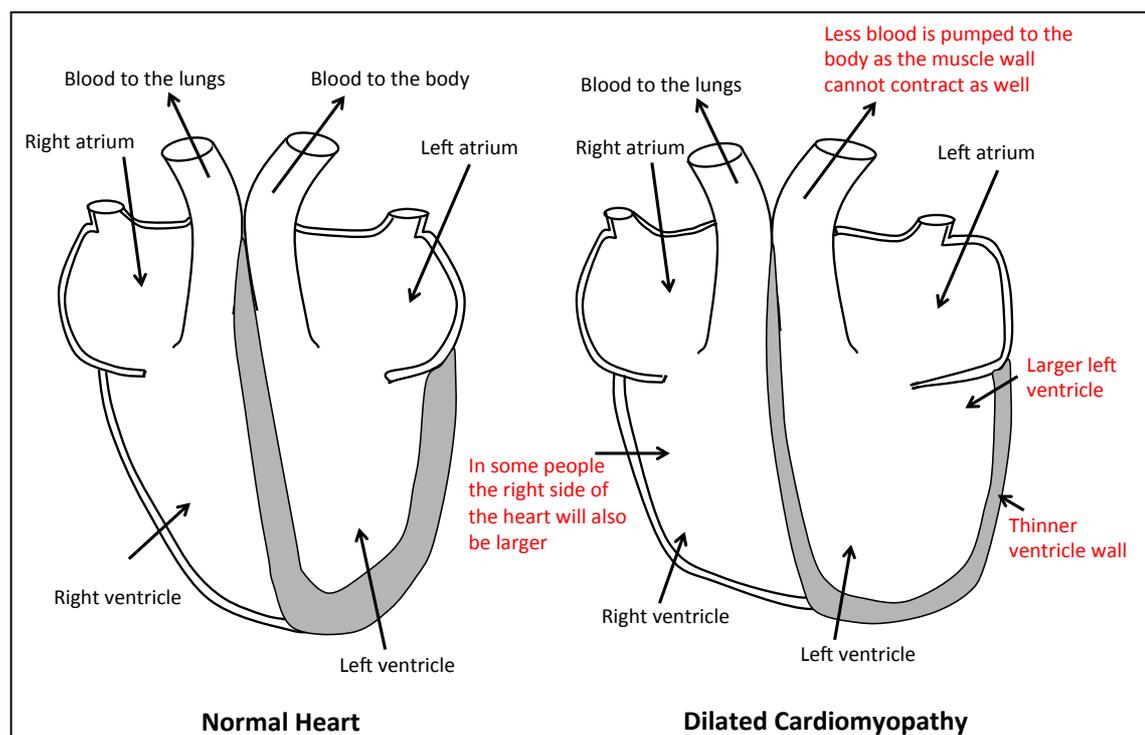


## Familial Dilated Cardiomyopathy (FDC or DCM)

### What is dilated cardiomyopathy (DCM)?

Dilated cardiomyopathy is a type of heart muscle condition affecting the left ventricle (the main pumping chamber of the heart). The left ventricle enlarges (dilates) and this affects the ability of the heart to effectively pump blood (shown below). DCM can affect both adults and children, and the symptoms range from none, to mild to severe. DCM can also lead to heart failure. When the pumping function of the heart is very poor it can lead to symptoms such as shortness of breath, swelling of the ankles and legs, and fatigue. In some cases DCM may affect the electrical system of the heart, leading to rhythm problems, and this may require implantation of a pacemaker/defibrillator. Rarely, DCM can cause sudden death.



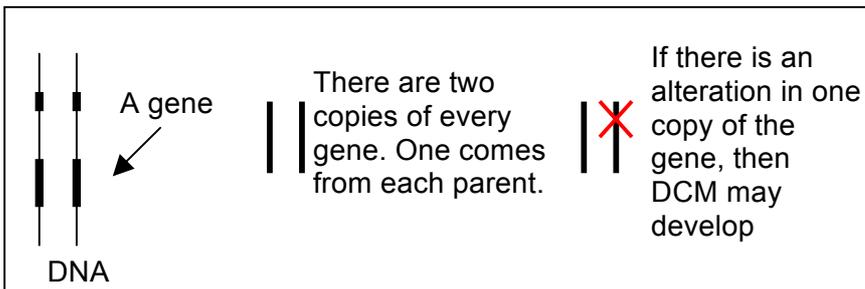
### What causes dilated cardiomyopathy?

There are numerous conditions that lead to a DCM. These include prolonged alcohol or drug use, viruses and thyroid disease. However in up to 50% of cases no cause can be identified. Familial (inherited) DCM accounts for approximately 20-50% of these cases and is caused by an inherited gene fault. In these cases clinical investigation of first-degree family members (parents, siblings, children) is very important. Early detection of familial DCM is important so that disease progression can be carefully monitored and treated to hopefully prevent future complications.

## Genes and familial dilated cardiomyopathy

Our body is made up of millions of cells, including brain cells, liver cells and heart cells. Each cell contains 46 chromosomes; chromosomes contain genetic information that guides our body's growth and development. These 46 chromosomes are grouped into 23 pairs, one of each pair coming from mum and the other from dad. One of these pairs is known as the sex chromosomes, and these decide whether we are male or female.

If you imagine a chromosome as being like a ball of wool, you could stretch it out into one long strand, which is known as the DNA. Along the length of DNA there are regions called genes. As there are two copies of every chromosome, there are also two copies of every gene (one inherited from each parent). Genes act as recipes to make certain things in the body, and each recipe is unique based on the order of units it is made up of. If there is a mistake in one of these genes (a



mix-up in the order of these units) it may lead to the development of disease. This mistake is known as a gene fault or alteration.

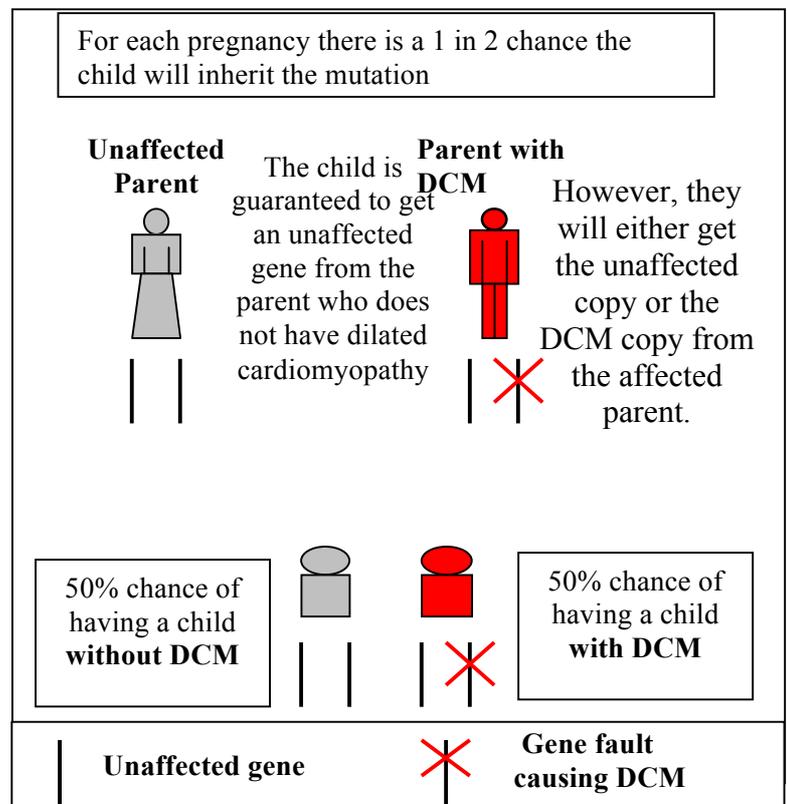
In the case of familial DCM, we know a single alteration in any one of 30 genes can cause the condition.

## How is familial dilated cardiomyopathy inherited?

In most cases, familial DCM is known as an autosomal dominant condition, whereby the gene with the fault “dominates” over the unaffected (normal) gene, causing the condition. This means that an affected person has inherited one gene fault causing familial DCM from one parent, and one unaffected gene from the other parent. Likewise, if a person who carries a DCM gene fault has a child, then **the chance that the child will be affected is 50%, or 1 in 2**. This is because they have a 1 in 2 chance of inheriting the gene fault causing DCM from the parent with the condition. This is illustrated in the diagram.

In some cases familial DCM can be inherited as an X-linked trait. This means that one gender may be more prone to developing the condition.

In rare instances a person may have a gene fault that has not been inherited from their parents. Instead, the fault has arisen some time after conception. This means that their brothers and sisters are not likely to develop the disease, however the gene fault can still be passed down to any children of the affected person.



## What is genetic testing?

Genetic testing involves looking for the alteration in the genes known to cause familial DCM. It is a long and difficult task that has been likened to looking for a single spelling mistake in an encyclopaedia set. This is because most families have their own unique gene alteration that may exist in one of the genes we have already identified, or even in a gene not yet discovered to be involved in this condition. To date over 30 genes have been found to cause DCM, however these genes account for a small proportion of this condition. Genetic testing in DCM is expensive and at the moment there is only a small chance (up to 20%) of finding the gene alteration causing the condition. Genetic testing for familial DCM is therefore not routinely offered at present, however, this may change in the near future as the technology for carrying out the tests improves. In some cases, particularly in families where there is also a history of conduction condition, then genetic testing is expected to be more informative.

If a gene alteration is identified in the affected person we can then offer a gene test to other family members. This test takes 4-6 weeks for a result, and will quickly determine if the family member has inherited the DCM copy or the unaffected copy of the gene. This is called predictive genetic testing, and allows a person to know if they are likely to develop the condition even before symptoms are present. If they do not carry the gene alteration then we can say that they will not develop DCM, and future ECG's and echocardiograms are not necessary. If they are shown to have the DCM gene alteration then there is a good chance they will develop the condition, and close follow-up would be recommended.

If you would like to know more about centres offering commercial genetic testing, please look at our Information Sheet on Cardiac Genetic Services, contact the Registry Coordinator on [registry@centenary.org.au](mailto:registry@centenary.org.au) or (02) 9565 6185 or speak to your regular cardiologist or doctor.

Genetic testing can affect applications for life insurance. If you would like to know more, please visit the Financial Services Council website at [www.fsc.org.au](http://www.fsc.org.au) for current guidelines (go to the Search bar and type in "Genetic") or call (02) 9299 3022.

## Research into hypertrophic cardiomyopathy (HCM)

**The Agnes Ginges Centre for Molecular Cardiology in Sydney has a large research program focusing on inherited heart conditions.** If you would like more information on this research program, please contact Ms Laura Yeates Cardiovascular Genetics Coordinator (02 9565 6187).

## Will a diagnosis of familial dilated cardiomyopathy change my lifestyle?

There are key recommendations we advise people about when they are diagnosed with familial dilated cardiomyopathy (FDC or familial DCM). These guidelines help to reduce the risk of the most devastating outcome – sudden cardiac death.

- **Avoid competitive sports and strenuous physical activity:** Competitive sports include those that require significant exertion, for example: Touch football, Basketball, Netball, Squash, running and even social games that you may not think are too strenuous. The international guidelines recommend people with dilated cardiomyopathy avoid competitive sports because high level exercise can trigger rhythm problems leading to sudden death.

- **Regular light exercise:** Although there is a lot of emphasis on avoiding sport, it is still important to maintain a healthy lifestyle. Regular light exercise such as walking is not only good for your heart, but also good for your general health.
- **Regular check-ups with your cardiologist:** It is important to monitor the progression of your condition, and you should also report any new symptoms.
- **Surgical procedures:** You should inform the attending doctor of your condition before any surgical or dental procedures.
- **Consult with your GP or cardiologist before taking any new medications:** It is important to talk to your doctor before taking any new medications, including over-the-counter (OTC) medications, complementary medicines, natural products and herbal remedies.
- **Avoid excessive amounts of alcohol:** Alcohol can have adverse effects on the body and can worsen the symptoms of DCM.
- **Encourage relatives to be screened:** As mentioned, siblings and children of an affected person have a 1 in 2 (50%) chance of being affected. Screening of relatives involves seeing a cardiologist to have a physical examination, an ECG and an Echocardiogram (ultrasound of the heart). The relative must inform the doctor there is a family history of hypertrophic cardiomyopathy to ensure no signs are missed. Find a specialist cardiac genetic service like the Genetic Heart Disease Clinics held at RPAH's Sydney Heart Centre (tel 02 9517 2011). See our Information Sheet on Cardiac Genetic Services to find your local service or speak to your regular cardiologist about screening family members.

### Further information:

If you have any further questions, please contact:

Your local GP or cardiologist

**The Cardiomyopathy Association of Australia** [www.cmaa.org.au](http://www.cmaa.org.au)

For your nearest contact person, contact the Heart Foundation's Heartline on 1300 362 787

**Australian Genetic Heart Disease Registry** [www.heartregistry.org.au](http://www.heartregistry.org.au)

Tel: 02 9565 6185 or email [registry@centenary.org.au](mailto:registry@centenary.org.au)

### Other helpful websites:

[www.cardiomyopathy.org](http://www.cardiomyopathy.org) Cardiomyopathy Association (UK)

[www.heartfoundation.org.au](http://www.heartfoundation.org.au) National Heart Foundation of Australia

[www.hearts4heart.net.au](http://www.hearts4heart.net.au) Hearts4Heart support group for young people with heart disease