SICKLE CELL ANEMIA

1. What is sickle cell anemia

Sickle cell anemia (or *drepanocytosis*) is a **genetic blood disorder**.
It affects **red blood cells**, the cells that carry oxygen from the lungs to the entire body.

The disease is caused by a problem in a protein called **hemoglobin**.
Patients affected by this disease have an unusual type of hemoglobin called **hemoglobin S**.

Disease forms

- **Disease (homozygosity βS/βS)**
  People have two copies of the altered form of the gene.
  Their red blood cells break down easily and can "sickle" often.
- **Healthy carriers (heterozygotes βS/β)**
  They have only one copy of the altered gene.
  They usually **have no symptoms** and live a normal life.

Diagnosis

To understand if a person has the disease or is a carrier, two tests are needed:

- **Complete blood count** (CBC)
- **Hemoglobin electrophoresis**: shows if there are abnormal forms of hemoglobin

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2. How red blood cells look in healthy people and in patients

- In a healthy person, red blood cells are **round, soft, and elastic**.
  They flow easily even through the smallest blood vessels.
- In people with sickle cell anemia, under certain situations, red blood cells:
  - change shape and become "**sickle-shaped**", elongated, and rigid
  - stick to each other
  - block small blood vessels
  - can no longer deliver oxygen to tissues

Situations that promote sickling include:
low oxygen, cold, dehydration, infections, and intense physical exertion.

Consequences

- Tissues do not receive oxygen→ **intense pain** and potential organ damage.
- Red blood cells break down easily and live for only **10–20 days** (normally they live for about 4 months).
- This causes **chronic anemia**.

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3. What symptoms can a person with sickle cell anemia have?

Symptoms due to chronic anemia

- tiredness
- fatigue
- difficulty concentrating
- slower growth in children
- **jaundice** (yellow skin and eyes)

Pain crisis: VOC

The most frequent and important symptom consists of:
sickle cell crisis or **vaso-occlusive crisis (VOC)**.

These crises are:

- painful
- recurrent
- unpredictable
- possible at any age
- can last **up to 10 days**

VOC is the **primary cause of emergency room visits and hospitalization**.

Other possible acute complications

They can be very serious:

- **acute chest syndrome**
- **stroke** or other cerebrovascular problems
- **severe acute anemia** due to "sequestration" of red blood cells in the spleen or liver
- **severe infections**
- bone and joint problems
- **priapism** (prolonged and painful erection, unrelated to desire)

Chronic complications

Over the years, the disease can damage various organs:

- heart
- lungs
- kidneys

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4. ⚠️ When should you be concerned?

A person with sickle cell anemia must **contact immediately** their primary care physician, pediatrician, reference center, or go to the **emergency room** if the following appear:

- **severe paleness** or **jaundice**, with or without a high fever
- **severe abdominal pain**, with vomiting and/or fever