heartgene

Cardiomyopathy Genetic Testing: HCP Ordering Guide

Why Heartgene exists

Genetic testing can meaningfully change care in cardiomyopathy, yet in real-world practice, only about 1.1% of patients receive genetic testing.[1,2]

Heartgene's mission is to close this gap by designing a testing model that works for patients first while remaining easy for clinicians to use.

Interpretation is performed using the **Heartgene Cardiomyopathy and Channelopathy Panel**, a 57-gene panel that includes genes associated with hypertrophic (HCM), dilated (DCM), and arrhythmogenic cardiomyopathies, as well as key cardiac channelopathies linked to arrhythmia and sudden cardiac death risk.

Who this testing is for

Heartgene provides clinical-grade genetic testing for patients with suspected or confirmed cardiomyopathy, as well as eligible family members when a pathogenic variant is identified.

This program is designed to make genetic testing accessible without adding burden to clinical workflows. No insurance is required, and there is no billing.

→ **Testing is supported by sponsored programs designed to expand access.**

Two ways to get testing to your patients

You can choose the approach that best fits your practice. Both options use the same testing process and clinical partners.

A. Order kits to your clinic:

- Request kits for your practice at heartgene.com
- Distribute kits directly to patients during visits

B. Send patients directly to Heartgene:

- Patients request a kit shipped to their home from heartgene.com

1. Epidemiology of the inherited cardiomyopathies
2. Real-world genetic testing utilization among patients with cardiomyopathy

The most important step!

This program is patient-centered by design.

Patients are the primary participants in the process. They receive:

- Their own secure portal
- Their test results
- Genetic counseling when results are positive

To enable this, **patients must register their kit and complete required consent forms before mailing it back.**

Registering the kit and completing all the steps will:

- Activate patient consent
- Link the sample to the patient
- Enable delivery of results and counseling

Unregistered kits or incomplete consents cannot be processed, and samples may be discarded.

Tip: Encourage patients to complete registration and consents before leaving the visit, or immediately upon receiving the kit at home.

What happens after the sample is returned

Once a registered kit is mailed back:

- Sequencing is performed by Broad Clinical Labs.
- Interpretation is completed by the Mass General Brigham Laboratory for Molecular Medicine.
- Results are returned to the care team 4–8 weeks after the sample is received.

How results are delivered

Pathogenic or likely pathogenic results

- Sent directly to the ordering or referring provider
- Reviewed with the patient during genetic counseling

Negative results

- Available to the patient
- Patients are encouraged to share results with their care team

Variants of uncertain significance (VUS)

- Tracked internally
- Can be discussed with providers if clinically relevant

Genetic counseling

Post-test genetic counseling is provided for patients with positive results.

- Delivered by DNAVisit
- Designed to support patients and in understanding results and next steps

Building the next phase together

Heartgene was created to bring sustained focus to cardiomyopathy genetics and to improve the experience for both clinicians and patients.

We are grateful to the clinicians who have been early adopters of the program. Your use of the testing and real-world feedback have shaped where Heartgene is headed. Over the coming months, our focus includes:

- Improved visibility into sample status and results
- A dedicated provider portal designed for cardiology workflows
- Ongoing workflow refinements informed directly by clinician input
- Expansion of the genetic panel to include core channelopathy genes

Questions or feedback

If you have questions or feedback, please reach out at support@heartgene.com. We are building this program with clinicians and value your input.