Sickle Cell Anemia

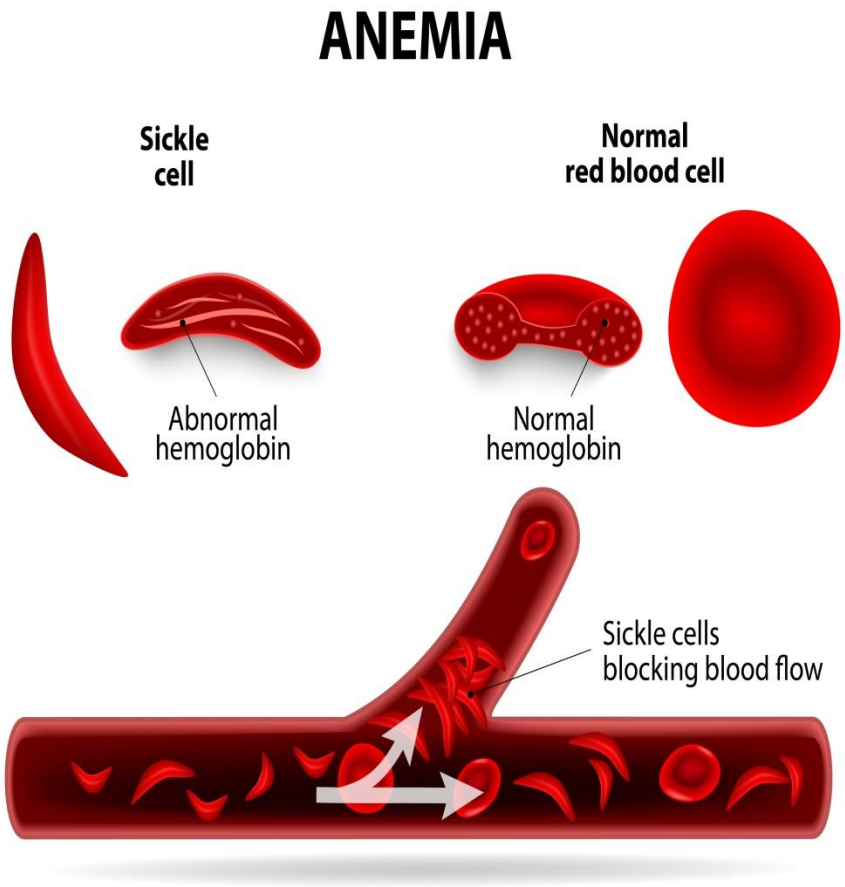
Dr. S.D. SARASWATHY
Assistant Professor
Department of Biomedical Science
Bharathidasan University
Tiruchirappalli

Objectives

- Definition
- Introduction
- Pathophysiology
- Symptoms and Signs
- Diagnosis
- Treatment

Definition

- Sickle cell anemia (SCA) is an inherited blood disorder which causes red blood cells (RBCs) to become "sickled."



https://aholdencirm.files.wordpress.com/2015/03/shutterstock_236959819.jpg

Introduction

- SCA a genetic disease that is caused by the substitution of a normal hemoglobin (HgbA) for an abnormal form of hemoglobin called hemoglobin S (HgbS).
- Over a time abnormal type of hemoglobin cause the RBCs to become rigid and change shape like crescent moon or sickle.

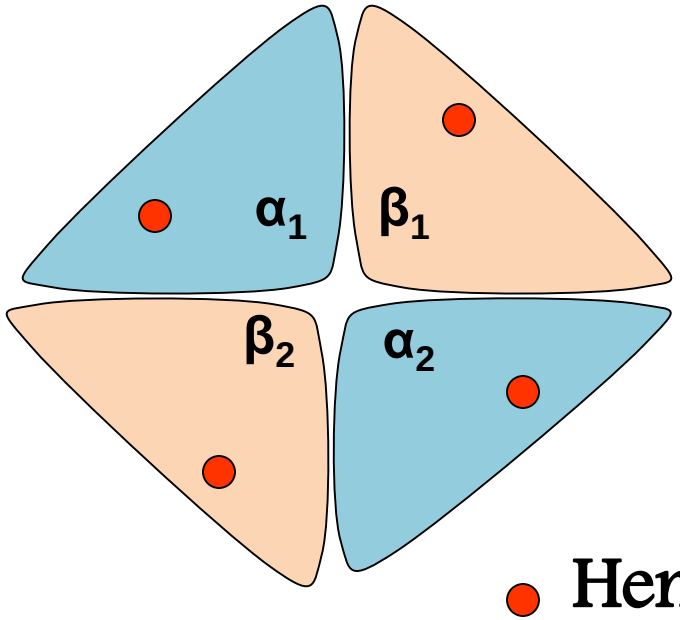
Sickle Cell Anemia (SCA)

- Normal RBCs are soft and round, and can squeeze through tiny blood tubes (vessels).
- The sickle-shaped RBCs have difficulty in passing through the small blood vessels.
- Normally, RBCs life span is about 120 days.
- RBCs with HgbS, life span is too short (normally about 16 days).

Hemoglobin structure

- **Hemoglobin A (HbA) present in Normal Adults:**
- Large Protein (64.4 kDa) attached to four heme moieties
- Tetramer – $\alpha_2\beta_2$
- Globulin portion consists of four polypeptide chains (α with 141 AA and β with 146 AA) arranged in pairs.

- **Hemoglobin** is the main component of the RBC, that carries oxygen from the air in our lungs to all parts of the body.

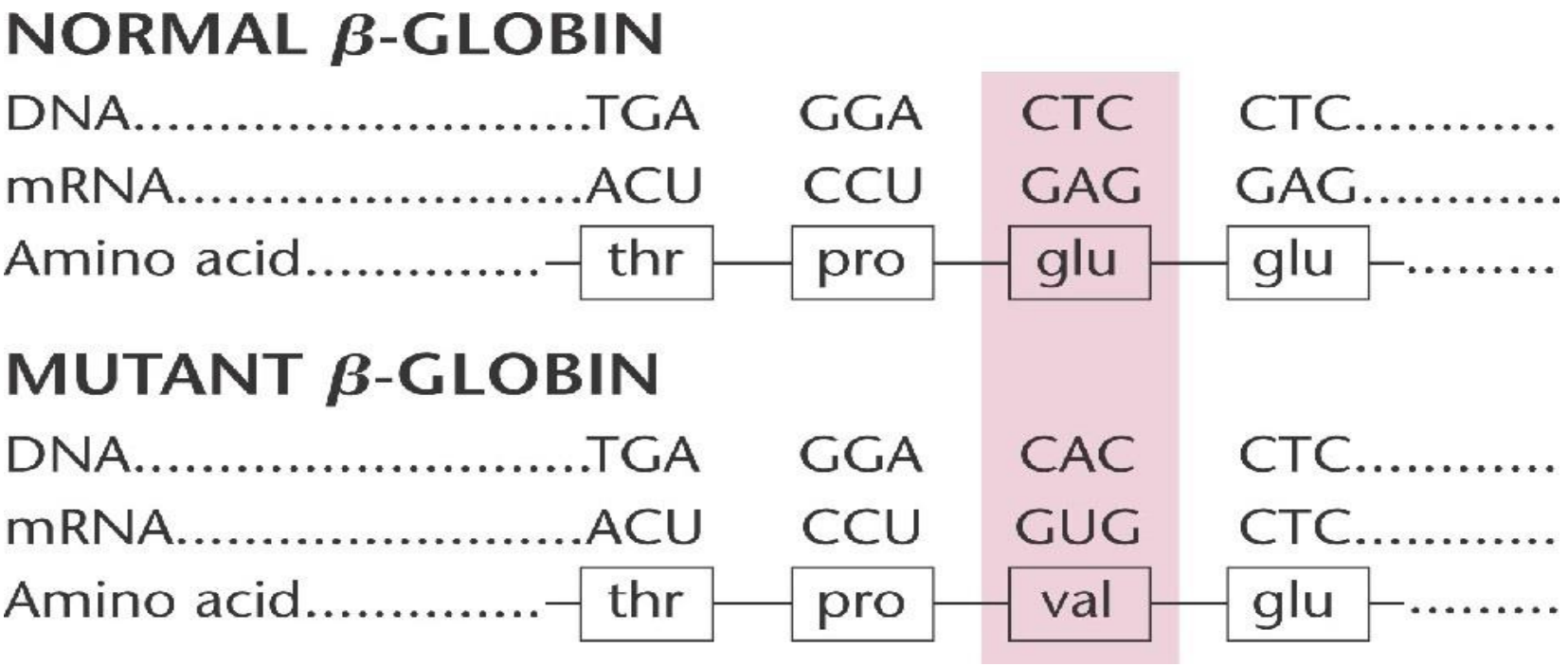


Sickle Cell Anemia - Causes

- Sickle cell disease is the result of a **point mutation**, of the HBB gene (β-globin gene), located on the short arm of chromosome 11.
- *HBB gene* mutation produces an abnormal version of β-globin known as hemoglobin S (HbS).
- β-globin gene is a member of the globin gene family, involved in oxygen transport.
- In Sickle cell disorder at least one of the β-globin subunits in hemoglobin is replaced with HbS.

Sickle Cell Mutation

- A single base pair mutation in the β-globin gene that converts a GAG codon into GUG, which encodes the amino acid valine rather than glutamic acid.



http://www.dokimiscience.com/b---mutation.html

Sickle Cell Trait

- SCA is an autosomal recessive condition.
- A person would have a mixture of HbA and HbS in their RBCs without having sickle cell disease. This condition is called "**sickle cell trait**"
- These people will have enough normal hemoglobin in their RBCs to prevent the cells from sickling.
- SCA is passed on to the children only if both the parents have the defective hemoglobin (HbS) in their RBCs.

Symptoms and Signs

Symptoms usually show up at a young age, vary from person to person and change over time

- **Anemia:** Decrease in RBCs production with increase in destruction of defective RBCs.
- Excessive fatigue or irritability, from anemia.
- **Episodes of Pain:** The sickle cells also block the flow of blood through vessels, thus resulting in periodic pain (chest, arms, legs, abdomen and bone).
- Joint pain that resembles arthritis
- Painful swelling of hands and feet (Hand-foot syndrome
- Delayed growth and puberty

SCA – Clinical features

1. Hemolysis
2. Occlusion of blood vessels by sickled red cells
3. Hematuria, isosthenuria (renal papillary necrosis)
4. Sometimes results in Jaundice.
5. Life-threatening infections.

Diagnosis

- **Hemoglobin Electrophoresis** (using blood sample) will determine the type of hemoglobin.
- Distinct hemoglobins move different distances, depending on their composition.
- This technique differentiates between normal hemoglobin (A), Sickle hemoglobin (S), and other different kinds of hemoglobin (such as C, D, E, etc.).

Treatment

- No cure for sickle cell anemia.
- However, there are treatments that have reduced the death rate and the levels of pain caused by the disease.
- Treatment of complications often includes antibiotics, pain management, intravenous fluids, blood transfusion and surgery.