snapdragon?
(1) Pink
(2) Red
(3) White
(4) Both (2) and (3)

- Sugar polymers protrudes from the surface of plasma membrane of the red blood cells and The formation of sugar is controlled by the gene.
- The gene I has three alleles I^A, I^B and i.
- I^A and I^B are completely dominant over i
- A' blood group have 'A' type of glycoproteins (a-antigen).
- 'B' type blood group have 'B' type glycoprotein (b-antigen)
- 'AB' have both types of antigens whereas 'O' group do not have any glycoproteins on RBC.
- Thus, six genotypic combinations are possible with three alleles.

Alleles of blood group of parent	Alleles of blood group of parent	Possible Genotypes of offspring	Possible Blood groups of offspring
I^AI^A	I^AI^A	I^AI^A	A
I^AI^A	I^AI^B	I^AI^A, I^AI^B	A, AB
I^AI^A	ii	I^Ai, I^Ai	A
I^Ai	ii	I^Ai, ii	A, O
I^AI^A	I^BI^B	I^AI^B	AB
I^BI^B	I^AI^B	I^AI^B, I^BI^B	AB, B
I^BI^B	ii	I^Bi	B
I^Bi	ii	I^Bi, ii	B, O
I^BI^B	I^AI^A	I^AI^B	AB
I^Bi	I^AI^B	I^AI^B, I^BI^B, I^Ai, I^Bi	A, AB, B
I^AI^B	ii	I^Ai, I^Bi	A, B
I^AI^B	I^AI^A	I^AI^A, I^AI^B	A, AB
I^AI^B	I^BI^B	I^AI^B, I^BI^B	AB, B

Alleles of blood group of parent	Alleles of blood group of parent	Possible Genotypes of offspring	Possible Blood groups of offspring
I^AI^B	I^AI^B	I^AI^A, I^AI^B, I^BI^B	A, AB, B
ii	*ii*	*ii*	O
ii	I^BI^B	I^Bi	B
ii	I^AI^A	I^Ai	A
ii	I^AI^B	I^Ai, I^Bi	A, B

Codominance

- In ABO blood group, both I^A and I^B are dominant over 'i'. As the person with genotype I^A and I^B produces both antigens; the allele I^A or I^B are called as codominant and are expressed as AB.
- All cases of multiple allelism need not to be codominant.

Pleiotropic Genes

- A single gene controlling many phenotypic characters is called pleiotropic gene and this ability of a gene to have many effects is called pleiotropy.
- For example-Starch synthesis in pea seeds is controlled by one gene. 'B' and 'b' are its two alleles.
- Starch is synthesised effectively by homozygous (BB) and large starch grain is produced.
- Homozygenous (bb) have lesser efficiency in starch synthesis and produce smaller starch grains.
- After maturation of the seeds, BB genotype seeds are round and the bb genotype seeds are wrinkled.
- Heterozygotes (Bb) produce round seeds, so, B seems to be the dominant allele. The starch grains produced are of intermediate size in Bb genotype seeds.

Previous Year's Question

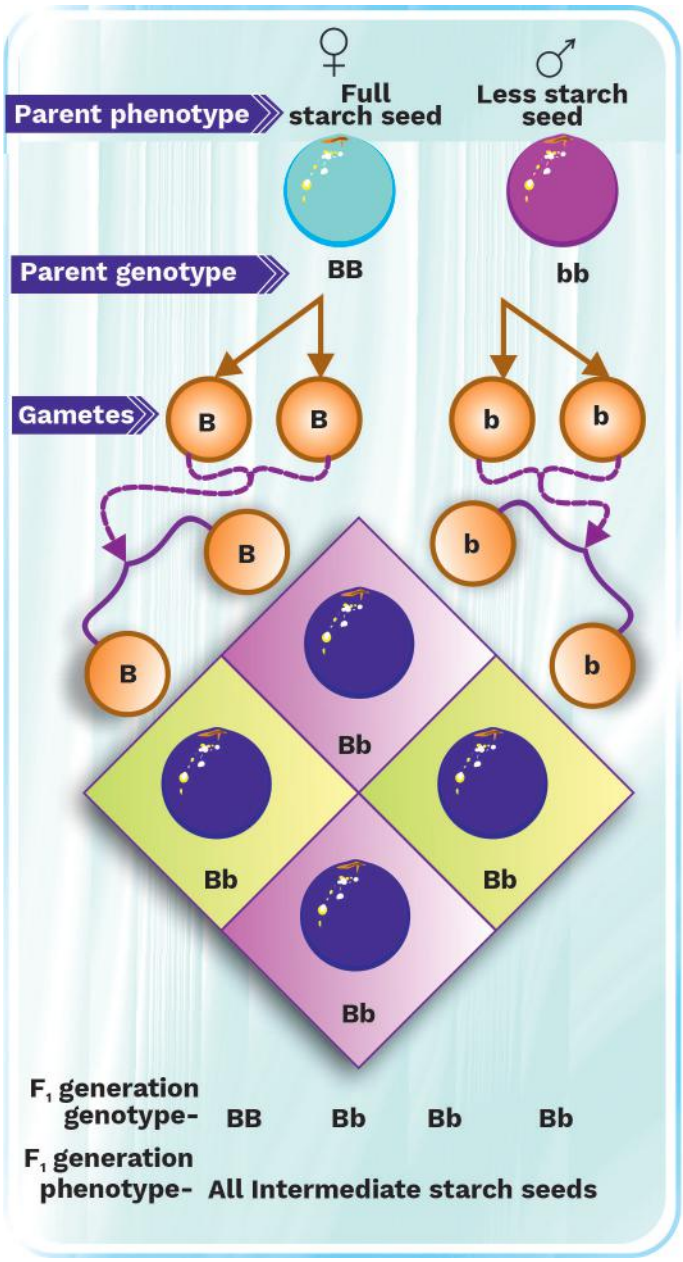
ABO blood groups in humans are controlled by the gene *I*. It has three alleles I^A, I^B and i. Since there are three different alleles, six different genotypes are possible. How many phenotypes can occur?

(1) Three (2) One
(3) Four (4) Two

Definition

Pleiotropic Genes: A single gene expressing more than one phenotypic expression .

- The starch synthesis shows incomplete dominance while the seed shape shows dominant recessive inheritance pattern.



Previous Year's Question

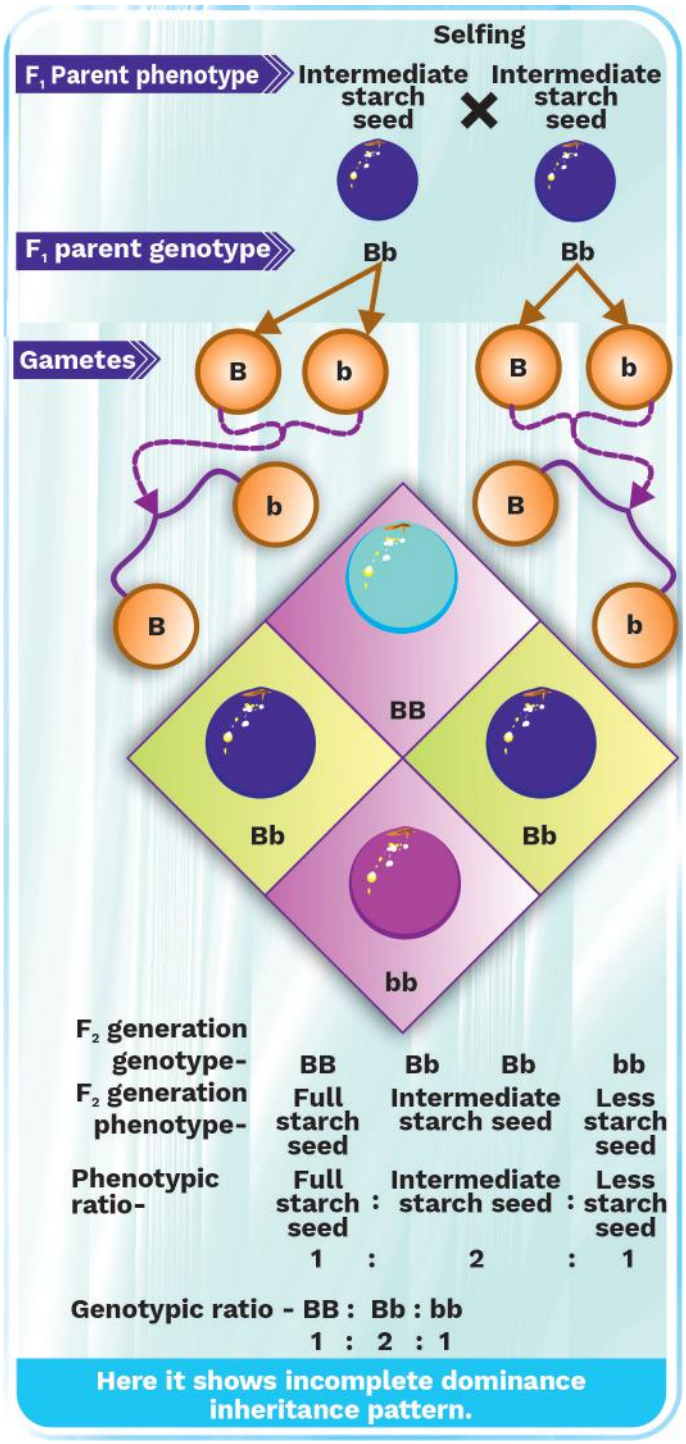
Which of the following is an example of pleiotropy?
(1) Haemophilia
(2) Thalassemia
(3) Sickle cell anaemia
(4) Colour blindness

Previous Year's Question

Pleiotropic gene
(1) inhibits crossing over
(2) promotes crsooing over
(3) controls only one phenotype
(4) controls several phenotype

Previous Year's Question

An agent that promotes occurrence of mutation is called
(1) carcinogen
(2) mutagen
(3) muton
(4) both (2) and (3)



Previous Year's Question

In Drosophila, gene for white eye mutation is also responsible for depigmentation of the body parts.Thus the gene that controls several phenotype is called

(1) oncogenes
(2) epistatic gene
(3) hypostatic gene
(4) pleiotropic

Previous Year's Question

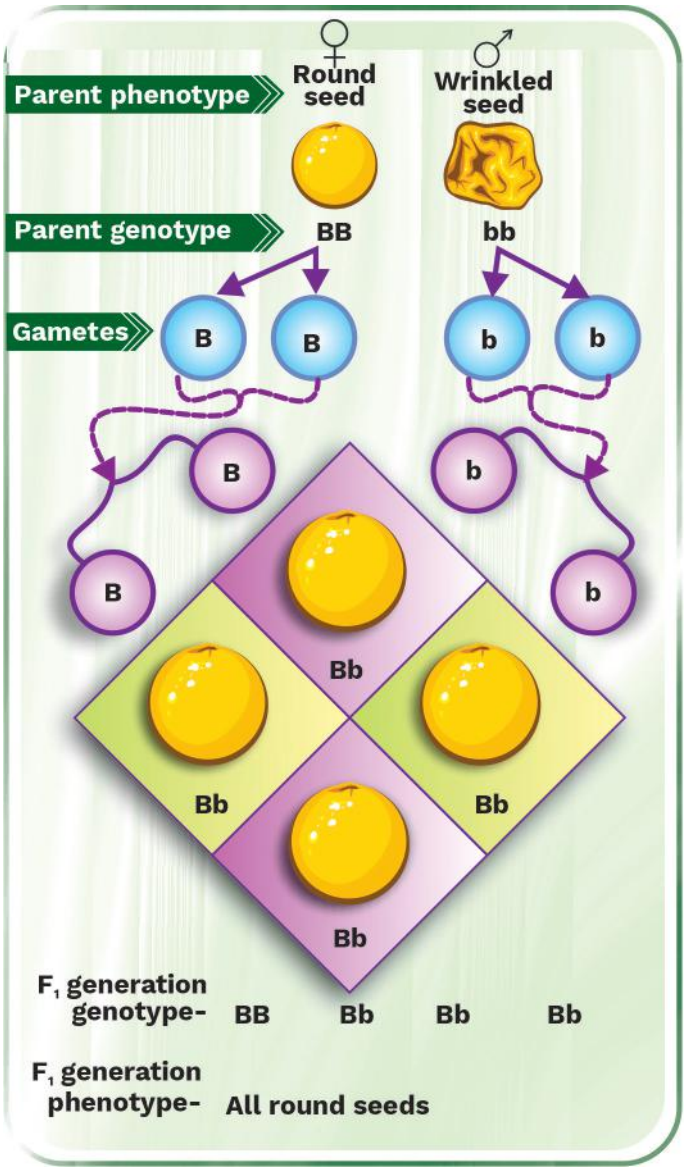
What is true in case of honey bee?

(1) Male diploid, female haploid
(2) Male diploid, female diploid
(3) Male haploid, female haploid
(4) Male haploid, female diploid

Previous Year's Question

The genetic ratio of 9:3:3:1 is due to

(1) segregation of characters
(2) crossing over of chromosomes
(3) independent assortment of genes
(4) homologous pairing between chromosomes



Previous Year's Question

In a plant, red fruit (R) is dominant over yellow fruit (r) and tallness (T) is dominant over shortness (t). If a plant with RRTt genotype is crossed with a plant that is a rrtt

(1) 25% will be tall with red fruits
(2) 50%will be tall with red fruits
(3) 75% will be tall with red fruit
(4) All the offsprings will be tall with red fruit

Previous Year's Question

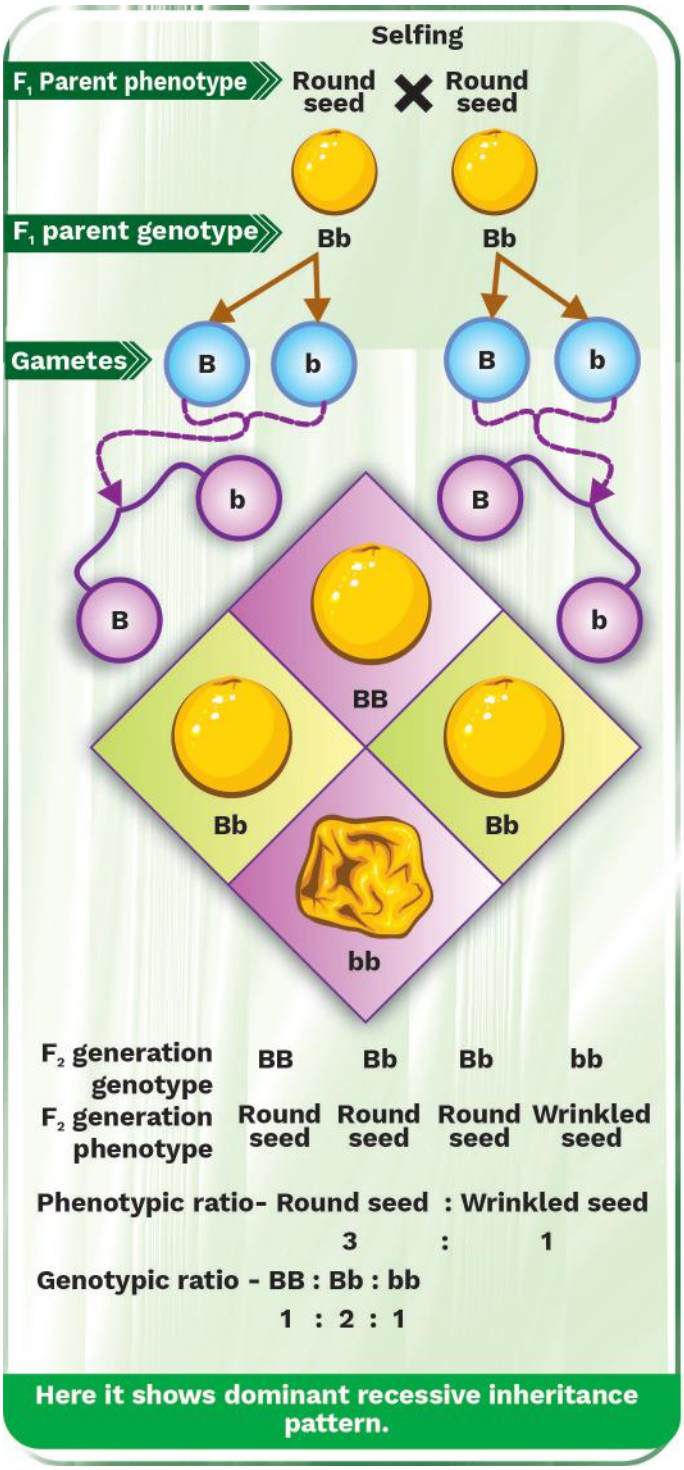
A single recessive allele which can express is effect should occur on

(1) any autosome
(2) any chromosome
(3) X-chromosome of female
(4) X-chromosome of male

Previous Year's Question

Right handedness is dominant over left handedness.Most probable gene types with two right handed parents having left handed child is

(1) RRx rr (2) RRxRr
(3) Rr xRr (4) Rrx RR



Rack Your Brain

Name the respective pattern of inheritance where phenotype
(i) is between the two parents
(ii) resembles only one of the two parents.

Previous Year's Question

A colour-blind man marries a woman who is homozygous for normal colour vision, the probability of their son being colour-blind is
(1) 0 (2) 0.5
(3) 0.75 (4) 1

Previous Year's Question

The polygenic genes show
(1) different karyotypes
(2) different genotypes
(3) different phenotypes
(4) none of these

Previous Year's Question

When a dihybrid cross is fit into a Punnett square with 16 boxes, the maximum number of different phenotypes available are
(1) 3 (2) 9
(3) 4 (4) 1

- Sickel cell Anaemia is also an example of pleitropic gene.In this the person with genotype HBsHBS produced sickel celled red blood cells and also there is damage to the internal organs of the person.
- Disease phenylketonuria, which occurs in
- humans is also an another exaple of pleiotropic gene.
- The disease is caused by mutation in the gene that codes for the enzyme phenyl alanine hydroxylase.
- Due to the mutation of the gene the person shows mental retardation and a reduction in hair and skin pigmentation.

POLYGENIC INHERITANCE

- If more than one gene controls the expression of a single trait then they are known as polygenic genes.
- The inheritance of a trait is called quantitative inheritance or multiple factor inheritance or polygenic inheritance (AA, BB, CC).
- Example- Height in humnas.In humans we just do not have tall and short but a whole range of different heights.
- The inheritance of skin colour in man (aa, bb, cc).
- The skin pigmentation is determined by three two pairs of alleles.
- AABBCC and aabbcc
- The genotype aabbcc expresses very light skin colour .
- The genotype AABBCC expresses very dark skin colour.
- The effect of the other different combination of the allleles is additive.
- The effect of all the genes is additive.

Definition

Polygenic Inheritance: The inheritance in which more than one gene controls the expression of a single trait.

Previous Year's Question

A gene is said to be dominant if it
(1) never expresses in any condition
(2) it expresses only in heterozygous condition
(3) it expresses its effect only in homozygous condition
(4) it expresses both in heterozygous and homozygous condition

Previous Year's Question

Which one of the following traits of garden peastudied by Mendel is a recessive trait?
(1) Green pod colour
(2) Round seed shape
(3) Green seed shape
(4) Axial flower position

Previous Year's Question

The phenotypic ratio obtained in quatitative inheritance of a dihybrid cross is
(1) 1:2:1
(2) 9:3:3:1
(3) 1:4:6:4:1
(4) 1:6:15:20:15:6:1

♀ \ ♂	ABC	aBC	AbC	ABc	abC	Abc	aBc	abc
ABC	AABBCC Very dark	AaBBCC Dark	AABbCC Dark	AABBCc Dark	AaBbCC Fairly dark	AABbCc Fairly dark	AaBBCc Fairly dark	AaBbCc Intermediate
aBC	AaBBCC Dark	aaBBCC Fairly dark	AaBbCC Fairly dark	AaBBCc Fairly dark	aaBbCC Intermediate	AaBbCc Intermediate	aaBBCc Intermediate	aaBbCc Fairly light
AbC	AABbCC Dark	AaBbCC Fairly dark	AAbbCC Fairly dark	AABbCc Fairly dark	AabbCC Intermediate	AAbbCc Intermediate	AaBbCc Intermediate	AabbCc Fairly light
ABc	AABBCc Dark	AaBBCc Fairly dark	AABbCc Fairly dark	AABBcc Fairly dark	AaBbCc Intermediate	AABbcc Intermediate	AaBBcc Intermediate	AaBbcc Fairly light
abC	AaBbCC Fairly dark	aaBbCC Intermediate	AabbCC Intermediate	AaBbCc Intermediate	aabbCC Fairly light	AabbCc Fairly light	aaBbCc Fairly light	aabbCc Light
Abc	AABbCc Fairly dark	AaBbCc Intermediate	AAbbCc Intermediate	AABbcc Intermediate	AabbCc Fairly light	Aabbcc Fairly light	AaBbcc Fairly light	Aabbcc Light
aBc	AaBBCc Fairly dark	aaBBCc Intermediate	AaBbCc Intermediate	AaBBcc Intermediate	aaBbCc Fairly light	AaBbcc Fairly light	aaBBcc Fairly light	aaBbcc Light
abc	AaBBCc Intermediate	aaBbCc Fairly light	AabbCc Fairly light	AaBbcc Fairly light	aabbCc Light	Aabbcc Light	aaBbcc Light	aabbcc Very light

Phenotypic ratio - Very Dark : Dark : Fairly Dark : Intermediate : Fairly light : Light : Very light
1 : 6 : 15 : 20 : 15 : 6 : 1

TABLE : NON MENDELIAN INTERGENIC GENE INTERACTION

Type of Gene Interaction	Definition	Example	Ratio
Epistatic Gene Interaction	A gene that masks the action of other gene is termed as epistatic gene. The process is epistasis . The gene whose effect is masked is known as hypostatic gene. Epistasis is of two types- **Recessive epistasis -** The recessive allele in homozygous condition masks the activity of the dominant allele. **Dominant Epistasis-**The dominant allele masks the expression of all the other alleles.	The coat colour in mice(Agouti and albino) Types of fruit colour in *ucurbita pepo* (Yellow, white and green)	**9 : 3 : 4** **12 : 3 : 1**
Complementary Gene Interaction (Duplicate Recessive Interaction)	Two genes when present together produce an effect but neither of them alone can produce the same effect.	Purple colour flower in sweet pea	**9 : 7**
Supplementary Gene Interaction	A pair of genes in which the dominant allele produces its effect irrespective of the other gene. The second allele can produce its effect only in the presence of the first allele.	Coat colour of mice	**9 : 3 : 4**
Duplicate Dominant Genes Interaction	The dominant alleles of two gene produce the same effect when present together or separately.	The shape of the capsule of Shepherd (Triangle and top shaped)	**15 : 1**

CHROMOSOMAL THEORY OF INHERITANCE

- Walter Sutton and Theodore Boveri noted that the behaviour of chromosomes was parallel to the behaviour of genes and used chromosome movement to explain Mendel's laws.
- The important things to remember are that chromosomes as well as genes occur in pairs. The two alleles of a gene pair are located on homologous sites on homologous chromosomes.
- Chromosomes as well as genes occur in pairs.
- The two alleles of a gene pair are located on homologous sites on homologous chromosomes
- The chromosomes segregate at the time of gamete formation and along with them the alleles present on the chrosomosomes also segregate out .

Previous Year's Question

Chromosomal Theory of inheritance was based on
(1) sex linkage
(2) segregation of genes
(3) diploidy and haploidy
(4) presence of sex chromosome

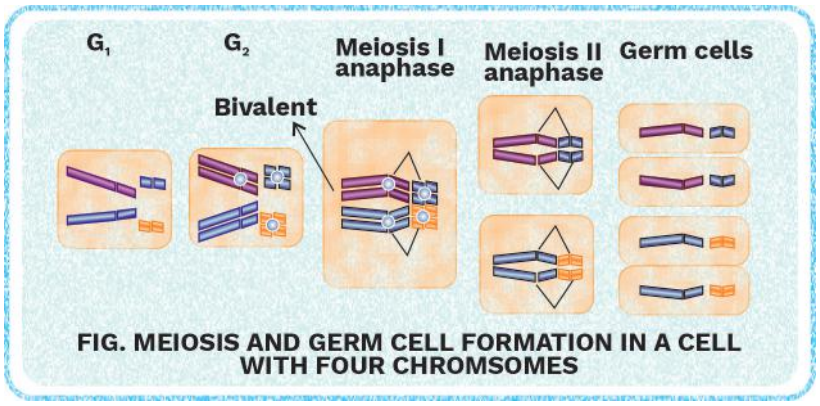


FIG. MEIOSIS AND GERM CELL FORMATION IN A CELL WITH FOUR CHROMSOMES

- Both the chromosomes and alleles occur in pairs One pair of chromosomes segregate independently of another pair.While in genes independent pairs segregate independently of each other.
- During gamete formation in meiosis I ,during Anaphase the two chromosome pairs align at the metaphase plate independently of each other.
- Each gamete has only one allele of each gene present in the chromosome.

Previous Year's Question

Which genotype and phenotype is a result of aneuploidy in sex chromosomes?
(1) 22 pairs + XXY male
(2) 22 + XX female
(3) 22 + pairs + XXXY female
(4) 22 pairs + Y female

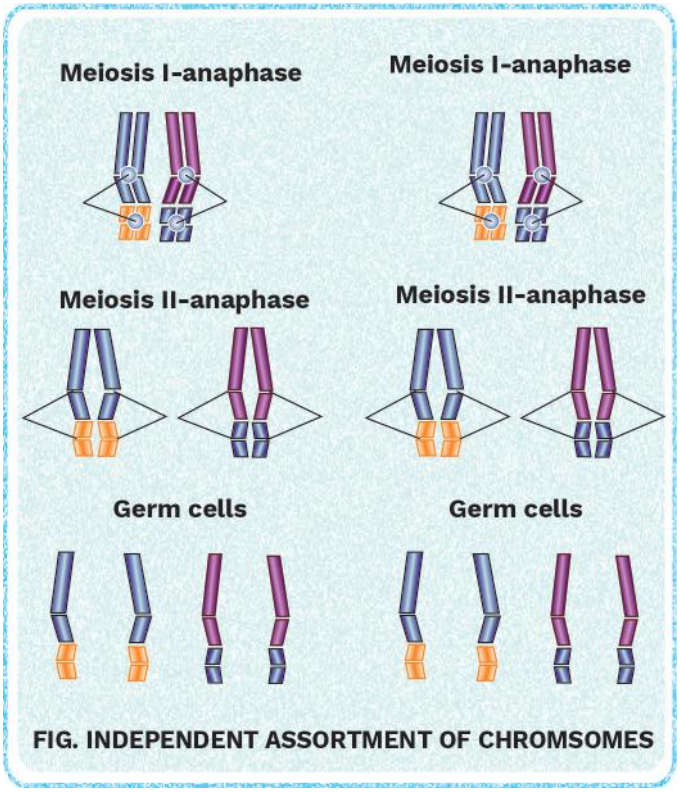


FIG. INDEPENDENT ASSORTMENT OF CHROMSOMES

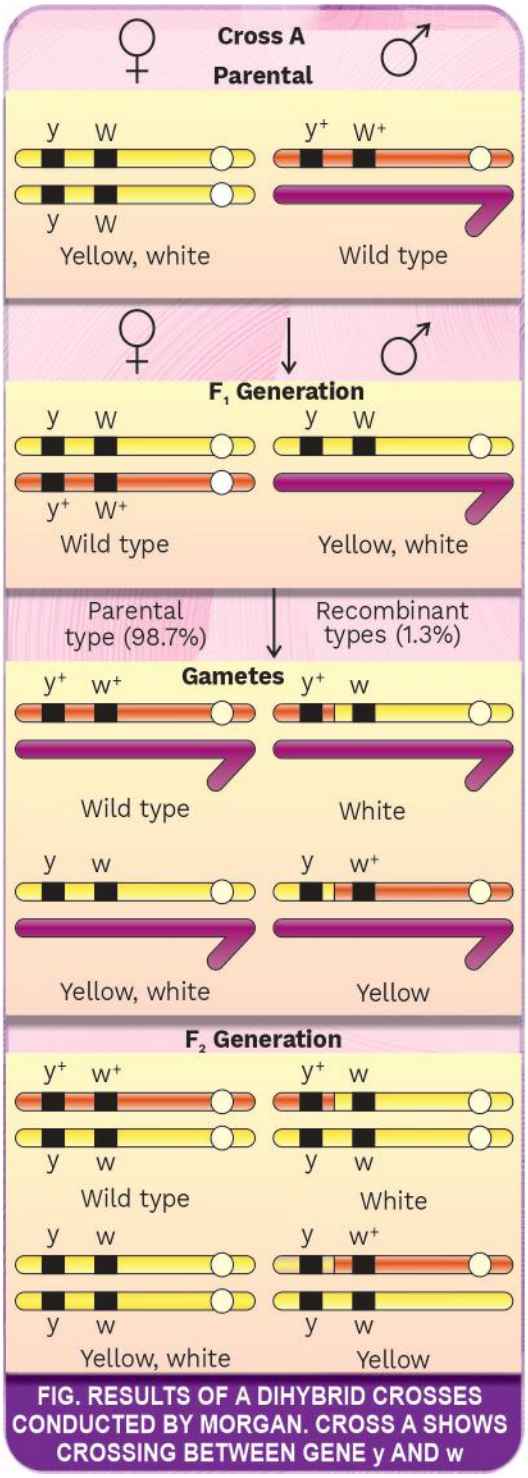


FIG. RESULTS OF A DIHYBRID CROSSES CONDUCTED BY MORGAN. CROSS A SHOWS CROSSING BETWEEN GENE y AND w

LINKAGE AND RECOMBINATION

- T.H. Morgan conducted experiments with *Drosophila* (fruit fly) to show that genes are located on chromosomes.
- He selected *Drosophila melanogaster* for his experiments due to following reasons:
- *Drosophila* has only 4 chromosomes.
- Life cycle is short of *Drosophila*.
- A large number of progenies may be produced in one mating only.
- Easy laboratory conditions are sufficient for experiment.
- Each cell of *Drosophila* consists of four pairs of chromosomes. They all are different in size.
- Out of four pairs, three are autosomes and similar in males and females. The fourth pair is allosome or sex chromosome.
- T.H. Morgan conducted following experiments (crosses) on Drosophila (normally red-eyed).

- Cross A-He crossed yellow- bodied and white eyed female with brown -bodied,red eyed male(wild type) .
- The F1 progeny was intercrossed.The result did not follow the mendelian ratio of a dihybrid cross i.e 9:3:3:1.
- The parental combination were 98.7% and recombinats were 1.3%.
- He again conducted across between crossed white-bodied female having miniature wings with yellow-bodied male with normal wings(wild type).
- The parental combination were 62.8% while the recombinants were 37.2%.
- Morgan knew that the genes were located on the X chromosome and understood that when the two genes in a dihybrid cross were situated on the same chromosome, the proportion of parental gene combinations were much higher than the non-parental type.
- He described such an association between two genes that expresses more of the parental characteristics as linkage.
- Thus he concluded from the two crosses that to genes body colour and wings size are loosly linked and thus, more crossing over and more number of recombinants.

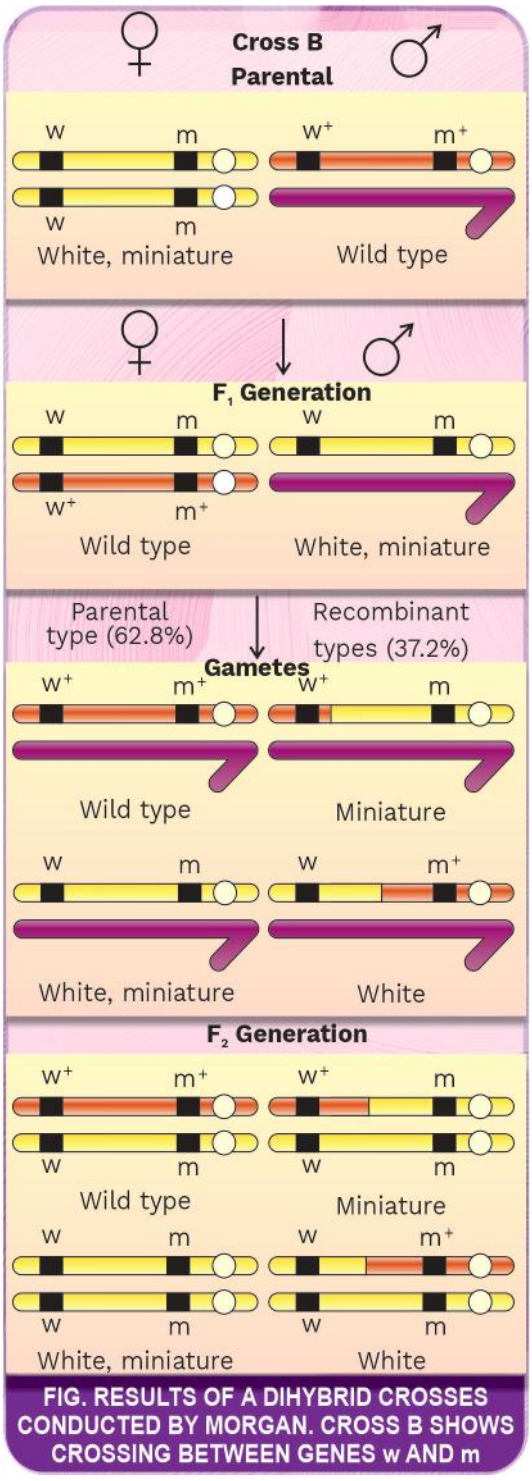


FIG. RESULTS OF A DIHYBRID CROSSES CONDUCTED BY MORGAN. CROSS B SHOWS CROSSING BETWEEN GENES w AND m

Linkage and Crossing Over

Linkage

- The tendency of genes present on a chromosome to inherit together in the next generation is known as linkage.
- Types of linkage-
  - Complete linkage
  - Incomplete linkage

Complete Linkage

- In this process, two or more genes on a chromosome are close to each other and inherit together in the next generation. In this process, there is no crossing over between the genes.

Incomplete Linkage

- In this process, the genes are not too close to each other and a little crossing over takes place between the genes. They segregate and are inherited independently, and thus, a new type of combinations are produced in the successive generation.

Linkage Group and Linkage Map

Linkage group

- The group of linked genes inherited together in the progeny is called linkage group.
- Each linkage group is related to a particular chromosome.
- In *Drosophila,* there are 4 pairs of chromosomes, so, 4 linkage groups in man, there are 23 linkage groups.

Linkage map

- The frequency of crossing over between the two genes is directly proportional to the relative distance between them. Morgan and Sturtevant proposed "let 1% crossing over (recombination) be equal to 1 map unit".
- Alfred Sturtevant used the frequency of recombination between gene pairs on the same chromosome as a measure of the distance between genes and he also mapped their position on the chromosome.
- Genetic maps have been used in th esequence of DNA in Human Genome Project

SEX DETERMINATION

- Establishment of sex of the individual through different ways is known as sex determination.
- In some animals like turtle it is determined by the temperature i.e the incubation temperature.
- While in Drosophila,humans,grasshopper,birds sex ids determined genetically.

Previous Year's Question

Bateson and Boveri have given us
(1) Multiple allelism
(2) Chromosomal theory of Inheritance
(3) Polygenic Inheritance
(4) Law of Dominance

Previous Year's Question

Number of linkage groups in Pisum sativum is
(1) 2 (2) 5
(3) 7 (4) 9

Previous Year's Question

The genes of different traits located on different loci on the same chromosome are
(1) alleles
(2) linked
(3) mutated
(4) pleiomorphic

Previous Year's Question

A woman has an X-linked condition on the one of her X chromosomes. This chromosome can be inherited by
(1) only grandchildren
(2) only sons
(3) only daughters
(4) both sons and daughters

- The studied on some insects lead to the understanding of genetic/chromosomal mechanism of sex determination.
- Henking found a nuclear structure during spermatogenesis of a few insects. He also found out that 50 per cent of the sperm received this structure after spermatogenesis while the other 50 per cent sperm did not receive it.
- He called the structure as the X body but he could not explain its importance.
- Later some scientists found out that the 'X body' was in fact a chromosome of sex determination and was named X chromosome.
- In *Bonnelia* the larvae do not have any sex.The larva which grows in the mud matures into female while the larva that develops near the proboscis of female develops into male.

Autosomes and Allosomes

- The chromosomes other than sex chromosomes in body cells (diploid) are called autosomes.
- The sex chromosomes which are morphologically different and are associated with sex determination are called allosomes e.g. X and Y.

Heterogametic and Homogametic Organisms

- The individual having two morphologically different sex chromosomes and producing two types of gametes is called heterogametic (XY).
- The individual having two morphologically similar sex chromosomes and producing all gametes of the same type is known as homogametic (XX).

XY Type

- In Drosophila and mammals the males have XY sex chromosome while the female have XX sex chromosome.
- The males are heterogametic as half their gametes have X and the other half have Y sex chromosomes.

Previous Year's Question

There are three genes a, b, c. Percentage of crossing over between a and b is 20%, b and c is 28% and a and c is 8%. What is the sequence of genes on chromosome?
(1) b, a, c
(2) a, b, c
(3) a, c, b
(4) None of these

Previous Year's Question

Pick out the correct statements.
(a) Haemophilia is a sex-linked recessive disease.
(b) Down's syndrome is due to aneuploidy.
(c) Phenylketonuria is an autosomal recessive gene disorder.
(d) Sickle cell anaemia is an X-linked recessive gene disorder.
(1) (a), (c) and (d) are correct.
(2) (a), (b) and (c) are correct.
(3) (a) and (d) are correct.
(4) (b) and (d) are correct.

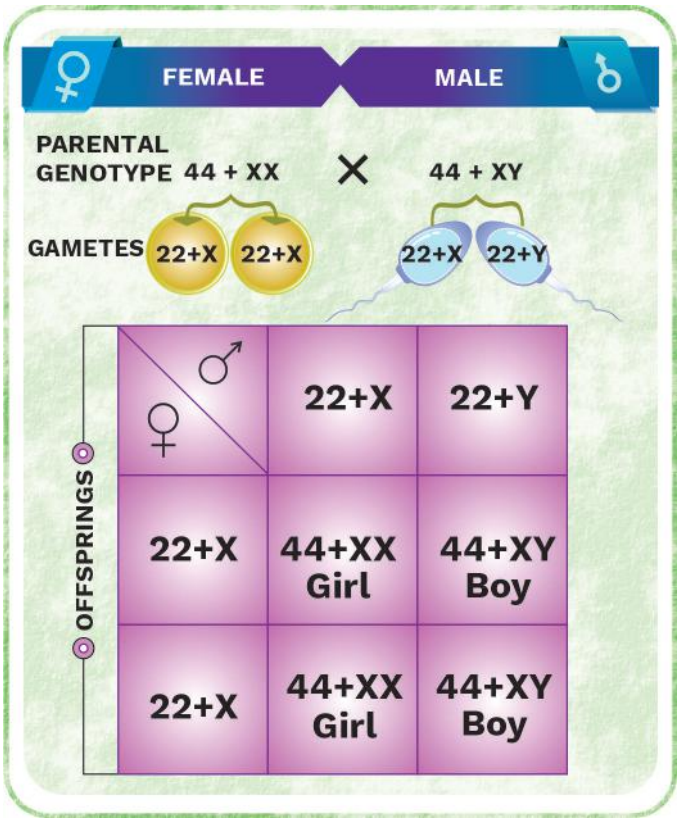
Previous Year's Question

Genetic identity of a human male is determined by
(1) sex-chromosome
(2) cell organelles
(3) autosome
(4) nucleolus

- The female is homogametic as all her gametes contain only one X sex chromosome.

Sex Determination in Humans

- Humans have 23 pairs of chromosome.Out of which 22 pairs are autosomes and the 23rd pair is the sex chromosome.
- During spermatogenesis,half the gametes in male have X sex chromosome i.e 22+X while the other half gametes have Y sex chromosome i.e 22 +Y.
- The female during oogenesis produce ova that has always will have X sex chromosome.Thus the gametes will be 22+X.
- Eggs fertilised by sperm having 22+X chromosomes become females while those fertilised by sperms that have 22+Y chromosome become males.



- It is the genetic makeup of the sperm that determines the sex of the child.

Rack Your Brain

Which mechanism causes a gene to move from one linkage group to another?

Previous Year's Question

Sex chromosomes of males are
(1) autosomes
(2) hemizygous
(3) homozygous
(4) heterozygous

Previous Year's Question

Human female possesses 44+XX chromosomes. The secondary oocyte shall have
(1) 22 (2) 44
(3) 22+X (4) 44+XX

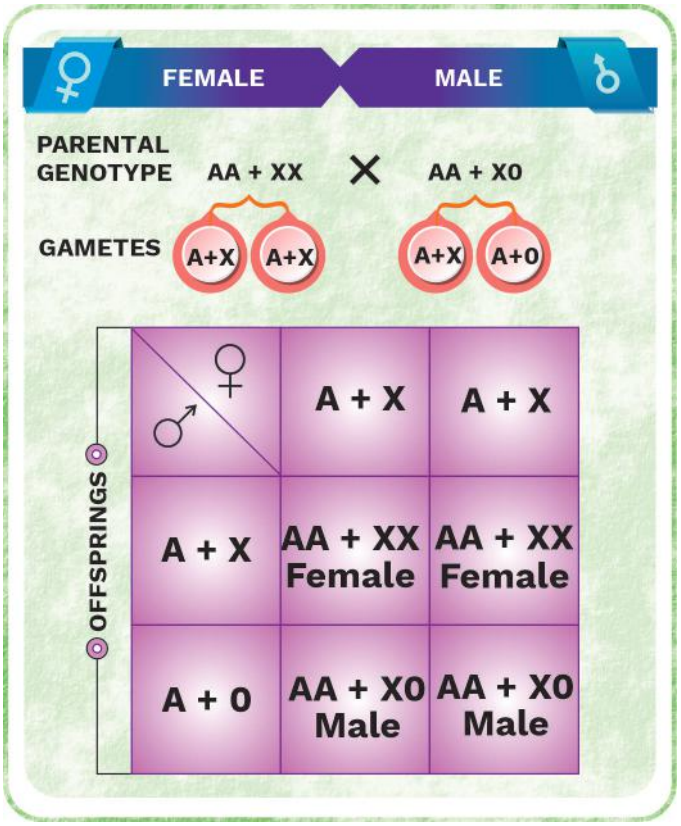
Previous Year's Question

A hereditary disease which is seldom passed from father to son is
(1) autosomal linked disease
(2) X-chromosomal linked disease
(3) Y-chromosomal linked disease
(4) none of the above

- During pregnancy there is always 50 per cent probability of either a male or a female child.
- But in India in some places female is blamed for the birth of a girl child.

"XO" Type

- This type of chromosome is found in certain insects and roundworms. It is produced due to non-disjunction of chromosomes during meiosis and then the egg is fertilised by a sperm lacking X-chromosome (Male having only one X chromosome i.e., XO and the female having two X-chromosomes).



"ZW" Type

- In birds, two different sex chromosomes of a female bird has been designated to be the Z and W chromosomes. In these organisms, the females

Previous Year's Question

The males of grashopper and bugs possess two sets of autosomes and
(1) only Y-chromosome
(2) only X-chromosome
(3) Xand Y chromosome
(4) Neither X nor Y chromosome

Previous Year's Question

Sex chromosomes are also called
(1) allosomes
(2) autosomes
(3) hybridization
(4) all of these

Definition

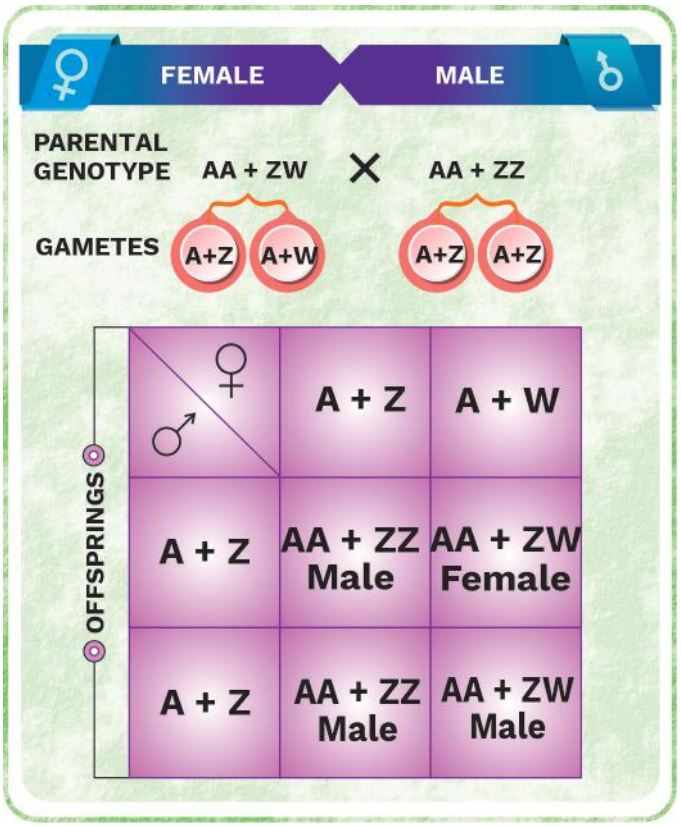
Mutation: A heritable change in the structure of a gene or the chromosome or a change in the number of chromosomes.

Previous Year's Question

There are three genes *a, b, c*. Percentage of crossing over between *a* and *b* is 20%, *b* and *c* is 28% and *a* and *c* is 8%. What is the sequence of genes on chromosome?
(1) *b, a, c*
(2) *a, b, c*
(3) *a, c, b*
(4) None of these

have one Z and one W chromosome, whereas males have a pair of Z-chromosomes besides the autosomes.

- The female is heterogametic as half the gametes produced have Z sex chromosome and half the gametes have W sex chromosome.
- The male is homogametic as all the gamertes have only Z sex chromosme.



Sex Determination in Honey Bee

- Sex determination in honey bee is based on the number of sets of chromosomes received by an individual.
- Worker or the queen has diploid number of chromosome i.e 32 while the male i.e the drones has only haploid number of chromosome i.e 16 as they are produced parthenogenetically.
- Sex determination in honey bees is haplodiploid.

Previous Year's Question

Sex chromosomes of a female bird are represented by

(1) XO (2) XX
(3) ZW (4) ZZ

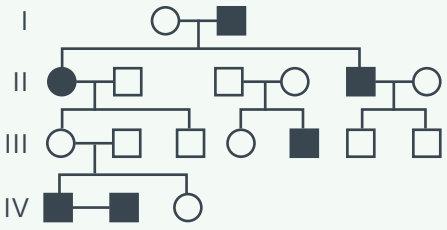
Previous Year's Question

Heterogametic male condition does not occur in

(1) birds
(2) human
(3) Drosophila
(4) honey bee

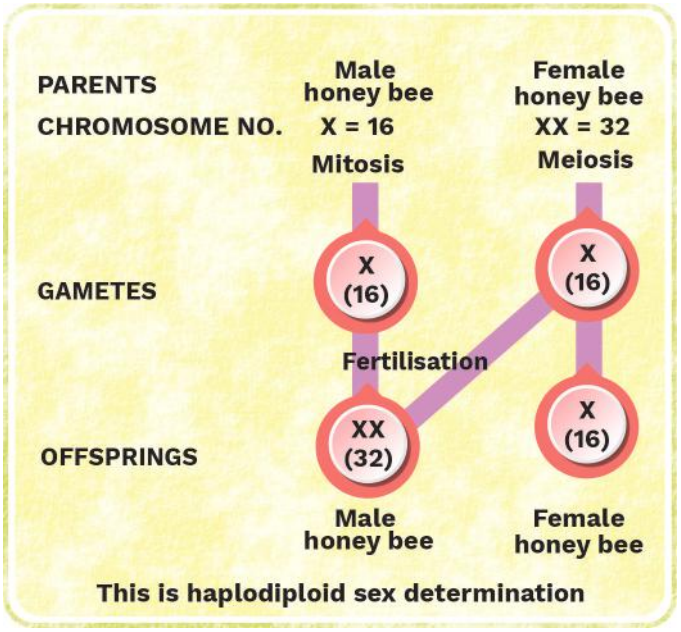
Previous Year's Question

In the following human pedigree, the filled symbols represent the affected individuals. Identify the type of given pedigree.



(1) X-linked dominant
(2) Autosomal dominant
(3) X-linked recessive
(4) Autosomal recessive

- Egg is produced by meiosis while the sperms are produced by mitosis.
- When an egg fuses with the sperm a female
- (queen or worker) is produced and an unfertilised egg develops as a male (drone).
- Male do not have father and thus cannot have sons, but they have a grandfather and can have grandsons.



MUTATION

- It may be defined as "a heritable change in the structure of a gene or the chromosome or a change in the number of chromosomes".

Causes of mutations

- Mutations can be caused by different agents and they are known as mutagens.Mutagens can be physical or chemical.
- Physical mutagens like high temerature and radiations like UV rays,alpha,beta and gama rays,X-rays.
- Chemical mutagens like mustard gas and nitrous gas.

Previous Year's Question

Thalasemia and sickle cell anaemia are caused due to a problem in globin molecule synthesis. Select the correct statement.

(1) Both are due to a quantitative defect in globin chain synthesis.
(2) Thalassemia is due to less synthesis of globin molecules.
(3) Sickle cell anaemia is due to a quantitative problem of globin molecules.
(4) Both are due to a qualitative defect in globin chain synthesis.

Previous Year's Question

Haplodiploidy pattern of sex determination is observed in

(1) birds
(2) humnas
(3) honey bees
(4) butterflies

Previous Year's Question

In humans,the key factors in sex determination is

(1) Y chromosome
(2) X chrpomosome
(3) Both (1) and (2)
(4) None of these

Types of Mutation

- **Point Mutation (gene mutation)-**Due to a change in the structure of a gene. This type of mutation normally alters the information conveyed by a gene; it alters its message.
- In this there is mutation in only one nitrogenous base of the triple codon and thus known as point mutation.
- **Frame Shift Mutation**-In this there is addition or loss of more than one nitrogenous bases.
- It result in shift of the nitrogenous bases and the reading of the codon is affected and hence the protein formed is different or may not be formed at all.
- **Chromosomal Mutation-**Due to a change in the structure or number of chromosomes. It alters only the number or position of existing genes.
- Chromosomal mutation may take place in the following four ways:
  - Deletion-Loss or deletion of a part of chromosome.
  - **Duplication-**Addition of a part of homologous chromosome to the chromosome.
  - **Inversion-**In this case, a part of chromosome may be broken and then again joined in the reverse direction.
  - **Reciprocal Translocation-**Joining of the non-homologous chromosome to the chromosome and vice versa (between non-homologous it may be called as illegitimate crossing over).
- Mutations arise by changes in chromosome number-Any change in the number of chromosomes is called ploidy.
- Types of ploidy-
  - Aneuploidy
  - Euploidy

Aneuploidy

- A change in the chromosome number which is not the exact multiple of basic haploid chromosome number is called Aneuploidy.

Previous Year's Question

Which one of the following conditions in humans is correctly matched with its chromosomal abnormality/linkage?

(1) Erythroblastosis foetalis
X-linked

(2) Down's syndrome
44 autosomes + XO

(3) Klinefelter's syndrome
44 autosomes + XXY

(4) Colour blindness Y-linked

Previous Year's Question

Genes located on differential region of Y-chromosomes are called

(1) XY linked genes

(2) holandric gene

(3) autosomal genes

(4) mutant genes

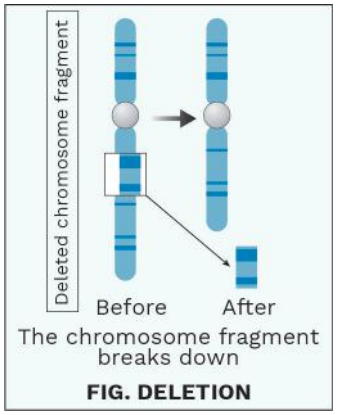


FIG. DELETION

- For example:
  - **Monosomics-**One chromosome less than the normal diploid number (2n–1) e.g., X O condition.
  - **Trisomics-**One chromosome extra (2n + 1) e.g., Down syndrome, super male (XYY), super female (XXX)

Euploidy

- A change in the chromosome number that is an exact multiple of basic haploid chromosome number is called euploidy (Polyploidy) e.g.,
  - **Triploidy-**Addition of one extra set of chromosomes (2n + n = 3n).
  - **Tetraploidy-**2 additional sets of chromosomes. Polyploidy plays an important role in the evolution of plants and is more common in plants.

PEDIGREE ANALYSIS

- An analysis of serial records of the occurrence of a trait in several generations of a human family is called pedigree analysis.
- The first thing in this analysis is to collect the family history regarding trait of a particular concern.
- Then, the family tree is made after some generations.
- By convention, the male members of the family are shown by squares and the females by circles. Solid symbols show the trait being investigated and open symbols denote normal individual for the trait.
- A symbol with a cross line indicates a carrier for a recessive trait. Parents are joined by horizontal lines and their progenies are connected to horizontal lines below the parents in order of their birth.

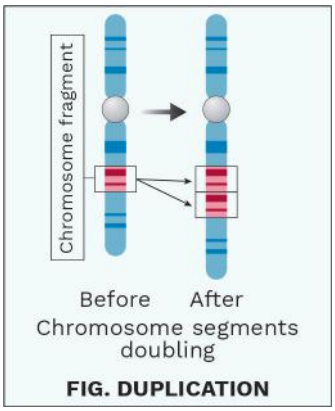


FIG. DUPLICATION

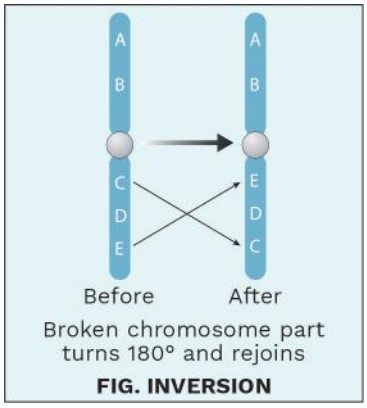


FIG. INVERSION

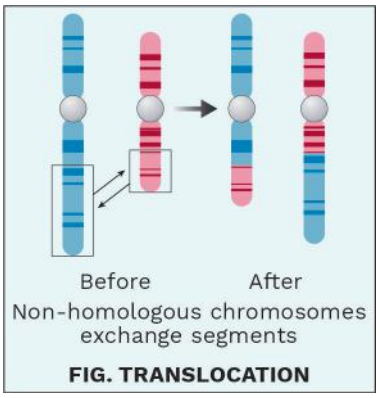


FIG. TRANSLOCATION

Utility of Pedigree Analysis

- It helps genetic counsellors to advice couples who are worried of the possibilities of having genetically defective children.
- The genotype of an individual may be known by this analysis.
- It may be useful for hereditary studies in sexually reproducing organisms having small number of offsprings.
- The information for would be couples in regard to certain genetic defect may be known.

DISORDERS

- They can be mendelian or chromosomal.

Mendelian Disorder

Sickle-Cell Anaemia

- It is an autosomal recessive disease.
- It is caused due to point muatation.
- In this the glutamic acid in the 6th position in the globin chain is replaced by valine.It is caused due to the mutation of to .
- In this disorder the red blood cells become sickel shaped.
- The genotype of the healthy individual is HB^AHB^A while the affected individual is HB^SHB^S.
- Homozygous individual for the disease die early due to destruction of the red blood cells.
- The individual who is heterozygous i.e HBAHBS is a carrier.
- The advantrage of heterozygous genotype is that they are protected from malaria.
- The malarial parasite i.e Plasmodium cannot live in the sickel celled red blood cells.

Previous Year's Question

Which of the following is not a hereditary disease?
(1) Cystic fibrosis
(2) Thalasaemia
(3) Haemophilia
(4) Cretinism

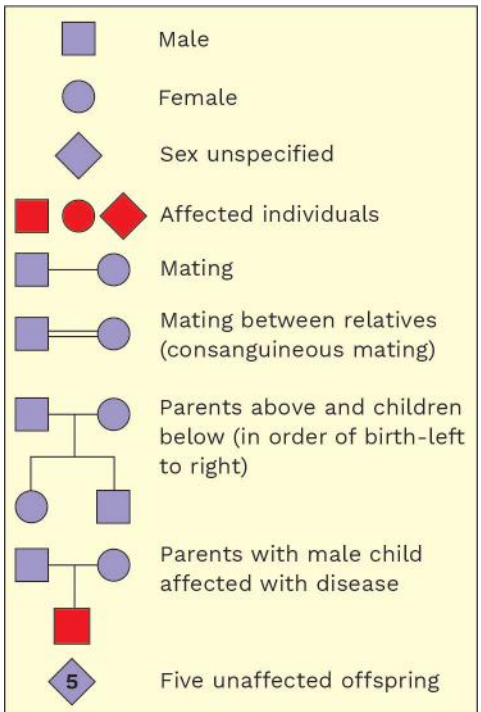


FIG. SYMBOLS USED IN THE HUMAN PEDIGREE ANALYSIS

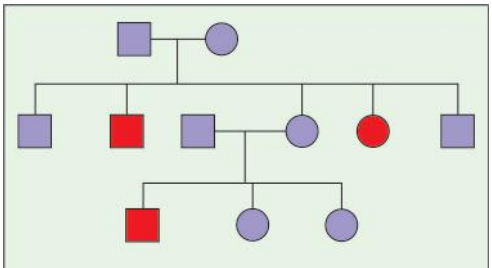
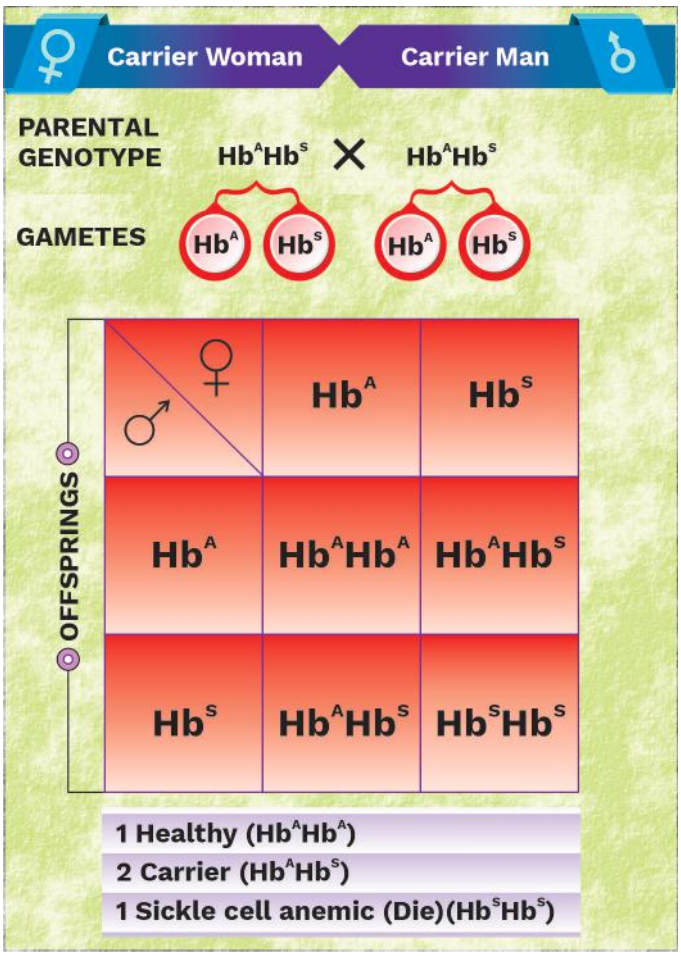


FIG. PEDIGREE ANALYSIS OF AN AUTOSOMAL RECESSIVE TRAIT



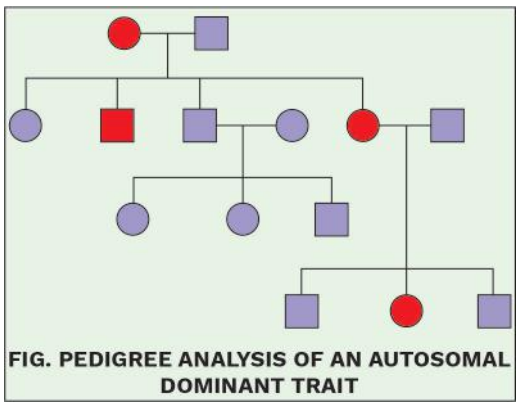


FIG. PEDIGREE ANALYSIS OF AN AUTOSOMAL DOMINANT TRAIT

Previous Year's Question

Which of the following conditions though harmful in itself,is also a potential saviour from a mosquito borne infectious disease?

(1) Pernicious anaemia
(2) Leukaemia
(3) Sickel -ceel Anaemia
(4) Thalassemia

Thalasemia

- It is an autosomal mutantional gene disorder.
- It results from defective synthesis of subunits of haemoglobin .
- Haemoglobin has two alpha and two beta chains.
- In Thalassemia the synthesis of the chains gets affected and thus it is quantitave defect.
- It is of the following types-
- Alpha thalassemia-The genes HBA1 and HBA2 is present on two loci and thus are four alleles on chromosome 16.
- A defect in the alleles on the 16th chromosome results in less or no synthesis of the alpha globulin and thus resulting in fewer aplha chains and more beta chains.

Previous Year's Question

Rearrangement of genes occurs due to

(1) translocation and duplication
(2) translocation and deficiency
(3) deletion and deficiency
(4) translocation and inversion

- Beta thalessemia-It is controlled by a single gene HBB on chromosome 11.
- Mutation in the gene would result in less synthesis of bet achain and thus more of alpha chain synthesis.
- Delta Thalessemia-In some humans delta chain is present and alpha chain synthesis does not take place.

Phenylketonuria

- It is an error that is autosomal recesssive.The affected person does not produce the enzyme phenylalanine hydroxylase that convetts amino acid phenylalanine into tryosin.
- Thus phenylalanine accumulates in the body and is converted to phenylpyruvic acid .
- This cause mental retardation and in some cases phenylpyruvic acid is secreted out in urine as the kidney is not able to absorb it.

Sex Influenced Traits

- The traits whose expression is influenced by sex hormones are known as sex influence traits e.g., Baldness is generally found in males and not in females.
- Baldness is controlled by a gene.The presence of a single dominant gene in male causes baldness while in female two recessive allele is responsible for it.Thus female usually are not bald.

Sex-linked Traits

- The traits whose inheritance occurs due to sex chromosomes are called sex-linked traits. The gene for the the trait is located on X chromosome.
- There is no corresponding gene present on the Y chromosome
- If the trait is recessive ,then one recessive allele in the X chromosome of male causes the disease but two recessives alleles in female can only cause the disease .

Previous Year's Question

The most popularly known blood grouping is the ABO grouping. It is named ABO and not ABC, because "O" in it refers to having

(1) overdominance of this type on the genes for A and B types
(2) one antibody only either anti A or anti B on the RBCs
(3) no antigens A and B on RBCs
(4) other antigens besides A and B on RBCs

Previous Year's Question

In *Drosophila*, the sex is determined by

(1) the ratio of number of X chromosomes to the sets of autosomes
(2) X and Y chromosomes
(3) the ratio of pairs of X chromosomes to the pairs of autosomes
(4) whether the egg is fertilized or develops parthenogenetically

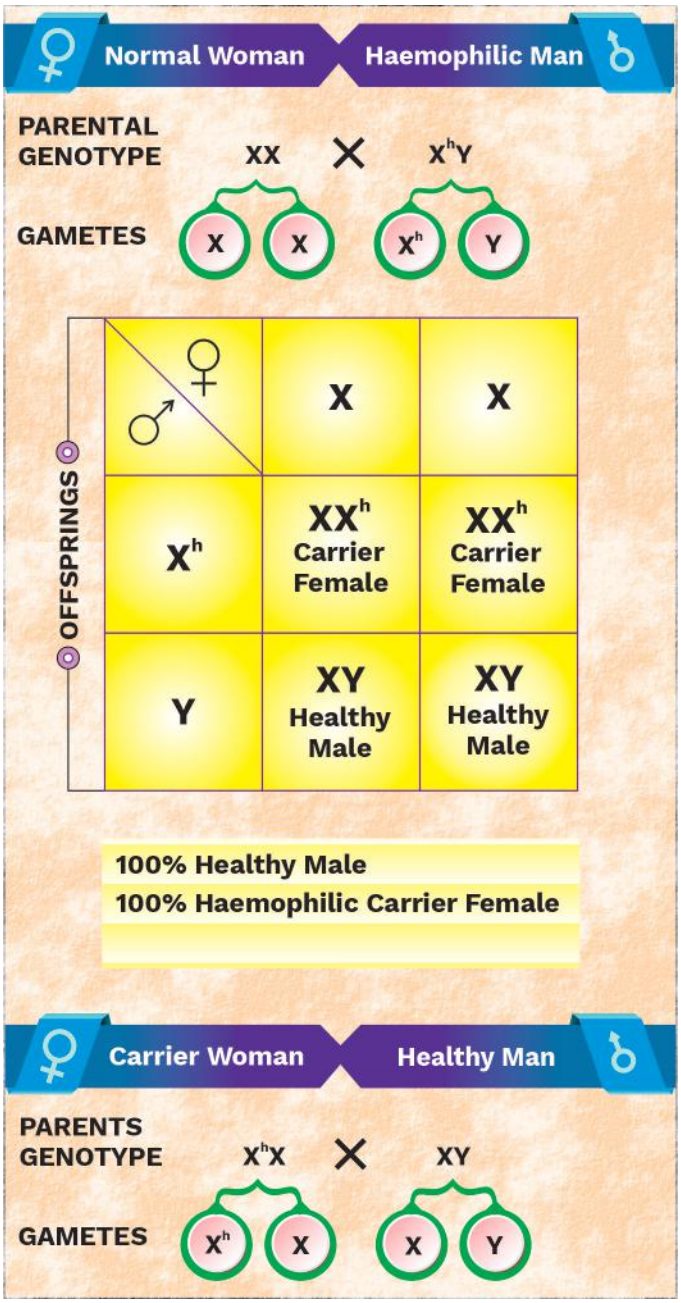
Previous Year's Question

Sex chromosomes of birds are

(1) ZZ - ZW (2) ZZ - WW
(3) XX - XY (4) XO - XX

- X linked inheritance shows a criss- cross inheritance.The male transmits the X linked gene to his grandson through his daughter.
- For example-Haemophilia, colour blindness

Haemophilia



Previous Year's Question

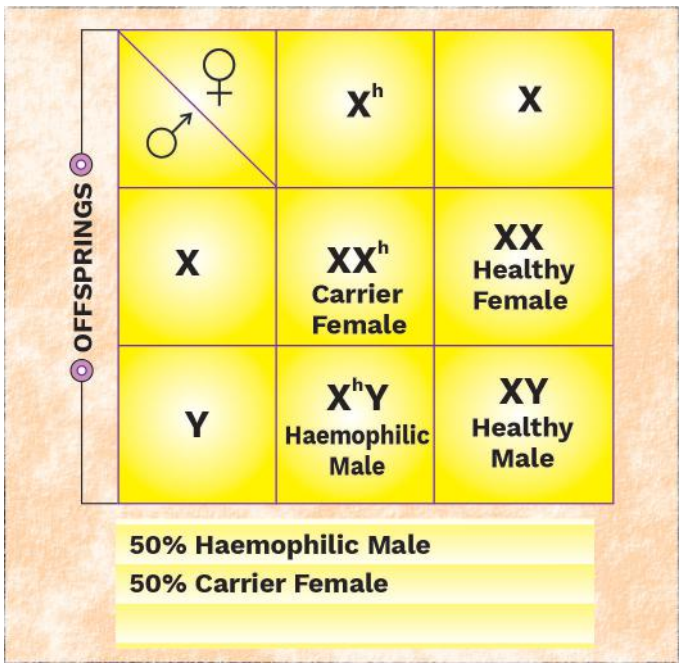
A marriage between normal visioned man and colour blind woman will produce offspring
(1) colour blind sons and 50% carrier daughter
(2) 50% colourblind sons and 50% carrier daughter
(3) normal males and carrier daughters
(4) colour blind sons and carrier daughters

Previous Year's Question

Which one of the following symbols and is representative used in human pedigree analysis is correct?
(1) O = unaffected male
(2) ... = unaffected female
(3) ● = male affected
(4) ... = O = mating between relatives

Previous Year's Question

Haemophilia is more common in males because it is a
(1) recessive character carried by Y-chromosome
(2) dominant character carried by Y-chromosome
(3) dominant trait carried by X-chromosome
(4) recessive trait carried by X-chromosome



- It is a sex linked recessive inheritance .
- The blood does not clot in the people suffering from this disorder.
- Male carrying a single recessive allele is affected while a female will be haemophilic only in the homozygous condition.
- A female carrying asingle recessive allele is a carrier.
- A female is usually not affected,as she may become haemophilic only if her father is haemophilic and mother a carrier or haemophilic.
- The male inherit the diseases from the female.

Colour Blindness

- It is also a sex linked recessive disorder.
- In this disorder the person cannot perceive red and green light.
- Sometimes blue colour is also not perceived by a person with this disorder.
- It shows the same criss-cross pattern of inheritance. Female should be homozygous recessive for the disorder to be expressed while a heterozygous male would be affected.

Rack Your Brain

Why the human male never passes the gene of haemophilia to their sons?

Previous Year's Question

Of both normal parents, the chance of a male child becoming colour blind are

(1) no
(2) possible only when all the four grand parents had normal vision
(3) possible only when father's mother was colour blind
(4) possible only when mother's father was colour blind

Previous Year's Question

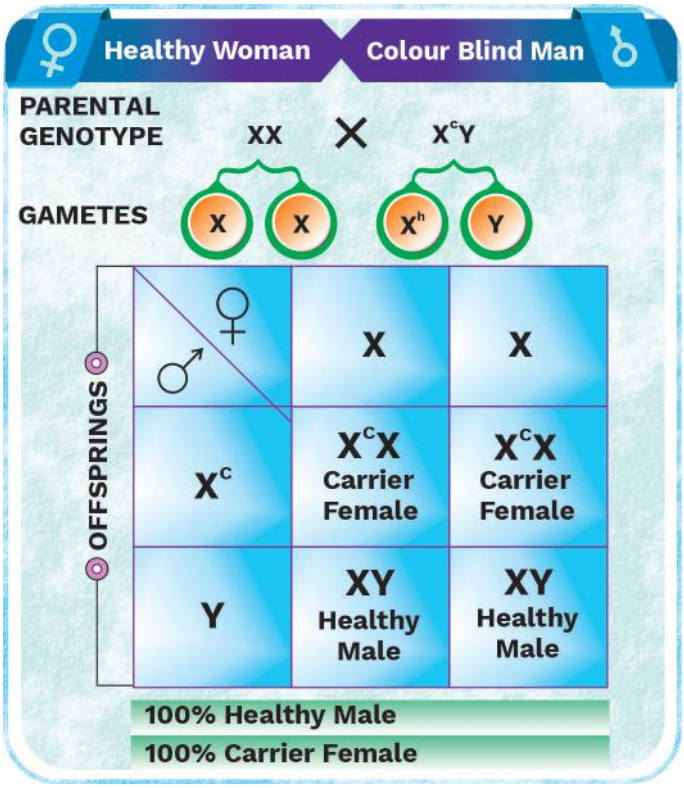
Distance between two linked genes is measured in map units that depict

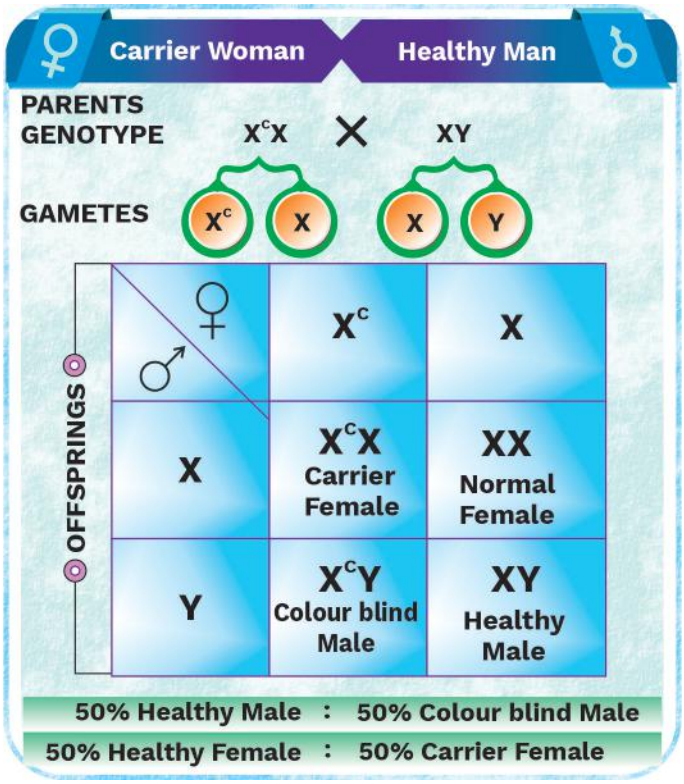
(1) ratio of crossing over between them
(2) cross-over value
(3) number of genes between them
(4) both (2) and (3)

Previous Year's Question

In Down's syndrome of a male child, the sex complement is

(1) XO (2) XY
(3) XX (4) XXY





Previous Year's Question

Sickle-cell anaemia is

(1) caused by substitution of valine by glutamic acid in the beta globin chain of haemoglobin

(2) caused by a change in a single base pair of DNA

(3) characterized by elongated sickle like RBCs with a nucleus

(4) an autosomal linked dominant trait

Rack Your Brain

A human male produces sperms with the genotypes AB, Ab, aB, and ab pertaining to two diallelic characters in equal proportions. What is the corresponding genotype of this person?

Previous Year's Question

Both husband and wife have normal vision though their fathers were colour blind. The probability of their daughter becoming colour blind is

(1) 0% (2) 25%

(3) 50% (4) 75%

CHROMOSOMAL DISORDERS

Down's Syndrome (Autosomal Disorder)

- The cause of this genetic disorder is the presence of an additional copy of the chromosome number 21 (trisomy of 21st chromosome).
- This disorder was first described by Langdon Down (1866). The affected individual is short-statured with broad flat face, wrinkled tongue and partially open mouth.
- Palm is broad with characteristic palm crease. Physical, psychomotor and mental development is retarded.

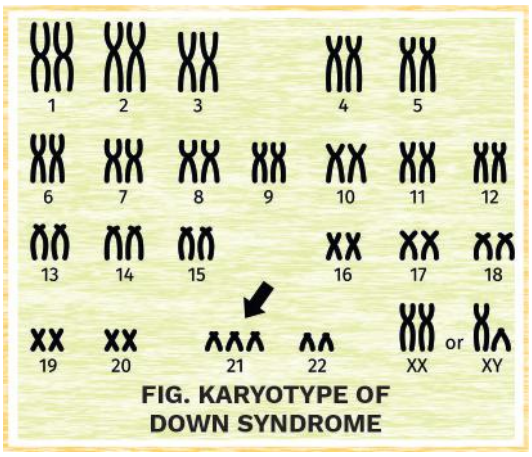


FIG. KARYOTYPE OF DOWN SYNDROME

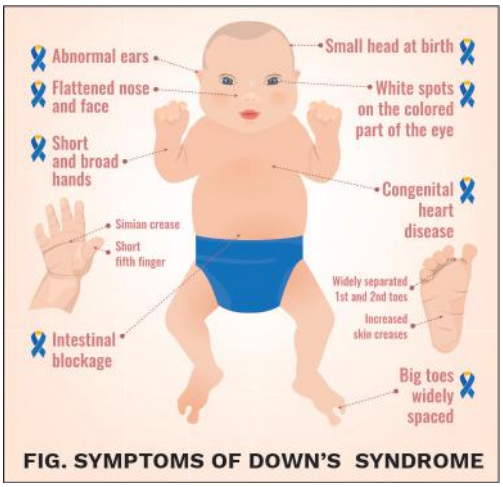


FIG. SYMPTOMS OF DOWN'S SYNDROME

Klinefelter's Syndrome

- This genetic disorder is caused due to the presence of an additional copy of X-chromosome resulting in a karyotype of 47, XXY.
- Such an individual has overall masculine development, however, the feminine development (development of breast, i.e., Gynaecomastia) is also expressed. Such individuals are sterile.

Previous Year's Question

Non genetic sex determination occurs in

(1) *Bonellia* (2) cow

(3) birds (4) fruit fly

Previous Year's Question

A normal woman, whose father was colour-blind is married to a normal man. The sons would be

(1) 75% colour-blind

(2) 50% colour-blind

(3) all normal

(4) all colour-blind

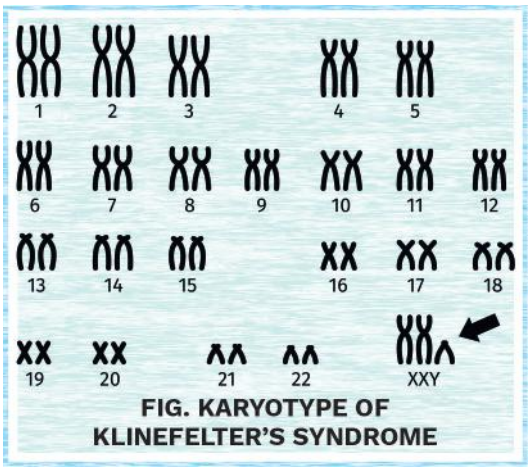


FIG. KARYOTYPE OF KLINEFELTER'S SYNDROME

Turner's Syndrome

- Such a disorder is caused due to the absence of one of the X chromosomes, i.e., 45 with X0, Such females are sterile as ovaries are rudimentary besides other features including lack of other secondary sexual characters.

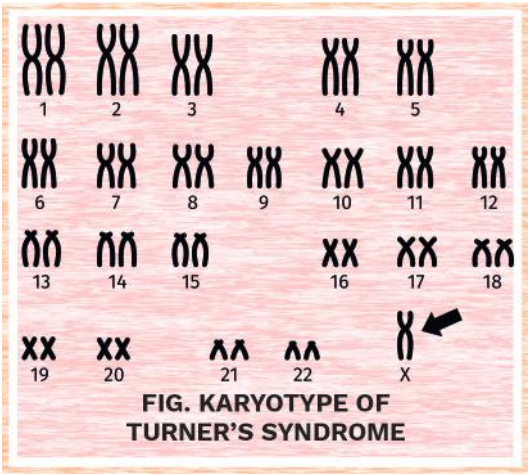


FIG. KARYOTYPE OF TURNER'S SYNDROME

Rack Your Brain

Why an abnormal human baby with 'XXX' sex chromosomes is born?

Previous Year's Question

Mr. Kapoor has Bb autosomal gene pair and d allele sex-linked. What shall be proportion of Bd in sperms?
(1) Zero
(2) 1/2
(3) 1/4
(4) 1/8

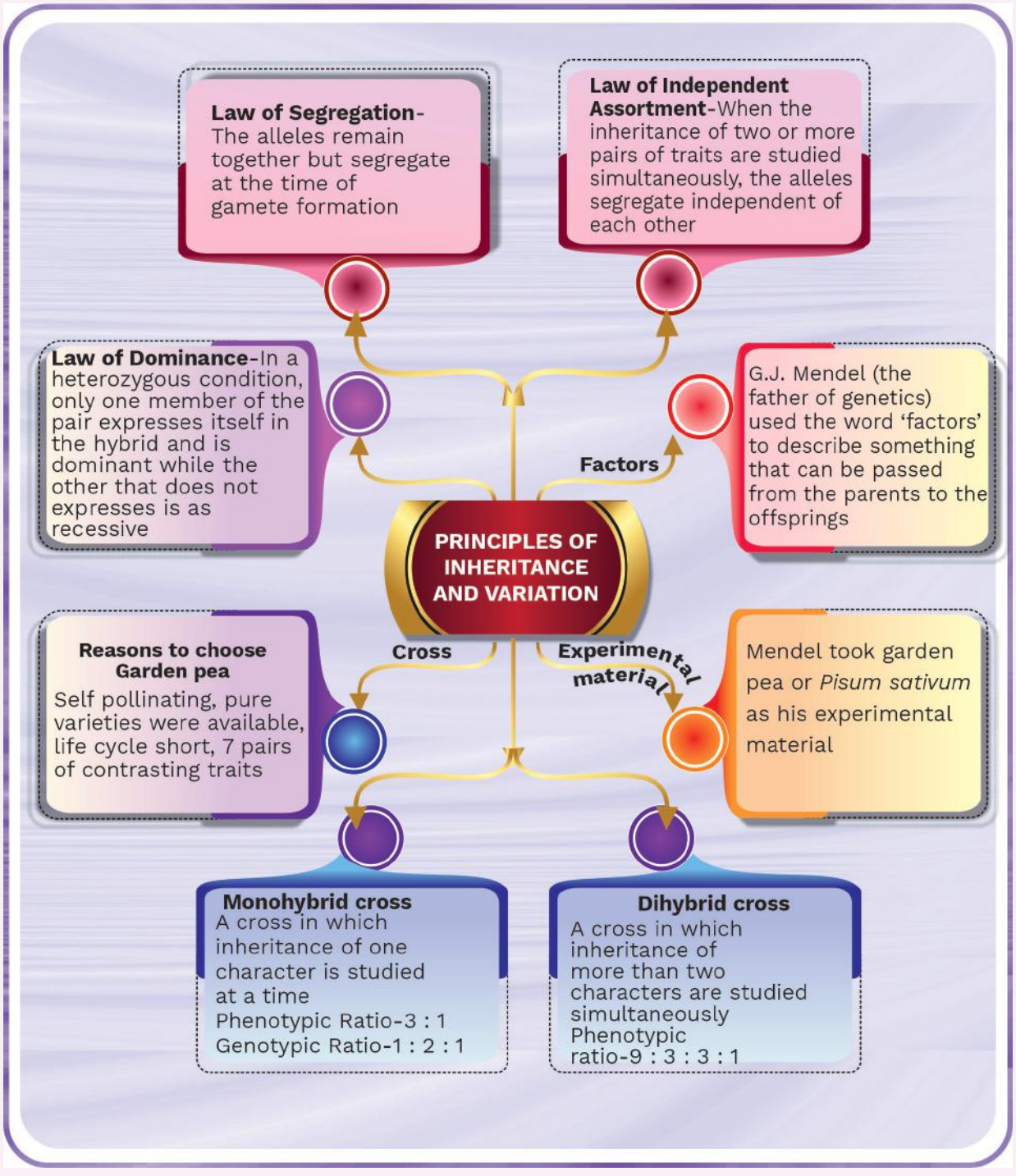
Rack Your Brain

Turner's syndrome is a sterile female, generally with normal intelligence and known viable monosomy in humans. Is the combination '44+YO' is also viable. Justify?

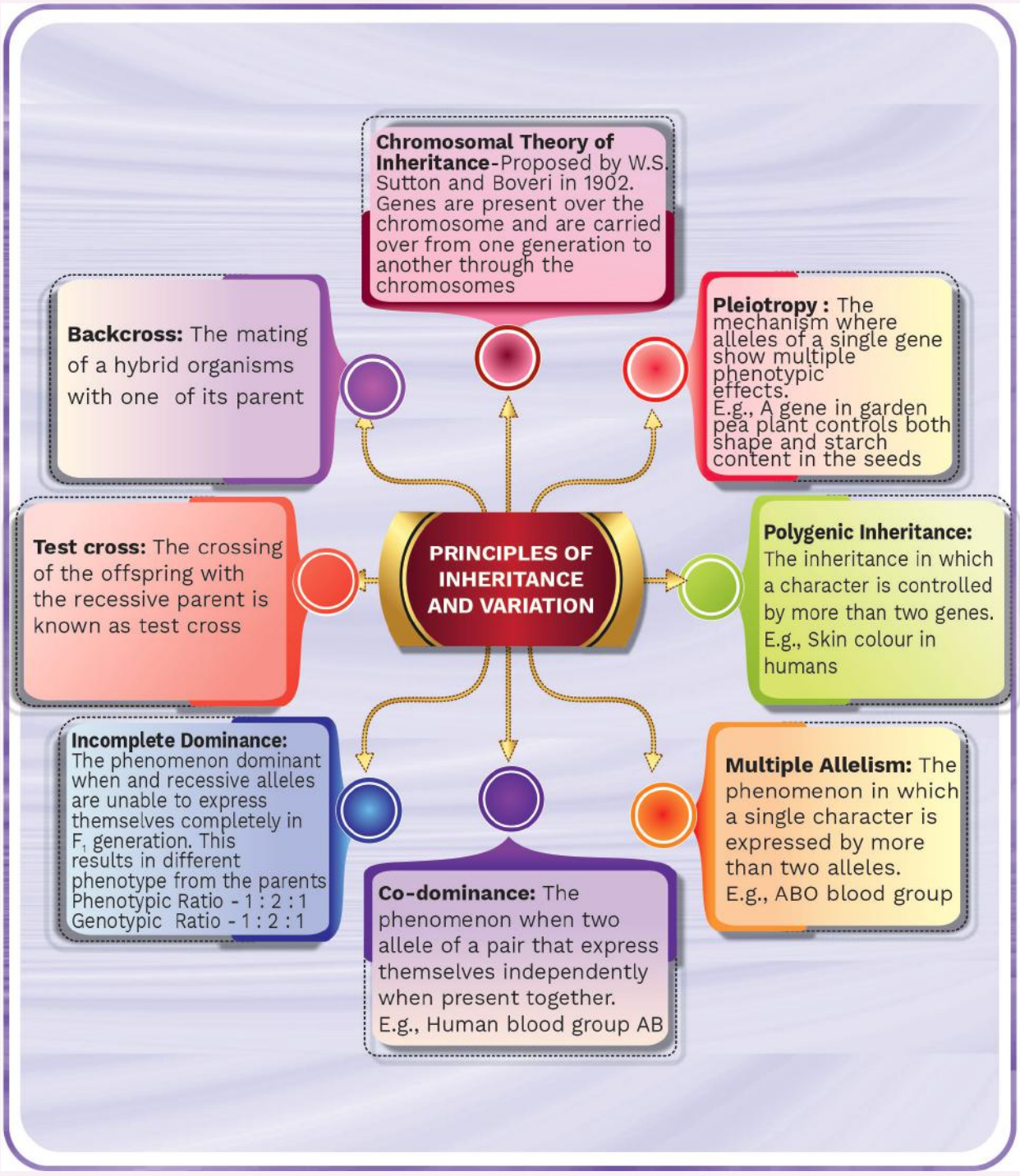
Gray Matter Alert!!!

Cri-du-chat Syndrome: It is an autosomal disorder in which the short arm of one of the 5th chromosome is deleted. The person suffering the disorder is mentally and physically impared. The crying of them is cat like and thus the name given to the disorder.

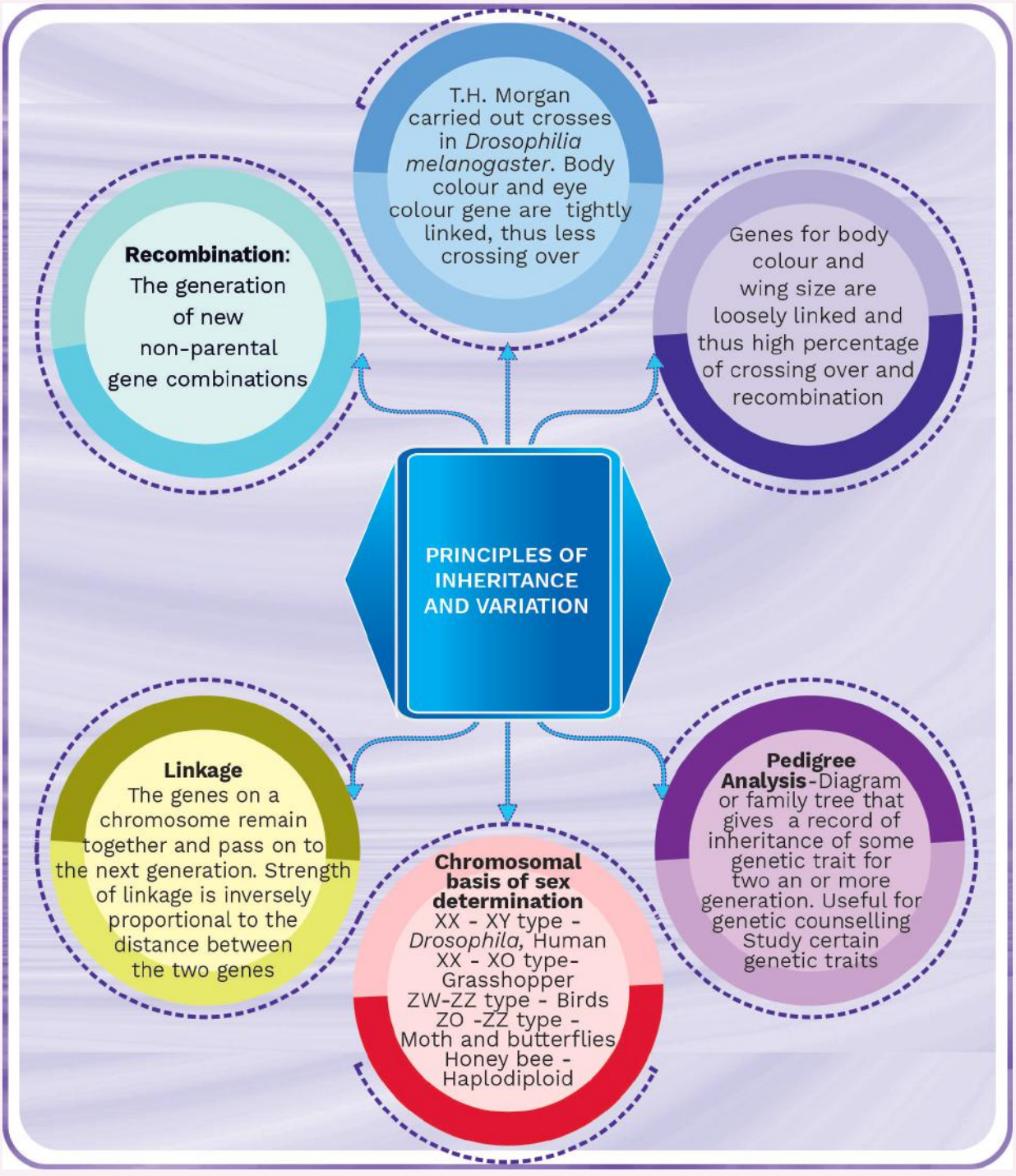
Summary


Law of Segregation- The alleles remain together but segregate at the time of gamete formation
Law of Independent Assortment-When the inheritance of two or more pairs of traits are studied simultaneously, the alleles segregate independent of each other
Law of Dominance-In a heterozygous condition, only one member of the pair expresses itself in the hybrid and is dominant while the other that does not expresses is as recessive
G.J. Mendel (the father of genetics) used the word 'factors' to describe something that can be passed from the parents to the offsprings
Factors
PRINCIPLES OF INHERITANCE AND VARIATION
Cross
Experimental material
Reasons to choose Garden pea
Self pollinating, pure varieties were available, life cycle short, 7 pairs of contrasting traits
Mendel took garden pea or Pisum sativum as his experimental material
Monohybrid cross
A cross in which inheritance of one character is studied at a time
Phenotypic Ratio-3 : 1
Genotypic Ratio-1 : 2 : 1
Dihybrid cross
A cross in which inheritance of more than two characters are studied simultaneously
Phenotypic ratio-9 : 3 : 3 : 1

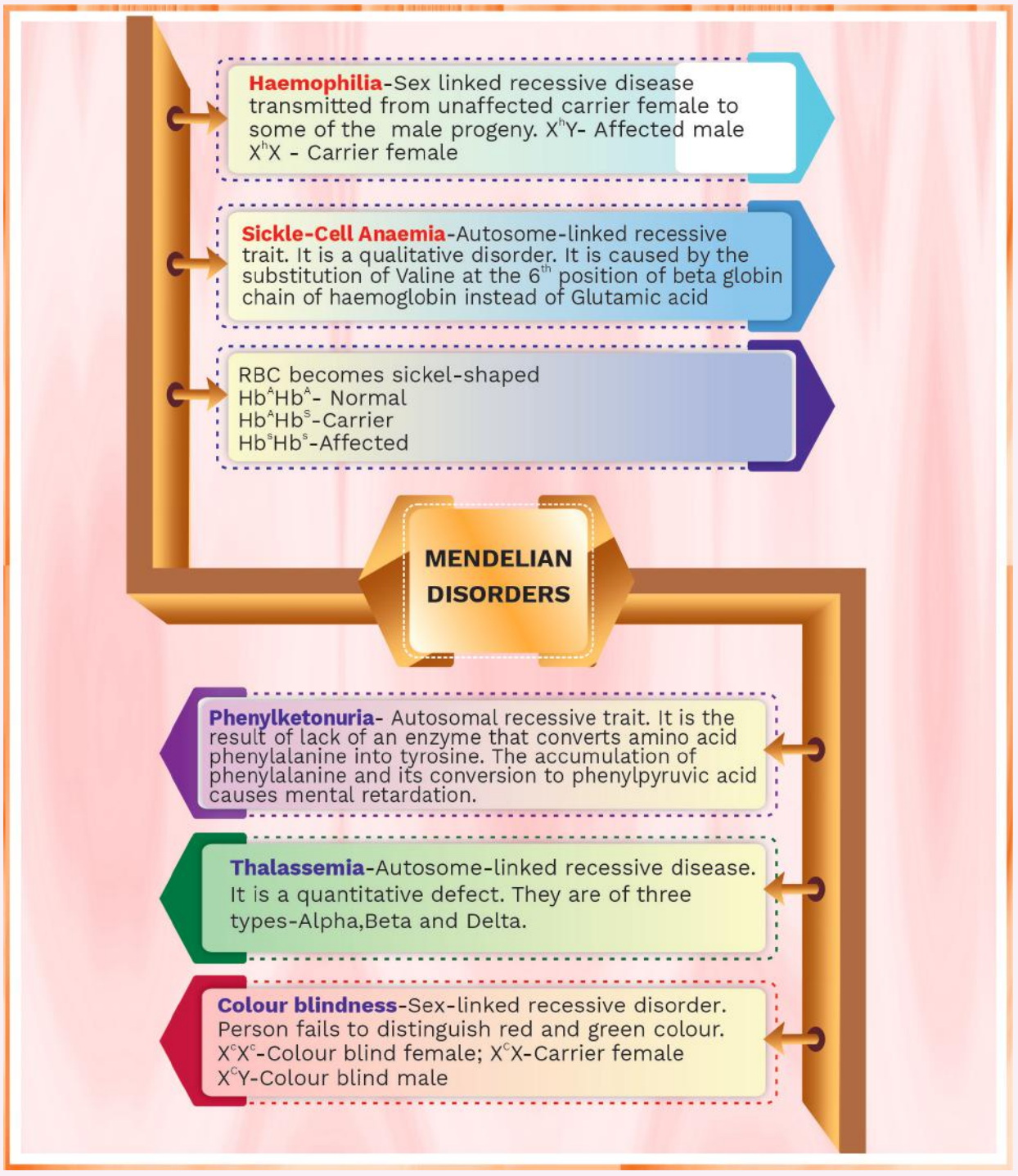
Summary


PRINCIPLES OF INHERITANCE AND VARIATION
Chromosomal Theory of Inheritance-Proposed by W.S. Sutton and Boveri in 1902. Genes are present over the chromosome and are carried over from one generation to another through the chromosomes
Backcross: The mating of a hybrid organisms with one of its parent
Pleiotropy : The mechanism where alleles of a single gene show multiple phenotypic effects. E.g., A gene in garden pea plant controls both shape and starch content in the seeds
Test cross: The crossing of the offspring with the recessive parent is known as test cross
Polygenic Inheritance: The inheritance in which a character is controlled by more than two genes. E.g., Skin colour in humans
Incomplete Dominance: The phenomenon dominant when and recessive alleles are unable to express themselves completely in F1 generation. This results in different phenotype from the parents Phenotypic Ratio - 1 : 2 : 1 Genotypic Ratio - 1 : 2 : 1
Co-dominance: The phenomenon when two allele of a pair that express themselves independently when present together. E.g., Human blood group AB
Multiple Allelism: The phenomenon in which a single character is expressed by more than two alleles. E.g., ABO blood group

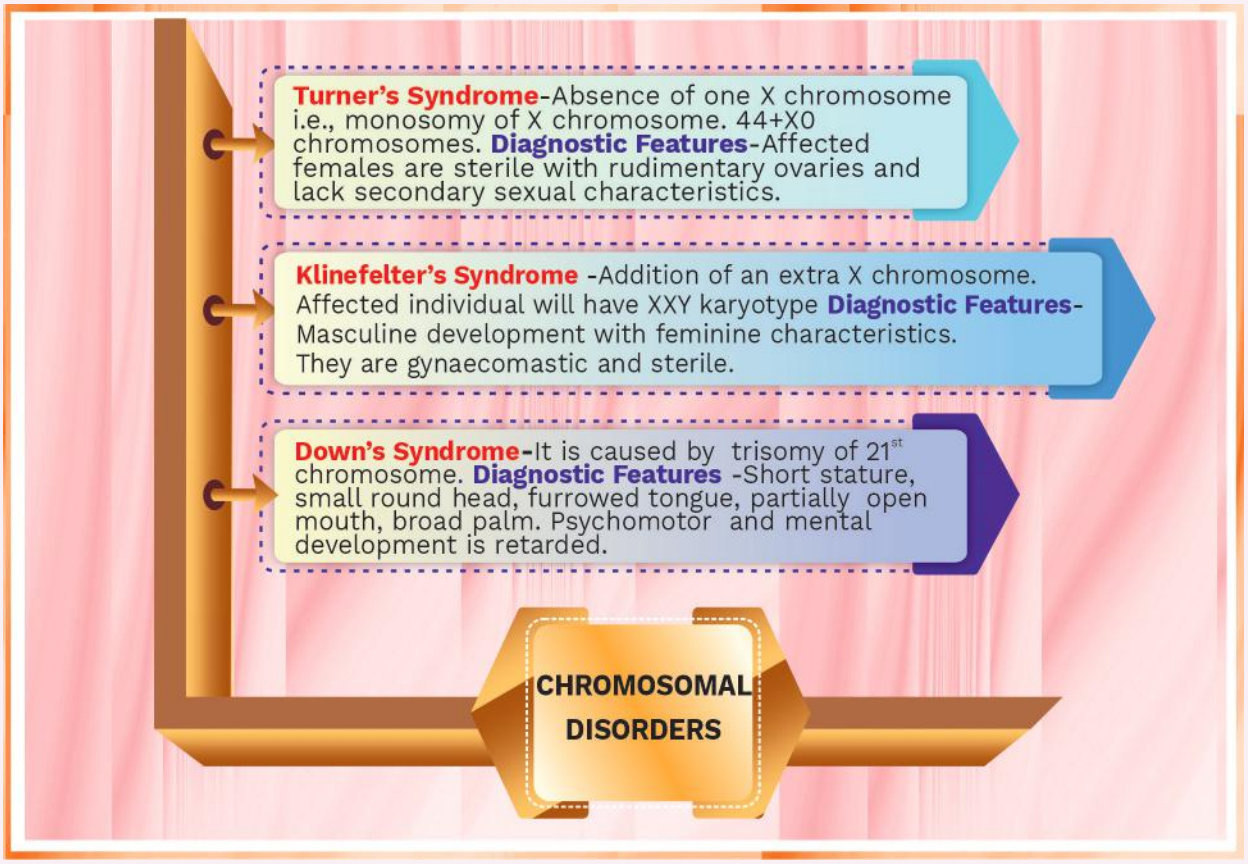
Summary


PRINCIPLES OF INHERITANCE AND VARIATION
T.H. Morgan carried out crosses in *Drosophilia melanogaster*. Body colour and eye colour gene are tightly linked, thus less crossing over
Recombination: The generation of new non-parental gene combinations
Genes for body colour and wing size are loosely linked and thus high percentage of crossing over and recombination
Linkage The genes on a chromosome remain together and pass on to the next generation. Strength of linkage is inversely proportional to the distance between the two genes
Chromosomal basis of sex determination XX - XY type - *Drosophila*, Human XX - XO type- Grasshopper ZW-ZZ type - Birds ZO -ZZ type - Moth and butterflies Honey bee - Haplodiploid
Pedigree Analysis-Diagram or family tree that gives a record of inheritance of some genetic trait for two an or more generation. Useful for genetic counselling Study certain genetic traits

Summary


MENDELIAN DISORDERS
Haemophilia-Sex linked recessive disease transmitted from unaffected carrier female to some of the male progeny. X^hY- Affected male X^hX - Carrier female
Sickle-Cell Anaemia-Autosome-linked recessive trait. It is a qualitative disorder. It is caused by the substitution of Valine at the 6th position of beta globin chain of haemoglobin instead of Glutamic acid
RBC becomes sickel-shaped
Hb^AHb^A- Normal
Hb^AHb^S-Carrier
Hb^SHb^S-Affected
Phenylketonuria- Autosomal recessive trait. It is the result of lack of an enzyme that converts amino acid phenylalanine into tyrosine. The accumulation of phenylalanine and its conversion to phenylpyruvic acid causes mental retardation.
Thalassemia-Autosome-linked recessive disease. It is a quantitative defect. They are of three types-Alpha,Beta and Delta.
Colour blindness-Sex-linked recessive disorder. Person fails to distinguish red and green colour.
X^cX^c-Colour blind female; X^cX-Carrier female
X^cY-Colour blind male

Summary


Turner's Syndrome-Absence of one X chromosome i.e., monosomy of X chromosome. 44+X0 chromosomes. Diagnostic Features-Affected females are sterile with rudimentary ovaries and lack secondary sexual characteristics.
Klinefelter's Syndrome -Addition of an extra X chromosome. Affected individual will have XXY karyotype Diagnostic Features- Masculine development with feminine characteristics. They are gynaecomastic and sterile.
Down's Syndrome-It is caused by trisomy of 21st chromosome. Diagnostic Features -Short stature, small round head, furrowed tongue, partially open mouth, broad palm. Psychomotor and mental development is retarded.
CHROMOSOMAL DISORDERS

Solved Exercise

1. **A man of A blood group marries a woman of AB blood group. Which type of progeny would indicate that man is heterozygous A?**
(1) AB
(2) A
(3) O
(4) B

Sol. (4)
Blood group B in the progeny will indicate that man was heterozygous.

2. **Multiple alleles control inheritance of**
(1) phenylketonuria
(2) colour blindness
(3) sickle cell anaemia
(4) blood groups

Sol. (4)
IA,IB and I control the blood group.

3. **The contrasting pairs of factors in Mendelian crosses are called**
(1) multiple alleles
(2) allelomorphs
(3) alloloci
(4) paramorphs

Sol. (2)
Contrasting pairs of factors of a character are known as allelomorph.

4. **First geneticist/father of genetics was**
(1) De Vries
(2) Mendel
(3) Darwin
(4) Morgan

Sol. (2)
G.J Mendel is known as the father of genetics.

5. **Mendel's last law is**
(1) segregation
(2) dominance
(3) independent assortment
(4) polygenic inheritance

Sol. (1)
Law of segregation states that alleles remain together but separate at the time of gamete formation.

6. **Blue eye colour is recessive to brown eye colour. A brown eyed man whose mother was blue eyed marries a blue-eyed women. The children will be**
(1) both blue eyed and brown eyed 1 : 1
(2) all brown eyed
(3) all blue eyed
(4) blue eyed and brown eyed 3 : 1

Sol. (1)
This is possible as the man and his mother were heterozygous for the eye colour.

7. **The allele which is unable to express its effect in the presence of another is called**
(1) codominant
(2) supplementary
(3) complementary
(4) recessive

Sol. (4)
Recessive alleles can only express themselves when both the alleles are the same type.

8. **A colour blind girl is rare because she will be born only when**
(1) her mother and maternal grand father were colour blind
(2) her father and maternal grand father were colour blind
(3) her mother is colour blind and father has normal vision
(4) parents have normal vision but grand parents were colour blind

Sol. (2)
It explanation only possible if this is the scenario.

9. **Cross between AaBB and aaBB will form**
(1) 1 AaBB : 1 aaBB
(2) all AaBB
(3) 3 AaBB : 1 aaBB
(4) 1 AaBB : 3 aaBB

Sol. (1)
The gametes formed will be AB, aB and aB. When they fuse together they will give the mentioned result.

10. **tt mates with Tt. What will be the characteristic of offspring?**
(1) 75% recessive
(2) 50% recessive
(3) 25% recessive
(4) All dominant

Sol. (2)
50% will be recessive.