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Genetics

This is a branch of biology that tries to explain the cause of similarities and difference between parents and their off springs. The first quantitative experiments on heredity of any significance were carried out in the middle of the nineteenth century by Gregor Mendel on the garden peas

Specific objectives

The learner should be able to

- Explain the concept of inheritance
- Define genetics terms
- Explain inheritance of trains using the monohybrid and dihybrid crosses.
- Discuss the challenges of inheritable disorders
- Explain gene interaction, sex linkage, sex determination, sex limitation, lethal genes and polygenes.

Terminologies.

1. Agene

This is the basic unit of hereditary and occupies a discrete position on the chromosomes. The gene controls the production of enzyme which in turn determines the process that goes on in a cell and eventually in the organ and the entire organism. In sexually reproducing organism, genes occur in pairs, where each member of a pair is contributed by the female and male parents.

2. Alleles (Allelomorphs)

This is one of the pair of a gene that occupy the same locus (position). Alleles are genes that are responsible for the production of contrasting characteristics such as tallness and shortness in plants and animals.

3. Genotype

This is the genetical constitution of an organism i.e. the particular set alleles, leading to observable characteristics

4. Phenotype

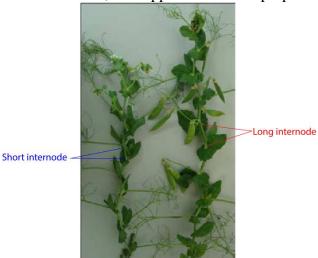
This is the physical characteristic of an organism determined by the genotype and the environment.

Monohybrid inheritance

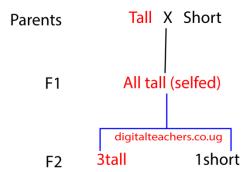
This an inheritance that deals with a single pair of contrasting characteristics such Tallness and shortness when concerned with height of peas.

In his investigation of a single pair of contrasting characteristics, Mendel observed that, in the first filial (F1) generation one of the characteristic never appeared only to appear in the second filial (F2) generation in small proportion compared to the one that appeared in the first filial (F1) generation.

For example; he crossed peas with long internode, with peas of short internode. He observed that in the F1 generation, all plants had tall internodes. When selfted to produce F2, the peas with short internodes, then appeared in small proportions.



The result of these crosses can be illustrated as follows,



It was concluded from the results that inheritance is a process in which discrete structure or particle (genes) which may or may not show themselves in the outward appearance of the organism are transmitted from parent to off spring.

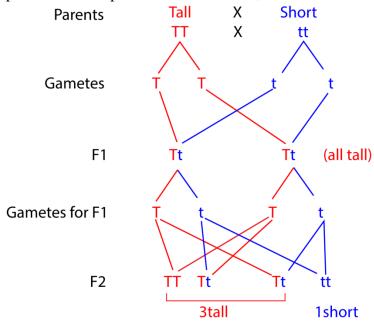
The evidence of existence of inheritable particles is got from the observation that they can be combined in one generation but separate in the next, as in witness by the recovery of the short form in F2 generation despite its absence in the F1 generation,

The characteristic that show in F1 generation (tallness in this case) is described as being **dominant** and while that which masked F1 generation (shortness) is said to be **recessive**.

Genes and their transmission

Gene normally occur in pair each of which is obtained from each parent. The cross of a tall plant and short plant is shown diagrammatically by the two methods below.

The allele for tallness (dominant) character is represented by a capital letter **T** while the allele for shortness (recessive character) is represented by small letter **t**. It is assumed that each parent plant contains a pair of identical alleles; **TT** in case of tall plant and **tt** in case of short plant.



Pannet square to show fusion of F1 gametes

	½ T	¹⁄2 t
½ T	1/4 TT	¹⁄4 Tt
½ t	½ Tt	½ tt

In terms of probability, there are 3 chances out of four for a tall plant to appear in F2 generation; and one chance of four for a short plant to appear.

Mendel's first law of segregation states that an organism's characteristics are controlled by two genes (alleles) and only one can be carried by in a gamete.

Mendel's second law of Independent Assortment:

During the formation of gametes, alleles in a pair may combine with another allele from another pair randomly.

Breeding True

Phenotypically TT and Tt are the same i.e. Tall. When an organism contains identical alleles like TT and tt is said to be **homozygous** and with dissimilar allele is **heterozygous**.

Since the homozygous (TT) and heterozygous (Tt) peas are both tall there is no way we can distinguish between the two genotypes from their external appearance.

One way of establishing whether a given tall plant is homozygous or heterozygous is to self-pollinate it. If the resultant off springs are all tall, we can conclude that the parent has the genotype TT.

If, however, we get a mixture of Tall and short plants; the parent plant must have the genotype Tt

The point is that when an organism which is homozygous at a particular locus is self- fertilized it produce off spring all of which are identical with parent. Exactly the same results occur if organismis crossed with another organism that is homozygous is said to breed true, The organism is said to belong to a pure line for the characteristics in question.

Test crosses

This is the crossing of an individual having **homozygous recessive** genotype with an individual showing a dormant trait to determine whether that individual is homozygous or heterozygous for the trait.

The homozygous individual produces all offspring having dominant trait while a heterozygous individual produces a mixture of offspring with dominant and recessive traits.

Back cross

This is a cross between hybrids in F1 generation with one of the parents or an organism genetically equivalent to the parents. Back crossing is mainly aimed at increasing the genetic contribution of one particular parent to the off spring.

Monohybrid inheritance human

A number of human conditions are known to be associated with a single pair of alleles which are inherited in Mendelian fashion.

1. Albinism

1. Albinism

This is a condition in human beings where the individual fail to produce skin pigments called melanin.

Albinos have;

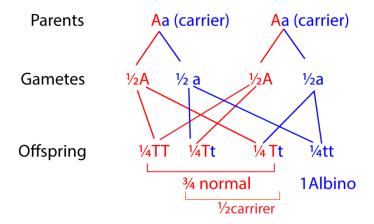
- ✓ Light skin
- ✓ White hair
- ✓ Pink eyes
- ✓ They are sensitive to bright light



Albino

The allele for albinisms is caused by recessive [a] and so only exert its effect in the homozygous state [aa]. The allele for melanin production [A] is dominant.

Suppose a couple each with normal pigmentation have an albino child. For this to happen the child must have [aa]. Therefore, unless for rare mutation, the parents must both heterozygous [Aa] so each produces A and a gametes in about equal number. Therefore, randomly to produce three type of genotype AA, Aa, aa.



- 2. Congenital disease such as cystic fibrosis in which the connective tissue develops in glands of the body.
- 3. Chondrodystrophic dwarfs are characterized by shortened and deformed legs and arms. It is cause by a dominant gene and hence affect in homozygous and heterozygous state

CO-DOMINANCE

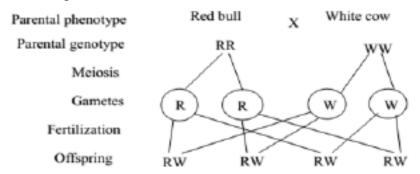
This is a condition where genes determining a particular character all show up such that the phenotype of the offspring is a mixture of that of the parents. *It mainly occurs in animals*. Co-dominance is where in the heterozygous state neither allele is completely dominant over the other i.e. the 2 alleles are co-dominant. This results in the phenotype intermediate between the parent's appearances. The alleles for each trait are represented with different capital letters.

Questions:

1) In animals, the genes for fur colour are co-dominant. What will be the offsprings when a red bull is crossed with a white cow?

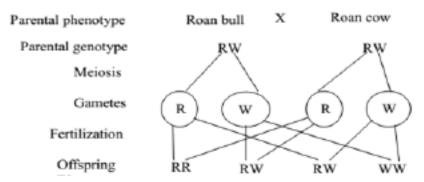
Solution:

Let R represent the allele for red bull Let W represent the allele for white cow



F1 generation are all roan

2) What would be the off springs in the 2nd generation?



F2 generation 1red, 2 roan, 1 white

INCOMPLETE DOMINANCE

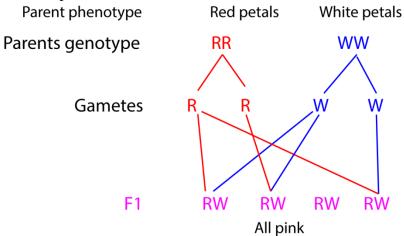
This is a condition in the heterozygous where neither of the alleles is dominant over the other and the phenotype of the offspring is an intermediate between that of the parents. *It mainly occurs in plants*.

For example, consider petal colour in flowers: when a red flowered plant is crossed with a white flowered plant, the offspring produced are all pink coloured petal flowers.

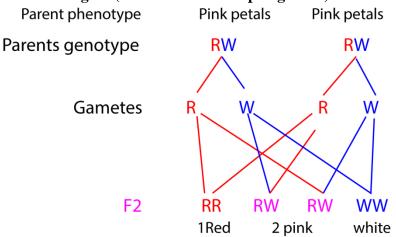
Example:

Let R represent the allele for red petal colour.

Let W represent the allele for white flowers



Then Selfing F1 (Cross between offspring in F1)



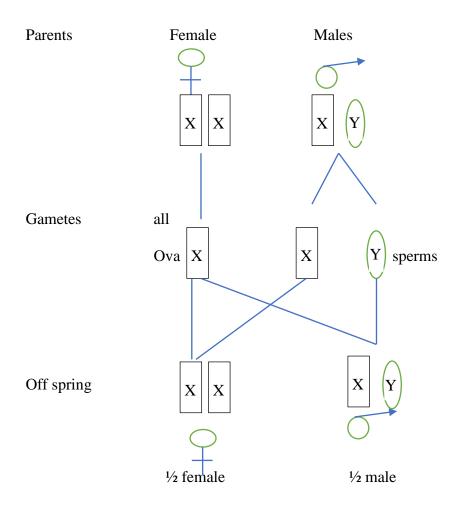
F2 Phenotypic ratio: 1 red: 2 pink: 1 white. (1:2:1)

Sex determination.

The medium size chromosome in drosophila melanogaster determine the individual's sex for which reason they are called the **sex chromosome**. In the female the two sex-chromosome, both rod shaped in appearance, are identical and are known as **X-chromosomes**. In the male, however the two sex-chromosome differ from each other one is rod-shaped X-chromosome, the other is hook-shaped and is called **Y-chromosome**.

The sex chromosomes are exception to the rule that homologous chromosomes are identical in appearance. Being different they are described as **heterosomes**; All the other chromosomes, which are identical in appearance, are called **autosomes**.

Despite this difference, the sex chromosomes are transmitted in a normal mendelian manner as shown below.



Generally, a female produces only one kind of gamete as the chromosomes are concerned; all her eggs contain an X chromosome. For this reason, in human and many other species, the female is said to be homogametic (same gametes). A male on the other produces two kinds of gametes as far as chromosome are concerned: half of the sperm contain an Y chromosome, the other half a Y. The male is therefore heterogametic (different gametes) on fusing randomly, approximately

half the zygotes receive two X chromosome and develop into female, the rest receive Y chromosomes and give rise to males. In some insects, females are XX and male XO

Sex linkage

Sex linked characters are those whose genes that are carried on sex chromosomes usually X-chromosomes and is inherited along with sex for examples:

- (i) red-green color blindness the decreased ability to see color or differences in color. Simple tasks such as selecting ripe fruit, choosing clothing, and reading traffic lights can be more challenging. People with total color blindness (achromatopsia) may also have decreased visual acuity and be uncomfortable in bright environments
- (ii) hemophilia

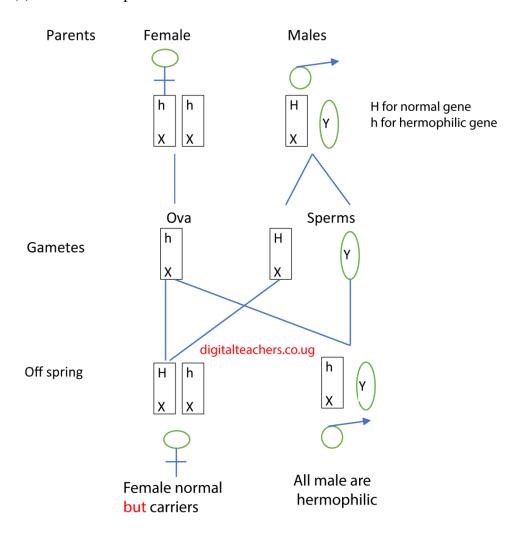
Symptoms of hemophilia are

- Unexplained and excessive bleeding from cuts or injuries, or after surgery or dental work
- Many large or deep bruises
- Unusual bleeding after vaccinations
- Pain, swelling or tightness in your joints
- Blood in your urine or stool
- (iii) eye color in drosophila

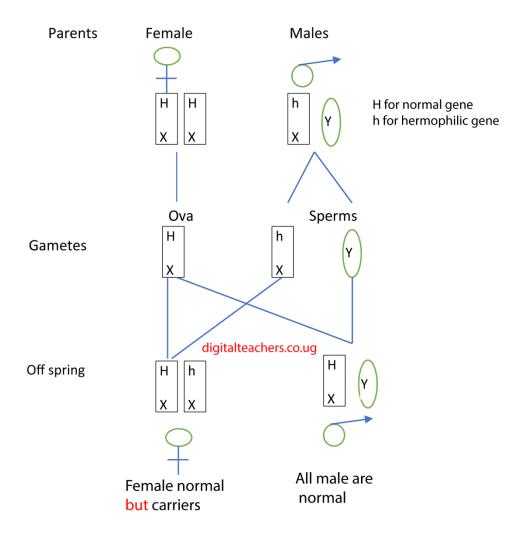
Transmission

Hemophilia can be represented as follow

(a) Female hemophilic and normal man



(b) Female normal X male hemophilic



THE Y-CHROMOSOME.

- 1.If a sex-linked trait is associated exclusively with Y-chromosome, it is expected to show up exclusively in males. In general, most Y-chromosome are empty. However, the 'porcupine' man is said to have transmitted hard and spine skin exclusively to his male children.
- 2. The hair pinna or ear is a characteristic common in India and it is transmitted to male off spring only.

Sex linked are more common in male than female because

- (i) Males have only 1 X chromosome, from their mother. If that X chromosome has the gene for red-green color blindness (instead of a normal X chromosome), they will have red-green color blindness.
- (ii) Females have 2 X chromosomes, one from their mother and one from their father. To have red-green color blindness, **both** X chromosomes would need to have the gene for red-green color blindness

Sex limited characters

These are characters that that show up exclusively in one sex only e.g. ovary in female

Note: A sex – limited character is one which is controlled by a gene located on any chromosome but expresses itself in only one of the two sexes.

Multiple alleles

Multiple alleles are two or more alternative forms of a gene controlling a particular characteristic, of which any two may occupy the same gene loci on homologous chromosomes.

An example of such multiple allele is provided by the alleles controlling the ABO blood group system in humans. The ABO system is controlled by three alleles generally referred to as I^A , I^B , and I^O .

The I^A allele is responsible for production of type A antigens in the person's red blood cells, and the I^B allele for type B antigen. The I⁰ produces neither antigen.

But only two of the three alleles can be present at any one time, an individual may thus, possess any of the following six genotype; I^AI^A , I^AI^O , I^BI^B , I^BI^O , I^AI^B , and I^OI^O .

I^A and I^B show equal dominance with respect to one another [i.e. they are codominant] but each is dominant to I^O thus:

- A person belongs to blood group A has genotype I^AI^A or I^AI^0

- A person belongs to blood group B has genotype I^BI^B or I^BI^O

- A person belong to blood group AB has genotype IAIB

- A person belongs to blood group O has genotype I^OI^O

The fact that there more than two alleles responsible for determining the blood group makes no difference to their transmission, which take place in a normal mendelian fashion.

Thus, a child whose parents are both blood group O must be group O. However, a parent with blood A or B, the child may have any of the blood group has shown below.

Parents' phenotypes blood group A x blood group B IAIO BO Parentas' genotypes X digitalteachers.co.ug Gametes IAIO IBIO lolo Off springs genotype IAIB **Blood** groups AB A В 0

Blood transfusion

This the giving of blood to a patient who has lost a lot of blood due to accident or who lacks lack enough blood due to other causes. The person who gives blood is called a donor while the person who receives blood is recipient.

A doctor needs to careful to match blood of the donor and recipient, because when some blood is mixed, red blood cell stick together of **agglutinate**, which may be fetal. The blood that do not agglutinate when mixed are said to be **compatible**.

Agglutination is caused by presence or absence of two antigen called A and B in red blood cell, and antibodies, called anti-A and anti-B in the sera.

Everybody can be placed in one of the four groups according to the antigen and antibodies their blood contains shown in the table below

Person's blood group	Antigen on the red blood cells	Antibodies in the sera
A	A	Anti-B
В	В	Anti-A
AB	A +B	None
0	none	Anti-A + Anti-B

The shows compatible transfusion

Recipient blood group	Donor blood group
Group A	Group A or O
Group B	Group B or O
Group AB	Group A, B, AB or O
Group O	Group O

Variation, in biology

This is any difference between cells, individual organisms, or groups of organisms of any species caused either by genetic differences (genotypic **variation**) or by the effect of environmental factors on the expression of the genetic potentials (phenotypic **variation**).

Importance of variation

Genetic **variation** is an **important** force **in** evolution as it allows natural selection to increase or decrease frequency of alleles already **in the** population. ...

The higher the genetic variations, the higher the chances of survival of a population in a changing environment.

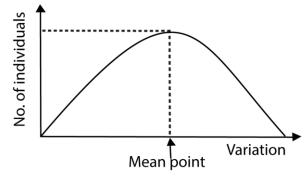
Categories of variation

There are two types of genetic variations.

(i) Continuous variations.

These are variations that show a gradual change in individuals without a clear-cut division between the two extremes. It results into formation of intermediates. Such variations include height, intelligence, skin colour, yield in plants, etc. In such variations, organisms are usually very many around the mean/average point.

Graphic illustration of continuous variation



(ii) Discontinuous variation.

This is a variation, which shows a clear-cut difference between the two extremes without intermediates. This results into expression of only two phenotypes. Examples of discontinuous variations include, tongue rolling, blood groups, sex, etc.

Causes of variation

Some variations are inherited and are called inherited variations while others are occupied as a result of the environment hence called environmental variations.

Examples of inherited variations are blood groups, eye colour, albinism, hair, etc.

Examples of environmental variations are knowledge, etc.

Environmental factors that cause variations

- ✓ Diet
- ✓ Pathogens
- ✓ Altitude
- ✓ Light

Factors that cause inherited variations

✓ Mutation

A **Mutation** occurs when a DNA gene is damaged or changed in such a way as to alter the genetic message carried by that gene.

A Mutagen is an agent of substance that can bring about a permanent alteration to the physical composition of a DNA gene such that the genetic message is changed.

- ✓ Crossing over
- ✓ Fertilization

Exe

D. 20

erc	ise						
1.	A mother of twins has blood group A. one of the sons has blood group AB while the other has blood broup O. which one of the following is the blood group of the father?						
	A. Group A						
	B. Group B						
	C. Group O						
	D. Group AB						
2.	In plants, tallness is dominant to shortness. When two heterozygous parents from F2						
	generation were crossed, 2508 offspring were produced. What was the approximate						
	number of tall plants among the off springs?						
	A. 627						
	B. 826						
	C. 1254						
•	D. 1881						
3.	A person of blood group A can safely receive blood from a person of blood group						
	A. B and AB						
	B. A and O						
	C. A only						
1	D. AB only When a red flowered plant is crossed with a white flowered plant, the offenring are pink						
4.	When a red flowered plant is crossed with a white flowered plant, the offspring are pink flowered plants. What would be the proportion of the red flowered plants among the						
	offspring if two pink flowered plants were crossed? A. ½ B. 1/3 C. ¼ D. 2/3						
5							
٥.	In plants, tallness is dominant over shortness. What will be the proportion of short plants among the offspring if two heterozygous tall plants were crossed?						
	A. 1 B. ½ C. 1/3 D. ¼						
6.	Which one of the following reactions is likely to occur when a person of blood group A						
	donates blood to a person of blood group B?						
	A. Antibodies a would react with antigen A						
	B. Antibody b would react with antigen B						
	C. Antibody a would react with antigen B						
	D. Antibodies b would react with antigen A and B						
7.	1						
	plants with only pink flowers. If the pink flowered plants were selfed and produced 60						
	plants, how many of them would be expected to bear pink flowers						
	A. 30						
	B. 10						
	C. 40						

- 8. What percentage of offspring will have blood group O if a man heterozygous for blood group B marries a woman heterozygous for blood group A?
 - A. 100%
 - B. 75%
 - C. 50%
 - D. 25%
- 9. A couple produced children with blood groups A, B, AB and O. Which of the following were the genotypes of the parents?
 - A. BB and BO
 - B. AO and BO
 - C. BO and AA
 - D. AB and OO
- 10. Which of the following are likely blood groups of children from parents of blood group A and B?
 - A. O only
 - B. AB only
 - C. A and AB only
 - D. O, A, B and AB
- 11. A gene which does not express itself unless in homozygous state is
 - A. Dominant
 - B. Sex linked
 - C. Codominant
 - D. Recessive
- 12. In peas, yellow seed color is dominant over green seed colour. What would be the phenotype of offspring if true breeding yellow-seeded plant is crossed with a green-seeded plant?
 - A. 2 yellow: 2 green
 - B. 2 yellow:1greens
 - C. 3 green: 1 yellow
 - D. All yellow
- 13. An albino offspring is likely to result from a cross between
 - A. An albino and normal mother
 - B. Both parents who are carrier for albinism
 - C. A carrier mother for albinism and normal father
 - D. Both parents who are normal
- 14. In human, the gene for brown eyes is dominant to gene for blue eyes. If heterozygous brown eyed male married a blue eyed female, what is the probability of producing blue eyed offspring?
 - A. 0%
 - B. 25%
 - C. 50%
 - D. 75%

- 15. A man of blood group AB marries a woman of blood group A. what would be the possible blood groups of the children?
 - A. A and AB
 - B. A only
 - C. AB
 - D. A and B
- 16. A pure breeding red-flowered plant was crossed with a pure breeding white flowered plant and all the resulting F1 generation had pink flowers. What percentage of F2 plants would have red flowers if F1 plants were self-pollinated?
 - A. 100%
 - B. 50%
 - C. 33%
 - D. 25%
- 17. When homozygous red flowered plants were crossed with homozygous white flowered plants, all plants produced had pink flowers. What would be the phenotypic ratio of plants resulting from a cross of pink-flowered plants?
 - A. 3 red-flowered: 1 white flowered
 - B. 1 red flowered: 3 shite flowered
 - C. 1red flowered: 2 pink flowered plant: 1 white-flowered
 - D. 2 red flowered: 1 pink flowered: 1 white flowered
- 18. Which of the following is true about a person of blood group O?
 - A. Receives blood from people of all blood groups
 - B. Donates blood people of all other groups.
 - C. Receives blood from only people of blood groups AB and O
 - D. Donates blood to only people with blood group AB.
- 19. In cattle, when a white bull is mated with a red cow, the offspring is roan. This indicates that the gene for white is
 - A. Dominant to that for red
 - B. Recessive to that for red
 - C. Codominant with that for red
 - D. Mutated to show roan
- 20. What would be the ratio of phenotypes if a roan bull and a roan cow from the offspring referred to in question 19 were mated
 - A. 1 red: 2roan: 1 white
 - B. 2 red: 1 roan: 1 white
 - C. 1 red: 1 roan: 2 white
 - D. 1 red: 1 white
- 21. A man of blood group A marries a woman of blood group B and produced a child of blood group AB. Which of the following is true about this family?
 - A. The mother could donate blood to the father but not to the child
 - B. The child could donate blood to the mother and the father
 - C. The child could receive blood from mother and father.
 - D. No blood transfusion between the members of the family are possible

- 22. Which one of the following is likely to occur when a donor of blood group A gives blood to a recipient of blood group B?
 - A. Antibody A reacts with antigen B
 - B. Antigen B reacts with antibodies b
 - C. Antibodies b reacts with antigen A
 - D. Antigen A reacts with antigen B
- 23. When homozygous black mouse (BB) was crosses with homozygous white mouse (WW), the offspring were all brown. What would be the color of the mice produced if F2 offspring is mated heterozygous white parents?
 - A. 2brown: 1 white
 - B. 1 brown: 3 white
 - C. 1 brown: 1 white
 - D. All white
- 24. If a man of blood group A marries a woman of blood group O, what is the possible genotype of their children
 - A. AA, OO
 - B. AA, AO
 - C. AO, OO
 - D. AO only
- 25. The serum of a universal donor contains
 - A. Antigen A
 - B. Antigen B
 - C. Neither antigen A nor B
 - D. Both antigen A and B
- 26. Heterozygous flowered plant (Rr) is crossed with white flowered plant (rr). If R is dominant over r, what will be the phenotype of the offspring?
 - A. All red
 - B. All white
 - C. Pink and white
 - D. Red and white
- 27. In cattle a gene for red color coat R is codominant with a gene for white color coat W. If a red cow was mated with white bull, what would be the phenotype of F1 generation?
 - A. All red
 - B. All white
 - C. 3red: 1white
 - D. Intermediate coat color (roan)

Structured questions

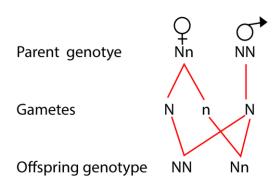
- 28. (a) What is mutation?
 - (b) The gene for production of normal hemoglobin is dominant to the mutant gene that causes sickle cell anemia. If a female heterozygous to sickle cell anemia marries a normal man. Illustrate with suitable symbols, the possible phenotype and genotype.

- 29. In an experiment, a long winged male drosophila was crossed with a short winged female drosophila. All offspring in the F1 generation were long winged. When two members of F1 were mated, the F2 generation consisted of 62 long winged flies and 21 short winged flies.
 - (a) Suggest an explanation why all F1 generation flies were long winged.
 - (b) (i) what type of flies would develop from a mating between short winged flies in the second generation
 - (ii) Give a reason for your answer.
 - (c) Mating between a short winged fly in F2 with a long winged fly in F1generation produced 90 flies. How many were long winged? Show your working.
- 30. (a)(i) Which chromosomes are responsible for determining sex in human?
 - (ii) using appropriate symbols show how sex is determined in human.
 - (b) Red-green color blind is a defect caused by a recessive gen carried on the X-chromosome. What would be the phenotype of offspring when normal woman marries a color blind man. Show your working.
- 31. (a) what is meant by genotype
 - (b) A mon of blood group A married a woman homozygous for blood group B and they produced a son of blood group B.
 - (i) work out the genotype of the father and of son (04marks)
 - (ii) the son married a wife of blood group O. Showing your working, give the percentage of their offspring. (03marks)
- (c) Blood groups in humans, show discontinuous variation. Explain what you understand by this statement. (02marks)
- 32. (a) Distinguish between a homozygous and heterozygous state in genetics (01mark)
 - (b) When a tall pea plant was crossed with a short pea plant, only tall plants were produced in F1 generation. When two plants from F1 generation were crossed, the resulting F2 generation had a mixture of tall and short plants.
 - (i) Using suitable symbols, show genetic crosses to produce F1 and F2 generations. (10marks)
 - (ii) If F1 generation had 81 plants determine how many would be short and tall. (02marks)
 - (c) Giving a reason state the phenotype which is recessive in pea plant. (02marks)
- 33. (a) Differentiate between continuous and discontinuous variation, giving an example, in human. (04marks)
 - (b) Distinguish between incomplete dominance and co-dominance in hereditary. (04marks)
 - (c) A man of blood group A married a woman of blood group B but they produced a child of blood group O among others. What were the genotype of parents and offspring? Show working) (07marks)
- 34. (a) distinguish between dominance and codominance in genetics (02marks)

- (b) When tall pea plants were crossed with short pea plant, all the plants in F1 were tall. When two plants of F1 generation were crossed, both tall and short plants were produced in F2 generation.
- (i) why were all plants tall in F1 generation (02mark)
- (ii) using suitable symbols show the crosses to produce F1 and F2 generation. (07 $\frac{1}{2}$ marks)
- (c) In rose plant, when a red flowered plant is crossed with white flowered plant, all plants produced bear pink flowers. Using suitable symbols show the result of crossing a pink flowered plant and a white flowered plant. (03 ½ marks)

1	В	6	A	11	D	16	D	21	С	26	D
2	D	7	A	12	D	17	С	22	В	27	A
3	В	8	D	13	В	18	В	23	С		
4	С	9	В	14	С	19	С	24	С		
5	D	10	D	15	A	20	A	25	С		

- 28. (a) A **Mutation** occurs when a DNA gene is damaged or changed in such a way as to alter the genetic message carried by that gene.
 - (b) Gene for normal hemoglobin = N Gene for sickle cell = n



Offspring phenotype: all normal

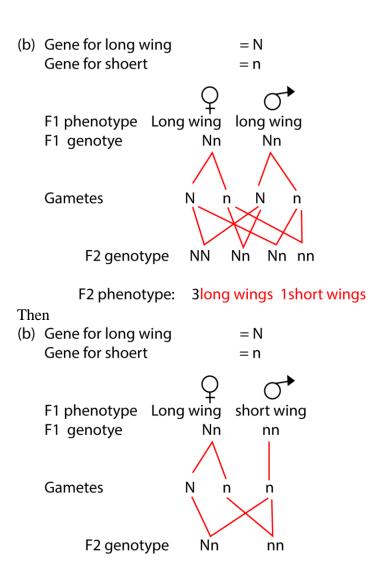
- 29. (a) A gene for long wings is dominant to a gene for short wings
 - (b) Gene for long wing = N
 Gene for shoert = n

 Parent phenotype Long wing short wing Parent genotye NN nn

 Gametes N n

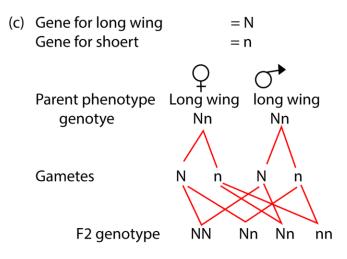
F1 phenotype: all long wings

Then



F2 phenotype: 1long wings 1short wings

All will have short wings when short winged flies are mated with short winged flies of F1 generation because short winged flies are homozygous. Or equal proportion of long winged and short winged flies if a long winged fly of F1 is used since F2 long winged flies are heterozygous.



F2 phenotype: 3long wings 1short wings Number of flies with long wings = $\frac{3 \times 90}{4}$ = 67

30. (a) (i) X- chromosome and Y- chromosome

