

# ① Pleiotropism Or Pleiotropic Gene

**Introduction :-** When a gene produces more than one phenotypic effect totally unrelated, it is called pleiotropic gene & this phenomenon of controlling more than one character at the same time called pleiotropism.

Mendel propose law of unit character which means each character is controlled by one gene but a pleiotropic gene along with controlling the expression of its phenotypic trait also influences some related or unrelated character / characters. The main character which a pleio-tropic gene influences represents its major effect while a less evident phenotypic expression of its other trait is called minor effect.

- eg~
1. In house rat, the recessive gene  $dd$  for dwarfness also influences the development of eosinophilous cells of pituitary gland. Due to this secretion of pituitary hormone

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is checked.

2. Sickle cell anaemia is a hereditary disease found in tropical Africa but is now spreading in American blocks where whose ancestor came from that part of Africa & Kerala of India. Persons suffering from disease have sickle cell haemoglobin ( $Hb^S$ ) in their RBCs. Such RBCs become sickle shaped under low  $O_2$  concentrations.

Sickle cell haemoglobin or haemoglobin 'S' molecule ( $Hb^S$ ) is different from normal haemoglobin called haemoglobin A ( $Hb^A$ ). When RBCs containing  $Hb^S$  face  $O_2$  shortage, the  $Hb^S$  molecule aggregate & form stiff fibres. These fibres caused distortion of RBCs which become sickle shaped. The distorted stiff RBC failed to stick squeeze through narrow capillaries unlike the normal flexible RBCs. The sickled RBCs get stucked blocking the fine capillaries & reduce circulation. Sickled RBCs break down more easily & this decreases the number of RBC in blood causing anaemia.



(b)

The gene  $Hb^s$  that produces sickle cell haemoglobin has pleiotropic effect. Its major expression is production of Hb<sup>s</sup>. The other effects are sickling of RBC, poor circulation, anaemia, jaundice & low resistance to infection.

Inheritance of sickle-cell anaemia ~ the disease

Sickle cell anaemia is caused by gene which is lethal in homozygous condition.

$Hb^A Hb^A$  — normal (haemoglobin A only, no sickling)

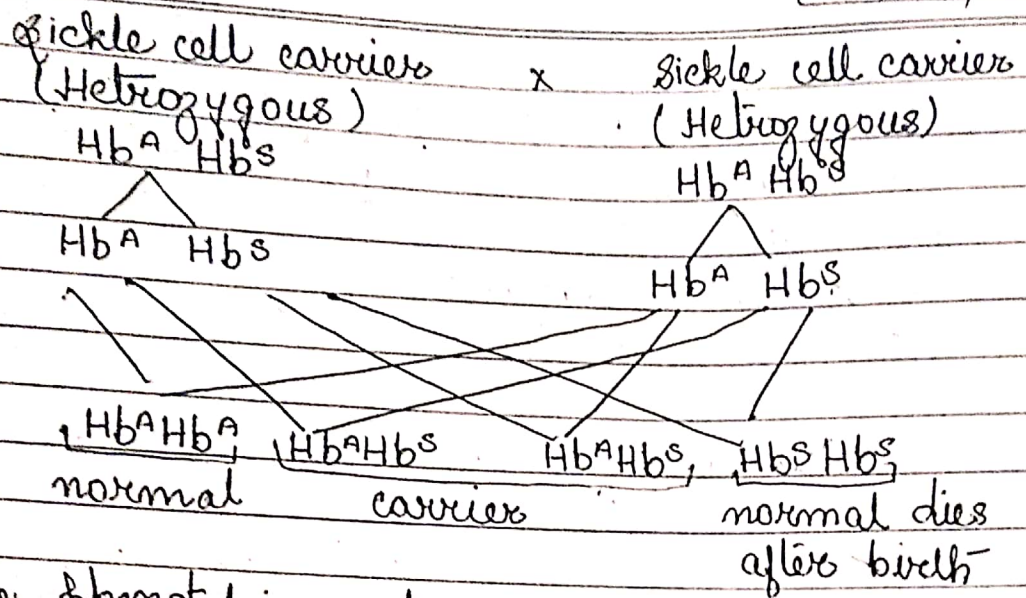
$Hb^A Hb^S$  — carrier i.e.; sickle-cell trait & haemoglobin A & S are found.

Because of both type of haemoglobin intermediate, forms are formed produce i.e., a case of co-dominance. In it sickling under reduced  $O_2$  tension occurs.

$Hb^S Hb^S$  — sickle-cell anaemia (fatal).  
Haemoglobin S only & sickling occurs under normal  $O_2$  tensions.

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(4)



∴ phenotypic ratio = 1:2:1

Marriages between the two carriers produce normal, carriers & sickle cell anaemic person in the ratio of 1:2:1. But sickle cell anaemia bearing carriers & normal ratio is 2:1. Birth of children with sickle cell anaemia can be avoided by discouraging marriages among heterozygotes i.e. among persons with sickle cell trait. The heterozygote carriers can be identified by microscopic examination of blood.

Genes for sickle cell anaemia is lethal in homozygous condition but produces sickle cell trait in heterozygous carrier.



(5)

It means it is a case of co-dominance. A lethal gene is defined as a gene which causes death of the bearer.

Difference between  $Hb^A$  &  $Hb^S$  — Haemoglobin has 2  $\alpha$  &

2  $\beta$  polypeptide chains.  $Hb^S$  Gene  $Hb^S$  codes for polypeptide chain of sickle cell haemoglobin. This allele is a mutant of normal  $Hb^A$  gene & produced by a change in one nucleotide pair in the 6th codon in its cistron DNA. This causes change or substitution of 6th amino acid — Glutamic acid by ~~alanine~~ <sup>valine</sup> in the  $\beta$ -polypeptide chain of haemoglobin. NEEL & BEET 1949 independently proposed that sickling was caused by single mutant gene.

$\beta$ -chain of normal haemoglobin ( $Hb^A$ )

$\beta$ -chain of sickle cell haemoglobin ( $Hb^S$ )

1 Valine

valine

2 Histidine

Histidine

3 Leucine

Leucine

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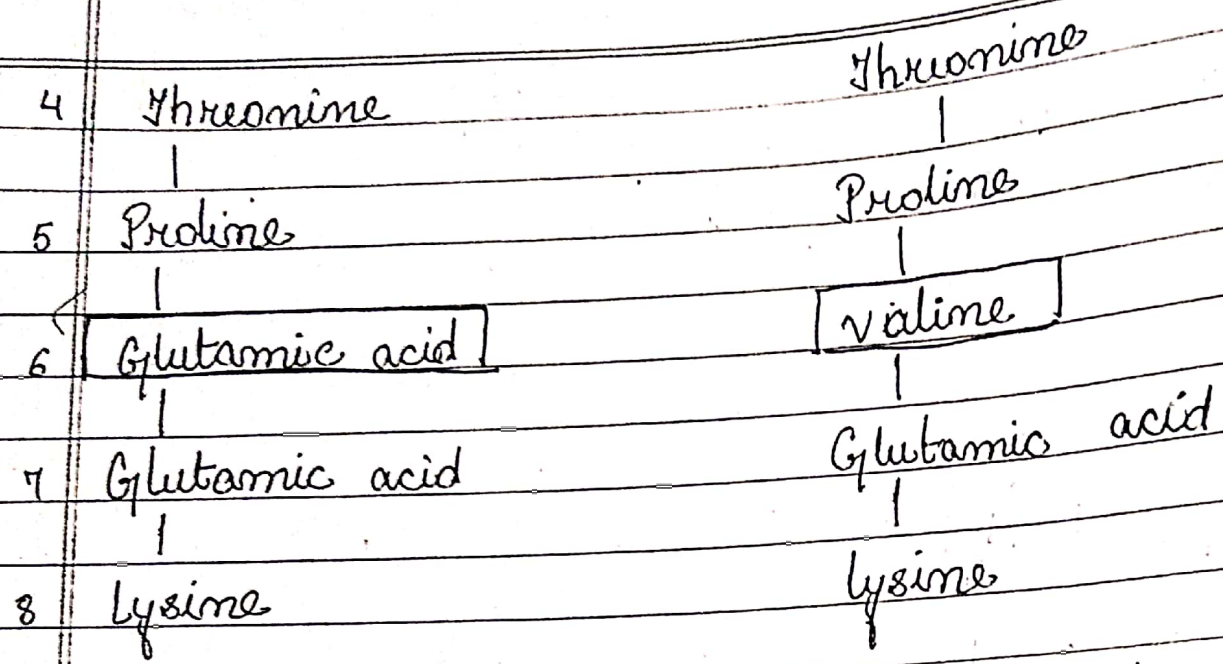


fig: showing difference in the amino acid sequence of HbA & HbS

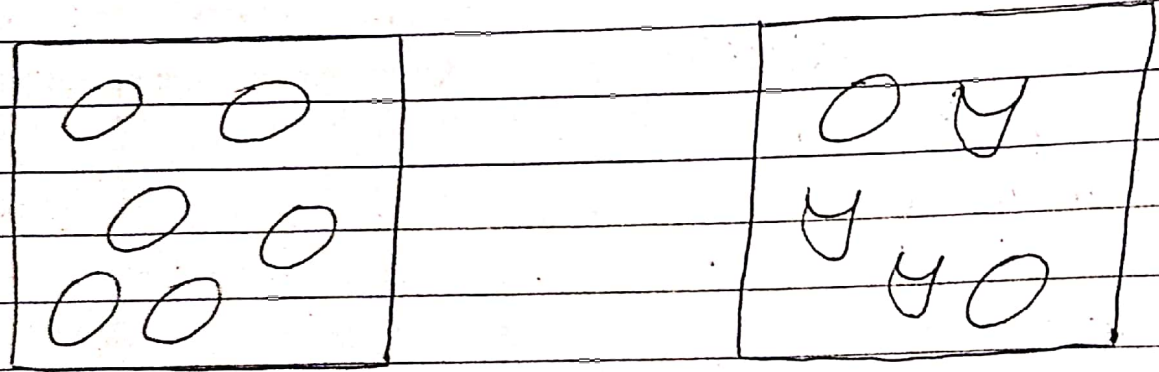


fig: RBC of a normal person

fig: RBC of a person suffering from sickle cell anaemia (carrier)

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