Should My Child(ren) Have Predictive Genetic Testing For Hypertrophic Cardiomyopathy?

This is an information package to help guide parents in a discussion with their genetic counsellor or healthcare provider about predictive genetic testing for their children.

What is predictive genetic testing?

Predictive genetic testing is a test done before a child has any symptoms of hypertrophic cardiomyopathy (HCM). The test looks to see if your child has inherited the genetic variant (difference) causing HCM in your family. Not all children who carry the genetic variant will end up having symptoms of HCM.

It's your decision

Hypertrophic cardiomyopathy (HCM) is a heart condition that usually runs in families. The children of a person with HCM have a higher chance of developing this heart condition.

If a genetic cause (genetic variant/difference) has been found in your family, there are 2 choices for your child or children:

1 **Children can be followed by a heart doctor and have heart screening tests (e.g. echocardiogram, ECG/EKG).**

OR

2 **Children can have predictive genetic testing. If a child is found to carry the genetic variant, they will be followed by a heart doctor. If they do not carry the genetic variant, heart screening is not necessary for most children.**

Some parents wonder about not doing genetic testing or heart screening for their child(ren) until their child(ren) have symptoms of HCM. This choice may mean missing the chance to avoid serious heart problems, or even sudden death (rare). Early diagnosis and education improve the long-term health of children with HCM. Because of this, it is very important that all children who could have HCM are seen by a heart doctor.

Having heart screening only or having predictive genetic testing are both good choices. Making the decision might be easier if you:

1. Think about what your child's medical care will look like with each choice.
2. Think about how each choice fits with your beliefs and values.
3. Talk with your child's other parent, your child (if they are old enough) and your genetic counsellor. You might also want to talk to your family, family doctor, or other people that you trust.

Information to help you make the decision

What is Hypertrophic Cardiomyopathy (HCM)?
HCM is a medical condition that affects the thickness of the heart muscle and the rhythm of the heart. People with HCM may have shortness of breath, chest pain, heart palpitations (funny beats), fainting, cardiac arrest (heart stopping beating), or heart failure. They may also have no symptoms. HCM can present at any age but usually presents before 35 years of age.[1]

People with HCM have a higher chance of cardiac arrest compared to the general population. The risk in the general population is 0.4% (4 in 1000 people).[2]

The chance of cardiac arrest for a person with HCM is approximately 6%.[3]

6 in 100 people.

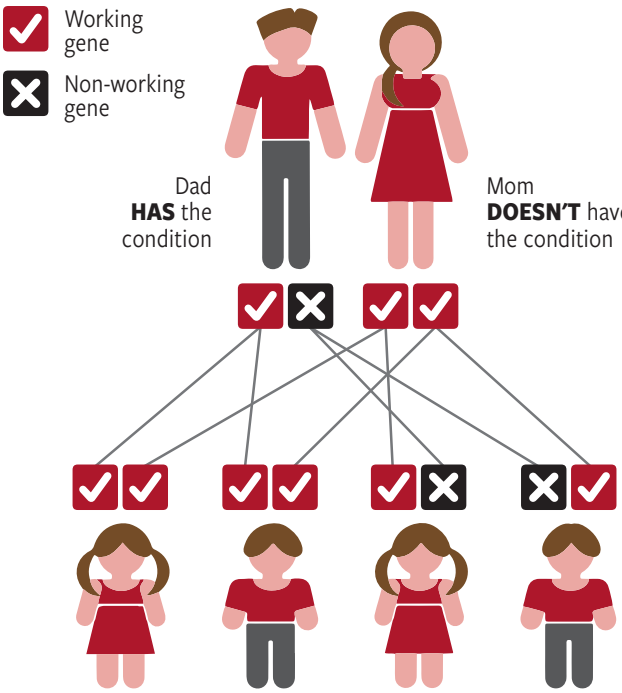
People with HCM have a higher chance of heart failure compared to the general population. The lifetime risk of heart failure in the general population is ~2-3% (2-3 in 100 people).[4]

The lifetime risk of heart failure for people with HCM is approximately 5-10%.[5]

5-10 in 100 people.

What causes HCM?
HCM is a genetic condition. It is caused by genetic variants (differences) in genes that are important for the heart muscle. A genetic variant that makes a gene function abnormally is called pathogenic or likely pathogenic.

Inheritance of HCM



What is the chance that my child inherited the genetic variant found in our family for HCM?
Both males and females can have HCM. If one parent has the genetic variant for HCM, each of their children have a 50% chance of inheriting the genetic variant and a 50% chance of not inheriting the genetic variant. Children who inherit the genetic variant have a higher chance of developing HCM.

If my child inherited the genetic variant found in our family, what is the chance that they will develop HCM?
The chance of developing HCM depends on the genetic variant in the family, the results of their heart tests, and their age, sex, and health history. Your child's heart doctor will talk to you and your child about their specific risk.

What would happen next if my child inherited the genetic variant found in my family for HCM?
If you choose predictive genetic testing for your child and they are found to carry the genetic variant in your family, they would be followed closely by a heart doctor. The heart doctor would talk to you about different management strategies that can reduce the health risks associated with HCM. Not all people who carry a genetic variant will show signs or symptoms of HCM.

What would happen next if my child did NOT inherit the genetic variant for HCM?
If you choose predictive genetic testing for your child and they do not carry the genetic variant found in your family, they would no longer need ongoing heart screening for HCM. Although they would no longer need to be followed by a heart doctor, heart assessments may be recommended in some situations.

What would happen next if my child does not have predictive genetic testing?
If you choose heart screening only for your child, they would be seen by a heart doctor. The heart doctor would talk with you about how often your child needs heart screening tests and how it helps to protect your child's health. It is important to know that predictive genetic testing remains an option for your child at any point in the future.

What can we do to reduce the risks associated with HCM for our child(ren)?

Management
- Watch for symptoms such as shortness of breath, chest pain, heart palpitations (funny beats), or fainting. If your child has any of these symptoms take them to see a doctor immediately.
- Take your child for regular heart screening.

Education
- Encourage family and caregivers to take CPR training.
- Be aware of the location of automated external defibrillators (AEDs).
- Educate family and friends about the condition.

Advantages and disadvantages of doing heart screening only VS predictive genetic testing

Heart Screening Only (without predictive genetic testing)

Advantages
- Allows you to know if your child has signs of HCM.
  "Having children followed by a heart doctor is important to get a good baseline and then check for changes."
- Possibly avoids additional worry for you and your child unless your child has signs of HCM.
  "We didn't want to live with the burden of having a positive genetic test if our child didn't show signs of heart problems."
- Allows your child to take part in the decision about genetic testing when they are older.

Disadvantages
- Requires time and costs for ongoing medical appointments that may not be needed.
  "The cost and time of having each of our children screened was greater than that of having them gene tested, and following those who tested positive."
- May create unnecessary worry if your child is not at risk of HCM.
- Does not detect all individuals affected by HCM. As a result, an opportunity may be missed to make lifestyle changes to decrease the risk of heart symptoms.

Predictive Genetic Testing

Advantages
- Avoids unnecessary heart screening for your child if genetic testing is negative.
- Reduces worry for you and your child if genetic testing is negative.
  "We wanted to reduce worry for those in our family who were not gene positive."
- Allows your child to make lifestyle changes to decrease the risk of heart symptoms if genetic testing is positive.

Disadvantages
- May lead to your child being treated differently (e.g. overprotected).
- Doesn't allow your child to take part in the decision, if your child is young.
  "I was 5 when I had genetic testing and unfortunately wasn't part of the decision making process."
- Potential risk of genetic and employment discrimination.
  "Genetic discrimination was a big worry for us."
- Rarely, overtime, our understanding of a genetic test result can change.

Genetic Discrimination
Genetic discrimination is when a genetic test result is used in a negative way against a person. Canada and the United States both have laws to stop insurance companies and employers from using genetic test results in certain situations. These laws do not prevent the use of family history and medical information by these groups.

Other things to consider:
- How did the condition present in your family?
- Is the timing right?
- How does your child feel about genetic testing?
- Has your child had any heart screening done?

Making a decision

What are the advantages and disadvantages that matter most to you?

DOING Heart Screening Only

Advantages:

Disadvantages:

DOING Predictive Genetic Testing

Advantages:

Disadvantages:

What are the advantages and disadvantages that matter most to your child?

DOING Heart Screening Only

Advantages:

Disadvantages:

DOING Predictive Genetic Testing

Advantages:

Disadvantages:

Do you have all the information you need?

Do You:

- Know the chance that your child has inherited the genetic variant (difference) for HCM?
- Know what happens next if your child has inherited the genetic variant for HCM?
- Understand what could happen if your child doesn't have heart screening or predictive genetic testing?
- Know the advantages and disadvantages that are most important to you and your child?

What is your decision?

- ○ Heart screening only
- ○ Predictive genetic testing
- ○ I don't know

Are you comfortable with this decision?[5]

Sure of yourself
Do you feel sure about the best choice for you?
○ Yes ○ No

Understand the information
Do you know the advantages and disadvantages of doing or not doing heart screening and genetic testing?
○ Yes ○ No

Advantages and Disadvantages
Are you clear about which advantages and disadvantages matter most to you?
○ Yes ○ No

Encouragement
Do you have enough support and advice to make a choice?
○ Yes ○ No

Agreement
Are you and your child(ren)'s other parent in agreement with your decision?
○ Yes ○ No

References

[1] Lopes LR, Rahman MS, Elliott PM. A systematic review and meta-analysis of genotype-phenotype associations in patients with hypertrophic cardiomyopathy caused by sarcomeric protein mutations. Heart. 2013 Dec; 99(24):1800-11.

[2] Goldenberg I, Horr S, Moss A J, Lopes C M, Barsheshet A, McNitt S, Zareba W, et al, 2011. "Risk for Life-Threatening Cardiac Events in Patients with Genotype-Confirmed Long-QT Syndrome and Normal-Range Corrected QT Intervals." Journal of the American College of Cardiology 57 (1): 51-59.

[3] Ho CY, Day SM, Ashley EA, Michels M, Pereira AC, Jacoby D, Cirino AL, Fox JC, Lakdawala NK, Ware JS, Caleshu CA, Helms AS, Colan SD, Girolami F, Cecchi F, Seidman CE, Sajeev G, Signorovitch J, Green EM, Olivotto I, et al. "Genotype and lifetime burden of disease in hypertrophic cardiomyopathy. Insights from the Sarcomeric Human Cardiomyopathy Registry (SHaRe)." Circulation. 2018; 138:1387–98.

[4] Lloyd-Jones D, et al. "Heart disease and stroke statistics—2010 update: a report from the American Heart Association." Circulation. 2010; 121:e46–e215.

[5] Harris KM, Spirito P, Maron MS, Zenovich AG, Formisano F, Lesser JR, Mackey-Bojack S, Manning WJ, Udelson JE, Maron BJ. "Prevalence, clinical profile, and significance of left ventricular remodeling in the end-stage phase of hypertrophic cardiomyopathy." Circulation. 2006; 114:216–25.

[6] The SURE Test ©O'Connor and Légaré, 2008.

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Hypertrophic Cardiomyopathy Association – *www.4hcm.org*

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