In its place, the dark-colour allele became more predominant, because dark moths could camouflage themselves better on the stained trees and avoid being eaten by their bird predators. The population evolved to a higher adaptive state with the change in gene frequencies (light–colour allele frequency went down while dark – colour allele frequency went up).

Questions

1. A village has 4% albino people. Find the % of people homozygous to normal skin and heterozygous to normal skin.
2. A disease is caused by a recessive allele. Dominant give normal health. In a population the recessive allele is 0.3. Find the homozygous dominant genotypic frequency.
3. 49% of a population is double recessive for a trait. Find the genotypic frequency of double dominant individuals.
4. Incapability of rolling of tongue is due to double recessive genotype. If 16% of the population is incapable of rolling the tongue then find the frequency of double dominant individuals. Find the genotypic frequency of heterozygous individuals.
5. If q = 0.6, find the heterozygous genotypic frequency.
6. q of a population is 0.2. What is the q valve of this population after 10 generations? Assume that this population follows Hardy-Weinberg equation.

MCQ

1. In sex linked inheritance of humans.
   (1) most or the X- linked recessive disorders are expressed in the heterozygous genotype of females.
   (2) most of the X- linked recessive disorders are expressed in males.
   (3) mothers transfer X- linked characters only to their daughters.
   (4) fathers transfer X- linked characters only to their sons.
   (5) males express only X- linked dominant disorders. (2022 AL/32)

2. Certain plants of a particular species bear purple flowers while other plants of the same species hear white flowers. To explain the inheritance of the flower colour of this plant Species
   (1) a monohybrid cross is sufficient. (2) a dihybrid cross is sufficient.
   (3) a monohybrid cross and a dihybrid cross are necessary.
   (4) knowledge of incomplete dominance is necessary.
   (5) knowledge of gene linkage is necessary. (2020 AL/31)

3. In man, sickle cell anaemia is an example for
   (1) heterozygous dominance. (2) polygenic inheritance. (3) epistasis. (4) pleiotropy. (5) epigenetics. (2019 AL/30)

4. Which of the following statements regarding the cross Rr x Rr is correct?
   (1) The probability of having the allele r in both the egg and sperm at fertilization is %.
   (2) This is a dihybrid cross because two alleles are involved.
   (3) According to Mendelian inheritance, the probability of having dominant phenotype in F2 generation by interbreeding of Fl is
   (4) If 1:2:1 ratio of phenotypes was obtained in F2 generation by interbreeding of F1 generation, it may be due to codominance.
   (5) R and r are linked. (2019 AL/31)

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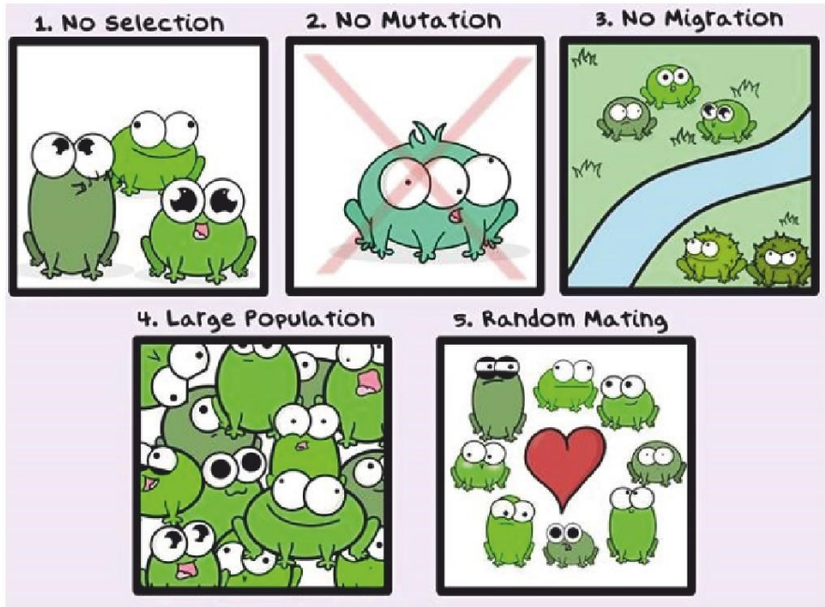
sampath lankadheera

Live Biology

Biology 3

NEW SYLLABUS

English Medium



UNIT 06

Genetics

Linkage/Polulation Genetics

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Naturally, most populations do deviate from Hardy-Weinberg equilibrium except for certain genetic loci. Slowly evolving populations may also do not deviate much from Hardy-Weinberg equilibrium and therefore, they remain as predicted for a non-evolving population.

Evolution and Change in Gene Frequency

Evolution can be explained in terms of changes in ……. (gene) ……... over generations. A species evolves when changes in gene frequencies drive the species into a higher level of adaptation for a specific ecological niche.

…….. …….. within the …….. is the key to evolution. …….. will create new …….. and migration will include it into the population to increase variation. …….. ……. will then choose the better adapted individuals based on their phenotypic variations, causing population to evolve.

As a result, after the evolution the population is at a higher adaptive level compared to the level of adaptation they showed before being evolved.

This concept can be illustrated with the evolution of the peppered moth in England during the time of industrialization.

The moth had two phenotypic varieties based on their colour; dark and light.

Prior to the industrialization of central England, the light-coloured allele was most prevalent.

The light-coloured moths had an advantage over the dark coloured ones as they could hide on the white-barked trees to avoid predation from birds.

Due to the pollutants generated parallel to the industrialization, the light-coloured trees were stained dark.

This exposed the light-coloured moths to predation, reducing their numbers. As a result, the light- colour allele became less prevalent.

If the egg provides the C^W allele and the sperm the C^R allele, the resulting C^RC^W heterozygotes in the progeny, q x P = 0.2 x 0.8 = 0.16 = 16%

Thus, the total frequency of heterozygote in the progeny

$$pq + qp = 2pq = 0.16 + 0.16 = 0.32, \text{ or } 32\%$$

In the above example, only three kinds of genotypes are possible.

When conditions suits for Hardy-Weinberg equilibrium, the sum of frequencies of all three genotypes equals 1.

Thus, the equation for Hardy-Weinberg equilibrium can be written as below;

$$\mathbf{p^2 + 2pq + q^2 = 1}$$

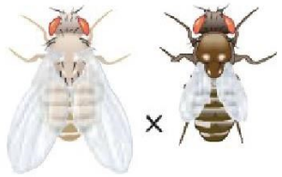
Conditions for Hardy-Weinberg Equilibrium

The Hardy-Weinberg approach describes a hypothetical population that is not evolving and fulfilling the following conditions.

1. ……....
   Mutations result changes in alleles. Insertion, deletion, or substitution of nucleotides result altered alleles. This leads to modified gene pool.
2. ……..
   Breeding occurs randomly without any influence that causes selectiveness. Mating of closely related individuals may alter the allele frequencies.
3. ……....
   All genotypes of the progeny are expected to survive irrespective to their differences, abilities and the environmental conditions. Variations in survival and reproduction of some genotypes may alter the frequency of alleles.
4. ……...
   In small populations, particular genotypes may disappear due to death or infertility. Therefore, larger the population, more likely to favor the Hardy-Weinberg equilibrium.
5. ……..
   Individuals moving in and out of the populations may cause appearance of new genes and disappearance of existing genes. This is called gene flow and it may alter allele frequencies.

Genetic linkage

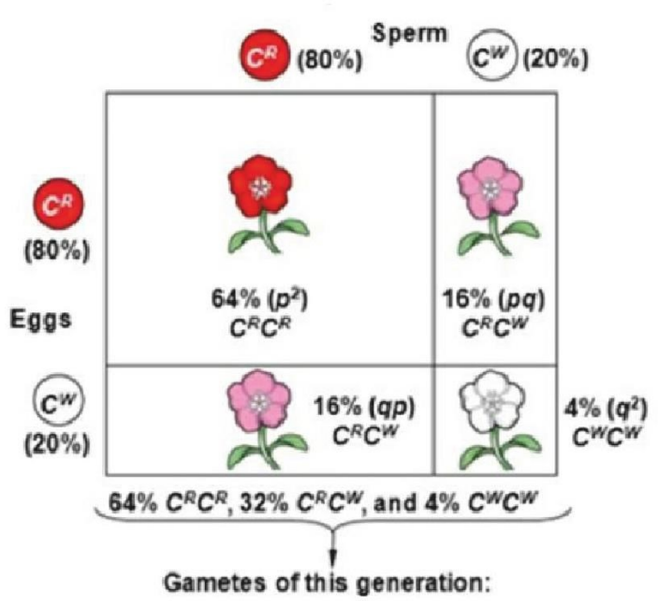
- Some genes coding for particular characters are located on the …….. …….. and also at a closer distance. Thus, they …….... from ……. …….. and …… ……... occur during the meiotic cell division at gametogenesis and inherit together. This results in …….. from Mendel's law of independent assortment. The above phenomenon is called …… ……...
  Eg. Inheritance of body colour and wing size in the fruit fly Drosophila.



In *Drosophila,* wild-type flies are found to have gray bodies and normal-sized wings. Due to mutation for the above traits body colour becomes black and wings become vestigial. Both characters are determined by genes of autosomal chromosomes.

In this example, the mutant alleles are recessive to the wild-type alleles. The alleles for body colour are indicated as G (gray) and g (black), and those for wing size are indicated as N (normal) and n (vestigial).

To examine the above, wild type flies were crossed with flies which are mutant to both body color as well as wing size and followed by a dihybrid test cross.



During random fertilization, gametes fuse together randomly. Therefore, rule of multiplication can be applied to calculate the probability for each genotype combination.

According to Hardy-Weinberg equilibrium, if a character is determined by two alleles, the three genotypes will appear in the following proportions;

P^2 = Frequency of dominant homozygotes

q^2 = Frequency of the recessive homozygotes

2Pq = Frequency of the heterozygotes

The probability that two C^R alleles will come together, P x P= P^2 = 0.8 x 0.8 = 0.64

Hence the proportion of C^RC^R genotype in the progeny = 64%

The probability that two C^W alleles will come together, q x q = q^2 = 0.2 x 0.2 = 0.04

Hence the proportion of C^WC^W genotype in the progeny = 4%.

C^RC^W heterozygotes can arise in two different ways.

If the sperm provides the C^R allele and the egg provides the C^W allele, the resulting C^RC^W heterozygotes in the progeny, P x q = 0.8 x 0.2 = 0.16 = 16%

Population Genetics

Hardy-Weinberg Equilibrium

Hardy-Weinberg Equilibrium is used to assess whether a population is evolving with respect to a particular characteristic/ genetic locus. The genetic makeup of a trait in a population would remain unchanged, if they are not evolving at that genetic locus. Therefore, the predicted data for a particular trait of a population can be compared with the actual data obtained from the same population. The comparison of both data as mentioned above may help to determine whether the population evolves or not for the considered trait.

Hardy-Weinberg Equilibrium Principle

In 1908 British mathematician G.H. Hardy and German physician W. Weinberg, independently showed that, in a population that is not evolving, allele and genotype frequencies will remain constant from generation to generation. This is now considered as key concept in population genetics and referred to as Hardy-Weinberg Equilibrium Principle.

To determine whether the allele and genotype frequencies have changed in consecutive generations, a Punnett square can be drawn, considering the combination of alleles in all possible crosses in a population.

The following example can be used to work out the Hardy Weinberg equilibrium. A wild flower plant population showing incomplete dominance for the flower colour alleles have distinct phenotypes indicating their genotypes.

Plants homozygous for the C^R allele (C^RC^R) produce red pigment and have red flowers.

Plants homozygous for the C^W allele (C^WC^W) have white flowers .

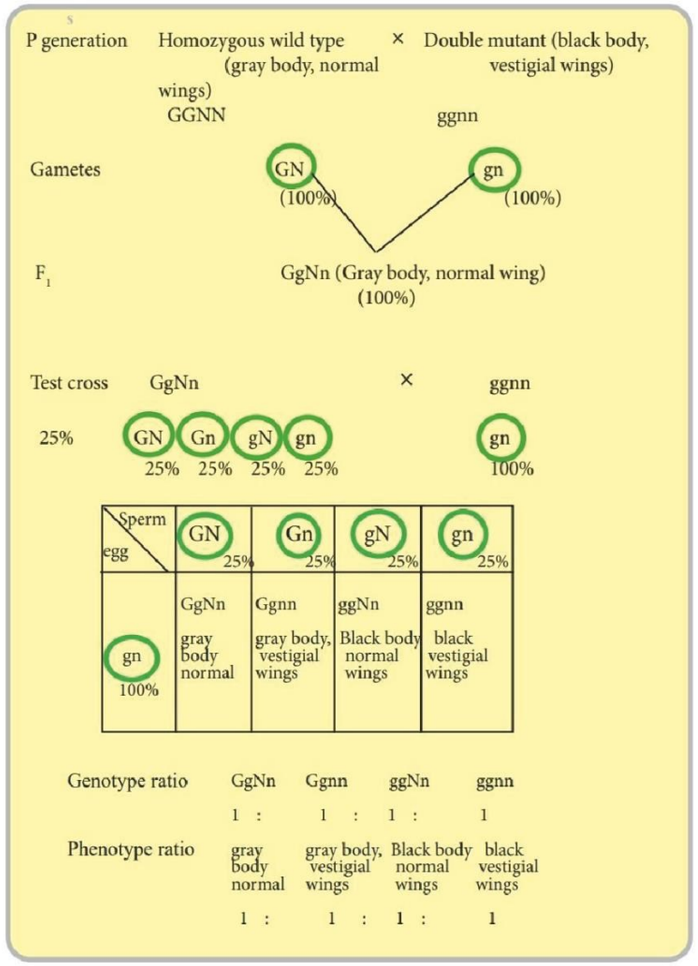
Heterozygous plants (C^RC^W) produce some red pigment and have pink flowers.

In the population of 500 flowers, there were 800 C^R alleles and 200 C^W alleles. Since the flower colour of the above plant type is determined by pair of alleles, 500 flowers may consist of 1000 alleles for their flower petal's pigmentation.

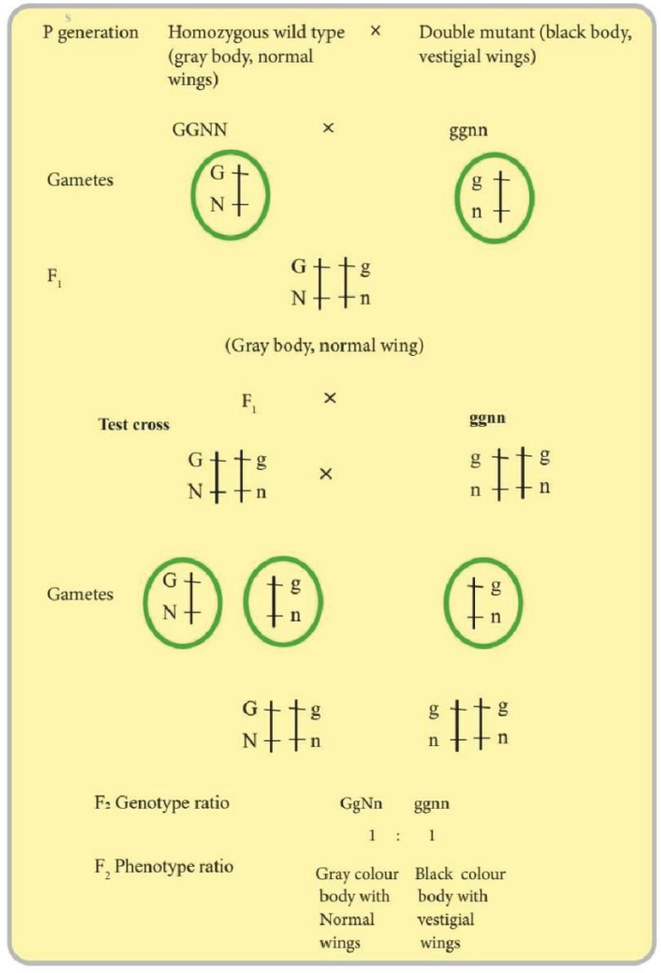
Therefore,

- allele frequency of C^R alleles (p) = 800 /1000 = 0.8
- 1000 allele frequency of C^w alleles (q) = 200/1000 = 0.2

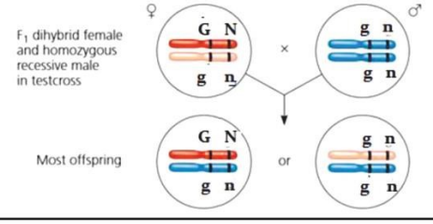
If the gametes are formed at random, the probability that an egg or sperm contains a C^R or C^W allele is equal to the frequency of each of these alleles in the population. Thus, each egg has an 80% chance of containing a C^R allele and a 20% chance of containing a C^W allele; the same is true for each sperm.



Most off spring had a parental (P generation) genotype, indicating that the genes for body colour and wing size are genetically linked on the same chromosome.



Though, the genes for body colour and wing size are linked, in some occasions, they get assorted independently due to …… ……... Therefore, the above test cross may results recombinant off springs in ……... frequency. For example, in Morgan's experiment recombinant phenotypes Gray colour body with vestigial wings (Ggnn) and Black colour body with normal wings (ggNn) were observed in lesser number.



The production of a relatively …….. number of offspring with …….. phenotypes indicated occasional breaks in the genetic linkage. This is due to the crossing over occurs between the homologous chromosomes.

Questions

1. In *Drosophila,* gray bodies (G) dominant to g (black). Normal-sized wings (N) dominant to n (vestigial). If 2 dihybrid flies were crossed, find out the probable genotypic and phenotypic ratios of offspring's when genes are linked and crossover absent.
2. Tall tomato plants having red fruits were crossed with short tomato plants having orange fruits. All plants in F_1 generation were tall with red fruits. When F_1 plants were crossed with each other the following phenotypic ratio was obtained in the F_2 generation.
   Tall plants having red fruits : Short plants having orange fruits
   3 : 1
   Explain this result.
3. Homozygous dominant tomato plant with red (R) spherical (S) fruits was crossed with a homozygous recessive tomato plant with yellow (r) elongated (s) fruits. Resulting F1 were back crossed with the double recessive mother plant. Progenies are as follows.
   Red / spherical - 24 Yellow / elongated - 25 Explain this result.

Human sex determination

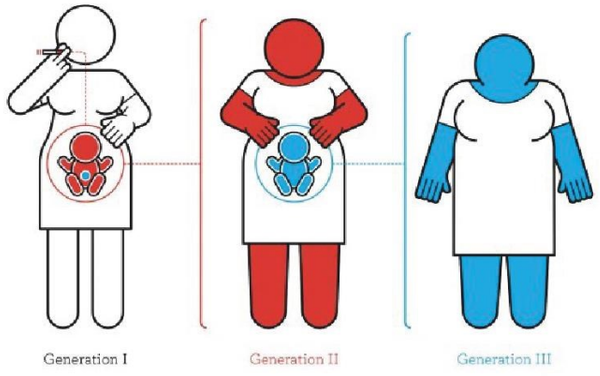
Sex is determined by the expression of sex chromosomes. In humans, each individual carries …….. pairs of ……. chromosomes and one pair of ……. chromosomes. Type of sex chromosomes expressing male traits are named Y chromosome and the other as X chromosome. Comparatively X chromosome is bigger than Y chromosome. They both code for …… traits except their …….. regions. When X and Y chromosomes pair up, they remain homologous only in specific regions. On the other hand when chromosome X, X pair up, they both remain homologous to each other.

On the occurrence of gametogenesis in females, meiosis yields haploid eggs carrying 100% X chromosomes, whereas in males, half the number of the haploid sperms produced carry X and the remaining half Y chromosomes. During the fertilization of male and female gametes, the occasion where both egg and sperm carry X chromosomes results in a female zygote, and on the other hand, an occasions where an egg fuses with a sperm carrying Y chromosome results in a male zygote.

Epigenetics (2020 AL Short Notes)

Study of occurence of certain phenotypes of certain characters controlled by factors other than their DNA sequence or genetic code is called epigenetics. This is due to '……..' and '…….. ' of certain genes by modifying nucleotides of a DNA sequence by …….. and ……., where methyl groups are added to ……. type DNA sequence or else removed from a methylated DNA sequence. The above random occasions result different modified expression for a single DNA sequence.

Epigenetics results due to either inherited signals from parents or signals arising due to the environmental factors. Inheriting epigenetic traits from parents to the children's generation is called epigenetic inheritance. This may get reversed by various external stimuli from the environment. Some epigenetic influences result in inappropriate gene expressions leading to cancers.

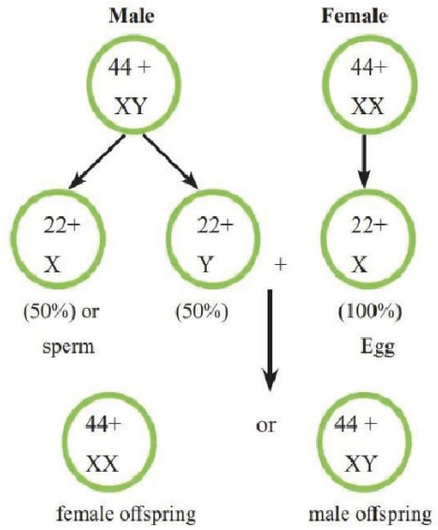


Schizophrenia is a mental disorder that occurs due to the genetic defects. In some identical twins, only one of them gets schizophrenia and the other does not get it. This is due to two types of expressions for same DNA sequence, called epigenetics.

Cystic fibrosis

Cystic fibrosis is a disease condition causing …….. and ……... …….. than its …….. nature. As a result, mucus get …….. in the ……..., ……..., …….. ……., and ……... organs which cause lung infections, respiratory failure, poor digestion, and infertility. The thickening of mucus is due to the excess chlorine secretion of defected chloride channels of the plasma membrane. The defect in the trans-membrane chloride channel occurs as a result of the ……..… ……... ……..(CFTR) protein. The altered CFTR protein is due to the mutation of CFTR gene. This is identified as autosomal recessive disorder.

Thus, any mating occasions between male and female organisms of same species leading to fertilization can have …… percent chance for yielding either male or female zygotes. Anatomical sex signs develop in humans based on the expression of XX and XY chromosomal combination.



Human sex linked characteristics

Certain characters of humans are carried on the genes located on the sex chromosomes. Those ……. located on the ……. chromosomes are called ……. …….. genes and the characters expressed by them are called sex linked characters. Characters expressed by or carried on the X chromosome are called X- linked characters and the genes expressing or carrying those characters are called …….- linked genes. On the other hand, characters which are expressed by or carried on the Y chromosome are called …….. linked genes and the genes expressing or carrying those characters are called Y linked genes. Y chromosome carry only …….. …….. other than those related to the sex. Some disorders carried on the Y-linked genes are transferred and expressed only through male progeny.

Eg: ……

……

In addition to sex related characters, X chromosomes carry many other characters which are not relevant to individual sex.

Red green colour blindness

An X-linked recessive disorder characterized by the difficulty in perceiving differences between red and green colours.

Haemophilia:

An X-linked recessive disorder where one or more of the proteins required for blood clotting are absent. Haemophilic person run the risk of severe bleeding during injuries due to the delay in clot formation. Campbell et al, 2015)

Inheritance of X-linked genes

The inheritance of sex linked characters or genes differ for male and female due to the ……. genotype of …….... and the …..……. genotype of ……..…..

During the fertilization, X chromosome from both of their biological parents result a female zygote and on the other hand, X chromosome from the female parent and Y chromosome from the male parent results a male zygote. Thus, for females, X-linked recessive disorders are expressed only at their homozygous genotype. However in males, due to the presence of only one X chromosome, have only one recessive X-linked allele. Therefore, having a recessive X-linked allele with disorder is sufficient for expression.

Questions

1. Colour blindness is due to presence of a recessive allele on X chromosome. A normal man whom has a colour blind father got married to a normal woman whom has a colour blind father. What is the probability of their 1st child to be a colour blind?
2. Father of a woman was suffering from hemophilia and her mother comes from a non hemophilic family.
   If this woman married to a normal man, then find the probability of their 1st child to be hemophilic. Find the probability of 1st and 2nd child to be hemophilic.

Pleiotropy (2020 AL)

……

……

The above phenomenon is called as Pleiotropy.

Pleiotropic alleles are responsible for the multiple symptoms associated with certain hereditary diseases in humans, such as cystic fibrosis and sickle-cell disease.

Sickle-cell disease

Sickle cell disease is caused by an alteration in the haemoglobin protein of red blood cells. A single gene mutation is responsible for the above condition. In homozygous recessive individuals, all the haemoglobins are of the sickle-cell variety. People living in high altitudes or under physical stress experience low oxygen content in their blood. Low oxygen content in the blood may induce the sickle-cell haemoglobin proteins to get accumulated and results sickle shape in red blood cells.

Sickle cells may clump and clog small blood vessels causing tissue and organ damage in several body parts. This may result renal failure, heart failure and thrombosis.

(Hb^A—Normal, Hb^S– Sickle)