

Press Pack



It's one of the most common inherited neurological disorders, affecting around 1 in every 2500 people, so why haven't you heard of it?

CMT Awareness Week 2010 will raise the profile of this secret condition to improve the lives of those with the CMT and increase the rate of diagnosis.



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Awareness Week will see the uk come out of the CMT closet

CMT is one of the most common, inherited neurological conditions in the world, affecting around 23,000 people in the UK, but GPs are struggling to diagnose it and people are suffering further health complications as a result. CMT Awareness Week (19 -25 April 2010) will bring this condition to the fore for the first time.

Charcot Marie Tooth Syndrome (CMT), also known as Hereditary Motor and Sensory Neuropathy (HMSN), or Peroneal Muscular Atrophy, is a heterogeneous inherited disorder of nerves that is characterized by loss of muscle tissue and touch sensation, predominantly in the feet and legs but also in the hands and arms in the advanced stages of disease. Presently incurable, it is estimated that one in every 2500 people in the UK are affected.

Symptoms and progression of the disease can vary. Scoliosis is common and other orthopaedic complications such as foot problems are common, breathing can be affected in some but this rare.

CMT United Kingdom, a registered charity, set up in 1987 decided to launch the awareness week, after a recent survey of its 1,500 members revealed that 70% of people with CMT struggled with the condition for five or more years before receiving a diagnoses.

Karen Butcher from CMT United Kingdom comments: *"In many ways CMT is a secret condition, it's never spoken about and few know that it even exists. There's a general lack of awareness in the medical profession, so people fail to be diagnosed quickly enough, meaning that many endure painful limbs, falling over and balance problems, for many years. Those that have been diagnosed don't speak of CMT, because the lack of awareness makes them feel embarrassed and isolated. By raising awareness of this condition we hope to alleviate these problems."*

Dr Mary Reilly, Consultant Neurologist at the MRC Centre for Neuromuscular Diseases, UCL Institute of Neurology and The National Hospital for Neurology and Neurosurgery, is backing the campaign, she said: *"CMT does not describe a single disorder, but a group of conditions. It is important to determine exactly what kind of CMT someone has, in order to improve their quality of life. This can be done fairly easily through examination, taking a family history, nerve conduction studies and genetic studies on blood samples. However, this sort of assessment can only be done once the diagnosis is considered in a patient, which based on research by CMT United Kingdom is taking longer than we would hope. Many people put up with CMT for a long time thinking that they are clumsy or have funny feet, suffering in silence when they could be receiving help and support."*

CMT United Kingdom is asking all of its members to wear badges during CMT Awareness Week to prompt questions from the general public. Information packs will be sent to medical professionals as well as a poster campaign in hospitals, health centres, schools and libraries.

Fast Facts

- Charcot-Marie-Tooth disease is named after the three neurologists who first described the condition in 1886.
- CMT is thought to affect approximately 23,000 people in the UK.
- CMT is sometimes referred to as hereditary motor and sensory neuropathy (HMSN).
- There are two main forms of CMT: Demyelinating (CMT1) – affects the myelin sheath insulating and nourishing the nerve's axon. Axonal (CMT2) – directly affects the axon.
- CMT affects people very differently, even within the same family.
- CMT slowly gets worse over time, causing gradual deterioration of both the motor nerves and the sensory nerves.
- CMT can cause foot drop walking gait, foot bone abnormalities (including high arches and hammer toes), problems with hand function and dexterity, balance problems, occasional cramping in the legs and arms and loss of some normal reflexes.
- CMT may cause long term pain and chronic tiredness.
- CMT is usually passed on from parent to child, with a 50% chance of the child inheriting the condition.
- CMT affects all ethnic groups equally throughout the world.



CMT in detail^{1#2}

What is Charcot-Marie-Tooth disease?

Charcot-Marie-Tooth disease is named after the three neurologists who first described the condition in 1886. Many different names have been used to describe CMT but the other commonly used name is hereditary motor and sensory neuropathy (HMSN).

CMT or HMSN therefore is used to describe a group of conditions that give rise to weakness and wasting of the muscles below the knees and often those of the hands. Many affected people also have loss of feeling in the hands and feet, and this is the 'sensory' component. The term neuropathy refers to the fact that it is the peripheral nerves (which connect the spinal cord to the muscles, joints and skin, carrying messages in both directions), which do not function normally. As the name implies, these are inherited disorders.

CMT is also referred to as peroneal muscular atrophy, because the peroneal muscles on the outer side of the calves are particularly affected. Other names include Dejerine-Sottas disease and hereditary hypertrophic neuropathy.

The different types of CMT

Peripheral nerves can be thought of as being electrical cables: the fibres (like wires) run down the middle and are wrapped in insulating material (myelin).

If the myelin is damaged the nerve impulses tend to be conducted more slowly than usual. If the fibres (also called axons) are damaged, the speed of conduction is normal but the size of the signal is reduced.

These changes can be detected by electrical tests. Traditionally, the commonest forms of CMT have been divided into two types: in type 1 the damage is to the myelin resulting in slow conduction, and this is referred to as the demyelinating type of CMT, whereas in type 2 it is the nerve fibres that are at fault and the term axonal type CMT is used.

Although major advances have been made in the last decade in the identification of the genes responsible for CMT, not all of the genes associated with CMT have yet been identified. This means that the electrical test (nerve conduction studies) is still often very useful in making the diagnosis. An exception is when there is a clear family history of autosomal dominant inheritance, when DNA testing for the commonest form of CMT (type 1A) might be performed as a first investigation.

Although the electrical tests are usually clearcut, allowing a diagnosis of CMT type 1 or CMT type 2, in some cases it may be difficult to decide from the electrical test which type of CMT a patient has and the term intermediate CMT is used. Although this can be confusing for patients it is useful for doctors in deciding which genes to screen. In the future, when all the causative genes are identified, a comprehensive classification of CMT will be available but in view of the fact that there are likely to be numerous genes involved it is probable that electrical tests will remain important in the initial assessment of patients.

How does CMT affect people?

The first evidence of CMT is usually between the ages of 5 and 15 but sometimes may not be until very much later, even into middle-age. Usually the first symptom is slight difficulty in walking because of problems with picking up the feet, and this may well be noted by parents first. Many people, particularly those with CMT type 1, have high arched feet (referred to as pes cavus) and this may be obvious from a very early age. It tends to become particularly noticeable at the time of the growth spurt associated with puberty. Weakness of the hands occurs in some people, but this does not usually cause symptoms until after the age of 20.

Patients can experience numbness of the feet and hands (usually noticed in the feet first) which is not often troublesome, but the tendency to have cold feet is frequent. Very rarely the numbness can be severe, and it is then easy for affected individuals to injure themselves without knowing it; painless ulcers of the feet may develop as a result of poorly fitting shoes, or burns on the hand from hot cups etc. Pain is not a common feature of CMT and if present may be due to secondary effects on the joints or muscles.

The reflexes (such as the knee jerk) are commonly lost. This does not cause any trouble for the individual, but is often noted early on by doctors. A few people with CMT 1 have shakiness of the hands (tremor) and the combination of tremor and CMT is sometimes referred to as the Roussy-Levy Syndrome.

Mild curvature of the spine (scoliosis) occurs in some people and tends to be more severe in those with early onset of limb problems.

[More >](#)

CMT in detail_{2#2}

The types of CMT which run through the generations in families (see section on dominant inheritance) are not usually severely disabling disorders and often do not change a great deal after people have finished growing. It is unusual for people with CMT to lose the ability to walk, although some older people need a stick or other walking aids. It is important to stress that the disorder often varies enormously in severity, even in members of the same family, and 10 to 20% of affected individuals have no symptoms at all but are found to have evidence of the condition on examination or using electrical tests.

How is CMT inherited?

The commonest forms of CMT are inherited in a way that is referred to as autosomal dominant (AD). This type of inheritance is the most common type in CMT 1 and CMT 2, each child of an affected parent has a 50% chance of inheriting the abnormal gene and being affected. People of either sex can have the condition.

However in occasional families with CMT 1 and CMT 2 the inheritance is autosomal recessive (AR). In AR inheritance a person needs two abnormal copies of the gene to be affected unlike AD inheritance where the person only needs one copy of the abnormal gene to be affected. AR inheritance is only seen if both parents are 'carriers' of the faulty gene but these parents do not themselves have any symptoms. Both parents therefore have one abnormal and one normal copy of the gene. The condition develops only if a child inherits the abnormal gene in a double dose, i.e. one from each parent.

Males and females can be affected. Many people with autosomal recessive CMT do not have affected relatives as each child has only a 1 in 4 chance of being affected and most families are quite small. It is therefore common for only one child to be affected.

In some families, CMT is caused by an X-linked gene (X-linked inheritance) which is carried on the X chromosome, one of the so-called sex chromosomes which determine the sex of the child (females are XX, males are XY). The result is that boys inherit the disease from their mothers who are known as carriers. Carriers may show no sign of disease, although sometimes they are mildly affected, but each of their sons has a 50% chance of having CMT and each of their daughters has a 50% chance of being a carrier. Affected males cannot transmit the disease to their sons.

In these families therefore, males are more severely affected than females and males cannot pass on the disease to their sons.

Problems and management

There is no specific treatment at present for the underlying genetic defect in CMT even in those patients in whom a genetic diagnosis has been made although there are many groups researching this area. This does not mean that patients with CMT cannot be treated or helped in many other ways. It is very important that problems patients experience such as foot problems are addressed appropriately and this may greatly improve a patient's quality of life. Accurate genetic diagnosis and genetic counselling is the other area of management in which there has been rapid development in recent years.

Ideally children and teenagers with CMT should be seen annually by a neurologist or a paediatrician to ensure that severe problems with the feet do not develop. Surgery may be helpful for very highly arched feet, either to reduce the arch and the curling of the toes which often goes with it, or to fuse together some of the foot bones. After procedures of this sort, and any other operation, it is essential to minimise periods spent in bed, as increased difficulties in walking are often noticed afterwards.

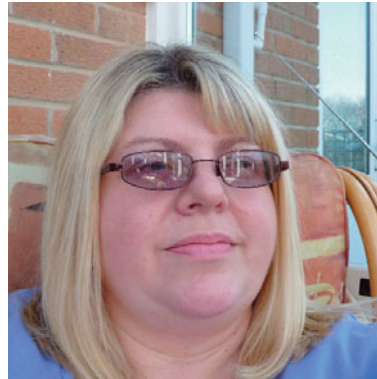
Just as rest may enhance difficulty in walking, active exercise and maintaining fitness help to maintain mobility. Surgery is not usually needed for scoliosis but may have to be considered in the very few cases in which this is severe. In those with a lot of numbness of the feet, it is important to take great care of the feet, washing and drying them carefully, and inspecting the skin for small ulcers. The inside of the shoes should be shaken out to remove small stones etc., and the insides felt for irregularities that could damage the skin.

Case studies 1#2

Karen Morris

I was diagnosed when I was 20, the GP kept telling me it was stress as I kept getting cramps from my hips to my toes in both legs and getting a cramp in my tongue. At first I was sent away with just paracetamol. I ended up getting a second opinion and paying to find out what was wrong with me.

I've also experienced a general lack of support from my GP in the past, when I asked for a referral to a neurologist, I was met with the reply; "there's nothing they can do for you so what's the point?".



Bev Harrison

When I was about 16 I started having joint problems and was admitted into hospital when I was 17. They shortened a tendon and put a pin in my left leg. Due to continued problems I then had a pattelectomy carried out when I was 21 which was not very successful. This was due to chronic pains in my legs and lower limbs but it was not until my son, who was born in 1980 got to the age of 7 when it was brought to our attention that he had an awkward gait at his school medical and their first thoughts were that he had Friedreich's Ataxia.

Various tests and appointments were carried out by the Neurologist and we honestly thought that he had a very serious disabling and debilitating condition - first he was tested for Friedreich's Ataxia Muscular Dystrophy and then when he had blood tests carried out it was ascertained that he had Charcot Marie Tooth Disease and as it was a hereditary condition. I was also tested and it was also ascertained that I also had the condition and was told that the operations that I had when I was 17 should not have been carried out.

I have since been plagued with chronic pain and fatigue a lot of it caused by the CMT but also caused by Chronic Arthritis and had a knee replacement carried out in April 2009 which again has not been very successful. I have been walking with crutches for some 8 years now and occasionally use a wheelchair/mobility scooter. My home had to be adapted to include a stairlift, wet room shower and downstairs WC and a bath lift which has been

done with very many grateful thanks to the local authority. I do however manage to work on a part time basis and try to keep myself going as much as I can. I have however found that the CMT is now affecting my hand/arms co-ordination at times.

My son's condition is settled at the moment but the CMT does not only affect his legs and his gait but he has problems with his hands and he had to have special needs assistance throughout school. He now works in IT and has had knee braces prescribed which he wears when he feels that he needs to.



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Case studies 2#2

Karen Sewell

I first of all noticed there was something wrong with my son's feet at the beginning of 2008. Todd my youngest son went to have his feet measured for new shoes as normal and it showed that his feet were 2 sizes different. So, I took him to see our GP I had also noticed that his foot was starting to turn outwards. This was also the same for my oldest son. When I took Todd to the G.P I explained the difference in size and also the foot turning outwards. He then told me that there was nothing that could be done. So I just went away.

I still was not happy so I took Todd to the school nurse to ask for her advice. She then, thankfully, referred Todd and Jed to the podiatrist at our local clinic. Who suggested that they should both be referred to the orthopedic consultant at Broomfield hospital, Chelmsford, Essex.

We finally got an appointment in January 2009. We saw an excellent consultant called Mr. Tuite. Who sent the boys for a number of tests which included nerve conductive tests and an MRI scan. He had told us that he thought it was a nerve problem.

While the boys were having their nerve test the consultant also tested me. As looking back in my family history there were clues that this had been in our family for at least four generations and we knew nothing about it. Just put weakness in the hands and feet down to something that we seemed to suffer from but didn't know it was actually called something. And that we weren't just being clumsy when we trip over virtually nothing.

What a sudden relief.

So the next step was for Todd to have tendon transfer on his left foot which was the worst affected and the right foot just had the tendons released. This seems to have done the trick.

As for Jed his problem is a bit more advanced so Mr. Tuite referred him to Mr. Hill at Great Ormond Street. Jed is still waiting to go in for an operation. He has got to have tendon transfer, hindfoot osteotomy and soft tissue release.

I have now had the genetics test and I have CMT type 1a. We have also found out that my brother is also affected and also his daughter. He has two other children who are not at present showing any signs. I think it is a shame that the G.P's don't seem to know anything about the disease.



Media spokespeople

Dr Mary Reilly

UCL Institute of Neurology and The National Hospital for Neurology and Neurosurgery

Dr. Mary M. Reilly graduated from University College Dublin in 1986. She started her clinical training in Neurology in Dublin after gaining her MRCPI in 1988. She moved to the National Hospital for Neurology and Neurosurgery, London in 1991 where she completed an MD thesis on Familial Amyloid Polyneuropathy. She then completed her clinical neurological training subspecialising in peripheral nerve diseases. Since 1998, she is head of the peripheral nerve services in the National Hospital for Neurology and Neurosurgery and is actively involved in research in genetic neuropathies. She received her FRCP in 2002 and her FRCPI in 2003.



Gita Ramdharry PhD MSc BSc(Hons) MCSP

Senior Lecturer, Faculty of Health and Social Care Sciences, St George's University of London/Kingston University

NIHR Fellow, MRC Centre for Neuromuscular Diseases, Institute of Neurology, UCL

Gita qualified as a physiotherapist in 1995 and specialised in treating people with neurological disorders in 1997. In 2001 she worked at the National Hospital for Neurology and Neurosurgery as a senior physiotherapist. This is where she first started treating people with CMT in the out-patients and orthotics clinic.

Based on her experiences of working with people with CMT, Gita started a PhD at the Institute of Neurology, UCL in 2004 investigating how people with CMT compensate for muscle weakness while walking, and the effects of splints. On completion of the PhD in 2007 Gita moved to St George's school of physiotherapy to work as a senior lecturer.

She maintained her links with the institute of neurology and collaborated with Dr Mary Reilly to run an exercise trial looking at the effects of strengthening hip muscles in people with CMT.

In December 2009, Gita was awarded an National Institute for Health Research fellowship for research into balance and falls in people with neuromuscular disorders. Gita is now working part time at the new MRC neuromuscular centre in London to pursue this research.





For more information, to set up an interview with a spokesperson or case study please contact Kate Pearson: email kate@cmtuk.org.uk or telephone **07894 055 959**

