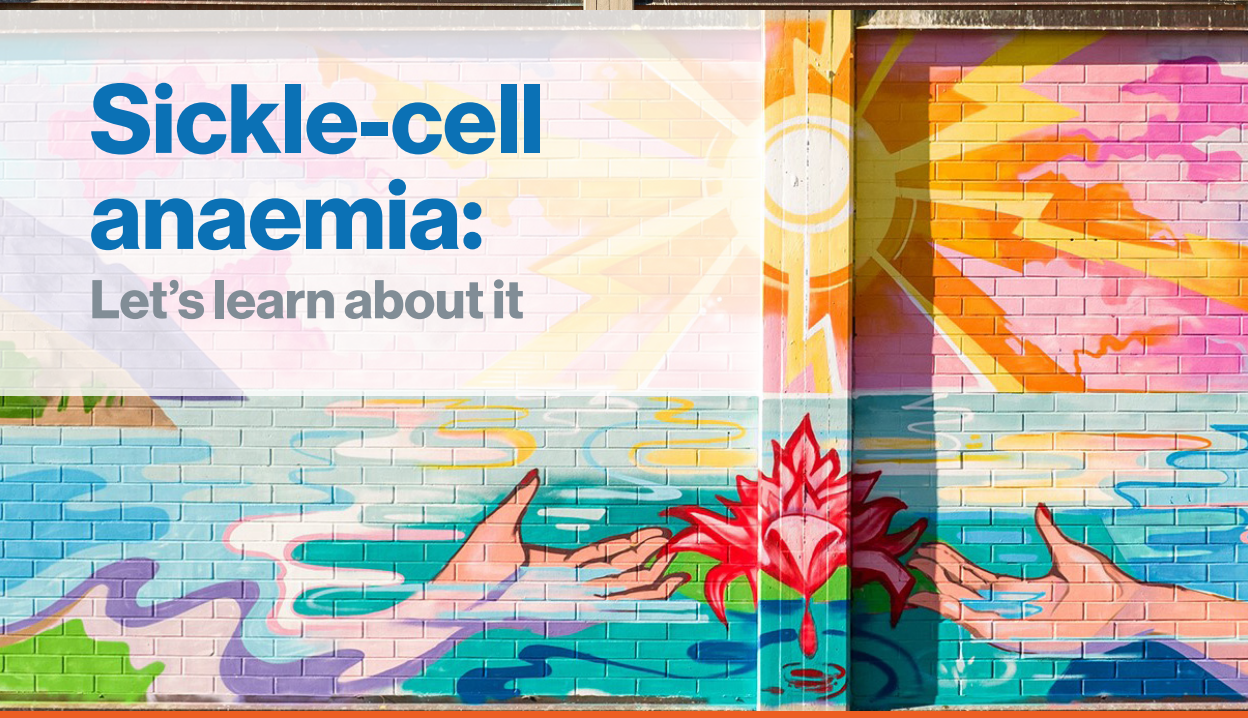
Sickle-cell anaemia:

Let's learn about it

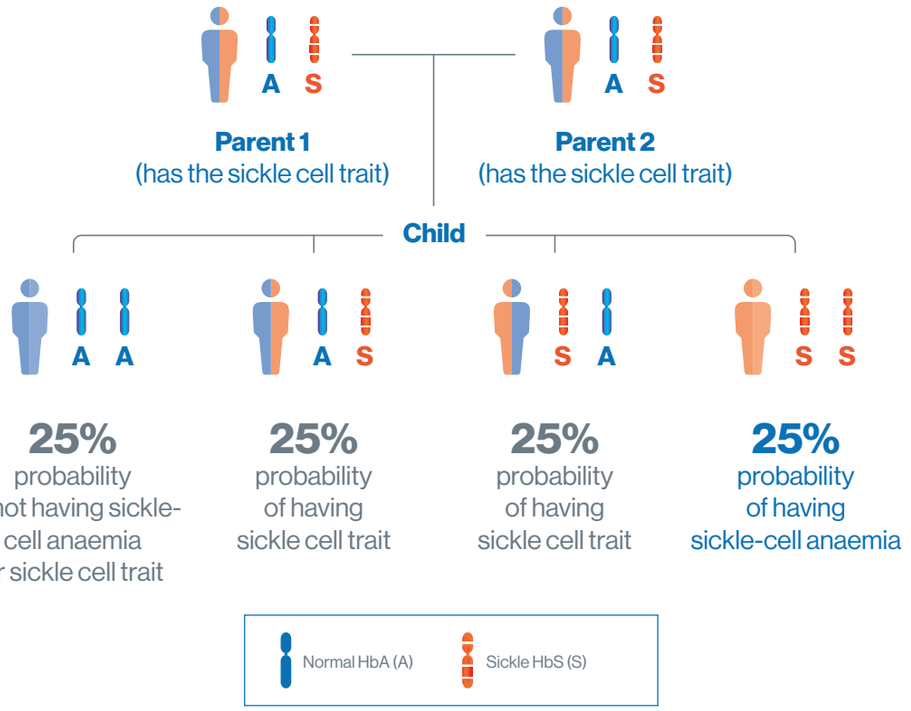


What is sickle-cell anaemia?

Sickle-cell anaemia, also known as drepanocytosis, is a **hereditary disease**, caused by the presence, inside the red blood cells, of a different form **of haemoglobin**, a protein that carries **oxygen** from the lungs to all the tissues of the body. [1]
The presence **of sickle haemoglobin (HbS)** is due to an **alteration** in the **Hb gene**, which results in the characteristic sickle shape of the red blood cells and can be **inherited** from parents. [2]

To be born with sickle-cell anaemia, a **child must inherit** a copy of the mutated gene from **both parents**.This can happen even if both parents are "**healthy carriers**", in other words, they do not suffer from sickle-cell anaemia but have a copy of the mutated gene (a condition known as "**sickle cell trait**"). [3]

What can happen if both parents are healthy carriers of the sickle cell trait:

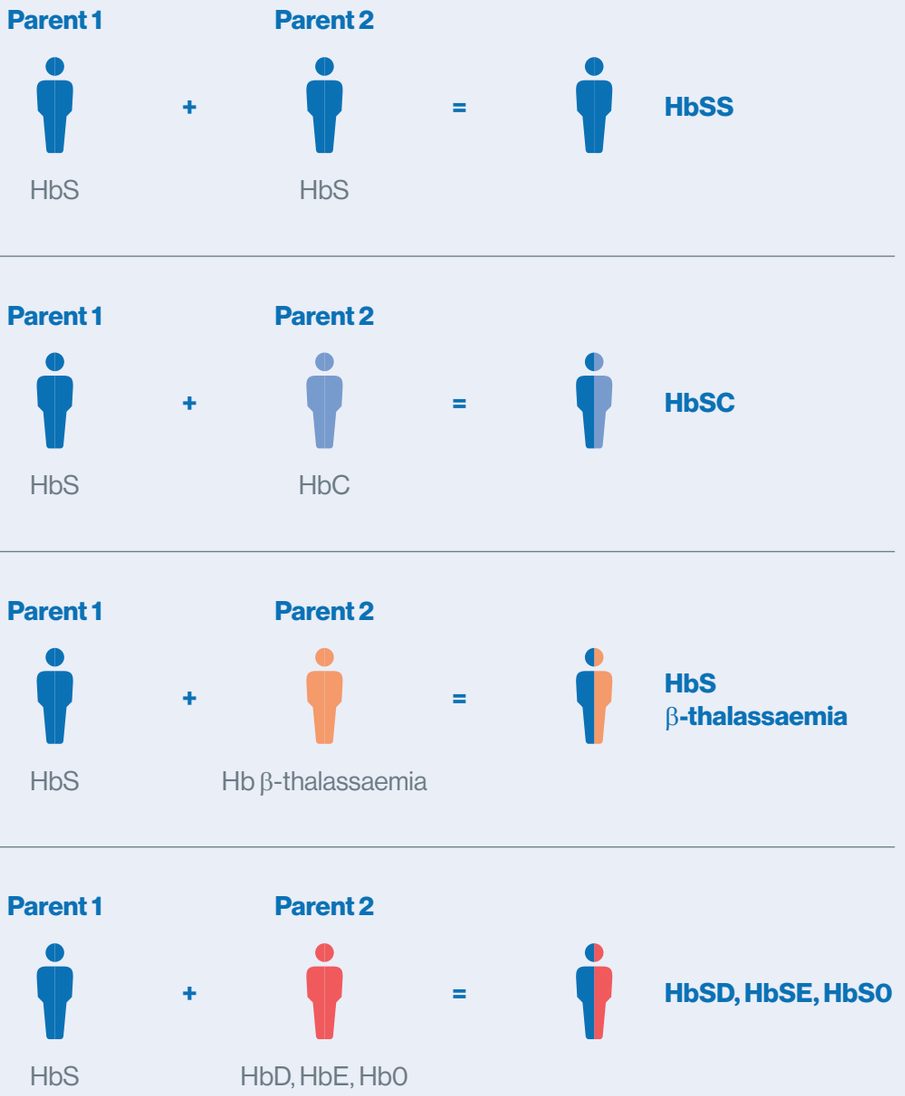


If one parent is a healthy carrier and the other is not (i.e. does not have either sickle-cell anaemia or sickle cell trait), **it is impossible that any children will be born with the disease**. [1]

The different types of sickle-cell anaemia

There are different alterations of the Hb gene that when combined with the HbS gene cause **different types of sickle cell anaemia:** [4]

- Hb SS
- Hb SC
- Hb S β-thalassaemia
- Hb SD, HbSE, HbS0



Sickle red blood cells

Normal red blood cell

Normal red blood cells are shaped like disks, they are **flexible** and **they slide easily** even through the smallest blood vessels. [1]

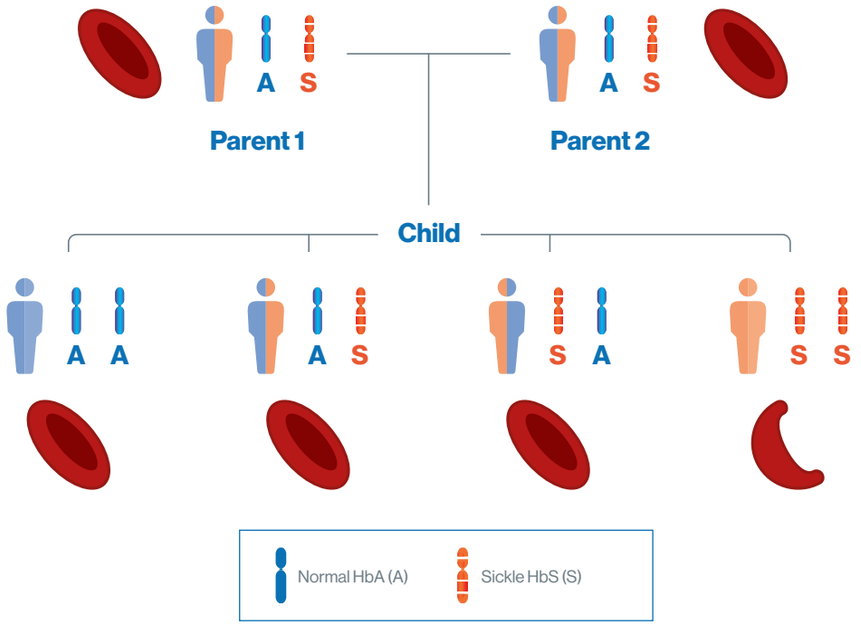
Sickle red blood cell

Sickle red blood cells have an **unusual**, **sickle-like** or **crescent** shape, and they are sticky and stiff. [1]

Haemolysis and anaemia

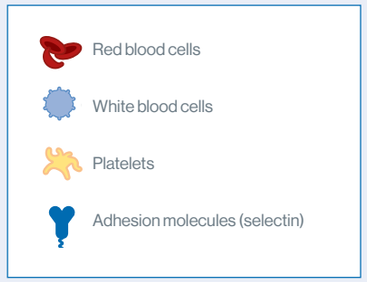
Sickle red blood cells break up faster than healthy ones (a process known as "**haemolysis**"), resulting in chronic **anaemia** (which can make a person feel weak and tired). [1]

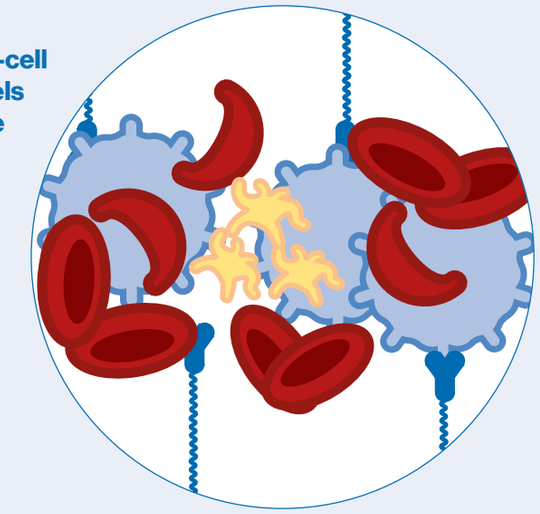
The **defective** HbS gene makes the red blood cells **stiff**, **sickle-shaped** and unable to slide properly in the blood. [1]



Vaso-occlusive crises (VOCS): the distinguishing sign of sickle-cell anaemia [5]

In addition to the red blood cells, **sickle-cell anaemia** also involves the **blood vessels** and other blood cells, such as the **white blood cells** and **platelets**. [5]

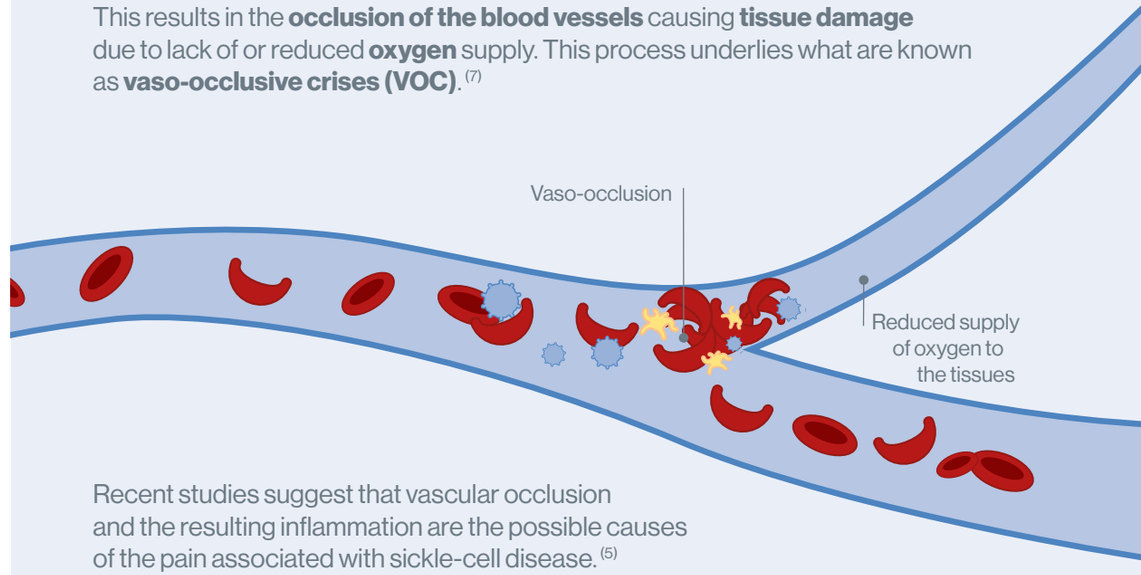




Formation of cell aggregates

Due to the effect of adhesion molecules (**selectin**), which are particularly **plentiful** and **active** in subjects with the disease, the blood cells tend to **clump together** and **adhere** to the vessel walls. [6]

This results in the **occlusion of the blood vessels** causing **tissue damage** due to lack of or reduced **oxygen** supply. This process underlies what are known as **vaso-occlusive crises (VOC)**. [7]



Recent studies suggest that vascular occlusion and the resulting inflammation are the possible causes of the pain associated with sickle-cell disease. [5]

What vaso-occlusive crises (VOCs) look like

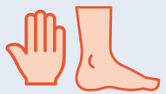
Vaso-occlusive crises (VOCs) are recurrent, unpredictable episodic events associated with **acute pain**.[8]
They occur with varying **frequency** and **intensity** in the different parts of the body.[5]

VOCs can last up to **10 days**.[9]

VOCs are the **main reason for hospitalisation** and for **emergency department** admission among patients with sickle-cell anaemia.[10]

Crises managed at home can also be VOCs. As a matter of fact, a study has shown that about **25%** of VOCs are managed **at home**.[11]

VOCs can occur in any part of the body, for example:



Hands and feet (especially in children)[5]

Chest (ribs and sternum)[5]

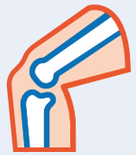
Musculoskeletal sites[12]

It is important to monitor and tell your doctor about all pain crises.

What are the main symptoms of sickle-cell anaemia?

Pain[5]

Fatigue[13]



Joint disorders[13]

Headache[13]

Stress[1]

Dyspnoea[14]

Anxiety and depression[15]

Insomnia[14]

Jaundice (yellow eyes and skin)[1]

Dactylitis (painful swelling of the hands and feet)[5]

Sickle-cell anaemia is a complex condition that **can affect all organs and tissues of the body**.[16]

The disorders caused by sickle cell anaemia **can be managed** and **prevented** with pharmacological therapies and actions that improve quality of life.[1]

Even simple actions can make a big difference

Treatment

Follow a **treatment programme** at a **specialised centre**.[17]

Attend regular check-ups with your specialist.[17]

Be sure to follow **the treatment plan prescribed by your doctor**.

Personal care

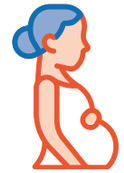
Do **relaxation** and **breathing** exercises **to reduce stress**.[1]

Health

AN adequate **fluid intake** (at least 2 L/day, but not more than 4 L/day) is recommended, especially in the summer.[12]

Avoid exposure to **extreme temperatures** (e.g. hot-cold, bathing in cold water, air-conditioned environments).[12]

Have the **preventive vaccinations** included in the vaccination plan for patients with sickle-cell anaemia.[12]



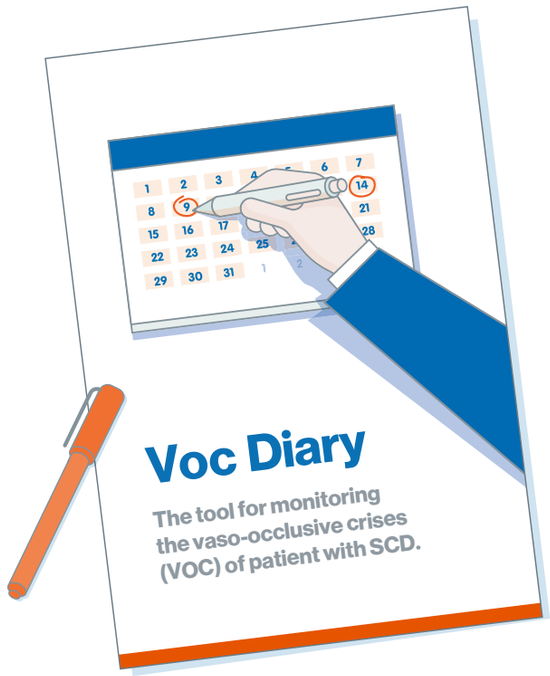
Before starting a pregnancy it is advisable to have **genetic counselling**, and if you are already pregnant, counselling should be arranged as soon as possible.[12]

Monitoring

Remember to **keep track** and **tell your** doctor about all **pain episodes**, even those managed on your own at home.

In the case of an emergency, tell **Emergency** Department **staff** about your condition and remember to always take all **useful documentation** with you.

Never **underestimate** even the mildest of symptoms.



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