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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 d^{dw} dw 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.

Sickle cell anaemia is a hereditary disease found in tropical africa but is now spreading in American blocks where whose ansester 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 concentration.

Sickle cell haemoglobin or haemoglobin S molecule (Hb^S) is different from normal haemoglobin called haemoglobin A (Hb^A). When RBCs containing HbS face O_2 shortage, the HbS molecule aggregate & form stiff fibres. These fibres caused distorsion of RBCs which become sickle shaped. The distorted stiff RBC failed to stick squeeze through narrow capillaries unlike the normal flexible RBCs. The sickled

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The gene Hb^S that produces sickle cell haemoglobin has pleiotropic effect. Its main expression is production of HbS. The other effects are sickling of RBC, poor circulation, anaemia, jaundice & low resistence 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 haemoglo-bin intermediate forms are foun produce. i.e; a case of co-domin-ance. In it sickling under redu O_2 tention occurs.

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

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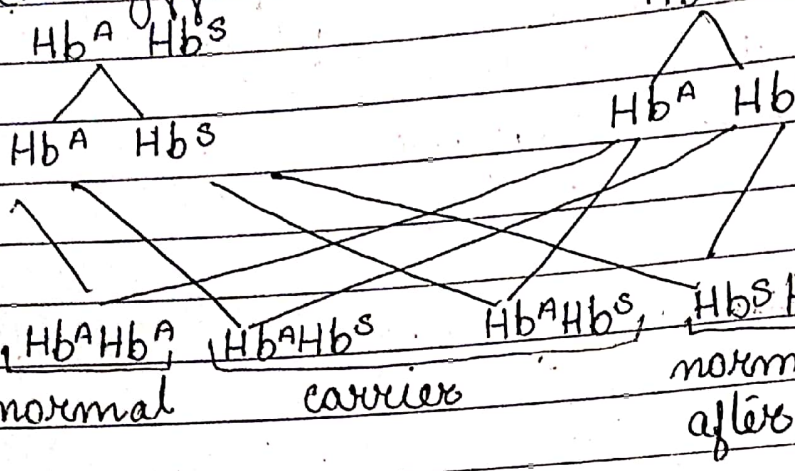
Sickle cell carriers (Hetrozygous) $Hb^A Hb^S$ × Sickle cell carriers (Hetrozygous) $Hb^A Hb^S$



Ratio 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 hetrozygotes i.e; among persons with sickle cell trait. The hetrozygote carrier can be identified by microscopic examination of blood.

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

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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 α & 2 β polypeptide chains. 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 valine in the β-polypeptide chain of haemoglobin. NEEL & BEET 1949 independently proposed that sickling was caused by single mutant gene.

	β-chain of normal haemoglobin (Hb^A)	β-chain of sickle cell haemoglobin (Hb^S)
1	Valine	Valine
2	Histidine	Histidine
3	Lucine	Lucine

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4	Threonine	Threonine
	|	|
6	Proline	Proline
	|	|
6	Glutamic acid	valine
	|	|
7	Glutamic acid	Glutamic acid
	|	|
8	Lysine	Lysine

fig: showing difference in the amino acid sequence of Hb^A & Hb^S

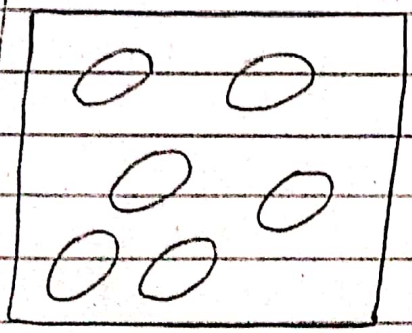

fig: RBC of a normal person

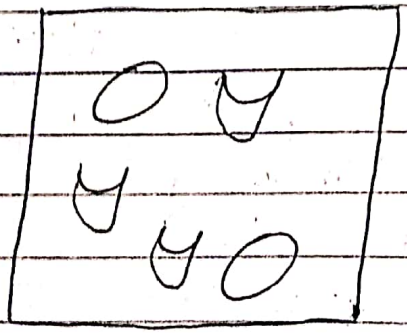

fig: RBC of a person suffering from sickle cell anaemia (carrier).

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