GeneSeq®: Cardio

Helping you provide better patient care

Testing for **more than 100 genetic causes** of familial cardiac disease.

Treatment That May Help

Familial cardiac diseases are associated with up to 80% of cases of sudden cardiac death in young patients.[1] Identification of individuals with pathogenic mutations in genes associated with cardiac disease may allow timely initiation of screening and treatment that may help prevent myocardial infarction, stroke, and sudden cardiac death.

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can be a useful prognostic tool in the presence of a positive family history and symptoms of cardiomyopathy, arrhythmia, aortopathy, Noonan syndrome, RASopathies, congenital heart disease, early-onset coronary artery disease, or familial hypercholesterolemia.

Clinical Utility

- Establish/confirm a diagnosis of familial cardiac disease.
- Identify the need for regular cardiac screening, lifestyle changes, or pharmacological or surgical intervention to prevent the progression of cardiac disease and secondary complications.
- Identify first-degree relatives of the proband who have inherited a disease-causing genetic variant and may be at risk for myocardial infarction, stroke, or sudden cardiac death.
- Facilitate appropriate genetic counseling for probands and their first-degree relatives.

Sample Requirements

- 10 mL whole blood or 30 mL if ordering multiple tests.

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Six indications for testing, available separately or in combination

Test No.	Test Name	Genes Included In the Profile
451422	**GeneSeq®: Cardio - Familial Cardiomyopathy Profile** (48 genes)	
	Hypertrophic cardiomyopathy	*ACTC1, CAV3, CSRP3, GLA, LAMP2, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, PRKAG2, TNNC1, TNNI3, TNNT2, TPM1*
	Arrhythmogenic right ventricular dysplasia/cardiomyopathy	*DSC2, DSG2, DSP, JUP, PKP2, RYR2, TGFB3, TMEM43*
	Dilated cardiomyopathy	*ABCC9, ACTC1, ACTN2, ALMS1, BAG3, CSRP3, CTF1, DES, DNAJC19, EMD, EYA4, FKTN, LAMP2, LDB3, LMNA, MYBPC3, MYH7, PLN, RBM20, SCN5A, SGCD, TAZ, TCAP, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TTN*, VCL*
	Restrictive, left ventricular non-compaction, and cardiac amyloidosis	*ACTC1, APOA1, DES, DTNA, MYH7, TAZ, TNNI3, TNNT2, TTR*
451412	**GeneSeq®: Cardio - Familial Arrhythmia Profile** (29 genes)	
	Long QT syndrome/ Brugada syndrome	*AKAP9, ANK2, ATP1B1, CACNA1C, CACNB2, CAV3, GPD1L, KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, LIG3, NOS1AP, PLN, SCN1B, SCN4B, SCN5A, SNTA1*
	Arrhythmogenic right ventricular dysplasia/cardiomyopathy	*DSC2, DSG2, DSP, JUP, PKP2, RYR2, TGFB3, TMEM43*
	Catecholaminergic polymorphic ventricular tachycardia	*CASQ2, KCNJ2, RYR2*
	Atrial fibrillation	*KCNE2, KCNQ1, NPPA*
451432	**GeneSeq®: Cardio - Familial Aortopathy Profile** (11 genes)	
	Marfan syndrome, Loeys-Dietz syndrome, vascular Ehlers-Danlos syndrome	*COL3A1, FBN1, SMAD3, TGFB2, TGFBR1, TGFBR2*
	Thoracic aortic aneurysms and dissections	*ACTA2, MED12**, MYH11, MYLK, TGFB2, SLC2A10, SMAD3, TGFBR1, TGFBR2*
451441	**GeneSeq®: Cardio - Noonan Syndrome/RASopathies Profile** (20 genes)	
	Noonan syndrome, cardiofaciocutaneous syndrome, Costello syndrome, LEOPARD syndrome	*A2ML1, BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MRAS, NF1, NRAS, PTPN11, RAF1, RASA2, RIT1, RRAS, SHOC2, SOS1, SOS2, SPRED1*
451402	**GeneSeq®: Cardio - Familial Congenital Heart Disease Profile** (4 genes)	
	Atrial septal defects	*GATA4, NKX2-5, TBX5*
	CHARGE syndrome	*CHD7*
452040	**GeneSeq®: Cardio - Familial Hypercholesterolemia Profile** (4 genes)	
	Familial Hypercholesterolemia	*APOB†, LDLR, LDLRAP1, PCSK9*
451416	**GeneSeq®: Cardio - Early-onset Coronary Artery Disease/ Familial Hypercholesterolemia Profile** (8 genes)	
	Familial Hypercholesterolemia/Coronary Artery Disease	*ABCA1, APOA2, APOB†, APOC3, LDLR, LDLRAP1, PCSK9, PON2*

*For TTN, exons 154 and 155 are not reported.
**Only the c.3020A>G mutation is sequenced.
†Only a 566 base pair region of exon 26 is sequenced.

A la carte gene sequencing and known mutation testing options

452028	FBN1 (Marfan Syndrome) Full Gene Sequencing	*FBN1* full gene sequencing
452053	GeneSeq®: Cardio - Gene Specific Sequencing	Full gene sequencing for any gene(s) on any of the GeneSeq: Cardio profiles
451382	Mutation-specific Sequencing, Whole Blood	Known mutation testing for any gene(s) on any of the GeneSeq: Cardio profiles

Reference
1. Shephard S, Semsarian C. Advances in the prevention of sudden cardiac death in the young. *Ther Adv Cardiovasc Dis*. 2009;32(Suppl 2):S2-S5.

For pricing and ordering information, please contact Client Services at **866-647-0735.**

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Visit the online Test Menu at **www.LabCorp.com** for full test information, including CPT codes and current specimen collection requirements.