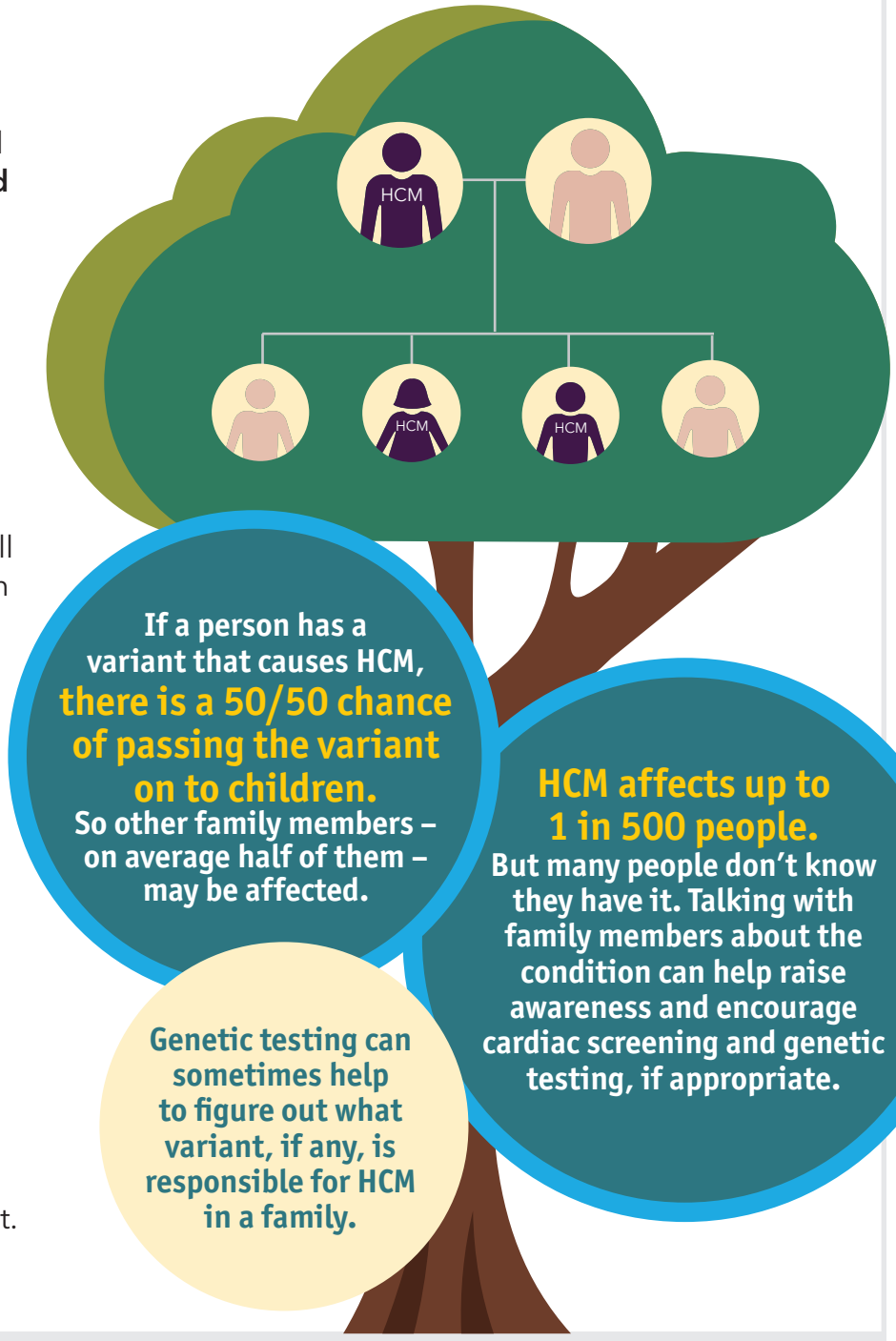
CardioSmart
American College of Cardiology

HCM family screening and genetic testing: What you need to know

Hypertrophic cardiomyopathy (HCM), which can lead to a thickening of the heart muscle and other complications, is often passed down in families. **If you've been told you have HCM, this handout will help you understand more about how this condition happens, and screening and genetic testing for your family.**

We have over 20,000 genes. These genes, which act like instruction manuals, shape what we look like, how tall we are, as well as our overall health and risk for disease. HCM can happen if there is a typo or change (called a variant) in one of the many genes involved with heart muscle growth. These changes can then be passed down to children.

What's interesting is that HCM can look and feel very different even among family members. Some people have clear signs or symptoms, while others may not have any symptoms at all. And not everyone will develop HCM. Still, knowing whether someone has inherited a variant for HCM and could be at risk of developing related health problems is important.



What do genetic test results mean?

Because HCM runs in families, once one person is diagnosed it is important to talk about family screening and share information about the condition – especially with parents, brothers, sisters, and children. The steps they should take will depend on family history of HCM and the results of your genetic test if you had one. Think of it as:

Step 1. If you have HCM, genetic testing can help to figure out the gene involved.

Step 2: If your genetic test is positive, then family members can be tested for the specific gene change that was found in you.

If you have HCM, **your genetic test results** will guide next steps for family members.

Then your family members should ...			
	Talk with your HCM team about how to monitor or follow up; there could be a gene for HCM that current testing doesn't pick up yet.	Have genetic testing to see if they carry the same variant. If they do, then additional testing and imaging to determine if they also have HCM.	Check back with your HCM team over time to see if interpretation of the gene variant changes as more information is available.

** Always be sure to seek care for any heart symptoms even if a genetic test is negative.*

What is the difference between screening and genetic testing?

Screening usually includes:	Genetic testing involves:
• Physical exam by a heart doctor who knows about HCM	• Meeting with a genetic counselor
• Thorough health history	• Giving a sample of blood or saliva
• Electrocardiogram + echocardiogram, which are repeated every 1-5 years for adults (interval varies and is individualized for each patient), and once a year for children and adolescents even if they feel OK and don't have symptoms. These are continued over time as HCM may not develop until later.	• A lab runs tests to look for the gene variant found in your family member.

Whether a family member undergoes genetic testing is a personal decision. A genetic counselor can help you understand the test and weigh the risks and benefits.

What is a genetic counselor?

A genetic counselor can help educate you about how HCM is passed down in families, when screening and genetic testing is needed, and what the results might mean for you and your family.

Genetic counselors have specific training in genetics and counseling and can help:

- Review the benefits and limitations of genetic testing
- Explain how the test results will be used
- Give advice on how to talk with family members about HCM and the need for screening or testing even if they feel OK
- Determine the next person in your family who should be tested and the right test to use (for example, if an unaffected family member tests negative then their children may not need to be tested)

Benefits and drawbacks of genetic testing

+ Positives or Benefits	– Negatives or Challenges
• Identifies the cause of HCM in some families. • Helps you and your family members get early monitoring and treatment. • Gives you and your health care team more information to guide screening, treatment recommendations and decisions. • Prevents unnecessary tests, follow-up and worry when someone is found not to have the gene variant. • Helps inform family planning/decisions about having children.	• Learning you or a family member has HCM can be difficult; you may feel sad, anxious, guilty, or depressed. • Some people worry about getting life insurance or disability insurance. (Note: There is a law that protects for health insurance and employment.) • A positive result does not necessarily change your treatment recommendations. • While there are some genes commonly linked to HCM, genetic tests don't always detect a variant for HCM. There are many genes that don't have strong evidence for being linked to HCM, but researchers are learning more over time.

Questions to ask

- Is genetic testing right for me
- Who in my family needs to be screened or tested? When and how often?
- How might the results affect me or my family emotionally?
- How can a genetic counselor help?
- What is the risk of passing the variant on to my children?
- How can I talk with my family about HCM and the need for screening or genetic testing?
- How long does it take to get genetic test results?
- Who will have access to my genetic test results?
- Who will pay for screening or genetic testing?
- Do I need to be worried about getting life insurance for myself or my family members?

Ask for help in talking with family

It's not always easy to talk about inherited conditions, and many parents say they feel guilty about the possibility of passing it on to their children. Sample letters and talking points can be helpful. Your HCM center, specialist or genetic counselor can offer more tips.

For more information visit *CardioSmart.org/HCM* or use the *HCM Family Screening and Testing Worksheet* to see how strongly HCM runs in your family and which relatives need to be screened or tested.

This information should not take the place of clinical advice about HCM screening or genetic testing given to you by your health care team.