LABupdate

Familial Cardiomyopathy

Cardiomyopathies are generally characterized by weakening and impaired contractile function of the myocardium that leads to ventricular hypertrophy or dilation. Myocardial dysfunction associated with cardiomyopathy can either be of mechanical or electrical etiology.[1,2] The 4 major types of cardiomyopathy include dilated cardiomyopathy (DCM), hypertrophic cardiomyopathy (HCM), arrhythmogenic right ventricular dysplasia/cardiomyopathy (ARVD/C), and restrictive cardiomyopathy (RCM).[2,3] Rarer types include left ventricular noncompaction (LVNC)[1,3] and the amyloid-associated cardiomyopathies, such as transthyretin (TTR) amyloidosis and apolipoprotein A-1 amyloidosis (AApoA-1).[4]

Patients with cardiomyopathy are at increased risk for arrhythmias, thrombolytic events, and sudden cardiac death (SCD).[1,3] Age of onset and life expectancy vary with etiology.[1,2,4-7] Symptoms of cardiomyopathy are typical of those associated with heart failure and may include fatigue, shortness of breath, orthopnea, paroxysmal nocturnal dyspnea, cough, and edema.[2] Patients may be asymptomatic in the early stages, and, in some cases, SCD is the initial presentation.[2]

Prevention of heart failure, thrombolytic events, and SCD in cardiomyopathy patients can be achieved through administration of antiarrhythmic and/or anticoagulation drugs, implantable cardioverter defibrillator (ICD) therapy, and certain lifestyle changes[2,5-7]; however, identification of at-risk individuals who may benefit from these treatments can be difficult, since abnormal electrocardiogram or echocardiogram profiles may not always be clear, at-risk patients may be asymptomatic, and SCD may be the presenting clinical manifestation.[1,6,7]

Many cardiomyopathies are now recognized as familial conditions that may be transmitted in an autosomal dominant, autosomal recessive, X-linked, or mitochondrial manner.[1,2,4-7] Genetic testing for the presence of germline mutations in the genes known to be associated with cardiomyopathy may[1,4-7]:

- Confirm a diagnosis of a familial cardiomyopathy.
- Identify which subtype of a particular cardiomyopathy a patient may have.
- Identify family members of an index patient who harbor the familial mutation and may wish to undergo cardiac screening at regular intervals.
- Facilitate appropriate genetic counseling for family members.

Relevant Assays*

Test Name	Test No.
GeneSeq®: Cardio Familial Cardiomyopathy Profile	451422
GeneSeq®: Cardio Gene Specific Sequencing, NGS**	452053
Mutation-specific Sequencing, Whole Blood†	451382

* Visit the online Test Menu at www.LabCorp.com for more information, including a current list of included genes, test methodology, and specimen requirements. To request a sample shipping kit, please call 866-647-0735.
**Full Gene Sequencing for any gene(s) on any of the GeneSeq: Cardio panels
†Known mutation testing for any gene(s) on any of the GeneSeq: Cardio panels

References

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