'Our understanding of brain function in both health and disease is growing at a very fast rate.'

Professor PERRY BARTLETT, director of the Queensland Brain Institute at the University of Queensland

Health & Science

The heirloom no one wants

Charcot-Marie-Tooth disorder, the most common inherited neurological condition, is poorly understood, writes **Eleanor Learmonth**.

'My feet hurt," is one of the most frequently moaned sentences in the English language. It is such an everyday problem, particularly for women, that few of us stop to consider it may be a symptom of a serious genetic condition: Charcot-Marie-Tooth disorder. CMT, named after the scientists who discovered it, is the world's most commonly inherited neurological condition.

Yet in many respects, it is a confounding and confusing illness. It is poorly understood, even by doctors, and the symptoms span a spectrum of everyday problems.

Take a quick quiz …

1. Did you hate sport at school?
2. Do you have trouble opening jars?
3. Do you feel the cold badly?
4. Do your feet often hurt and do you have trouble finding comfortable shoes?
5. In all honesty, do you think your feet look a bit weird?

If you were nodding your head to any of those questions, or if they apply to your children, you may want to note that 60 per cent of Australians who have it are undiagnosed and have probably never heard of it.

So what is CMT? Simply put, it is a degenerative and progressive disorder of the nervous system that primarily affects the parts of the body furthest from the brain: the hands and feet. While the brain is unaffected, the feet and hands become weaker and insensitive over time.

It is a multi-gene disorder. Australian and international research has identified more than 50 different kinds of CMT, and the number is still rising. CMT Type 1A is the most common form and is not life threatening.

Other variants, for example CMT Type 3, are far more severe. Luckily, there are only a few kinds that are fatal. Other forms are confined to very specific groups, such as families where a cultural tradition of marriage between first and second cousins is common and long-standing.

Worldwide it is estimated that about 2.6 million people suffer from CMT, and it is found equally across race and gender.

The disorder also varies greatly between individuals. Patients usually have only some of the wide variety of symptoms and they can range from almost unnoticeable to severely disabling, even within the same family.

The most visible symptom is typically a highly-arched foot. This, in turn, can cause painful feet, trips and falls, as well as problems with running, walking, and extended periods of standing.

Sufferers may also be clumsy, have an odd way of walking, slow reflexes and bad balance. Simply put, we are not likely to be members of the Bolshoi ballet or on anyone's Olympic team.

I say "we" for the obvious reason. Diagnosed with CMT at 29, I became the first-known sufferer in my family – the gateway patient of my clan. CMT has subsequently been found in a grandparent, a parent, a sibling and both my children.

Like many other CMT patients, I knew something was wrong but had no idea that my various idiosyncrasies were all attributable to one source. Growing up, one just gets used to the weirdness.

My hands shook so I avoided becoming a waitress and learned quickly that authority figures tended to consider shaking the sign of a guilty conscience.

On the plus side, I got to ride in a Japanese police car and experience a lights-in-the-eyes police interrogation. On the down side, my luggage frequently gets a little more attention than is normal at customs.

Pain in my feet eventually drove me into the hands of doctors, but a definitive diagnosis took two years, and many wrong turns. By that time I was also having trouble with my hands.

Another common symptom is the gradual loss of sensation and weakness in the fingers, causing trouble with chopsticks, pegs, buttons or zippers as the finger muscles gradually atrophy.

As the peripheral nervous system deteriorates, the CMT sufferer starts to feel the cold badly in their hands, feet and legs.

Given the range of physical problems that CMT causes, why does it remain largely unrecognised and undiagnosed in Australia? Part of the problem lies in the nature of genetic conditions.

"You often just think you're normal because your mother or father was like that too," says Dr Joshua Burns, a CMT researcher undertaking a study into the disorder at the Children's Hospital at Westmead.

"You might think, say, having highly-arched feet or weak hands is just normal within your family."

He says people are therefore more inclined to put up with the problems rather than actively seek a diagnosis.

The condition slowly manifests itself during childhood or adolescence, even into the late 20s, making it harder to notice, and easier to ignore.

Often a child's classmates will detect something is wrong long before the family has noticed. In the playground, diagnosis often takes the shape of a nickname.

Mine was "Parko", a jocular allusion to Parkinson's disease. My school friends had acknowledged there was a problem 15 years before my doctor.

Many CMT patients report similar stories. In an online CMT forum, five different people, from Ireland to Iowa, reported being known as "chicken-legs" at school, all before they had been diagnosed.

The condition also moves between generations in a complex manner.

"It shows all forms of inheritance – dominant, recessive and X-linked," says Professor Garth Nicholson, a leading researcher into neurogenetic disorders from Sydney's Anzac Research Institute.

In the case of dominant inheritance, children have a 50 per cent chance of developing CMT. In recessive inheritance the possibility is cut to 25 per cent.

If the genetic defect is carried on the mother's sex chromosome (X-linked inheritance), the sons have a 50 per cent chance of developing CMT, while any daughters have a 50 per cent chance of becoming a carrier.

"It can also occur spontaneously, where there is no family history," Nicholson says. This new mutation can then be passed on to children.

While research on CMT has increased exponentially over the past decade, many doctors haven't kept pace.

"Most doctors still don't know much about CMT; a lot can't recognise it," Burns says. "Patients tend to get misdiagnosed with other things like post-polio, or told there is nothing wrong."

Alarmingly, the condition can be mistaken for drug or alcohol addiction.

Although being diagnosed with a genetic disorder can be a big, multi-generational drag, not knowing what's wrong may be worse. Correct treatment will minimise the harm and may prevent misdiagnosis and mistreatment.

"If left unmanaged, many CMT sufferers end up having joint replacements later in life that could have been avoided," Nicholson says.

Once diagnosed, treatment may include physiotherapy, orthotics and occasionally surgery.

The Charcot-Marie-Tooth Association is holding a seminar and awareness day at Concord Hospital on Sunday.
Phone 9767 5105 or visit its website – www.charcot-marie-tooth.org

Back on his feet … Grant Magnus, who has the rare genetic condition of Charcot-Marie-Tooth disease, which affects his feet and hands, with his daughters, Eliza and Scarlett. Photo: Fiona Morris

I've got my life back – diagnosis and surgery ease the pain

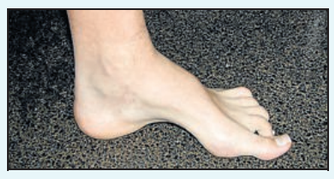
MISDIAGNOSIS plagued Grant Magnus, a 36-year-old defensive driving instructor from Holsworthy. "Aged five, I was initially diagnosed with autism and muscular dystrophy, because I could do long division but couldn't walk up and down steps."

Magnus had tendon transfer surgery in 2000, severing and reattaching more than a dozen tendons in his calves, feet and toes to alleviate pain, straighten his feet and toes and increase his mobility.

The operation was successful and Magnus celebrated by taking part in a cancer charity bike ride from Coffs Harbour to Sydney.

"The surgery changed my life," Magnus said. "I can't speak highly enough of it … without it I probably wouldn't be walking now.

"Before the surgery it was very painful to walk and my feet were all twisted. The biggest problem for me now is winter; my feet and recently my hands ache when it's cold."



Telling sign … a CMT-affected foot.

Magnus's daughters, Eliza, 9, and Scarlett, 7, have the same condition as their father, CMT, but thanks to an understanding, helpful school and support at home, they are thriving.

Self-esteem can be a big problem for CMT children, particularly if the parents and school don't understand what's wrong. Slow, illegible handwriting can be a big problem and teachers aren't always sympathetic when there is no diagnosis; a messy notebook can be attributed to laziness or a bad attitude, not DNA.

Magnus's children's school was made aware of the girls' condition so they would not have to endure being misdiagnosed like their father was.

"They can't run as fast as the other kids and they have some trouble writing, but at the moment, they are coping well," he said. "The school is talking about getting an expert in to talk to the kids about what CMT is."

CMT symptoms can increase with age, so the girls may eventually require surgery.

HANDED DOWN Some other common inherited neurological conditions

▸ **HUNTINGTON'S DISEASE** causes gradual deterioration of the central nervous system and death. There is no cure. It mainly affects adults, usually appearing between the ages of 30 and 50. In Australia, seven to 10 people in every 100,000 will be affected. Huntington's is caused by a defective gene that results in the destruction of brain cells. The gene can be passed from one generation to the next. If a parent has it, a child has a 50-50 chance of inheriting it.

▸ **JUVENILE MYOCLONIC EPILEPSY** is one of the most common epilepsy syndromes. It accounts for 7 per cent of all cases of epilepsy. JME seizures usually begin around puberty; 15 per cent of children with childhood absence epilepsy develop JME. It is more likely in people who have family members with generalised epilepsy. The seizures of more than 80 per cent of people with JME are well controlled with medication.

▸ **DUCHENNE MUSCULAR DYSTROPHY** occurs in one in 3500 live births. Duchenne muscular dystrophy and Becker muscular dystrophy cause similar patterns of weakness, although Duchenne is more severe. They usually lead to wheelchair dependence and early death.

▸ **MYOTONIC DYSTROPHY** occurs in one in 8500 births and is the most common adult form of muscular dystrophy. It causes muscle wasting; symptoms include the inability to release the hand from a grip. No specific treatment has been found, though braces can be used to support muscles. Associated heart problems, cataracts and other abnormalities can be treated.

NEWS SCAN

Compiled by Deborah Smith

On deaf ears

More than seven out of 10 children will have at least one case of middle ear disease, or otitis media, by their first birthday, a report by Access Economics shows. And more than half a million Australians will be affected by mild to moderate temporary hearing impairment due to the condition. "What is particularly confronting is the number of antibiotics being prescribed for middle ear disease and the long-term implications otitis media can have on children's health and learning," Clinical Associate Professor Harvey Coates of the University of Western Australia said.

Bears in space

Water bears are the first animals to have survived exposure to the vacuum and radiation of space. Last year researchers at Kristianstad University in Sweden sent 3000 of the multicellular, invertebrate animals, about one millimetre in size, on a 12-day space trip. "Our principal finding is that the space vacuum, which entails extreme dehydration and cosmic radiation were not a problem for water bears. On the other hand, the ultraviolet radiation in space is harmful to water bears, although a few individuals can even survive that," they reported in the journal *Current Biology*.

Chimps good chums

After an argument there's often nothing better than a hug from a friend. Now it seems chimps use the same comforting techniques. British researchers observed 22 adult chimps at Chester Zoo over 18 months, recording 256 aggressive incidents. In half of the cases, once the fight was over, another animal would console the victim. – Guardian N&M

Women avoid Pap smears

Australian women still let embarrassment and busy lifestyles get in the way of Pap smears, with many avoiding the procedure for more than three years. One woman in 10, aged 28 to 34, has never had a Pap smear, which is recommended every two years. The online survey of 1003 women conducted for the Cancer Council found more than half the respondents cited embarrassment as the main reason they delayed Pap smears, while 45 per cent thought they did not have the time. – AAP

Bipolar risk for older dads

The older a man is, the more likely he is to father children who develop bipolar disorder as adults, a large Swedish study shows. The researchers examined health records of more than 7 million people with known biological parents to find 13,428 with bipolar disorder. The highest risk was in fathers 55 and older. – NY Times

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