

Screening for Hereditary Hemochromatosis: Recommendations from the American College of Physicians

Summaries for Patients are a service provided by *Annals* to help patients better understand the complicated and often mystifying language of modern medicine.

The full reports are titled “Screening for Hereditary Hemochromatosis: A Clinical Practice Guideline from the American College of Physicians” and “Screening Primary Care Patients for Hereditary Hemochromatosis with Transferrin Saturation and Serum Ferritin Level: Systematic Review for the American College of Physicians.” They are in the 4 October 2005 issue of *Annals of Internal Medicine* (volume 143, pages 517-521 and pages 522-536, respectively). The first report was written by A. Qaseem, M. Aronson, N. Fitterman, V. Snow, K.B. Weiss, and D. Owens, for the Clinical Efficacy Assessment Subcommittee of the American College of Physicians; the second report was written by B. Schmitt, R.M. Golub, and R. Green.

Who developed these guidelines?

The American College of Physicians (ACP) developed these recommendations. Members of ACP are internal medicine doctors (internists), specialists in the care of adults.

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 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 ACP develop these recommendations?

Authors reviewed published studies to see whether the benefits of screening for hemochromatosis outweigh the risks. They looked for studies that addressed the following issues: 1) frequency of hemochromatosis, 2) risk for organ damage in patients with hemochromatosis but no symptoms, 3) usefulness of hemochromatosis tests, 4) effectiveness of early treatment of hemochromatosis in preventing organ damage, and 5) risks of screening and early treatment.

What did the authors find?

No studies directly measured the risks and benefits of hemochromatosis screening. About 1 of every 169 to 556 patients presenting for general medical care have hemochromatosis. The frequency of organ damage in patients without symptoms but with the abnormal gene is unknown. No studies clearly identified the benefits of routinely removing blood from people without symptoms who had hemochromatosis.

What does the ACP suggest that patients and doctors do?

Because existing information does not clearly identify the risks and benefits of screening, whether to look for the disease in people who have no symptoms is unknown. If doctors think a patient might have hemochromatosis, they should make a diagnosis by using blood tests to measure ferritin level and transferrin saturation. If these test results are abnormal, the patient and doctor should discuss the risks and benefits of genetic testing. Patients with family members with hemochromatosis should discuss the risks and benefits of screening with their doctors.

What are the cautions related to these recommendations?

These recommendations could change as more information becomes available about the benefits and risks of screening for hemochromatosis.

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