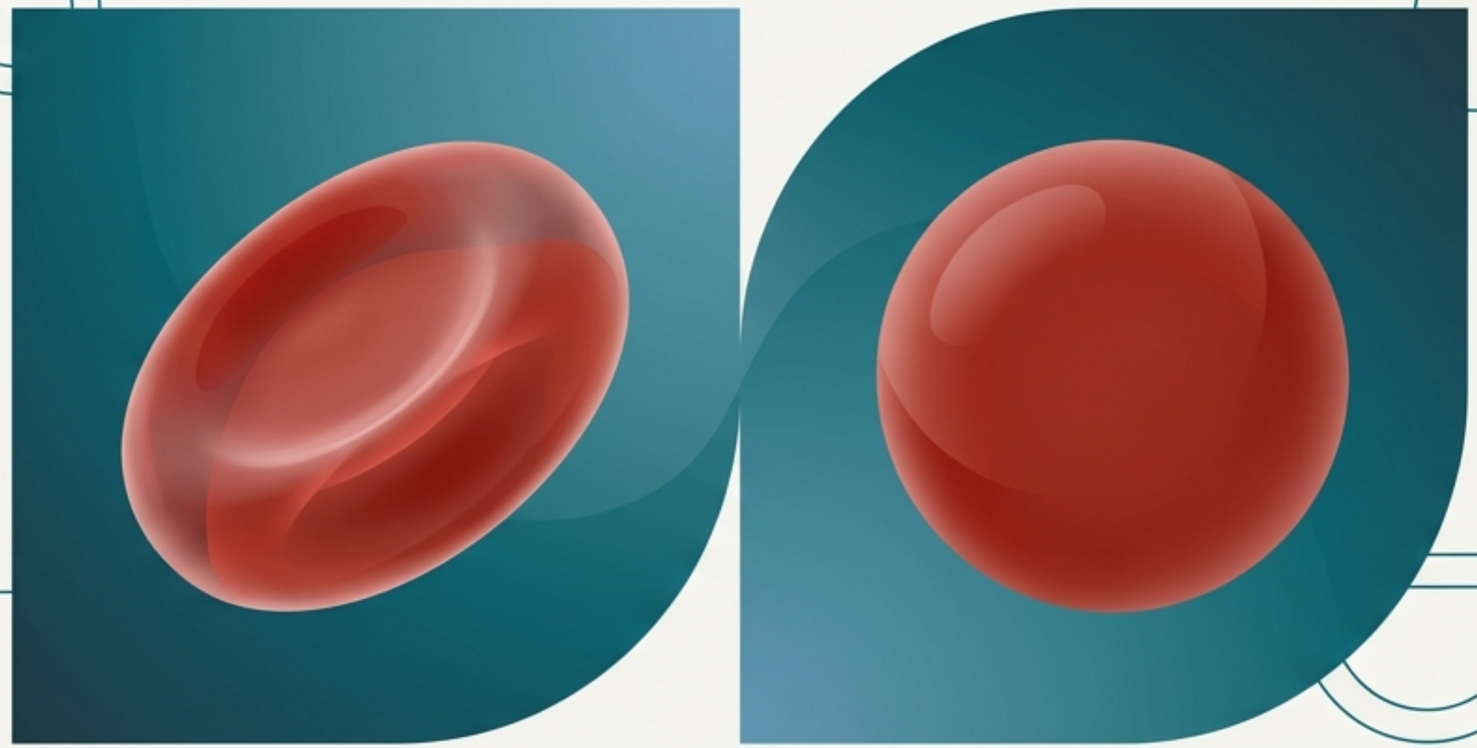
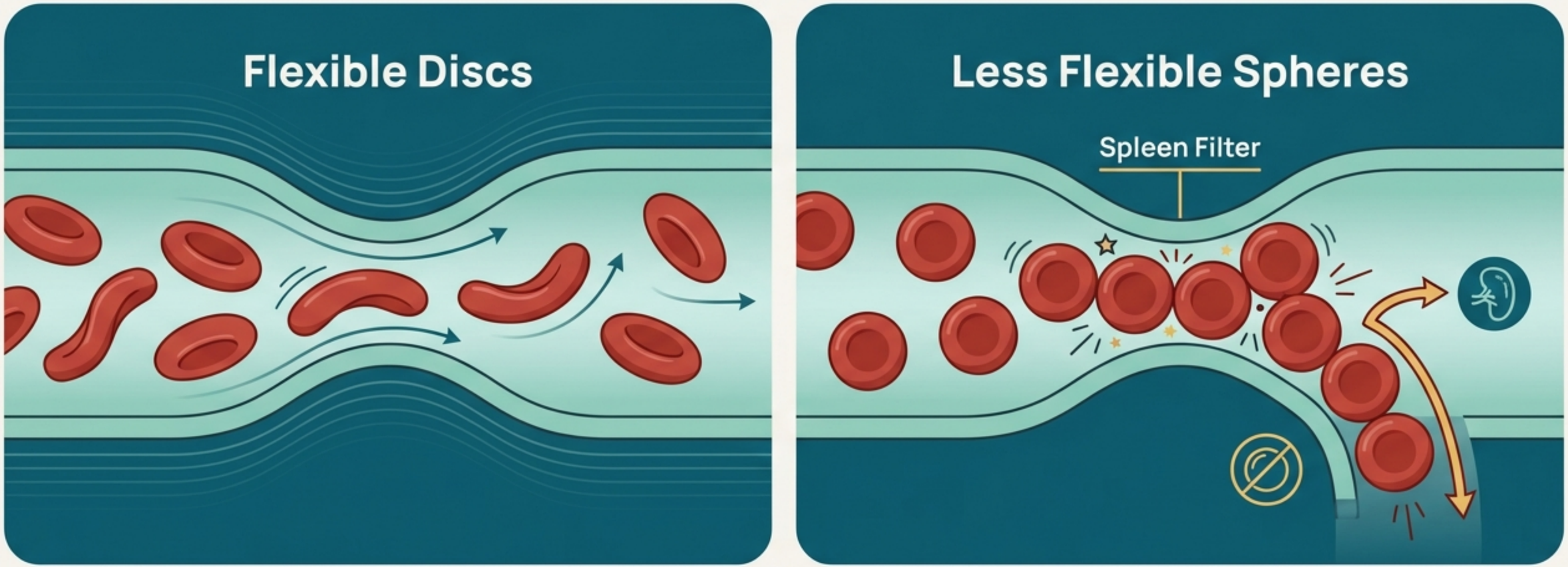
Understanding Hereditary Spherocytosis

A Guide to Your Health and Well-being



This guide is designed to help you understand Hereditary Spherocytosis (HS). Most people with HS have a stable condition and lead full, active lives with the right monitoring and care. Our goal is to help you understand your pattern and support you over time.

It All Comes Down to Shape



Think of your red blood cells as **flexible disks** that easily **bend to travel** through the body's tiny pathways. In Hereditary Spherocytosis, the cells become **round** and **less flexible**. Because of this shape, the **spleen**—your body's blood filter—removes them from circulation sooner than it should.

What is Hereditary Spherocytosis?

Genetic Condition: HS is a genetic condition affecting the membrane, or outer shell, of your red blood cells (RBCs).

Altered Shape: Instead of flexible discs, the cells become rounder ‘spherocytes’ that are less able to squeeze through small spaces.

The Spleen’s Role: Your spleen recognizes these cells as abnormal and removes them from the bloodstream earlier than usual.

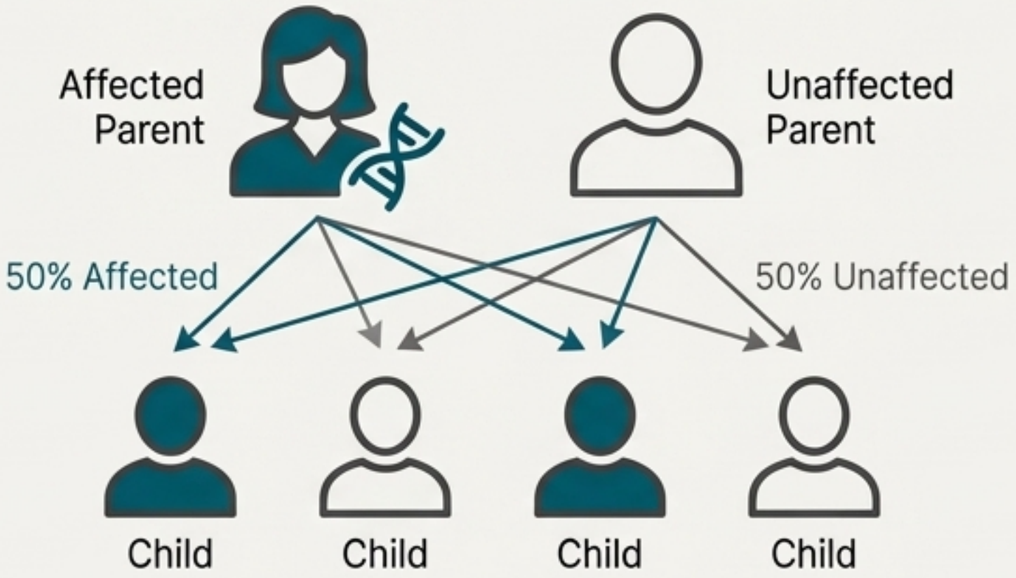
Result: Hemolysis: This process of ongoing red blood cell breakdown is called hemolysis.

Why It Happens: A Look at the Genetics

HS is caused by changes in the genes that create the protein scaffolding for the red blood cell membrane. These proteins include spectrin, ankyrin, and others.

How It's Inherited

Most people inherit HS in an **autosomal dominant** pattern. This means a child has a 50% chance of inheriting the condition if one parent has it.



New Mutations

In some cases, the genetic change appears for the first time in a person with no family history of HS. This is called a ***de novo*** mutation.

HS is Different for Everyone

Mild HS

Minimal or no symptoms. Hemoglobin levels are normal or near-normal.

Moderate HS

Noticeable anemia and jaundice (yellowing of the skin/eyes). Increased risk of developing gallstones.

Severe HS

Significant anemia that may require blood transfusions. Spleen removal is often considered as a treatment.

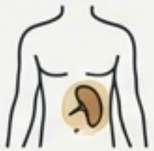
Common Signs and Symptoms

Fatigue or low energy

Pale skin (pallor)

Yellowing of the eyes or skin (jaundice)

Darker-than-usual urine



Abdominal discomfort or feeling full early (from an enlarged spleen)

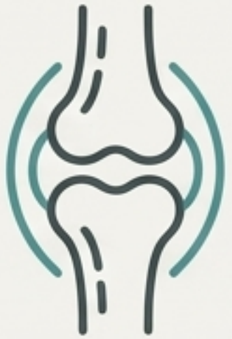
During Illness: Episodes of worsening anemia.

Possible Gallstones: Abdominal pain (especially after fatty foods), nausea, or vomiting.

In newborns, HS can cause jaundice that is more noticeable or lasts longer than typical newborn jaundice. Close monitoring is key.

Understanding and Watching for Complications

Most people with HS do well with regular care. However, it's important to be aware of a few potential complications.



Aplastic Crisis

When bone marrow temporarily stops making RBCs, often triggered by a virus. **Urgent medical evaluation is needed** for sudden fatigue, extreme paleness, or shortness of breath.

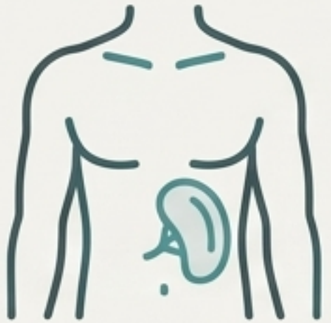
Hemolytic Crisis

When RBCs break down faster than usual, often during an infection.

Look for increased jaundice, darker urine, or worsening anemia.

Gallstones

Caused by high levels of bilirubin from RBC breakdown. Report any abdominal pain, nausea, or vomiting to your doctor.



Enlarged Spleen

The spleen can grow from overwork. This may cause discomfort. Your doctor may advise avoiding certain high-impact activities.

How Your Doctor Confirms the Diagnosis

Family History: A review of your family's health history often provides the first clue.

Complete Blood Count (CBC): Measures hemoglobin levels and other blood components to assess anemia.

Blood Smear: A microscopic look at your blood to identify the presence of round spherocytes.

Markers of Hemolysis: Blood tests for bilirubin and reticulocyte count show how fast RBCs are breaking down and being replaced.

Specialized Tests: Tests like EMA binding or osmotic fragility can confirm the diagnosis.

Abdominal Ultrasound: Used to check the size of the spleen and look for gallstones.

A Note on Genetic Testing

Most people are diagnosed based on their blood tests and clinical features alone. Genetic testing is not always required.

When Genetic Testing May Be Used

- If the diagnosis is unclear after other tests.

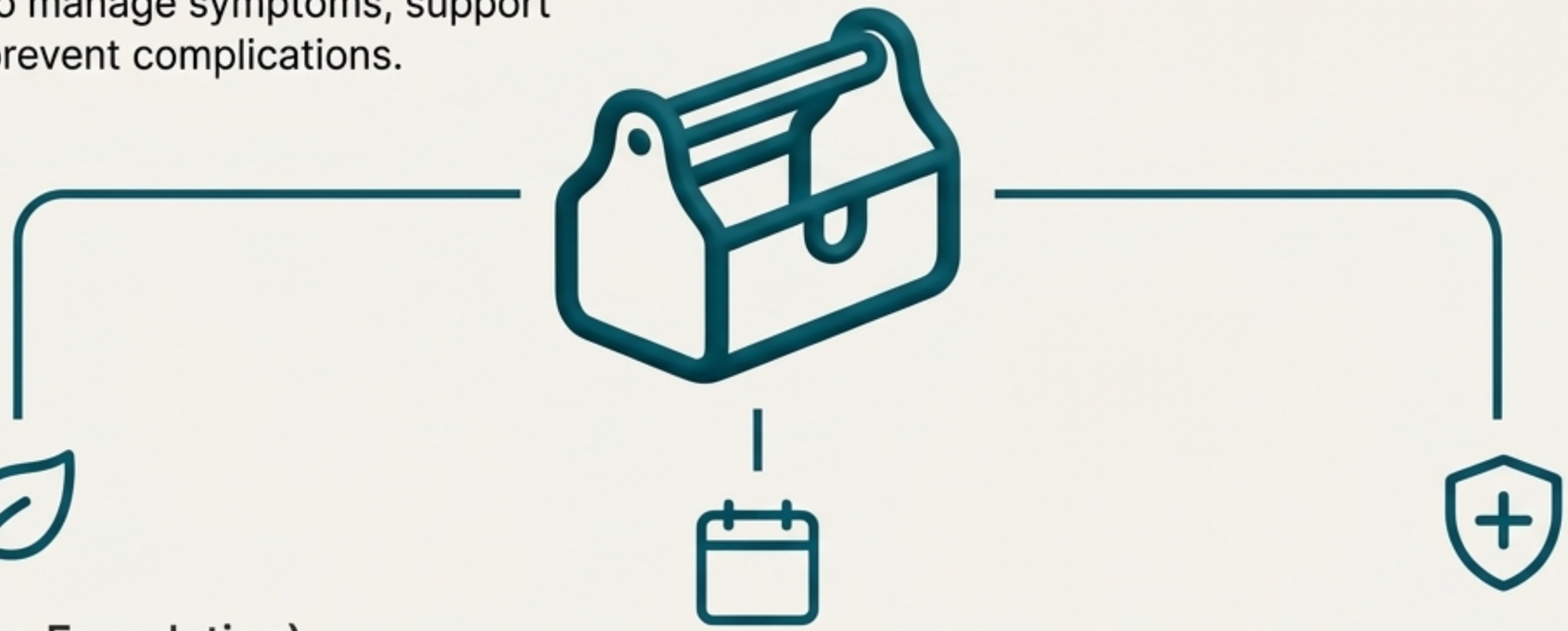
- When symptoms begin very early in life.

- If there is no known family history of HS.

- For families who want detailed information for future planning.

Your Management Toolkit: A Partnership in Care

Treatment is tailored to the severity of your HS. The goal is to manage symptoms, support your body, and prevent complications.



Folic Acid (The Foundation)

Because your bone marrow works harder to replace RBCs, folic acid is essential to support this increased production. Most people take a daily supplement.

Monitoring & Follow-Up

Regular visits with your doctor are key to tracking your health and catching any issues early.

Surgical Options

For more significant symptoms, other treatments like splenectomy may be considered. We'll explore these next.

Considering Splenectomy

Why is it an option?

Removing the spleen (splenectomy) is the most effective way to reduce the destruction of red blood cells. This can improve anemia, reduce jaundice, and increase energy levels.

Who is it for?

It is typically recommended for those with **moderate to severe HS** whose symptoms significantly affect daily life.

Types of Splenectomy

Full Splenectomy

The entire spleen is removed.

Partial Splenectomy

In some children, removing only part of the spleen can reduce symptoms while preserving some of its immune function.

Managing Gallstones

The Connection

The rapid breakdown of red blood cells increases a substance called bilirubin in your body. Over time, this can lead to the formation of "pigment gallstones" in the gallbladder.

Symptoms to Watch For

- Abdominal pain, particularly after eating fatty foods
- Nausea or vomiting

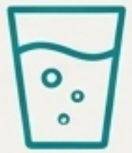
Treatment

If gallstones cause pain or infection, the gallbladder may need to be surgically removed. Sometimes this is done at the same time as a splenectomy.

Living Well with HS: Your Daily Plan

Most people with HS live normal, active lives.
Consistent self-care is your strongest tool.

Take Folic Acid Consistently: As prescribed by your doctor.



Stay Hydrated: Good hydration is always important for your health.

Attend Regular Follow-ups: Stay connected with your medical team.

Report New Symptoms: Keep your doctor informed about any changes.

Know Your Body: Pay attention to your energy levels and listen to what your body needs.

When to Contact Your Medical Team

Contact Your Doctor If You Notice...

- Worsening fatigue or unusual paleness
- Increasing yellowing of skin or eyes
- New or persistent abdominal pain
- Very dark urine

Seek Urgent Care For...

- Severe weakness or difficulty catching your breath
- Fainting or dizziness
- Sudden, severe abdominal pain

Special Alert: If you have had a splenectomy, **a fever is always an urgent concern** and should be evaluated by a medical professional promptly.

The Outlook: A Lifelong but Manageable Journey

Hereditary Spherocytosis is a lifelong condition, but it is highly manageable. Long-term follow-up with your care team is the key to preventing complications and living a full, healthy life.

- HS makes red blood cells round and fragile, causing them to break down sooner.
- Most cases are mild to moderate, and an active life is the norm with routine care.
- Anemia and jaundice are common, especially during illness.
- Be mindful of complications like gallstones and report abdominal pain.
- Splenectomy is an effective option for more severe symptoms.
- Regular follow-up is your most important tool for long-term wellness.