Cardiomyopathy Association of Australia Ltd

MARCH 2024 - THE LATEST IN BIG-HEARTED NEWS

AT A GLANCE

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Letter from the President

Hello everyone,

March already! The year has started off with a bang here at the CMAA. I'm pleased to bring you an update about some of the initiatives that are under way. We are delighted that the CMAA has successfully secured a grant from Microsoft providing free access to their full suite of Office products. While seemingly small, such grants allow more money to go towards patient-centric initiatives including our upcoming website redesign. We know it can be very helpful to hear from other patients, to not feel so alone, and to hear how others manage, and we've reached an agreement with a pharmaceutical company to produce patient story videos for the website. With a focus on Hypertrophic Cardiomyopathy initially, additional funding sources for other cardiomyopathy types are being pursued. If you are interested in telling your story, either in writing or on camera, please e-mail info@cmaa.org.au.

In our last newsletter we mentioned gene therapy trials, and had some queries in response about how gene therapies work. We've included some more information here, and will go into further detail in future editions.

Last edition we mentioned the current enquiry into genetic discrimination in life insurance. The CMAA made a detailed submission to ensure the voice of cardiomyopathy patients was heard. The government is now considering the submissions received, and we'll keep you up to date as announcements are made.

We know that one of the most common questions asked when diagnosed with cardiomyopathy is 'which cardiologist should I see?'. In an attempt to assist, we've decided to produce a register of recommended specialists for our website. If you have had an excellent experience with a dr, or know of a great dr by recommendation, please let us know. Read more below.

There are some patient events coming up which we're please to share with you. We've also included some info about current research. It is encouraging and hopeful to know that there are people working to better diagnose and treat cardiomyopathy, and we'll try to include info about new research, developments, and trials regularly.

If you'd like to get in touch, please do. Our e-mail is info@cmaa.org.au .

What is Gene Therapy?

What are Genes?

The majority of our genetic material is housed in 23 pairs of chromosomes within the nucleus of our cells. DNA within each chromosome contains instructions that define our individual traits. Genes, which are distinct segments of DNA, play a crucial role in regulating various characteristics, such as hair colour and height. Typically, individuals inherit two copies of each gene from their biological parents. It is estimated that humans possess between 20,000 to 25,000 genes in total.

What is a Gene Variant, or Mutation

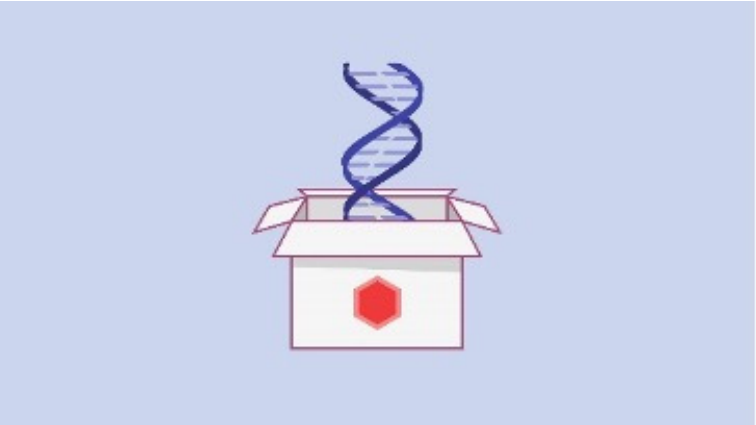
Proteins play a crucial role in the functioning of our body. Genes serve as the instructions for producing proteins for the body's functions. However, these instructions may not always be accurate. A gene variant, also referred to as a gene mutation, is a small alteration in the DNA within our genes that can modify the instructions for protein assembly and function. Gene alterations can be inherited from parents, occur with aging, or result from environmental factors like chemicals and radiation. While genetic changes are a natural occurrence, certain rare alterations can lead to diseases such as genetic cardiomyopathy by affecting the functioning of specific proteins.

What is Gene Therapy?

Gene therapy aims to address the underlying cause of disease, by addressing changes in genes. If genes are like instructions for our body, gene therapy can fill in missing parts or correct errors in the instruction's details.

Gene therapy is the use of genetic material to treat or prevent disease. The genetic material that is delivered to the patient has instructions to change how a protein, or group of proteins, is produced by the cell. For some diseases, this means making changes to account for too much, not enough, or incorrect essential proteins being produced within cells.

This new genetic material, such as a working gene, is delivered into the cell using a vector. A vector is like a package used to deliver a specific message. Viruses can be used as vectors because they have evolved to be very good at getting.into cells. Scientists have learned how to remove the viral genes and use this same ability to treat or prevent disease. All viral vectors are tested many times for safety prior to being used in humans.



A vector is like a package used to deliver a specific message

The vector can be delivered in one of two ways:

- ex-vivo treatment removes the person's own cells and delivers the genetic material to these cells outside the body. The modified cells are then returned to the body.
- in-vivo treatment means the genetic material is delivered directly into the person, such as through an injection.

Next newsletter, we'll delve further into one of the technologies currently undergoing human trials for one gene in hypertrophic cardiomyopathy patients

Source information and picture: American Society of Gene and Cell Therapy

Cardiologist / Specialist Register

Other patient organisations have registers of recommended specialists available, and we recognise this is something highly valued by patients. We aim to develop a register for our website of those drs who come highly recommended by cardiomyopathy patients. We will also include those specialists who are working in the specific types of cardiomyopathies, for example, have published research or have expressed a particular interest in aspects of cardiomyopathy management. The CMAA will act as both a conduit for patients to share their recommendations, and a source of information about those who are working in the cardiomyopathy space. If you have had a great experience with a cardiologist / surgeon / electrophysiologist etc, please e-mail us at Info@cmaa.org.au with your type of cardiomyopathy, and why you like that particular dr. (Your name and details will be kept confidential). We will write to the cardiologist to gain their permission and ensure we have correct contact details before placing their details on the register.

If you have had a particularly bad experience you'd like to share, feel free to let us know. We will keep this information confidential and it won't appear in any register, but we're interested to know about the patient experience, good and bad, in order to ensure we are well informed.

Upcoming Events

The Royal North Shore Hospital free ICD Education Session

Online via Zoom

Date: Tuesday 5th March 2024 3pm – 4pm

Speaker: Dr Logan Kanagaratnam, Cardiologist, RNSH

Topic: Ventricular Arrhythmias, Causes, Diagnosis & Treatment

For more information click here. If you would like to attend please register via the link below:

https://us02web.zoom.us/meeting/register/tZAscumvrzwrGdZasaY4ks25ZIUKYzHyvfRN

Marfan and Aortopathy Conference Brisbane

Dr Yong Shen Wee is hosting a patient conference for Marfan and Aortopathy patients and families. From the flyer:

Join us for a special one-day conference dedicated to raising awareness of Marfan Syndrome, aortopathy, and associated conditions. Tailored to people impacted by these conditions, this conference is an opportunity to gain insights from leading specialists as we come together to share knowledge, foster understanding, and promote advancements in the field.

For more information and to register, click here

Research Spotlight

Alfred Health Artificial intelligence (AI) assisted voice analysis for the evaluation of breathlessness.

This research project is aiming to develop a new test to help evaluate the cause of breathlessness. The test involves analysing the patterns of speech and breathing whilst talking. Your voice will be recorded using a mobile phone and analysed using artificial intelligence.

Who can participate?

You may be eligible for this study if you:

- are aged over 18
- have heart failure.

What is involved?

Your involvement will take a few minutes and doesn't require a clinic visit. Participants will be instructed to download an app on their smartphone and record their voice for a minute every week for up to 52 weeks.

Your involvement can help people with chronic heart failure access an easier and cheaper home monitoring tool that can save lives and enhance quality of life.

If you would like further information, please contact the Study Coordinators:

T: (03) 9076 3040
E: hfresearch@alfred.org.au

Cardiomyopathy Genetics Research - NSW Hearts is now a nation-wide study

NSW Hearts began as a study within NSW but has now expanded to all of Australia.

It is a research study aiming to better understand the causes of inherited cardiomyopathies. We plan on doing this by recruiting over 3000 people living in Australia with an inherited cardiomyopathy.

We are asking you to consider enrolling:

1. If you are over 17 years old
2. Diagnosed with inherited cardiomyopathy
3. Live in Australia

In this study we will ask you to complete surveys and individuals in NSW will provide a blood sample.

If you are interested in learning more about the study or participating, you can contact the

study team at nswhearts@populationgenomics.org.au

Reach Out

For questions, feedback, article ideas, or story contributions, email info@cmaa.org.au, **and we'll be in touch.**