Manual for
Sickle Cell Disease

NOVARTIS

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ASAFE

What is sickle cell disease?

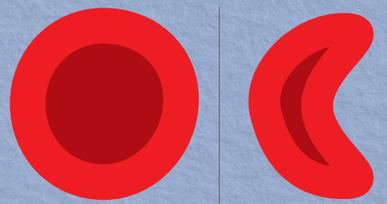
- **Sickle cell disease** (SCD), also known as sickle cell anaemia, is a hereditary blood disease, which is transmitted by genes[1].

- People with SCD have a problem in the gene that produces haemoglobin[1].

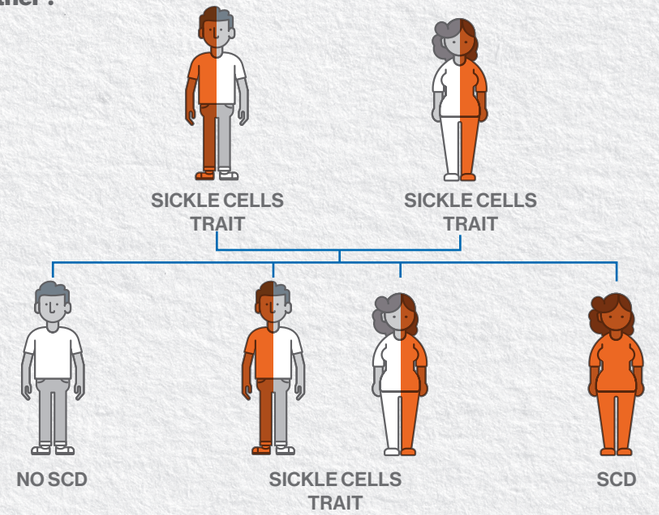
- **Haemoglobin** is a protein present in red blood cells, the blood cells responsible for transporting oxygen from the lungs to every part of body[1].

- In healthy red blood cells, haemoglobin causes these cells to be flat, round and flexible. In contrast, in a person with SCD, abnormal haemoglobin, also called **sickle haemoglobin, causes red blood cells to have a curved and rigid shape** (it is also said that they are sickle shaped)[1].

- Sickle **red blood cells can appear in the blood vessels,** slow down or block blood flow to different parts of the body, possibly causing pain and other health complications[1].



- A person **inherits the sickle cell trait** or SCD in the same way as he or she inherits the colour of his or her hair or eyes, **through the genes they receive from their father and mother[1]:**

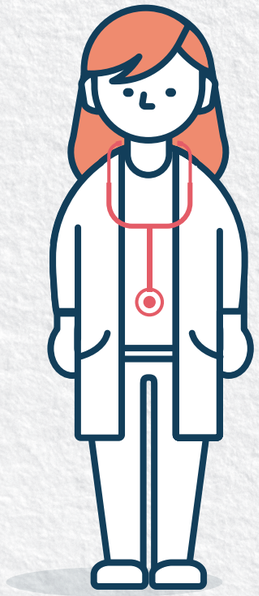


- A person is considered to have **the sickle cell trait** when they inherit a gene that produces normal haemoglobin and another gene that produces sickle haemoglobin. People with the sickle cell trait generally have no symptoms and, in most cases, they are healthy, but they can transmit the altered gene to their children.[1]

- A person with SCD inherits an abnormal haemoglobin gene from each of their parents[1].

SCD is a genetic disease that is suffered if two abnormal haemoglobin genes are inherited, one from each parent[1].

How is it diagnosed?



A **blood test** may show whether SCD or the sickle cell trait has been inherited. Diagnostic confirmation is performed using various methods[1].

The **detection of SCD in newborns** helps to best manage the disease from its early diagnosis.[1]

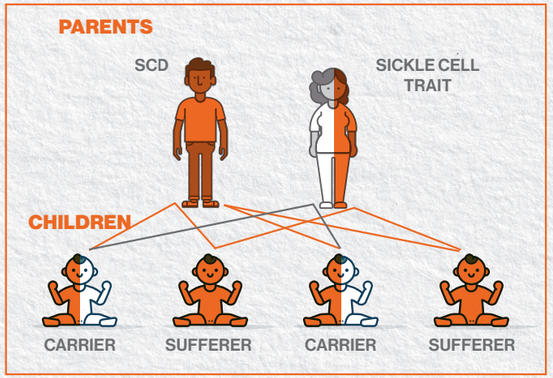
In Spain, families are also informed if it is detected that the baby is a healthy carrier of the sickle cell trait, so that they can receive general recommendations and **genetic counselling,** which is very important when planning a pregnancy.[1]

If a neonatal screening test has not been performed, people with SCD are generally diagnosed when they have a symptom or disease-related complication[1].

Why is genetic counselling so important?

Each person has two genes for haemoglobin, one received from the father and another from the mother[1].

1. When **one of the parents** has a **sickle cell trait,** i.e. they have the gene but do not have the disease, **and the other has the disease[1]:**



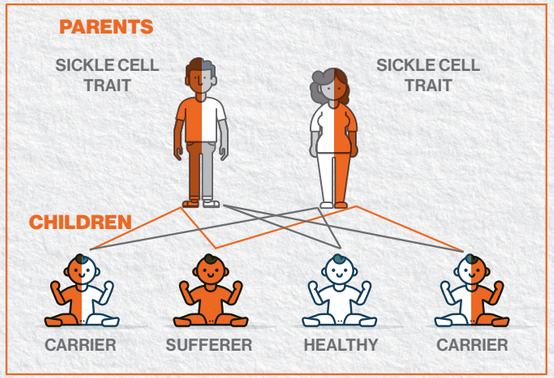
In every pregnancy, there is a[1]:

50% chance of your child inheriting the disease.

50% chance of your child carrying the disease gene (without suffering from the disease).

This probability is the same whether it is the father or the mother who has the disease.

2. When **both parents** have a **sickle cell trait,** i.e. they have the gene but they don't have the disease[1]:



In every pregnancy, there is a[1]:

50% chance of your child carrying the disease (without suffering from it).

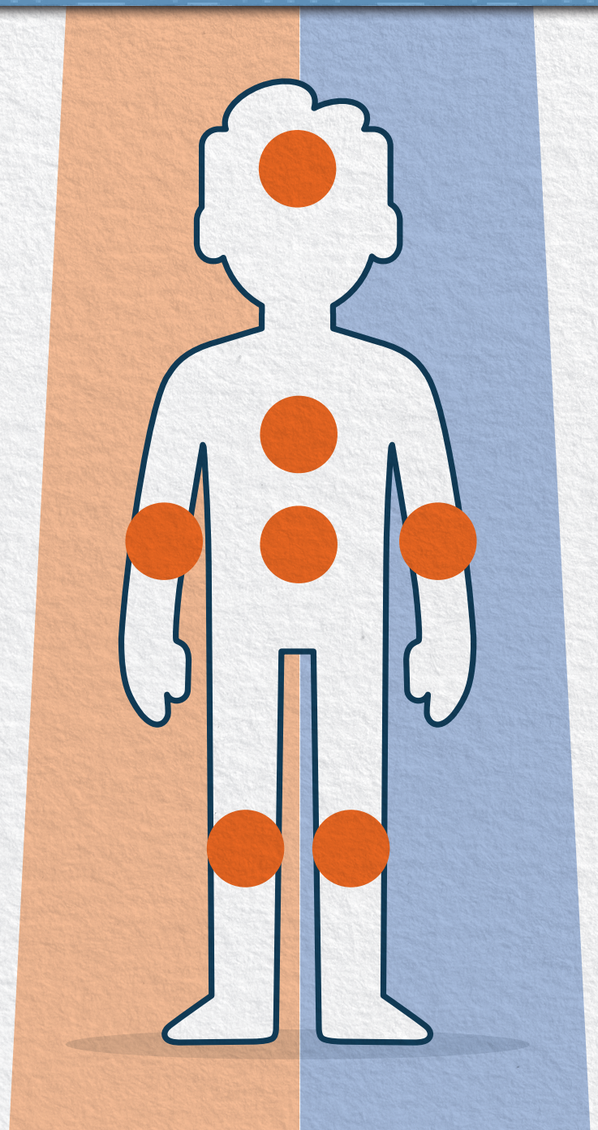
25% chance of being healthy and not carrying the trait (does not inherit the gene from the father or mother).

25% chance of their children having the disease (inheriting both genes of the disease).

Genetic counselling is very important. If the father and mother suffer from the disease, all their children will inherit the disease[1]. If you are planning on having children, seek genetic counselling.

What are the symptoms and complications of SCD?

- **The signs of SCD appear during the first year of a child's life,** at approximately 5 months of age[2]:
  - Crying.
  - Irritability.
  - Yellowing of the skin (jaundice).
  - Swelling of the hands and feet.
- **Symptoms of SCD are different for each person.** Most are related to the complications of the disease.[2]
- The **sickle red blood cells are destroyed faster than normal**. The body may have problems creating enough new cells and replacing those that are lost, causing what is known as **anaemia.** Anaemia results in a state of fatigue, irritability, extreme tiredness and difficulty breathing[2].
- As they are sickle-shaped, sickle red blood cells **cannot flow freely through the blood vessels.** As a result, oxygen does not reach all body tissues and organs, which can cause different types of problems[2].



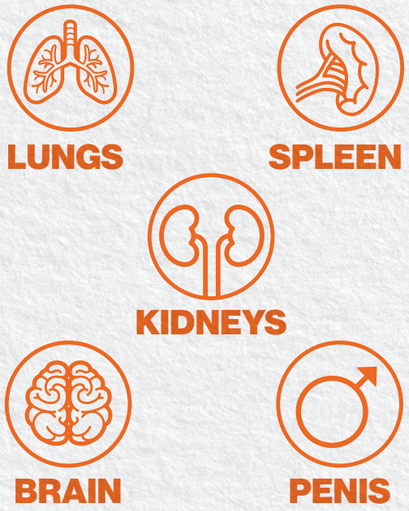
- Blockage of blood flow may cause severe pain, called **pain episodes or vaso-occlusive crises,** one of the most common complications associated with SCD[3]
- The severity, duration and frequency of vaso-occlusive crises varies depending on the person[4].
- **You may feel pain in any part of the body** and possibly in more than one place at the same time. It often occurs in the[4]:
  - Arms and legs.
  - Abdomen.
  - Chest.
  - Hands and feet (most typical in small children).
  - Lumbar region.

It is important to have these crises properly checked and not to suffer with them at home, and seek professional help whenever necessary.

What are the symptoms and complications of SCD?

Vaso-occlusion

DAMAGE TO ORGANS[5]



Other complications that may occur in SCD are as follows[4]:

- Headache or dizziness.
- Painful erections.
- Weakness or difficulty moving some parts of your body.
- Yellowing of the skin or whites of the eyes (jaundice).
- Infections (which mainly affect children).

Throughout your life, SCD can damage the spleen, brain, eyes, lungs, liver, heart and other organs[5].

It is therefore **very important to maintain frequent check-ups with your medical team and comply with the treatments prescribed to you, as well as the recommendations given.**

Advice and recommendations

Paediatric patients with SCD require comprehensive care that includes the usual care of any child in primary care and **specialised multidisciplinary** nursing, haematology, psychology, social workers and other specialist departments[1].

During adolescence, preparations are made for the **transition** to follow-up units of adult patients with SCD[1].

To stay as healthy as possible, make sure you receive regular medical care, maintain a healthy lifestyle and avoid situations that may trigger a pain episode[2].

DON'T FORGET:

- To take measures to **prevent infections[6].**
- **Stick to the general vaccination schedule** and receive the recommended vaccines for patients with SCD to strengthen the immune system and prevent you from getting ill[1].
- **Lack of adherence to treatment** has been associated with a **higher incidence of vaso-occlusive crises** and hospitalisation[1].
- **Treatment and follow-up in symptom-free periods, compliance with medications and attending periodic check-ups are important in order prevent the incidence and decrease the severity of complications[1].**

ALSO[1,4,7,8]:

- Maintain a healthy diet.
- You can exercise regularly, but not intensively. You should also always drink plenty of fluids.
- Always drink plenty of water, especially in cases of fever, vomiting or diarrhoea.
- Keep an eye on your physical tiredness and rest whenever necessary.
- Limit the amount of alcohol you drink.
- Use techniques to reduce stress.
- Wash your hands frequently.

It is very important to follow all the recommendations of your medical team and attend all visits and laboratory tests to ensure optimal management of your condition[1].

Recommendations for preventing and managing SCD and pain crises

- Make and **attend regular appointments with your medical team.** These visits will help to reduce the number of acute problems that require immediate care. Your medical team may help to **prevent complications and improve your quality of life**[5].

- On the other hand, many diverse circumstances have been identified as **trigger factors** for vaso-occlusive crises. **Some precautions may help to reduce these crises, such as avoiding** [1,3,4,7,8]**:**

- Exposure to cold climates or cold water.
- Dehydration.
- Infections.
- Intense physical exercise.
- High altitudes.
- Hypoxia situations (decrease in oxygen levels).
- Smoking.
- Emotional stress.

- To manage the pain, you could try a heating pad, a hot bath or a massage. Physical therapy may also provide some relief[4].

- **Follow your specialist's recommendations** when you have a pain episode. If necessary, go to hospital so you can be treated with stronger painkillers[4].

PAIN EPISODE DIARY

- To control SCD, it is important to monitor **your pain episodes.** You can use a diary like the one you will find at the end of this manual to record your pain episodes, symptoms, triggers and other important information.

- **Remember to share this information with your medical care team.** This will help the healthcare professionals to provide **personalised care** and help **minimise long-term complications.**

More relevant information

You may also find useful information about SCD using the following links:

AEAL, Asociación Española de Afectados por Linfoma [Spanish Association for Lymphoma, Myeloma and Leukaemia Patients]

www.aeal.es

Asociación Española de Pediatría [The Spanish Paediatrics Association]

https://enfamilia.aeped.es/temas-salud/anemia-celulas-falciformes

Grupo Español de Eritropatología [Spanish Erythropathology Group]

https://eritropatologia.com/

MedlinePlus

https://medlineplus.gov/spanish/sicklecelldisease.html

Sociedad Española de Hematología y Oncología Pediátricas [Spanish Society of Paediatric Haematology and Oncology]

www.sehop.org

Sickle Cell Disease (SCD)

https://www.asafefalciforme.org/

Pain episode diary

Write down the month and note the day when the pain episode began and ended.

MONTH _____

1	2	3	4	5	6	7
8	9	10	11	12	13	14
15	16	17	18	19	20	21
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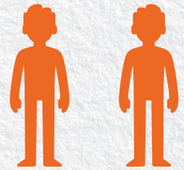
How severe was the pain? Circle the number and description.

1 2 3 4 5 6 7 8 9 10

SLIGHT MILD MODERATE SEVERE EXTREME

Where did you feel it? Circle the affected areas.



FRONT BACK

What were you unable to do during this time? Mark those which apply.

SCHOOL

WORK

SLEEP

CARING FOR FAMILY

OTHER _____

Any other symptoms? Please indicate them.

☐ TIREDNESS ☐ FEVER ☐ SHORTNESS OF BREATH

☐ DIZZINESS ☐ NAUSEA ☐ OTHER SYMPTOMS

Did something happen before? What were the triggers? Mark those which apply.

CHANGE OF TEMPERATURE

WIND

ALTITUDE

STRESS

EXERCISE

DEHYDRATION

INFECTION

NOTHING

OTHER _____

What did you do to relieve the pain?

MEDICATION FOR PAIN

VISITED A&E

APPLIED HEAT

RELAXATION TECHNIQUES

OTHER _____

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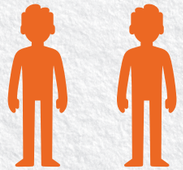

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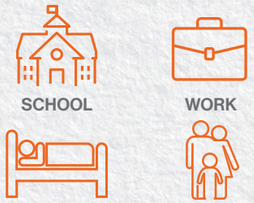

SCHOOL WORK SLEEP CARING FOR FAMILY

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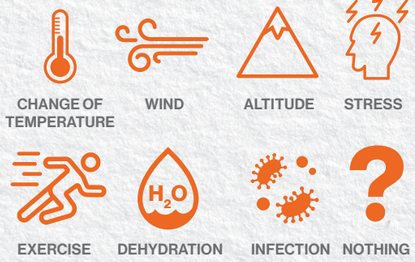

CHANGE OF TEMPERATURE WIND ALTITUDE STRESS

EXERCISE DEHYDRATION INFECTION NOTHING

OTHER _____

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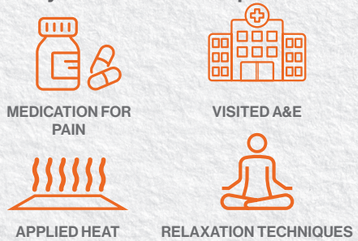

MEDICATION FOR PAIN VISITED A&E APPLIED HEAT RELAXATION TECHNIQUES

OTHER _____

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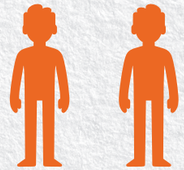

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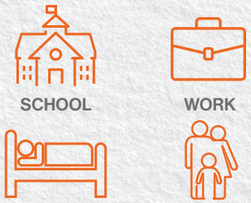

SCHOOL WORK SLEEP CARING FOR FAMILY

OTHER _____

Any other symptoms? Please indicate them.

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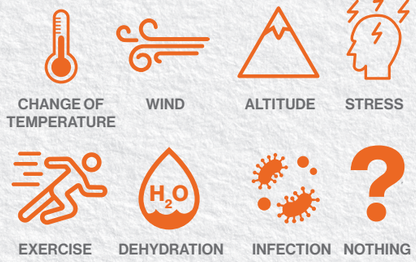
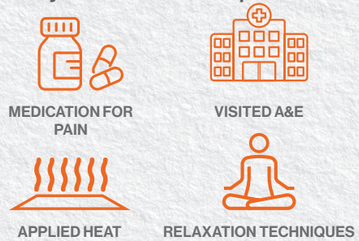


CHANGE OF TEMPERATURE WIND ALTITUDE STRESS

EXERCISE DEHYDRATION INFECTION NOTHING

OTHER _____

What did you do to relieve the pain?



MEDICATION FOR PAIN VISITED A&E APPLIED HEAT RELAXATION TECHNIQUES

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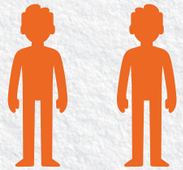

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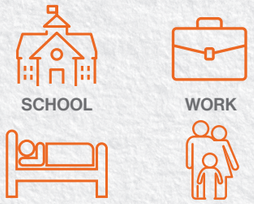

SCHOOL WORK SLEEP CARING FOR FAMILY

OTHER _____

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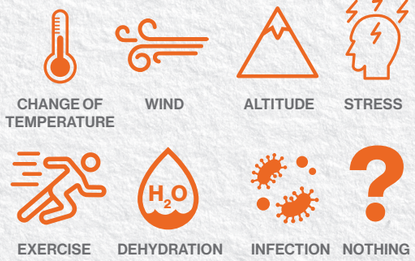
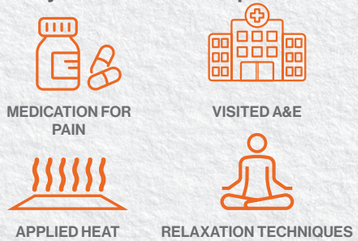


CHANGE OF TEMPERATURE WIND ALTITUDE STRESS

EXERCISE DEHYDRATION INFECTION NOTHING

OTHER _____

What did you do to relieve the pain?



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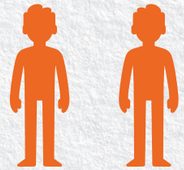

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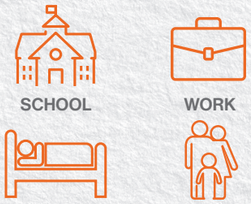

SCHOOL WORK SLEEP CARING FOR FAMILY

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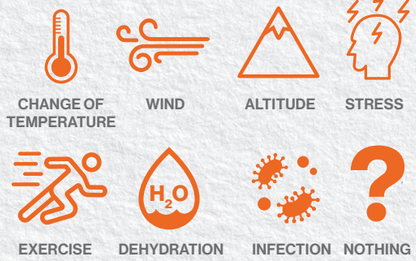

CHANGE OF TEMPERATURE WIND ALTITUDE STRESS

EXERCISE DEHYDRATION INFECTION NOTHING

OTHER _____

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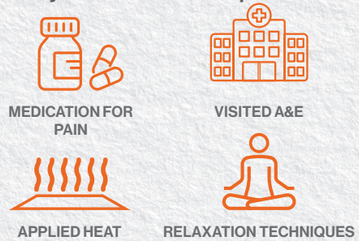

MEDICATION FOR PAIN VISITED A&E APPLIED HEAT RELAXATION TECHNIQUES

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REFERENCES:

1. Sociedad Española de Hematología y Oncología Pediátricas [Spanish Society of Paediatric Haematology and Oncology] Enfermedad de Células Falciformes - Guía de Práctica Clínica. [Sickle cell disease - Clinical Practice Guidelines]. SEHOP 2019. Available at: http://www.sehop.org/wp-content/uploads/2019/03/Gu%C3%ADa-SEHOP-Falciforme-2019.pdf. Last accessed: June 2020.

2. U.S. National Library of Medicine. MedlinePlus. Sickle cell disease [Internet]. 2020. Available at: https://medlineplus.gov/spanish/sicklecelldisease.html. Last accessed: June 2020.

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