# AMYLOIDOSIS news

CARING FOR PEOPLE WITH AMYLOIDOSIS AND THEIR FAMILIES

Leukaemia Foundation
VISION TO CURE
MISSION TO CARE

*Autumn 2016*


# Waiting for a new heart

**At only 57 years old, Mario Falcone of Sydney was not only diagnosed with life-threatening amyloidosis, but was faced with news that a heart transplant was his only option for a good outcome.**

*Mario and Leanne Falcone*

According to his devoted wife and carer, Leanne, the first signs of Mario's amyloidosis began in early 2014 when on his daily walk he found himself out of breath and experiencing sharp chest pains.

"In the following days Mario would come home from his walk and tell me that he had chest pains and was really struggling to breathe," Leanne said.

"Soon after, Mario ended up in hospital with what they said was a mild heart attack."

After a range of tests showed nothing, Mario's cardiologist decided to do a heart biopsy which revealed amyloid deposits around his heart.

Heart muscle biopsy involves the removal of a small sample of heart muscle that is then examined under a microscope to detect amyloid deposits. It is the 'gold standard' for diagnosing amyloid deposits in the heart.

"We had never even heard of amyloidosis, but were told the shocking news that Mario had only 12 to 18 months to live.

"The cardiologist referred Mario to the Westmead Amyloidosis Clinics in Sydney to find out what type of amyloidosis it was and the extent of amyloid around Mario's heart so they could work out the best treatment," Leanne said.

"Genetic testing was also done to see if it was hereditary. We have three sons and two grandsons and were so relieved when we found out it could not be passed on."

Mario was found to have wild type ATTR amyloidosis (previously known as senile amyloidosis), which is usually found in men over 70, not in their 50s like Mario.

When the thickness of amyloid around Mario's heart was measured it was found to be much worse than first thought. Mario and Leanne were confronted with the news that there was no cure or treatment.

Mario was put on a trial for the drug diflunisal which they hoped would slow the progress of the amyloid.

Mario's cardiologist also referred him to the St Vincent's Hospital Heart Lung Transplant Unit when it became apparent that Mario's only option for a good outcome was a heart transplant. Heart transplantation is not an option for a large majority of patients with wild type ATTR amyloidosis because they are normally much older.

Mario was told he would not be considered for a transplant unless he lost weight and in the last 18 months has lost around 50kg and continues to lose more.

"We didn't realise how complex the process of receiving a new heart was. The heart has to be the right size for your body and compatible, and there are many other factors that are taken into consideration," Leanne said.

"It has been a long, gruelling process but fortunately Mario is now on the transplant list and we are waiting for a new heart. ▶

# A message from the editor

**Welcome to our first edition of *Amyloidosis News* for 2016.**

**This edition focuses on cardiac amyloidosis with our front page article about 59-year-old Mario Falcone who has wild type ATTR amyloidosis. An article on page 3 about cardiac amyloidosis was written by Mrs Pat Neely, Honorary Support and Education Officer, Amyloidosis Centre, Princess Alexandra Hospital, and Dr Matthew Burrage, Cardiology Registrar.**

We understand amyloidosis treatment is very complex and can be not only physically gruelling but also emotionally draining. Suddenly the things in our lives that we could confidently predict and look forward to, become uncertain. On page 4, Clinical Psychologist Lynda Katona discusses ways those affected by a disease like amyloidosis can learn to live with uncertainty.

Unfortunately, as many of our readers know, peripheral neuropathy is a common problem for people living with amyloidosis and on page 6 and 7 we look at its causes and treatments.

To ensure that as many patients and their families have access to our support services, no matter where they live in Australia, we are recording a number of our support seminars that patients and their families can watch in the comfort of their own homes. Two of the latest seminars which can be viewed on our YouTube channel at youtube.com/leukaemiaqld feature peripheral neuropathy and kidney involvement in amyloidosis. DVD versions of these presentations are also available by phoning the Leukaemia Foundation on 1800 640 240.

Please remember that you are not alone as you live with amyloidosis. The Leukaemia Foundation is here to support you whether you are newly diagnosed, going through treatment, or months and years down the track. Please contact us if you need support or information.

*Sheila Deuchars*

Look out for support groups in your own state – smaller states have general blood cancer support groups which also welcome amyloidosis patients to attend. See the back page for more information.

**Sheila Deuchars**
Amyloidosis and Support Coordinator
Leukaemia Foundation

"Mario still visits the Westmead Amyloidosis Clinic regularly where they look after his amyloidosis, although a majority of his care is now taking place at St Vincent's which are looking after keeping his heart stable enough while he waits for a transplant."

According to Leanne, when someone is diagnosed with a disease like amyloidosis, it is important to have support from family or a carer.

"I always go to appointments with Mario to be another listening ear. Sometimes I think the information is so overwhelming that he just tunes out and I have to explain it to him later," Leanne said.

"When people say your life can change in a heartbeat – it is very true.

"Amyloidosis has changed Mario's life and that is very hard for him to deal with when he is only 59.

"We own the hospitality business and Mario was always very busy, but he has had to slow down which he finds really difficult."

However, Leanne is quick to add they have also had beautiful moments throughout their journey with amyloidosis.

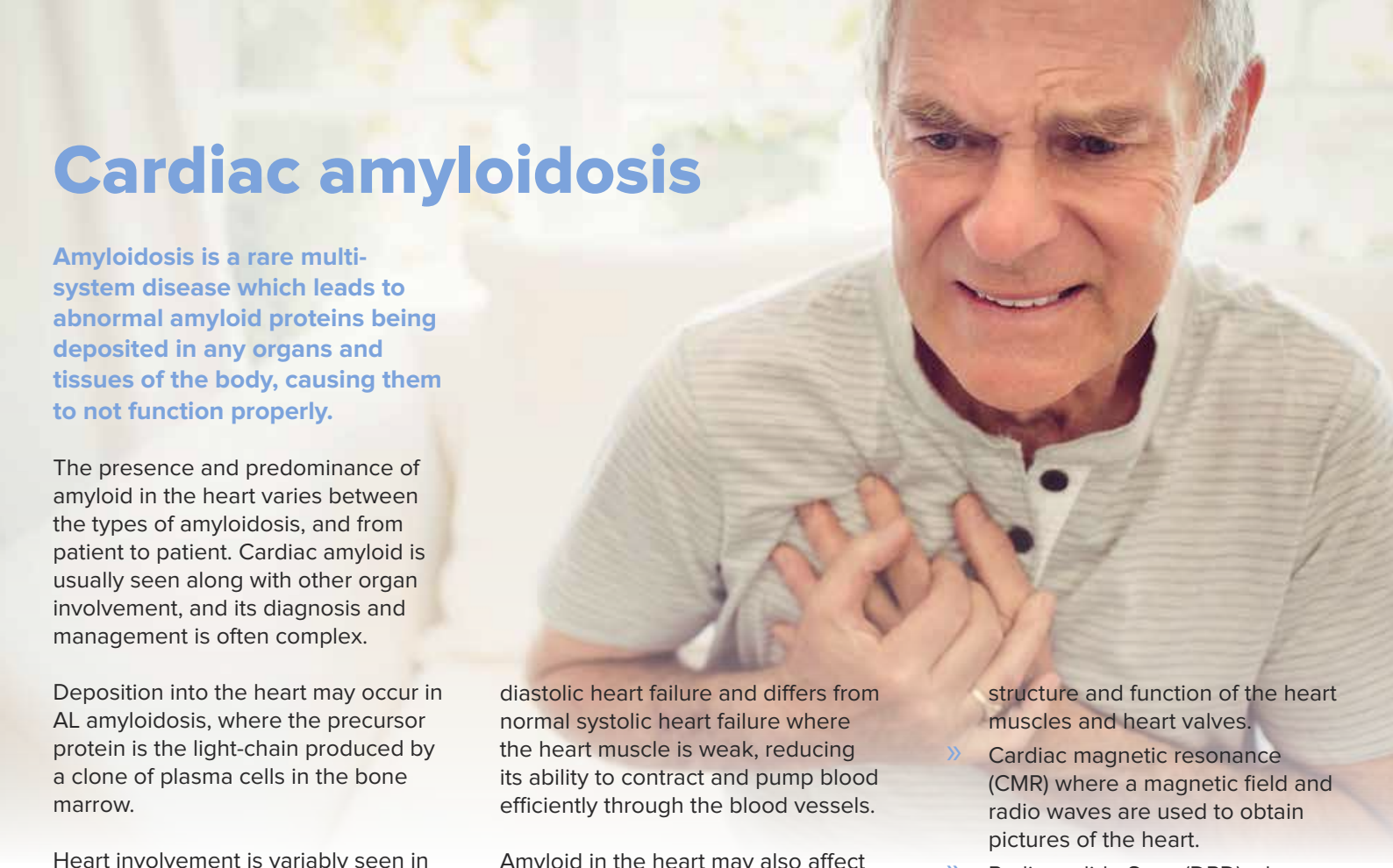
"We cherish our family. Our wonderful grown up sons, daughters-in-law, and grandchildren, aged 2½ and 4 months, bring us so much joy. Children have a way of making you happy even when you've had a bad day.

"We see life a little more clearly – we appreciate the people in our life and don't worry about the silly, trivial things that don't matter.

"For now, we have Mario's bag packed and ready when a heart becomes available – so we wait and hope."

**Transthyretin (TTR) is a normal blood protein, present in everybody. In healthy people, normal so-called 'wild type' TTR functions as a transporter of thyroid hormone and vitamin A (retinol) within the bloodstream. In wild type ATTR amyloidosis, formerly known as senile systemic amyloidosis, the normal 'wild type' TTR proteins clump together and form amyloid deposits, mainly in the heart and often also in the part of the wrists called the carpal tunnel. Wild type ATTR amyloidosis is not an inherited condition (does not run in families).**

# Cardiac amyloidosis



**Amyloidosis is a rare multi-system disease which leads to abnormal amyloid proteins being deposited in any organs and tissues of the body, causing them to not function properly.**

The presence and predominance of amyloid in the heart varies between the types of amyloidosis, and from patient to patient. Cardiac amyloid is usually seen along with other organ involvement, and its diagnosis and management is often complex.

Deposition into the heart may occur in AL amyloidosis, where the precursor protein is the light-chain produced by a clone of plasma cells in the bone marrow.

Heart involvement is variably seen in all types of familial ATTR amyloidosis, which is caused by the inheritance of an abnormal (mutant) transthyretin protein made in the liver.

ATTR wild type amyloidosis (or senile amyloidosis), typically seen in older men and not inherited, causes heart disease by a build-up of these proteins in the heart.

AA amyloidosis, occurring in some people with long-standing chronic inflammatory disorders, very rarely deposits in the heart.

Prognosis varies amongst the different types of amyloidosis and from patient to patient with the same condition. It does seem however that cardiac involvement in AL amyloidosis progresses faster than in senile amyloidosis or the hereditary type.

The amount by which amyloid affects the heart varies from patient to patient. In some cases there may only be minimal amyloid protein deposition, with no significant symptoms.

In more serious cases there can be widespread infiltration, which causes the heart muscle to become stiff. This results in the heart muscle being unable to relax properly, with impaired filling and function. This is known as diastolic heart failure and differs from normal systolic heart failure where the heart muscle is weak, reducing its ability to contract and pump blood efficiently through the blood vessels.

Amyloid in the heart may also affect the heart's conduction system by interfering with the electrical signals. This may result in an irregular heart beat (arrhythmia), palpitations, and blackouts secondary to heart block.

**Symptoms of cardiac involvement may include:**

- lethargy and extreme tiredness
- breathlessness on exertion
- swelling of the legs and ankles
- feeling light headed when standing
- chest pain mimicking angina
- weight loss
- loss of appetite.

Diagnosing cardiac amyloidosis can be complex. The diagnosis of amyloid is initially established through a tissue or organ biopsy. Protein staining with the Congo Red stain appears red under normal light, but becomes apple green under special polarised light. The tissue is then tested further to establish the type of amyloidosis.

Tests are also run to establish organ damage including the heart. These may include:

- an ECG to detect the electrical impulses controlling the heart contractions.
- A scan using ultrasound Echocardiogram (ECHO) which allows assessment of the heart's structure and function of the heart muscles and heart valves.
- Cardiac magnetic resonance (CMR) where a magnetic field and radio waves are used to obtain pictures of the heart.
- Radionuclide Scan (DPD) where a radioactive marker (DPD) hones in on the amyloid protein in the heart. This is most useful for diagnosing the ATTR amyloid protein and is less useful in AL amyloidosis. The DPD scan combined with a method called single positron emission computerised tomography (SPECT) allows much clearer evaluation of deposits in the heart.
- Blood tests. Two highly sensitive biomarkers: B-type natriuretic peptide (BNP) or N-terminal pro-BNP) and cardiac troponin (T or I) may detect the degree of damaged heart muscle.
- A heart biopsy performed to establish amyloid in the heart. This is the gold standard test but is highly invasive.

Goals of treatment are three-fold: to stop or slow the production of the amyloid protein, to care for the damaged organs and enhance quality of life.

In AL amyloidosis the extent of the amyloid load in the heart will influence all treatment decisions.

A combination of drugs may be used and in highly selected patients with very early cardiac involvement ▶

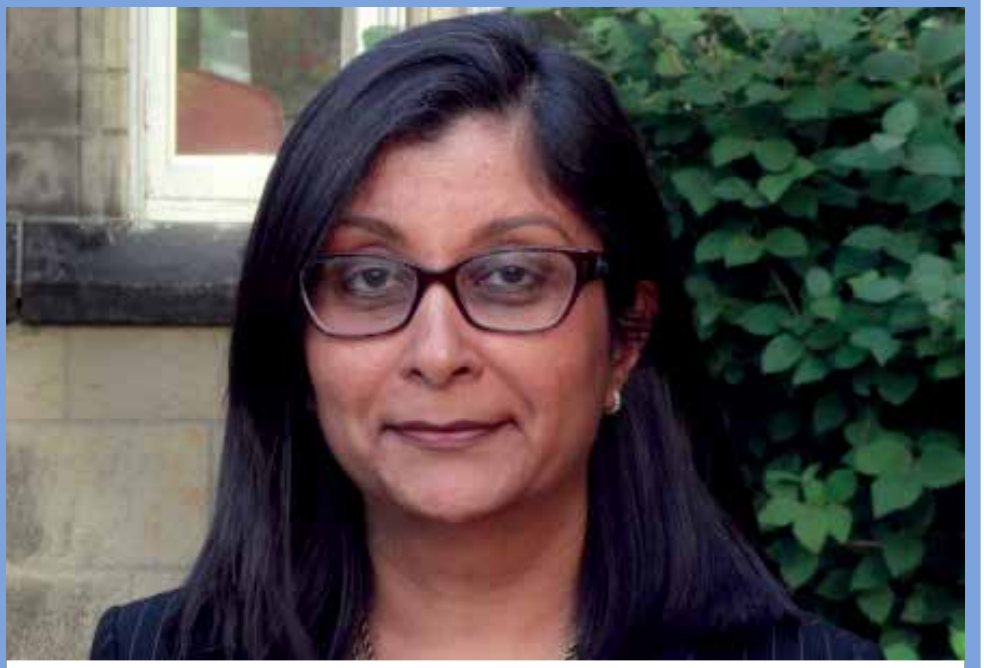
# High dose chemo and stem cell transplant improves survival for AL amyloidosis patients

**Patients with light-chain (AL) amyloidosis who are treated with high-dose chemotherapy (melphalan) and autologous (using their own stem cells) transplantation (HDM/SCT) have the greatest success for long-term survival, according to new research.**

The findings, which appeared in the journal Blood, report on the largest number of patients in the world receiving high-dose chemotherapy and stem cell transplantation as treatment for this rare disease.

HDM/SCT was considered an innovative treatment approach when it was first developed at Boston University School of Medicine (BUSM) and Boston Medical Center (BMC) in 1994. Since that time, 629 patients with AL amyloidosis have undergone this treatment at BMC.

Hematologic responses (the first indicator that treatment is beginning to work) were assessed in 543 patients at 6-12 months following HDM/SCT treatment. Forty per cent achieved a hematologic complete response (CR) post stem cell transplantation. Hematologic relapse occurred in 18.2 per cent at a median of 3.97 years post treatment.



*Vaishali Sanchorawala, MD, Professor of Medicine, Associate Director, Amyloidosis Center Boston University School of Medicine*

Long-term survival, up to 20 years, was achieved in nearly one-third of patients.

"While survival is strongly dependent upon achieving hematologic complete response (CR), the survival of patients who did not achieve a CR and of those who relapsed after CR is notable, suggesting a benefit of aggressive treatment," lead author Vaishali Sanchorawala, MD, Professor of Medicine and Associate Director of the Amyloidosis Center at BUSM, said.

"Strategies to better understand which patients may benefit the most from this treatment and reducing treatment-related mortality, as well as using combination therapies with novel agents to increase the CR rate, will likely improve outcomes in the future for patients who just a few years ago were considered to have a rapidly fatal diagnosis."

a stem cell transplant may be recommended in an attempt to rapidly suppress the production of the abnormal plasma cells, especially if the cardiac disease appears to be progressing. If this is achieved there is a chance that the amyloidal fibrils in the heart may leach out and the heart function will improve. This can be slow and may occur quite some time after haematological response.

In familial ATTR amyloidosis liver transplantation remains the established treatment for variant TTR–related cardiomyopathy.

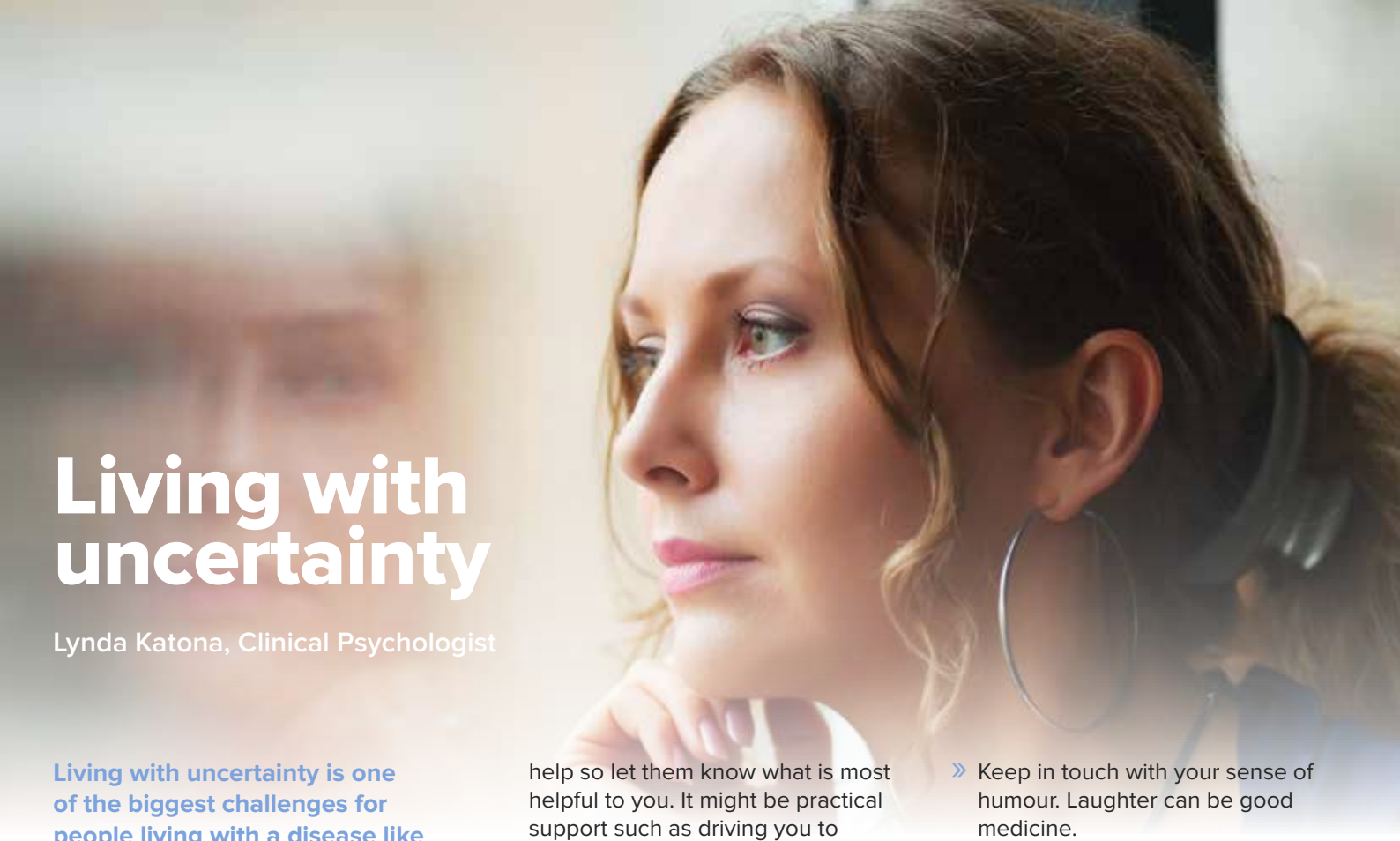
In ATTR wild type there is no treatment at this time to stop the normal wild type ATTR depositing in the heart.

Pacemakers may be recommended for certain patients with problems of the cardiac electrical system, and cardiac transplantation may be discussed in highly-selected younger patients.

A number of new drugs to slow the development of cardiac amyloidosis and neuropathy in all types of ATTR are being trialled. Exciting trials are also underway to withdraw the amyloid out of the body in all types of amyloidosis.

In the meantime supportive treatments remain extremely important in treating cardiac amyloidosis such as:

- limiting fluid intake and keeping fluid intake steady
- recording the patient's weight regularly
- diuretic drugs to help the body get rid of excess fluid and salt via the urine
- limiting salt and foods containing high salt content
- wearing support stockings
- using medications to lower blood pressure and deal with other symptoms. Many standard heart failure drugs are not appropriate in treating cardiac amyloidosis. Cardiologists along with the other amyloidosis specialists will advise the patient about what is appropriate for their disease.



# Living with uncertainty

Lynda Katona, Clinical Psychologist

**Living with uncertainty is one of the biggest challenges for people living with a disease like amyloidosis and their loved ones.**

Those things in life we thought we could confidently predict and look forward to become uncertain.

You may stop planning to do things because you don't want to disappoint others or yourself. As a result you risk becoming isolated and depressed.

## How can you learn to live with this uncertainty?

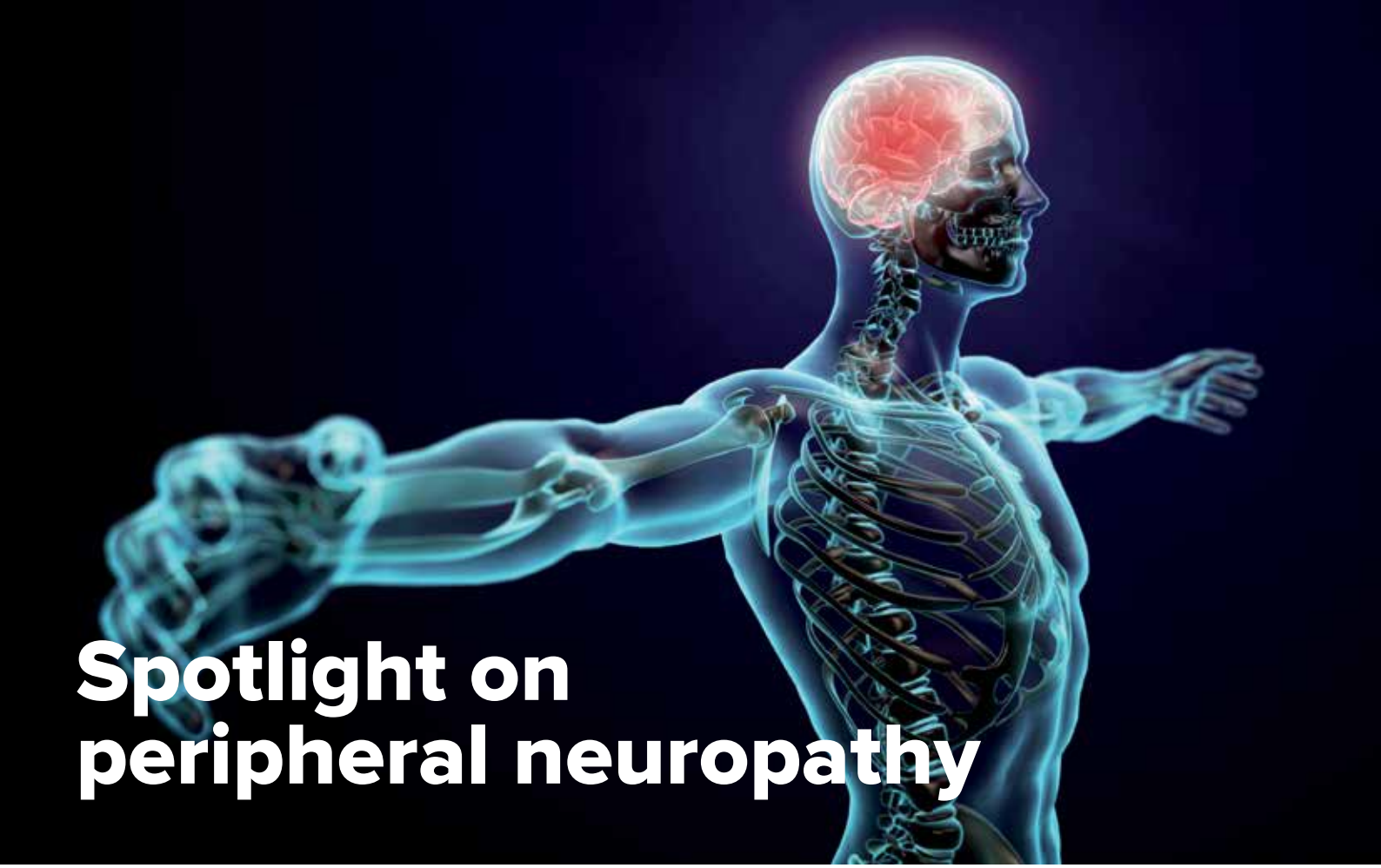
There isn't one easy answer to this question however there are some things that you can do that can help you to live well with uncertainty.

- Look after yourself physically. Aim for a balanced, healthy diet and engage in regular exercise within your capability. Speak to a physiotherapist or exercise physiologist if you feel unsure about what you can do.
- It is important to acknowledge the changes and losses that have resulted from your illness. While it can be helpful to have an overall positive perspective, it is important to acknowledge and experience the difficult feelings. If ignored, they tend to hang around leaving you stuck in sadness, anger, fear and grief.
- Family and friends often want to help so let them know what is most helpful to you. It might be practical support such as driving you to medical appointments or making a meal, or it might be listening to you when you are having a down day.
- Remind yourself of the things that you still have control over. While you need to accept the things you can't control, such as the course of your illness, it is important to remember you can choose things like what you wear and eat.
- Establishing a daily/weekly routine can help you to feel more in control and give you a sense of purpose. It is still important to plan to do things even if you have to change your plans at short notice.
- Be realistic about what you can do considering your energy levels. You may not be able to do all the things you used to do prior to diagnosis; don't beat yourself up about this.
- Engaging in daily relaxation or meditation exercises can help you feel relaxed and manage anxiety, pain and help you to sleep. Yoga and Tai Chi are also helpful forms of relaxation.
- If you have spiritual beliefs, connecting with them and a spiritual community can be a source of comfort and support.
- Keep your thinking balanced. Don't be unrealistically positive, but don't be unrealistically negative.
- Keep in touch with your sense of humour. Laughter can be good medicine.
- Don't be defined by your illness. It is part of your life and who you are, but it is not all of you. Keeping in touch with your pre-illness interests and activities can help remind you that you are still the same person just with some added challenges.
- Reconsider your priorities. Put your time and energy into people and activities that are really important to you.
- Focus on the here and now rather than getting caught up with past regrets or fears for the future. No one chooses to have cancer but there can be positive things that come from the experience.

If you need help dealing with your illness consider seeing a psychologist or counsellor. You can also contact the Leukaemia Foundation on 1800 620 420.

Remember life is never the same again after the diagnosis of amyloidosis, however it can still be rich and rewarding.

*Lynda Katona is a Clinical Psychologist with over 25 years experience. She worked for ten years in the oncology service at The Alfred, and has a particular interest in patients with blood cancers and those undergoing stem cell transplant.*



# Spotlight on peripheral neuropathy

**Amyloidosis treatment, or sometimes the disease itself, can cause peripheral neuropathy (PN). It causes damage to nerves of the peripheral nervous system, which transmits information from the brain and spinal cord to every other part of the body.**

It causes pain, numbness, tingling, swelling or muscle weakness in different parts of the body. It usually begins in the hands and feet and gets worse over time.

There are three main types of peripheral neuropathy that are not mutually exclusive and can often overlap.

**Sensory neuropathy** leads to loss of temperature, increased pain and altered pressure sensations. It can result in a general sense of numbness, especially in the hands and feet.

This can be the most disturbing and uncomfortable for people. It may be described as pins and needles, shooting pains, hypersensitive to touch, or pain that will not go away.

**Autonomic neuropathy** leads to changes in sweat glands, moisture and texture in the skin. There can also be an inability to control muscles that expand or contract blood vessels to maintain safe blood pressure levels. This can lead to a lowered blood pressure with symptoms of dizziness, light-headedness, or even fainting when a person moves suddenly from a seated to a standing position. If the nerves located in the gut are affected then this can lead to diarrhoea, constipation or incontinence.

**Motor neuropathy** can lead to a loss of motor function, muscle weakness, decreased foot stability, painful cramps, muscle wasting and altered foot structure.

## The peripheral nervous system

The peripheral nervous system consists of all the nerves outside the brain and spinal cord including the nerves in the face, arms, legs, chest, and some nerves in your skull. It acts like an information highway with messages being sent and received through the nervous system to and from the brain.

When the nerves within the peripheral nervous system become damaged the messages they carry can become distorted or interrupted. This is what occurs in peripheral neuropathy.

## What causes peripheral neuropathy?

There are numerous factors causing peripheral neuropathy:

- Underlying causes such as diabetes, older age, excessive consumption of alcohol, kidney disease and other pre-existing autoimmune and inflammatory diseases.
- Treatments such as thalidomide, Velcade (bortezomib) and vincristine (part of the VAD chemotherapy regime), can damage the nerve cells, particularly when given in high doses.
- Paraprotein can be deposited on nerve tissue and damage the nerve cells. High levels of paraprotein can also lead to thickening of the blood, called hyperviscosity. This may reduce the circulation of the blood and can also lead to the symptoms of peripheral neuropathy.
- A common viral infection called shingles.
- AL amyloidosis or a previous diagnosis of MGUS (monoclonal gammopathy of unknown significance).

- Myeloma can cause holes in bones which can lead to compression of nerves.
- Amyloid deposits.

## What are the symptoms?

Symptoms will vary from person to person. The hands and feet are the most commonly affected areas.

- Pain, which can vary in intensity and is often described as sharp, burning or stabbing.
- Pins and needles may start in your toes or the balls of your feet and travel up your legs. This sensation may also start in your fingers and work its way up your hands and arms.
- Unusual sensations or an increased sensitivity to touch, often worse at night. Some patients say they cannot stand the feeling of the bed sheets on their feet.
- Altered sensation, such as a feeling of pain or heat when touching something cold.
- Numbness in your hands or feet.
- Muscle cramps, weakness and tremor, interfering with your ability to perform everyday tasks.
- Lack of co-ordination or sense of proportion. It may seem that your body is not doing what you want it to do. You may also find your sense of where things are in your surroundings can become distorted.
- Loss of dexterity which interferes with simple tasks that require intricate movements of the fingers and hands, such as doing up buttons.

## Treatment

Medication may help reduce pain in the feet, legs, and arms although it doesn't bring back full sensation.

Some over-the-counter painkillers may help, too. Your doctor may prescribe medication that is used to treat other medical problems, such as epilepsy or depression, which can also have an effect on neuropathic pain. The drugs to prevent seizures include gabapentin, pregabalin, phenytoin, and carbamazepine. The antidepressant drugs may include amitriptyline or duloxetine. Injections of local anaesthetic such as lignocaine or topical patches containing lignocaine may relieve more intractable pain.

Your doctor may also refer you to a pain specialist or neurologist. Active and passive forms of exercise can reduce cramps, improve muscle strength, and prevent muscle wasting in affected limbs. Massage may improve circulation which can reduce pain.

## Supplements

Supplements that may be helpful in managing the symptoms of neuropathy include:

- vitamin B complex that includes B1, B6, B12 and folic acid.
- magnesium.
- amino acids including Acetyl-L-carnitine and Alpha-lipoic acid.
- fish oils, omega-3 fatty acids (EPA and DHA), flaxseed oil.

We advise that you discuss taking any supplements with your doctor to ensure they are safe and don't interact with other medicines.

## Self-management tips

- Take care of your hands and feet. Wear well-fitting protective shoes and keep hands and feet warm.
- Be careful when getting into the bath or shower and check the temperature of the water first. Wash up with rubber gloves.
- Do regular, gentle exercise to keep your muscles toned and to improve circulation.
- Stop smoking - ask your doctor for advice.
- Low resistant bed clothing such as silk. sheets can be more comfortable.
- Eat a well-balanced diet that includes essential vitamins and minerals.
- Avoid falls by reducing the risks in your own home. Make sure hallways and stairs are well lit and free of clutter and have hand rails fitted in your home.
- Ask for a referral to an occupational therapist to look at possible assistive devices and assess any risks around your home.
- Stay focused on the task at hand such as chopping vegetables and don't get distracted.
- Adopt good posture and avoid sitting with legs crossed for long periods of time.
- Transcutaneous electrical nerve stimulation (TENS) machines can sometimes help reduce your level of pain by delivering tiny electrical impulses to specific nerve pathways.
- Complementary therapies such as acupuncture, reflexology and gentle massage may all help to relieve some of your symptoms.
- Relaxation techniques such as meditation, visualisation, relaxation or a combination of these can be helpful in reducing muscle tension which may be contributing to your pain.

Go to our YouTube channel at **youtube.com/leukaemiaqld** and watch Cancer Care Coordinator Carmel talk about peripheral neuropathy.

# Wear an amyloidosis ribbon

An amyloidosis ribbon has been developed by the Leukaemia Foundation to help promote awareness of this rare disease. We urge people with amyloidosis to wear one as a way of connecting with and showing support for other people affected by this disease. They are available for a gold coin donation at our education and support sessions and by calling the Leukaemia Foundation on **1800 620 420.**

# The importance of accurate diagnosis

**Bruce Townsend tells the story of his diagnosis with wild type ATTR amyloidosis and how difficult it can be to get an accurate diagnosis.**

*Bruce and Glenda Townsend (right) at a recent Leukaemia Foundation amyloidosis support group in Brisbane.*

Looking back, my 65th birthday in 2009 may have been the beginning of my journey with amyloidosis. Not that I had any idea what amyloidosis was at that time.

I had been part of a veterans' exercise group which exercised twice a week for one to two hours. The age of the group ranged from 62 to 66 and I was noticing more and more that, compared to my peers, I seemed to take twice as long to recover.

My GP suggested a stress echocardiogram, which I underwent a couple of weeks later. Although it revealed a slight thickening of the left ventricle wall (13mm), I was not made aware of this as my cardiologist considered it not to be a problem.

Several years later in early 2013, I underwent a bone scan to check for cobalt poisoning that may have resulted from a faulty knee replacement. However, the scan didn't reveal cobalt poisoning but rather an abnormality in my heart.

My cardiologist ordered another echocardiogram, a bone marrow biopsy and a rectal biopsy. While the bone marrow biopsy was clear, the echo and rectal biopsy checks showed signs of amyloidosis and heart thickening that had increased to 17mm. A heart MRI followed which again revealed cardiac amyloidosis and thickening up to possibly 20mm.

More tests confirmed the presence of amyloidosis in my heart although finding what type of amyloidosis I had was proving difficult. At that time my doctors felt I had AL Amyloidosis and I commenced a six-month course of chemotherapy that consisted of a combination of the drugs cyclophosphamide, prednisolone and thalidomide. Half way through this course of chemotherapy, it was suggested I may need a stem cell transplant and was briefed on what was required and what to expect for this procedure.

Prior to the stem cell transplant I had ten heart biopsy samples taken in an attempt to determine a definite typing of my amyloidosis. Again this was not possible so the stem cell transplant was cancelled.

Further investigation indicated that I possibly had wild type ATTR, previously known as senile amyloidosis. I completed my chemotherapy treatment in October 2013. During the next 12 months, I had four more echocardiograms plus a bone marrow biopsy to check on my heart condition and any signs of multiple myeloma.

In November 2014, I arranged for a second opinion and transferred across to the Amyloidosis Clinic at The Princess Alexandra Hospital (PAH) in Brisbane. They sent some of my heart biopsies to London, and the diagnosis of wild type ATTR wild type was confirmed.

Due to possible future heart complications related to this form of amyloidosis I had a pacemaker fitted in August 2015.

During the last two-and-a-half years since the initial confirmation of my cardiac amyloidosis, I have undergone 11 echocardiograms, three bone marrow biopsies, two colonoscopies/ gastroscopies, a rectal biopsy, six months of chemotherapy, at least ten ECGs and had an MRI pacemaker implanted. I am also on a trial drug called diflunisal that hopefully may slow the deposition of the amyloid into my heart.

I am still under regular supervision from a cardiologist and a haematologist but lead a reasonably normal and active life with the help of a supportive and understanding wife.

So what have I learned through my experience with wild type ATTR amyloidosis.

The exact typing of your amyloidosis can be very difficult, very challenging and may take some time to diagnose, but early diagnosis is paramount for your future health. Do not be afraid to seek a second opinion.

The medical staff and support workers at the PAH Amyloidosis Centre offer a wealth of knowledge, assistance and personal help.

The Leukaemia Foundation offers an excellent support system for amyloidosis patients and their families or carers. They hold seminars and support lunches throughout the year, which my wife, Glenda, and I attend. Getting involved with a support group is a great way to help you through those 'off' days.

# Travelling while on kidney dialysis

**Q.**

**My husband is on dialysis because of the damage his kidneys have sustained from the deposition of the amyloid protein. Otherwise he is well and we would love to go on holiday in Australia. Is it possible to get dialysis in other centres around Australia? Where are these centres and how do we arrange this?**

A list of the dialysis units in Australia is listed on the Kidney Health Australia website at kidney.org.au/your-kidneys/support/dialysis/dialysis-and-travel and can be searched by state.

You need to contact the unit close to where you are travelling and enquire about whether they have vacancies. You also need to check whether they take private and public patients because each state has different arrangements between its health department and the private and public providers of dialysis services.

**You can also ring 1800 454 3639 or email info@kidney.org.au.**

# What do misfolding proteins in amyloidosis mean?

**Throughout our lifetime, our DNA is coding for the manufacture of small molecules called proteins. These proteins provide the structure and function for nearly all of life's biological processes.**

Enzymes that facilitate our cells' chemistry, hormones that affect our body's growth and regulation, and antibodies that form our immune response are all examples of proteins in action. Just about everything in our bodies – from the colour of our eyes, to carrying oxygen in our blood, to whether we can digest milk – is determined by the proteins we make.

Once produced within the body, proteins will naturally fold into a particular shape. This natural form of a protein molecule is what allows for its specific function. Put simply, when proteins are folded properly, they work as they should, and we enjoy relatively good health. When proteins are misfolded, it affects our body's ability to function, and problems may arise over time.

Misfolded proteins can be produced because of genetic causes, or because of other factors related to chronic inflammation or increasing age. Regardless, our bodies are usually capable of identifying and removing these abnormal proteins. In some cases though, we either produce too much of the abnormal proteins for our body to handle, or we are not able to break down and clean up the proteins at all. Such defects in protein production and processing are associated with many diseases.

Broadly speaking, amyloidosis is one class in a growing list of protein folding disorders. While there are many distinct types of amyloidosis, in all cases the misfolded proteins, called amyloid (meaning 'starch-like'), take on a particular shape that makes it difficult for the body to break down. Because of this misfolding, the amyloid proteins bind together to form rigid, linear fibres (or fibrils) that accumulate in our body's organs and tissues.

Depending on where the amyloid builds up, such as in the kidney, heart and nerves, different symptoms and potentially life-threatening conditions become manifest.

While amyloidosis has been known since the 19th century, it is only within the last few decades that our understanding of it has matured. Presently, there are over 25 different proteins that have been identified as contributing to amyloidosis. Additional types of precursor proteins that can lead to amyloid formation continue to be discovered through ongoing research.

# Proteomics assay being tested in Australia

by Dorothy Loo, The Proteomics Core Facility Manager, UQ Diamantina Institute

**The Princess Alexandra Amyloidosis Centre (PAAC) in Brisbane is currently testing a novel proteomics assay which has been used at the Mayo Clinic in the United States and which can diagnose and classify amyloidosis with great accuracy.**

The importance of early and accurate diagnosis and typing is critical. The treatments depend on the type of amyloidosis and the organs affected, as well as on the patient's condition, age, and personal preference. If not treated in a timely manner, amyloid deposits will continue to damage tissues and may result in organ failure and death.

## Why is amyloidosis so difficult to classify?

Amyloidosis is a group of diseases which present with tissue and organ damage caused by abnormal but non-cancerous protein deposits. Treatment of amyloidosis requires the reduction of these protein deposits in the body, however this is not an easy process as there are over 20 different amyloidosis causing proteins.

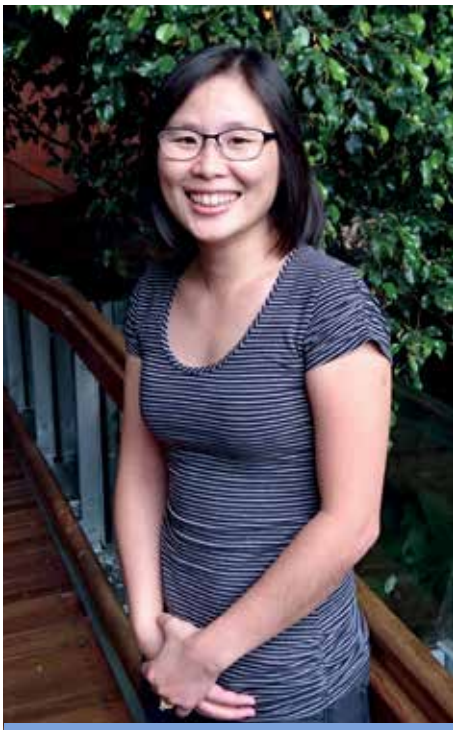
Each protein is involved with a different type of amyloidosis and requires a different treatment. Treating patients for the wrong type of amyloidosis can be detrimental to the health of an already ill person. Therefore, it is essential that treatment only be implemented after the correct amyloidosis type has been determined.

Misdiagnosis can occur because amyloidosis is a very rare disorder and the different types of amyloidosis can present with a similar clinical picture. In addition, diagnosis with traditional laboratory tests is difficult as reagents normally used to identify different proteins are ineffective against abnormally folded proteins in amyloid plaques.

## Proteomics assay

The new proteomics assay being tested at the PAAC requires a multidisciplinary collaboration between clinicians and pathologists from the hospital and proteomics scientists from the Translational Research Institute. The various specialist procedures include: a patient biopsy of the affected tissue and organ; formalin fixation to preserve patient biopsies; Congo red staining of amyloid proteins in biopsies; excision of the Congo red stained amyloid plaque with laser capture microdissection; and mass spectrometry proteomic identification of amyloid plaque proteins.

The clinician at PAAC, Dr Peter Mollee, interprets the proteomics results



*Dorothy Loo*

with the patient's clinical history to diagnose the amyloidosis subtype. They have tested over 136 samples since 2010 with most patient biopsies from the heart, kidney and small bowel, hence the biopsies are usually very small. Only 129 biopsies had sufficient tissues for the proteomics assay. The current proteomics assay has had a 94 per cent success rate in the 129 cases tested. The PAAC is the only centre in Australia to provide proteomics results for amyloidosis patients.

## ORAL DOXYCYCLINE SHOWS PROMISE IN TREATING AL CARDIAC AMYLOIDOSIS

**A paper was presented at the 2015 American Society of Hematology (ASH) Annual Meeting and Exposition on the improved outcomes for patients with cardiac AL amyloidosis being treated with oral doxycycline.**

The paper reported that previous findings have now been confirmed by a larger cohort which suggested that oral doxycycline improved the survival in cardiac AL amyloidosis based on the possible cardio-protective effect of doxycycline on the heart from toxic amyloidogenic light chains.

Thirty patients with cardiac AL amyloidosis who were treated at the Royal Free Amyloidosis Treatment Centre, London, received oral doxycycline as adjuvant to standard chemotherapy. Doxycyline was given orally 100mg twice daily until completion of treatment and continued as long as tolerated without unacceptable toxicity or patient preference.

The median age was 65 years (range 47-87 years), all patients had cardiac involvement with six having Mayo stage II (2 doxycycline and 4 controls), and the remainder with Mayo stage III disease including 28 with Mayo stage IIIb disease (high risk).

The study showed treatment with doxycycline in combination with chemotherapy significantly improve the overall survival in patients with advanced cardiac stage IIIa (low risk) AL amyloidosis but not in those with very advanced stage IIIb (high risk) disease. This larger study confirms the previous preliminary results of using adjuvant doxycycline in AL amyloidosis and strongly supports the rationale to proceed with a randomised trial.

# Avoiding germs during chemotherapy

**When you have chemotherapy, catching a cold or the flu can wreak havoc with your body. But, it's not always easy to avoid germs when you are at work or in public. And it doesn't help that lots of people go out in public or to work even when they're ill. Here are some tips that help the people you come in contact with to keep their germs to themselves.**

**Avoid the communal lolly jars and salad bars.** You have no idea whether they were prepared in a sanitary way or whether other people touching the food have washed their hands.

**Keep your desk area at work clean.** Step up the disinfecting. Studies show that a desk may boast 400 times more bacteria than the average toilet seat - which means you may be sharing your workspace with a lot of germs. Most cleaning crews don't wash desks, so be sure to use disinfectant wipes daily to clean the areas in your workspace that you touch often - phone, keyboard, desk, cabinet handles and doorknob.

**Use your own dishes.** Sure you know that using your own utensils, glasses and dishware is a smart move, but did you know that making sure they're dried thoroughly is key? That's because moisture breeds germs. And dishes that are put away wet can become contaminated with bacteria.

**Stay away from the sick.** Email them or call them rather than visiting them. If you must meet with someone who is unwell, sit as far away as you can and, whatever you do, don't touch anything they touch.

**Wash your hands.** Everyone knows "Happy Birthday," and you should sing it to yourself twice while you wash your hands as that's how long it takes for your hands to get truly clean. And don't forget to intertwine your fingers and wash beneath your nails.

**Be careful what you touch.** When you touch things that many other people touch, such as taps and door handles, use hand sanitizer or wash your hands right after touching.

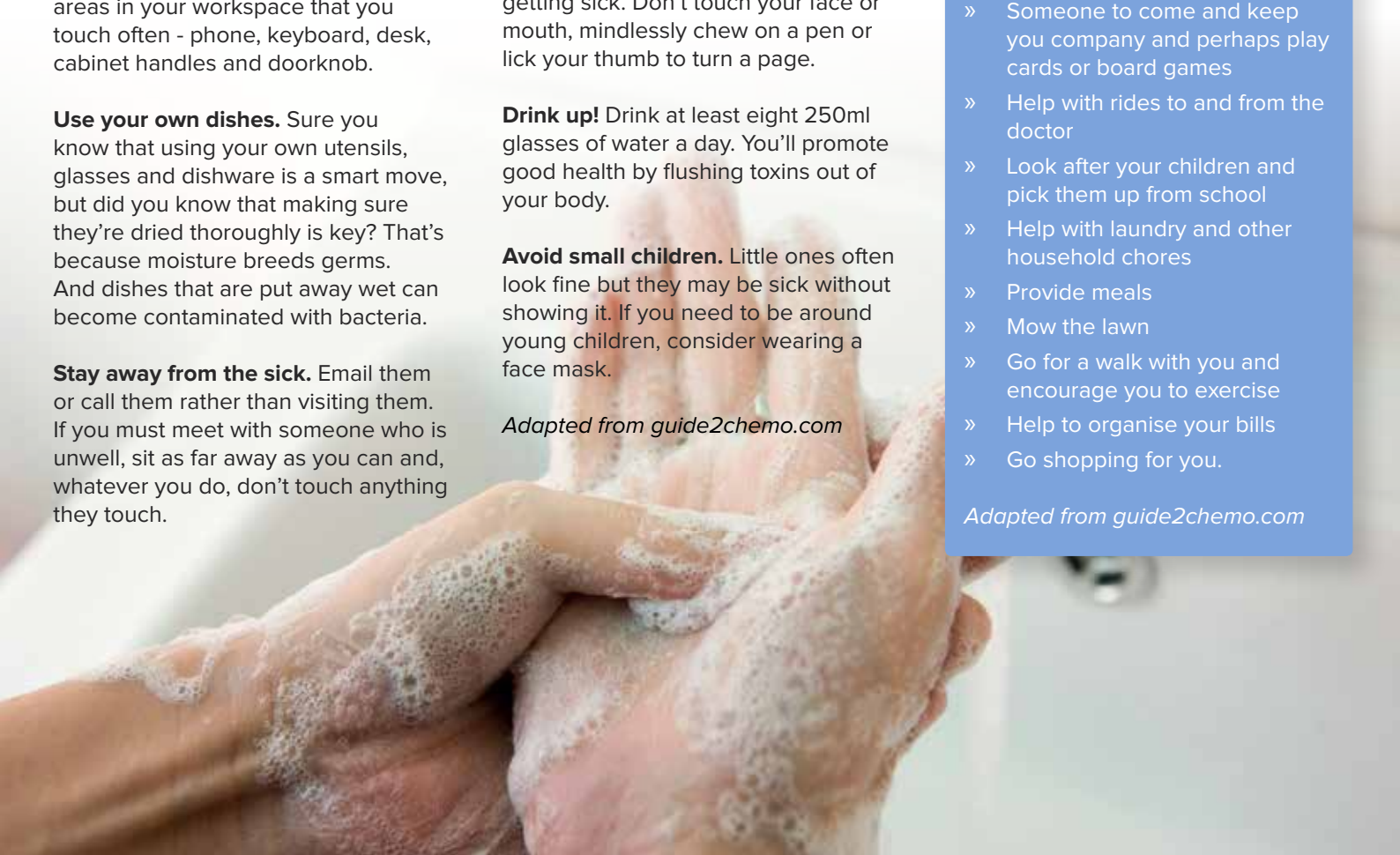
**Take some time off.** Staying home from work or avoiding crowded places like shopping centres reduces your exposure to foreign germs. Consider using your holidays or sick days when it seems like something is going around at work (such as in the winter, when people have colds or the flu). If you can't take time off, see whether you can work from home to avoid these germs.

**Watch where you put your hands.** Keep your hands away from your face to reduce your risk of getting sick. Don't touch your face or mouth, mindlessly chew on a pen or lick your thumb to turn a page.

**Drink up!** Drink at least eight 250ml glasses of water a day. You'll promote good health by flushing toxins out of your body.

**Avoid small children.** Little ones often look fine but they may be sick without showing it. If you need to be around young children, consider wearing a face mask.

*Adapted from guide2chemo.com*



## Accept help with open arms

"What can I do to help?" When you begin chemotherapy or other treatment for amyloidosis, friends, family and even casual acquaintances will ask you this question. If you're the kind of person who hates to ask for help, remember that friendship is a matter of give and take. Let your friends give back to you — not just for your sake, but for theirs, too!

As you begin treatment, it's perfectly OK to tell people you have no idea what you'll need. Tell them when your treatments will begin, and ask them to check back with you then.

**Be aware there are many different ways your friends and family can help you such as:**

- Help with your elderly parents
- A friendly ear just to listen to you
- Someone to come and keep you company and perhaps play cards or board games
- Help with rides to and from the doctor
- Look after your children and pick them up from school
- Help with laundry and other household chores
- Provide meals
- Mow the lawn
- Go for a walk with you and encourage you to exercise
- Help to organise your bills
- Go shopping for you.

*Adapted from guide2chemo.com*

# What's on around Australia

## NEW SOUTH WALES

Fifteen patients and their families attended an amyloidosis education and support event on 1 April hosted by the Leukaemia Foundation at the Wenty Leagues Club in Sydney.

Guest speaker at the event was Linda Mekhael, Clinical Research manager at the Amyloidosis Clinic at Westmead Hospital.

People attended, some from as far away as Tamworth and country Victoria, were able to discuss topics including the role of the clinic in coordinating biopsy samples between various medical specialities, trials of new treatments being conducted and the length of time taken for these to be completed and the possible creation of a central register of patients, privacy laws permitting, to facilitate further diagnosis and treatment.

### Community forum

To raise awareness of amyloidosis in the community, the Westmead Amyloidosis Clinic will hold a Community Forum on Friday 6 May.

The forum will take place at the Lowewenthal Auditorium, Education Block, Level 2, Westmead Hospital from 6.30-9pm and will coincide with the inaugural National Amyloidosis Day on 8 May.

The forum is a night of dialogue rather than lectures and an opportunity to discuss major advances in research and the treatment of amyloidosis. There will be an opportunity for questions to be asked of experts in each of the medical specialties involved in the diagnosis and care of the disease.

If you would like to attend please email Linda Mekhael at linda.mekhael@health.new.gov.au Feel free to attach a question that you would like answered on the night.

## SOUTH AUSTRALIA

Amyloidosis / myelodysplastic syndrome (MDS) / myeloproliferative neoplasms (MPN) and Waldenstrom's macroglobulinaemia (WM) support groups will be held on 8 June, 10 August, and 12 October, 10am-12pm, at the Bridgestone Australia Leukaemia Foundation Village, 39 Folland Avenue, Northfield, South Australia. For more information and to **RSVP please phone 08 8169 6036 or 1800 620 420 or email kharvey@leukaemia.org.au**

## QUEENSLAND

Queensland amyloidosis patients and their families and carers can meet together at regular support lunches and share stories as well as hear about the latest treatments and research from experts. Dates for amyloidosis lunches are Wednesday 22 June, 7 September and 30 November. **Please call 07 3055 8233 to RSVP or find out more information.**

## TASMANIA

Amyloidosis patients and carers are welcome to attend the Hobart blood cancer support group on 27 July, 11am-1pm at the Leukaemia Foundation office, 153 Collins St, Hobart. **RSVP or find out more information by phoning Jane Anderson on 03 6231 0620 or janderson@leukaemia.org.au.**

## Support Services team

**Queensland Amyloidosis Coordinator**
Sheila Deuchars

**Victoria/Tasmania Blood Cancer Support Manager**
Tennille Lewin

**New South Wales/ACT Blood Cancer Support Manager**
Snezana Djordjevic

**South Australia/Northern Territory Blood Cancer Support Manager**
Peter Diamond

**Western Australia Blood Cancer Support Manager**
Tanya Harris

Leukaemia Foundation
VISION TO CURE
MISSION TO CARE

## Contact us

**If you need support or would like to make an enquiry please contact the Leukaemia Foundation on 1800 620 420 or go to leukaemiaqld.org.au (Queensland patients) or leukaemia.org.au.**