Familial dilated cardiomyopathy (DCM) is a health condition that can run in families that impacts the heart's ability to function how it should due to the heart muscle in the left ventricle becoming thin and weak. Because the muscle becomes more thin, the space inside the left ventricle, called the chamber, gets bigger (dilated).



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When the space inside the heart is bigger and the heart muscle is weak, the heart can't pump blood as efficiently as it should. This means that the heart has to work harder to try to pump a normal amount of blood, which further causes the heart muscle to thin and grow weaker. Eventually, DCM can lead to heart failure.

<u>Cardiomyopathies</u> (diseases of the heart muscle) can come in several forms, but DCM is the most common. It typically affects people who are between the ages of 20-60 years old, and can also lead to <u>arrhythmias</u> (irregular heartbeat), blood clots, or <u>sudden death</u>. Some studies have estimated that approximately 1 in 250 people will have DCM, and males are three times more likely to have DCM than women.

## Causes

About half of all cases of DCM are inherited, or genetic, and can be passed down in families. The other half are either acquired through environmental causes (such as exposures or medications), or the cause is unknown. Approximately 30 genes have thus far been discovered that can cause DCM in families, including:

- *TTN* (10-20% of all DCM)
- <u>LMNA</u> (6% of all DCM)
- <u>MYH7</u> (4.2% of all DCM)
- *MYH6* (3-4% of all DCM)
- <u>MYBPC3</u> (2-4% of all DCM)
- <u>SCN5A</u> (2-4% of all DCM)
- <u>TNNT2</u> (2.9% of all DCM)
- *BAG3* (2.5% of all DCM)
- ANKRD1 (2.2% of all DCM)
- <u>TPM1</u> (unknown percentage of all DCM)
- <u>TNNI3</u> (unknown percentage of all DCM)
- <u>ACTC1</u> (unknown percentage of all DCM)
- <u>DSG2</u> (unknown percentage of all DCM)

## **Diagnosing Dilated Cardiomyopathy**

While genetic testing for DCM can be helpful to establish a diagnosis or to determine what form of DCM someone has, a medical provider who has experience with DCM can usually diagnose it by using other tests. An <u>echocardiogram</u> or <u>cardiac MRI</u> can look to see if the left ventricle is measuring larger than expected, as well as how much blood the left ventricle is pumping (called systolic function). If the left ventricle is enlarged AND someone's systolic

function is less than 50% of what it should be, that would lead to a clinical diagnosis of DCM.

People who have DCM usually start to show symptoms in their 40s, 50s, or 60s, although symptoms can begin at any age.

## **Medical Management for Dilated Cardiomyopathy**

Treatment for DCM can sometimes vary depending on the individual person and their specific health concerns, and should be discussed with a medical provider who is familiar with DCM. Some options for medical management can include medication, regular cardiac monitoring, a surgically-implantable <u>defibrillator</u>, and a heart transplant. People who have DCM should try to avoid things like strenuous physical activity and excessive alcohol consumption.

Click <u>here</u> to learn more about scheduling a genetic counseling appointment for questions about pediatric or adult genetic conditions.

## **Additional Resources**

American Heart Association

Children's Cardiomyopathy Foundation

Heart Failure Society of America

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