



SPARK

SICKLE CELL UNDERSTANDING

A brochure that builds on your knowledge of sickle cell

GIA
*Living With
Sickle Cell*

GINA
Parent

WELCOME

This brochure is designed to build on your foundation of sickle cell knowledge, deepening your understanding of the disease and how it can affect your body.

This brochure can help you:

- **EXPLORE** what sickle cell is
- **UNDERSTAND** the impact hemoglobin Sickle has on the body
- **LEARN MORE** about available treatment options to discuss with your doctor
- **EXPERIENCE** a stronger partnership with your care team

These topics can help to reinforce and build on your sickle cell knowledge, allowing you to **spark more meaningful conversations with your care team.**

Spark a deeper understanding of sickle cell with the latest information, tools, and resources at sparksicklecellchange.com/sign-up

LET'S SPARK CHANGE IN SICKLE CELL

Living with sickle cell takes an **incredible amount of courage and strength.** Whether you have sickle cell or are a parent, caregiver, or ally of someone living with sickle cell, you have the power to learn more and change the way you approach the disease.

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“Having sickle cell has really pushed me to do better. And if I have complications doing it, I may just have to maneuver differently, but I'm still going to make sure I get it done. So that's the biggest thing, **sickle cell has made me stronger as a person.**”

—Cory



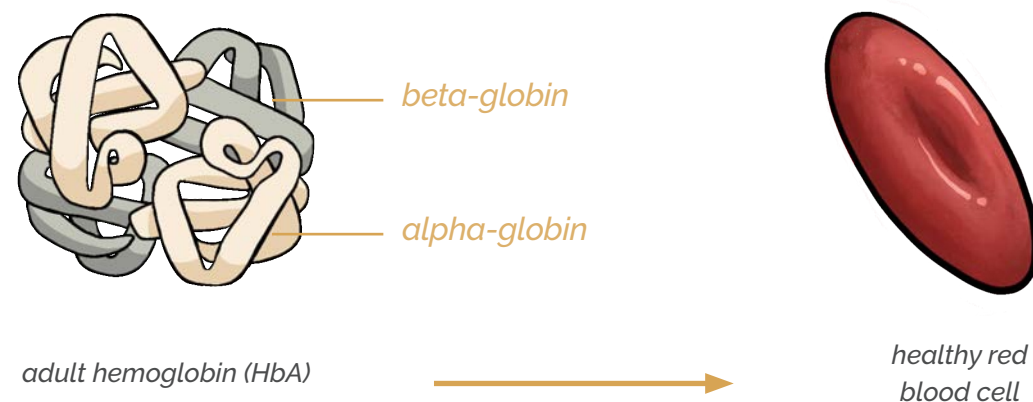
CORY
Living With Sickle Cell

WHAT IS SICKLE CELL?

RED BLOOD CELLS AND HEMOGLOBIN

Red blood cells play an important role in your body. They carry oxygen from the lungs to your organs and move carbon dioxide from your organs back to your lungs. This cycle gives your organs the oxygen they need to function.

Hemoglobin (Hb) is a protein in red blood cells that does the actual work of carrying oxygen from the lungs to the cells and organs throughout your body. When red blood cells are healthy, each cell is densely packed with the adult form of hemoglobin called HbA. **HbA consists of 2 functional alpha (α)-globin and 2 functional beta (β)-globin subunits.** Healthy red blood cells are smooth and disk-shaped, allowing them to move through blood vessels with ease.

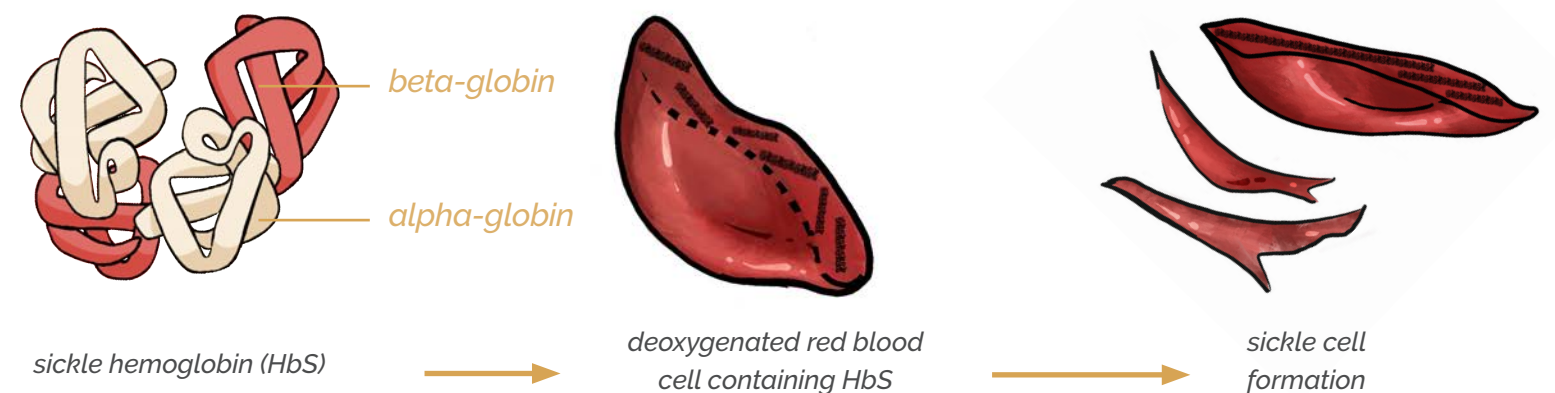


HEMOGLOBIN AND SICKLE CELL

People with sickle cell have a **change (mutation) in both copies of the *HBB* gene** that they inherit from their parents. In people with sickle cell, this mutation affects the beta-globin subunits of the hemoglobin protein. This causes people with sickle cell to produce an abnormal form of hemoglobin called **hemoglobin Sickle (HbS)**.

Unlike HbA molecules, HbS molecules only look and work normally in high-oxygen states, such as when carrying oxygen to your cells and organs. In low-oxygen states, such as after delivering oxygen, HbS molecules change shape (undergo a conformational change) and stick together. When there are many HbS molecules with a change in shape, they clump together and form long **chains of polymers**, in a process called **polymerization**. These clumps (polymers) of HbS molecules dehydrate and distort red blood cells, giving them the characteristic sickle shape.

Sickled red blood cells are fragile, rigid, and sticky. It can be difficult for them to move through small blood vessels, resulting in slowed or blocked blood flow through these vessels. This can prevent oxygen from circulating properly throughout the body. Sickled cells are also fragile and can break down prematurely, lasting only 10 to 20 days compared with 90-120 days for healthy red blood cells.



A person who inherits **1 copy of the sickle *HBB* gene and 1 copy of a normal *HBB* gene** has **sickle cell trait**. They can experience some of the symptoms of sickle cell under extreme circumstances.

People with sickle cell trait can pass it along to their children. When both parents carry the sickle cell trait, there is a **1 in 4 chance in each pregnancy of having a child with sickle cell disease**. A simple blood test from your doctor can determine if you are a carrier.

HOW SICKLE CELL AFFECTS YOUR BODY

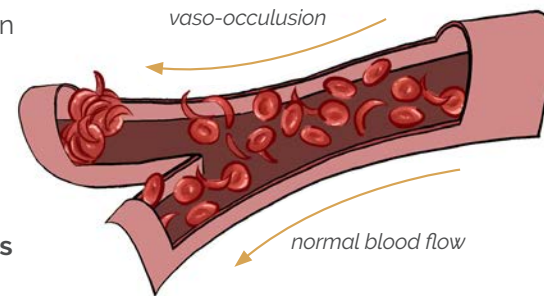
Sickle cell affects everyone differently. People with sickle cell may experience symptoms and complications differently, and these can change over time.

ACUTE COMPLICATIONS

In people with sickle cell, sickled-shaped red blood cells can block or slow blood flow. This blockage, known as a **vaso-occlusion**, can prevent organs and tissues from getting the oxygen they need and can cause **vaso-occlusive crises (VOCs)**.

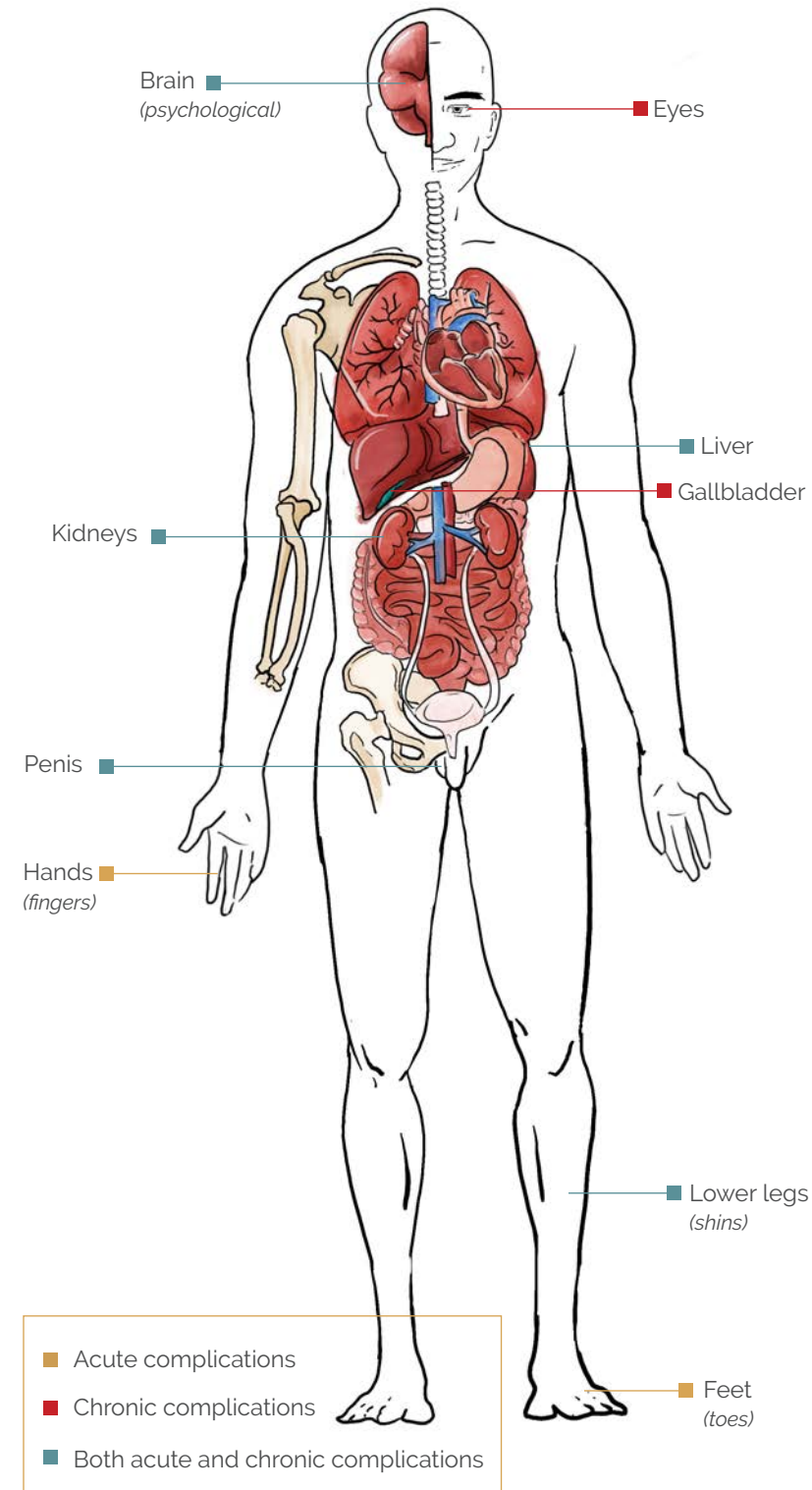
VOCs include pain crises, acute chest syndrome, and stroke—these can cause excruciating pain, as well as severe cardiovascular, lung, and infection issues that may require an emergency room visit. Acute pain crises are the most common complication of sickle cell, but all acute complications can have a significant impact on your organs and your overall health.

- **Acute pain crises**
- **Acute chest syndrome (ACS)**, lungs
- **Infections**, spleen
- **Liver complications (hepatic sequestration)**, liver
- **Prolonged painful erections (priapism)**, penis
- **Spleen complications (acute splenic sequestration)**, spleen
- **Stroke (overt and silent)**, brain



While acute complications may occur suddenly and resolve quickly, the underlying damage they cause may continue. The blockages and damage from sickled blood cells that cause acute complications can cause organ damage and eventually progress to organ failure, and sometimes cause early or sudden death. Recognizing the impact acute complications can have on your health now and in the future can help you and your healthcare team create a plan to better monitor, identify, and manage any complications over time.

Spark a deeper understanding of the acute and chronic complications associated with sickle cell at sparksicklecellchange.com/what-is-sickle-cell/symptoms-complications



CHRONIC COMPLICATIONS

Even when you are not in pain, ongoing vaso-occlusion can lead to chronic complications and can cause **organ damage or even organ failure**. Chronic complications of sickle cell may include:

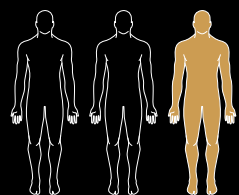
- **Anemia (chronic hemolytic anemia)**
- **Bone complications (avascular aseptic necrosis)**
- **Chronic pain**
- **Delayed puberty**
- **Fatigue**
- **Gallstones**, gallbladder
- **Dactylitis**, pain crisis in hands and feet
- **Jaundice**, skin and eyes
- **Kidney problems**, kidneys
- **Leg sores (ulcers)**, legs
- **Organ failure**
- **Vision problems (retinopathy)**, eyes

Chronic complications are a significant cause of illness in adults with sickle cell. Because chronic complications may go unnoticed and can cause irreversible organ damage, **it is important to routinely talk to your doctor about treatment options and a treatment plan that is right for your sickle cell.**

FOCUSING ON MENTAL HEALTH

Acute and chronic complications can have an emotional effect on people with sickle cell. Frequent pain, hospitalizations, and absence from school or work can cause people with sickle cell to have feelings of low self-esteem and hopelessness. In a study done in 2017, over one-third of adults with sickle cell had experienced depression and/or anxiety because of the disease.

Spark a conversation with your healthcare team to make sure you have the support you need. This may include adding a psychiatrist, social worker, or other mental health professional to your team.



1 in every 3 people with sickle cell experience depression and/or anxiety



“To be open and transparent is tough. To be vulnerable is tough. To show your weakness is tough. There’s a proverb that says that if you want to go fast, go alone. If you want to go far, go together.

And the only way that we as people can address sickle cell is if we go together.”

—Jimmie, Living With Sickle Cell



REV JIMMIE
Living With Sickle Cell

TREATING SICKLE CELL

Learning about the current treatment options can help you and your loved ones create a comprehensive care plan with your doctor to manage your sickle cell.

Current therapies mainly focus on managing the symptoms of sickle cell and require lifelong use. Recent progress in sickle cell has led to more treatment options that have the potential to help manage acute and chronic symptoms of the disease. **Treatment options may include:**



• **SICKLE CELL TREATMENTS** can help reduce the frequency of pain crises and hospitalizations or reduce the number of blood transfusions a person with sickle cell needs. A list for other supportive therapies includes, but is not limited to: antibiotics, vitamins, vaccinations, and IV and oral hydration



• **MEDICATIONS TO MANAGE PAIN** can help people with sickle cell manage acute and chronic pain



• **BLOOD TRANSFUSIONS** can help relieve symptoms of sickle cell and potentially prevent stroke in high-risk patients but can also lead to iron overload and other serious complications



• **HEMATOPOIETIC STEM CELL TRANSPLANT (HSCT)** can cure sickle cell using the stem cells of a person who doesn't have the sickle *HBB* gene mutation, but is only available to people who are under the age of 18 and have a matched related donor available

General health maintenance and preventative care:

- Drink plenty of water. Avoid getting too hot or too cold. Avoid exposure to high altitudes like flying or mountain climbing. Avoid situations where you may be exposed to low oxygen levels like exercising really hard such as training for an athletic competition or attending military boot camp
- Stay up to date on vaccinations
- Wash your hands frequently to prevent infections
- Attend regular checkups with your healthcare team

Significant strides have been made in the treatment of sickle cell but it is not enough; serious medical and social burdens remain. There is still a need for treatments that can eliminate lifelong symptom management and its unwanted side effects.*

*Nothing contained within this brochure should be considered medical advice.

GETTING THE SUPPORT THAT YOU NEED

Living with sickle cell can be challenging but it's important to **remember that you're not alone**. Beyond your family and friends, your care team and the sickle cell community are there to support you and to help you get the care you need and deserve.

WORKING WITH YOUR HEALTHCARE TEAM

Because sickle cell can affect every organ in your body, your care may require the help of a group of specialists who meet your individual needs. Your healthcare team is responsible for providing comprehensive care that monitors and manages all of your symptoms (not just pain) and addresses the acute and chronic complications before they progress. Building a healthcare team requires a strong partnership between you and all of the specialists who are involved in your care. **Here are some ways that you can work with your care team to build a strong partnership:**

- Be open and honest
- Start your visit by telling your doctor what you'd like to discuss (mention your top 2 or 3 issues)
- Take notes on what your doctor says and confirm next steps
- Learn how to access your medical records
- Keep notes of healthcare issues between appointments to bring to future visits
- Ask questions until you understand**

Taking an active role in your care is important. No one knows your body better than you do. If you believe something has changed or is wrong, you should reach out to your healthcare team right away. This can give your healthcare team a better opportunity to manage symptoms and complications.

ENGAGING WITH THE SICKLE CELL COMMUNITY

In addition to your care team, there is an entire sickle cell community, including patient advocacy, community-based organizations, and support groups, which are here to help. **You can find a list of organizations that can provide you with ongoing support and resources at sparksicklecellchange.com.**

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“I started researching, where can we go to an event? Where can he meet someone, or more kids, or more people like him to make him feel like he's not alone? And so I found an organization that was holding a sickle cell event in our area.”

—Crystal, Caregiver to Joey, Who Is Living With Sickle Cell



Spark your engagement with the sickle cell community at sparksicklecellchange.com/community-organizations

JOEY
Living With Sickle Cell

THE ORTIZ AND RIVAS FAMILY



Spark change in sickle cell by visiting
sparksicklecellchange.com

A heartfelt thanks to the patients, families, allies, and sickle cell community who have helped make Spark possible.

We hope their experience helps to inspire you and your family as we continue to Spark change in sickle cell.

The people featured in this brochure have been compensated for their time by bluebird bio.

