Genetic research could hold the key that will change many lives including Chris’s

When Chris’s husband Vic proposed, she warned him: “You’re marrying someone who’s sick and I’ve got no idea what’s going to happen with the rest of my life.”

Thankfully Chris and her husband have just celebrated their 20th wedding anniversary. But there were many times during their marriage where Chris believed she may not survive the year, let alone celebrate such an important milestone.

You see, Chris has a condition which means she could have a massive stroke or heart attack at any moment.

Your support could help change that and provide a future for Chris, and women like her, that is free of heartache and worry.

Chris’s symptoms began when she was in her 20’s. She started to suffer from terrible migraines and nausea, lost a lot of weight and had vertigo. And most concerning, she had extremely high blood pressure.

A normal reading for a healthy adult is 120/80. At times Chris’s blood pressure soared to 250/150 – almost twice as high as it should have been.

Each morning when she woke up, she would wonder whether that would be the day she would suffer a stroke.

Despite being on six different medications, her blood pressure refused to go down. Her doctors were stumped.

It took 15 long years of struggling with her symptoms before Chris finally received a diagnosis. A surprisingly common but little-known vascular disease called fibromuscular dysplasia (FMD). A disease which can make the artery walls too weak or too stiff and affect any medium-sized blood vessel anywhere in the body.

It’s estimated one in 20 Australians have mild forms of FMD and 90% are women. At present there are only a handful of genes linked to this disease, and no specific treatments.

As a result of years of high blood pressure one of Chris’s kidneys was so severely damaged it had to be removed.

She has had approximately 12 angioplasties, a procedure to reduce blockages in different blood vessels, including her kidney, arm and stomach.

For Chris and her husband, celebrating their recent 20th wedding anniversary, had a special poignancy. There was no guarantee when they said their vows that Chris would live to celebrate this milestone.

Thanks to research being conducted at the Victor Chang Cardiac Research Institute, Chris and her family finally have hope that a treatment for FMD might now be on the horizon.

Working with international collaborators, Professor Jason Kovacic was recently involved in the discovery of five genes implicated in FMD. This is the first important step to finding effective treatments for this potentially fatal disease.

This exciting breakthrough is just the start. With the continued support of generous donors like you, we can better understand the cause of FMD, and can work towards treatments for this common and sometimes fatal disease.
Thank you

We all hoped that 2021 would be a better year than 2020 but unfortunately, we have all continued to face unique and unprecedented challenges. For many it has meant isolation, lockdowns and not seeing friends and family for many weeks, months or even years. And it has provided many challenges in the workplace that have also been felt here at the Victor Chang Cardiac Research Institute.

But thanks to your wonderful support, vital breakthroughs and discoveries have continued to be made at the Institute, as we too have adapted to the new world we live in. The fight against heart disease has never ceased.

In this edition of The Beat, you will read about the exciting discovery by Professor Richard Harvey and his team that may lead to ways to repair damage after a heart attack.

We are also pleased to announce our research capability has grown with the establishment of the first dedicated hub of heart research in Western Australia. This will enable the Institute’s Professor Livia Hool to continue her important research into a treatment for hypertrophic cardiomyopathy, the leading cause of cardiac death in children between the ages of five and fifteen.

And there are some handy heart health tips to help you prevent heart disease. Surprisingly they are not as hard to do as you might think.

Thank you again for your loyal support, particularly in these difficult times, your commitment to heart research is invaluable.

Warm regards,

Professor Jason Kovacic
Executive Director

Solving the puzzle that is the cause of enlarged heart syndrome

Dr Helena Viola likens her role as a medical researcher to that of a detective. Both require time and perseverance to uncover the clues and carefully piece them together.

Cardiovascular biochemist Dr Helena Viola has been working alongside Professor Livia Hool to solve the puzzle of hypertrophic cardiomyopathy, otherwise known as enlarged heart syndrome and the leading cause of cardiac death in children aged five to 15.

She and Professor Hool have been researching the cause of this inherited heart condition together for the last 10 years.

Excitingly, Dr Viola believes life-saving answers are just around the corner. Dr Viola and Professor Hool have discovered a mechanism involving a ‘communication breakdown’ in the heart that occurs due to genetic mutations. They are currently working on therapies aimed at restoring these communications to prevent the development of hypertrophic cardiomyopathy and cardiac arrest.

Knowing her work may one day save lives keeps her focused and motivated.

“I have seen the effect this disease has on patients and their families. My main goal is to play a role in developing therapies to help people impacted by this disease.”

Dr Viola has always had an inquiring mind and was encouraged by her parents to always ask “why” and not accept things at face value. This curiosity and pursuit of answers shaped her interest in science and makes her an outstanding researcher.

Dr Viola is determined to solve the complex puzzle that is hypertrophic cardiomyopathy. Like many of us, her family too has been touched by cardiovascular disease, which has only added to her drive to try and find answers.

Your gifts are helping to support young researchers and the future of heart research.
You can mend a broken heart

Heart attacks cause permanent damage to the heart. Thanks to your support there may be a way to repair damaged hearts.

You, or someone you love may have had a heart attack. You know how distressing and confronting it is. For some people, the damage to their heart is so great it leads to heart failure or sudden cardiac arrest.

But a team of scientists led by Professor Richard Harvey at the Victor Chang Cardiac Research Institute has discovered an important protective response in the heart that could limit the damage done after a heart attack.

The study found that when our gene pathways sense a lack of oxygen (hypoxia) after a heart attack they mount a protective response. If this response is blocked, chemicals called oxidants build up in the heart’s cells called fibroblasts. Too much of this oxidant causes the fibroblasts to expand and this can lead to excessive scarring and damage to the heart muscle.

The next exciting stage of the research is to develop synthetic antioxidants to repair some of this damage to the heart.

Dr Vaibhao Janbandhu, the paper’s lead author says “Hopefully what we have discovered will lead to the pursuit of synthetic antioxidants that can specifically target different cell types in the heart after a heart attack, including fibroblasts, to limit and even repair some of the damage.”

Your generous support of heart research helps make studies like this possible, which will hopefully lead to new treatments for heart attack patients. It is truly life-saving support.

Open your heart and tell us your story

By sharing your experience of heart disease, you can help others. Your story can raise valuable funds and increase awareness about the dangers of heart disease.

Will you become a heart hero by sharing your story? You will help raise vital funds for heart research. And your story may just prompt someone to go the doctor and have a heart check-up. It may just save a life.

We’d love to hear from you. Rest assured we will only share your story once we have your full consent and approval.

Emma tragically lost her husband Jeff when he suffered a heart attack, their son Seb was just five years old at the time. Emma has since committed to sharing her story to raise awareness for the importance of heart health and to help raise funds for heart research.
You are helping to solve the mystery of why high blood pressure is one of the causes of heart disease

The link between high blood pressure and heart disease is well known. What is not known is how high blood pressure changes the cells that cause heart disease. Professor Boris Martinac is determined to find out.

High blood pressure remains the most common risk factor for heart disease, you or someone you love may have it. But little is known about how high blood pressure leads to heart disease, specifically heart hypertrophy.

Heart hypertrophy is the thickening of the heart muscle. If the heart muscle thickens it can weaken the heart’s ability to pump blood efficiently through the body. This in turn can cause heart failure and sudden cardiac death.

We’ve known for a long time that high blood pressure is a risk factor for developing heart hypertrophy. But what we haven’t yet been able to understand is the mechanism that causes this hypertrophy change in the cells.

Professor Boris Martinac is leading an important project into uncovering this crucial link. As Professor Martinac explains: “If we can solve this missing piece in the puzzle and identify this new cellular pathway, we will be far closer to understanding the origins of this condition.”

And Professor Boris Martinac and his team have just been awarded a vital grant from the National Health and Medical Research Council (NHMRC) to help solve this mystery.

With just one in ten of these grant applications being successful and medical funding becoming more and more competitive, your support is more vital than ever to the future of heart research.

Our scientists simply cannot rely on grants alone to fund potentially life-saving research, and we cannot thank you enough for what you and others like you help make possible through your generosity.

The research project will focus on investigating, at a molecular level, two ion channels called Piezol and TRPM4 which are important for regulating heartbeats. There is still a limited understanding of the role these ion channels play in causing heart and vascular disease.

By understanding what is happening to the heart at a cellular level, it’s hoped we will be able to develop new treatments for tackling heart hypertrophy.

Professor Boris Martinac is passionate about finding the link between high blood pressure and heart disease. Professor Martinac has received vital funding for his research into heart hypertrophy.

It is only through research that the answers to the causes of heart disease will be found and many lives saved. Thank you for your ongoing support to heart research that saves lives.

There are various ways you can support vital cardiac research. You can choose to become a One Heart Supporter by making a regular monthly gift, hold a fundraising event, organise donations in lieu of presents or even include a gift to the Victor Chang Cardiac Research Institute in your Will. To find out more please visit victorchang.edu.au/support-us or phone 1300 842 867.

If you would like to discuss including a gift in your Will, please call Bequest Manager Samantha Burns on (02) 9295 8753 for a confidential conversation.
Genetic testing may hold the key to early detection of a fatal inherited heart disease

In families with a history of dilated cardiomyopathy there is now hope for early diagnosis and treatment.

Inherited genes are increasingly recognised as important causes of heart disease including dilated cardiomyopathy.

As Professor Dianne Fatkin explains “If we can identify family members at risk even before any symptoms develop, we can intervene straight away, which may help delay or potentially stop their progression to severe heart failure.”

Thanks to a recent study Professor Fatkin and her team have shown that whole genetic testing provides an accurate method to detect a range of genes associated with dilated cardiomyopathy.

This gives hope of an early diagnosis for those with a family history of the disease. They can receive treatment before any symptoms are present which could not only prolong but possibly save lives.

In families with a history of the disease, there is often an anxious wait to find out whether it will affect them or their loved ones. If an early diagnosis isn’t made, they may face the trauma of heart failure or worse still, need a heart transplant.

More opportunities to find causes and treatments for heart disease in WA

As a valued supporter of heart research, you will be pleased to know the Institute is growing, through the establishment of a dedicated centre for heart research in Western Australia.

The Victor Chang Cardiac Research Institute, in collaboration with the University of Western Australia and Wesfarmers, has established Western Australia’s first dedicated centre for heart research.

Led by Professor Livia Hool, the centre will continue working on her breakthrough discovery in the treatment of hypertrophic cardiomyopathy which is the leading cause of cardiac death in children aged between five and 15.

“About one in 500 people carry a genetic mutation for hypertrophic cardiomyopathy – or an enlarged heart. Excitingly, my team has discovered a way to prevent the heart from becoming larger. If it is effective in human trials, it will be the first-ever treatment to prevent the development of the disease,” says Professor Hool.

The centre will also undertake new areas of research such as understanding the mechanisms that cause sudden cardiac death. As part of this project, in international collaboration, the team is building a 3D-model of the calcium channel to find the safest, most effective way to deliver treatments that target the calcium channel in people at risk.

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Gifts from our kind supporters are vital to the Victor Chang Cardiac Research Institute’s ability to tackle long-term research projects such this. It has taken two decades of heart disease research and three years of genetic analysis. Thanks to you, many families now have hope for early detection and treatment.
EXERCISE

Walking is a great way to maintain a healthy lifestyle.

But how many steps do we need to take per day to really make a difference to our health?

In 2015 The George Institute of Global Health conducted a study which monitored 3,000 Australians with pedometers over a five-year period. It found:

- A person who increased their steps from 1,000 a day to 10,000 a day had a 46% lower mortality risk
- A sedentary person who increased their steps to 3,000 a day five days a week had a 12% reduction in death during 2021 an article in the Journal of the American Medical Association showed middle-aged people taking at least 7,000 steps a day had a 50-70% lower risk of death. For older women the benefits appeared to max out at 7,500 steps a day.

What’s the conclusion? Start walking! If you can, aim to reach around 7,000 plus steps a day. Your heart will thank you for it.

DIET

Most people have heard of cholesterol.

But just what is it and why is it important for heart health?

Cholesterol is the fatty substance found in animal-based foods such as meat, eggs and dairy and our body needs some to function normally. But there is good and bad cholesterol:

- HDL Cholesterol is known as ‘good’ cholesterol because it removes excess cholesterol from the blood and protects against heart disease. It’s a bit like a vacuum cleaner for cholesterol in the body.
- LDL Cholesterol is known as ‘bad’ cholesterol because it is linked to cholesterol build up in our blood vessels and increases heart disease risk.

In general, the lower your LDL cholesterol and the higher your HDL cholesterol, the better your chances of preventing heart disease.

Luckily, there are some simple dietary changes that can help you maintain the right balance.

Choose healthy unsaturated fats

- Oily fish
- Olive oil
- Nuts
- Seeds
- Avocado
- Margarine

Eat foods high in fibre

- Oats
- Legumes
- Avocado
- Nuts and seeds
- Sweet potato
- Broccoli

Include plant sterol-enriched foods

2–3g of plant sterols is equivalent to:

- Two cholesterol-lowering Weet-Bix or
- One tablespoon of enriched margarine (e.g., Flora ProActiv) or
- 500 mL Dairy Farmers Heart Active

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