Hereditary Cardiovascular Conditions

GENETIC TESTING FOR UNDIAGNOSED DISEASES

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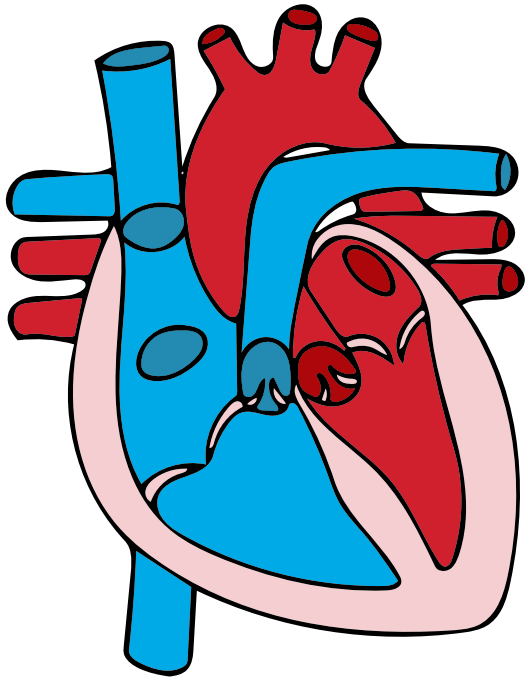
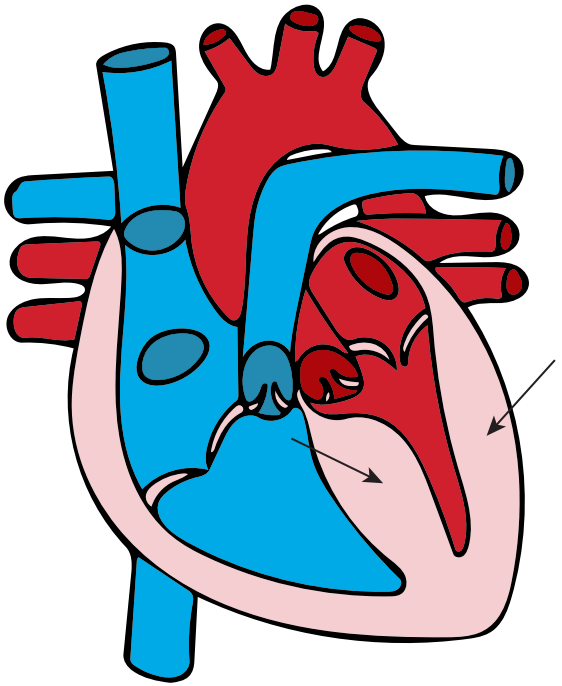
What is Hypertrophic Cardiomyopathy (HCM)?

NORMAL HEART



HEART WITH HCM



Extra or thick heart muscle
Typically in the left ventricle

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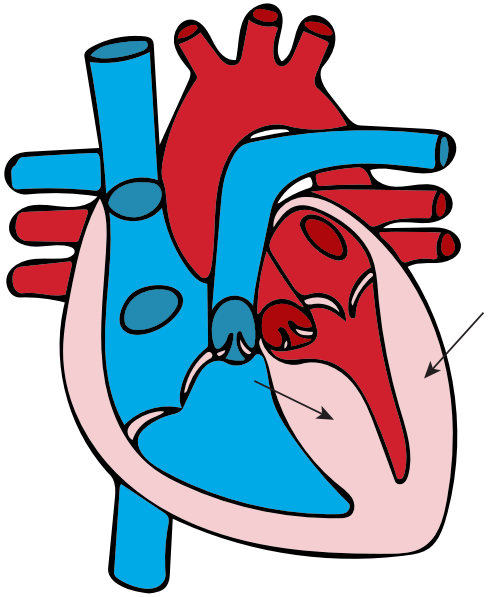
Symptoms of HCM

Shortness of breath

Risk for sudden cardiac arrest



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

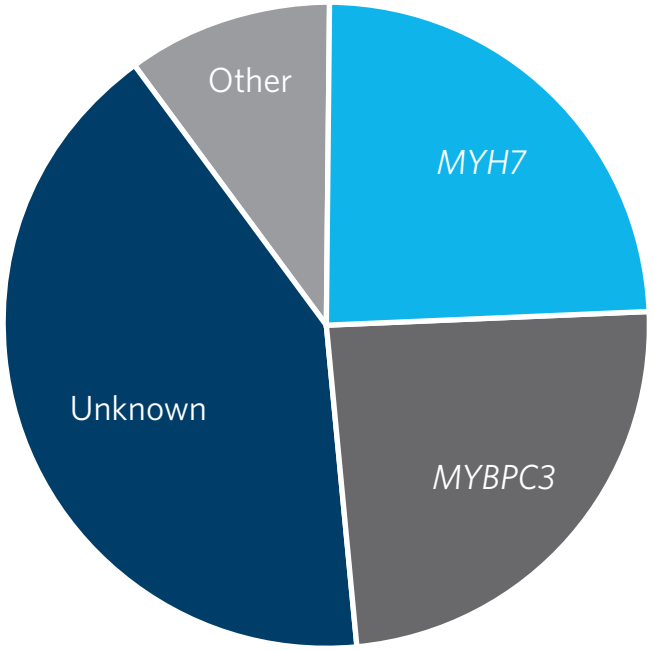
Arrhythmia
(abnormal heart rhythm)

Syncope
(fainting)

Genetic Causes of HCM

Mutations in many genes can cause HCM. These gene mutations cause the heart muscle to become too thick.

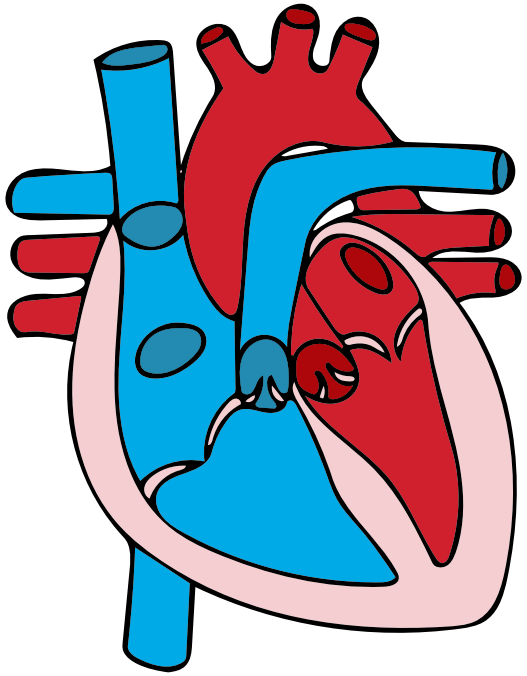
There are some genetic causes of HCM that are still not known at this time.



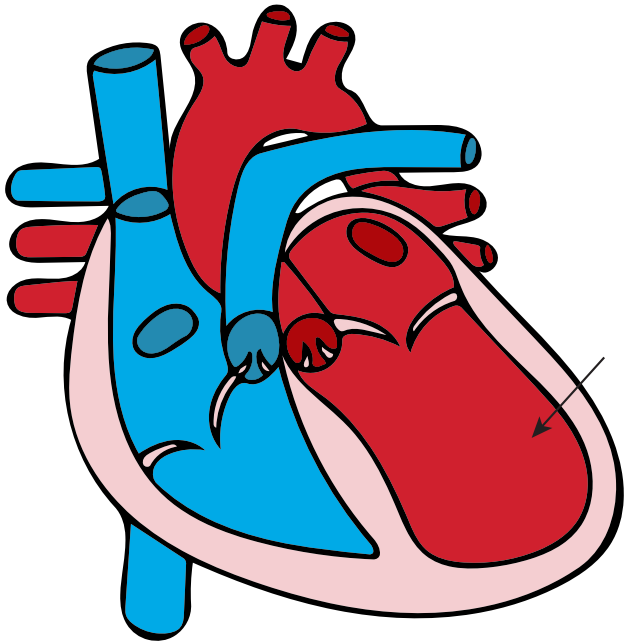
This chart shows the genetic causes of HCM. Most known mutations are in 2 genes (*MYH7* and *MYBPC3*). There are other genes known to cause HCM ('Other') and likely other genetic causes that are not known ('Unknown').

What is Dilated Cardiomyopathy (DCM)?

NORMAL HEART



HEART WITH DCM



Larger ventricle with thinner heart muscle
(mostly in left ventricle)

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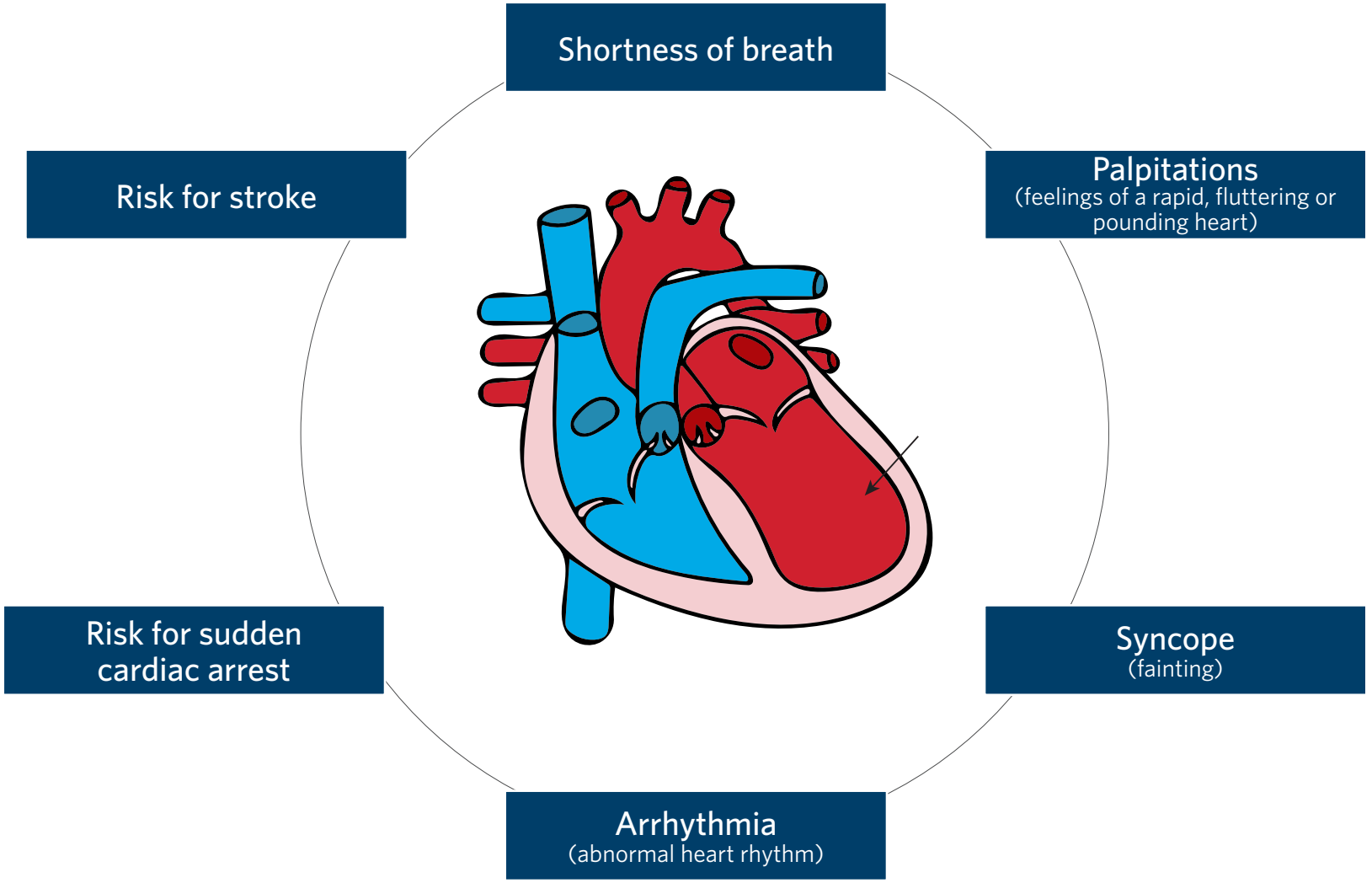
Symptoms of DCM



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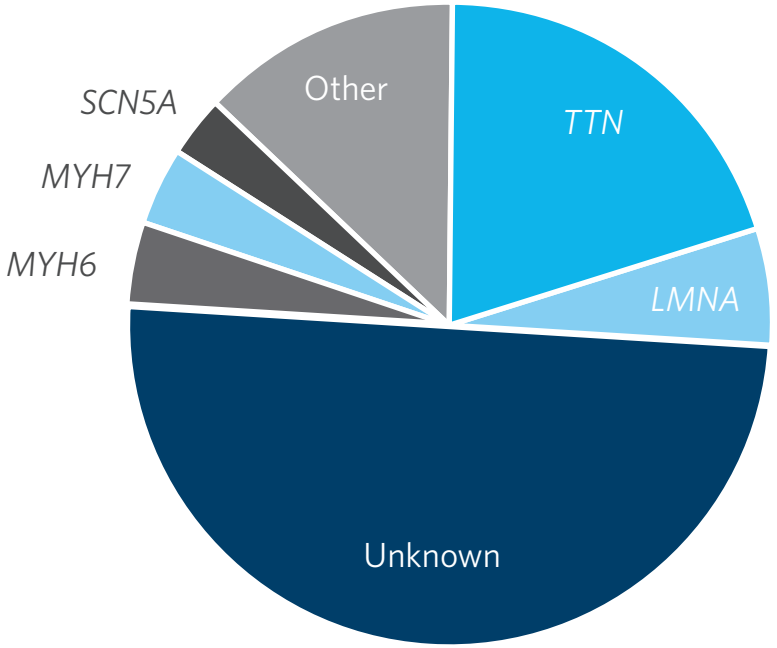
Genetic Causes of DCM

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').

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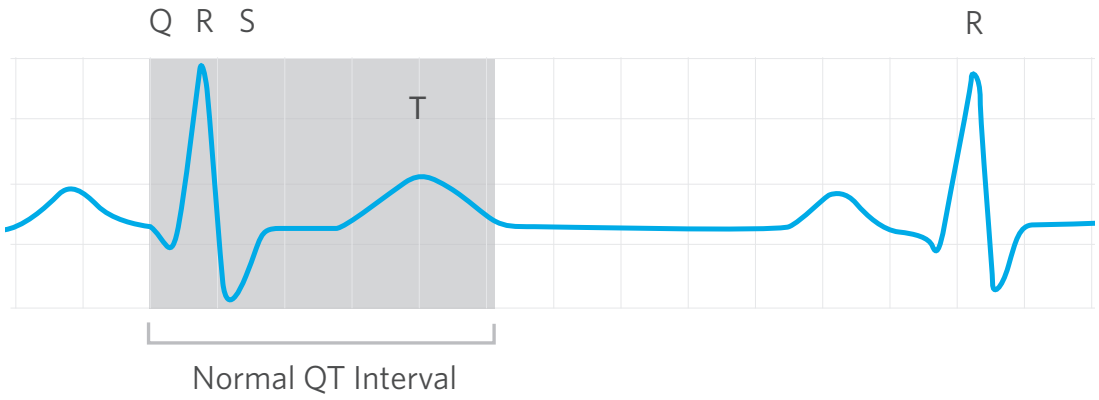
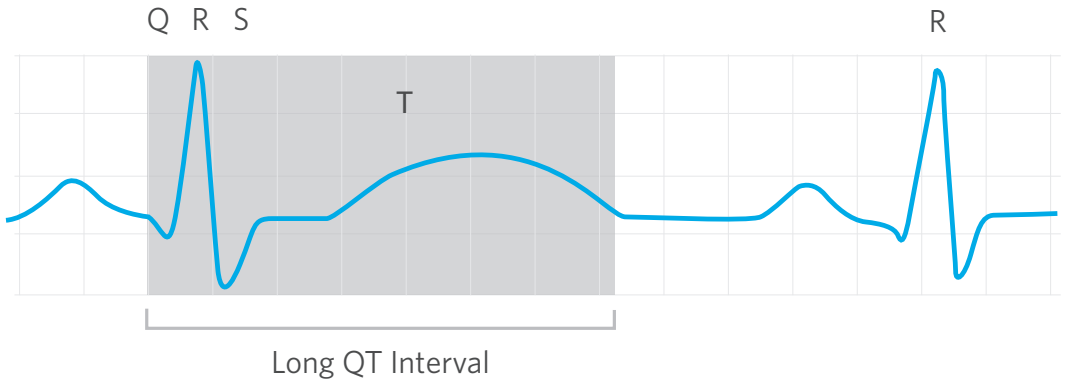
What is Long QT Syndrome (LQTS)?

NORMAL EKG



EKG SHOWING LQTS



Long QT interval seen on electrocardiogram (EKG)

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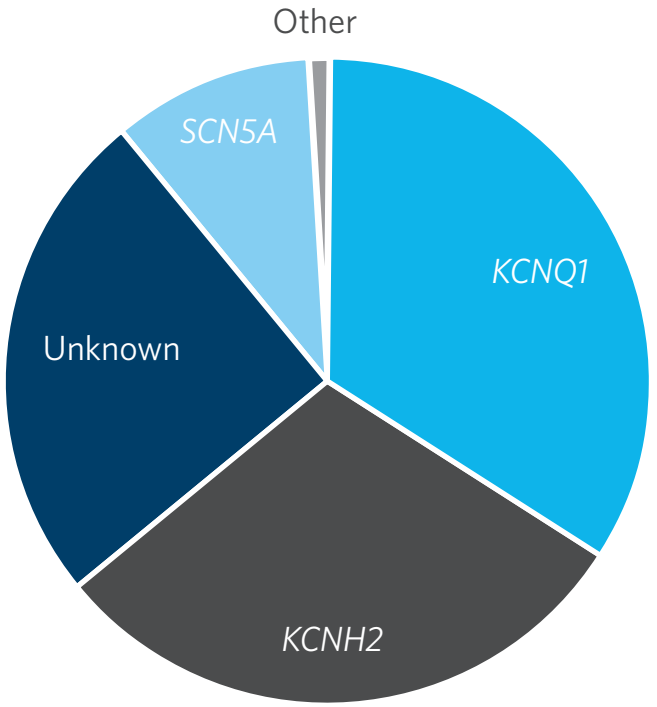
Genetic Causes of Long QT Syndrome (LQTS)

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

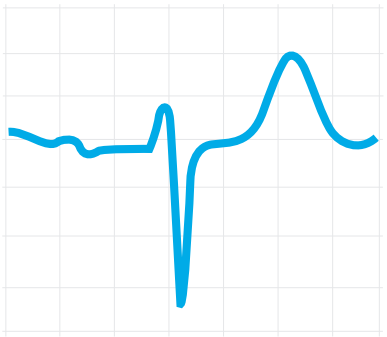
There are some genetic causes of LQTS that are still not known at this time.



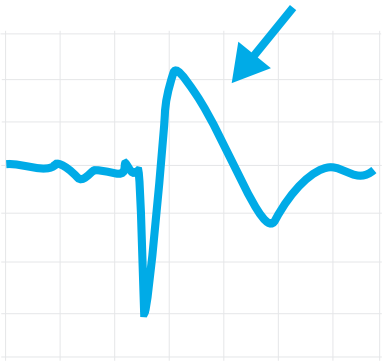
This chart shows the genetic causes of LQTS. Most known mutations are in 3 genes (*KCNH2, KCNQ1* and *SCN5A*). There are other genes known to cause LQTS ('Other') and likely other genetic causes that are not known ('Unknown').

What is Brugada Syndrome (BrS)?

NORMAL EKG



EKG SHOWING BrS, TYPE 1

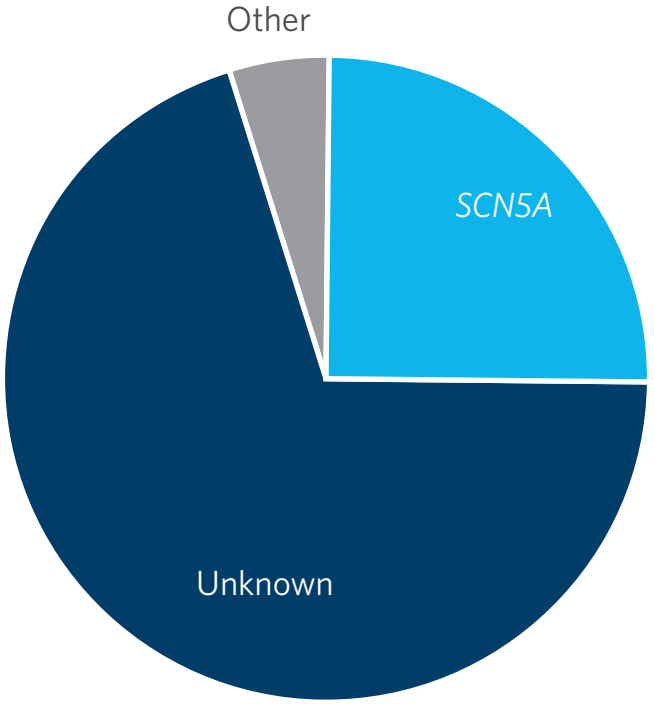


Specific pattern seen on electrocardiogram (EKG)

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').

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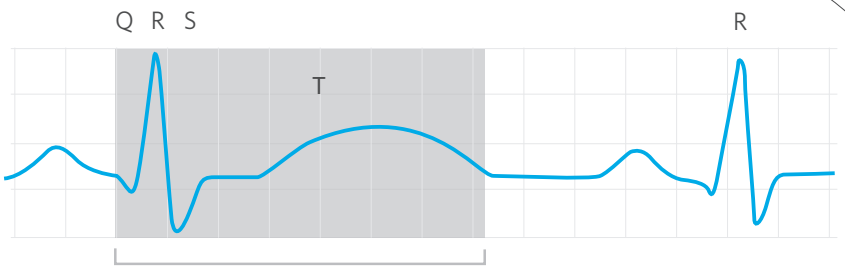
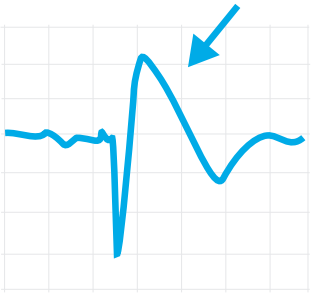
Symptoms of Arrhythmias

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



Risk for sudden cardiac arrest



Syncope
(fainting)

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Management for Inherited Cardiovascular Conditions

SCREENING OPTIONS

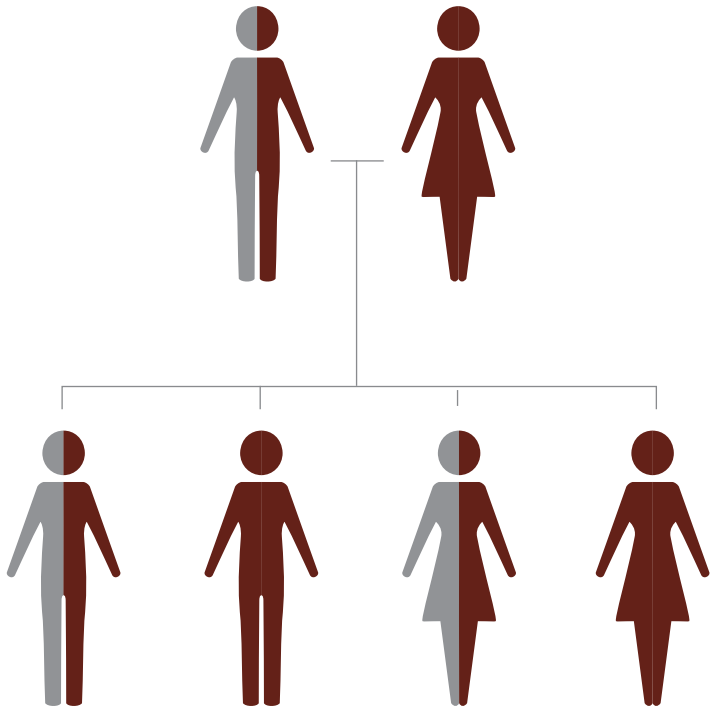
- ☐ Echocardiogram (echo)
- ☐ Electrocardiogram (EKG)
- ☐ Cardiovascular evaluation (physical exam)

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.

Autosomal Dominant Inheritance

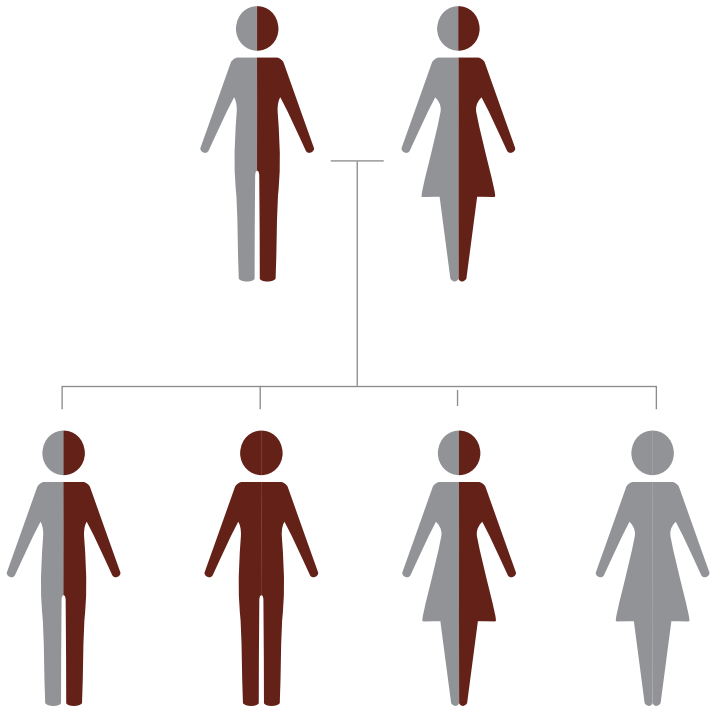


Has genetic mutation

No mutation

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.

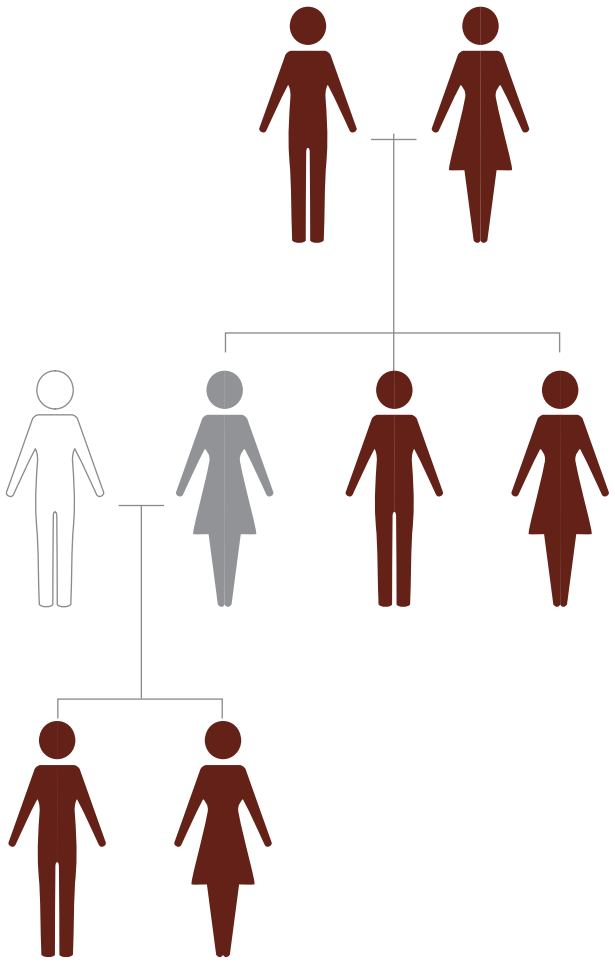
Autosomal Recessive Inheritance



- Carries genetic mutation, but has no symptoms
- No mutation
- Has symptoms of cardiovascular condition

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



- Has inherited cardiomyopathy or arrhythmia
- Should talk with his/her doctor about screening

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

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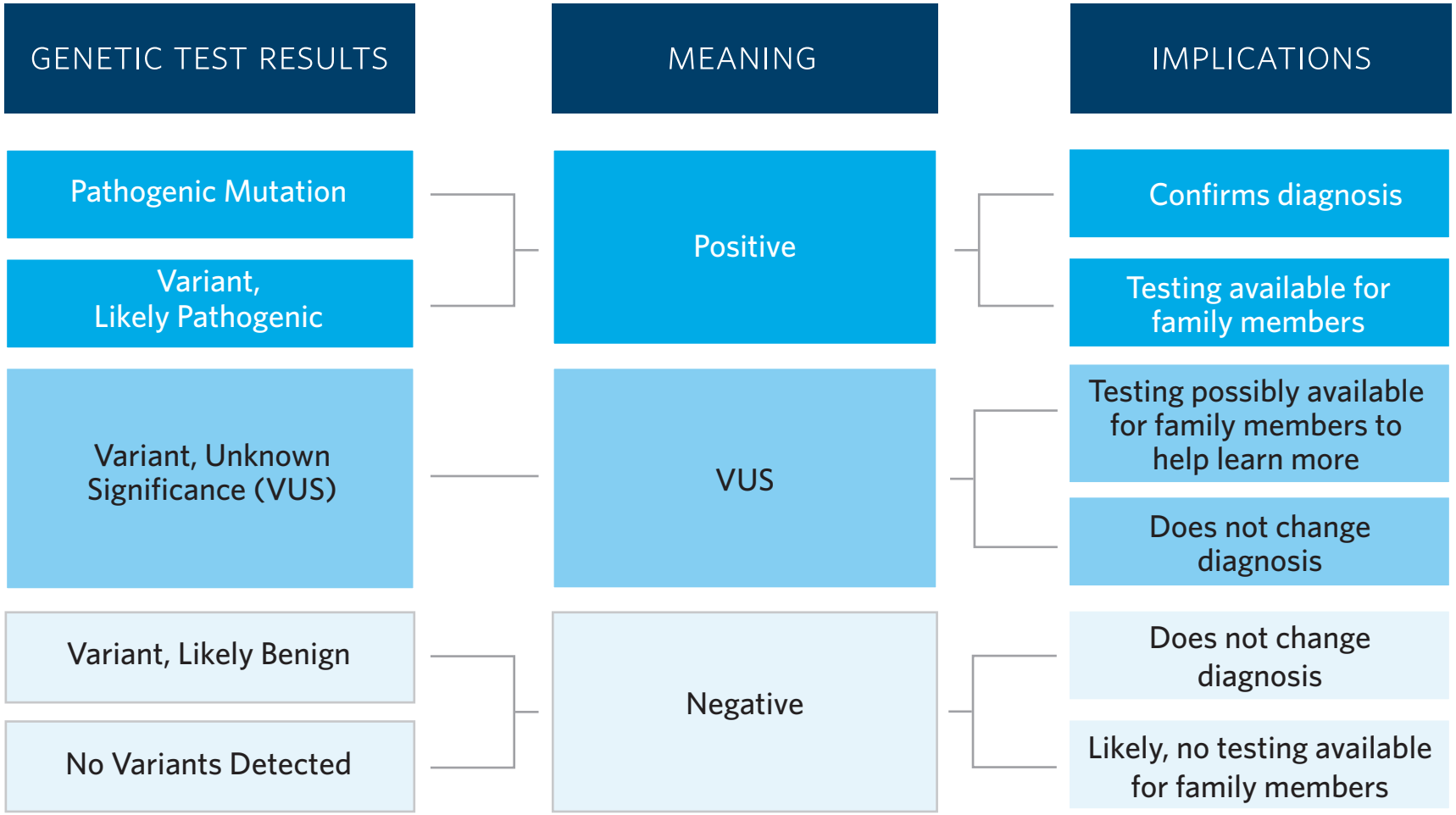
What do my Test Results Mean?



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Positive Test Results

Mutation found	Confirms diagnosis	Testing available for family members

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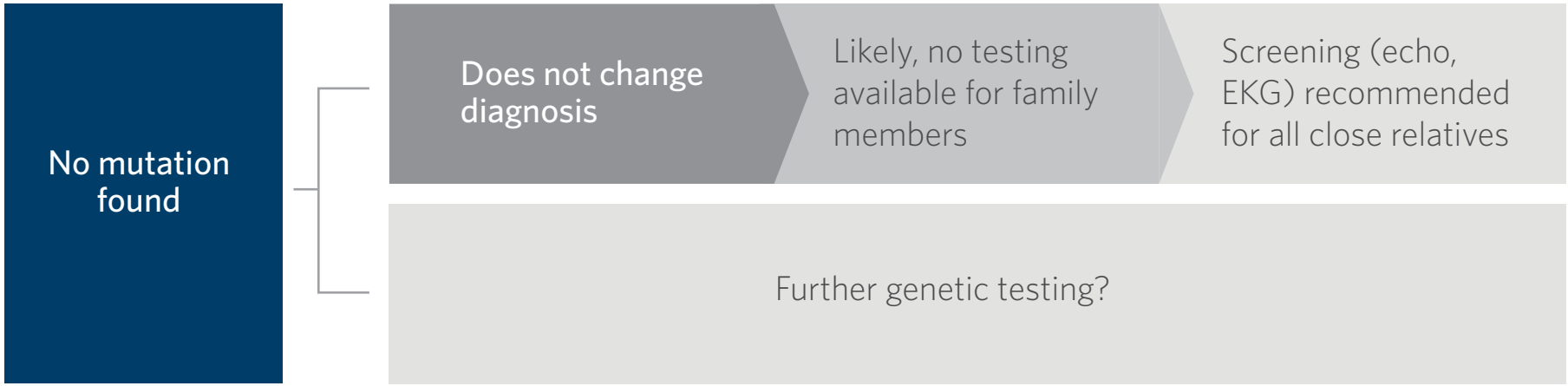
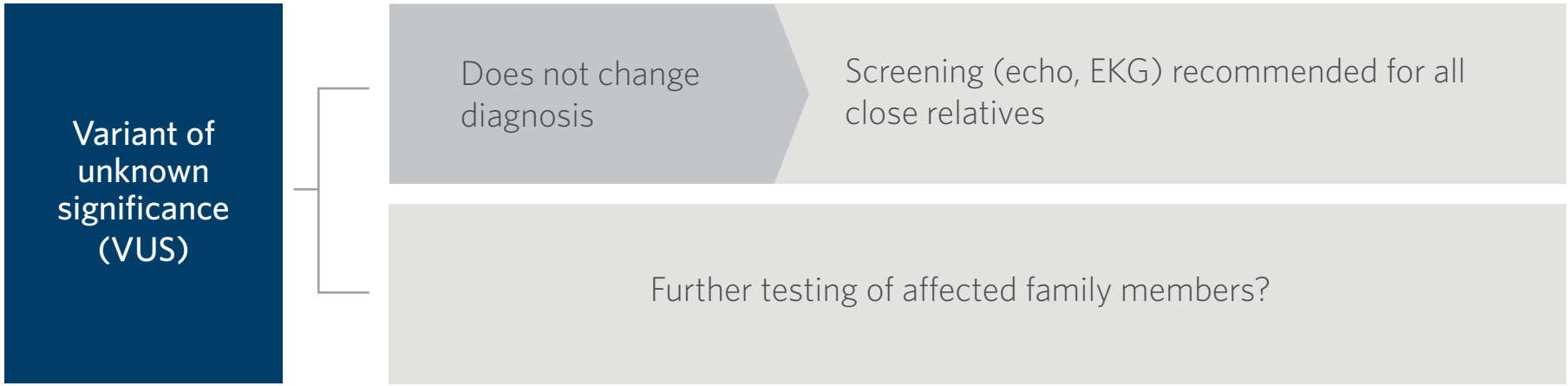
Negative Test Results



Variant of Unknown Significance Test Results



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