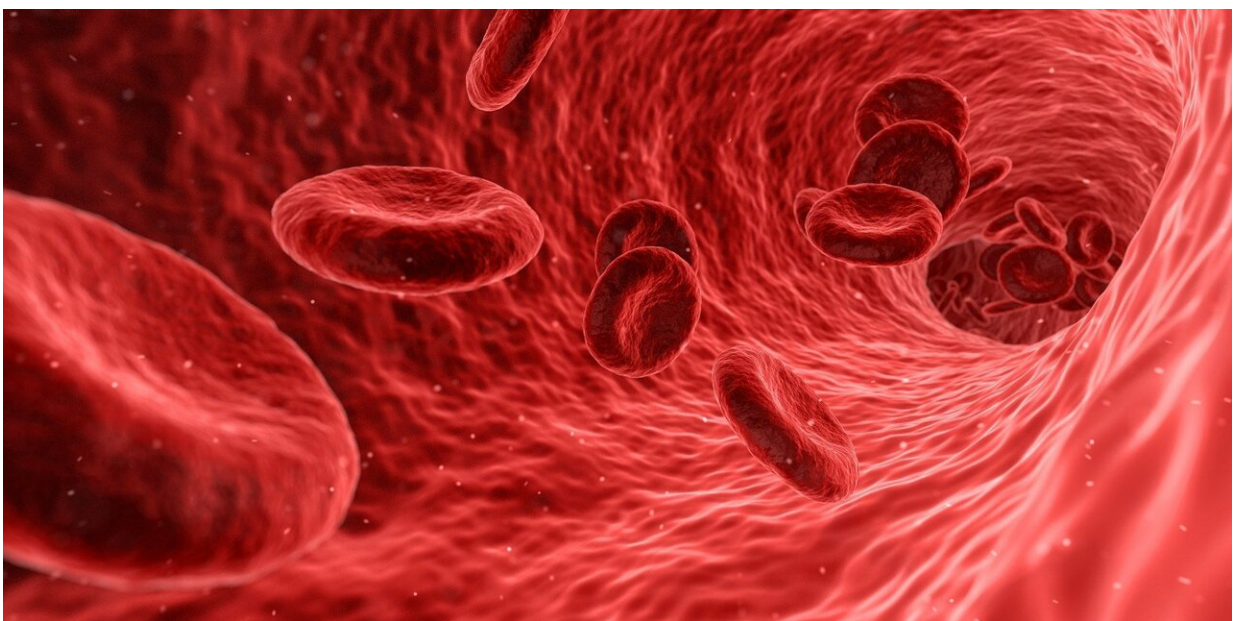


Genomic screening to identify iron overload encourages patients to seek treatment and condition management, study finds

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Genomic screening to identify hemochromatosis—a disorder that causes iron levels in the body to rise to dangerous levels—encourages people with the condition to seek treatment and ongoing management, a [Geisinger has study found](#).

Hereditary hemochromatosis type 1 (HH1), caused by a change in the

HFE gene, is underdiagnosed, often resulting in missed opportunities for early and consistent treatment. Without treatment, hemochromatosis can cause iron overload, a buildup of iron that can damage many parts of the body. Treatment of hemochromatosis includes regular monitoring of [iron levels](#) and removing excess iron from the body.

The Geisinger study evaluated genetic and electronic health record data for 86,601 participants enrolled in the MyCode Community Health Initiative. Of this group, 201 were found to have changes in the HFE gene, but only 28% of these had been previously diagnosed with HH1. Of those who were notified about their [genetic change](#) through MyCode, 69% proceeded with a recommended lab test, and 69% of those whose labs showed evidence of iron overload began subsequent treatment.

"Historically, people have been found to have hemochromatosis due to a personal or family history. We found that genetic screening for HFE across a health care population identifies underdiagnosed iron overload, encourages relevant management and offers the potential to reduce morbidity and mortality associated with [iron overload](#)," said Juliann Savatt, MS, CGC, assistant professor and co-director of the MyCode Genomic Screening and Counseling Program at Geisinger.

The results were published Oct. 23 in *JAMA Network Open*.

More research is needed to determine the long-term impact of genomic screening on [health outcomes](#) and cost-effectiveness for those with hemochromatosis, the study team wrote.

More information: Juliann M. Savatt et al, Testing and Management of Iron Overload After Genetic Screening–Identified Hemochromatosis, *JAMA Network Open* (2023). [DOI: 10.1001/jamanetworkopen.2023.38995](#)

Provided by Geisinger Health System

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