Hereditary Cardiovascular Conditions

GENETIC TESTING FOR UNDIAGNOSED DISEASES
What is Hypertrophic Cardiomyopathy (HCM)?

NORMAL HEART

HEART WITH HCM

Extra or thick heart muscle
Typically in the left ventricle
Symptoms of HCM

- Shortness of breath
- Palpitations (feelings of a rapid, fluttering or pounding heart)
- Risk for sudden cardiac arrest
- Syncope (fainting)
- Arrhythmia (abnormal heart rhythm)
Mutations in many genes can cause HCM. These gene mutations cause the heart muscle to become too thick.

There are some genetic causes of HCM that are still not known at this time.

This chart shows the genetic causes of HCM. Most known mutations are in 2 genes (MYH7 and MYBPC3). There are other genes known to cause HCM (‘Other’) and likely other genetic causes that are not known (‘Unknown’).
What is Dilated Cardiomyopathy (DCM)?

NORMAL HEART

HEART WITH DCM

Larger ventricle with thinner heart muscle (mostly in left ventricle)
Symptoms of DCM

- Shortness of breath
- Palpitations (feelings of a rapid, fluttering or pounding heart)
- Risk for stroke
- Risk for sudden cardiac arrest
- Syncope (fainting)
- Arrhythmia (abnormal heart rhythm)
Genetic Causes of DCM

Mutations in many different genes can cause DCM. These gene mutations cause the heart tissue to become weaker.

There are some genetic factors that cause DCM that are still not known at this time, and other cases of DCM that are caused by decreased oxygen to the heart (like a heart attack). These “ischemic” cases are not usually genetic.

This chart shows the genetic causes of DCM. There are other genes known to cause DCM (‘Other’) and likely other genetic causes that are not known (‘Unknown’).
What is Long QT Syndrome (LQTS)?

NORMAL EKG

EKG SHOWING LQTS

Long QT interval seen on electrocardiogram (EKG)
Mutations in many different genes can cause LQTS. These gene mutations cause the heartbeat to become abnormal.

There are some genetic causes of LQTS that are still not known at this time.

This chart shows the genetic causes of LQTS. Most known mutations are in 3 genes (KCNH2, KCNQ1 and SCN5A). There are other genes known to cause LQTS ('Other') and likely other genetic causes that are not known ('Unknown').
What is Brugada Syndrome (BrS)?

NORMAL EKG

EKG SHOWING BrS, TYPE 1

Specific pattern seen on electrocardiogram (EKG)
Genetic Causes of Brugada Syndrome (BrS)

Mutations in many different genes can cause BrS. These gene mutations cause the heartbeat to become abnormal.

There are some genetic causes of BrS that are still not known at this time.

This chart shows the genetic causes of BrS. Most known mutations are in the SCN5A gene. There are other genes known to cause BrS ('Other') and likely other genetic causes that are not known ('Unknown').
Symptoms of Arrhythmias

- **Palpitations**
  - (feelings of a rapid, fluttering or pounding heart)

- **Syncope**
  - (fainting)

- **Risk for sudden cardiac arrest**

- **Brugada syndrome, type 1**

- **Long QT Interval**
Management for Inherited Cardiovascular Conditions

SCREENING OPTIONS
- Echocardiogram (echo)
- Electrocardiogram (EKG)
- Cardiovascular evaluation (physical exam)

TREATMENT OPTIONS
- Medications for treatment and/or avoiding certain medications
- Surgery
- Avoiding certain competitive sports
- Pacemaker
- Implantable cardioverter defibrillator (ICD)

Your doctor or other healthcare provider can help create a plan specific to you and your family.
Most inherited cardiovascular conditions are inherited in an autosomal dominant pattern. This means that people with the cardiovascular condition have a 50% risk of passing the condition down to each child. Not everyone that inherits a mutation will develop the disease at the same age or to the same degree of severity.
Very few inherited cardiovascular conditions are inherited in an autosomal recessive pattern. In this case, each parent has no symptoms, but carries a genetic mutation and has a 25% risk of having a child with the cardiovascular condition.
No Known Genetic Mutation in the Family

If genetic testing does not find a mutation that causes cardiomyopathy or arrhythmia, or if you opt not to have genetic testing, your family members may still benefit from increased screening for signs or symptoms.

SCREENING RECOMMENDATIONS:
Echo, EKG and physical exam every 1-2 years
What do my Test Results Mean?

<table>
<thead>
<tr>
<th>Genetic Test Results</th>
<th>Meaning</th>
<th>Implications</th>
</tr>
</thead>
<tbody>
<tr>
<td>Pathogenic Mutation</td>
<td>Positive</td>
<td>Confirms diagnosis</td>
</tr>
<tr>
<td>Variant, Likely Pathogenic</td>
<td>VUS</td>
<td>Testing possibly available for family members to help learn more</td>
</tr>
<tr>
<td>Variant, Unknown Significance (VUS)</td>
<td>Negative</td>
<td>Does not change diagnosis</td>
</tr>
<tr>
<td>Variant, Likely Benign</td>
<td></td>
<td>Does not change diagnosis</td>
</tr>
<tr>
<td>No Variants Detected</td>
<td></td>
<td>Likely, no testing available for family members</td>
</tr>
</tbody>
</table>

Confirms diagnosis

Testing available for family members

Testing possibly available for family members to help learn more

Does not change diagnosis

Does not change diagnosis

Likely, no testing available for family members
Positive Test Results

- Mutation found
- Confirms diagnosis
- Testing available for family members
Negative Test Results

No mutation found

- Does not change diagnosis
- Likely, no testing available for family members
- Screening (echo, EKG) recommended for all close relatives

Further genetic testing?
Variant of Unknown Significance Test Results

Variant of unknown significance (VUS)

- Does not change diagnosis
- Screening (echo, EKG) recommended for all close relatives
- Further testing of affected family members?