Sickle Cell Anemia

It is hereditary disease, **microcytic** anemia called HbSS, caused by mutation occur in the beta globin chain, in the **6th** position on the chromosome **11**.

Person have HbSS may be homozygous or heterozygous, if an individual is homozygous mean have sickle cell disease, but when individual is heterozygous, have the Sickle cell trait

The bone marrow can't make new red blood cells fast enough to replace the dying ones. Typically appear during infant's (after the 6th months of the life).

Caused of HbSS:

The mutation occurs in gene coding for hemoglobin, lead to single base change in the DNA, (**adenine is replaced by thymine**) this leads to an amino acid change from **glutamic acid** to **valine** in **sixth position** of the **β- globin chain**.

Normal β- chain	Amino acid	pro	glu	glu
	Base composition	CCT	GAG	GAG
Sickle β- chain	Base composition	CCT	GTG	GAG
	Amino acid	pro	val	glu

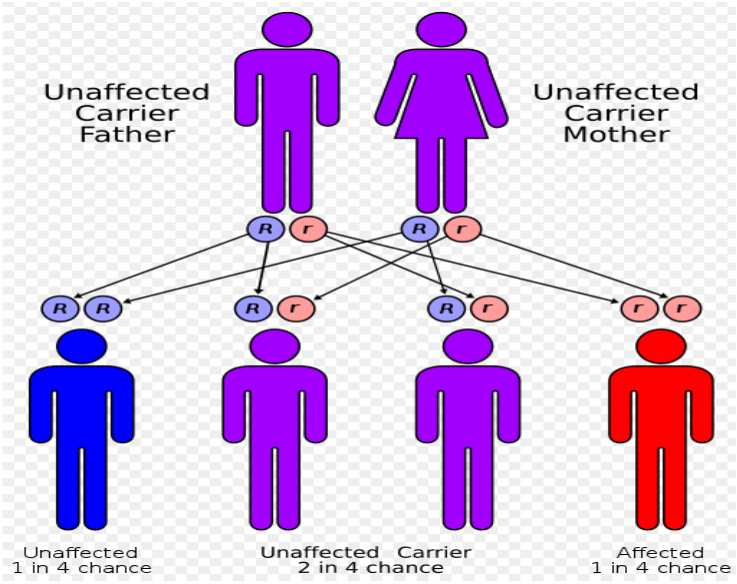
Sickle disease inheritance:

The presence of two defective genes (**SS**) is needed for sickle cell anemia.

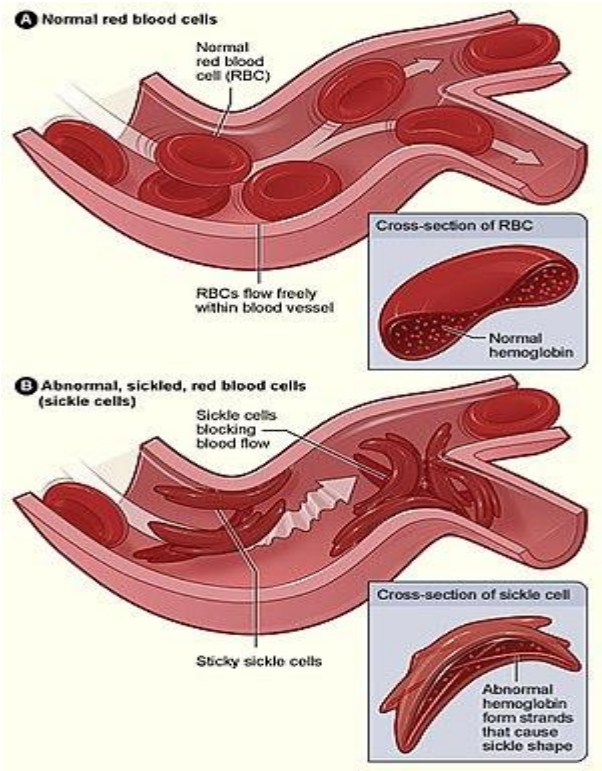
If each parent carries one sickle hemoglobin gene (**S**) and one normal gene (**A**), each child has a **25%** chance of inheriting two defective genes and having sickle cell anemia.

25% chance of inheriting two normal genes and not having the disease.

50% chance of being an unaffected carrier like the parents.



Symptoms:



1-**Anemia**: sickle cells break apart easily and die, red blood cells usually live for about 120 days before they need to be replaced. But sickle cells usually die in **10 to 20 days**, leaving a shortage of red blood cells (anemia).

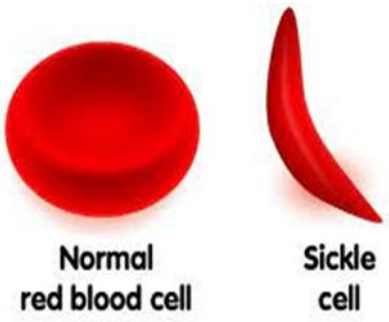
2-**Pain (Sickle Cell Crisis)**: sudden episode of pain throughout the body.

3-**Painful swelling of hands and feet**: the swelling is caused by sickle-shaped red blood cells blocking blood flow to the hands and feet.

4-**Delayed growth**: red blood cells provide oxygen and nutrients for body, that important for growth. A shortage of healthy red blood cells can slow growth in infants and children and delay puberty in teenagers.

5-**Vision problems**: tiny blood vessels that supply your eyes may become plugged with sickle cells, that can damage the retina (the portion of the eye that processes visual images), leading to vision problems.

Changing in Red blood cell of HbSS:



1-Because abnormal structure of hemoglobin, in sickle cell disease, **low oxygen tension promotes red blood cell sickling** and repeated damage of cell membrane and **decrease the cell's elasticity**.

2-The rigid blood cells are unable to deform as they pass through narrow capillaries, leading to vessel.

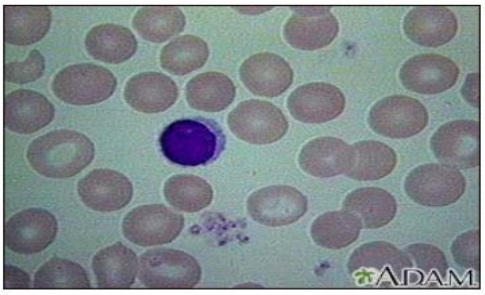
3-The actual anemia of the illness is caused by rapid hemolysis because short RBC life, the destruction of the red cells inside the spleen

Laboratory findings:

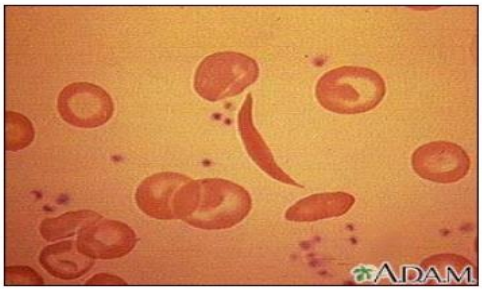
1-The hemoglobin is usually **6-9 g/dL-low**

2-Blood Film – indicate the presence of sickle cells

3-Hemoglobin electrophoresis in Hb SS, no Hb A is detected. The amount of Hb F is variable and is usually 5-15%,



Red blood cells, normal cell



Red blood cells, sickle cell

References:

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