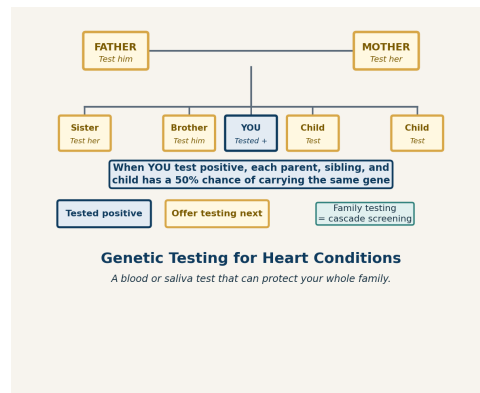


Genetic Testing for Heart Conditions

When It Is Ordered, How It Works, and What Your Result Means

Some heart conditions run in families. A simple blood or saliva test can find the gene change behind them. Testing does not change your DNA — but it can guide your care and protect the relatives who share your genes.



Online: <https://go.riasalimd.com/cardiac-genetics-guide>

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Names & Terms You Will Hear

Term	Meaning
Genetic (DNA) Testing	A lab test that reads your genes to look for a change that can cause a heart condition. Done on blood or saliva.
Inherited Heart Condition	A heart problem caused by a gene change passed down in a family. Also called a genetic or familial heart condition.
Gene	A short set of DNA instructions. A change in one gene can stop the heart from working the right way.
Variant (Mutation)	A change in a gene. Some variants cause disease. Many are harmless. Some we do not understand yet.
Pathogenic Variant	A gene change known to cause disease. This is a 'positive' result. It confirms the diagnosis.
Variant of Uncertain Significance (VUS)	A gene change found in your DNA that we cannot yet call harmful or harmless. It is NOT used to guide your care.
Cascade Screening	Testing your parents, brothers, sisters, and children after a gene change is found in you. Each one has a 50% chance of carrying it.
First-Degree Relative	A parent, a brother or sister, or a child. They share half of your genes. They are tested first in cascade screening.
Genetic Counselor	An expert who explains the test and helps you decide. They also help you understand what your result means for your family.
Genotype-Positive, Phenotype-Negative	You carry the gene change but have no signs of disease yet. You still need regular heart checkups.
GINA	The Genetic Information Nondiscrimination Act. A U.S. law that bars health insurers and employers from using your genetic results.
Penetrance	How likely a gene change is to actually cause disease. Some changes cause disease in nearly everyone; others in only some people.



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Three things to know before you test:

1. **Testing reads your DNA** from blood or saliva — it does not change your genes.
2. **One positive result helps the whole family** — relatives can be tested for that exact change (cascade screening).
3. **Your results are private** — the GINA law bars health insurers and employers from using them.



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Genetic Testing for Heart Conditions?

- Genetic testing reads your DNA to look for a change in a gene that can cause an inherited heart condition. The sample is usually a simple blood draw or a saliva (spit) tube. It does not hurt your DNA.
- Your doctor may order it for a few reasons. The heart condition runs in your family. You were diagnosed young. Or a close relative has an inherited heart problem or died young and suddenly.
- Many heart conditions can be inherited. Common ones are a thick heart muscle (HCM) and a weak, stretched heart (DCM). Others are rhythm problems like long QT and Brugada, and a scarred right-heart muscle (ARVC).
- A gene test can also find other inherited problems. These include inherited high cholesterol, aorta problems like Marfan syndrome, and amyloidosis (a protein that stiffens the heart).
- Your result comes back in one of three ways: positive (a known disease gene change was found), negative (none was found), or VUS (a change was found but we are not sure it matters).
- A positive result does the most good for your family. Once we know the exact gene change, your parents, siblings, and children can be tested for that one change. This is called cascade screening.
- A genetic counselor is part of the process. They explain the test before you decide. They also help you understand the result and what it means for the people you love.
- Your results are private. A U.S. law called GINA stops health insurers and employers from using them. Testing is a tool to guide care and protect family. It is not a label that follows you.



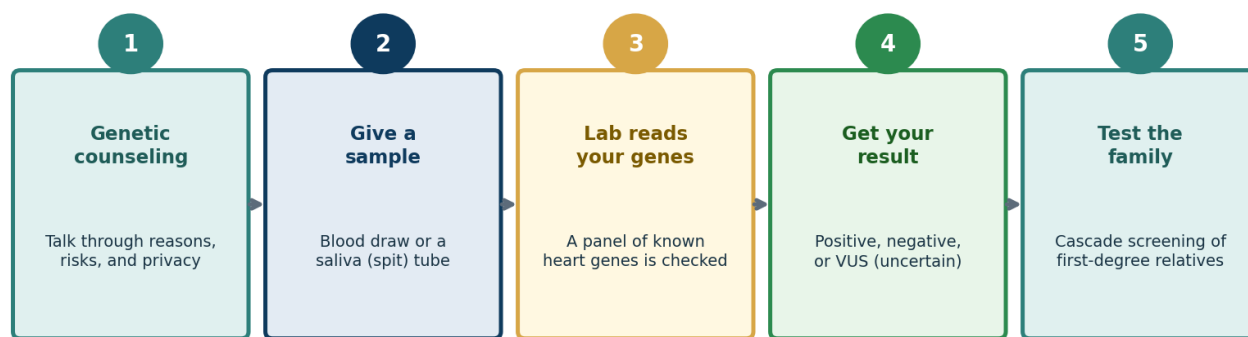
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The Genetic Testing Journey, Step by Step



The testing journey. Step 1: meet a genetic counselor. Step 2: give a blood or saliva sample. Step 3: the lab reads a panel of known heart genes. Step 4: you get a result (positive, negative, or VUS). Step 5: cascade screening tests first-degree relatives.

Source: 2022 EHRA/HRS cardiac genetic-testing consensus.

Inherited heart conditions: common genes and what we watch for

Condition	Common gene examples	What we watch for
Hypertrophic cardiomyopathy (HCM) — thick heart muscle	MYBPC3, MYH7	Fainting, palpitations; risk of dangerous rhythm; sometimes a defibrillator
Dilated cardiomyopathy (DCM) — weak, stretched heart	TTN, LMNA	Heart-failure signs; some genes (LMNA) carry higher rhythm risk
Long QT syndrome — slow electrical reset	KCNQ1, KCNH2, SCN5A	Fainting with exercise or startle; certain drugs are avoided
Brugada syndrome — sodium-channel rhythm problem	SCN5A	Fainting, rhythm events, often during sleep or with fever
ARVC — scarred right-heart muscle	PKP2, DSP	Palpitations, exercise limits; rhythm-event risk
Familial hypercholesterolemia (FH) — inherited high LDL	LDLR, APOB, PCSK9	Very high LDL from birth; early plaque buildup



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Condition	Common gene examples	What we watch for
Inherited aortopathy / Marfan — weak aorta wall	FBN1, TGFB1/2	Aorta widening; imaging and activity limits
Hereditary amyloidosis (hATTR) — stiffening protein	TTR	Stiff heart, nerve and stomach symptoms; gene-targeted drugs exist

The genetic counselor: your guide.

A genetic counselor is an expert who maps your family tree. They explain what a test can and cannot tell you. They review privacy and the GINA law before you decide. After testing, they help you read your result and plan family screening. You can find one at findageneticcounselor.org.



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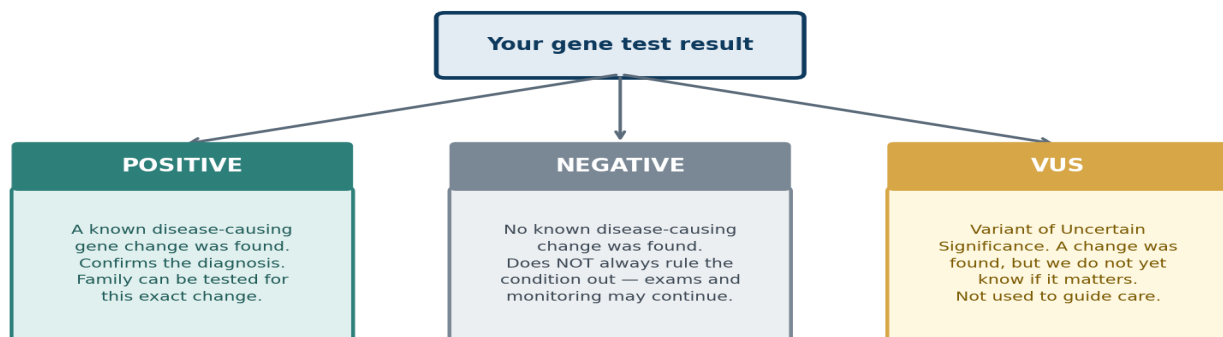


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Why It Matters

- Finding the gene change can confirm a diagnosis when tests like the echo or EKG are not clear. It can turn a 'maybe' into a 'yes,' so the right treatment starts sooner.
- It protects your family. For some inherited heart conditions, the first sign is a sudden cardiac arrest in a young person. Family testing can find at-risk relatives before that ever happens.
- A clear answer can guide big choices. It can show whether you need a defibrillator, which medicines to avoid, whether to limit hard sports, and how often to have heart imaging.
- A negative result in a relative can bring real relief. If you carry a known gene change but your child tests negative for it, that child usually does not need the lifelong monitoring you do.
- It does NOT change everything. A gene result is one piece of the picture. Your symptoms, exam, EKG, echo, and family history still matter. Testing rarely changes care on its own.
- Timing helps families plan. Some couples use results when they think about having children. A genetic counselor can explain the options. There is no pressure.

What Your Result Means: Positive, Negative, or Uncertain (VUS)



What your result means. **POSITIVE**: a known disease-causing gene change was found, confirming the diagnosis and letting family be tested. **NEGATIVE**: none found, but the condition may still be possible. **VUS**: a change of uncertain meaning that is NOT used to guide care. Colors here are neutral on purpose — no result is 'good' or 'bad' by itself. Source: ACMG variant classification, 2015.



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A POSITIVE Result: A Known Disease Gene Change Was Found

- A positive result means the lab found a gene change already known to cause your heart condition. It confirms the diagnosis, often when other tests were not clear.
- It does not start a new medicine on its own. Instead, it guides care: how closely to watch your heart, which drugs or sports to avoid, and whether a defibrillator may help.
- It opens the door to cascade screening. Now your relatives can be tested for the exact same change. Each first-degree relative has a 50% chance of carrying it.
- A positive result does not always mean you will get sick. Some gene changes cause disease in nearly everyone; others in only some carriers. This is called penetrance.
- Keep your lab report. It names the exact gene and variant. Relatives and future doctors will need that exact name to test for the same change.
- Your care team and counselor will make a clear plan: how often to image your heart, what to avoid, and when to involve family. You are not facing it alone.



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A NEGATIVE Result: No Known Change Was Found

- A negative result means the lab did not find a known disease-causing change in the genes it checked. That is often reassuring — but it must be read in context.
- It does not always rule the condition out. Science has not found every gene yet, and some conditions can occur without a change we can detect.
- Your doctor still uses your exam, EKG, echo, and family history. You may still need regular checkups based on what runs in your family.
- A negative result is most powerful when a relative's gene change is already known. If you test negative for the family's exact change, you usually do not need the lifelong monitoring that carriers do.
- If you have strong symptoms or a clear family pattern but a negative test, your doctor may still treat and monitor you as if you have the condition.
- Ask your counselor what your negative result means for you specifically. The answer depends on your family and on whether a family change was already identified.



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A VUS: A Change of Uncertain Meaning

- VUS stands for 'variant of uncertain significance.' The lab found a gene change, but there is not yet enough evidence to call it harmful or harmless.
- A VUS is NOT used to guide your care. Guidelines are clear: doctors should not start tests, devices, or treatment based on a VUS alone.
- Most VUS results are later found to be harmless. Labs re-check them over time as more is learned. A small number are eventually re-classified as disease-causing.
- Because a VUS is uncertain, it should not be used to test relatives the way a known change is. Family testing waits until the meaning is clearer.
- It is normal to feel unsettled by a VUS. A genetic counselor can explain why it does not change your plan and what to watch for as labs learn more.
- Ask to be kept informed. If your VUS is re-classified in the future, your care team can reach out and update your plan.



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Risk Factors

Some patients are more likely to develop this condition. Knowing your risk helps your care team take extra precautions.

Risk Factor	Why It Increases Risk
A close relative has an inherited heart condition	If a parent, sibling, or child has a known gene change, you may be offered testing for that exact change. Each first-degree relative has a 50% chance of carrying it.
Sudden death of a young family member	A sudden death before age 50 with no clear cause can point to an inherited rhythm problem. The risk is higher if it happened during sleep, exercise, or after a loud noise. Family testing may help.
Heart disease found at a young age	Some problems found early in life point to a gene cause. These include a thick or weak heart muscle, a dangerous rhythm, or very high cholesterol.
Unexplained fainting or seizures with exercise or startle	These can be signs of an inherited rhythm condition like long QT. A gene test, along with an EKG, can help find the cause.
A heart condition in several relatives	The same heart problem may show up in more than one generation. That makes an inherited cause more likely. Testing can find it.
Certain physical signs	Some body signs point to an inherited cause. A very tall, long-limbed build can mean Marfan. Tendon cholesterol lumps can mean familial high cholesterol. Nerve and stomach symptoms with a stiff heart can mean amyloidosis.
A known gene change waiting to be checked in you	If a relative's exact gene change is already known, your test is simple and very accurate — we look for that one change only.
Planning a family	Some couples want to know their status before having children. A genetic counselor can explain the choices.



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Treatment Options

- **Start with a genetic counselor.** Before any test, a counselor reviews your family tree. They explain what the test can and cannot tell you. They cover privacy and the GINA law. You decide whether to test. It is always your choice.
- **Give a sample: blood or saliva.** Most tests use a small blood draw. Some use a saliva (spit) tube you can even do at home. Both read the same DNA. Results usually take a few weeks to come back.
- **The lab checks a panel of heart genes.** For most conditions, the lab reads a group of genes known to cause that problem — for example, the genes behind hypertrophic cardiomyopathy or long QT. This is more useful than one gene alone.
- **A positive result confirms the diagnosis.** It does not start a new medicine by itself. Instead, it guides care: how closely to watch your heart, which drugs or sports to avoid, and whether a defibrillator may help down the road.
- **Cascade screening is the big payoff.** Once your gene change is known, relatives can be tested for that one change. Those who carry it start monitoring early. Those who do not are usually reassured and spared extra tests.
- **A negative result is read in context.** If no gene change is found, it does not always rule the condition out. Your doctor still uses your exam, EKG, and echo. You may still need regular checkups based on your family history.
- **A VUS is not acted on.** A 'variant of uncertain significance' is not used to make decisions. Labs re-check these over time. Some are later reclassified as harmless; a few become known causes. Your counselor keeps you posted.
- **Treatment follows the condition, not the test.** Genetic testing points to the right diagnosis. The actual care — medicines, devices, lifestyle steps — comes from each condition's own plan. See our companion guides below.



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Comfort Measures at Home (No Medication Needed)

These simple steps support healing and ease symptoms. Use them alongside any medication your doctor prescribes.

- Bring your family history to the visit. Write down which relatives had heart problems, at what age, and how. A family tree is one of the best tools your counselor has.
- Ask the genetic counselor your questions. There are no silly ones. Good ones include: What will a positive result change for me? What about a negative one? Who in my family should be tested?
- Know your privacy rights. GINA stops health insurers and employers from using genetic results. It does NOT cover life, disability, or long-term care insurance, so ask your counselor about timing.
- Take your time deciding. Testing is almost never an emergency. It is fine to think it over, talk with family, and come back. The choice is always yours.
- Keep a copy of your result. Save the lab report. Future doctors and relatives may need the exact gene and variant name to test for the same change.
- Tell your relatives if you test positive. A short, kind message lets them get tested and protected. Your counselor or doctor can give you a family letter to share.
- Keep up your heart checkups. A gene result does not replace your regular care. Echo, EKG, and clinic visits still find problems early and guide treatment.



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Risks, Benefits, and Alternatives

Every choice has trade-offs. Use this table to start the shared decision conversation.

Option	Risks	Benefits	Alternatives
Genetic testing (after counseling)	Can return a VUS that brings worry without a clear answer. May reveal a condition you did not expect. Rarely, family tension over who shares the gene.	Can confirm a diagnosis and guide care. A known gene change lets the whole family be tested. May prevent a sudden cardiac event in a relative.	Clinical follow-up alone (echo, EKG, exam over time) without a gene test. Test only the symptomatic person first.
Cascade screening of relatives	Relatives must decide whether to know. Some feel anxious while waiting. A positive result means lifelong monitoring.	Finds at-risk relatives before symptoms. Those who test negative for the family change are usually freed from extra testing. Saves lives in rhythm conditions.	Screen relatives with EKG and echo only, without gene testing. Less precise, and may miss carriers who look normal today.
Genetic counseling	Adds a visit before testing. Rarely, raises hard questions about family and future plans.	Helps you make an informed choice, understand your result, and protect your privacy. Reduces confusion and worry.	Order testing without counseling — not advised; raises the chance of a misread result and missed family screening.
Acting on a VUS	A VUS is uncertain. Treating it like a real cause can lead to needless tests, devices, or worry.	None proven. Guidelines say a VUS should NOT change your care.	Wait and watch. Labs re-classify VUS results over time as more is learned. Your counselor will update you.



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Option	Risks	Benefits	Alternatives
No testing	May miss an inherited cause and the chance to protect relatives. A first sign can be a sudden cardiac event.	No cost, no chance of a VUS, no privacy questions to weigh.	Test the person with symptoms first. Then test the family. This is the path we suggest when a condition looks inherited.



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Common Misconceptions

Myth	Reality
"A genetic test will change my DNA."	It will not. The test only reads your DNA to look for a change. It is a blood draw or a saliva tube. Nothing is added or altered. Your genes stay exactly as they are.
"A positive result means I will definitely get sick."	Not always. Some gene changes cause disease in nearly everyone; others in only some people. This is called penetrance. A positive result raises your risk and guides monitoring — it is not a certain sentence.
"A negative result means I am in the clear."	Not always. A negative test does not rule every condition out. We may not know all the genes involved. Your exam, EKG, echo, and family history still matter, and you may still need checkups.
"A VUS means I have the disease."	No. A 'variant of uncertain significance' is a change we cannot yet call harmful or harmless. Guidelines say it should NOT change your care. Most VUS results are later found to be harmless.
"My insurance company can drop me if I test positive."	A U.S. law called GINA bars health insurers and employers from using your genetic results to deny coverage or a job. Ask your counselor about life and disability insurance, which GINA does not cover.
"Only one person in the family needs to be tested."	Once a gene change is found in one person, each first-degree relative has a 50% chance of carrying it. Cascade screening tests them all — and it is the most powerful step after a diagnosis.
"Genetic testing is only for cancer."	Many heart conditions are inherited too — thick or weak heart muscles, dangerous rhythms, inherited high cholesterol, aorta problems, and amyloidosis. A cardiac gene panel checks the genes behind them.



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Myth	Reality
"If I test positive, there is nothing anyone can do."	The opposite is true. Knowing lets your team watch you closely, avoid risky drugs or sports, consider a defibrillator if needed, and protect relatives. Early action prevents harm.



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Possible Complications

Knowing what can go wrong helps you spot problems early. Most complications are uncommon, especially with treatment.

Where / What	What Can Happen
Uncertain result (VUS)	A change is found, but its meaning is unknown. This can cause worry and confusion. A good counselor explains that a VUS should not change your care, and that it may be re-classified later.
False reassurance	A negative result may be read as 'all clear' when the condition is still possible. Your doctor must read the result alongside your exam, EKG, echo, and family history.
Emotional impact	Learning you carry a gene change — or that you passed it on — can be stressful. Counseling and support help. Knowing also brings the power to act early.
Family stress	Results affect relatives, not just you. Some may not want to know. Sharing a result is a personal choice; a counselor can help you approach it gently.
Insurance gaps	GINA protects health insurance and employment, but not life, disability, or long-term care insurance. Timing of those policies is worth discussing before you test.
Unexpected findings	Rarely, a test reveals something you were not looking for. A counselor reviews this possibility with you before you decide, so there are no surprises.

Your privacy and the GINA law.

The Genetic Information Nondiscrimination Act (GINA) is a U.S. law. It bars **health insurers** from using your genetic results to deny coverage or raise your premium, and bars **employers** from using them in hiring or firing. GINA does **not** cover life, disability, or long-term care insurance — so ask your counselor about the timing of those policies before you test.



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Points to Know

If you remember nothing else, remember these key points.

- Genetic testing reads your DNA from blood or saliva. It does not change your genes. It looks for a change that can cause an inherited heart condition.
- Many heart conditions are inherited: thick or weak heart muscles, long QT and Brugada rhythm problems, ARVC, familial high cholesterol, Marfan aorta disease, and hereditary amyloidosis.
- Results come in three forms: positive (a disease gene change found), negative (none found), and VUS (a change found but its meaning is unknown). A VUS is not used to guide care.
- The biggest payoff is for family. Once your gene change is known, relatives can be tested for it. Each first-degree relative has a 50% chance of carrying it.
- A genetic counselor guides the whole process — before, during, and after. They help you decide and explain what your result means.
- Your results are private. GINA bars health insurers and employers from using them. It does not cover life, disability, or long-term care insurance.
- Testing guides care; it rarely changes it alone. Your symptoms, exam, EKG, echo, and family history still matter.
- Testing is your choice and is almost never urgent. Take your time, ask questions, and talk it over with people you trust.



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When to Call Us - and When to Call 911

If you are not sure, call. We would rather hear from you twice than miss a real problem.

- **Call 911** for fainting with no warning, a seizure during exercise or a loud startle, or sudden chest pain or trouble breathing. In inherited rhythm conditions these can be danger signs. Do not drive yourself.
- **Call our office (727-943-5200)** if a close relative was found to have an inherited heart condition or a known heart gene change. We can arrange testing for you for that exact change.
- **Call us** if a family member died suddenly and unexpectedly before age 50, especially during sleep, exercise, or after a startle. We can review whether family testing makes sense.
- **Call us** if you were diagnosed young with a thick or weak heart muscle, a dangerous rhythm, or very high cholesterol. Genetic testing may help confirm the cause.
- **Call us** if you have a genetic result you do not understand. We will go over it with you, or connect you with a genetic counselor.
- **Call us** if you carry a known gene change and have new symptoms — fainting, palpitations, chest pain, or breathlessness. These may mean the condition is now active.
- **Call us** if you want to talk through testing before starting a family. We can refer you to a counselor to review your options.

Office: (727) 943-5200

See also: companion guides from Dr. Ali.

[Familial Hypercholesterolemia](#) · [Hypertrophic Cardiomyopathy](#) · [Long QT Syndrome](#) · [Brugada Syndrome](#) · [Cardiac Sarcoidosis](#).



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Trusted Resources

The links below are the trusted resources Dr. Ali used to write this guide and that he recommends you visit for more information. Each is a clickable link in the digital version and a Short.io tracked URL on the printed version.

- [MedlinePlus — Genetic Testing \(NIH\)](#) - Federal plain-language overview of what genetic testing is, how it is done, and what the different results mean.
- [NHGRI — Genetic Discrimination and the GINA Law](#) - Official explanation of the GINA law: what it protects (health insurance and jobs) and what it does not (life, disability, and long-term care insurance).
- [Find a Genetic Counselor \(NSGC\)](#) - Search tool from the National Society of Genetic Counselors to locate a certified genetic counselor near you.
- [American Heart Association — Genes and Heart Disease](#) - AHA patient overview of inherited heart conditions and why family screening matters.
- [Cleveland Clinic — Genetic Testing](#) - Plain-language guide to who should consider testing, how it works, and how to read a result.
- [Mayo Clinic — Genetic Testing](#) - Mayo's patient page on how testing is performed, result types, and the role of genetic counseling.
- [MedlinePlus — What is Genetic Counseling?](#) - Explains what a genetic counselor does and how a counseling visit helps before and after testing.
- [Familial Hypercholesterolemia — Companion Guide](#) - Dr. Ali's guide to inherited high cholesterol — the most common inherited heart-risk condition, and a prime use for cascade screening.
- [Hypertrophic Cardiomyopathy — Companion Guide](#) - Dr. Ali's guide to the thick-heart-muscle condition — a leading reason cardiac genetic testing is ordered.
- [Long QT Syndrome — Companion Guide](#) - Dr. Ali's guide to this inherited electrical rhythm condition, where genetic testing helps confirm the type and guide treatment.



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Sources Used to Build This Guide

Every recommendation in this guide is grounded in one or more of the studies, guidelines, or trusted resources below. Short links are tracked so Dr. Ali can see which references patients are reading; nothing personally identifying is captured.

- [Cleveland Clinic — Genetic Testing for Heart Disease](#) [Patient-Education]
Who should consider cardiac genetic testing and what results mean
- [AHA — Genetics and Cardiovascular Disease](#) [Patient-Education]
Inherited heart conditions and family screening
- [MedlinePlus — Genetic Testing](#) [Patient-Education]
Plain-language overview of the genetic testing process
- [2022 EHRA/HRS/APHRS/LAHRs Expert Consensus on Cardiac Genetic Testing](#) [Guideline]
Current consensus on when to test, gene panels, and counseling for channelopathies and cardiomyopathies
- [HRS/EHRA Expert Consensus — Genetic Testing for the Channelopathies and Cardiomyopathies \(2011\)](#) [Guideline]
Foundational consensus on which inherited conditions have an established testing yield
- [ACMG — Standards for Interpretation of Sequence Variants \(Richards 2015\)](#) [Guideline]
Five-tier variant classification: pathogenic, likely pathogenic, VUS, likely benign, benign
- [NHGRI — Genetic Discrimination and the GINA Law](#) [Government]
GINA scope: protects health insurance and employment; does not cover life, disability, or long-term care insurance
- [Mayo Clinic — Genetic Testing](#) [Patient-Education]
How testing is performed, result types, and the role of genetic counseling
- [MedlinePlus — What is Genetic Counseling?](#) [Patient-Education]
What a genetic counselor does before and after testing
- [NSGC — Find a Genetic Counselor](#) [Patient-Education]
Patient-facing directory to locate a certified genetic counselor
- [AHA — Cardiomyopathy and Family Screening](#) [Patient-Education]
Cascade screening of first-degree relatives in inherited cardiomyopathy



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About This Guide

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Reviewer note: Reviewed Cleveland Clinic, Mayo Clinic, AHA, NIH MedlinePlus, and the 2022 EHRA/HRS/APHRS cardiac genetic-testing consensus. This guide adds: a cascade-screening family tree; a positive/negative/VUS result-meaning flow with severity-neutral colors; a step-by-step testing journey; an inherited-conditions table linking each condition to its common genes; and plain-language sections on the genetic counselor's role and the GINA privacy law that most patient pages omit.

Disclaimer

This guide is for educational purposes only and does not replace medical advice from your physician. Recommendations may not apply to every patient. Always follow the specific instructions of Dr. Ali and your care team. In an emergency call 911 or go to the nearest emergency department.



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