

COVER STORY

Peta Rasdien

Be alert, not alarmed by chance of rare illness



Tamara and Jason Robins with Harrison, left, and Noah.

Mitochondrial disease

Tamara and Jason Robins had never heard of mitochondrial disease before one of their twin boys, Noah, was diagnosed when he was 2½.

Certainly, alarm bells had been ringing. While his brother Harrison was achieving milestones at all the right times and learning to walk at nine months, Noah was what they called a "colicky baby", hard to settle, and appeared to be quite floppy, finding it difficult even to sit up.

After a course of physiotherapy and a range of blood tests and scans, doctors delivered the worst news.

Noah had Leigh's disease — the clinical presentation of a mitochondrial mutation.

"No cure, no treatment, and really not a lot of answers for the thousands of questions we had," Mrs Robins said.

Mitochondrial disease is the result of a genetic mutation in either the mitochondrial DNA or the nuclear DNA and can affect children and adults.

This genetic fault means mitochondria — often called the "cells' powerhouses" and responsible for producing 90 per cent of the body's energy — are unable properly to convert and deliver energy around the body, which can affect the central nervous system and cause progressive degeneration of motor functions.

Recognition of the disease is still relatively new. In 1988, scientists discovered that mutations in

mitochondrial DNA caused disease and it was only in 1992 that a nuclear gene mutation was found to cause mitochondrial disease.

At first thought to be a rare disease, recent research suggests the condition may affect one in 200 people, according to the Australian Mitochondrial Disease Foundation. A range of diseases are caused by problems in mitochondrial energy production, and symptoms can be varied and include muscle fatigue, hearing loss, migraine, strokes, diabetes, heart disease, gastrointestinal symptoms and eye disease. Because there are many more common diseases that cause these symptoms, a diagnosis can be difficult and sometimes delayed.

The family had sought advice from a naturopath and believed a tonic of vitamins and supplements had helped Noah. "We are so proud of every little baby step that he has made. Although, sadly, this is a degenerative disease, Noah has made little steps forwards which we pray he will continue to do," Mrs Robins said.

Noah's twin brother, Harrison, has high-functioning autism and Mrs Robins said doctors believed this may be due to the fact that he too has a mitochondrial mutation, although at a lesser loading than that of Noah.

Some diseases are out of the ordinary but their diagnosis depends on vigilance from both patient and doctor to optimise treatment and lead to a successful outcome

It may be a disease you've never heard of or something that's been talked about but you're not sure what it is, but lesser-known or unusual diseases deserve the spotlight too, according to Royal Australian College of General Practitioners spokeswoman Helen Wilcox.

And the people with these conditions warranted acknowledgment and public awareness, Dr Wilcox said.

"They are living with a little-known illness and managing on their own, without some of the system supports that are offered to the more high-profile illnesses," she said.

While it didn't happen often, diagnosing an illness that was out of the ordinary could be rewarding.

"Often when you do diagnose these things, even though they are rare, you feel very fortunate because delayed diagnosis can influence how sick the patients get and how much better they can get," she said.

It's believed that about 50 per cent of the time GPs deal with about 30 common diagnoses, but about 800 diagnoses for the remaining 50 per cent.

Dr Wilcox said many GPs worked on the "three strikes rule" in order to prevent patients with little-known diseases slipping through the diagnosis net. "If you are seeing a patient again for the third visit and things aren't better and you haven't got a clear diagnosis, it is a big trigger to spread the net, look at the literature, discuss it with your GP colleagues or refer off for a specialist opinion," she said.

It was also a good idea for patients to discuss with their doctors what other things they might be considering as a possible cause of symptoms and how long they should give treatment before opting for a "plan B".

"I think we've all got a few cases on our case load where the patient's vigilance has literally saved their life," she said.

She'd had a young patient who displayed all

the classic symptoms of gastro-oesophageal reflux disease.

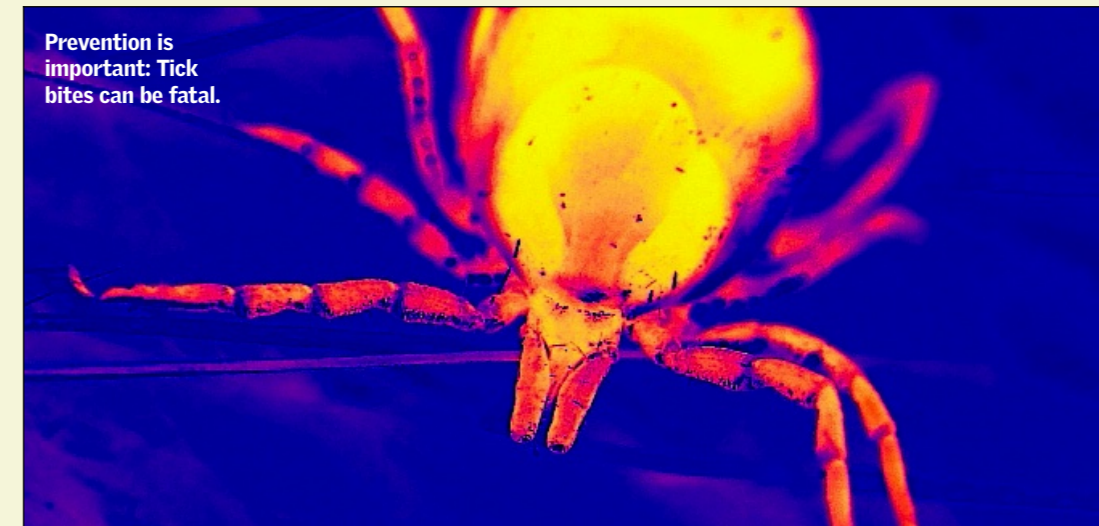
Dr Wilcox did the usual tests, prescribed medication to reduce the reflux and advised the patient that she should get back in touch in four weeks' time if her symptoms persisted.

"At the four-week mark she was vigilant, she rang me and said she wasn't better, we . . . did an endoscopy and found a very early stage oesophageal cancer and she was able to have curative surgery," Dr Wilcox said.

"If we'd left it another couple of months, it would have been inoperable, fatal. So she really saved her own life by sticking to the plan."

Dr Wilcox said it wasn't necessary to worry about developing or contracting a rare illness but it was worth being vigilant.

A good start was to know your family medical history — not just for illnesses like diabetes or heart disease, but for illnesses that might not cause daily symptoms such as haemochromatosis or deep-vein thrombosis.



Prevention is important: Tick bites can be fatal.

Lyme disease

Lyme disease is a rare tick-borne disease that can be fatal to humans caused by a bacterium called *Borrelia burgdorferi*.

This bacterium is transferred to humans through a tick bite.

In its initial phase, patients may experience an expanding reddish rash and flu-like symptoms. As the infection progresses, it can produce abnormalities in the joints, heart, and nervous system.

Lyme disease was first identified in 1982 following investigations into a group of children who lived near

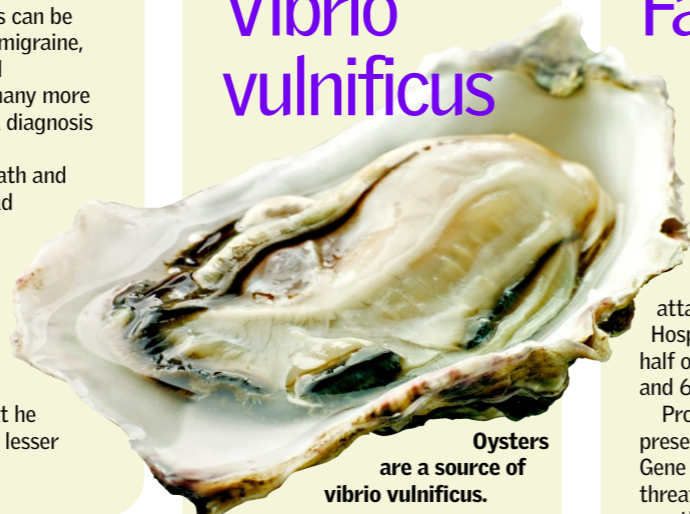
each other in Lyme, Connecticut, in the US, who had all been initially diagnosed with rheumatoid arthritis. Most cases of Lyme disease can be cured with antibiotics.

Prevention is also important. Experts recommend that people try to avoid tick bites by spraying insect repellent on to exposed skin, wearing long sleeves and long pants tucked into boots.

The disease is common in the US, however, debate is continuing about whether it exists in Australia.

While patients with illnesses that are clinically consistent with Lyme disease have been identified in Australia, there has been no confirmed evidence of the *Borrelia burgdorferi* bacterium in Australian ticks.

Vibrio vulnificus



Oysters are a source of vibrio vulnificus.

Vibrio vulnificus is a bacterium found in marine environments such as estuaries, brackish ponds or coastal areas and is in the same family as the bacterium that causes cholera.

Patients become infected after eating seafood (especially raw or undercooked oysters), swimming or wading in contaminated seawater with an open wound or via puncture wounds from the spines of fish such as tilapia.

A number of cases have been reported in Australia. First documented in 1979, *vibrio vulnificus* causes symptoms including vomiting, diarrhoea, abdominal pain and a blistering dermatitis that can require surgery to remove affected parts of the skin and underlying tissues, or even amputation.

While among healthy people it can be treated with antibiotics and does not have any long-term consequences, those people who are not diagnosed quickly or who are immune-compromised can experience septic shock and death.

Royal Australian College of General Practitioners spokeswoman Helen Wilcox said people with haemochromatosis were also more susceptible to infection with *vibrio vulnificus*.

Familial hypercholesterolemia

An estimated 4000 West Australians could be ticking time bombs, and up to 80 times more likely than the rest of the population to have a heart attack or develop premature heart disease because of a genetic disorder characterised by markedly elevated cholesterol levels.

Known as Familial hypercholesterolemia (FH), the disorder is considered a silent condition because many sufferers are not aware they have it until they develop angina or have a heart attack. Gerald Watts, head of the lipid disorders clinic at Royal Perth Hospital, said FH was a dangerous condition when left untreated, with half of its sufferers having heart attacks before the age of 50 for men and 60 for women.

Professor Watts said the gene mutation for FH only needed to be present in one parent for the disorder to be passed on to their children. Gene mutations in both parents made the disorder even more life threatening but this was very rare. Recent surveys had shown about one-third of people with FH also displayed other risk factors which increased their heart attack risk, including obesity, smoking, high blood pressure and diabetes, Professor Watts said.

About one in 20 people with premature heart disease was likely to also have the FH gene.

The best way to treat FH was to pick the condition up early with DNA testing in children and teenagers, usually after puberty in boys and after a girl's first period. Treatments for the condition included a healthy lifestyle and dietary intake and the use of statin drugs. "It's absolutely fundamental that young people with FH do not smoke nor become obese," he said. In severe cases, where FH patients carried the gene (homozygous FH) from both parents, those patients required intensive life-long treatment. Statin drugs were combined with regular apheresis, a process similar to dialysis, to remove cholesterol-containing particles from the bloodstream.

FH has been a part of Simone Poor's life since she was only four years old, diagnosed with the condition when she started developing cholesterol deposits on her hands, elbows, knees and ankles.

Ms Poor is one of only six people in WA to carry the FH gene inherited from both parents.

Connie Clarke

For more information about FH, contact the Genetic Support Council WA on 9485 8999 or visit www.familialhypercholesterolaemiasupportwa.websyte.com.au

Haemochromatosis

We hear a lot about the dangers of low iron but for about one in 300 Australians it is iron overload that is cause for concern.

These people are living with a genetic condition known as haemochromatosis.

While iron is essential for the production of oxygen-carrying haemoglobin, people with haemochromatosis absorb more iron from their food than is necessary. Royal Australian College of General Practitioners spokeswoman Helen Wilcox said if untreated, serious conditions could be caused by the excess iron in haemochromatosis including cirrhosis, liver cancer, diabetes, arthritis, heart disease and psychological problems.

Because iron builds up slowly, obvious symptoms may not appear until the age of 30-40 for men and later for women.

These symptoms can often be mistaken for other conditions and include chronic fatigue, joint pain

and bronze colouring of the skin.

Diagnosis can be made via a simple blood test to identify the mutated haemochromatosis gene HFE.

Treatment includes regularly removing blood in a procedure called venesection, where up to 500ml of blood is removed at a time until iron levels are normalised. Lifelong maintenance venesection is required, usually every three to six months.

Fortunately for Bayswater man John Lee, 59, his haemochromatosis has not led to any long-term effects, despite a delay in diagnosis.

Five years after he lost his employment in an office for falling asleep on the job, he was finally diagnosed in 1999.

Initial suspicions of chronic fatigue syndrome were ruled out when his GP ordered a full blood iron study.



John Lee: No long-term health effects from haemochromatosis.

Gastro-oesophageal reflux

The name gastro-oesophageal reflux disease, or GORD as it is also called, might not be well known but its symptoms certainly are.

Regular and frequent heartburn and/or acid regurgitation are common symptoms of GORD, a condition which, if not treated, can greatly impair quality of life and have long-term complications.

GORD is usually caused by temporary or permanent changes in the barrier between the stomach and the oesophagus; this can include abnormal relaxation of the lower oesophageal sphincter, which usually holds the top of the stomach closed. The acidic stomach contents are then able to "wash" back up into the oesophagus, where

they can damage the oesophageal lining, causing inflammation, known as oesophagitis, or in more severe cases ulceration. It can also cause a chronic cough and less commonly nausea, excessive belching or chest pain.

Treatment usually involves eliminating anything that makes symptoms worse. This can be alcohol, smoking, coffee, spicy or fatty foods, eating large meals or some medications.

Royal Australian College of General Practitioners spokeswoman Helen Wilcox said trigger foods were not always obvious. "Some people will get symptoms of GORD after seemingly innocuous foods like lettuce or capsicum," she said.

Munchausen syndrome

Those with the rare psychological condition Munchausen syndrome, also known as a factitious disorder, deliberately fake medical symptoms or cause themselves harm in order to gain attention.

The condition was first named in 1951 after Baron von Munchausen, famed for making up stories about imaginary exploits.

Clinical psychologist Zyron Krupenia said people with the syndrome were motivated by a desire for sympathy or attention, or to feel a sense of power gained by fooling their doctors. He said he had treated two patients with the related condition, Munchausen's by proxy, which was

where a caregiver faked symptoms, sometimes by causing injury to someone else, often a child.

"When you do have a genuine illness you get a lot of attention and so it is not a big leap for those people who are needy to figure out that they can get attention in that way because it pays off, they do get what they need, and it becomes self-perpetuating," he said.

The internet had made it even easier for people with the syndrome to look up symptoms in order to fake them.

While the condition was rare, Mr Krupenia said it was even rarer for a person with Munchausen to present to a psychologist.