Left Ventricular Noncompaction Cardiomyopathy

What is left ventricular noncompaction?

Left ventricular noncompaction is a disease of the heart muscle that has only recently been described. Left ventricular noncompaction can occur on its own (i.e. isolated) or along with other heart problems (i.e. congenital heart disease) and is characterised by deep trabeculations (finger-like projections) in the muscle wall of the left ventricle. These trabeculations can also occur in the right ventricle. The heart muscle abnormalities occur during the development of the heart in the embryo. Symptoms of the disease are variable; with some patients having no symptoms while others may develop shortness of breath, palpitations, chest pain, dizziness and fainting episodes. Occasionally the disease can cause heart failure, stroke (due to blood clots forming in the trabeculations then travelling to the brain) or sudden death.

![Diagram of normal heart and left ventricular noncompaction](modified.png)

It is not known how common left ventricular noncompaction is, although it is suspected to be relatively rare. There are generally two ages of presentation. The most common is during the first few years of life while other patients do not notice symptoms until adulthood. **It is very important that everyone with a family history of left ventricular noncompaction be seen by a cardiologist for testing.**

What causes isolated left ventricular noncompaction?

In many instances, left ventricular noncompaction is caused by abnormalities in our genes. Our body is made up of millions of cells. There are many different types of cells, including brain cells, liver cells and heart cells to name a few. Each cell contains 46 chromosomes, which hold the genetic material that decides features such as the colour of our eyes and whether we are tall or short. 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. A female has two X chromosomes (XX) while a male has an X and a Y (XY).

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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 (as mentioned, one from each parent). Genes act as recipes to make certain things (proteins) in the body. If there is a mistake in one of these genes it may lead to the development of disease. This mistake is known as a gene alteration, and such mutations may be responsible for left ventricular noncompaction.

**How is left ventricular noncompaction inherited?**
Families with left ventricular noncompaction (LVNC) have been shown to pass the disease on in two different ways, via autosomal dominant or x-linked inheritance.

**Autosomal Dominant Inheritance:** This means that an affected person has a 1 in 2 (50%) chance of passing the gene alteration on to children and males and females are affected equally. Most inherited heart diseases are passed on in this fashion.

**X-Linked Inheritance:** This type of inheritance has so far only been observed in a small group of patients who develop left ventricular noncompaction during childhood. In this case the gene alteration exists in a gene located on the X chromosome (sex chromosomes determine gender, i.e. females XX, males XY). Therefore, a father with left ventricular noncompaction has a 100% chance of passing the gene alteration on to his daughters but no chance of passing it on to his sons. Alternatively, an affected mother has a 50% chance of passing the gene alteration on to both daughters and sons. A female who inherits the gene alteration may have less chance of actually developing the disease (i.e. may never develop the disease). Rarely, a person may be the first in the family to have the mutation. In this case, brothers and sisters of that person are not likely to develop the disease, however children of the affected person are at risk of inheriting the gene mutation.

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<tr>
<th>AUTOSOMAL DOMINANT</th>
<th>X-LINKED</th>
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<td>For each pregnancy there is a 1 in 2 chance the child will inherit the gene alteration</td>
<td>The chance that a child will be affected depends on which parent has the disease. Males more often are affected</td>
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**Unaffected Parent**

- The child is guaranteed to get an unaffected gene from the parent who does not have LVNC

**Parent with LVNC**

- However, they will either inherit the unaffected copy or the altered copy from the affected parent.

**50% chance of having a child without LVNC**

**50% chance of having a child with LVNC**

- Unaffected copy
- Altered/affected copy causing LVNC

**Father with LVNC**

- All daughters will inherit the affected X, however may not develop the disease. The father is only able to pass on his Y to his sons

**Mother with LVNC**

- An affected mother has a 50% chance of passing on her affected X to both her sons and daughters. Again, her daughters may not develop the disease.

The gene mutation is carried on the X chromosome. Females have two X’s while males have an X and a Y.

Rarely, a person may be the first in the family to have the mutation. In this case, brothers and sisters of that person are not likely to develop the disease, however children of the affected person are at risk of inheriting the gene mutation.

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What is genetic testing?

Genetic testing involves looking for a mistake in the genes known to cause left ventricular noncompaction. At present, less than five genes have been identified to cause this disease and these genes are poorly understood. The Agnes Ginges Centre for Molecular Cardiology, Sydney is carrying out research to both identify gene mutations in patients with left ventricular noncompaction and to understand how these gene abnormalities cause heart disease. In addition to the genetic studies, clinical and family information is also being collected to help understand more about this disease. If you would like to take part in this research program or would like more information, please call Ms Laura Yeates, Cardiovascular Genetics Research Coordinator (Ph: 02 9565 6187 or email: l.yeates@centenary.org.au).

Will a diagnosis of left ventricular noncompaction change my lifestyle?

Relatively little is known about the clinical aspects of left ventricular noncompaction. Based on the limited available information on this condition, following are some of the Do's and Don’ts that we advise people when they are diagnosed with left ventricular noncompaction.

- **Avoid competitive sports.** 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. We recommend people avoid competitive sports because it may be associated with severe cardiac symptoms and possible sudden death.

- **Regular light exercise.** It is 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 the disease, and you should also report any new symptoms.

- **Surgical procedures:** You should inform the attending doctor of your condition before any surgical/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 alternative medications obtained from the ‘healing arts’ eg. herbal remedies.

- **Encourage relatives to be screened.** As mentioned, siblings and children of an affected person are at risk of also being affected. Screening of relatives involves them seeing a cardiologist where they will have a physical examination, an ECG, an echocardiogram (ultrasound of the heart) and possibly a magnetic resonance imaging (MRI) scan. The Genetic Heart Disease Clinic held at the Sydney Heart Centre, RPAH, is always available for the screening of relatives (Ph: 02 9517 2011).

The Agnes Ginges Centre for Molecular Cardiology carries out important research on left ventricular noncompaction and sudden cardiac death. Any donations towards this research would be gratefully received. Please contact Prof Christopher Semsarian if you would like to know more.

If you have any further questions, please contact:
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Australian Genetic Heart Disease Registry www.heartregistry.org.au

Cardiomyopathy Australia wishes to thank all of the above for their kind permission to use this important information for the benefit of all members
Our sincere thanks to members who have sent their appreciation of recent newsletters. It is really great to know that you are gaining information from our newsletters as that’s what it’s all about.

Here are some examples of what has been sent……..

Thank you Margot and John…. wonderful newsletters and I always get a lot out of them, as do our patients.
Margaret (nurse) Canberra

To all involved with the Newsletter:
Many thanks for another very informative and interesting newsletter, There must be so much involved in researching, collating etc: I have no doubt you are already thinking about the next one.
As a result of my HOCM, I’ve lost the sight in my left eye, so I have a love/hate relationship with my computer. Receiving the newsletter online means I can read the general content but print the articles of most interest to me and most relevant to my situation. An ideal arrangement and much more economical. Thank you.
My next challenge is to get a copy of Margot’s book—I’ll enlist help from a more savvy member of the family!
Thanks again for the help and encouragement and please know that your hard work is appreciated.
Alison ( NSW)

I have been in touch with Alison to let her know that when she gets a copy of my book she will be able to increase the size of the type if she has an Ebook reader. I don’t need my reading glasses when reading on my Kindle as I choose which size type I want. If Alison decides to read the book as a PDF on her computer she can also increase the type size.
Happy reading Alison
Cheers
Margot

When any of us are given the news that we have a cardiomyopathy (CM), we just don’t know what to do, what to tell our families and what it all means for our future. It is now nearly thirty years since I was told I had cardiomyopathy and the usual prognosis in those days was either “You can drop dead any minute,” or as was in my case “go home and get your affairs in order as you won’t last more than six months.”

Many people with a chronic illness can see no positives in the first instance but believe me as time progresses we usually console ourselves with the fact that “it could have been worse.” We all live varying lengths of time whatever our health situation is…. none of us really knows when it is our time to leave this life but it certainly doesn’t help ourselves or those around us to dwell on this fact. Live every day as it comes and particularly as you reach your senior years (and many of you will) even with cardiomyopathy as diagnostic techniques are continually improving, medication, is readily available and improving so as I often say to CM folk, “If there’s ever a good time to contract CM, it is now”… believe me. When I was diagnosed there was very little medication suitable for CM and what there was had so many side effects there were times when you felt you couldn’t continue with it. But then the reality hits and you ask yourself “what is the alternative?” So we all persevere and gradually get used to this new life.

So what are the positives?
If you’ve live in the same place since you were first diagnosed, you will be on first name basis with your pharmacist, your doctor’s receptionist and your friends will always keep in touch to see how you are unless of course you’ve bored them senseless with your complaining and blow by blow details of your illness, so watch out for that one. Eventually your life will carry on in a ‘new’ normality and there will be days when you actually forget you have CM. Now if you’ve moved around as we have in the time I’ve had CM, you have widened all the range of people I have mentioned above.
Make your CM your hobby or main interest and learn everything you can about it. Knowledge is power.

When or if you are hospitalised along the way, you are always an interesting patient because not everyone has CM. Newly qualified doctors and nurses are interested in your story as they learn a lot from chronic illness patients, particularly those with CM and knowledge of their condition. They enjoy speaking with those with a happy although ‘sometimes’ tired smile on their face.

CM people are usually told, “You don’t look like a cardiac patient.”
So remember to take every day as it comes and each day when you wake up look in the mirror, smile at your reflection and say to yourself, “I told you so… we’re still here and winning.” #
To those with cardiomyopathy….Have you been checked by your GP for your ability to Drive a Car.

An important reminder to all members both old and new.

We have had quite a few enquiries lately about regulations for driving if one has cardiomyopathy. A couple of years ago when the new legislation came into being we ran full particulars in the newsletter. Rather than repeat the large document we presented then, we have given the link for members to check out this government paper and suggest you be aware of the consequences of not seeing your GP to get a medical certificate stating you are fit to drive. No certificate—no driving otherwise no insurance for you or anyone else who might have an injury from being a passenger in yours or the other driver’s cars. An accident will cost you big money. It is vital that you read the full paper on the requirements but check the index and you can then go to the correct part instead of having to plough through a lot of unnecessary reading. There is no point in stating that you are a careful driver because there many out there driving who are not careful and could cause you to have an accident.
The misconception is, if you have cardiomyopathy, you can’t drive but this is not correct. You can drive as long as you carry your medical certificate. If your GP tells you that you are well enough to drive PLEASE tell him you want a letter to that affect and forward it to the State Motor Registry who will then send you a certificate for the doctor to complete. If the doctor does not act in this responsible manner he can held in breach of the act and that will cost him a lot of money.

Summarised below are the new standards relating to cardiomyopathy.

For more information about the standards, the full document can be downloaded from www.austroads.com.au You can also order a hard copy from this website.

What about your reporting responsibilities?
In ALL STATES AND TERRITORIES it is the responsibility of drivers to inform the licensing authority if they have a long term or permanent health condition that is likely to affect their driving ability. In addition, in some states (namely South Australia and the Northern Territory) health professionals are required to make a direct report to the licensing authority if a patient is diagnosed with a condition that may affect their fitness to drive (this is called mandatory reporting).
In previous CMAA newsletters we have drawn members’ attention to the fact that the medical standards impose licensing restrictions for various heart conditions and that people with cardiomyopathy for example should discuss with their doctor the impact of their condition on their ability to drive safely.
We have been aware that some CMAA members have been irritated by these requirements and so may have decided not to report to the licencing authority. Many people do not realize the implications of not reporting. There are for example heavy fines, and if you have a car accident and it becomes apparent that you have a chronic health condition that affects your driving and you have not reported it, your insurance may not be valid.
The reporting requirements are not dictated by the medical standards but reflect road safety legislation in the individual states and territories. These laws are summarized in the Appendix 3 in the standards for easy reference. Your medical professional will be aware of these requirements so PLEASE check with them and ensure that you have fulfilled the necessary reporting requirements and that you have fulfilled the requirements for a conditional licence.
Remember, the medical standards and the road safety legislation is designed to make sure you are safe on the road. Conditional Licences are designed to make sure your medical condition is monitored regularly so that you can remain independent for as long as possible.

Remember go to www.austroads.com.au. If you do not use a computer ask a friend or relative to checkout the website for you and print you a copy of the paper. You can also phone the Department and order a hard copy from this website.#