

Iron Reign Research

Hereditary Hemochromatosis Diagnostic Guide

This educational guide provides a visual overview of hereditary hemochromatosis (HFE-related iron overload), including the major blood tests and genetic tests commonly used to help determine risk and diagnosis.



DO I HAVE HEREDITARY HEMOCHROMATOSIS?

A STEP-BY-STEP GUIDE TO FIND OUT

Hereditary hemochromatosis (HH) is a genetic disorder that causes increased iron absorption and iron buildup in the body.



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KNOW THE RISK FACTORS



- Family history of HH
- Northern European ancestry
- Symptoms of iron overload (fatigue, joint pain, abdominal pain, skin bronzing, diabetes, heart issues, low libido, etc.)
- Unexplained liver problems

YOU MAY BE AT RISK IF YOU:



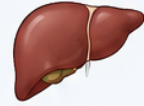
Have a close relative with HH



Are of Northern European descent



Have symptoms of iron overload



Have unexplained liver problems

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GET INITIAL BLOOD TESTS



These tests show how much iron is in your body.

- Serum Ferritin – shows iron stores
- Transferrin Saturation (TSAT) – shows iron in the blood
- Serum Iron
- Total Iron Binding Capacity (TIBC)

INTERPRETING THE RESULTS

Test	Normal Range (Typical)	Suggests Possible Iron Overload When:
Serum Ferritin	30 – 300 ng/mL (men) 15 – 150 ng/mL (women)	> 300 ng/mL (men) > 200 ng/mL (women)
Transferrin Saturation	20 – 45%	> 45%
Serum Iron	60 – 170 µg/dL	Elevated
TIBC	250 – 450 µg/dL	Low or normal

✓ If your ferritin is elevated and transferrin saturation is >45%, further testing is recommended.

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GET GENETIC TESTING



A simple blood or saliva test looks for mutations in the *HFE* gene.

The two most common mutations are:

- C282Y
- H63D

POSSIBLE GENETIC RESULTS

No Mutations
(C282Y-/H63D-)



Unlikely to have HH

One Mutation
(C282Y+/- or H63D+/-)



Carrier – usually does not cause iron overload

Two Mutations
(C282Y+/C282Y+)



High risk for HH and iron overload

Two Mutations
(C282Y+/H63D+) or (H63D+/H63D+)



Possible risk – may cause iron overload, especially with other factors

! Genetic results should always be interpreted with your blood test results and clinical picture.

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CONFIRM THE DIAGNOSIS



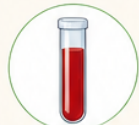
Your healthcare provider will consider:

- Blood test results
- Genetic test results
- Symptoms and medical history
- Physical exam

Sometimes additional tests are needed:

- Liver function tests
- Liver MRI (to measure iron)
- Liver biopsy (rarely needed)

DIAGNOSIS IS MOST LIKELY IF YOU HAVE:



High ferritin



High transferrin saturation (>45%)



Two *HFE* gene mutations (at risk genotype)



Signs or symptoms of iron overload or organ involvement

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ASSESS SEVERITY & ORGAN INVOLVEMENT



Iron can affect multiple organs.

Your doctor may check:

- Liver
- Heart
- Pancreas
- Joints
- Endocrine system (hormones)

POSSIBLE TESTS



Liver function blood tests



MRI for liver iron



Echocardiogram (heart)



Blood sugar (diabetes risk)



Hormone tests

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NEXT STEPS



- If diagnosed early, treatment can prevent complications.
- Main treatment is therapeutic phlebotomy (regular blood removal).
- Avoid iron supplements and excess vitamin C.
- Limit alcohol to protect your liver.

WORK WITH YOUR HEALTHCARE PROVIDER



Discuss results and options



Follow a treatment plan if needed



Monitor iron levels regularly



Maintain a healthy lifestyle



Consider testing at-risk family members



KEY TAKEAWAY: A combination of blood tests, genetic testing, and clinical evaluation determines if you have hereditary hemochromatosis.



This infographic is for educational purposes only and does not replace professional medical advice. Consult your healthcare provider for personalized evaluation and recommendations.

