Medical Policy

Genetic Testing for Predisposition to Inherited Hypertrophic Cardiomyopathy

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Policy Number: 909
BCBSA Reference Number: 2.02.28

Related Policies
None

Policy

Commercial Members: Managed Care (HMO and POS), PPO, and Indemnity
Medicare HMO BlueSM and Medicare PPO BlueSM Members

Genetic testing for predisposition to hypertrophic cardiomyopathy (HCM) may be considered MEDICALLY NECESSARY for individuals who are at risk for development of HCM, defined as having a first-degree relative with established HCM, when there is a known pathogenic gene mutation present in that affected relative.

Genetic testing for predisposition to HCM is considered NOT MEDICALLY NECESSARY for patients with a family history of HCM in which a first-degree relative has tested negative for pathologic mutations.

Genetic testing for predisposition to HCM is considered INVESTIGATIONAL for all other patient populations, including but not limited to individuals who have a first-degree relative with clinical HCM, but in whom genetic testing is unavailable.

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.*

Providers should report all services using the most up-to-date industry-standard procedure, revenue, and diagnosis codes, including modifiers where applicable.

**CPT Codes**
There is no specific CPT code for this service.

**HCPCS Codes**

<table>
<thead>
<tr>
<th>HCPCS Codes</th>
<th>Code Description</th>
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<tbody>
<tr>
<td>S3866</td>
<td>Genetic analysis for a specific gene mutation for hypertrophic cardiomyopathy (HCM) in an individual with a known HCM mutation in the family</td>
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**Description**
Familial hypertrophic cardiomyopathy (HCM) is an inherited condition that is caused by a mutation in one or more of the cardiac sarcomere genes. HCM is associated with numerous cardiac abnormalities, the most serious of which is sudden cardiac death (SCD). Genetic testing for HCM-associated mutations is currently available through a number of commercial laboratories.

The clinical diagnosis of HCM depends on the presence of left ventricular hypertrophy (LVH), measured by echocardiography or magnetic resonance imaging (MRI), in the absence of other known causative factors such as valvular disease, long-standing hypertension, or other myocardial disease. In addition to primary cardiac disorders, there are systemic diseases that can lead to LVH and thus “mimic” HCM. These include infiltrative diseases such as amyloidosis, glycogen storage diseases such as Fabry disease and Pompe disease, and neuromuscular disorders such as Noonan's syndrome and Friederich's ataxia. These disorders need to be excluded before a diagnosis of familial HCM is made.

Genetic testing has been proposed as a component of screening at-risk individuals to determine predisposition to HCM among those patients at risk. Patients at risk for HCM are defined as individuals who have a close family member with established HCM. Results of genetic testing may influence management of at-risk individuals, which may in turn lead to improved outcomes. Furthermore, results of genetic testing may have implications for decision making in the areas of reproduction, employment, and leisure activities.

**Summary**
For individuals at risk for HCM (first-degree relatives), genetic testing is most useful when there is a known mutation in the family. In this situation, genetic testing will establish the presence or absence of the same mutation in a close relative with a high degree of certainty. Absence of this mutation will establish that the individual has not inherited the familial predisposition to HCM and thus has a similar risk of developing HCM as the general population. These patients no longer need ongoing surveillance for the presence of clinical signs of HCM. Therefore, genetic testing may be considered medically necessary for first-degree relatives of individuals with a known pathologic mutation.

For at-risk individuals without a known mutation in the family, the evidence does not permit conclusions of the effect of genetic testing on outcomes, since there is not a clear relationship between testing and improved outcomes. Genetic testing is considered investigational for this purpose. For at-risk individuals who have a family member with HCM who tests negative for pathologic mutations, genetic testing is not indicated. Genetic testing is considered not medically necessary in this situation.
Policy History

<table>
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<tr>
<th>Date</th>
<th>Action</th>
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<tbody>
<tr>
<td>12/2013</td>
<td>Coding information clarified</td>
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<tr>
<td>2/2013</td>
<td>New policy describing coverage and non-coverage</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

36. Ackerman MJ, Priori SG, Willems S et al. HRS/EHRA expert consensus statement on the state of genetic testing for the channelopathies and cardiomyopathies this document was developed as a partnership between the Heart Rhythm Society (HRS) and the European Heart Rhythm Association (EHRA). Heart Rhythm 2011; 8(8):1308-39.