LABupdate

Familial Arrhythmia

Cardiac arrhythmias are generally characterized by abnormal electrical activity in the heart that puts patients at high risk for embolic stroke and/or sudden cardiac death (SCD). Commonly recognized arrhythmic disorders include atrial fibrillation (AF), long QT syndrome (LQTS), catecholaminergic polymorphic ventricular tachycardia (CPVT), arrhythmogenic right ventricular dysplasia/cardiomyopathy (ARVD/C), and Brugada syndrome (BrS). While their clinical presentations are generally similar and may include syncope, palpitations, dizziness, dyspnea, stroke, and/or SCD,[1] each of these disorders has a different etiology and prognosis. Age of onset varies by condition and can, in some cases, occur during early childhood or adolescence.[1-3]

Treatments such as antiarrhythmic and/or anticoagulation drugs, implantable cardioverter defibrillator (ICD) therapy, and certain lifestyle changes may prevent stroke and sudden cardiac death in patients with cardiac arrhythmias.[1,3]

Genetic testing for mutations in genes known to be associated with LQTS, CPVT, ARVD/C, AF, and BrS can be used in conjunction with standard cardiac testing to help:[1,3,4,5]

- Confirm a diagnosis.
- Differentiate between different arrhythmic disorders.
- Clarify the prognosis, alerting patients and physicians to the most common arrhythmia triggers, which may be specific to the underlying genetic cause.
- Guide therapeutic strategies.
- Identify family members who are at increased risk for arrhythmic disorder and may benefit from cardiac screening.

An estimated 30% to 50% of arrhythmia cases are familial.[2,6-11] Mutations responsible for arrhythmias are typically acquired in an autosomal-dominant manner.[1,3] Carrier screening for mutations in at-risk family members may help identify individuals — particularly those who do not have clinical signs or symptoms of disease — who would benefit from early intervention to reduce the risk of cardiac events.[1,3,5,9,11]

Relevant Assays*

Test Name	Test No.
GeneSeq®: Cardio Familial Arrhythmia Profile	451412
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

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