MARFAN MATTER

Newsletter of the Marfan Association Victoria Inc.
www.marfanvic.org.au

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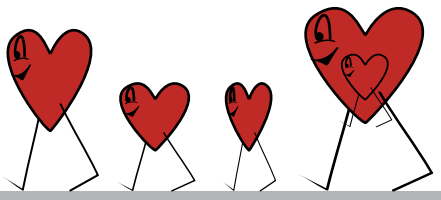
2017

New Committee

New Walk Location

New Beginning!



www.marfanvic.org.au Take a look at our website for news and updates

In This Issue

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A NEW CHAPTER BEGINS

It has been a long time between newsletters but we are pleased to announce we are back! The 2015/2016 year was a quiet one for the association. The committee were feeling drained after a few years of big projects such as the DVD and conference. The association was put on a hiatus year after we failed to get new interest in committee positions. We were in danger of folding. Fortunately a mother and daughter team Christine and Natasha showed interest, and long term member and former committee member Peter Laxby volunteered his time to join the committee once again.

The AGM was held last October at Lilydale Lake and the committee was elected:

President: Christine Adams

Vice President: Peter Laxby

Treasurer: Graeme Edwards

Secretary: Natasha Johnston

General Committee: Jean Edwards, Loretta Hecker, Lorraine Martini and Michael Crupi

There have been several committee meetings since October and the wheels are in motion to get the association back up and running. Plans are in full swing for our next Walk for Marfan event in October. Updates are being made to some of our printed material. A fun social get together is being discussed and opportunities to spread awareness are being investigated.

With genetic testing and medical advances some association members have had their diagnosis changed from Marfan Syndrome to other related disorders. Going forward the committee plans to expand our Marfan focus to be more inclusive of our "genetic cousins" with related disorders.

We look forward to the future and all the possibilities this new chapter has in store.

THANK YOU ROSSLYN

Last October our President Rosslyn Jablonsky handed over the reins to the newly elected committee. Despite her busy work and family commitments Ros has been dedicated to keeping the association operational and successful.

A Marfan group was initially established in 1989 but by the mid 90's was in hiatus. Ros' first position was as Secretary when she, Sally Ferguson, Justin Nix, Peter Laxby and Nancy Laxby re-established the group and incorporated the association in 1997. Ros says, 'As the first Secretary I wrote the original constitution and set up all the forms etc we needed. I didn't realise how big a job it was going to be until I was immersed in it. Having a five month old baby as well as a 3 year old at the time made it more challenging. I remember getting up to feed Alex at 5am and spending the time she was awake after her feed typing up the constitution etc. They were fun days!'.

Over the next eight years or so Ros held other executive committee positions sometimes more than one at time. She had a break for a few years from the executive committee when the demands of working full-time with two young kids was a bit too much. But Ros once again stepped up as the President around 2009/2010 when the association started to struggle once again.

'Initially we had lots of members on various sub-committees but after a couple of years the interest and commitment waned leaving the core executive members to take on all the load. We had meetings all over the place including at each others homes which were spread from Geelong to Broadford to Mitcham to Sunbury. For most of us it became a family affair, our Marfan Family. We had lots of great times together'.

We thank Ros for all her hard work and commitment over many years. Her dedication along with several other hard working members has ensured the association's operation and success over the last twenty years.

Introducing Tash Johnson

I'm Tash Johnston the new secretary for the Marfan Association Victoria. I live in Melbourne, have three beautiful girls and have been married for 13 years to a very intelligent man. In 2008 my second daughter came into the world in a spectacular fashion with her right knee hyper flexed backwards. Once the doctors and nurses recovered from the shock and my husband noticed my daughter had a cleft palate, my very pale, long limbed baby was handed to me without any instructions. From there I spent the next three years traveling from Daylesford to the Royal Children's to visit all of my daughter's 25 specialist's with a toddler in tow. After visiting a genetic specialist who spent the appointment on google, then seeing an orthopedic surgeon who noticed my daughter's blue tint in the white of her eyes, I decided to become my own genetic specialist. At 2am in the morning I started googling cleft palate, blue tint in white of the eye, hyper-flexible joints and hernia. I was directed to the Marfan website and then to a new site for Loeys-Dietz Syndrome. As the sun was rising I sent a photo of my daughter to Dr Hal Dietz at John Hopkins University in Boston USA describing my daughter's symptoms. The next day I received an email from Dr Dietz advising me my daughter had a lot of the symptoms and a list of suggestions to take to my next doctors appointment. At my next genetic appointment at 18 months my daughter was diagnosed with Loeys-Dietz Syndrome.

So why is a mum of a daughter with Loeys-Dietz Syndrome on the committee for the Marfan Association Victoria I heard you ask? Well there is a couple of reasons. The first one is because you guys have done such a great job of your awareness campaign that when I see a doctor or attend an emergency department I can quickly advise them my daughter has a Marfan like syndrome and they quickly know what I mean and how serious the syndrome is.

On my to do list has been to create a Loeys-Dietz Foundation, however I have noticed in the USA that the connective tissue disorders are starting to work together as the syndromes are rare and they have realised there is more power in numbers. Also before genetic testing, so many people have been misdiagnosed.

My interest in joining the committee began after attending the association's Marfan Conference a few years ago which I was very impressed with. I don't know about you but over the years I have struggled to educate doctors and advocate the needs of my daughter. I have argued with so many doctors, but have discovered that spending the time researching my daughter's syndrome and then presenting the details to my doctors has been the only way I have been able to get her the treatment she needs. Also sometimes this syndrome can be so lonely in so many ways. There's the ongoing blame game of what did I do while I was pregnant that my daughter ended up with this syndrome, the blame and sadness you share with your partner as you watch your child suffer, how do you manage to give your other children the attention they need and not show your daily fear of losing your child from a ticking time bomb.

Each week I receive phone calls from scared individuals from around Australia who are at a different stage of their journey. Every single person I have spoken to needed to be pointed in the right direction to get further information about Marfan Syndrome and other connective disorders so they could receive the right treatment. I've been fortunate enough to be able to direct them to some exceptional information on the Marfan website.

I look forward to working with you all and doing my best to continue the great work of the Marfan Association Victoria.

ADRIAN HECKER 11/2/86 - 13/5/17

Our deepest sympathy is extended to committee member Loretta Hecker on the passing of her son Adrian. Loretta writes:

On Saturday 13th May 2017 my son Adrian went about his day as normal, working on projects on his computer. About 3.30pm he walked from his room and said he did not feel well then collapsed. I immediately rang 000. Six ambulance officers worked on him but he could not be revived.

Adrian had a number of surgeries for aortic aneurysms including aortic heart valve replacement. During that time he completed his Bachelor of Science (Mathematics) with 1st Class Honors at RMIT followed by Master of Science at Melbourne University. He worked on projects at Peter MacCallum Cancer Institute developing programs that pin pointed the faulty cancer gene and received a scholarship for his work.

Adrian was such an intelligent gifted person with a wit and humor taken from us too early in life.
Thank you to the Marfan Association Victoria for their support and understanding for the past few years.

FROM THE PRESIDENT

Hi, I'm Christine Adams, President of the Marfan Association Victoria

"Where has she suddenly come from?" I'm sure you are asking, you have never heard of me and not seen me involved in the Marfan Association Victoria before.

Well you are right. I have only known of Marfan Syndrome from a friend who died from it over fifteen years ago. Then my daughter Natasha Johnston had her second baby 9 years ago and soon realised that something was wrong. At first it was discussed that there was a possibility of Marfan Syndrome. But after much research and lots of testing Felicity was eventually diagnosed with Loeys-Dietz Syndrome. The long road of loving, growing and working with a child with LDS started for the family. At the time all this was discovered, we were recovering from the 2009 bush fires. We didn't give as much time to Natasha and family as we should have and I always said "when I have got over this, I will give you more time"

The time came, when Tasha called me one day and said The Marfan Association Victoria is about to fold, they need people to get involved, and this was my time to get involved.

So who am I? I have lived in Marysville with my husband for thirteen years. We own the Marysville Caravan and Holiday Park. I have had a long history of being involved in many community and business groups, as chair of many of these groups. I very much enjoy being involved in organising events. I am hoping to use some of these skills in my involvement with the association. I am hoping over time that we will be able to look at other genetic disorders related to Marfan Syndrome and how we can work together and share our resources.

If at any time you want to have a chat, let me know what you are thinking or make a suggestion, I would love to hear from you.

The Sally Ferguson Award

The Sally Ferguson award was introduced in 2013. Sally Ferguson is one of the founding members of the association and served as president for ten years as well as other committee positions.

Congratulations to Narelle Phillips on receiving the award at the AGM last October. Narelle has been a committee member on and off for the past six years. In 2015 we held our first conference. Narelle worked tirelessly along with Ros, Kate and the rest of the committee who spent months organising the conference. She researched and oversaw the work done by our appointed marketing company and event planner for the conference as well as the work done in the lead up to the conference with our updates to the logo, website, and other literature. Without her help we would have struggled to reach the finish line and host a successful conference.

Narelle has readily worked behind the scenes on many projects including the DVD launch, administrates our facebook page and is a willing organiser and worker at the annual walk. We thank Narelle and the other volunteers like her who assure the continued success of our association.

The Marfan Association Victoria Inc. is a support group for adults, children and families affected by Marfan syndrome.

9TH Annual Walk For Marfan & AGM Sunday 29TH October 2017

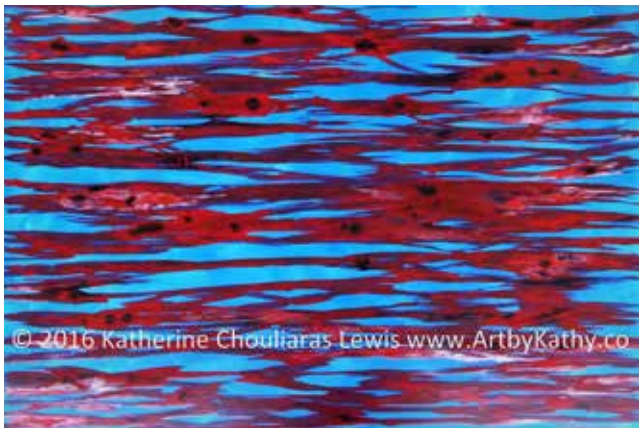
It has been two years since our last Walk for Marfan and we know you have missed it. We are pleased to inform you that the tradition shall continue with a bit of a twist. Anyone who attended our last Walk for Marfan will know how cold, wet and windy mother nature was for us. Some stayed home, many others still came and braved the elements. Thanks to the many determined volunteers and participants that still made the day a success.

Holding the walk at Albert Park Lake has been problematic due to Parks Victoria not allowing the reservation of a pavilion and the pavilion space being small. A volunteer is required to arrive at first light to reserve our usual spot and the transportation and set up of additional shelter along with tables and chairs is required. Then we have mother nature to contend with which at the last walk threatened to blow away our portable marquees.

The decision was made that we had to find another park location for our Walk for Marfan that had weather proof shelter and equipment on site that would ease the work required of our volunteers. That location had to be reasonably central, have good transport links and a pleasant walking track for us to enjoy our stroll for a good cause. We believe we have found just that at Caulfield Park!

As the committee was coming out of a hiatus year it was a challenge to organize a successful walk for our usual April timing. Winter isn't the best time of year for a fund-raising walk so we have pushed it out to spring. As our constitution required our annual AGM be held before the end of October, instead of planing for two event dates in spring we are combining the two.

The walk shall be held in the morning followed by lunch and social time. The afternoon shall finish with our AGM. Detailed specifics of the day will be communicated closer to the event, but for now mark Sunday 29th October 2017 in your calendar and be sure to invite your friends and family to participate in the Walk.



A Generous Donation

Rosanna based artist Katherine Chouliaras Lewis regularly holidays in Merimbula. She learned about Marfan syndrome from Carla Harris the owner of her accommodation in Merimbula and mother to Paris who has Marfan syndrome. Carla actively spreads awareness of Marfan syndrome including having our brochures and donation tin in her reception area. Carla also administrates an excellent facebook group called Marfan Network Australia.

Katherine felt moved to help our association's fund-raising effort and immediately started work on a painting with a connective tissue theme which she has donated to us. The painting titled "Connections" (acrylics on canvas 61cm x 91cm) can be viewed on Kathy's website www.ArtbyKathy.co and is easily identified among her other spectacular works by its prominent red and blue colour.

The painting will be used for a raffle or silent auction prize at the Walk for Marfan on 29th October 2017.

Thank you Kathy for your generosity.

Meet Your Gene:

An Introduction to the Marfan Gene and Current Research: A conversation with Hal Dietz, MD

Roanne Weisman, a healthcare writer from Boston who has Marfan syndrome and who is a long-time member of the Foundation, had a conversation with Hal Dietz, MD, Victor A. McKusick Professor of Medicine and Genetics, and Director, William S. Smilow Center for Marfan Syndrome Research, Johns Hopkins University School of Medicine, to get a basic understanding of Marfan research to share with our community.

What is a gene anyway?

A gene is a segment of our body's DNA that tells cells how to make proteins. DNA, (deoxyribonucleic acid) is the carrier of genetic information that makes us human, but also makes each of us unique. Every cell in a person's body has the same DNA. For most genes we each have two copies, one inherited from each parent.

What is a protein?

Proteins are the workhorses of the cells that make up our bodies. Each gene encodes (has instructions to make) a specific protein. Each protein has a defined task: Some proteins create our bodies' structures, such as bones, blood vessels, and organs. Other proteins, called enzymes, carry out specific biochemical activities, such as digesting food and converting it into energy.

What is the Marfan gene and how does it cause Marfan syndrome?

The Marfan gene encodes for fibrillin-1, which is a component of connective tissue—the material between the cells of the body that binds cells together and gives tissues form and strength. Besides acting like "glue," connective tissue proteins such as fibrillin-1 can provide instructions to neighboring cells that influence how they behave. There is strong evidence that fibrillin-1 participates in both types of functions. As with most other genes, every person normally has two copies of the fibrillin-1 gene. In people with Marfan syndrome, at least one of these copies has a defective sequence of DNA—also called a mutation—so the "instructions" to make fibrillin-1 are not quite right. As a result, the altered fibrillin-1 has a reduced ability to perform its intended structural and instructive functions.

What happens in the body when fibrillin-1 is not working properly?

The consequences of defects in fibrillin-1 vary depending on the tissue or stage of development being considered. Selected features of Marfan syndrome, such as eye lens dislocation, are thought to largely reflect failure of a structural glue-like function of fibrillin-1 aggregates (called ciliary zonules) that normal hold the lens in the center of the eye. In contrast, an altered ability of fibrillin-1 to regulate cell behavior is thought to contribute to many of the cardiovascular, lung, and skeletal features of Marfan syndrome. More likely than not, there is a complex combination of multiple factors that dictates when and where features of Marfan syndrome show up and how severe they will be.

If the defect is in only one gene, why doesn't every person with Marfan syndrome have the same symptoms?

There is no "common" Marfan syndrome mutation – indeed, more than 1,000 different mutations in the fibrillin-1 gene have been observed. The location and type of mutation can influence the quantity or quality of the fibrillin-1 protein that is made by the cell. This is thought to be a major reason why the severity of Marfan syndrome can vary between families. However, significant variation in severity can be seen among family members with the exact same fibrillin-1 mutation. This can reflect both "environmental" and "genetic" modification. Environmental modification means that our life experiences can influence our health in general and the influence of a genetic predisposition for a specific disease such as Marfan syndrome. It stands to reason that if someone makes good choices regarding medical care, exercise, and diet, for example, that they would be better off when compared to a relative with the same genetic predisposition, but less good lifestyle choices. Genetic modification relates to the fact that we each have many thousands of genetic variants that influence how our proteins behave. Those natural variants have functional consequences, even if they don't cause diseases themselves. That is why we look and behave differently from each other and from our parents and siblings. In essence, we have each been dealt a different hand with regard to natural gene variants. Sometimes these natural variants can protect people from the consequences of the Marfan mutation, making symptoms more mild. Others can accentuate the consequences and make symptoms worse. This means that even within the same family, two people with Marfan syndrome may have very different symptoms.

What is the focus of current research to improve the lives of people with Marfan syndrome?

Thanks to years of research, we came to the understanding that the fibrillin-1 protein also serves other important functions besides structural functions, and the course of our therapeutic focus has become much more promising. Currently, there is a strong research focus on the ability of fibrilllin-1 to regulate the activity of a class of molecules,

called growth factors. These molecules bind to the surface of cells and tell the cells how to behave. One growth factor, TGF-beta, has particular relevance for Marfan syndrome. Normally, TGF-beta is active during fetal development, encouraging growth, but is less active in adults, except at certain times, such as for wound healing. Normally functioning fibrillin is like a "traffic cop" for TGF-beta, signaling it to be active when it is needed and stopping or suppressing its activity when it is not needed. But in people with Marfan syndrome, this signaling system has trouble stopping the activity of TGF-beta. We learned that in both humans and mouse models with Marfan syndrome, there was clear evidence for too much TGF-beta activity.

What does too much TGF-beta activity to do the Marfan body?

We and others have focused on problems with the aortic wall enlarging and dissecting, but there is also good work that shows high TGF-beta activity contributes to problems in heart valves, skeletal muscles, and lungs.

Has this knowledge led to a therapeutic approach (medication)?

There has been a lot of activity to find potential mechanisms to adjust the level of TGF-beta in Marfan syndrome. Using mouse models, we found that a blood pressure drug called Losartan can potently suppress TGF-beta activity and can prevent aortic aneurysm and lung problems. Losartan not only prevented the structure of the aorta from getting too big, but it also fully preserved the normal architecture of the aortic wall. There was also evidence that it could stabilize damage, prevent new damage, and possibly even reverse existing damage.

What about Losartan for people?

Recent studies have suggested that medications such as losartan show strong promise for the care of people with Marfan syndrome and related disorders – performing as good or better than conventional therapies, such as beta blockers, in various studies. There is both room for improvement and many unanswered questions. Is losartan the best drug in its class (a group of medications called angiotensin receptor blockers or ARBs)? What is the optimal dose? Are there some people who will respond to ARBs and others who will not? Are there combination therapies that should be considered? Answers to these critical questions require more research and future clinical trials.

Is personalized medicine on the horizon for people with Marfan syndrome?

Individualized treatment is one of the most exciting and promising Marfan treatment research pathways right now. One day we may be able to design the treatment that would be right for each person. That is the goal of the ancillary studies that are still ongoing. We hope that we will be able to look at the genetic makeup of each person, as well as specific measurements of chemicals in the bloodstream, to determine how people are responding to treatments. This will help us predict whether we are on the right track or if we need to modify treatment.

What other research efforts have promise?

Now that we have discovered the connections between fibrillin-1 and TGF-beta, we are investigating what else might be "downstream" of TGF-beta that could or should be blocked. What are other biochemical pathways that are damaging to people with Marfan syndrome? Can we find effective ways to block those pathways? How does nature modify Marfan syndrome? If we understand how natural genetic variants can protect some people from the consequences of a fibrillin-1 mutation, perhaps we can identify drugs that can mimic nature's successful strategy. We anticipate that this will be a very exciting and fruitful research direction.

Roanne Weisman, MSW, has been writing for consumers and professionals in the areas of health, medicine, education, and science since 1985. (TheWriteWayToHealth.com.) Roanne has Marfan syndrome, as does her son, Ben Weisman, a Marfan Foundation Board member. The co-author of several award-winning trade health books with physicians, Roanne is a skilled "translator" of medical and scientific information. Her most recent book, In Sickness As In Health: Helping Couples Cope With the Complexities of Illness, is about the effects of serious injury and illness—including chronic conditions like Marfan Syndrome— on intimate relationships.

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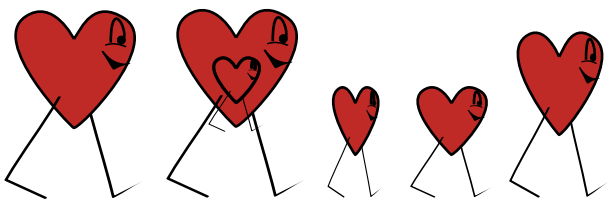

MEMBERSHIP RENEWALS

Membership renewals are due by 30th June for the 2017/2018 year

Why You Should Be A Member?

• To financially support the association's running costs. We are run by volunteers but we need financial support for operational costs such as website hosting, post and telephone charges, incorporation fees, insurance, meeting room hire etc.

• Boost the size of our membership. While it's true that membership payments assist our costs, they don't cover all of it. Donations and fund-raising meet the gap, but when we run projects such as our DVD and Conference, we rely on grant applications to fund the shortfall. Our membership level has fallen below the minimum level required by many grants to apply.

• Member only benefits. We offer open access to the public to our literature, online website and facebook information, telephone support, events etc so it may seem that you get little benefit in being a financial member. There are some perks for being a member such as this newsletter. Subsidised prices for items such as T-shirts and free or subsidised prices for events. Best of all, that good feeling you get from being a member of something that is assisting our community.

• Contribute to the future and success of the association. Time and effort is donated by the committee and the volunteers behind the scenes. Having you as a member makes that work feel appreciated and encourages more effort to keep the association working.

How much is membership?

• Annual membership cost is $20 for singles or $35 for a family (two adults and directly related children residing at one address). If your family has formally had only a single membership, upgrade to a family membership so we have a truer representation of our membership.

How do I renew or become a member?

• Complete the membership form on our website (contact the association for the form if you don't have web access).

• Payments can be made by:

1. POST: Cheque or money order to PO Box 477, Belmont, Vic 3216.

2. DIRECT DEPOSIT: NAB BSB: 083-644 Account: 68-896-4560 (It is vital that you include your NAME and MSHIP in the description).

Thank you for your membership and supporting your association!

CONTACT US TO CHECK YOUR MEMBERSHIP STATUS

The Marfan Association Victoria Inc. Was established in 1997. The goals of the Association:

- Provide accurate and timely information about Marfan syndrome to those affected by this genetic disorder, their families, friends, health professionals and other interested people
- Provide support and opportunities to share experiences and improve medical care
- Promote Marfan syndrome awareness within the community
- Support and foster research.

Please note: This newsletter is not intended for diagnostic purposes. Should you have any questions arising from information contained within the newsletter please refer to your personal physician.

MARFAN ASSOCIATION VICTORIA

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