

## Screening for Hemochromatosis: Recommendations from the U.S. Preventive Services Task Force

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The full reports are titled “Screening for Hemochromatosis: Recommendation Statement” and “Screening for Hereditary Hemochromatosis: A Systematic Review for the U.S. Preventive Services Task Force.” They are in the 1 August 2006 issue of *Annals of Internal Medicine* (volume 145, pages 204-208 and 209-223). The first report was written by the U.S. Preventive Services Task Force; the second report was written by E.P. Whitlock, B.A. Garlitz, E.L. Harris, T.L. Beil, and P.R. Smith.

### Who developed these guidelines?

The U.S. Preventive Services Task Force (USPSTF) is a group of health experts that reviews published research and makes recommendations about preventive health care.

### What is the problem and what is known about it so far?

Hereditary hemochromatosis is a disease in which an abnormal gene causes the body to absorb too much iron from food. Iron builds up in body organs, which can lead to serious liver damage (cirrhosis), diabetes, heart failure, arthritis, and skin discoloration. Most people do not know that they have hemochromatosis until organ damage occurs. If the condition is identified early, organ damage can be prevented or slowed by periodic removal of blood with the same procedure used during blood donation. Tests for hemochromatosis include blood tests that indicate body levels of iron (transferrin saturation and ferritin level). Another test looks for the most common gene abnormality in hemochromatosis.

People disagree about whether routine medical care should include screening for hemochromatosis. Screening means looking for the disease in people who have no symptoms. Screening is different from diagnosis, which refers to testing people who have symptoms of a disease. Knowing about hereditary hemochromatosis early enough might help to prevent organ damage. However, not all people who have the genetic abnormality develop organ damage. The risks of screening include complications of further screening in those who screen positive, unnecessary blood removal, and giving people who may never get sick the label “hemochromatosis.” Such labeling might cause anxiety or problems with health insurance.

### How did the USPSTF develop this recommendation?

The USPSTF reviewed published studies to decide whether there was good evidence that the benefits of screening for hemochromatosis outweigh the risks. The focus was screening for the gene associated with hereditary hemochromatosis.

### What did the authors find?

No studies directly measured the risks and benefits of hemochromatosis screening with genetic tests. The authors found fair evidence that disease due to hereditary hemochromatosis is rare in the general population. Only a small proportion of people with the hemochromatosis gene actually develop the disease. No studies clearly identified the benefits of routinely removing blood from people without symptoms who had hemochromatosis. Screening would identify a large number of people who have the high-risk gene but may never develop illness. This could result in unnecessary tests and procedures and anxiety.

### What does the USPSTF suggest that patients and doctors do?

The USPSTF recommends that doctors do not routinely do genetic testing to screen patients with no symptoms of hemochromatosis for the disease.

### What are the cautions related to this recommendation?

This recommendation could change as more information becomes available about the benefits and risks of screening for hemochromatosis. This recommendation does not apply to patients who have symptoms that might be explained by hemochromatosis or to patients with family members with the disease.

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