Genetic Testing for Hereditary Hemochromatosis

Definition
Hereditary hemochromatosis (HHC) is an autosomal recessive condition primarily caused by mutations of the HFE gene and characterized by excessive iron absorption from the gastrointestinal tract, iron overload and consequent deposition in multiple tissues. Deposition of excess iron results in target end organ damage presenting as non-specific clinical features including malaise and lethargy, and complications of cirrhosis, diabetes, cardiomyopathy, gonadal failure and arthritis. HHC is the most common identified genetic disorder in Caucasians; which may be seen in approximately 1 in 250.

Related Medical Guideline
Genetic Counseling and Testing

Guideline
Genetic testing for HHC is considered medically necessary for any of the following indications:

1. Elevated serum ferritin level (> upper limit of normal)
2. ≥ 2 consecutive elevations of fasting serum transferrin saturation (> 45%)
3. Positive family history of hemochromatosis in a first-degree relative

Limitations/Exclusions
Genetic testing for HHC is considered experimental and investigational for all other indications not meeting the above criteria.
Applicable Procedure Codes

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

Applicable ICD-10 Diagnosis Codes

<table>
<thead>
<tr>
<th>Code</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>E83.10</td>
<td>Disorder of iron metabolism, unspecified</td>
</tr>
<tr>
<td>E83.110</td>
<td>Hereditary hemochromatosis</td>
</tr>
<tr>
<td>E83.118</td>
<td>Other hemochromatosis</td>
</tr>
<tr>
<td>E83.119</td>
<td>Hemochromatosis, unspecified</td>
</tr>
<tr>
<td>E83.19</td>
<td>Other disorders of iron metabolism</td>
</tr>
</tbody>
</table>

References

7. Specialty matched clinical peer review.