EmblemHealth

Genetic Testing for Hereditary Cardiomyopathy

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Definitions

Familial hypertrophic cardiomyopathy (HCM)	A disease of the myocardium commonly caused by a mutation in one or more of the cardiac sarcomere genes and characterized by left ventricular hypertrophy (LVH) in the absence of predisposing cardiac or systemic diseases. The clinical features of HCM vary for each individual and range from asymptomatic to progressive heart failure to sudden cardiac death. Common symptoms include dyspnea on exertion, palpitations, chest pain, orthostatic hypotension, presyncope, and syncope. It is the most common hereditary cardiac condition in the U.S., affecting 1 in 500 adults and possibly the most common cause of sudden cardiac death (SCD) in young athletes and others 35 years of age and younger.
Familial dilated cardiomyopathy (DCM)	A type of dilated cardiomyopathy associated with heterogenous genetic mutations, characterized by left ventricular enlargement and dilatation, systolic dysfunction, leading to clinical manifestations of heart failure. Primary clinical manifestations of DCM are heart failure, arrhythmias and/or conduction system disease, and/or thromboembolic disease.
Arrhythmogenic right ventricular dysplasia/cardiomyopathy (ARVD/C)	A myocardial disorder of the right ventricle associated with a heterogenous number of genetic mutations including PKP2 (plakophilin-2), DSG2 (desmoglein-2) and DSP (desmoplakin) genes. ARVD/C is characterized by progressive fibrofatty replacement of the myocardium that predisposes affected individuals to ventricular tachycardia and sudden death, especially in young persons and athletes. The pathology in ARVD/C may also extend to involve the left ventricle.

Guideline

Genetic testing for HCM is considered medically necessary when the following criteria are met; **all**:

1. Member has a first-degree relative with a documented history of HCM, and the first degree relative also has a genetic mutation with strong evidence for pathogenicity
2. Member tests negative for HCM with specific clinical investigations (e.g., with EKG, echocardiogram or cardiac magnetic resonance imaging [MRI])
3. Member has received genetic counseling

Limitations/Exclusions

1. Genetic testing for HCM is considered investigational for all other indications not meeting the above criteria.
2. Genetic testing for all other hereditary cardiomyopathies including, DCM, ARVD/C, restrictive and left ventricular noncompaction cardiomyopathies is considered investigational and not medically necessary for all indications.

Applicable Procedure Codes

81403	Molecular pathology procedure, Level 4 (eg, analysis of single exon by DNA sequence analysis, analysis of 10 amplicons using multiplex PCR in 2 or more independent reactions, mutation scanning or duplication/deletion variants of 2-5 exons) (Revision eff. 01/01/2018)
81405	Molecular pathology procedure, Level 6 (eg, analysis of 6-10 exons by DNA sequence analysis, mutation scanning or duplication/deletion variants of 11-25 exons, regionally targeted cytogenomic array analysis) CPOX (coproporphyrinogen oxidase) (eg, hereditary coproporphyria), full gene sequence CTRC (chymotrypsin C) (eg, hereditary pancreatitis), full gene sequence PKLR (pyruvate kinase, liver and RBC) (eg, pyruvate kinase deficiency), full gene sequence (Revision eff. 01/01/2018)
81406	Molecular pathology procedure, Level 7 (eg, analysis of 11-25 exons by DNA sequence analysis, mutation scanning or duplication/deletion variants of 26-50 exons, cytogenomic array analysis for neoplasia) ANOS1 (anosmin-1) (eg, Kallmann syndrome 1), full gene sequence HMBS (hydroxymethylbilane synthase) (eg, acute intermittent porphyria), full gene sequence PPOX (protoporphyrinogen oxidase) (eg, variegate porphyria), full gene sequence (Revision eff. 01/01/2018)
81407	Molecular pathology procedure, Level 8 (eg, analysis of 26-50 exons by DNA sequence analysis, mutation scanning or duplication/deletion variants of >50 exons, sequence analysis of multiple genes on one platform)
81479	Unlisted molecular pathology procedure
S3865	Comprehensive gene sequence analysis for hypertrophic cardiomyopathy
S3866	Genetic analysis for a specific gene mutation for hypertrophic cardiomyopathy (HCM) in an individual with a known HCM mutation in the family

Applicable ICD-10 Diagnosis Codes

I42.0	Dilated cardiomyopathy
I42.1	Obstructive hypertrophic cardiomyopathy
I42.2	Other hypertrophic cardiomyopathy

References

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