Genetic Insights: quick reference guide for healthcare providers

This guide is intended to facilitate a discussion between a provider and their patient.

Genetic Insights test results: **Cardiomyopathy**

Key results

A pathogenic or likely pathogenic variant associated with an increased risk of developing cardiomyopathy was found in the *MYH7* gene.

Next steps

Clinical recommendations	Resources
Genetic Insights is a screening test and is not intended for diagnosis. A follow-up genetic test should be performed in a clinical setting before any other action is taken.	**Ready to order?** Check with your institution and/or patient's insurance about the preferred testing laboratory. Blueprint Genetics® offers genetic testing. To confirm this test result, **targeted variant testing** for the variant identified is available. You can order a confirmation test here: Blueprint Genetics/TVT **Have questions?** Call **1.866.GENE.INFO** (1.866.436.3463) to speak to a specialized Quest genetic counselor or geneticist available to healthcare providers to discuss test selection and results.
Refer your patient to a genetic counselor.	Your patient can schedule a 1-on-1 remote genetic counseling session through their online Genetic Insights Cardiomyopathy Report at no additional cost.
Genetic counselors can provide counseling on the implications of this test result and next steps for your patient.	To find a genetic counselor practicing in your patient's area for an in-person session, please visit FindAGeneticCounselor.NSGC.org

Patient conversation starters:

Patient conversation starters summarize the preceding information in plain language to support meaningful conversations between you and your patient.

FindAGeneticCounselor.NSGC.org

Your Genetic Insights test is a screening test. The next step is to have your result confirmed with a second genetic test.

It's also important that you talk with a genetic counselor. Genetic counselors are experts in genetics and can help you understand this result and potential next steps.

You can access a genetic counselor through your Genetic Insights portal at no additional cost to you, and we can discuss a referral to a local genetic counselor.

Visit QuestDiagnostics.com/Genetic-Health-Screening for more information about this test.

What is cardiomyopathy?

Cardiomyopathy is a disease of the heart muscle that can present with different types and have different etiologies.[1]

Hypertrophic cardiomyopathy (HCM) and dilated cardiomyopathy (DCM) are the most common types and can be caused by DNA variants.[1]

People with a DNA variant in the *MYH7* gene have an increased risk of developing cardiomyopathy in their lifetime. The exact risk is currently unknown.

Signs and symptoms of cardiomyopathy may vary and can include arrhythmia, fainting, shortness of breath, chest pain, and more rarely sudden cardiac arrest or death. Cardiomyopathy may progress to heart failure.[1]

See the Management options section for more detail.

Patient conversation starters:

Cardiomyopathy is a type of heart disease that damages the heart muscle.

People with a confirmed DNA variant in the *MYH7* gene have a higher than typical chance of developing cardiomyopathy. This can cause problems ranging from shortness of breath to heart failure.

Not everyone with a DNA variant will develop cardiomyopathy. It's very important to make a plan to monitor your heart health.

What this result means for family members

Family members may have the same DNA variant. The DNA variant was most likely inherited from a parent. Full siblings and children have a 50% chance of having this variant.

In people with a confirmed DNA variant associated with an increased risk for cardiomyopathy, cascade genetic testing for other family members may help inform their risks and screening protocols.[3,4]

A genetic counselor can help determine the most appropriate testing options. Therefore, it is strongly recommended that people share their results with their biological relatives.

Patient conversation starters:

DNA variants in the *MYH7* gene can run in families.

That means the DNA variant can be inherited or passed down from parents to their children. Your close relatives, like your parents, full siblings, and children, have a 50% (or 1 in 2) chance of having the same DNA variant.

Other relatives might also have the same DNA variant.

Sharing these results with your family is important so family members can decide if they want to have genetic testing and monitor their heart health.

Management options

People with a DNA variant linked to cardiomyopathy have options for monitoring and managing their heart health. Select clinical guidelines are listed below.

Clinical scenario	Guidelines for people with the *MYH7* variant detected
Individuals with confirmed DNA variant	Focused cardiovascular evaluation including, but not limited to, physical examination, collection of detailed family history, echocardiogram, electrocardiogram, consideration of stress testing[2,4,5]
Asymptomatic individuals with confirmed DNA variant	Consider routine cardiac screening[3,4]
Individuals with a confirmed DNA variant and signs or symptoms of cardiomyopathy	Treatment is highly individualized, and expert centers should be consulted[3,5] Treatment and management typically follow the American College of Cardiology Foundation/American Heart Association Guidelines[2,3,5]

Patient conversation starters:

It's recommended that people with a DNA variant linked to cardiomyopathy make a plan to evaluate their heart health. This way, any heart problems can be found as early as possible, and a treatment plan can be made. If your genetic test result is confirmed, it's important to work with the right specialists—like a cardiologist and a geneticist—to make a plan that's right for you.

Refer to current guidelines for complete recommendations. Recommendations and guidelines may change over time.

If the test result is confirmed, local centers of excellence in cardiovascular disease should be consulted for further clinical management.

Additional resources

The following patient advocacy groups have additional information and resources about cardiomyopathy:

HCM Association: 4HCM.org

DCM Foundation: DCMFoundation.org

References

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3. Hershberger RE, Givertz MM, Ho CY, et al. Genetic evaluation of cardiomyopathy: a clinical practice resource of the American College of Medical Genetics and Genomics (ACMG). *Genet Med*. 2018;20(9):899-909. Published correction appears in *Genet Med*. 2019;21(10):2406-2409. doi:10.1038/s41436-018-0039-z
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5. Ommen SR, Mital S, Burke MA, et al. 2020 AHA/ACC Guideline for the Diagnosis and Treatment of Patients With Hypertrophic Cardiomyopathy: A Report of the American College of Cardiology/American Heart Association Joint Committee on Clinical Practice Guidelines. *Circulation*. 2020;142(25):e558-e631. Published correction appears in *Circulation*. 2020;142(25):e633. doi:10.1161/CIR.0000000000000937