



# UNDERSTANDING HEREDITARY HEMOCHROMATOSIS

*A brief guide for patients with HFE-related iron overload*

Hereditary hemochromatosis is common and very treatable.  
Most people diagnosed early remain healthy with simple monitoring and phlebotomy.

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## What It Is

Hereditary hemochromatosis is a genetic condition that causes the body to absorb more iron from food than it needs.

It is most often due to changes in the **HFE gene**, usually the **C282Y variant** inherited from both parents. Over time, excess iron can build up in organs unless it is removed.

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## Symptoms

Many people have **no symptoms**, especially early on.

Possible symptoms include:

- fatigue or weakness
- joint pain
- abdominal discomfort
- decreased libido or erectile dysfunction
- bronze or gray skin color
- symptoms of diabetes
- heart rhythm problems

If untreated over many years, excess iron can damage organs such as the liver, heart, and pancreas.

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## Diagnosis

Hereditary hemochromatosis is often discovered during routine blood testing.

Key tests include:

- **Transferrin saturation (TSAT)** greater than 45%
- **Ferritin** above the usual range
- **HFE genetic testing**

Additional evaluation may include liver enzyme tests or imaging, depending on iron levels and other risk factors.

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## Is It Dangerous?

**Usually not**, especially when detected and treated early.

Risk is higher:

- ferritin is very high (often above 1,000 ng/mL)
- there is heavy alcohol use or underlying fatty liver disease

With treatment, most people never develop organ damage.

## What Is the Treatment?

The main treatment is **phlebotomy**, which safely removes iron by removing blood.

- **Iron-reduction phase:** weekly or every 1–2 weeks until iron levels fall into the target range
- **Maintenance phase:** phlebotomy a few times per year to keep levels stable
- **Target ferritin:** 50–100 ng/mL

Iron-binding medications (chelation) are used only when phlebotomy is not possible.

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## Self-Care

- avoid **iron supplements**
  - avoid **high-dose vitamin C supplements**
  - limit alcohol
  - avoid **raw shellfish** (risk of serious infection)
  - a strict low-iron diet is **not** required
  - maintain a healthy, balanced diet
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## When to Call Your Doctor

Contact your doctor if you notice:

- worsening fatigue or joint pain
- abdominal swelling
- darkening skin
- symptoms of diabetes
- changes in menstrual cycles or erectile function

**Seek urgent care** for vomiting blood, black stools, severe abdominal pain, jaundice, chest pain, or confusion.

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## Plan Going Forward

Most people need:

- periodic ferritin and TSAT testing
- scheduled phlebotomy
- routine liver monitoring.

**Family screening:** Parents, siblings, and adult children should discuss testing with their doctor.

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## Key Points to Remember

- **highly treatable:** most people live normal, healthy lives with monitoring and phlebotomy
- **early detection matters:** treatment started early helps prevent organ damage
- **phlebotomy works well:** removing small amounts of blood safely lowers iron levels
- **simple self-care helps:** avoid iron supplements, high-dose vitamin C, and raw shellfish
- **family screening is important:** relatives may benefit from testing and early care