Medical Policy

Genetic Testing for Hereditary Hemochromatosis

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Policy Number: 908
BCBSA Reference Number: 2.04.80

Related Policies
None

Policy
Genetic testing for HFE gene mutations may be considered MEDICALLY NECESSARY in a patient with abnormal serum iron indices indicating iron overload.

Genetic testing for HFE gene mutations may be considered MEDICALLY NECESSARY in individuals with a family history of hemochromatosis in a first-degree relative.

Genetic testing for hereditary hemochromatosis in screening of the general population is considered INVESTIGATIONAL.

Prior Authorization Information
Commercial Members: Managed Care (HMO and POS)
Prior authorization is NOT required.

Commercial Members: PPO, and Indemnity
Prior authorization is NOT required.

Medicare Members: HMO BlueSM
Prior authorization is NOT required.

Medicare Members: PPO BlueSM
Prior authorization is NOT required.

CPT Codes / HCPCS Codes / ICD-9 Codes
The following codes are included below for informational purposes. Inclusion or exclusion of a code does not constitute or imply member coverage or provider reimbursement. Please refer to the member’s contract benefits in effect at the time of service to determine coverage or non-coverage as it applies to an individual member. A draft of future ICD-10 Coding related to this document, as it might look today, is included below for your reference.
Providers should report all services using the most up-to-date industry-standard procedure, revenue, and diagnosis codes, including modifiers where applicable.

CPT Codes

<table>
<thead>
<tr>
<th>CPT codes</th>
<th>Code Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>81256</td>
<td>HFE (hemochromatosis) (e.g., hereditary hemochromatosis) gene analysis, common variants (e.g., C282Y, H63D)</td>
</tr>
</tbody>
</table>

ICD-9 Diagnosis Codes

<table>
<thead>
<tr>
<th>ICD-9-CM diagnosis codes:</th>
<th>Code Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>275.01-275.09</td>
<td>Disorders of iron metabolism code range</td>
</tr>
<tr>
<td>V18.19</td>
<td>Family history of other endocrine and metabolic diseases</td>
</tr>
</tbody>
</table>

ICD-10-CM Diagnosis Codes

<table>
<thead>
<tr>
<th>ICD-10-CM diagnosis codes:</th>
<th>Code Description</th>
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<tbody>
<tr>
<td>E83.110</td>
<td>Hereditary hemochromatosis</td>
</tr>
<tr>
<td>E83.111</td>
<td>Hemochromatosis due to repeated red blood cell transfusions</td>
</tr>
<tr>
<td>E83.118</td>
<td>Other hemochromatosis</td>
</tr>
<tr>
<td>E83.119</td>
<td>Hemochromatosis, unspecified</td>
</tr>
<tr>
<td>E83.10</td>
<td>Disorder of iron metabolism, unspecified</td>
</tr>
<tr>
<td>E83.19</td>
<td>Other disorders of iron metabolism</td>
</tr>
<tr>
<td>Z83.49</td>
<td>Family history of other endocrine, nutritional and metabolic diseases</td>
</tr>
</tbody>
</table>

Description

Hereditary hemochromatosis, a common genetic disorder of iron metabolism, can lead to inappropriate iron absorption, toxic accumulation and organ damage. Genetic testing is available to assess mutations in the HFE gene, which are responsible for the majority of clinically significant cases of hereditary hemochromatosis.

Iron overload syndromes may be hereditary, secondary to some other disease (e.g. iron-loading anemias, parenteral iron overload, chronic liver disease or dysmetabolic iron overload syndrome), or due to other miscellaneous conditions (e.g., neonatal iron overload, aceruloplasminemia, congenital atransferrinemia).

Iron overload, if left untreated, can lead to secondary tissue damage in a wide range of organs resulting in chronic liver disease (hepatic fibrosis, cirrhosis, hepatocellular carcinoma), endocrine dysfunction (diabetes, hypogonadism), arthralgia or arthritis (typically involving the second and third metacarpophalangeal joints), and cardiomyopathy (either with symptomatic cardiac failure or arrhythmias).

Hereditary hemochromatosis (HH), an autosomal recessive disorder, is the most common, identified, genetic disorder in Caucasians, and may be seen in approximately 1 in 250 Caucasians. HH leads to inappropriate iron absorption from the intestine and progressive increase in intracellular iron concentrations. Untreated HH leads to premature death, usually by liver complications. Treatment by removing excess iron with serial phlebotomy is simple and effective, and if started before irreversible end organ damage, restores normal life expectancy.

Genetic testing can confirm a hereditary nature of the iron overload.
Summary
Hereditary hemochromatosis is a common genetic disorder in the Caucasian population. Abnormal serum iron indices, clinical symptoms of iron overload or a family history of hereditary hemochromatosis, may provoke testing for diagnosis. Testing for mutations in the \textit{HFE} gene, which contributes to the majority of cases of hereditary hemochromatosis, can confirm a genetic etiology; if clinically indicated, serial phlebotomy may be initiated, which can lead to a restored normal life expectancy. Therefore, genetic testing for \textit{HFE} gene mutations may be considered medically necessary for patients with a clinical suspicion of hemochromatosis (signs and symptoms of iron overload) or in patients with fasting serum iron indices that are suggestive of iron overload, as well as in individuals with a family history of hemochromatosis.

As hereditary hemochromatosis is common, general population screening has been proposed because of the high prevalence of the disease, the lack of early clinical or nonspecific early clinical findings, the simplicity and effectiveness of treatment, and the low success rate of late diagnosis and treatment. However, because the penetrance of the genotype is low, and the natural history of untreated individuals cannot be predicted, there is a lack of support for population-based screening. Therefore, genetic testing for hereditary hemochromatosis in screening of the general population is considered investigational.

Policy History

<table>
<thead>
<tr>
<th>Date</th>
<th>Action</th>
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<tbody>
<tr>
<td>7/2014</td>
<td>New references added from BCBSA National medical policy.</td>
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<tr>
<td>6/2014</td>
<td>Updated Coding section with ICD10 procedure and diagnosis codes, effective 10/2015.</td>
</tr>
<tr>
<td>6/2013</td>
<td>New references from BCBSA National medical policy.</td>
</tr>
<tr>
<td>5/2013</td>
<td>New references from BCBSA National medical policy.</td>
</tr>
<tr>
<td>2/2013</td>
<td>New policy describing covered and non-covered indications</td>
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Information Pertaining to All Blue Cross Blue Shield Medical Policies
Click on any of the following terms to access the relevant information:

- Medical Policy Terms of Use
- Managed Care Guidelines
- Indemnity/PPO Guidelines
- Clinical Exception Process
- Medical Technology Assessment Guidelines

References


