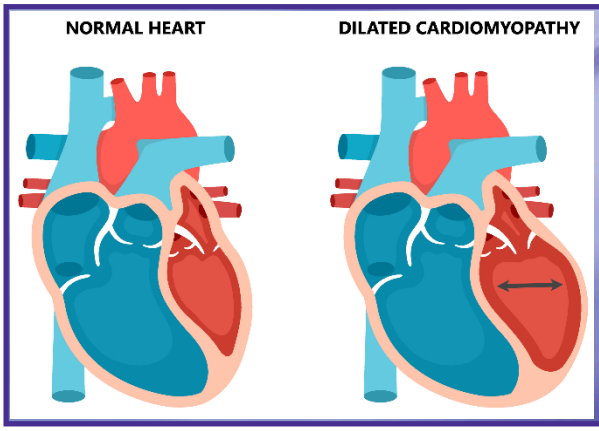

Genetic testing for Dilated Cardiomyopathy

Dilated cardiomyopathy (DCM) is a complex type of heart disease. It can include:

- Thin, weak heart muscles
- Very large heart chambers (especially the bottom part of the heart called the ventricles)

These changes make it hard for the heart to pump blood and can sometimes lead to heart failure. DCM can also lead to heart valve problems, abnormal heart rhythms, and blood clots in the heart.

Common symptoms of heart failure include shortness of breath, fatigue, and swelling of the ankles, feet, legs, abdomen, and veins in the neck.

How is DCM diagnosed?

DCM is diagnosed by a doctor based on your medical history, your family history, a physical exam, an ultrasound of your heart (echocardiogram), and heart rhythm testing (electrocardiogram or ECG). Large heart chambers can be seen by ultrasound and an irregular heartbeat may be found by ECG.

What causes DCM?

There are both non-genetic and genetic causes of DCM.

Non-genetic causes:

- Infection (myocarditis)
- High blood pressure
- Thyroid disease or diabetes
- Heart valve disease
- Alcohol or drug abuse
- Heart attack or coronary artery disease
- Certain toxins, including some chemotherapy drugs and radiation

Genetic cause:

- **Inherited (genetic) conditions**

What is inherited DCM?

Inherited DCM is due to a mutation in the genetic code (DNA) that causes the heart chambers to slowly get larger (dilate). The laboratory calls these mutations “pathogenic variants.” There are many genes that can cause DCM. When one of the DCM genes is not working right, the heart muscles get thinner and weaker over time. This makes it harder for the heart to pump blood.

How is DCM inherited?

DCM can run in the family. If a person has a mutation in one of the DCM genes, others in the family may have the same mutation. When a parent has an inherited form of DCM there is usually a 50% (1 in 2) chance to pass the mutation to any child. A person’s sex does not affect who can pass on or inherit a mutation. This is called autosomal dominant inheritance. Rarely, DCM is due to a gene with a different type of inheritance.

GATHERING YOUR FAMILY HISTORY

It is not always easy to recognize DCM. Many people with DCM are not aware of the condition and may not notice any symptoms. It is helpful to ask family members about medical problems that might be related to DCM.

Ask about:

- Unusual shortness of breath
- Fainting or dizziness
- Palpitations (fast, fluttering or pounding heartbeat)
- Chest pain or pressure during or after exercise
- Heart enlargement or dilation
- Swelling (edema) in the legs, ankles, feet, or belly (abdomen)
- Sudden cardiac death or unexplained death
- Early heart failure

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How is genetic testing done?

A blood or saliva test can look for mutations (pathogenic variants) in genes that cause DCM. Genetic testing works best when it is done on a person who has DCM with no known cause. A genetic cause is even more likely if there is familial DCM. This is when two or more people in a family have unexplained DCM. Current testing finds a mutation in about 30% to 35% of people with DCM who have this type of family history. You can learn more about genetic testing by talking with a genetic counselor.

What are the benefits of genetic testing?

When mutation is found in a person with DCM, other people in the family can be offered genetic testing. Testing can then identify which family members are at-risk for DCM, even if they have no heart symptoms. Cardiac screening lowers the risk for heart problems in healthy people with a gene mutation. Family members who did not inherit the mutation can be reassured.

What if testing does not find a genetic change that causes DCM?

Many people with familial DCM get a negative (normal) result, where no mutation is found in any of the tested genes. However, current testing does not find all genetic causes of DCM. There could still be a genetic cause for DCM in some families. For this reason, cardiac screening is still recommended for close relatives.

What happens if I inherit a pathogenic variant in a DCM gene?

A person who inherits a mutation is more likely to develop DCM during their lifetime. However, some people with a mutation never develop DCM or any related heart problems. Regular cardiac screening is recommended for anyone with a mutation that can cause DCM.

SCREENING FOR DILATED CARDIOMYOPATHY

Screening recommendations for those at risk for DCM include echocardiogram (heart ultrasound) and ECG (electrocardiogram). The frequency of screening depends on your age and medical history.

Echocardiogram is a heart ultrasound. It is the most common test used to diagnose DCM. This test uses sound waves to produce images of the heart. Your doctor can see the size of the heart muscle, how the blood flows through your heart, and how the heart valves move.

ECG (Electrocardiogram) is a test that checks the electrical activity (rhythm) of your heart as it beats. An ECG can detect abnormal electrical signals that may happen with a dilated heart. A Holter monitor or ZIO patch is a portable form of ECG that records your heartbeat for a much longer period of time. These monitors may be on the body for 24 to 48 hours or as long as 14-30 days.

What happens if I develop DCM?

Many people with DCM are able to live a normal life without severe heart problems. However, some people with DCM will have symptoms that can include breathing trouble, chest pain, fatigue, palpitations, swelling in the legs, ankles, feet, or belly. DCM can also lead to heart valve problems, abnormal heart rhythms, and blood clots in the heart. Treatment can help reduce or prevent symptoms and lower the risk of complications. Treatment includes regular follow-up, lifestyle changes, medications, and procedures, as needed. An automatic implantable cardiac defibrillator (AICD) may be placed when there are concerns about dangerous heart rhythms.

The information is not intended to diagnose health problems or to take the place of professional medical care. If you have persistent health problems or if you have further questions, please consult your health care provider.

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