ALBERTA PRECISION LABORATORIES
Leaders in Laboratory Medicine

Cardiomyopathy Gene Panels: Information for Ordering Providers

Cardiomyopathy is defined as disease of the heart muscle and has many different presentations.

1. Hypertrophic cardiomyopathy (HCM) has an incidence of 1/500 and is characterized by idiopathic myocardial hypertrophy and myocyte disarray.
2. Dilated cardiomyopathy (DCM) has an incidence of 1/250 and is characterized by ventricular enlargement and systolic dysfunction, a reduction in the myocardial force of contraction.
3. Arrhythmogenic right ventricular dysplasia/cardiomyopathy (ARVD/C) has an incidence of 1/1000 and is characterized by progressive fibrofatty replacement of the myocardium that predisposes to ventricular tachycardia and sudden cardiac arrest.

Cardiomyopathies are genetically heterogeneous, meaning that the conditions can be caused by a pathogenic variant(s) in any one of a number of genes. Therefore, testing is based on the patient's phenotype and family history and includes a number of related genes.

Since not all genes associated with a given cardiomyopathy are known or included in the panel, a pathogenic variant will not be identified for every patient. **The absence of a pathogenic variant does not exclude a clinical or genetic diagnosis. Individuals who carry a pathogenic variant in a cardiomyopathy gene have an inherited form of heart disease and their at-risk family members should be offered genetic testing.**

Associated Disorders

Cardiomyopathy can be associated with a variety of metabolic and syndromic conditions. Examples include: inborn errors of metabolism (e.g. Fabry disease, Pompe disease), neuromuscular diseases (e.g. Emery-Dreifuss Muscular Dystrophy) and other genetic syndromes (e.g. Noonan syndrome). Consultation with a relevant specialist may be indicated if a pathogenic variant is identified in an associated gene.

Cardiomyopathy Gene Panels

There are 5 cardiomyopathy gene panels. Please reference the APL Test Directory (https://www.albertahealthservices.ca/webapps/labservices/indexAPL.asp?zoneid=1&SearchText=&submit=Submit+Query&upperTest=-1&lowerTest=-1) for a list of genes on each panel and ordering instructions.

- Arrhythmogenic right ventricular dysplasia/cardiomyopathy (LAB4772) - 17 genes
- Dilated cardiomyopathy (LAB4771) - 42 genes
- Hypertrophic cardiomyopathy (LAB4770) - 43 genes
- Pan cardiomyopathy (LAB4773) - 89 genes, includes genes with less evidence
- Comprehensive cardiac (LAB4774) - 99 genes, includes all genes on the pan cardiomyopathy panel and the pan arrhythmia panel

Indications for Testing

Individuals with a diagnosis of idiopathic cardiomyopathy are eligible for testing. Each patient is eligible to have only one cardiac gene panel. Please select the most appropriate test for your patient. Requests to reanalyze uninformative results for other genes/panels will be reviewed on a case by case basis and may not be accommodated. Refer to algorithm below for the most appropriate test for your patient.

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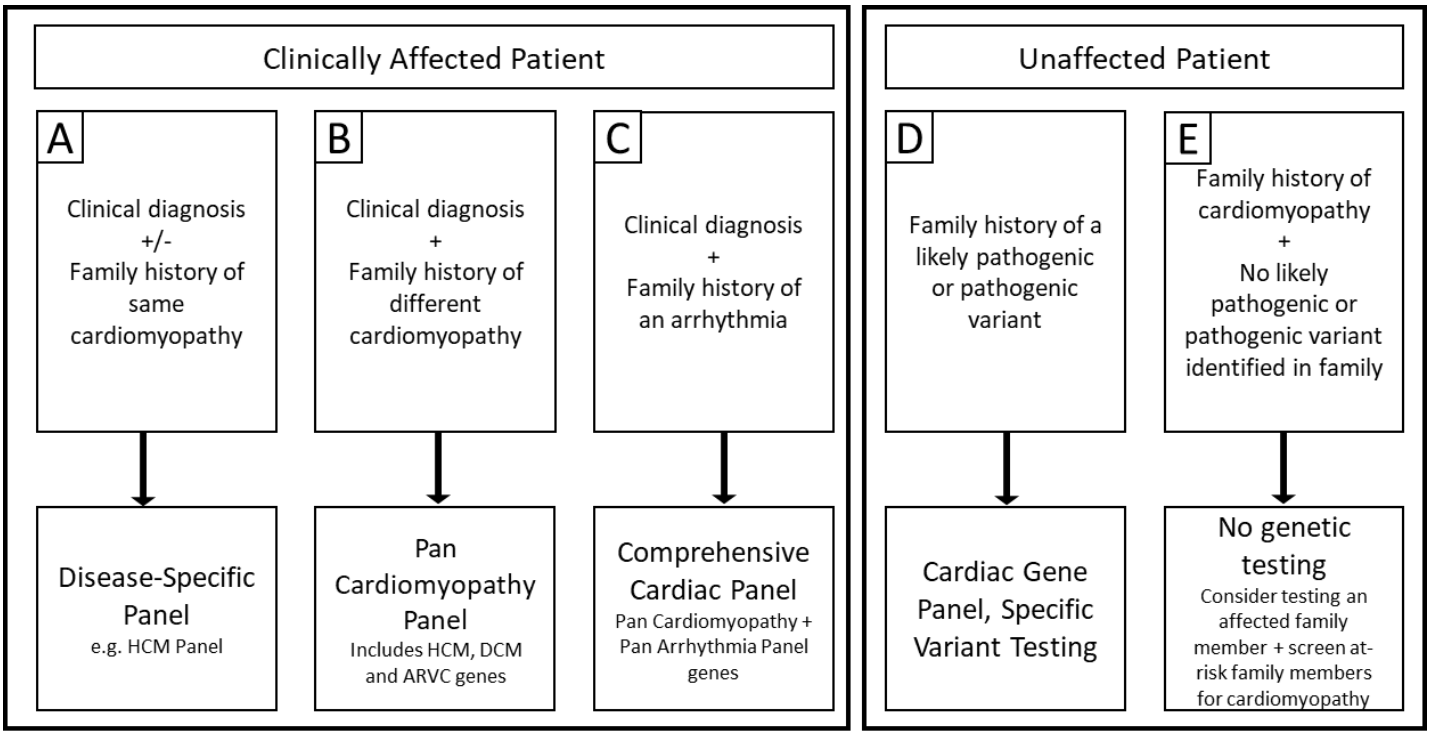

Adapted from Yogasundaram et al. Cardiomyopathies and Genetic Testing in Heart Failure: Role in Defining Phenotype-Targeted Approaches and Management. Can J Cardiol. 2021 Apr;37(4):547-559.

Ordering Privileges
Diagnostic testing can be ordered by Clinical Genetics or the Medical Examiner's Office.
Predictive testing for a known pathogenic/likely pathogenic variant is restricted to Clinical Genetics.

When can I expect results?
Results may take up to 4 months.

Can testing be expedited to facilitate medical management of a patient?
Expedited testing (a minimum of 4 weeks from the time the *sample* is received in the testing lab) is available if required for immediate medical management. Please provide details on the requisition form regarding the reason for expedited testing as well as a target date for results.

How are results reported?
Results are sent to the ordering provider and available in Netcare and Connect Care.

Requisition forms, contact information and other resources can be found at Genetics & Genomics

Contact Information
Laboratory Genetic Counsellors, Genetics & Genomics North Sector at 780-407-1015.

Resources
Canadian SADS Foundation
Hypertrophic Cardiomyopathy Association