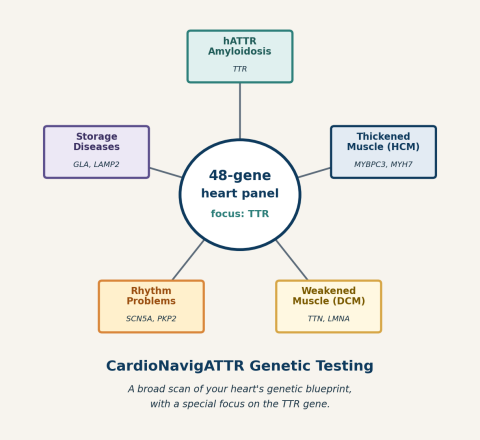
Understanding CardioNavigATTR Testing

A No-Cost Heart Gene Test, with a Special Focus on TTR

A sponsored (no-charge) cheek-swab gene test offered in our office. It scans 48 heart genes, mainly to check for hATTR amyloidosis.



Online: https://go.riasalimd.com/cardionavigattr-guide

Rias KS Ali, MD FACC

Board Certified in Interventional Cardiology & Cardiovascular Disease

4740 Mile Stretch Drive, Holiday FL 34690 | Office: (727) 943-5200 | Fax: (727) 943-5201 | RiasAliMD.com

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Names & Terms You Will Hear

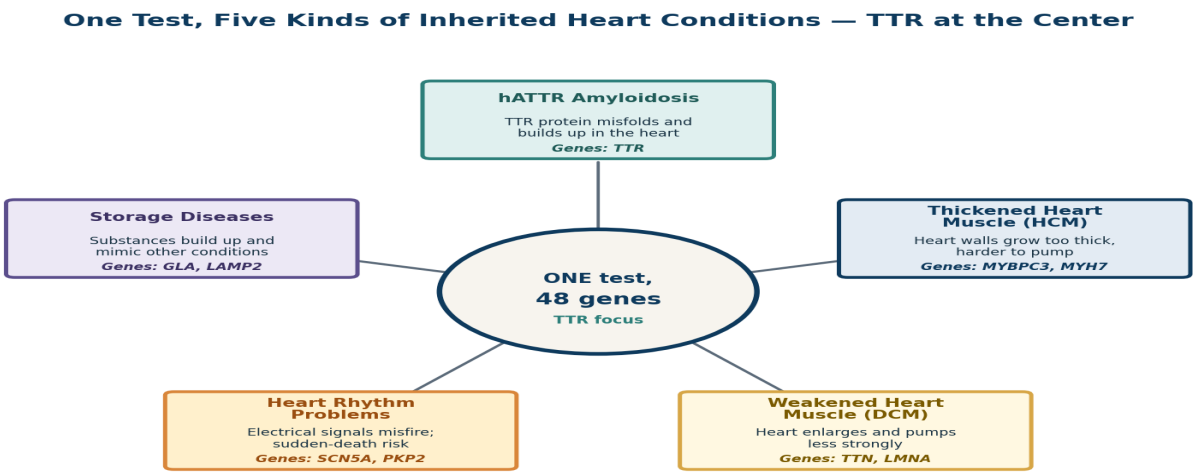
Term	Meaning
CardioNavigATTR Panel	A sponsored (no-charge) heart gene test. It scans many genes linked to inherited heart conditions, with a special focus on the TTR gene.
Sponsored / No-Charge Testing	The test and the genetic counseling are paid for by the program's sponsor (the NavigATTR program). There is no cost or bill to you.
PreventionGenetics	The lab that runs the test. It is part of Exact Sciences. The lab reads your genes from the cheek-swab sample we collect.
hATTR Amyloidosis	Hereditary transthyretin amyloidosis. A protein called TTR folds the wrong way and builds up in the heart and/or nerves.
Transthyretin (TTR)	A protein made by the liver. The TTR gene holds its instructions. A change in that gene can cause hereditary ATTR. It is this test's focus.
HCM (Thickened Heart Muscle)	Hypertrophic cardiomyopathy — the heart walls grow too thick. The panel checks genes like MYBPC3 and MYH7.
DCM (Weakened Heart Muscle)	Dilated cardiomyopathy — the heart enlarges and pumps less strongly. The panel checks genes like TTN and LMNA.
Storage Disease	A condition where substances build up inside heart cells and mimic other problems — such as Fabry (GLA gene) or Danon (LAMP2 gene).
Buccal (Cheek) Swab	A soft swab rubbed on the inside of your cheek to collect cells. It is painless and needs no needle.
Genetic Counseling	A confidential talk that explains the test before and your result after. Strongly recommended and included with the program.
Cascade Testing	Testing close relatives after a gene change is found in you. The right steps depend on which gene is involved.

Term	Meaning
Variant of Uncertain Significance (VUS)	A spelling change was found in your DNA, but it is not yet known if it matters. It is not a diagnosis and is not used to guide care.
GINA	A U.S. law that bars health insurers and employers from using your genetic results. It stands for the Genetic Information Nondiscrimination Act.

An honest word about the name. "CardioNavigATTR" sounds TTR-focused, and the program is backed by the makers of TTR drugs. But the test itself is a **broad cardiac gene panel** — it can find changes tied to thickened or weakened heart muscle, rhythm problems, and storage diseases, not just TTR. That is why your consent and counseling should cover the **whole scope** of the test, not amyloidosis alone.

What Is CardioNavigATTR Testing?

- CardioNavigATTR is a sponsored heart gene test you can have done right in our office. **Sponsored means no charge** — the test and the genetic counseling are paid for by the program. There is no bill to you.
- Think of it as **a broad scan of your heart's genetic blueprint, with a special focus on the TTR gene**. It is ordered mainly to check for hATTR amyloidosis — but it checks other inherited heart conditions too.
- The test reads **48 genes** linked to inherited heart conditions. (The program is listed as about a 36-gene panel. The current lab version reports 48 — a panel that has grown over time. Your lab report lists the exact genes checked.)
- Those genes group into **five kinds of inherited heart conditions**: hATTR amyloidosis, thickened heart muscle (HCM), weakened heart muscle (DCM), heart-rhythm problems, and storage diseases that mimic others.
- The **TTR gene** is the special focus. In hATTR amyloidosis, the TTR protein folds the wrong way and builds up in the heart and nerves. That is the main reason the test is usually ordered.
- The lab is **PreventionGenetics**, part of Exact Sciences. It runs the panel on a method called PGxome (next-generation sequencing) in a CLIA-certified lab. Your doctor orders the test; the lab reads your genes.
- The sample is a **cheek (buccal) swab** — a soft swab rubbed inside your cheek. No blood draw is needed. We collect it in the office and mail it to the lab.
- Results usually come back in about **2 to 3 weeks**. A genetic counselor and your cardiologist help you understand what your result means for you and your family.



One test, five kinds of inherited heart conditions, with TTR at the center. The panel reads 48 genes — not TTR alone.

The five kinds of inherited heart conditions this panel checks

Category	What it means	Key genes
hATTR Amyloidosis	A protein (TTR) misfolds and builds up in the heart and nerves.	TTR
Thickened Heart Muscle (HCM)	Heart walls grow too thick, making it harder to pump.	MYBPC3, MYH7
Weakened Heart Muscle (DCM)	Heart enlarges and pumps less strongly.	TTN, LMNA
Heart Rhythm Problems	Electrical signals misfire, raising sudden-death risk.	SCN5A, PKP2
Storage Diseases	Substances build up in heart cells and mimic other conditions.	GLA (Fabry), LAMP2 (Danon)

What Is hATTR Amyloidosis?

- hATTR stands for **hereditary transthyretin amyloidosis**. A protein made by the TTR gene folds the wrong way and builds up in the heart and/or nerves over time.
- In the heart, it makes the walls **thicken and stiffen**, so the heart cannot relax and fill the way it should.
- In the nerves, it can cause **numbness, tingling, or weakness** in the hands and feet. Carpal tunnel in both hands — often years before heart symptoms — is a classic early clue.
- This is why the test focuses on the TTR gene: catching hATTR matters because there are now **FDA-approved medicines** — like tafamidis — that slow it down if it is diagnosed.
- If your TTR gene shows no known disease-causing change, hereditary ATTR is unlikely. Any ATTR you do have would more likely be the age-related (wild-type) form, which is managed differently.

Why It Matters

- Knowing the cause changes care. Finding a gene change can **confirm why your heart is affected** and point to care aimed at that specific condition instead of treating symptoms alone.
- For hATTR, there are real treatments. **TTR stabilizers** like tafamidis and acoramidis help the protein hold its shape. **TTR silencers** lower how much TTR the body makes. Finding the cause early opens the door to them, before the heart is badly affected.
- Because the panel is broad, it can also explain **other inherited conditions** — such as thickened heart muscle (HCM) or weakened heart muscle (DCM). Each gene has its own care and family-testing plan.
- It protects your family. Many inherited heart conditions run in families. A clear gene finding means relatives may **benefit from testing too** — the right steps depend on which gene is involved.
- Found early, at-risk relatives can be **watched over time**. If a condition ever starts, care can begin sooner.
- You do not face this alone. The counselor explains the full scope of the test. A privacy law called GINA guards your results from health insurers and employers.
- A negative or uncertain result is still useful. But it does **not** fully rule out an inherited condition. Your doctor reads it with your history, your symptoms, and your other heart tests.

Your result, what it means, and the next step

Result	What it means	Next step
Positive	A disease-causing change was found — in TTR or another panel gene. This points to an inherited heart condition.	Plan care aimed at that condition. Offer gene-specific testing to relatives.
Negative / Indeterminate	No clear disease-causing change was found. This does not fully rule out an inherited condition.	Keep following your heart based on symptoms and other tests.
Uncertain (VUS)	A 'spelling change' was found, but it is not yet known if it matters. Not a diagnosis.	The counselor explains it. It is tracked over time and not used to guide care for now.

Your privacy is protected. A U.S. law called **GINA** bars health insurers and employers from using your genetic results. Its full name is the Genetic Information Nondiscrimination Act. Your counselor can explain what it covers.

What Each Result Type Means

POSITIVE	NEGATIVE / INDETERMINATE	UNCERTAIN (VUS)
• A disease-causing change was found.	• No clear disease-causing change was found.	• A 'spelling change' we cannot yet judge.
• It may be in TTR or in another panel gene.	• Lowers the odds of an inherited cause.	• It is NOT a diagnosis.
• Confirms the cause and guides gene-specific care.	• Does NOT fully rule one out.	• Common with broad panels.
• Opens cascade testing for relatives.	• Your doctor reads it with your other tests.	• Tracked over time; you are told if it changes.

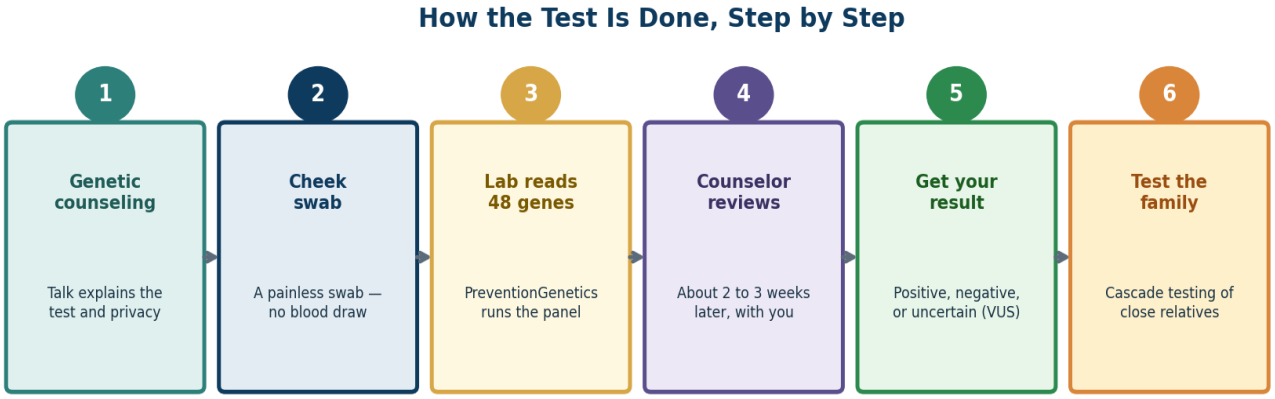
Risk Factors

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
Diagnosed or suspected cardiac amyloidosis	An abnormal PYP (pyrophosphate) bone scan, a heart biopsy showing amyloid, or strong suspicion of ATTR makes you a candidate.
Unexplained thick heart with heart failure (HFpEF)	Thick heart walls with a normal squeeze and heart-failure symptoms (preserved ejection fraction) are a key red flag for ATTR.
Carpal tunnel in both hands with heart disease	Carpal tunnel in both hands — often years before heart symptoms — paired with heart disease is a classic ATTR warning sign.
A relative with hereditary ATTR	If a close relative carries a TTR gene change, you may carry it too. Testing tells you your status.
Two or more amyloid 'red-flag' features	Lower-back spinal stenosis, a biceps tendon rupture, nerve symptoms, or a low-voltage EKG with thick walls can add up to qualify you.
West-African or Caribbean ancestry with heart findings	The V122I TTR change is more common in these groups. With heart findings, testing may be especially worthwhile.

Treatment Options

- **Step 1 — Genetic counseling first.** Before testing, a genetic counselor explains the **full scope** of the test — not just TTR, but all the heart conditions it can find — what a result could mean, and how your privacy is protected. Counseling is strongly recommended and included at no charge.
- **Step 2 — The cheek swab.** In the office, we rub a soft swab inside your cheek to collect cells. It is quick and painless. There is **no blood draw and no needle**.
- **Step 3 — Mail to the lab.** We package the swab and mail it to PreventionGenetics. You do not have to do anything else.
- **Step 4 — The lab reads 48 genes.** The lab runs the panel using next-generation sequencing. Results usually come back in about **2 to 3 weeks**.
- **Step 5 — Review your result with a counselor.** A genetic counselor and your cardiologist go over the result with you — what it means for your treatment and what it means for your family.
- **Step 6 — Act on the result.** If a gene change is found, we plan care aimed at that specific condition and offer the right family testing. If the result is negative or uncertain, we keep following your heart based on your symptoms and other tests.
- **This is a test, not a treatment.** The test looks for a cause. The treatments — such as tafamidis for hATTR — are separate. Your doctor decides what is right for you.



The path from counseling to family testing. The cheek swab is collected in the office; results come back in about 2 to 3 weeks and are reviewed with a genetic counselor.

A real buccal (cheek) swab being collected — the actual method used for CardioNavigATTR testing. No blood draw and no needle. Image: U.S. Air Force photo by SSgt. Nicole Leidholm / DVIDS (Public Domain).

The In-Office Cheek Swab and Genetic Counseling

- The sample is a **buccal (cheek) swab** — a soft swab rubbed inside your cheek. It is painless, takes a moment, and needs **no blood draw and no needle**.
- We collect the swab in the office and mail it to PreventionGenetics. The lab runs the 48-gene panel using next-generation sequencing in a CLIA-certified lab. You do not have to do anything else.
- **Genetic counseling is strongly recommended** and included. Before the test, the counselor explains the **full scope** — that it checks far more than TTR — what a result might mean, and your privacy. After, they help you understand your result.
- Counseling is **confidential**. It is a conversation to help you and your family, not a record that follows you around. GINA also protects your results from health insurers and employers.
- Results usually return in about **2 to 3 weeks**. Your cardiologist and the counselor review them with you together.

Risks, Benefits, and Alternatives

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

Option	Risks	Benefits	Alternatives
CardioNavigATTR gene test (cheek swab)	A result can cause worry. Because the panel is broad, it can find changes in genes beyond TTR, and uncertain results (VUS) are common. A positive result raises questions for relatives. Rarely, the sample must be repeated.	No cost and no blood draw. Can confirm hATTR or another inherited condition, guide care, and let family get tested. Counseling is included.	Blood or saliva sample instead of a swab. A different genetics lab. A narrower (TTR-only) test. Choosing not to test.
Genetic counseling	Takes time. Some find the questions about family and the full scope of the test hard to think about.	Explains the **whole panel** in plain words, protects your privacy, and helps you and your family understand the result. No charge.	Reviewing results with your cardiologist alone. Declining counseling.
Family (cascade) testing of relatives	Relatives must choose for themselves. A positive result in a relative brings its own news and decisions.	Finds at-risk relatives early. A negative relative may avoid years of monitoring. Helps families plan ahead.	Watching relatives with heart imaging without gene testing. Testing only those with symptoms.
Not testing	The cause of the heart problem may stay unknown. Relatives are not warned. Treatment may be delayed.	No test, no result to process. Some people prefer to wait.	Testing now. Testing later when you feel ready. Counseling-only first.

Common Misconceptions

Myth	Reality
"This test only checks for amyloidosis."	No. The name sounds TTR-focused, but it is a **broad heart-gene panel**. It checks 48 genes across five kinds of inherited heart conditions — including thickened and weakened heart muscle, rhythm problems, and storage diseases — not TTR alone.
"Sponsored testing must have a hidden cost."	There is no cost to you. The program's sponsor pays for the test and the genetic counseling. You will not get a bill for it.
"A gene test means a blood draw."	Not here. CardioNavigATTR uses a cheek (buccal) swab. It is painless and needs no needle. We collect it in the office.
"A VUS means I have a heart condition."	No. A variant of uncertain significance is like a word spelled slightly differently in your DNA — we do not yet know if it changes the story. It is **not a diagnosis**, and it is not used to guide your care. VUS are common with broad panels.
"A negative test means I do not have an inherited condition."	Not exactly. A negative or indeterminate result lowers the odds but does **not** fully rule one out. Some DNA changes are not detected by this kind of test, and science keeps evolving.
"My genetic result could be used against me by insurance."	A U.S. law called GINA bars health insurers and employers from using your genetic results. Your genetic counselor can explain what GINA covers.
"This single test treats my heart."	The test looks for a cause; it does not treat anything. If a condition like hATTR is found, treatments such as tafamidis are a separate step your doctor decides on.

Possible Complications

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

Where / What	What Can Happen
More uncertain results (VUS)	Because this is a broad 48-gene panel, it finds more variants of uncertain significance than a single-gene test. A counselor explains them. A VUS is not a diagnosis and is not used to guide care.
Unexpected findings beyond TTR	The panel can turn up a change in a gene for thickened or weakened heart muscle, a rhythm problem, or a storage disease — not just TTR. This is why counseling covers the whole panel before you test.
Worry while you wait	Waiting 2 to 3 weeks for a result can feel stressful. Your care team and counselor are here for your questions.
Family news that is hard to share	A positive result means relatives may carry the same change. Deciding how and when to tell family can be difficult. Counseling helps.
A negative test is not a full all-clear	A negative or uncertain result does not fully rule out an inherited condition. Your doctor still follows your heart based on your other tests.
Eligibility is decided by your doctor	Not everyone qualifies for the sponsored program. Your doctor decides if you meet the program's criteria.

What Happens Next — and What This Test Cannot Tell Us

- **Genetic counseling** is the first next step — strongly recommended — to put your result in the context of your health, symptoms, and family history.
- **Family awareness and cascade testing** come next when a gene change is found. This is **gene-specific**: a non-TTR finding follows that gene's own testing plan, not the TTR plan.
- **Follow-up heart tests** — such as an echocardiogram, MRI, or ECG — may be ordered based on your symptoms, whatever the result.
- **A VUS is monitored.** As research grows, an uncertain variant may be reclassified. If that happens, you will be notified.
- **What this test cannot tell us:** a negative or uncertain result does not fully rule out an inherited condition. The test does not catch every kind of DNA change. A few types are missed — repeat expansions, certain large rearrangements, changes in the cell's mitochondrial DNA, and low-level mosaics. Your doctor reads the result with your history and exam.

Questions to Ask Your Doctor

- "This test checks many genes — what conditions besides hATTR does it look for, and what would each mean for me?"
- "If a variant of uncertain significance is found, what does it mean for my day-to-day life?"
- "Should my family members be tested, and would the steps depend on which gene is involved?"
- "Do I need any additional heart tests — like an echocardiogram, MRI, or ECG — based on my result?"
- "How will I be notified if the meaning of an uncertain variant changes over time, and is a genetic counselor referral right for me?"

Points to Know

If you remember nothing else, remember these key points.

- CardioNavigATTR is a no-cost, sponsored cheek-swab gene test offered in our office. The lab is PreventionGenetics.
- It is a **broad scan** of 48 heart genes — with a special focus on the TTR gene — not a TTR-only test.
- It checks five kinds of inherited heart conditions: hATTR amyloidosis, thickened muscle (HCM), weakened muscle (DCM), rhythm problems, and storage diseases.
- A positive result can confirm a cause and guide care; a negative or uncertain result does not fully rule out an inherited condition.
- A VUS is a 'spelling change' we cannot yet judge — not a diagnosis. VUS are common with broad panels and are tracked over time.
- Genetic counseling — covering the whole panel — is strongly recommended before and after the test, and your results are confidential.
- Family testing is **gene-specific**: the right steps depend on which gene is involved. GINA protects your results; your doctor decides eligibility.

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 our office** if you want to know whether the CardioNavigATTR sponsored test is right for you. Your doctor decides eligibility.
- **Call our office** if you have questions about your result, or if you would like to speak with the genetic counselor again.
- **Call our office** if a relative was found to have an inherited heart condition and you want to arrange family (cascade) testing.
- **Call our office** if your heart symptoms change — more shortness of breath, swelling in the legs, or a 3-pound weight gain in 2 days.
- **Call our office** before stopping or starting any heart medicine based on something you read about amyloidosis or genetic testing.
- **Call 911** for fainting, severe shortness of breath, or chest pain that does not go away.

Office: (727) 943-5200

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.

- PreventionGenetics — CardioNavigATTR Program - The official page for the sponsored testing program and gene list.
- Amyloidosis Foundation - Patient-friendly information on hereditary ATTR amyloidosis.
- Hypertrophic Cardiomyopathy Association - Patient support and information on thickened heart muscle (HCM).
- National Society of Genetic Counselors — Find a Counselor - Find a genetic counselor and learn what counseling involves.
- Our Cardiac Amyloidosis guide - Learn more about ATTR and AL amyloidosis and how they are treated.
- Our Tafamidis guide - Details on the main pill used to treat hATTR cardiac amyloidosis.
- Our Genetic Testing for Heart Conditions guide - How inherited heart conditions are tested and what results mean.
- Our Inherited Heart Conditions guide - An overview of heart conditions that can run in families.
- Our Heart Failure with Preserved EF (HFpEF) guide - A common reason ATTR testing is considered — stiff heart with normal squeeze.

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.

- PreventionGenetics — CardioNavigATTR Sponsored Testing Program *[Patient-Education]*
  Official program page for the sponsored, no-charge genetic testing + counseling program offered in the office; confirms the lab (PreventionGenetics/Exact Sciences), the buccal-swab sample, ~2-3 week turnaround, CLIA-certified PGxome NGS platform, and the gene panel (listed as ~36 genes; current report version covers 48)
- NavigATTR Sponsored Testing Program (AstraZeneca / Ionis) *[Patient-Education]*
  Describes the sponsor-funded NavigATTR program (AstraZeneca/Ionis, makers of TTR-directed therapies) that pays for the test and genetic counseling — the basis for the 'no cost to you' framing and the honest note that the sponsor's focus is hATTR while the panel scope is broader
- AHA — Genetic Testing and Counseling for Hereditary Transthyretin Amyloidosis (hATTR) *[Guideline]*
  American Heart Association scientific statement on genetic testing and counseling for hATTR — supports the 'counseling before and after, strongly recommended' framing and cascade (family) testing for TTR variants
- HFSA Practice Guideline — Genetic Evaluation of Cardiomyopathy (2018) *[Guideline]*
  Heart Failure Society of America practice guideline on genetic evaluation of cardiomyopathy — establishes that broad cardiomyopathy panels (HCM, DCM, ACM, storage) require pre/post-test counseling and gene-specific cascade testing; the evidence base for treating CardioNavigATTR as a broad panel, not TTR-only
- Genetics in Hypertrophic Cardiomyopathy (Cardiac Failure Review) *[Reference]*
  Reviews the sarcomere genes (MYBPC3, MYH7) most often responsible for thickened heart muscle (HCM) — supports the HCM category of the five-category panel table and the 'most common HCM genes' framing
- Genetic Architecture of Sudden Cardiac Death (MDPI, Genes) *[Reference]*
  Reviews the rhythm/channelopathy and arrhythmogenic-cardiomyopathy genes (SCN5A, PKP2 and others) tied to sudden-death risk — supports the 'heart rhythm problems' category of the panel and the sudden-death framing
- ICER — Disease-Modifying Therapies for Transthyretin Amyloidosis (ATTR) *[Reference]*
  Independent review of TTR stabilizers (tafamidis, acoramidis) and TTR silencers — supports the 'finding the cause opens the door to FDA-approved treatments that slow progression' framing for hATTR
- Patient Perspectives on Genetic Testing in TTR Amyloidosis (Journal of Cardiac Failure) *[Reference]*
  Patient-voice data on how people experience TTR genetic testing — informs the plain-language counseling, privacy, and family-communication framing and the 'worry while you wait' and 'hard to share' complication notes
- 2023 ACC Expert Consensus Decision Pathway for Cardiac Amyloidosis (Kittleson, JACC) *[Guideline]*
  Current ACC pathway on diagnosing transthyretin cardiac amyloidosis (ATTR-CM), the red-flag features (HFpEF with thick walls, bilateral carpal tunnel, abnormal PYP scan), and TTR genotyping to separate hereditary (hATTR/ATTRv) from wild-type — the eligibility and 'who qualifies' framing
- NIH GeneReviews — Hereditary Transthyretin Amyloidosis (hATTR / TTR gene) *[Guideline]*
  Authoritative reference on the TTR gene, V122I (p.Val142Ile) and other variants, autosomal-dominant inheritance (50% per first-degree relative), heart-and-nerve involvement, and cascade testing of at-risk relatives

- U.S. EEOC — Genetic Information Nondiscrimination Act (GINA) *[Reference]*
  Authoritative source on the GINA privacy protections that bar health insurers and employers from using genetic test results — the privacy reassurance in the guide

About This Guide

Rias KS Ali, MD FACC | Board Certified in Interventional Cardiology & Cardiovascular Disease

4740 Mile Stretch Drive, Holiday FL 34690 | Office: (727) 943-5200 | Fax: (727) 943-5201 | RiasAliMD.com

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Reviewer note: CardioNavigATTR is a sponsored program: the lab is PreventionGenetics (an Exact Sciences company), run through the NavigATTR program backed by AstraZeneca and Ionis, the makers of TTR drugs. We reviewed the official PreventionGenetics program page, the AHA statement on genetic testing for hATTR, the HFSA guideline on genetic evaluation of cardiomyopathy, and the 2023 ACC consensus on cardiac amyloidosis. This guide adds a plain-language orientation the program does not: that this is a broad heart-gene panel (not TTR alone), what each gene category and result type means, why counseling covers the full scope, and how gene-specific family testing works.

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.

Take This Guide With You

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