Cardiovascular Diseases Genetic Testing Program

Molecular Genetics Laboratory

Shipping Instructions

Please enclose a test requisition form with sample. All information must be complete before sample can be processed. Samples may be shipped at room temperature by overnight Federal Express to arrive Monday through Friday.

Ship to:

Molecular Genetics Lab
Cincinnati Children's Hospital
3333 Burnet Ave. NRB 1042
Cincinnati, OH 45229

Phone: 513-636-4474
Fax: 513-636-4373

Comprehensive Cardiomyopathy Panel

The Comprehensive Cardiomyopathy Panel offers Next Generation Sequencing of 37 genes associated with dilated cardiomyopathy (DCM), hypertrophic cardiomyopathy (HCM), left ventricular noncompaction (LVNC), and restrictive cardiomyopathy (RCM).
The Comprehensive Cardiomyopathy Panel can be ordered as a first line test, or can be ordered as a cost-effective reflex following targeted disease testing (DCM, HCM, LVNC, or RCM).

HCM
Hypertrophic Cardiomyopathy (HCM) is relatively common, with a prevalence of 1 in 500 adults. HCM is a primary disorder of the heart muscle characterized by left ventricular hypertrophy. The most classic finding in HCM is asymmetric septal hypertrophy, with or without left ventricular outflow tract obstruction. The disease demonstrates extensive clinical variability with regard to age of onset, severity and progression of disease. HCM can affect infants and children although it is more typically identified in adolescence or adulthood. Approximately 50-65% of individuals with a known or suspected diagnosis of familial HCM have a mutation in one of a number of genes encoding components of the sarcomere and cytoskeleton.

DCM
Dilated Cardiomyopathy (DCM) is characterized by left ventricular enlargement and systolic dysfunction. DCM can either be acquired though environmental causes (most commonly through ischemic injury following myocardial infarction or coronary artery disease), or can be inherited. DCM is commonly an adult-onset disease, but demonstrates extensive variability with regard to age of onset and reduced penetrance. It is estimated that 20-50% of cases of DCM have a genetic basis.

LVNC
Left Ventricular Noncompaction (LVNC) is characterized by a spongy morphologic appearance of the myocardium, occurring primarily in the left ventricle with the abnormal trabeculations typically being most evident in the apical portion of the left ventricle. LVNC can be seen in isolation or in association with other cardiomyopathies (HCM or DCM) or congenital heart disease. It is estimated that up to 44% of LVNC cases are inherited.

RCM
Restrictive Cardiomyopathy (RCM) is characterized by increased stiffness of the ventricular chambers leading to abnormal filling and relaxation. Generally the ventricular wall thickness and systolic function remains within normal limits. Approximately 35% of individuals with RCM will have a mutation identified with currently available genetic testing.

Test Indication
This test is indicated for individuals with a primary cardiomyopathy in the absence of identifiable acquired causes.

Genes on Comprehensive Cardiomyopathy Panels

- Comprehensive Cardiomyopathy Panel – 37 genes
- HCM Panel – 23 genes*
- DCM Panel – 30 genes*
- LVNC Panel – 13 genes*
- RCM Panel – 9 genes*

**Optional reflex to remaining genes*

Methodology:

Next Generation Sequencing: DNA sequences are assembled and compared to the published genomic reference sequences in Genome Reference Consortium Build 37. Dideoxy DNA sequencing is used to provide data for bases with insufficient coverage and to confirm the reported variants from next-generation sequencing. This assay does not detect variants in the promoter regions, deep intronic regions, or other regulatory elements, and does not detect large deletions or mosaicisms. Variants are reported according to HGVS nomenclature (www.hgvs.org/mutnomen).

Sensitivity & Accuracy:

Validation testing indicates an analytic sensitivity of over 99% and an analytic specificity of 100%.

Clinical Sensitivity
Based on the literatures, the clinical validity is determined to be:

- Greater than 93% for HCM
- Greater than 26% for DCM
- Approximately 30% for LVNC
- Approximately 35% for RCM

Specimen:

Peripheral blood in EDTA tube
Adult: 3-5mL
Child: 3-5mL
Infant: 1-3mL
For other specimen types, please contact us at 513-636-4474

Turnaround Time:

Full Panel Analysis 8-10 weeks
Known Mutation Analysis 1-2 weeks

CPT Codes:

Comprehensive Cardiomyopathy Panel: 81403, 81404, 81405x12, 81406x5, 81407x4, 81479x14

Dilated Cardiomyopathy Panel: 81403, 81405 x9, 81406 x4, 81407 x4, 81479 x12

Hypertrophic Cardiomyopathy Panel: 81403, 81404x2, 81405x9, 81406x3, 81407x3, 81479 x5

Left Ventricular Noncompaction Panel: 81405 x5, 81406 x4, 81407 x2, 81479 x2

Restrictive Cardiomyopathy Panel: 81404, 81405 x3, 81406, 81407 x2, 81479 x2

Known Mutation Analysis: 81403

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Comprehensive Cardiomyopathy Panel Gene List

Gene	DCM	HCM	LVNC	RCM
ABCC9	x			
ACTC1	x	x	x	x
ACTN2	x	x	x	
ANKRD1*	x	x		
BAG3	x			x
CAV3		x		
CRYAB	x			x
CSRP3	x	x		
DES	x		x	x
EMD	x			
LAMP2	x	x		
LMNA	x		x	
MYBPC3	x	x	x	x
MYH6	x	x		
MYH7	x	x	x	x
MYL2		x	x	
MYL3		x	x	
MYPN	x			
NEBL*	x			
NEXN	x	x		
PLN	x	x		
PRKAG2		x		
RBM20	x			
SCN5A	x			
SCO2		x		
SGCD	x			
SURF1		x		
TAZ	x		x	
TCAP	x			
TNNC1	x	x		
TNNI3	x	x		x
TNNT2	x	x	x	x
TPM1	x	x	x	
TTN	x			
TTR		x		x
VCL	x	x	x	
ZASP/LDB3	x	x	x	

*CCHMC was the lead in identifying these novel genes

www.cincinnatichildrens.org/heartdx Phone: 513-636-4474 Fax: 513-636-4373