Charcot-Marie-Tooth -

A Practical Guide

Researched and Written by John Isitt

“Working to support those who are affected by Charcot-Marie-Tooth Disease, also known as Hereditary Motor and Sensory Neuropathy or Peroneal Muscular Atrophy”
Charcot-Marie-Tooth - A Practical Guide

second edition

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Preface

2007 brings the 21st Anniversary of CMT United Kingdom and we head towards that landmark in buoyant shape and mood. The support group has come a long way since our formation, indeed in the six years since Charcot-Marie-Tooth Disease: A Practical Guide was first written. Membership has increased from 980 to over 1250 since then, and we have revised and extended our website which has now become our “window” on the world. We remain committed to helping support and inform people affected by CMT both within and beyond our membership. Between the website and this new edition of “CMT: A Practical Guide” we aim to do just that.

Once again the content and style have been led by members of the National support group, all with direct experience of living with the condition, problems faced and solutions. While bringing together information on all aspects of CMT and how to manage it, we have sought to keep medical terminology to a minimum, making it even more readable and relevant than before. We believe this really is a practical guide, and a valuable bible for those who have CMT or are interested in learning more about it, including professionals.

Available in loose-leaf format and on CD-Rom, the Guide is designed to be easily updated and extended – reflecting the ever-changing world of medical knowledge and services. Our thanks to all those who contributed, including the book’s focus group, the many medical advisers and John Isitt, of Resonant Media, who researched and compiled the book.

The Trustees of CMT United Kingdom

April 2006
Foreward

Charcot-Marie-Tooth Disease (CMT) is a condition that causes deterioration of the peripheral nerves controlling sensory information and muscle function in the hands, forearms, lower legs and feet. It can lead to foot problems such as high arches and foot-drop, weakness, problems with balance and hand function, and loss of some normal reflexes. It can also cause pain and fatigue and occasionally more severe disability, but is not life-threatening and does not affect life expectancy.

Coping with any medical condition can be daunting, but even more so when there is little information and even the doctors find it hard to diagnose or know little about it. Despite being the most common inherited neurological condition, affecting 1 in 2500 worldwide, that is the problem faced by many people with Charcot-Marie-Tooth Disease (CMT).

This book aims to bridge the gap, acting as a guide for both those affected by CMT and medical staff who want to find out more about it. It covers theoretical and practical issues - from understanding the genetics and mechanics of the condition, to diagnosis, the latest treatments available, and advice on coping and managing the condition day to day.
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“Working to support those who are affected by Charcot-Marie-Tooth Disease, also known as Hereditary Motor and Sensory Neuropathy or Peroneal Muscular Atrophy”
What is CMT?

CMT is a condition that damages your peripheral nerves. These nerves are responsible for passing on commands from your brain to your muscles (motor nerves) and for passing information to the brain about sensations, such as pain, heat, cold and touch (sensory nerves).

Because of the nerve damage, people with CMT may find that some of their muscles become slowly weaker over the years, particularly in their feet and hands. And some people find that their sense of feeling becomes duller, or numb, in the same areas.

In the UK, some 25,000 people are thought to have CMT, making it the most common inherited neurological condition.

Other key points about CMT are that it:

- is hardly ever life threatening, but often becomes slowly worse over the years
- affects people very differently, even in the same family
- can cause the muscles in the foot, lower leg, hand and forearm to get weaker (waste)
- can cause foot drop walking gait, foot bone abnormalities (eg high arches and hammer toes), problems with hand function, balance problems, occasional lower leg and forearm muscle cramping, and loss of some normal reflexes
- may cause long-term pain and tiredness (fatigue)
- is usually passed on from parent to child
- affects all ethnic groups throughout the world
- is the focus of major genetic research, bringing us close to answering the CMT enigma.

What causes CMT?

CMT is caused by a genetic fault (mutation) that leads to damage of the nerves in your legs and arms.

In order for you to move with speed and precision, messages have to be relayed between your brain and the rest of your body within a fraction of a second. These messages are relayed through your nerves.

For example, if you want to move your leg, an electrical message is sent from your brain, via the spinal cord, to the muscles in your leg along a motor nerve.

If you cut or burn your leg, you feel it because an electrical signal is sent from the affected area, up the sensory nerves, via the spinal cord, to your brain.

The nerves in your arms and legs, called peripheral nerves, can be compared to electrical cables. The central “wire” is known as the axon and the “plastic outer” is called the myelin sheath.

Axons transmit the electrical signals to and from the brain, and myelin sheath acts as insulation, speeding up the signal and nourishing the central axon.
Some forms of CMT affect the axon – making the signal to and from the brain weaker and less efficient. Other forms of CMT affect the myelin sheath, slowing down the signal.

Damage to the axon causes the symptoms of CMT. Without an intact axon and myelin sheath, your nerves are unable to activate target muscles or relay sensory information from your limbs back to the brain.

As at February 2006, 21 genes have been found to cause different types of CMT. Each one of these genes is responsible for making particular proteins that are essential to the axon or myelin sheaths.

Read more about the genetic causes of CMT in the chapter, Genes: what they mean for you.

The different types of CMT

There are two main types of CMT:

1. **Demyelinating (CMT1)** – this form of CMT affects the myelin sheath that insulates the central axon.
2. **Axonal (CMT2)** – this form of CMT affects the central axon.

There are two other variations of CMT1 and CMT2, known as:

- CMTX
- DI-CMT

You may have heard of other types of CMT, including CMT3 and CMT4 for example. Because of our better understanding of the genetic causes of CMT, we now know that all of these "different types are in fact simply variations of the two main types – demyelinating and axonal."
Demyelinating CMT (CMT1)

CMT1 is the most common form of CMT (six out of ten people with CMT will have CMT1A), affecting the nerve’s insulating myelin sheath.

People with CMT1 will usually notice symptoms developing in childhood or adolescence (usually between the ages of five and 15).

Some forms of CMT1 have an earlier age of onset and these forms used to be called HMSN3 in the old classification. Two of these forms are called Dejerine Sottas disease (DSD) and Congenital Hypomyelinating Neuropathy (CHN).

Axonal CMT (CMT2)

CMT2 is not as common as CMT1, but has similar symptoms. Rather than damaging the nerves’ insulating myelin sheath, CMT2 affects the axon. Because of this CMT2 is also known as “axonal CMT”.

Symptoms are often first noticed between the ages of 10 and 20.

CMTX

CMTX is a variation of CMT1 and CMT2. It is called “X” because the gene that causes this form of CMT is carried on the X chromosome. It usually affects men much more severely than women because a man has only one X chromosome and women have two. Men will usually develop symptoms beginning in late childhood or adolescence. Women may be completely unaffected, in which case they tend to be known as “carriers”; or they may be affected; but usually not as severely as males.

DI-CMT

DI stands for “dominant intermediate” and this form of CMT is rare and usually has similar symptoms as the common versions of CMT1 and CMT2. The reason it is called intermediate is that both the myelin sheath and the axon are damaged equally.

For both CMT1 and CMT2 there are many different subtypes and you may hear about CMT1A or CMT2AR, for example. These are all different genetic variations of the main types.

Find out more about the different types of CMT and the genetic background to each, read about genes in the chapter, Genes: what they mean for you.

CMT – the axon and myelin sheath

Now we know more about the types of CMT, it may be helpful to find out a little more about how the myelin sheath and the axon work together.

To recap, our nerves can be compared to an electrical cable. The wire running down the inside of the cable is called the axon and the insulating plastic is called the myelin sheath.
Simply put, damage to the axon means that the signal becomes weaker; whereas damage to the myelin sheath slows down the signal (doctors call this nerve conduction velocity.

What is not so commonly known is that it is only when the axon itself is damaged that you get the symptoms of CMT. So, why does CMT1, in which the myelin sheath is damaged, lead to the symptoms of CMT?

The role of the myelin sheath

As well as insulating the axon, the myelin sheath also nourishes it. Eventually, if the damage to the myelin sheath continues, the axon is damaged as the myelin withers. This is known as secondary axonal damage. Only when this happens do the symptoms of CMT become apparent.

What this means is that, if you have CMT1, although the speed that your nerves pass on messages may be slow, this in itself will not cause CMT. In fact you can live with slow nerves for decades with no sign or symptom of CMT. It is only when the damage to the myelin sheath becomes so severe that the axon is also damaged that you will be affected.

Knowing your type – is it important?

It is not always possible to know what type of CMT you have but nerve conduction tests can usually tell you whether you have CMT1 or CMT2. An accurate diagnosis is often only possible with a genetic test, but not all the genes that cause the different types of CMT have been found.

So far, 21 genes involved with CMT have been identified (more are being found every year). Of these, only one is widely available to be tested in regional genetic laboratories. Three more can be tested in about six laboratories in the UK, the rest are only usually available to be tested as part of medical research.

From a day-to-day point of view, it is not particularly important to know what type of CMT you have, as current treatment options are not based on what type of CMT you have, but rather on your particular needs. (In the future this could change, as it is possible that future treatments may become available that only benefit people with certain genetic types of CMT.)

Currently, there are four practical benefits of having an accurate genetic diagnosis:

1. It can help give you a better idea of the course of your CMT in the future. People within the same family can have very different symptoms. But, certain genes give a spectrum of likely symptoms and how the condition is likely to change over the years. For example, this could be important for teenagers trying to make a career choice as they could get a better understanding of how the CMT will be affecting them in 10 or 20 years.
The symptoms of CMT

The exact symptoms that you may experience may vary hugely from other people with CMT. This is because there are many different subtypes of CMT. And it is perfectly possible that you may never have any symptoms, as anywhere between one in 10 and one in five people with CMT will never have any symptoms. (Some people may only get symptoms later in life, even if they have a type that ‘normally’ shows symptoms by the teenage years.)

Early symptoms may include:

- slight difficulty walking because of problems picking up the feet (foot drop)
- children may experience difficulty with running and general agility before any other noticeable symptoms – including being “clumsy”
- high arches (some people will have abnormally flat feet)
- weakness in the hand and forearms, although the feet are usually affected first.

Other symptoms can include:

- some loss of feeling in the feet, lower legs, hands and forearms, although this is rarely troublesome
- loss of fine control in the hands, making it difficult to write, do fiddly things or open jars, for example
- some muscle tremor
- tiredness (fatigue) because of the extra effort needed to do daily activities
- slight curve to the spine
- increased difficulty walking – aids may be needed, such as orthoses and walking sticks
- hip or knee problems.

Rarely, people with CMT can develop more severe symptoms. These can include:

- a severe curve of the spine (scoliosis)
- speech and swallowing difficulties
- some difficulty breathing, particularly at night.
What does CMT stand for?

CMT is an acronym for Charcot Marie Tooth, the surnames of the three doctors that first described CMT in the late 1800s: Frenchmen Jean-Martin Charcot and Pierre Marie; and Howard Henry Tooth from the UK.

CMT has many other names, the most common being:

- **Hereditary motor and sensory neuropathy (HMSN)** – so called because it is passed on through families (hereditary) and affects both the motor and sensory nerves (neuropathy).

- **Peroneal muscular atrophy (PMA)** – so called because one of the muscles that is often first affected, becoming weaker and wasted (atrophy) is called the peroneus muscle, found in the shin.

Today the term CMT is the most commonly used, although HMSN is still used in some medical literature.

**HNPP**

Although once thought to be an entirely different condition to CMT it is now understood that HNPP is in fact genetically very similar. In fact, in some classifications, HNPP is listed as a subtype of CMT1.

Both HNPP and CMT1A are genetically close and cause problems in the insulating myelin sheath because of a defect in the PMP22 gene (in HNPP there is one too few and in CMT1A there is one too many).

The symptoms of HNPP, however, are usually quite different. People with HNPP often get irregular attacks of numbness or paralysis (palsy) mostly in the arms or legs which usually clear up after a few weeks. At other times people with HNPP have normal strength and sensation in their limbs.

Usually the symptoms of HNPP are triggered by a stretching or pressure on the nerves, although sometimes there is no obvious cause. Occasionally HNPP can get worse slowly (progressive) and resembles CMT.

For more information about HNPP see Appendix 2.

**CMT and other conditions**

There are a number of conditions that have some similarities to and can be confused with CMT.

**Distal Hereditary Motor neuropathy (dHMN)**

dHMN, also known as distal Spinal Muscular atrophy (DSMA), and CMT both lead to muscle wasting and weakness usually affecting the feet first and then the hands. dHMN differs from CMT in that it only affects the motor nerves (nerves to muscles) and never the sensory nerves (nerves for sensation) whereas CMT affects both motor and sensory nerves. There are many different types of dHMN
described and some types are caused by the same genes that cause some forms of CMT2. Genetic testing is available in this type of CMT on a research basis.

**Charcot’s foot/joint**

Despite its name Charcot’s joint should not be confused with Charcot-Marie-Tooth (CMT) - they are very different conditions. Charcot’s joint is a complication of a severe sensory neuropathy (diabetes is the most common cause) leading to a damaged and swollen joint. Charcot’s joint is only rarely seen in CMT.

**Muscular dystrophy**

It is a common mistake to think of CMT as a muscular condition. However, despite muscle weakness in the legs and arms, CMT does not directly affect the muscle, unlike muscular dystrophy (MD). CMT is a neurological condition, meaning that it damages the nerves. This damage to the nerves then leads to weakness in the muscles.

**Multiple sclerosis (MS)**

CMT1 and multiple sclerosis (MS) are outwardly similar as they both lead to damage of the myelin sheath that insulates the nerves. But CMT only affects the nerves in the legs and the arms (peripheral nerves), whereas MS affects the central nervous system (the spine and the brain).

Another difference between CMT and MS is how they are caused. In most cases CMT is passed on from parent to child whereas, in MS, no single cause has been identified. There seems to some genetic link, but it seems that MS is triggered by something like a virus or allergy.

**Questions and answers**

I’m sure I’d heard that there were more than just the two types of CMT. Why aren’t they mentioned here?

Not so long ago, it was thought that there were many more different types of CMT; you may have heard of CMT3 and CMT4. In fact, once there used to be eight types. Things are much simpler now, as doctors and geneticists understand that many of the old types – 3 and 4 for example, were in fact just more severe forms of type 1 and 2.

The key point to remember is that, CMT1 affects the myelin sheath (if you think of an electrical cable, the myelin sheath is the insulating plastic). Whereas CMT2 affects the axon (using the same cable analogy, the axon is the wire).

CMTX is just a variation of either CMT1 or CMT2 – but is passed on from parent to child in a different way. DI-CMT (intermediate) is a form of CMT that seems to affect both the axon and the myelin sheath.

I’ve heard CMT described as ‘progressive’. What does this mean?

In medical terms, progressive means that a condition will get worse over time. But what is not defined is how much worse or over how much time. The problems linked with CMT do usually get
worse over time, but usually very slowly over the years, if not decades, often with little or no change from young adulthood to late adulthood.

“Like a lot of progressive conditions you can go through a period of tremendous progression and then remission. It’s strange sometimes it hits you and you realise you’ve got worse – but whether it’s some form of denial I don’t know. I realised about four or five years ago that I was having problems tying my shoelaces and it dawned on me it had probably been that way for a while.” Margaret

Is CMT a disease?

CMT is not a disease like flu, HIV or measles. It cannot be caught from another person. Although the medical definition of a disease is “a disorder with a specific cause and recognisable signs and symptoms”, most people with CMT prefer not to refer to it as a disease, but rather as a condition.

Does CMT affect the autonomic nervous system?

Yes, although the main affect is on our peripheral nerves.

The autonomic nervous system is a system of nerves over which we have no direct control, and sometimes people call it the automatic nervous system. It is responsible for controlling blood pressure, and it does that, in part, by dilating and contracting the blood vessels, thereby directing blood to where it is needed.

In CMT and other disorders that affect the autonomic nervous system, this tends to cause reduced blood flow in the skin of the feet and shins. It is this that causes the coldness and blue discolouration. It can be uncomfortable, but doesn’t cause any serious problems.
Diagnosis

If you and your GP think you may have CMT you will be referred to a doctor who specialises in problems of the nerves, known as a neurologist.

Steps to a diagnosis

When you see a neurologist, he or she will follow a number of steps to confirm whether you have the condition or not.

1. As a first step, your doctor will take a standard patient history, including family history. Your doctor will ask you about your symptoms – what they are like and how long you have had them – and whether any of your family have CMT or have had similar symptoms.

2. Next, you will be given a neurological examination. Your doctor will look for evidence of muscle weakness in the arms, legs, hands and feet; as well as signs of muscle wasting, reduced reflexes and any sensory loss. Your doctor will also look for any signs of foot problems – such as high arches, hammer toes, inverted heel or flat feet. Any sign of enlarged nerves will also be looked out for (these may be seen or felt through the skin).

3. If your doctor suspects CMT, you may be asked to undergo some tests called electro-diagnostic tests. These consist of two parts:
   a) Nerve conduction tests – electrodes are placed on your skin over your peripheral motor or sensory nerves. These electrodes will give you a mild electrical shock, which may be uncomfortable. The shock stimulates your sensory and motor nerves and the doctor will be able to measure the speed at which the signal was transmitted and the size of the signal.
   b) Electromyography (EMG) – a needle electrode is inserted through your skin to measure the electrical activity of the muscles.

In the UK, the next stage of diagnosis will be to do a genetic test to try and determine which type of CMT you have. Not all the genes to do with CMT have been identified yet, or are commonly available to test, so this test may not be conclusive.

Although it used to be common practice, in the vast majority of adults a nerve biopsy is no longer necessary, although it is still sometimes useful in children. A nerve biopsy involves removing a small piece of peripheral nerve through a cut in the skin, usually from the calf. The nerve is then examined in a laboratory for any signs of abnormalities.

For more information about genes and genetic testing, see the chapter on Genes: what they mean for you.

“I noticed I couldn’t wear high heels – I was trying to walk and I couldn’t understand why I couldn’t. But you rationalise it – you say, ‘Oh it must be because I twisted my ankle so many times’. But of course, twisted ankles are another symptom. This was the late 80s and I was in my early forties.” Rose
What should happen now?

Once you have been diagnosed the following steps, or similar, should happen. (It may be a good idea for you to photocopy or print this list out and talk it through with your neurologist and GP to make sure that you get the proper care that you deserve.)

Ideally, you should have the following:

1. You should receive a copy of your clinic letter. Ideally this will be phrased in such a way that you can show it to your other family members to explain your diagnosis and to help explain to them that it would be a good idea for them to see a specialist too. (Read about telling people about CMT in the chapter on Coping with CMT.)

2. Follow up appointment within a month of diagnosis – probably with a specialist nurse, geneticist.

3. Find out as much information as you can about CMT. A very good source of information is the CMT United Kingdom website, otherwise find out more about finding good health information in the chapter on Managing your CMT.

4. Get a referral to see a physiotherapist with good understanding of neuromuscular conditions and an orthotist, if necessary.

5. A system should be put into place that if you discover other, new, symptoms they can be checked to see whether these symptoms have anything to do with CMT or not.

6. Within two or three months, you should have a follow up clinic. And as part of this clinic you should be given information on:
   a) CMT United Kingdom
   b) Disability living allowance (if needed)
   c) Mobility issues, including the DVLA and Motability
   d) Exams – how to get more time if you are taking GCSE, A’ Levels or other exams, for example.
   e) Occupational help, if necessary.

In the real world, what may happen on diagnosis?

Unfortunately, in the real world, it is quite likely that your diagnosis will be given to you by someone who does not know much about CMT. It is quite likely that you will be told: “CMT is a progressive condition, there is no cure and there are no treatments – go away and live with it”. This is unhelpful and wrong.

CMT can be well managed and well treated. But you may need to knock some heads together to get the care you deserve. Remember, if you are having difficulty getting the treatment you need, contact CMT United Kingdom.
Reacting to your diagnosis

Everyone reacts differently when told they have CMT. Just as the condition affects each person differently, so everyone has a unique reaction to the news. Nobody – not your doctor, partner, family or friends – can tell you what you should or should not be feeling or how you should react.

“My parents knew I had something wrong when I started to walk. I walked slower and by the time I was five or six there was obviously something wrong I couldn’t run as fast as the other children. When I was about 12 a doctor said I had peroneal muscular atrophy. It was only when I was an adult and living away that I found out what it was.” Carolyn

The important thing to remember is that your emotional response is perfectly normal and could include:

- shock
- denial
- confusion
- fear
- wanting to avoid the issue
- anger
- grief
- guilt
- wanting to tell everyone or no-one
- relief.

A feeling of relief upon diagnosis is quite common, as many people with CMT have lived with the symptoms and problems of the condition for many years before discovering it is CMT. Learning that there is a name for the problems experienced over the years can help to understand and define the symptoms you have been experiencing. Some people call this “closure” and explain that they felt relief on diagnosis to know that the problems they had been living with for years were not symptoms of something more serious.

“In the 1960s I was told that I had a rare form of Muscular Dystrophy. But finally, when I came to Cardiff in 1981, I was told it was Charcot Marie Tooth (CMT). When I asked about the original diagnosis they said, ‘Oh we knew what you had, but thought you’d understand Muscular Dystrophy better’. Margaret
How you cope with the news of the diagnosis is also up to you. But you may want to think about the following five steps:

1  Take the time you need

   Do not rush into making important decisions about your health and life in general. You have time to think about your options and decide what is best for you.

   Taking the time you need to make decisions can help you:
   - feel less anxious and stressed
   - avoid depression
   - cope better with CMT
   - feel more in control.

2  Get the support you need

   When you are ready, talk with your family and friends. And to other people with CMT: the people who are going through the same thing as you and who may have “been there” already. They may be able to help you make informed decisions.

   CMT United Kingdom runs a number of local groups across the UK. Or, why not use the charity’s online Forum to talk to other people coping with the condition.

   “People with CMT have the usual mental health baggage which we all bring with us from our growing up years. It may or may not be CMT-related, but it does impact on our ability to function and make decisions as mature adults. It may have a positive impact (many of us consider ourselves “stronger”’ for having grown up “different”).

   “It may also have a negative impact, making us more vulnerable to stresses at work or at home. There are mental health professionals who are trained in various techniques to assist us in realising who we are and how to manage our lives within our strengths and weaknesses. We can be grateful that in these times it is not a stigma to see a therapist to focus on an issue we have defined as troublesome.” Susan

3  Talk with your doctor and other medical staff

   Try and strike up a good relationship with your doctor and other members of your healthcare team (neurologist, physiotherapist, etc). It will help you feel more satisfied with the care you receive. Research shows it may have a positive effect on symptoms and pain.

   Read the chapter on Treating your CMT to find out how specialist healthcare professionals can help you.

   If you feel that it would help, get a second opinion. See “Getting the most out of your doctor” in the chapter on Treating your CMT.
4 Seek out information

Many people find that discovering as much as possible about CMT really helps. If this suits you, and it may not, then make sure you look for information that is based on the latest reviewed scientific findings (find out more about good sources of evidenced based health information in the chapter on Managing your CMT).

5 Decide on a treatment plan

Work with your doctor and healthcare team to decide on a treatment and care plan that best suits you. Remember, research shows that most people with a long-term condition, like CMT, do better if they are involved in their healthcare.

Top 10 questions to ask your doctor after diagnosis

1. What is the technical name of my specific condition, and what does it mean in plain English?
2. What is my outlook for the future (prognosis)? When can I get a genetic test?
3. How soon do I need to make any decisions about treatment?
4. Will I need any additional tests, and if so, what kind and when? When can I see a specialist physiotherapist who understands about neurological conditions?
5. What are my treatment options?
6. What are the pros and cons of my treatment options (including risks of not having the treatment)?
7. Is there a research study (clinical trial) that I could benefit from?
8. Now that I have this diagnosis, what changes will I need to make in my daily life?
9. Apart from CMT United Kingdom, what organisations could help with support and information?
10. What resources do you recommend for further information?
“Working to support those who are affected by Charcot-Marie-Tooth Disease, also known as Hereditary Motor and Sensory Neuropathy or Peroneal Muscular Atrophy”
The mechanics of CMT

Whatever form of CMT you have the mechanics are broadly similar.

The damage caused by CMT to your peripheral nerves may lead to two underlying problems (known as primary symptoms):

1. Muscle wasting and weakness, usually first noticed in your feet and hands. Because the muscles in your legs and arms stop receiving signals from your brain, due to the damage to the peripheral motor nerves, they start to waste away through lack of use, leading to muscle weakness.

2. Loss of some degree of sensation, again usually starting in your feet and hands, although this is often not noticed until it is severe or has caused skin problems.

Muscle wasting and weakness

As the muscles weaken through wasting there are two problems that arise:

- contractures
- strain on other muscle groups.

1. Contractures

Nearly all muscles have an opposing muscle that balances the body as it moves. These pairs of muscles are called antagonists and allow for precise body control.

For example, your biceps are the muscles that bend your arm at the elbow (the classic bodybuilder’s pose) and your triceps do the exact opposite; straightening your arm.

Antagonistic muscles control every single joint – elbow, ankle, hip and those in your toes and fingers.

Muscles have a natural tendency to contract and tighten, only being stretched by their opposing muscle. Problems arise when a muscle weakens (wastes); the opposing muscle that continues to function is not stretched by this weak muscle, allowing the stronger muscle to become tighter and shorter. This mismatch can pull the joint out of shape.

One of the most important objectives of managing your CMT is to stop this tightening of a muscle before it damages the joint, reduces flexibility and leads to deformity. This can best be achieved by daily stretches and regular exercise. (Read more about this in the chapter, Exercise and stretching.)

Problems caused by an imbalance between muscles usually start as flexible deformities and progress to fixed deformities.

1. Flexible deformities – means that a joint, although damaged, can still be moved and it may be possible to prevent further damage, such as a fixed deformity, through managed stretching, physiotherapy and orthotics.
2 Fixed deformities – means that the joint has “welded” together. Usually the only effective treatment at this point is surgery, although orthotics and physiotherapy can both help to prevent any further problems occurring.

In CMT the most common changes can be found in the foot and ankle, due to wasting and weakness of the shin muscles. The ankle is mainly controlled by the shin muscles (at the front of your lower leg), which pull the ankle up, and the calf muscles (large muscles at the back of your lower leg), which pull the ankle down.

Because of weakness in the shin muscles people develop foot drop as it becomes harder for the shin muscles to pull up the ankle. Often this is accompanied by the heel turning in so that, when viewed from behind, it looks as though the person is walking on the outside edge of the foot, causing instability and balance problems. (Medically this is sometimes called heel varus.)

At the same time, the Achilles tendon (at the back of the foot) and calf muscles meet less and less resistance from the shin muscles and become shorter and stiffer through lack of use. If the calf muscle and the Achilles tendon are left to tighten and contract they will pull the foot and toes out of shape, leading to very high arches (medically known as pes cavus or cavus foot) and clawed toes.

Strain on other parts of the body

The body is very adaptable. If one muscle stops working another muscle will try to take over its job. This has a twofold effect. Firstly it makes you tire more quickly (fatigue) and secondly it puts your body under a lot of strain as one part of the body tries to compensate for another.

Loss of sensation

Because CMT damages the sensory nerves, many people with CMT are less sensitive to heat, touch and pain in the feet, lower legs and, less frequently, in the hands.

Although this lack of feeling is rarely severe enough to cause numbness, it can make people with CMT less likely to notice problems such as splinters or cuts to the feet, or accidental burns (hot
cups burning the hand, for example or over-hot water bottles burning the feet).

As well as loss of feeling (numbness) damage to the sensory nerves affects your awareness of your joint position and tension in your muscles, resulting in poor balance (a combination of not knowing where your limbs are and not being able to move them as efficiently). This partially explains why people with CMT feel they need to look down to see where their feet are. It also makes vision a more important aspect of balance and many people with CMT have far poorer balance with eyes shut or in the dark as they depend more on sight for balance.

Cold feet are also often a problem for people with CMT, caused by lack of muscle bulk, lack of movement and by poor circulation.

Summary of problems due to muscle wasting and sensory loss

<table>
<thead>
<tr>
<th>Complication</th>
<th>Why</th>
<th>Ways to prevent</th>
<th>Ways to treat</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ulcers, sores and burns</td>
<td>Not noticing damage to the hands or feet due to loss of sensation</td>
<td>Particular care with hot drinks and water bottles</td>
<td>Chiropody, medical intervention</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Checking feet daily</td>
<td></td>
</tr>
<tr>
<td>Very high arches</td>
<td>Calf muscle and Achilles tendon tighten (contracture)</td>
<td>Stretching; physiotherapy, Orthoses</td>
<td>Orthoses, surgery</td>
</tr>
<tr>
<td>Flat feet</td>
<td>Collapse of the foot arch</td>
<td>Stretching, physiotherapy</td>
<td>Podiatry, orthoses</td>
</tr>
<tr>
<td>Foot drop</td>
<td>Shin muscle weakens</td>
<td>Orthoses</td>
<td>Surgery (if severe)</td>
</tr>
<tr>
<td>Sprained or fractured ankles</td>
<td>Weakened muscles</td>
<td>Orthoses</td>
<td>Orthoses and surgery</td>
</tr>
<tr>
<td>Fatigue</td>
<td>Compensation by other muscles for CMT weakened muscles</td>
<td>Exercise, Orthoses, Energy conservation techniques</td>
<td>Coping strategies, orthoses, physiotherapy, OT</td>
</tr>
<tr>
<td>Loss of fine control in the hands</td>
<td>Muscle weakness</td>
<td>Hand orthoses; exercises &amp; stretching; alternative techniques suggested by OT</td>
<td>OT, physiotherapy</td>
</tr>
</tbody>
</table>

Read more about caring for your CMT in the chapters, Managing your CMT and Caring for common problems.
“Working to support those who are affected by Charcot-Marie-Tooth Disease, also known as Hereditary Motor and Sensory Neuropathy or Peroneal Muscular Atrophy”
Managing your CMT

Having any disability can be difficult, particularly if it is a long-term condition like CMT.

You may find that at different times in your life you face different problems, or you may find something you have been dealing with perfectly adequately for many years becomes more awkward.

However CMT affects you, somebody has probably dealt with that particular problem before and there is always an answer, technique, or method for dealing with it and living life to the full. It may take a bit of time to find the right solution for you, but do ask.

Remember you are not alone and that there are people who are on hand to offer you support when you need it. Apart from your friends and family, there are people with CMT who will be able to share their experiences and methods of overcoming the difficulties you may face (contact CMT United Kingdom). And of course there are a number of other voluntary organisations, not to mention professional carers on hand.

What you can do

It is your body. You are the person who best knows how your CMT affects you on a day-to-day basis. If you don’t take responsibility for it, no one else will. (Read more about taking control in the chapter, Coping with CMT.)

Professionals, like social workers and doctors for example, are there to offer their specialised advice and support on issues such as treatments, drugs and benefits, but they will only be with you for a few short hours. The rest of the time your well being, both physical and mental, is up to you.

You will understand how symptoms affect you, both mentally and physically. And you are the best person to keep tabs on any changes in your condition.

You can use your personal understanding to:

- feel more confident and in control of your life
- manage your condition and its treatment in partnership with healthcare professionals more equally
- help prevent further complications
- communicate effectively with professionals and share responsibility on treatment
- become more realistic about the impact of your disease on you and your family
- use your skills and knowledge to lead a fuller life.

Remember that getting the right information about CMT is important; helping you to make informed decisions for yourself. (Read about “Getting the right treatment” in this chapter.)

If you are interested in learning about how to take a more active role in your general health and well being, as well as managing your CMT, you may find that the NHS’ Expert Patient Programme helpful.
You and your GP

Your GP (general practitioner) is the “gate keeper” to many health services.

GPs are not specialists and it is quite possible that you will know far more about CMT than your GP, so do not expect her/him to understand the details. But your GP holds your medical records and can help you deal with some of your symptoms, referring you on to other specialists (physiotherapists, for example) when you need.

It is your GP’s role to look after your general health and make sure you see the specialists you need, so you get the right treatment, including:

- neurologist
- physiotherapist (with an interest in neuromuscular conditions)
- orthotist
- chiropodist
- occupational therapist
- orthopaedic surgeon.

“I don’t think my GP had seen anyone else with the condition. I know more than him.”
Lisa

Getting the right treatment

It is important that your CMT is treated by people who know about CMT. A consultant neurologist with a specialist interest in a peripheral neuropathy (which includes CMT) should oversee your treatment.

A neurologist specialises in understanding how your nerves work, what can go wrong with them, the likely course of any problems, and what treatments and care options should help. And, importantly, a neurologist can refer you to other specialists who can help with specific problems as needed.

In the real world it can be very difficult to see the right neurologist as there are just not enough doctors with this type of knowledge (there are only 10 neurologists in the UK with particular speciality in peripheral nerves). But there are 250 to 300 neurologists who will be able to give you very good care and who will be able to liaise with their specialist colleagues on any issues they are unclear about.

In effect you may go for long periods of time without seeing a helpful neurologist. In this case, do not despair as there are many other healthcare professionals who should be able to help. These include:

- Geneticist with an interest in neurology – they may not have the breadth of clinical knowledge, but will have a deep understanding about genes and the possible course of your CMT.
- Specialist paediatrician – a paediatrician is a doctor who specialises in child health and is probably the best person for a child with CMT to see.
Physiotherapist – preferably one with a special interest in neuromuscular conditions, but even if this is not possible, a physiotherapist can work with you to help your body function despite the effects of CMT.

Orthotist – an orthotist is someone who works with braces, supports and splints to help provide you with support and avoid some of the secondary complications of CMT.

Occupational therapist (OT) – OTs are expert problem solvers, helping you to continue to do day-to-day tasks despite any disability.

Podiatrist - (sometimes called chiropodist) are specialists in foot care.

Orthopaedic surgeon – a surgeon who specialises in surgery of the foot and ankle.

Rehabilitation specialist – is a doctor who works to get you back on your feet despite a recent health setback.

To help you, you can always get information from CMT United Kingdom or the British Peripheral Nerve Society or the Neuropathy Trust on which specialists are interested in neuropathies. Then ask your GP to be referred directly to your nearest specialist (although the waiting list for these specialists may be very long).

For more information on how these specialists can help you read the chapter, Treating your CMT.

“In Glasgow I always saw my neurologist right away. Down here [in Wales] there was no regular follow up – it was only when I decided that I wasn’t having this and complained that I got into a six to 12 month regular appointment. Now I’m usually seen once a year but I can phone and make an appointment. But I know the system and so many people don’t.” Margaret

General health tips for CMT

By taking care of your general health you are more likely to avoid problems with CMT and be able to lead a healthier and fuller life.

Keeping active

The human body is designed to be active. Long periods of rest or inactivity will actually damage your body. Read more about staying active and find out about some daily stretches and gentle exercises in the chapter, Exercise and stretching.

Keeping the weight off

Being overweight or obese – is bad for anyone’s health whether they have CMT or not. For people with CMT carrying extra weight can make matters worse for the following reasons:

- makes it more difficult to exercise or stay active
- puts more strain on already weakened muscles and joints
• increases the chance of back pain
• increases the risk of diabetes, which can lead to other neurological problems, particularly in the legs and feet
• puts more pressure on your heart and lungs
• cuts the amount of oxygen that is available to your body – to work effectively every cell needs a good supply of oxygen.

The best way to keep unnecessary weight off is to keep active, rather than trying to diet alone, perhaps taking up a sport such as swimming. Read more on exercise and eating healthily in the chapter, Exercise and stretching.

Alcohol

Alcohol was removed from the neurotoxic drug list in July 2004. While people with CMT generally suffer no ill effects from the moderate consumption of alcohol, they should be particularly mindful of the fact that alcohol affects balance and coordination. Heavy drinking or getting drunk is generally not recommended under any circumstances, as it can damage your nerves. This effect is likely to be exaggerated for people with CMT. If you have questions about alcohol and your health, consult your doctor.

Read more about sensible drinking in the chapter, Healthy eating.

Recreational drugs

As with alcohol abuse, recreational drugs are thought to have a damaging effect on the nervous system and this is likely to be worse for people with CMT.

Falls

CMT can increase the chance of you tripping and falling. Also, any inactivity because you are recovering from a fall will mean you lose stamina and strength. Although you should be able to recover this once you are up and about again, it will take time and effort. The danger is that any loss of stamina or strength will mean you are less likely to keep active, leading to more loss of stamina and strength. And so on.

Making sure you are wearing good shoes (with orthoses, if necessary), clearing any trip hazards from your home and taking particular care on uneven ground can all help you avoid falls.

A 15 point action plan to avoid falls:

1. Take regular exercise, even if this is only a short walk, to keep muscles strong (as possible!) and joints supple.
2. Fit easy grip handrails on both sides of the stairs. Avoid/minimise climbing stairs if it makes you feel unsafe.
3. Keep stairs and living areas well lit. Keep a torch by the bed.
4. Never leave objects which may be tripped over, on stairs or in walking areas. Avoid flexes and cables crossing walking areas.
Charcot-Marie-Tooth: A Practical Guide

5 Use non-slip rubber mats in the bath/shower. Fit a handrail near the bath/toilet. Avoid small rugs in the bathroom.
6 Replace worn rugs and carpets. Nail or tape down the edge of rugs to avoid slips and trips.
7 Minimise bending/climbing. Keep frequently used items on racks or in drawers at an easy level. Have a letter tray and rack for milk deliveries fitted.
8 If you must climb, use proper steps.
9 Get up from chairs/bed slowly. Blood pressure falls as you get up, and your body may take longer to adjust as you get older.
10 Avoid poorly fitting shoes or slippers (talk to a podiatrist).
11 Have regular eye tests. It is possible in many areas now to have an eye test in your own home.
12 Avoid clothes which may trip you such as trailing nightdresses.
13 Don’t rush to answer the telephone. Warn friends that it may take you longer to reach the telephone. Have an extension socket fitted upstairs.
14 If prescribed medication is making you feel dizzy, keep taking it, but consult your GP.
15 Keep rock salt/grit handy to put on external paths in cold weather.

Chilblains

Chilblains and cold extremities (especially feet!) are a problem for many people with CMT.

It is due to two factors:

1 Loss of muscle bulk and lack of movement, and therefore the normal heat generated by muscle activity is missing.
2 The affect that CMT can have on the autonomic nerves which control the blood vessels and therefore blood flow. The skin is patchy also due to the autonomic nerves controlling blood flow.

The autonomic nervous system is a system of nerves over which we have no direct control, and sometimes people call it the automatic nervous system. It is responsible for controlling blood pressure, and it does that, in part, by dilating and contracting the blood vessels, thereby directing blood to where it is needed.

In CMT and other disorders that affect the autonomic nervous system, this tends to cause reduced blood flow in the skin of the feet and shins. It is this that causes the coldness and blue discolouration. It can be uncomfortable, but doesn’t cause any serious problems.

Pressure sores

If you sit a lot, perhaps because you use a wheelchair, take extra precautions to avoid pressure sores. Make sure you use a decent pressure-relieving cushion (talk to your occupational therapist) and stretch out on a bed from time to time. A physiotherapist or occupational therapist can teach you how to do wheelchair push-ups at regular intervals throughout the day using your forearms (rather than your hands/wrists) to take the pressure off your bottom.
Getting the right information

Although there is not much information on CMT around (the most comprehensive source of information can be found on CMT United Kingdom’s website) with the advent of the internet it is now easier than ever before to find information on almost every ailment imaginable. But not all information is good quality and can, in some cases, be positively dangerous.

Ultimately it is up to you to make the decision whether to trust a source of information or not, but you may find the following points helpful.

- If the claims seem to be too good to be true, then they probably are.
- Beware of information that advises you to hand over a lot of cash.
- Be extremely careful of information that advises you to stop the medication prescribed by your doctor. (Do not do it without consulting your doctor first.)
- Be careful of information from other countries. Even information from the USA should be treated with caution as it may not be appropriate to the UK.
- Before making any decision on treatment, talk it over with your doctor.
- Use your common sense. Talk it over with friends and family. If you smell a rat, then do not let hope overcome reason.

Be careful of adverts and false claims

If you come across information that claims any ‘miracle’ cure or treatment, take extreme care. It could cost you a lot of money, damage your health or delay you getting the right treatment. The following are good signs that the information may be something to stay clear of.

- Phrases such as “scientific breakthrough”, “miraculous cure”, “exclusive product”, “secret formula” or “ancient ingredient”.
- Claims that the product can treat a wide range of problems.
- Anything that uses a lot of impressive-sounding medical terms. They may be more smoke and mirrors than substance, designed to cover up the lack of hard facts behind the claims.
- Personal stories of people who claim “amazing results”.
- Claims that the product is only available from one source and for a limited time only.
- Money back guarantees – especially if you are sending the money abroad or to a PO Box.
- Adverts that fail to list the company’s name, postal address or other contact information.

The best advice is to make sure the information is evidenced based and that you talk it over with your healthcare team.

Evidenced based means that the information has been reviewed in light of the latest scientific findings in medical journals.
Some of the best health information sources on the internet are:

- **Besttreatments**: (www.besttreatments.co.uk) – probably the best evidence based source of information on what works and what does not, using information from the British Medical Journal. Very easy to understand it covers over 200 conditions. And also give a clear explanation of what evidence based information is.

- **Prodigy**: (http://www.prodigy.nhs.uk/PatientInformation/) – Prodigy is the source of clinical knowledge put together by the NHS. It has a number of patient information leaflets, as well as giving you access to the information that a lot of doctors will use. Excellent.

- **MedlinePlus**: (http://medlineplus.gov/) – the official US government health information site. An excellent and easy to use source of information on a massive range of information. It also gives links to other trusted, but US only, sources of health information.

- **NHSOnline**: (http://www.nhsdirect.nhs.uk/) – gives a good overview of a range of conditions, but probably not as detailed or useful as MedlinePlus.

If you want to delve deeper the following portals are very good, but may take a bit more effort:

- **OMNI**: (http://130.88.13.169/) – a free catalogue of hand-selected and evaluated Internet resources in Health and Medicine, put together by information specialists at the University of Nottingham.

- **Cochrane Collaboration**: (http://www.cochrane.org/index0.htm) - is an international not-for-profit organisation, providing up-to-date information about the effects of health care. You can read plain English consumer summaries of the reviews at www.informedhealthonline.org

- **PubMed**: (http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?db=PubMed) – this is probably the most comprehensive source of clinical papers available. It can be very heavy going, but if you want to find the original research, this is a good place to start.

For something slightly off the beaten track, you may find Bad Science an interesting read. Set up by a single doctor, it aims to debunk some of the mumbo jumbo that passes itself off as health information: http://www.badscience.net/. Enjoy.
“Working to support those who are affected by Charcot-Marie-Tooth Disease, also known as Hereditary Motor and Sensory Neuropathy or Peroneal Muscular Atrophy”
Exercise and stretching

Stretching and exercise play a very important part in managing CMT and helping to prevent or ease the complications of the condition.

Exercise

So should people with CMT exercise? Yes, because exercise is more important for people with CMT than for friends and family without the condition.

The benefits include:

- Exercise, combined with a healthy diet, is the main way of controlling body weight, which is important as being overweight or obese puts extra pressure on already weakened muscles and joints.
- Exercise is a great way of tackling fatigue — by exercising regularly you will find you are able to carry on your day-to-day tasks without getting so tired.
- Muscles are prone to becoming weak without use, making it harder to get around — exercise keeps muscles strong.
- Exercise helps with balance and posture.

There is no universal rule of thumb about which exercises or activities will suit you. Do not let your worries about your CMT restrict you from trying different things. Just bear the following points in mind.

- If you are doing something that causes you any pain, stop immediately.
- Get to know your own limitations and understand the difference between getting naturally tired from exercise (a good thing) and excessive fatigue (a bad thing).
- See your physiotherapist or your gym instructor and talk to them about what would suit you.
- If one exercise does not suit you, try another (eg if walking doesn’t work, try cycling).

Which one for me?

Exercise does not mean you have to go to the gym. You can achieve aerobic benefits from changes to your daily routine, such as walking a bit further or climbing the stairs instead of taking the lift or standing on the escalator.

The key to getting the right amount of daily exercise is to make physical activity part of your normal routine. For example, getting off the bus one stop early, walking up the last flight of stairs, cycling to the station, walking to the shops or school are all good ways of reaching the 30 minute daily minimum.

A good form of exercise is ‘aerobic’ exercise that works your heart and lungs, for example, walking, swimming and cycling. All of these help to increase endurance, reduce fatigue, improve mood and increase your ability to do day-to-day activities.
“The Sports Centre also has a pool and after a gap of 20 years I can still swim – this is excellent as you feel so confident in water. You can also use the Health facilities (sauna and steam room) so there is no excuse for being a bit overweight and blame it on lack of exercise!” Ian

To help with your sense of balance, exercise that strengthens your posture muscles is important, for example, yoga, Pilates and Tai Chi. These forms of exercise will also train your balance responses.

“About six months ago I joined the gym and now go the Midland Sports Centre for the Disabled. Initially the sight of all those exercise machines frightened the life out of me. But once you’ve worked out a programme it’s surprising what you can do and how much confidence it can give you. Walking still frightens the life out of me; however once I start on a recumbent bike there is no stopping me.” Ian

**Stretching**

If you have CMT, then daily stretches are vital in order for you to keep flexible in your hands and ankles. They are a vital way of helping to prevent your muscles tightening and shortening, which can lead to loss of movement and pain and deformity in your joints, particularly in the hands, feet and ankles.

**Hands**

In the hands, the small muscles in the palm weaken first. Over time the bigger muscles in the forearm take over hand movements, allowing you to continue with day to day tasks. The result is that the large forearm muscles overpower the smaller hand muscles setting up an imbalance. Eventually, this imbalance can be observed in the resulting flat palm, flattened knuckles and bent fingers.

Due to the number of joints, the human hand has the ability to make complex movements. But, if some of those joints become stiff, you are likely to find that you lose the ability to do everyday manipulation tasks. In CMT, weakness of the hands makes doing everyday tasks more difficult. This becomes doubly difficult if the joints stiffen up as well. Daily stretching will ensure that the joints remain supple and slow down the development of hand deformities. It is important that you get into the habit of doing these stretches BEFORE deformity and stiffness develops.

** Ankles**

In the lower leg, the muscles on the shin (front of your lower legs) pull the foot and toes up, whereas the calf muscles (back of your lower legs) point the toes. The calf muscles attach onto the heel bone via the Achilles tendon (at the very back of your ankle).

In CMT the muscles on the shin tend to get weaker first which results in a “drop foot”. The stronger calf muscles overpower the weaker shin muscles setting up an imbalance between the two. Because of this the calf muscle will gradually get shorter and stiffer, as will the Achilles tendon, further increasing
the foot drop. Because the ankle needs to be at a right angle for the toes to clear the floor when walking, the result is an increased chance of tripping when walking, increased difficulty getting the heel to the floor and a greater chance of sprained ankles.

Stretching the calf daily is something that everybody with CMT should get into the habit of doing to keep the calf muscles lengthened and slow down the development of the deformity. These stretches are easy to perform and can be incorporated into your daily routine, for example during your morning shower when your muscles are warm and relaxed (just be careful not to slip) or standing up to a work surface while waiting for the kettle to boil.

Orthoses and stretching

Orthoses can play an important role in helping to maintain flexibility, joint range and prevent the muscles tightening and shortening. This can be by means of insoles which may be thicker on the outside of the foot than the inside so the foot is stretched when weight is put on it. If stretching is particularly difficult, night splints are sometimes used to stretch the calf by pulling toes up and strapping the heel down and at the same time holding the ankle straight. Similar orthoses used through the day can provide a very effective stretch as well as holding the foot in the best position for walking.

Daily stretches and exercises for people with CMT

Remember that these exercises are only a general guide. It is strongly recommended that you consult a physiotherapist to put together an exercise programme tailored to your individual needs.

If you experience any pain or difficulty doing these exercises, stop immediately and seek advice from your family doctor or physiotherapist.

Stretches

1. Calf stretch:

- With finger tips hold onto a wall or work surface.
- Keep your head up and back straight.
- Place one foot forward and one foot back with the back foot and heel fully on the floor. Make sure your toes are pointing forward.
- Let your front knee bend but keep your back knee straight.
- You should feel a stretch in the calf.
- Hold still for 20 to 30 seconds. Repeat three times and then swap legs.
2  Lower calf stretch:
   • Get in the same position as above.
   • Step your back leg forward so the toes are in line
     with the heel of the other foot.
   • Letting both knees bend, sink down with your weight
     on your back leg.
   • The stretch will not be as strong as with exercise 1.
   • Hold still for 20 to 30 seconds. Repeat three times
     and then swap legs.

3  Hip stretch:
   • Sit on the end of your bed with your feet on the floor.
   • Lie back, keeping your legs over the edge and
     feet on the floor.
   • Lift one leg and hug the knee to the chest.
   • Hold still for 20 to 30 seconds. Repeat three times
     and then swap legs.
   • Do not continue with this exercise if you experience back pain.

4  Hand stretch:
   • Sit up to a table and place both hands on top.
   • Keeping the fingers straight, bend forward at the knuckles.
   • You may use your other hand to help but don’t force the position. You should feel a
     stretch, NOT pain.
   • Hold for 20 to 30 seconds. Repeat three times and then swap hands.
5  Finger stretch:
   - Keeping you knuckles bent forward, use your other hand to straighten the ends of your fingers.
   - It is VERY important that you avoid bending the knuckles back when stretching the fingers.
   - Hold for 20 to 30 seconds. Repeat three times and then swap hands.

Exercises for posture

6  Standing posture:
   - Stand with your back against a wall. Make sure the back of your shoulders and head are against the wall.
   - Hold this position and then slowly slide the back of your head up the wall to stretch the back of the neck.
   - Repeat the movement 10 times.

7  Sitting posture:
   - Sit on the edge of a firm chair. Look straight ahead.
   - Slowly sit up as straight as possible then slowly slouch down.
   - Repeat the movement 10 times.
   - A good sitting posture is the middle position between these two movements.
Strengthening exercises

8 Upper legs:
- Sit on the edge of your bed or a dining chair.
- Keep feet hip width apart. Keep your arms by your side.
- Stand up fully then slowly sit down.
- Repeat the movement 10 times.

9 Trunk and hip:
- Lie on your back with your knees bent and feet firmly on the floor.
- Lightly pull in your navel. With one hand, feel the arch in your lower back. Squash that arch into the floor using your hand for feedback. This is called a pelvic tilt.
- Hold the pelvic tilt position, and then lift your bottom off the floor as far as is comfortable, without straining your neck and shoulders.
- Repeat the movement 10 times.
- If you find raising your bottom too difficult or you can’t hold the pelvic tilt, just practise the pelvic tilt movement with your bottom on the floor.

Balance exercise

10 Standing balance:
- Stand near a work surface or wall.
- Stand with your feet together keeping an upright posture. Hold for as long as possible using fingertip support on the work surface/wall as required.
- If you are able to, keep this position and turn your head right and left. Repeat the movement 10 times.
- If this is easy, extend your arms and rotate them around to the right and left. Repeat the movement 10 times.
Before exercising

Before you start to do any exercises, think about the following:

- Have you talked to your physiotherapist, family doctor or gym instructor about the right exercises and level for you?
- If needed, have you considered orthoses? The right one can make exercising more efficient and enjoyable.
- Remember to pace yourself – don’t overdo it. And if you have worked hard one day, think about relaxing the next.
- Put together an exercise plan – build up slowly so as not to injure yourself. (The 10% rule is a good one to stick to – aim to increase your exercise levels by no more than 10% to 15% each week.)

General benefits of exercise

Physical activity not only contributes to well-being, but is also essential for good health.

It is not just your physical health that can benefit from physical activity; your mental health can benefit as well.

All in all, physical activity is one of the best tonics available. And yet in the past 25 years we have done less and less of it – with walking and cycling falling by a quarter. And despite what we say, this decline in physical activity has less to do with the ‘modern world’, but more to do with our attitudes. We are less inclined to do activities that take effort and prefer to drive a car, or stand on the escalators.

Benefits of regular physical activity

- Overall reduces the risk of premature death by up to a third.
- Reduces the risk of coronary heart disease, stroke and type 2 diabetes by up to half.
- Combined with a balanced diet, it is the best way of keeping to healthy weight.
- Reduces the risk of osteoporosis.
- Helps treat mild to moderate depression.
- Helps people feel better and helps to beat stress and give you a good night sleep.
- Helps to keep our mental faculties sharp, particularly into old age.
- For older people, regular physical activity reduces your risk of falling and of being seriously injured.)
Questions and answers

I’ve heard that over exercising (overwork) can actually be harmful. Is this true?
Research into conditions such as CMT has shown that there is no increase in weakness with low to moderate intensity exercise, with the benefits far outweighing the risks. You can reduce your risks by:

- exercising at low to moderate levels, as advised by your gym instructor or physiotherapist
- not exercising to exhaustion
- recognising that if your muscles are sore for longer than 48 hours after the exercise that you have probably worked too hard.

Will any muscle that I build up rapidly waste away due to CMT?
Muscle wasting associated with CMT is slow and will tend to be at the extremities (hands and lower legs). If, for example, you built up your upper arm strength through weight training, we would not expect you to lose this effect due to the CMT if you have the more common types of CMT. However do remember that any muscle will lose strength if you stop exercising it. If you don’t use it, you lose it.

Are there any parts of my body that I shouldn’t exercise?
There are no parts of the body to avoid, although it may appear from research studies that there is little benefit to trying to strengthen severely weak muscles. However, physiotherapists would recommend general exercise as well as (or instead of) weight training, particularly exercise that strengthens muscles important for good posture and balance, such as yoga, Pilates and Tai Chi.

Am I able to do weight training?
In the past there has been some argument whether people with CMT should do any weight training, but studies show that low to moderate intensity weight training improves upper and lower body strength with few injuries. The key is not to overdo it, probably using lighter weights with between eight and 15 repetitions.

Before starting any weight training – ideally before you start any exercise – you should talk to a gym instructor or physiotherapist to help you start at the right level. Advice from a professional will ensure that you perform the exercise in an optimal and safe position for your muscles to work effectively. This is particularly important if you have any reduced sensation in your arms or legs.

How much is a good thing?
The health and well-being benefits that we gain from exercise only last for a short time. If you stop exercising you will quickly begin to lose the health benefits. This is one good reason that we need to exercise regularly – doing a little often – rather than doing a lot of exercise now and then.
In order to benefit from exercise the general recommendation is that we all need at least 30 minutes of moderate intensity physical activity five times or more a week. But that doesn’t mean it can’t do you any good if you can’t achieve that much. If you are at a lower level, and try to increase your level of activity over time, it should still be beneficial. Even chair based exercises can help.

You may find it helpful to rate how hard you are working on a scale such as the one below (the Borg scale):

<table>
<thead>
<tr>
<th>Level</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>6, 7, 8</td>
<td>Very, very light</td>
</tr>
<tr>
<td>9, 10</td>
<td>Very light</td>
</tr>
<tr>
<td>11, 12</td>
<td>Fairly light</td>
</tr>
<tr>
<td>13, 14</td>
<td>Somewhat hard</td>
</tr>
<tr>
<td>15, 16</td>
<td>Hard</td>
</tr>
<tr>
<td>17, 18</td>
<td>Very Hard</td>
</tr>
<tr>
<td>19, 20</td>
<td>Very, very hard</td>
</tr>
</tbody>
</table>

The words describe how hard you feel you are working when exercising. Light exercise would be working at levels 11-12. Moderate exercise would be levels 13-14.

In general the amount of exercise that we need varies with our age. Adults need between 45 to 60 minutes of moderate exercise every day to prevent obesity (you do not need to do this all at once, but over the course of the whole day). Children and young people should be getting 60 minutes of moderate exercise over the day, every day, as well as doing two activities a week of higher impact exercise – such as running or sports - in order to develop bone health and muscle strength and flexibility.

Older people should exercise as often as younger adults but with a focus on strengthening and balance exercises. As we get older we slowly lose muscle and our ability to balance is less effective. (Studies have shown that regular physical activity reduces your risk of falling and of being seriously injured.)

**It’s all very well going on about walking, but I find it tough. What would you advise?**

If you find walking difficult, then you may find that swimming, or water-based exercise, is the answer. Even if you can’t swim! This is because water-based exercises take the pressure off your joints. Talk to your local leisure centre about activities appropriate for you.
“Working to support those who are affected by Charcot-Marie-Tooth Disease, also known as Hereditary Motor and Sensory Neuropathy or Peroneal Muscular Atrophy”
Healthy eating

You need food to power your body, giving it energy and the material it needs to grow and repair itself. When you eat food it is broken down in your stomach and intestine (gut) and four main nutrients are extracted:

- carbohydrates
- fat
- protein
- vitamins and minerals.

A well-balanced diet is one that is low in fat, sugar and salt and high in fibre. (Fibre is the part of fruit, vegetables or cereal that passes through your body undigested and helps to prevent constipation.) Your diet should also contain enough protein and a wide range of vitamins and minerals.

Healthy eating means getting the right balance between different foods. For many people this means a change towards eating more fruit, vegetables and higher fibre starchy foods.

The five food groups

There are five food groups, each one providing a different combination of the three essential nutrients – carbohydrates, fat and protein – as well as vitamins and minerals.

1 Carbohydrates, including bread, cereal, potatoes, rice, pasta, noodles, chapatti (good for slow release energy)

Choose one of these foods at each meal – they release energy slowly into the bloodstream. High-fibre versions will keep you feeling fuller for longer.

The only fat we get from these foods is the fat we eat with them, such as butter on bread or potatoes, full cream milk on cereal. Watch out for these added fats – opt for low fat versions instead.

2 Fruit and vegetables (high in fibre, vitamins and minerals)

Aim for five portions a day. Fruit and vegetables provide essential vitamins and fibre, helping to protect the body against heart disease and some forms of cancer. A ‘portion’ could be one of the following:

- one glass of orange juice
- two tablespoons of vegetables – raw, cooked, frozen or canned
- one dessert bowl of salad
- one apple, orange, banana or similar sized fruit
- two small fruits – plums, apricots
• a small handful of grapes or cherries
• a half-tin of tinned fruit in natural juice or dessert bowl of stewed fruit.

3. Dairy, including milk, yoghurt, eggs and cheese (high in protein, good for calcium for healthy bones and teeth)

You can choose low-fat versions to help you keep to a healthy weight.

4. Protein, including meat, poultry, fish, nuts, pulses, beans, tofu, cheese

Beans and pulses also provide useful fibre. Try to have two helpings of protein-rich foods a day.

Red meat is higher in fat than chicken and fish but is a very good source of iron so should be included in the diet at least twice a week. Good sources of iron for vegetarians include wholegrain cereals and flours, leafy green vegetables, blackstrap molasses, pulses such as lentils and kidney beans.

5. High-calorie foods, such as fried and sugary foods (high in fat and sugar)

These foods include cakes, biscuits, chocolate, crisps, fried foods and pastries which are high in both fat and calories. Where possible choose low-fat, low-calorie versions or enjoy these occasionally as a treat.

It is important to choose a variety of foods from the first four groups every day to get a wide range of nutrients. For most people, food from the fifth group should only be eaten as treats. If you are underweight and need more calories, you may be advised to eat more of these foods.

Food for energy, repair and growth

Different foods give us different benefits. If you are looking for a healthy energy boost, try eating a banana – it is full of carbohydrate, fibre and almost no fat. Proteins, found in meat and pulses for example, are needed to help build and repair muscles.

For energy

Carbohydrate

Carbohydrate comes from starch and sugar and is found in bread, potatoes, rice, pasta, cereals, fruit and sweets. Carbohydrate is broken down in the liver to glucose, a form of sugar, which is used to make energy.

Any glucose not used immediately for energy is stored as glycogen in the liver and in the muscles. When your body needs extra energy – when running for a bus, for example – the glycogen is quickly converted back to glucose.

As well as storing glycogen, the liver helps control the level of glucose in the blood.
**Fat**

Fat comes from butter, cheese, oil, animal fat and from many “hidden” sources, for example, biscuits, pastry, crisps and cakes.

Fat can be used as a long-term energy store. It also provides the fat-soluble vitamins A, D, E and K and essential fatty acids.

**For repair and growth**

**Protein**

Protein comes from foods such as meat, fish, eggs, nuts, pulses and dairy products. It is made up of units called amino acids and when these reach the liver they provide building blocks to make cells and tissues throughout the body.

**Vitamins and minerals**

Your body carries out millions of chemical reactions every day. To do this it needs a mix of vitamins and minerals in addition to the essential nutrients, carbohydrates, fat and protein.

**Supplements**

Most people can get all the vitamins and minerals they need by choosing a variety of foods from a normal, healthy, well-balanced diet. Taking supplements cannot mimic the positive effects of food and fluids.

Mega doses of supplements maybe harmful. People with CMT should be particularly cautious of mega doses of vitamins A, B6 (Pyridoxine) and D. (A megadose is defined as ten times the recommended daily allowance (RDA). The RDA for vitamin A = 800 micrograms; B6 = 2mg; D = 5mg.)

Get advice from a dietitian before taking regular supplements. This is particularly true if you are thinking of taking a combination of supplements.

There is no evidence to recommend any particular supplements for people with CMT.

**Keeping to a healthy weight**

To sustain a healthy weight, you need to balance the amount of food you eat with the energy you need. If you eat fewer calories than your body needs (especially if you are physically active) you will lose weight. If you eat more than you need, your weight will increase.

The amount of energy we need differs according to our sex, age, weight and the amount of physical activity we take. For example, a small elderly woman will need less food than a young, active man.

If you have been ill or have lost a lot of weight you may not feel like eating and keeping to a well-balanced diet may be difficult. Try to keep eating as much as you can and, if necessary, ask your doctor or dietitian for advice.
Calories

In the same way that we measure distance in centimetres (cm), the energy in food is measured in units of calories (kcal). For example an apple will have about 50 calories and a Mars chocolate bar will have about 300 calories.

As a basic rule of thumb, adult men are advised to eat about 2,500 calories every day and women 2,000 a day. If you are not active you will probably need fewer calories every day.

Dealing with too much weight

The following factors all contribute to being overweight:

- less mobility
- fatigue
- convenience foods
- comfort eating
- drinking alcohol (a glass of wine has between 80 and 100 calories, and a pint of beer between 250 to 300 calories)
- lack of exercise.

There is no easy answer to help you keep the weight off, but the following tips will all help:

- eat a healthy balanced diet
- do not eat large portion sizes
- exercise as you are able (read the chapter, Exercise and stretching)
- plan your meals
- do not go to the shops when hungry and keep to a shopping list
- avoid having tempting foods at home
- choose healthy and low calorie snacks, such as fruit, vegetables, plain cereal bars and plain pop corn (do not add salt)
- choose healthy eating options – read the labels carefully.

If you’re having trouble with keeping weight off, you can find some good information at the British Dietetic Association’s Weightwise website.

Being underweight

Being underweight should be avoided as much as being overweight. Remember, it is not always easy to recognise that you are underweight (our friends and family are better at pointing out when we are carrying a few extra pounds).

Some of the problems associated with being underweight include:

- becoming physically weaker (fatigue)
- less able to fight infections
Charcot-Marie-Tooth: A Practical Guide

- less mobile
- depression

If you are underweight you should be referred to a registered dietitian. And the healthy eating tips are a little different, including:

- use full fat and sugar products
- try to eat little and often meals and snacks
- fortify meals.

What about alcohol?

Alcohol is a toxin (poison) and can damage muscle and nerves. But, if you are generally healthy, not taking certain medication and drink sensibly, then alcohol should not harm you.

But what is sensible drinking? The answer depends on whether you are a man or a woman. (The limits are different for men and women mainly because men are bigger and so can, in general, safely handle a little more alcohol than women.)

**Women**

- A maximum of 2 to 3 units of alcohol in a single day (no more than 14 units in a week)
- A minimum of two days a week without any alcohol.

**Men**

- A maximum of 3 to 4 units of alcohol in a single day (no more than 21 units in a week)
- A minimum of two days a week without any alcohol.

Units have been used for 25 years in the UK to describe amounts of alcohol. In the past, a unit could be identified as a drink – one unit (8 grams of pure alcohol) was a measure of spirits, half a pint of beer or a glass of wine. However, the alcohol content (abv) of drinks and the standard measures of drinks served have increased over time. Spirits used to be served in 25ml quantities – now it is often 35ml; wine used to be served in 125ml quantities - today it is usually 175ml or 250ml glasses; the strength of lager used to be 3.5% abv – now it is commonly 5% abv.

The abv (shown on the bottle, box or can) tells you how many units there are in a litre, ie 6% abv means there are 6 units in a litre. If you drink half a litre (500ml) – just under a pint – of beer of this strength – then you have had 3 units.
Calculating units of alcohol

An accurate way of calculating how many units you are drinking is to multiply the abv figure by the size of your drink. For example, a typical can of beer these days is 440ml, at 5% abv strength. Therefore: 440 x 5 = 2,200; divide this by 1,000 = 2.2 units.

You can use this formula to work out other drinks:

- a standard glass of wine (175ml) at 12% abv is 2.1 units and a large 250ml glass is 3 units
- one measure 35ml of spirits (40% abv) is 1.4 units
- a pint of low strength (3.5 - 4% abv) beer or lager is 2.3 units
- a pint of regular cider or lager (alcohol 5% abv) is 3.4 units
- a standard measure of port or sherry (50ml) is 1 unit.
Treating your CMT

In a nutshell

At present, there is no specific treatment or cure for the underlying genetic cause of CMT. Neither are there any drugs to stop or reverse the damage caused to the peripheral nerves.

But, although there is no cure for CMT, there are a number of treatments available. These can slow the development, or ease, some of the secondary complications linked to CMT, greatly improving your quality of life.

Good general advice is to look for the least invasive way to treat your problems using a combination of the following:

1. **Self-management**
   - Stretching and exercise.
   - Healthy eating.

2. **Physical therapies**
   - Physiotherapy.
   - Orthotics.
   - Occupational therapy.
   - Podiatry.

3. **Surgery**

A consultant neurologist in partnership with your GP (general practitioner) should oversee your treatment.

The aim of all of these treatment options is two fold:

1. **Prevent unnecessary problems**

   Through a combination of exercise, healthy eating, physiotherapy, occupational therapy and orthotics, the aim is to stop the primary symptoms of CMT developing into major secondary problems. Or limiting the way you live your life by causing problems such as having difficulty walking, back strain, difficulty handling things or uncomfortable ulcers on your feet.

2. **Treat any problems quickly**

   If secondary problems do develop, such as severely arched feet or weak ankles, then orthotics, surgery, pain management, counselling and other adaptations or aids (eg wheelchairs) may be helpful.

Because CMT is an ongoing condition, it is useful to be aware of the mechanics behind the condition to help prevent secondary complications developing. As always, prevention is better than treatment.

“My dad used to say, ‘I’m in good shape for the shape that I am’,” Susan.
Physiotherapy

Physiotherapists are specialists in how the human body functions and moves. They are specially trained to understand how the joints, muscles, tendons and ligaments of the body work together and to spot any problems.

If you have not had a session with a physiotherapist, you should ask your doctor to refer you.

Although physiotherapists (often called physios) cannot stop the progression of CMT, they can often spot a potential problem – like any stress you may be putting on other joints like your knees or hips – early enough so that it can be treated and, possibly, prevented. This can help prevent secondary weakness causing other problems.

To see a physiotherapist, try one of the following:

1. Get a referral from your GP or consultant neurologist.
2. Go private.

If you do decide to go privately contact the Chartered Society of Physiotherapy for a physiotherapist near you. And remember, ideally you should see a physiotherapist who is a specialist in neuromuscular conditions, especially CMT.

Orthotics

Orthotics is a specialist care area dealing with:

- splinting
- supports
- braces
- insoles
- special footwear.

All of which have been designed to optimise position and support as appropriate, to help you avoid some of the secondary complications of CMT.

Orthoses used to be cumbersome, large, unattractive and, often quite uncomfortable to wear. Now, however, the technology behind them offers lightweight, flexible, often unobtrusive and comfortable supports that can add substantially to your quality of life. (Although some people still do best in what may be described as bulky, less attractive orthoses.)

The best person to see regarding selection and provision of the most appropriate orthosis for you is the orthotist. An orthotist is a qualified, registered health professional, specialising in assessing people and their particular need for orthoses. Everyone with CMT has different needs and the orthotist will take care that you receive the right support for you.

“Soon after I left work I needed braces on my leg to walk any distance. And I have now got splints for my hands which I use if I get weak.”
There is a large range of different orthoses, each one specially designed to help overcome a particular problem associated with CMT. (All orthoses should be fitted to you personally to take into account your own needs. If they are not right for you, or you have not been offered an orthosis, then you may have to push for it.) Because orthotics often help change the way you move – allowing a more natural motion – it is best to work with your physiotherapist when starting with a new orthosis.

A new orthosis may not wear comfortably. If it is uncomfortable go back to your orthotist to change how it fits. An orthosis should not cause any discomfort. (If you are able to see a physiotherapist, he or she should be able to help you adapt your movement to the new orthosis.)

It is worth bearing in mind nothing needs to be forever. While it may be nice to be straighter, safer and more efficient when walking to work, it is quite acceptable to discard your orthosis for the sake of elegance for an evening out. Just accept you will not do so much walking. Similarly you may manage very well within the safe environment of your office or home but it may be useful to have some orthotic support when out shopping or on a golf course. In addition, because a splint can stop you using some of your muscles, it is best not to wear them the whole day – so as to maintain the strength and stamina you still have.

Some of the orthoses you might hear about include:

- FO - Foot Orthosis (insoles)
- FFO - Functional Foot Orthosis (insoles)
- SMO Supra-malleolar Orthosis (ankle brace)
- AFO - Ankle Foot Orthosis
- DAFO - Dynamic Ankle Foot Orthosis
- SAFO - Silicone Ankle Foot Orthosis
- JAFO - Jointed Ankle Foot Orthosis
- KO - Knee Orthosis
- KAFO - Knee Ankle Foot Orthosis
- HKAFO - Hip Knee Ankle Foot Orthosis.

Background to common orthoses

There are a vast range of different types of orthoses – coming in different materials, colour and size – so if the pictures and descriptions on the following pages do not match what you have, do not be surprised. The key point to remember is that your orthosis should work for you. If it does not, talk to the orthotist or physiotherapist.

Insoles

Insoles may offer correction and stability by trying to change position of your feet or accommodating the existing shape and thereby offering pain relief. Insoles may have wedges and pads to try and...
stretch, correct and realign the feet and ankles or may be moulded to the existing shape of the feet to try and offer comfort and pressure distribution to collapsed or deformed feet.

For example, you may be offered an arch support for flat feet, to help maintain a better ankle position, reducing the chance of joint strain. Whereas an insole raised on one side may help stretch your foot and help prevent your foot leaning over to one side.

If your foot is badly deformed, special footwear can be designed to give you maximum stability and comfort if wearing of normal footwear is no longer possible. But, where possible, your own shoes can be adapted in various ways to tip the foot or provide stability as needed.

An ankle brace which supports the dynamic arches of the foot and creates a more stable base. Useful in a mobile foot but can be uncomfortable and intolerable on a rigid foot.

Ankle braces

Ankle braces may offer some support to prevent too much movement if you ankles are weak. They may also help to counteract foot drop in mild cases. Bespoke moulded orthoses may provide more control and offer a degree of correction and stretch.

Two types of ankle brace, the one on the left gives some degree of support to sideway collapse of the ankle. The one on the right offers more support and provides a small amount of resistance to footdrop.
Ankle foot orthoses (AFOs)

AFOs come under the foot, and part way up the back of the calf, usually strapping at the ankle. Calf length AFOs may be made of plastic which may be of flexible design and can give you a lot of help to counter foot drop. Alternatively, a more rigid design can help provide improved alignment of your joints, not only of the feet and ankles but have an effect on knees, hips and general posture.

Metal AFOs, or callipers, may be of use when more pressure is needed to realign a limb. Generally, these offer correction by leather straps rather than hard plastic. Although heavier, they can be more comfortable and tolerable.

Overall, made to measure AFOs fit better as they have been designed fit the exact shape of your limbs.

AFOs come as either full foot length or three quarter foot length, allowing you to continue bending your foot during walking. Some people find full length hard to get on with and say the three quarter length makes walking and running easier.

Rigid AFOs made to plaster casts which can influence alignment of knee, hip and trunk by rigidly fixing the ankle in an optimum position.

Three types of readymade flexible footdrop splints (AFOs) which are designed to fit in the shoe (although not necessarily your usual size), they are more effective than the ankle braces for controlling footdrop.
Knee ankle foot orthoses (KAFOs)

Knee ankle foot orthoses (KAFOs) or full length callipers can be useful for knee and hip weakness, they generally have a knee lock so the knee can be held straight and stable for standing and walking but released for bending for sitting. These are generally combined with the previously mentioned designs for ankle support and can be of metal or plastic. A lot of work is currently being done on “intelligent” KAFOs that allow knee bend when required in walking but stability when at rest.

If you have difficulty working out whether you are wearing an orthosis or an orthotic, the following quote from Paul Charlton, CMT specialist orthotist, should help:

“The specialist working in the field of orthotics, who may fit an orthosis from a selection of orthoses, is known as an orthotist.”

For more information about how orthoses can help you read the chapter, Caring for common problems.

Occupational therapy

Occupational therapists (OT) are expert problem solvers. An OT can help you carry on doing day-to-day tasks, despite any problems you may have due to illness or disability. They focus on the tasks that are important to you, whether it is so that you can continue to dress yourself or use a computer.

Central to OT is activity analysis. An OT will carry out an assessment and will break down the activities you are having difficulty with into their single parts. They will then work with you to tailor an individual solution so that you can carry on that task independently. These tasks could be in the area of:

- work
- home
- self-care
- leisure.

Often people with CMT find it most helpful to see an OT if their physical ability changes. For example, if their CMT has made it more difficult to do things that, until then they had been able to do with little problem, such as opening a jam jar.

OTs work in various settings including community teams, social services and hospitals. If you would like to see an OT, talk to your GP, local social services (social care in Scotland) or your consultant. They can refer you to the most appropriate service. Or you can arrange to see a private OT by contacting Occupational Therapists in Independent Practice.
Generally, an OT based in a hospital or primary care setting will focus on rehabilitation and coping strategies. And an OT based in social services will focus on home modifications and equipment.

Depending on where you live, the equipment recommended by an OT might not be available on the NHS or through social service and may have to be bought privately. If this is the case, and you have difficulty paying, the OT may be able to advise you on charitable trusts that may help financially (or can refer you to a Social Worker for this).

To see an OT you can do so in one of four ways:

1. Referral from your GP or neurologist.
2. Referral from another healthcare specialist, such as a physiotherapist.
3. Go private.
4. Through social services, where you can self refer.

If you are interested in going privately, you can contact Occupational Therapists in Independent Practice. (See the chapter, Useful organisations.)

**Podiatry (chiropody)**

Podiatrists are specialists in foot care. They diagnose, treat and manage a variety of foot conditions and will be able to assess your nerve function, circulation and foot function, giving you information regarding foot care.

A podiatrist can treat hard skin to prevent it breaking open and treat any areas of ulceration. They may provide insoles or work with orthotists in the provision of specialist footwear.

Surgical Specialist Podiatrists may be involved with foot surgery.

Podiatrists used to be called chiropodists in the UK and the two names are generally used synonymously. In the UK these practitioners are now registered with the Health Professions Council, the independent, UK-wide health regulator.

You can get to see a podiatrist in one of two ways:

1. On the NHS through a referral by your GP
2. Privately. You can find a private podiatrist through the Health Professions Council. (Find their details in the chapter, Useful organisations.)

If you can get a referral, or can afford a private podiatrist, it is a good idea for everyone with CMT to see a podiatrist once a year for a check up.
Surgery

Although foot surgery is fairly common in people with CMT, it should usually be carried out only when less invasive measures, such as orthoses, have failed. Even minor surgery has some risks, whether it is from the surgery itself, the anaesthesia or the recovery period.

The aim of surgery is to:

- help you walk with the entire lower surface of the foot on the ground
- reduce pain
- improve balance and agility
- halt the development of deformity.

Some things to bear in mind before going ahead with any surgery:

- Make sure that the surgeon doing your operation knows all about CMT. Ideally the surgeon should be an orthopaedic surgeon, meaning that they specialise in ankle and foot surgery. If in doubt talk to CMT United Kingdom.
- Take a friend along with you when you discuss why you need surgery – they can help take notes and remind you to ask all your pre-agreed questions.
- Find out how likely it is for the operation to succeed (and chances for failure), as well as the success rates of the individual surgeon who will be operating on you.
- Make sure you find out how long you will take to recover. Will you be able to get around or work during this time? And will you need help from a physiotherapist to recover? If so, make sure that one is lined up for you.

Operations range from straightening the toes, particularly the big toe, to fairly major surgery on the ankle joint. Terms you may hear include:

**Tendon transfers** – moving a working tendon and muscle group and attaching the end of the tendon to a new place so that it works in a different manner. Often the tendon/muscle causing the deformity is moved to work in the opposite direction to prevent the chance of deformity occurring again.

**Triple arthrodesis** – stabilising the hind foot joint by stiffening (welding) three bones together (arthrodesis is another term for “fusion”), preventing movement in three directions (drop-lift, left-right and ankle tilt). A triple arthrodesis is only carried out once the foot has stopped growing – usually age 12 in girls and age 14 in boys.

**Osteotomy** – cutting the bone and repositioning it. Usually the bone is fixed in its new position with plates, screws and other devices.

**The foot problems that are most usually considered for foot surgery include:**

**High arched foot (cavus foot)**

If there are no bone deformities, the goal of surgery is to release the tightened muscles and ligaments, relaxing the bottom of the foot so that it flattens, as well as releasing the pressure on the toes so they do not become clawed. Tendon transfers may also be necessary.
If there are bony deformities some removal or cutting of the bone (osteotomy) will be needed, as well as muscle and tendon transfers, removing the pressure that leads to a high arched foot.

**Turned in heel (heel varus)**

If the heel is rigid, some cutting of the bone may be needed. A wedge of bone will be removed from the heel bone so that the heel can be straightened.

**Claw toes**

If the toes are still flexible, then the cavus correction, above, should fix the clawing of the toes.

If, however, the toes are rigid, then tendons may need to be transferred to release the pressure causing the clawing, as well as fusing the joint in the middle of the toes.

**Help yourself**

Use this checklist to make sure you get the right treatment for you:

- Do the anaesthetist and the doctor carrying out the operation – usually a surgeon – and their team understand that you have CMT?
- Do they all understand what CMT is?
- Are they aware of the list of drugs that should not be used on people with CMT? (Ask CMT United Kingdom for a list.)
- Have you told them about all the drugs and treatments you are taking, including any over the counter treatments (eg cough mixtures) and any complementary or alternative remedies?
- Have they discussed with you all the possible options (including no treatment) and their benefits and risks? This is known as “informed consent” and the Department of Health recognises that you have a “fundamental legal and ethical right to determine what happens to [your body].” This means that everything needs to be explained in a way that you feel comfortable with.

**Anaesthesia**

Anaesthetics – general or local – are powerful drugs that have a particular effect on your nervous system and muscles. Because CMT is a condition that affects part of your nervous system you need to take special care that the anaesthetist understands well in advance of any operation that you have CMT. This holds true even if you only have mild symptoms, no symptoms at all or have a family history of CMT.

By making sure you have told the anaesthetist well in advance he or she can work out with you what type of pain relief is best for you. As an extra precaution you will probably be asked to stay in hospital longer after the operation to make sure everything is okay, which is why people with CMT are often not able to have day treatment.

In theory local anaesthetics (which work directly on the nerve fibres) will be affected by CMT. There have been reports of greater sensitivity to local anaesthetics in people with CMT, namely a prolonged
action of the anaesthetic and a more intense effect. Also, in current practice, local anaesthetics are injected around nerves after locating them using nerve stimulators. This technique may need to be modified or may not work at all on nerves that have been significantly affected by neuropathy.

Non-depolarising muscle relaxants (the other type of muscle relaxant is known as depolarising) are likely to have a prolonged effect on people with CMT. The effect would be to prolong an anaesthetic since someone with CMT would need to be kept anaesthetised until the relaxant wore off or could be reversed.

Get the most out of your doctor (or other specialist)

Your visits to your doctor can be quite fraught. To quote Blaxter (1983), “a consultation presents incompatible obligations: to be brief and helpful, not waste time which is manifestly in short supply, and yet somehow to tell the story of a life in all its long detail”.

To help you get the most of your time with your doctor, think about the following:

**Prepare for your visit**

You may only have five or 10 minutes with your doctor, so think about what you want from the appointment. Make a list of questions and important symptoms and think about taking along a trusted friend or relative (they can help take notes and remind you of questions you wanted to ask).

It is often helpful if you take along a list of any medication you are taking.

**Give information**

Tell your doctor everything he or she needs to know about your health, even the things that you may feel embarrassed about. (It is often helpful to have made a list of key points before the consultation.)

**Get information**

Feel at ease to take notes and to ask questions if there is anything you do not understand.

**If need be, get a second opinion**

Do not feel shy about asking for a second opinion, especially if you have to make an important decision about treatment options, like surgery for example.

**Get information about what comes next**

Make sure your doctor shares any information from tests and tells you what you need to do next.
Your consent

Before having any treatment, particularly surgery, you will be asked for your consent (permission). And before giving your permission you need to understand:

- what are the percentage success rates (and failures)?
- what will happen during the treatment?
- why you need it?
- what could go wrong?
- are there any alternative treatments?
- what would happen if you did not have the treatment?

This is called informed consent and is a legal procedure that all medical staff in the UK should follow before giving you treatment. Even after giving your consent, you are within your rights to change your mind.

Drugs to be cautious of

In theory some drugs and medication may affect people with CMT more than the general population. But, the evidence is slight or non-existent. The only drug proven to have a detrimental effect on someone with CMT is Vincristine - a chemo-therapy drug.

It is good practice to make sure that any doctor who is about to treat you knows that you have CMT. And understands what CMT is.

Talk to your doctor or pharmacist before taking any new medication, getting them to check for any known problems for people with CMT (known as contra indications), in particular ask them to check for the following words: “could cause peripheral neuropathy”. In almost all the conditions in which these drugs are used an alternative is available.

To put the possible risk into context: alcohol and illegal recreational drugs, rather than prescription medication, are much more likely to have a harmful affect on people with CMT.
National Service Framework for Long-term Conditions (England)

The needs of people with long-term conditions have often been overlooked by health and social services. To address this in 2005 the Department of Health in England published a national service framework (NSF) targeting the 10 million people living with a long-term neurological condition, such as CMT.

The NSF is made up of 11 Quality Requirements which the health and social services in your local area are expected to deliver over the next 10 years. How quickly they do this will depend upon local priorities.

These 11 Quality Requirements are:

1. A person-centred service – this is the key theme to the NSF, which is supposed to help ensure that the full needs – health and social care – of a person with a neurological condition are assessed and met.
2. Early recognition followed by prompt diagnosis and treatment.
4. Early and specialist rehabilitation.
5. Community rehabilitation and support.
6. Vocational rehabilitation.
7. Equipment and accommodation.
8. Personal care and support.
9. Palliative care.
10. Support for family and carers.
11. Care during admission to hospital or other health and social care settings.

No one is quite sure how the NSF will practically improve the service received by people with CMT and other neurological conditions as there is:

- no ring-fenced funding
- no plan to address the shortage of neurologists and nurses
- no apparent way of measuring progress
- no guarantee that the same services will be accessible in every area. (Services will be planned and shaped locally in response to different population needs).

However, as the Neurological Alliance points out, if you feel that you are not receiving the treatment promised under the NSF, you are quite within your rights to complain. In addition, the NSF will mean that primary care trusts are not penalised for improving their service to people with CMT.

People with CMT living in Northern Ireland, Scotland and Wales are not covered by NSF for long-term conditions (it only applies to England).
Complementary and alternative treatments

Some people swear by complementary or alternative treatments. However, the medical evidence is often flimsy or non-existent and you should approach them with care as they can be expensive for little or no benefit.

As with all treatments or medicines there are two main dangers with complementary treatments:

1. Not receiving the proper treatment – your indirect risk.
2. Taking something that harms you – your direct risk.

To be as safe as possible, it is a good idea to go through this checklist before trying a complementary treatment:

- Get a conventional diagnosis as well as relying on a complementary or alternative diagnostic method. If you don’t you run the risk of missing a potentially serious problem.
- Do not stop taking any medication that has been prescribed by your doctor without telling her/him. Stopping medication suddenly can be dangerous.
- Make sure the practitioner you are planning to see is a member of a professional association (and check with the association that the practitioner is really a member). Your GP may be able to give you list of therapists in your area.
- Tell your GP before starting any treatments as some herbs and other substances don’t mix well with conventional medicines, including over the counter remedies.
- Stop taking any herbal remedies at least two weeks before surgery as they could interfere with the anaesthetic. (Make sure you tell the anaesthetist if you are using any complementary treatments.)
- Take particular care if you are pregnant. Treatments may have a harmful affect on your baby.
- Keep a careful eye on hygiene, especially if your therapist is using any products such as acupuncture needles, for example.
Treatment hopes for the future

Our understanding of CMT is increasing rapidly every year, with more and more genes being identified that have an affect on our nerves. With this greater understanding comes an increasing hope that effective treatments for CMT may become available.

In February 2006 a Europe wide (the UK may not be included due to scarcity of funds) trial is taking place to see whether very high doses of vitamin C may benefit people with CMT1A.

Also, a hormone has been discovered that, although not currently safe to be used in humans, has been discovered to have some benefit on CMT. Research is going on to find off shoots of this drug and doctors expect trials to start within the next three years.

The most realistic hope is that drugs and treatments will become available that will stop CMT getting worse. The greatest expectation is that treatments will be specific to the genetic cause of CMT, which is why finding out which gene is responsible for your CMT will become more important.

The most likely chance is that a treatment for CMT1A will become available in the next 10 years that will stop the condition getting worse. While it is very unlikely that a cure will be discovered in this time, without a doubt, partially effective treatments will be available.
Caring for common problems

Although no two people with CMT are likely to have exactly the same symptoms there are some common problem areas that affect people to a lesser or greater extent, most of which are best treated by a variety of healthcare specialists.

Feet

Although easily overlooked the human foot is a complex and marvellous piece of engineering that needs proper care. Each foot is made up of 26 bones bound together by ligaments, supported by muscles and supplied with blood and nerves. Unfortunately the feet are usually the first area to be affected by CMT and need particular care.

CMT leads to three possible problems in the feet.

1. **Muscle wasting and weakness**

The shin muscles (at the front of the lower leg) which pull the feet and the toes up are usually the first muscles to be affected due to the breakdown in their connecting nerves.

**Common secondary problems**
- Foot drop.
- Very high arches (cavus foot)
- Flat feet
- Hammer toes or claw toes.
- Weak ankles.
- Corns and calluses.

All of the above can become painful and make it increasingly difficult to get around, leading to a higher risk of tripping and sprained ankles – not to mention strain on the knees, hips and lower back (see the section, Getting around), if not prevented or treated early.

**Measures to ease or slow down the onset of problems**
- Daily home stretching of the calf.
- Exercise.
- Physiotherapy.
- Orthoses.

**Treatments for existing problems**
- Orthoses.
- Physiotherapy.
- Stretching under supervision.
- Surgery.
2. **Loss of sensation**

Even if at first it is not very noticeable, most people with CMT may find that they lose some sensation in their feet due to the damage to the sensory nerves.

**Common secondary problems**
- Unnoticed damage to the feet (eg blisters, cuts, splinters), which can lead to:
  - sores
  - ulcers.

**Measures to ease or slow down the onset of problems**
- Proper fitting shoes.
- Daily feet checks.
- Podiatry.
- Orthoses.

**Treatment for existing problems**
- Wound dressings and drug treatments, either from your GP or in hospital.
- Podiatry.
- Orthoses.

3. **Problems with circulation**

Some people will find that their CMT leads to poor circulation in their lower legs and feet, so that they feel the cold more and the healing process is not as good as it should be (making ulcers more likely if damage to the feet goes unnoticed).

**Common secondary problems**
- Chilblains.
- Dry or cracked skin.

**Measures to ease or slow down the onset of problems**
- Keep feet warm (if using a hot water bottle take particular care that you do not burn your feet due to lack of sensation).
- Moisturise your feet, particularly in winter.

**Treatments for existing problems**
- Podiatry – a podiatrist will be able to diagnose and treat common problems that occur with your feet. They can assess nerve, circulation and muscle function and will also advise on foot care.
- Ulcers will usually be cared for by a district nurse, unless very bad when plastic surgery may be needed.
Key points to daily foot care

The all-important day-to-day care comes down to you and should include the following rules of basic foot care:

- wear shoes that are comfortable and offer good support
- make sure that your arches are fully supported
- keep your feet clean
- apply an unscented moisturiser, baby lotion or olive oil to dry skin
- avoid using hot water and strong soaps
- dry your skin carefully – don’t rub hard with a towel
- do not cut corns, calluses or ingrown toenails – see your doctor or podiatrist
- avoid bruises, burns, cuts, cracks, chilblains and frostbite. If you get any of these seek professional advice
- any signs of a problem with a mole on the foot should be checked by your doctor or podiatrist immediately
- avoid exposure to cold and dampness
- seek immediate professional advice if you ever get an ulcer or sore on the foot or leg.

If you ever have any loss of feeling (or feel numb) in your feet or legs, the following should also help:

- see a podiatrist at least once a year
- check your feet everyday; particularly look out for any cuts, splinters, abrasions or blisters. If you have any of these, make sure they are healing properly, otherwise talk to your doctor sooner rather than later. (A mirror on the floor propped up against the wall can help you see your feet. Otherwise ask a friend or a partner to check for you.)
- before putting on your shoes, shake them out to get rid of any pebbles and then check the insides with your hands to feel for any rough spots.

Orthotics and your feet

There are many different types of orthoses available to help with foot drop, flat feet, arch and ankle instability.

The most appropriate type for you may depend on how severe your foot problem is and, to some degree, personal preference. The different orthoses often give different degrees of control and it may require some discussion and experimentation to find the most appropriate degree of control. If you feel that the first orthosis offered is not right for you, do not give up or be afraid to ask to try other types.

If the alignment of your toes cannot be corrected and you have “clawed feet”, it is possible to use orthoses purely to provide comfort. This may involve special insoles, shoes or a combination with more extensive splinting.
Fatigue

We all get tired out sometimes. But fatigue due to a condition like CMT can really make it difficult to get on with the everyday tasks in our lives.

The medical definition of fatigue is when your muscles are unable to produce the same amount of force over time. In other words, they become tired quickly: something that affects many people with CMT, becoming tired even when doing mundane tasks around the house.

On a cautionary note, it is important to distinguish between exercise or activity that makes us healthily tired; and fatigue. The former is a good thing, helping us to keep our stamina and generally fit (read more in the chapter, Exercise and stretching) and cutting it out will lead to under activity.

“One of the biggest impacts is the constant feeling of tiredness. I’ve recently been given a project at work which involves sitting down for long periods and I find I’ve got extra energy – even to cook a nice meal. Whereas [before] I was always on my feet and at the end of the day I was so tired.” Carolyn

For people with CMT there are three main causes of fatigue:

- physical problems
- emotional problems
- pain.

1. Physical reasons

Your muscles can become increasingly prone to fatigue due to:

- CMT: as the nerves in your arms and legs become increasingly damaged, they are unable to relay messages to your muscles in these areas. As a result the muscles begin to waste away – becoming weaker and less able to produce the same amount of force over time as before (fatigue).

- the wrong muscles doing the wrong exercise: each of our muscles are designed to work in a particular way; if one muscle is forced to compensate for another muscle that is not working because of the effects of CMT, then it will become tired faster. For example, if you have foot drop (caused by weakness in your shin muscle) and you have to lift your leg off the ground higher so that your toes clear the ground, you will be putting extra strain on the knee, upper leg and hip, leading to fatigue.

2. Emotional reasons

Anxiety, depression, tension and stress, as well as poor sleeping patterns, can all lead to fatigue.
3. **Pain and fatigue**

Long-term pain can lead to fatigue either directly, or by contributing to depression.

**Measures to ease or slow down the onset of problems**

It is not possible to ward off fatigue entirely, but the following can help.

- Planning your day.
- Exercise.
- Healthy eating.
- Orthoses – by treating foot drop, for example.
- Occupational therapy.

**Treatment for fatigue**

- Gentle exercise.
- Special diet supervised by a registered dietitian.
- Medication for depression.
- Pain management.

**Exercise and fatigue**

Exercise can help keep fatigue at bay by increasing your stamina, strength and flexibility.

Surprisingly, even if you feel fatigued, a little bit of exercise (perhaps no more than a few stretches or getting out of the house) can get the blood flowing and give you more energy. The key is to get the balance right. Too much or too little exercise can both cause fatigue.

Keep the following pointers in mind:

- Regular, light exercise such as walking has been shown to reduce fatigue and can help some people to sleep better.
- Plan some activity or light exercise into your day.
- If exercise is impossible try to stay active in your daily routine.
- Pay attention to how your body reacts to exercise: How did you sleep? How did you feel the next day?
- Drink plenty of fluids before, during and after exercise.
- Try keeping an exercise diary of activities to share with your doctor or nurse, so they can help monitor your progress.
- It is important to find a balance between activity and rest, and exercise in a way that allows the muscles to recover after activity.
Healthy eating and fatigue

Making sure you are eating a well balanced diet, including the five food groups — carbohydrates, fruit & vegetables, protein, dairy, and fat — will help stave off fatigue.

Other pointers to bear in mind include:

- Keeping to a healthy weight (being overweight can make you more tired as you carry around more, whereas being underweight can leave you with little energy).
- Plan daily meals
- Organise the kitchen:
- cooking equipment and food within easy reach
- sliding equipment along the work surface
- avoid scrubbing pots etc, line with tinfoil, etc.
- Cook larger portions and freeze the extra in handy portions.
- Avoid sugar and caffeine hits as they tend to make you feel sluggish.
- Make sure you drink enough fluid (not caffeine or alcohol), about 8 glasses a day.
- Do not spend too much time preparing or cooking meals that you are too tired to enjoy them.
- Keep a food diary, along with a wider “fatigue diary”.

Read more in the chapter, Healthy eating.

Tips on beating fatigue

The following tips should help you save your energy. They are only a guide and you may not find that all of them are suitable for you. For more specific advice on how to conserve energy it would be a good idea to consult an occupational therapist.

Arranging the world to suit you

- Keep frequently used items where they are easy to reach. (Try duplicating household supplies in different areas, eg cleaners and dustpans in several locations.)
- Try to replace heavy items with lighter ones; for example a lighter vacuum cleaner.
- Put long handles on your taps and doorknobs.
- Make sure your work surfaces are at the right height for you as poor posture (bending over, for example) drains energy.
- Store frequently used items at a height between “your hips and your lips” and store things where they will be used.
- Install swing-out shelving into your cupboards, use stacking storage bins on wheels and wire shelf units that hook on the backs of doors.
Wear an apron with pockets or a builder’s belt to carry around tools and other kit.

Avoid deep-pile carpets or rugs that can slip – they are trip hazards and if you use a wheelchair they are harder to get around on.

Think about moving your bed to the ground floor, if there is a toilet on this floor, so you do not have to climb any stairs.

Cutting out needless effort

- Sit rather than stand whenever possible as it takes up a quarter less energy. Try sitting in the shower, while preparing meals, washing dishes, etc.
- Use special equipment to make everyday tasks easier. For example, try a jar opener, a shower chair or a hands-free phone.
- Soak your dishes before washing them and then let them drip dry.
- Use a trolley or lightweight luggage cart for moving things around the home – particularly for laundry, cleaning items or moving heavy items from the house to car, etc.
- Use the internet to get things delivered direct to your door.

Planning ahead

- Gather all the supplies you need for a task before starting.
- Phone the shops to make sure they have everything in stock.
- Cook in larger quantities and freeze/refrigerate the rest in easy to use portions.
- Schedule in breaks, making sure you rest before getting tired. (Try setting an alarm to go off after 30 minutes to remind you and then have a break for 15 minutes and then start again.)
- Take your time – rushing around really does take up more energy.
- Keep a diary to try and identify what activities take it out of you.

Prioritise

- Cut out tasks that are not important.
- Delegate tasks where possible.
- Talk to your occupational therapist to see if any help can be organised for you.

Rest

- Make sure you plan in rest during your day, resting before you become fatigued.
- Try not to be over ambitious on days you are feeling well as this can tire you out on the following days.
- Try and make sure you sleep well (read the Royal College of Psychiatrists’ information on sleep).
Hands

Although the feet are usually affected first, people with CMT often find that problems also occur in their hands. As with the feet the main problems are:

- muscle wasting
- loss of sensation.

Circulatory problems are rarely a problem in the hands.

Muscle wasting and weakness

Usually the small muscles in the hand that straighten the fingers are affected first, resulting in the classic “CMT hand”, with hollows where there should be bulk, for example at the base of the thumb. At the same time, people may notice that fine movements become more difficult (affecting writing, fastening buttons, holding a knife and fork, etc).

Common secondary problems

- Claw fist.
- Strain and tightness in shoulder and neck (as muscles in the upper arm try to compensate for loss of hand strength).

Measures to ease or slow down the onset of problems

- Stretching.
- Orthoses.
- Occupational therapy.
- Physiotherapy.

Treatments

- Orthoses (to help with hand function).
- Splints.
- Surgery.

Orthoses and hands

There are a range of orthoses for the hand that can help with:

- function (eg helping you hold a pen)
- maintain good position of the hand at rest, especially at night
- provide a stretch to specific hand muscles as part of a hand programme.

Rather than an orthotist, it is often an occupational therapist that will provide orthoses for your hands.
Balance

Another muscle that is often affected by CMT is the one that runs down the outside of the calf (peroneus longus). It is responsible for pulling your foot out. If it weakens, your foot can “turn in” at the ankle, so if seen from behind, it looks as though you are walking on the outside edge of your foot. This can cause instability (the “wobble board” feeling) and balance problems and also pain and calluses.

If the weaker ankles are combined with loss of some sensation in the foot, this makes balance much harder. This is because as well as relying on the stability of the ankle the body also relies on sensory information from the foot to keep balanced (without the sensory information, it takes the body longer to recognise that you are about to topple over, making recovery slower and harder).

You may find the “15 steps to prevent a fall” helpful in the chapter, Managing your CMT.

Common secondary problems
- Ankle sprains (and fractures).
- More falls.
- Calluses.
- Locked, or painful, knees. (Standing and walking with knees locked back (hyper-extended) causes the muscles at the front of the thigh and sometimes higher up to stop working.)

Measures to ease or slow down the onset of problems
- Orthoses.
- Occupational therapy (including handholds, assessment of your house and walking aids).

Treatment
- Orthoses.
- Plaster casts.
- Surgery.

Orthoses to support your ankles and your balance

Some people feel more balanced and can walk better if all of the arches of the foot are propped up, your orthotist may know of these as dynamic insoles or neuro-physiological footplates.

If you are unable to stand still without having contact with something stable to give you a reference point, it is sometimes useful to explore having very rigid orthoses to block ankle movement, eliminating the “wobble board” effect. It may be appropriate to use temporary orthoses or plaster casts to see how you manage before trying this option. It can initially feel very strange, but can have many benefits. Many people are put off by the looks of these orthoses as they are bulky and often require bigger shoes. It is however worth considering what it does to the appearance of walking as this can often be far more normal and have a bigger impact than the appearance of the orthoses.
By addressing problems at the ankle, it is often possible to improve alignment and get stronger at the knees, hips and trunk. This is an area where it can be very useful to combine use of orthoses with physiotherapy.

**Getting around**

It is very unlikely that you will ever lose the ability to walk (unless you have a rare form of CMT called autosomal recessive CMT1). However, over the years, you may find that getting around becomes more difficult and tiring. This is usually due to a combination of reasons, including problems with your feet, balance, muscle strength (fatigue) and pain in your knees, hips and lower back.

Specific problems related to walking include:

- Having to raise your feet higher off the ground to clear the toes, which droop downwards due to foot drop. This puts added strain on the hips and lower back.
- Not being able to place your feet properly: rather than the heel of your foot landing first, the toes – due to foot drop – will land first. Additionally, if your calf muscle is allowed to tighten it will become increasingly difficult to bring your foot at right angles, making it difficult to place your foot flat on the ground.
- Weak ankles and poor balance can put strain on your gait.

**Common secondary problems**

- More falls.
- Unwillingness to get out and about.
- Lack of confidence walking.
- Getting tired quicker when you do walk, due to lack of exercise.
- Increased weight (which in itself, can make getting around more difficult and adds to the strain on weakened joints).

**Measures to alleviate or slow down the onset of problems**

- Daily stretching and exercise.
- Orthoses.
- Good shoes.
- Physiotherapy.

**Treatments**

- Walking aids.
- Rarely, wheelchair.
- Adaptations in the house, handrails for example.
- Orthoses.
- Surgery.

Read about keeping mobile out of the home in the chapter, Practical issues.
Pain

It is not uncommon to find that people affected by CMT experience pain, to some degree, at some point in their life – sometimes briefly, but it can become a long-term problem. Generally pain experts believe the earlier you treat pain, the better.

Pain is a very personal experience. What causes your pain and how you feel it is likely to be very different from the person next to you. Evidence, for example, shows that women feel pain differently from men and that some of the drugs that work well for men do not benefit women as much.

There are two types of pain that people with CMT might experience:

1. Pain due to tissue damage. It may be caused by stresses and strains on your body due to the CMT, especially the bones, joints, tendons and ligaments (may be called mechanical pain). If, for example, you are walking with difficulty due to foot drop, you are likely to be putting extra pressure on other parts of your body.

2. Neuropathic pain, which means that the pain is caused by a problem with the nerves themselves.

However pain affects you it is important to remember that there are many different treatments available, including:

- orthoses
- re-training how you move with a physiotherapist
- various drugs
- specialist pain clinics.

If you have had your pain for a long time you may find that various psychological, social and behavioural training methods and counselling could help you to manage your pain and generally function better. Also, physical therapy and exercise can help control pain.

Drugs for pain

Everyone responds to drugs differently; if one doesn’t work another may. Remember to take especial care if you have another medical condition or take other drugs (including over the counter and complementary treatments).

Paracetamol

Paracetamol is very effective at relieving pain and is recommended as first option pain relief. It is cheap, easily available and gentle on your stomach.

- Paracetamol is safe as long as you follow the directions on the packet.
- Paracetamol can seriously damage your liver if you take too many.

Non-steroidal anti-inflammatory drugs (NSAID)

NSAIDs, which include ibuprofen, reduce swelling (inflammation) and pain. There are many different brands, so if one does not suit you talk to your doctor about trying another.
Taking NSAIDs regularly can irritate your stomach and cause problems like ulcers, especially if you are over 65 or take high doses. About one in ten people suffer these problems.

- Serious side effects can include stomach pain and bleeding. Talk to your doctor immediately if you have either of these.
- People with asthma, high blood pressure, stomach problems, kidney and heart failure may not be able to take them.

Stronger painkillers

If paracetamol or NSAIDs do not work stronger painkillers, like codeine or tramadol, may be recommended. You may hear them called opiates, opioids or narcotic analgesics. They are sometimes combined with paracetamol.

- Constipation is a common side effect of strong painkillers affecting up to half of people. Plenty of water and foods with high fibre may prevent constipation.
- Some people suffer from drowsiness, nausea and vomiting.
- Opioids can be addictive so could give withdrawal symptoms when stopping (although this is less likely with weaker opioids like codeine, but can still happen).
- Some people find that they have to take higher and higher doses of opiates to get the same level of pain relief, although tolerance is not that common.

Muscle relaxants

If your muscles spasm or are tense, a muscle relaxant or sedative, like diazepam, may help. They work effectively, but there is some argument about whether they do more harm than good because of their side effects and now they are not recommended for more than two weeks.

- Danger of addiction, even after a short course of a week.
- Seven out of 10 people get dizzy or drowsy within a week of taking them.

Other muscle relaxants such as Baclofen and Tizanidine are used in variants of CMT where there is increased tone in the legs. These should only be used when prescribed by your neurologist as you will need to be closely monitored.

Anti-epilepsy drugs

Anti-epileptic drugs such as gabapentin have started to be used for treating pain arising from some peripheral nerve disorders, either diseases or injuries. However, despite CMT damaging the nerves, the pain that people with CMT experience is mainly due to mechanical problems (stresses and strains relating to deformity, etc).

Antidepressants

Certain antidepressants, in particular a type called Tricyclic antidepressants (TCAs) can be extremely helpful in the management of long-term pain.

- Can give you a dry mouth dry and can make you drowsy, or constipated. Symptoms normally clear up after taking them for a short time.
Complementary treatments for pain

The forms of complementary treatments that are most likely to help relieve pain include:
- TENS machines (which block pain carrying nerve impulses)
- acupuncture
- osteopathy
- massage
- herbal remedies, such as devil’s claw or willow bark (the original source of aspirin).

Stress

Stress is the mental or emotional strain that we feel when, for whatever reason, the demands upon us are greater than our ability to cope.

Stress is usually caused by an external factor, such as a deadline to finish a piece of work, moving house, or the death of a loved one. However, none of these events will cause stress in themselves. That depends on how we react to the event. A single event for one person will be nothing more than a bit of excitement; for another it will lead to a feeling of major stress.

Short-term stress is not bad for us. It is only when it goes on for some time that it can become harmful, with a twofold result:
- If you are already feeling stressed, then you are likely to have less resistance to other forms of stress.
- Constant stress brings about changes in the chemicals in the body, which can lead to the dampening of the body’s defences to illness and disease.

Some of the common causes of stress include:
- long-term illness
- uncertainty for the future
- the unpredictability of CMT or other condition
- disability
- lack of control
- financial difficulties.

Common symptoms of stress include:
- disturbed sleep
- fatigue
- body aches
- pain
- anxiety
- irritability
- tension
- headaches.
Measures to deal with stress

The important thing is to recognise stress and work out a way to deal with it – the longer you leave it the harder it is to solve the problem and the more damage it can do to you. These are some general ways that you may be able to deal with stress successfully:

- Change the factors that you can control in your life for the better. Most importantly learn to delegate and say “No”.
- Exercise regularly - the natural decrease in adrenaline after exercise may counteract the stress response. Exercise will also help make you fitter and healthier and so better able to deal with the problems of stress.
- Relax - use techniques such as guided imagery, meditation, muscle relaxation and relaxed breathing (a type of meditation).
- Find a friend - social support can help reduce stress and prolong life.
- Recognise when you need help - talk to your doctor or social worker, to help you gain control over your symptoms.
- Avoid nicotine, alcohol and caffeine – alcohol, tobacco and drinks like coffee, tea and cola are all stimulants. Rather than calming you down, they will all tend to add to your anxiety or stress. They also dehydrate you, which can make you feel more tired and less able to cope.
- Sleep – make sure you are getting enough. Too little sleep, or interrupted sleep, makes us less able to deal with stressful events. But remember, too much sleep is not good for us either. Try to keep to a regular pattern, going to sleep at the same time each night; avoiding caffeine in the afternoon and evening, alcohol and do not watch TV just before trying to sleep.
- Rest – if you are ill, do not carry on regardless. When you are ill you need your rest.
- Listen to your body – if you are feeling tired or thirsty, do something about it.
- Stress diary – keep a note of when you feel most stressed and what causes it. It sounds simple, but it may help you deal with the root cause of the stress.
- Manage your time – plan ahead and take things one at a time. You may find it helpful to plan in time “buffers” so that you can deal with unexpected events.
- Be realistic – “accept the things I cannot change, the courage to change the things I can, and the wisdom to know the difference”.
Top CMT tips

CMT can be a right pain in the backside. It can make everyday tasks frustrating, tiring and sometimes very difficult. But there are a number of gadgets that make life that much easier.

The following tips and gadgets have been suggested by people living with CMT. If you have a tip or a gadget you would like to share, tell us about it. (If you suggest a gadget, remember to tell us how and where to get it from.)

And if you are looking for a tip or gadget to make your life easier, why not post a question on our online Forum (www.cmt.org.uk). You are bound to get a whole host of suggestions.

Odd jobs

DIY can be a right pain so firstly always use the right tools and of a decent quality.

- To compensate for weakness in the hands a small extension to the tool helps.
- Screw drivers with a torque setting can give that added strength element. The best set I’ve got was from Maplins (ref: N53AW)
- Sack trucks are handy for moving heavy objects. If you don’t have a sack truck, and have laminate floor in your house, put the object on a large towel and you’ll be surprised how easy it is to pull. It won’t mark the floors either!
- I find difficulty in putting screws in; they seem to have a mind of their own and want to go anywhere but in the hole. To counteract this I came across a set of screwdrivers with grips on the end to hold the screw whilst you align and turn it. I have found this to be of great benefit particularly when dealing with fiddly little screws. I bought them in a local electronics shop (Maplins).
- Cylindrical foam (a closed cell foam which you can wrap around or insert items into to make a larger easier grip) works well for eating utensils, toothbrush, safety razor and various tools. This is similar to foam used to insulate pipes, but is available from stores and catalogues that carry “disability” items.

Miscellaneous

- Rather than have light switches, all the lights in the garage and on the patio are operated by light sensors. You will never have to face stumbling about looking for the light switch, which is particularly important when your legs are weak. The sensors are not expensive either.
- When cooking, use a steamer or a pasta saucepan to cook vegetables - no heavy pans of water to carry to the sink to drain, just lift out the perforated insert.
- To help picking up small metal objects I use a magnet on a telescopic tube which we keep in the kitchen. As well as helping to pick things up it also saves you having to bend down.
Another idea for picking things up off the floor is a proper picker-upper, from disability shops. It is an essential piece of equipment and good for picking up anything from shoes and spectacles, to the post on the doormat.

Loop or spring-loaded scissors. They return to the open position automatically. Use a gentle squeeze to cut with them. They give you more control with less power than conventional scissors.

Opening things

- My best buy is an implement called a “Jar-Key jar-opener”. You position the jar-key against the rim of the jar and gently lift the key to release the vacuum. The lid can then be easily unscrewed.
- My most useful tool is an ordinary nutcracker. I use it to open all kinds of bottles, eg small tablet bottles, eye drops, and larger tops on pop bottles, etc. I also use it on those awful tops on bleach bottles where the two sides of the cap have to be pressed in. It really is invaluable.
- Use a square of “Dycem” in each hand to make opening screw-top bottles and jars easier. Use a small piece to get a better grip on peel-off covers on the top of milk cartons or fruit juice containers. (Available from OT stores.)
- I’ve also got a metal jam-jar opener thing from any hardware shop, which can open several different size jars or bottles. It’s a bit more awkward to handle, but sometimes is better for the really tough jars.
- I’ve recently bought a ring-pull can opener from Lakeland Limited (ref 4418), which just slots under the rings, and gives easy leverage to pull them up and backwards, costs about £3 and is worth every penny.
- And for those tins which don’t have a ring-pull, a really good, stable electric can-opener is essential - one that has a heavy enough base to be able to free-stand even when holding a heavy tin.

Personal care

- When fastening buttons, particularly collar buttons, I use a device which looks like a stiff wire loop with handle attached. I bought it at my local chemist. It cuts out the frustration and tedium of trying to get buttons done up. It also stops me pestering my partner every time I put a tie on, which can be pushing your luck, especially if they are trying to get ready to go out.
- If you can’t bend down to reach your feet (either due to CMT or due to something else, a hip replacement for example), dressing becomes a problem: especially putting on the underwear. To remain independent in this department, try using a long handled shoe horn – about 18inches (45 cm) – with a hook at the end. By strategically dropping the underwear on the floor, it is possible to hook them, and pull them on over the feet. It also works with trousers!
Getting about

- To help pace yourself, use a step counter - not to do your 10,000 steps, but to work out what your daily tolerance level is. You can check how much you’re doing and ease off, before you become exhausted.

- My father has one tip that I thought I would pass on; try using the walking poles designed for hikers and walkers. By using two and placing the weight in the webbing straps he reckons that they are as good as a frame, but without being overly cumbersome. He is also able to walk on un-paved paths. Another advantage is that they are telescopic and pack away easily. They also have rubber stoppers that you can put over the end so that they can be used indoors.

- I have recently re-discovered my old Dr Marten boots. Although currently not in vogue, they are absolutely perfect support for those of us in the early stages of CMT who just need a little extra foot and ankle support but don’t yet want to wear an orthosis.

Useful shops and online stores

**Lakeland Limited:** have a huge range of kitchen gadgets which are all very user friendly - they also stock many of the “good-grips” range too - all available by mail order, usually within just a couple of days. Tel: 015394 88100; Web: www.lakelandlimited.co.uk.

**Maplins:** are a phone and online electronics store with over 12,000 products. Tel: 0870 4296000; Email: sales@maplin.co.uk; Web: www.maplin.co.uk.

**OT Stores:** an online shop for disabled equipment and daily living aids. Tel: 0845 260 7061; Email: info@otstores.co.uk; Web: www.otstores.co.uk.
Charcot-Marie-Tooth: A Practical Guide
Coping with CMT

Most information on CMT talks about the physical effects of the condition. Or looks at how to get practical help, such as benefits or aids to daily life. While these are important, it is easy to overlook your need for emotional and psychological support.

Looking after your psychological (mental) health is just as important as looking after your physical health. Doctors are increasingly aware of the way that one affects the other. Just as ongoing poor physical health can put you under emotional pressure; ongoing psychological ill health, such as stress, can directly undermine your physical health.

The good news is that CMT has no direct effect on our mental health. There is no reason why you should ever suffer from mental ill health. But, like many conditions, CMT may affect the way you feel, about yourself and about the way you interact with other people. And, from time to time, you may feel a range of different emotions. This is perfectly normal and most people go through these feelings at some stage.

Because CMT changes and develops over time the exact way you manage and cope with the condition will have to be flexible and adaptive. Not only to the changes in the condition itself, but also to the changes in your life, including work, family, home and age.

“I do occasionally get angry, downhearted or depressed – but that can be caused by many factors, not just CMT. I joined CMT United Kingdom about 10 years ago. It’s been a comfort and an encouragement – the conferences are like a family reunion because of our “genetic” connection to each other.” Carolyn

Telling people about your CMT

At some time you are going to want to, or have to, tell people you have CMT. These people could include:

- partner
- children
- brothers and sisters
- other family members
- friends
- employers
- other work colleagues
- financial institutions, such as mortgage lenders or insurers
- DVLA
- school.
When you tell them is largely up to you, although they may initiate the conversation with unsolicited questions, sometimes when you first meet. Usually people do not want all the facts of your story. It helps to have little scenarios in your head for different circumstances. But be prepared to educate them. They are likely to know nothing about CMT and what it means for you, so it may be helpful to have the facts about CMT at your finger tips.

For official business it is helpful to have a brochure or something from medical literature in hand-out form explaining the basics of CMT.

How you feel about telling people will depend on you and who you are telling. You may find it nerve wracking, or that you are confessing, or you may feel very blasé about it and just drop it into conversation.

“Prior to my diagnosis I needed a way to explain why I needed help descending the stairs at my college graduation. I was in my 20s, felt inferior about my ‘feet problems’ and had to carefully construct my wording. I decided to say that I had a ‘balance problem’ and was unable to descend more than two to three stairs without a rail. Looking back, it is hard to believe how much emotional turmoil this caused me.” Susan

You are likely to find that different people want different information from you about CMT. Friends and family may be more concerned about your health, how CMT is likely to affect you and whether they could get it.

Most people are just curious and the minimum explanation will suffice. For example, for questions such as, “Why do you walk like that?”, or “Why do you wear those shoes/AFOs”, you may find telling them: “I have weak ankles because of a problem with my nerves to my legs. This means that my nerves are firing inefficiently and so my muscles are weaker.”

Your insurance company, on the other hand, will probably want to understand about how CMT may affect your ability to work, whether it will shorten your life expectancy (it does not!) and how it is likely to develop over time.

**Telling your adult friends and family**

Whoever you are telling, be prepared for that person to react in a very different way than you expect, however well you know them. They could be very upset, even angry, or they may seem entirely uncaring.

Some of their reaction depends on your presentation. The closer you are to them (genetically and personally) the more likely they are to react emotionally. It might be up to you to defuse and support their worries if they are unable to support you.

Remember, although you are the one with CMT, it can affect the people you are telling; especially if there may be a chance that they have inherited the condition too. (Find out more about the Odds of passing on CMT by reading the chapter, Genes: what they mean to you.)
Telling children that you have CMT

Before telling children, you may want to read CMT United Kingdom’s leaflet written for younger children, called “A Simple Guide to CMT”. The leaflet may help you marshal your thoughts and you can always leave it with them.

The following are some points to bear in mind when thinking of telling a child you or a close family member has CMT:

- From an early age children usually know when something is wrong or different – being told the truth can prevent them from being isolated or excluded.
- Very young children may be relieved to be told that your CMT is not their fault (“If I’d been good”, etc)
- Children will have different levels of understanding and will need information in a language they will be able to relate to. Their teacher may be able to help you.
- If your children are of different ages you may want to tell them the details separately.
- Check as you go along that they have understood everything. Be prepared to keep covering the same questions in a different ways.

Telling a child that he or she has CMT

“To tell or not to tell” is the age old question. And is a question with no correct answer. Probably the best advice is to follow your heart and your logic within the parameters of your family and your child’s ability to understand the information.

“My personal belief is that it is better to know and to be informed. My CMT was not diagnosed until I was 27. I felt that if I had known as a youngster, I would have had better self esteem. I would have had a reason for my clumsy-like movements: better to have a ‘medical condition’ than to be a garden variety klutz. So, of course, I brought my son up with the full knowledge of CMT. As an adult he says he would rather not have known. His CMT is milder than mine and as a child he says he felt he had something very seriously the matter with him.” Susan

Children with CMT are often aware at a very early age that they are in some way different from other children. Some parents think that it is better to keep the fact that their child has CMT to themselves but this may not be the best policy. Children can see very quickly through the reaction of others and by the fact that they may not be able to do the same activities as others that there is something different.

There are good arguments that it is important to inoculate your child with knowledge befitting her/his maturity so that your child can answer the same kinds of questions you are asked. If your child is comfortable facing the public, he/she probably has a good self image.

It may help to remember that as soon as children enter school, they are always under scrutiny by their peers, and are mercilessly questioned for any perceived deviation (freckles, smaller or taller,
red hair, etc. If you think it appropriate (your child is shy, uses a wheelchair, or may need help or understanding in not-obvious ways) you can be more pro-active and help the teacher prepare the class for your child’s needs.

It is important that at all stages children do not equate “difference”, due to CMT, with “wrongness”. If a child does not know the reasons behind the symptoms of their condition then it may add to a sense of insecurity and even inferiority. And children can be very tactless and even cruel. A child that is kept in the dark about her/his CMT may not be best equipped to deal with an innocent enquiry or any teasing. Feelings of inferiority established early on in childhood can lead to low self esteem throughout adult life.

Honesty, a sense of humour and a lot of encouragement will help children come to terms with their physical selves and give them confidence.

It is not unusual for parents, when faced with their child’s diagnosis, to go through similar emotions as if they had been diagnosed with the condition themselves, including denial. This is perfectly normal. You can compare this process to Kubler-Ross’s stages of grief (see appendix 1). Your “ideal” child has “died” and you must realign your hopes and dreams with the child you have been given. Your child will probably not star as an athlete or ballerina. Is this important to you? Choices may be different without being inferior.

Other feelings that may occur are anger, fear of the future and depression. Some parents may also feel a sense of guilt if the condition was hereditary. This can be a negative if you, the parent, have not accepted your own CMT and have unresolved issues. A mature and stable parent can serve as a positive role model and advocate for her or his child. With the right help both parents and those with the condition will find that each of these periods of doubt, confusion and guilt will pass.

Telling your employer

There is no law saying that you do have to tell your employer that you have CMT, unless it poses a health and safety risk. However, it may be in your contract, so it is worth checking.

It may be good idea to tell your employer if you think you are going to need time off work or will need help or adjustments to continue working effectively. Remember, the Disability Discrimination Act (1995) came into full effect in October 2004 giving you protection under the law as soon as your CMT has an effect on your everyday activities. Under the DDA your employer has to make suitable adaptations so you can continue working, whether your employer is sympathetic to you or not.

Telling the DVLA

By law, when you are diagnosed with CMT, you have to tell the Driver & Vehicle Licensing Agency (DVLA) Drivers Medical Group. You must also tell them if your CMT gets worse or you have any changes in your physical abilities. Find out more about keeping mobile in the chapter, Practical issues.

Telling your insurer and other financial institutions

Most insurance polices demand that you tell the insurer of any medical condition. Although your CMT may have no direct bearing on the cover of the policy, not telling them may void your cover. You may want to take advice from your local Citizen’s advice bureau or a financial adviser.
Stress

Stress is the mental or emotional strain that we feel when, for whatever reason, the demands upon us are greater than our ability to cope.

A little bit of stress now and then is probably a good thing. It helps us keep sharp and respond to danger (stress is part of our “flight and fight” response to danger and problems). But persistent stress has been shown to be bad for both our mental and physical health – dampening our body’s natural defences. Although there is no direct link, ongoing negative stress is a contributing factor to depression and should, therefore, be tackled early on.

Learn more about stress in the chapter, Caring for common problems.

Depression

CMT itself has no direct affect on our brains or whether we are likely to become depressed or not. However, if the problems caused by CMT are not dealt with successfully, the ongoing strain and stress may make us more susceptible to mental illness, including depression. Despite our day to day use of the word “depressed” to describe our everyday feelings of being down in the dumps, off colour or Monday morning blues, depression is a recognised illness.

In the words of the Depression Alliance, if you are depressed you have an illness which means that “intense feeling of persistent sadness, helplessness and hopelessness are accompanied by physical effects such as sleeplessness, a loss of energy, or physical aches and pains.”

Common symptoms of depression include (with thanks to the Depression Alliance):

- feeling tired and loss of energy
- persistent sadness
- loss of self-confidence and self-esteem
- difficulty concentrating
- not being able to enjoy things that are usually pleasurable or interesting
- undue feelings of guilt or worthlessness
- feelings of helplessness and hopelessness
- sleep problems - difficulties in getting off to sleep or waking up much earlier than usual
- avoiding other people, sometimes even your close friends
- finding it hard to function at work/college/school
- loss of appetite
- loss of sex drive and/ or sexual problems
- physical aches and pains
- thinking about suicide and death
- self-harm.

If you think you are depressed you should seek professional help from your GP and a counsellor. Contact the Depression Alliance for more information on depression. (See the chapter, Useful organisations.)
Taking control

The first rule: as soon as you feel that you are losing control, seek help. What help best suits you, only you will know. The following may help.

Acceptance

Accepting the fact that you have CMT and that it is going to have an affect on your life is essential. Only through acceptance, can you start to take responsibility for your health and really start to put everything into perspective.

Taking control

Although you may have to give up control of some parts of your life, it is important to take control of others. Examples of how you may want to take more control include:

- Knowledge – generally, the more understanding you have of a condition, the better you feel and respond to treatment.
- Planning – you may not have the energy you used to have, but by planning your day, you can take back some control.
- Positive thinking – “pain is inevitable, but suffering is optional” may sound odd, but positive thinking about how you approach issues can make a major difference in how you feel.
- Problem solving – as an occupational therapist (OT) knows only too well, by getting proper aids and by breaking a task down into its individual components you may be able to find away to do the things you thought were impossible.

Contact with others

Talk to others, including:

- Your healthcare team (doctor, physiotherapist, OT). Although your CMT cannot be cured and the root cause of the condition cannot be treated, your healthcare team will be able to advise you on suitable ways to manage it.
- CMT United Kingdom – talk to others like you on our members only Forum, or visit one of our local groups or call us for a chat. There are many thousands of people with CMT who will be able to share their experiences and methods of overcoming difficulties you may face.

“Middle-age has brought self-acceptance and confidence – as it does for many, disabled or not. Although I cannot wear the clothes or shoes I would like, because of The Feet, I treat myself to good haircuts and enjoy doing my make-up.” Carolyn
Practical issues

Despite your best efforts, sometimes your CMT may mean that day-to-day activities become more difficult. If this is true for you, this chapter has been put together to give you an introduction to the range of help from local and central government that is available to keep you mobile and independent.

You may also find it helpful to read the chapter, Top tips, which gives a list of ideas from people living with CMT.

In the home

If your CMT is making it difficult for you to live independently there are various improvements and adaptations that may be available that could make life easier for you.

Probably the first step is to contact your local social services to arrange health and social care assessment from an occupational therapist (OT). They will assess your needs and advise you on your options, recommend types of equipment and ideas about adapting your house.

What exactly is available to you may depend on where in the UK you live and will be affected by whether you are living in:

- council housing
- housing association accommodation
- private accommodation, either as an owner occupier or renting from a private landlord.

In general housing associations will apply for grants from the housing corporation for funding of adaptations; whereas council tenants will have adaptations funded by their housing department.

If you are living in private accommodation you may be eligible to for one of the following grants:

- **House renovation grants** – is a discretionary grant (subject to means testing) to help you get your house fit to live in, especially if it is structurally unsound or in serious disrepair.

- **Disabled facilities grant** – is a mandatory grant (subject to means testing, unless the adaptations are for a disabled child). This means that if an occupational therapist from social services believes that changes are necessary then an application will be made for the grant. This grant is designed to make changes to your house to make it easier for you to get into and around your home, for example, to provide suitably adapted kitchen or bathroom facilities. There is a limit of £25,000 per application (£30,000 in Wales) although multiple grant applications may be made if your needs change again in the future.

- **Home repair grant** – is a discretionary grant and may be given to meet the costs of minor repairs, improvements or adaptations. The limit is £2,000 per application or £4,000 over three years. (In Scotland there is a mandatory grant to provide standard amenities to meet your needs as a disabled person. You can find information from your local authority.)
Mobility aids and wheelchairs

There are a large number of mobility aids available to help you, including walking sticks and walkers that may help you get around in the home. Even well-placed handrails around the walls can assist greatly.

Wheelchairs are often most helpful outside of the home, for covering longer distances and for crossing rougher terrain. Look upon this as a way of taking back some freedom of movement out and about, rather than any sort of failure on your part.

Understandably many people are reluctant to use a wheelchair, feeling that they are “giving up”. The flipside to a wheelchair is that it has many positive benefits, not least that it can help you get out and about and feel more mobile.

“Too many people want to stay out of a chair at all costs – I don’t really understand why some people are so terrified of them! But why walk (longer distances) when it hurts and is bloody hard work!” Karen

Wheelchairs are available through the NHS (not social services), but you will need to be assessed by an OT or a physiotherapist, either at your home, in hospital or at your local NHS wheelchair service.

You will be helped to decide on the right chair for you by the wheelchair service. If you need a more expensive chair than is on offer, and the therapist agrees, you may be able to get a voucher to pay the difference (only available in England).

Budget issues often mean you will be offered the cheapest wheelchair suitable, but the assessors should take into consideration all circumstances. For example if the user or the “carer” also has weakness and so difficulty lifting a heavier chair into the car, and/or the user also has weakness in the arms and thus weight would affect independent use, then a lighter one should be considered.

“Knowing to push for what they don’t offer can make a big difference!” Karen.

If you want an outdoor powered wheelchair or electric scooter, you will almost certainly have to pay for this yourself. If you are on the higher rate of the Mobility Component of the Disability Living Allowance you may be able to buy a wheelchair on preferential terms through Motability. (Read the chapter, Useful organisations.)

“I’m now a full time powerchair user. It was my decision and it enabled me to do more things because I have the energy. So what if I use a wheelchair – least it stops my feet getting tired!” Lisa
You can get help choosing the right wheelchair for you from the Disabled Living Foundation and the Disabled Living Centres Council. (Read the chapter, Useful organisations.)

“I’ve now had the wheelchair about 10 to 12 years. I never envisioned I might reach that stage. I use the wheelchair only when I go out. If I go to the shops I use it, mainly because people don’t tend to notice sticks. I always seem to find in a crowd that everyone seems to walk the opposite way to me and it’s a constant battle against people. If on my own I can get quite panic stricken. I now do most of my shopping mail order, but then I never was a big shopper.” Margaret

Keeping mobile out of the house

If you need help getting out the house, the key benefit is the Mobility Component of the Disability Living Allowance (DLA). It entitles you to money to spend on your transport/mobility priorities and also opens up other benefits and discounts, including:

- Vehicle Excise Duty (Road Tax) exemption. You will need form DLA404 (MLS303 in Northern Ireland) from the Benefits Agency and present it with your car log and form V10
- The Motability Scheme.

“I tend to use a stick when outside, if it’s in the country I use trekking poles. I’m sure I could manage without a stick a lot of the time, but I daren’t. It’s starting to impact on what I do – I want to go out but don’t want to because I might fall. I’ve had two bad falls.” Carolyn

Driving, CMT and the law and insurance

A diagnosis of CMT means that you must tell the Driver & Vehicle Licensing Agency (DVLA) Drivers Medical Group. You must also tell them if your CMT gets worse or if you have any changes in your physical abilities.

People with a neurological condition, such as CMT, will have to fill in form B1 and give written consent to DVLA for them to contact your doctor for information concerning your medical condition. (Form B1 can be downloaded from the DVLA website.)

Failure to inform DVLA is a criminal offence that may be subject to a fine of up to £1000.

The majority of people with CMT are considered safe to drive, both now and in the future and are given a full “till 70” driving licence. Occasionally DVLA may issue a licence subject to earlier medical review (one, two or three years) or restrict driving to automatic vehicles or vehicles with adaptations.

DVLA standards are reviewed by the Honorary Medical Advisory Panel every six months and if you are ever in doubt, read DVLA’s “At a glance guide to the current medical standards of fitness to
Charcot-Marie-Tooth: A Practical Guide

“Working to support those who are affected by Charcot-Marie-Tooth Disease, also known as Hereditary Motor and Sensory Neuropathy or Peroneal Muscular Atrophy”

*drive* booklet on medical conditions and restrictions. This can either be downloaded from their website (free of charge), or sent to you in the post (current fee £4.50 per copy).

If you’re unsure about how to manage driving, or what kinds of adaptations you might require, MAVIS (Mobility Advice and Information Service) provide a complete assessment service. MAVIS can also put you in touch with other centres around the country that provide this service.

The Forum of Disabled Drivers’ Assessment Centres, which includes Mavis, now has a central contact freephone number that can advise customers of the nearest centre (0800 5593636) or see Appendix 4

**CMT and your driving insurance**

Licence holders need to understand that many insurance companies take the view that insurance cover is invalid if a medical condition is not declared to themselves and DVLA.

There have been cases in the past of insurers trying to “dump” all neuromuscular conditions into one box. However, under the Disability Discrimination Act they are no longer permitted to charge higher premiums purely for having a disability: only if the disability actually affects your driving ability. Insurers have to look at the effect of the specific disability, and its impact on you as an individual, not how they perceive or generalise another disability or neuromuscular condition.

**Motability – getting a car or a wheelchair**

If you are getting the higher rate of the Mobility Component of the DLA (or the War Pensioners’ Mobility Supplement) you may be able to get help, on preferential terms, through Motability, with:

- hiring a new car
- buying on hire purchase new or used cars or wheelchairs.

Motability also runs its own lease and hire purchase scheme.

**Blue Badge**

The Blue Badge scheme provides help with parking for “people with severe walking difficulties who travel either as drivers or passengers”. It has fully replaced the old Orange Badge and is recognised, with variations, throughout the European Union.

The purpose of the Blue Badge is to allow people with walking difficulties to park within 30 metres of their destination. However, some other benefits of the badge include:

- some free use of parking meters and pay-and-display bays, although this is being phased out
- exemptions in some cases from limits on parking times (very rare outside of London)
- parking for up to three hours on yellow lines (unless there are loading or unloading restrictions)
- exemption from the Congestion Charge in London if you have registered with Transport for London in advance of your visit and your car is registered with the DVLA as your “primary mobility vehicle” and/or they have given you exemption from paying road tax.
There are certain places where the Blue Badge does not operate; this includes certain city centres (including some London Boroughs), some airports and private roads.

You may be able to qualify for a Blue Badge if you:

- get the higher rate of the Mobility Component of the DLA (or get the War Pensioner’s Mobility Supplement)
- use a motor vehicle supplied by a government health department
- are registered blind
- have a severe disability in both upper limbs and cannot turn the steering wheel even if the wheel is fitted with a turning knob
- have a permanent and substantial disability that causes inability to walk or very considerable difficulty walking.

To get a Blue Badge you need to talk to your local social services department. (You can get more information from the Department of Transport Mobility and Inclusion Unit.)

For more information about Blue Badges, contact the National Association of Blue Badge Holders (NABBH)

For a map of Blue Badge parking in central London send a cheque for £2.50 made out to “Association of London Government” to: Blue Badge Guide, ALG, 1st Floor New Zealand House, 80 Haymarket, London SW1Y 4TZ. Call 020 7747 4777 for enquiries.

Disabled Persons Railcard

A Disabled Person’s Railcard is valid for 12 months and will give you reduced rates of up to a third.

You need to meet one of the following criteria to qualify:

- are registered as visually impaired.
- are registered deaf.
- have epilepsy, and are disabled by repeated attacks even though you receive drug treatment.
- receive Attendance Allowance.
- receive Disability Living Allowance (in the Higher Rate for help with getting around, or in the Higher or Middle Rate for help with personal care).
- receive Severe Disablement Allowance.
- receive War Pensioner’s Mobility Supplement.
- receive War or Service Disablement Pension for 80 per cent or more disability.
- or are buying or leasing a vehicle through the “Motability” scheme.

Children, aged from 5 and under 16, may also be eligible for a Disabled Persons Railcard, if they can meet at least one of the required criteria. Although they only need to pay the normal child’s fare, an adult can travel with them at the discounted rate of one third off the adult fare.

If you need help travelling by train, for example getting from your car to the train, contact National Rail enquiries 24 hours in advance.
Many stations have toilets accessible to wheelchair users that the National Key Scheme key, available from RADAR, will open.

Visit the Disabled Persons Railcard (www.disabledpersons-railcard.co.uk) website or call 0191 218 8103 for more information.

Disability, benefits and employment

Disability Discrimination Act (1995)

The Disability Discrimination Act (DDA) 1995 aims to end the discrimination that many disabled people face. This Act gives disabled people rights in the areas of:

- employment
- education
- access to goods, facilities and services
- buying or renting land or property

The Act also allows the government to set minimum standards so that disabled people can use public transport easily.

In April 2005 a new Disability Discrimination Act was passed by Parliament, which amends or extends existing provisions in the DDA 1995, including:

- making it unlawful for operators of transport vehicles to discriminate against disabled people
- making it easier for disabled people to rent property and for tenants to make disability-related adaptations
- making sure that private clubs with 25 or more members cannot keep disabled people out, just because they have a disability
- extending protection to cover people who have HIV, cancer and multiple sclerosis from the moment they are diagnosed
- ensuring that discrimination law covers all the activities of the public sector
- requiring public bodies to promote equality of opportunity for disabled people

Some of the new laws - including the increased protection for people who have HIV, cancer and multiple sclerosis - came into force in December 2005. The Department for Work and Pensions (DWP) website has more about the December 2005 changes.

If you are having difficulties at work, contact the Disability Employment Adviser (DEA) at your local JobCentre Plus. DEAs can assist with many aspects of keeping you in work, working with your employer to make adjustments as necessary to your workplace.

If you ever face discrimination, in addition to groups like the Disability Alliance, you can also contact the Disability Rights Commission (DRC). The DRC was set up by the government to help secure civil rights for disabled people and produces guidance and further information on which aspects of life are covered by anti-discrimination law for disabled people.
Definition of disability

Are all people with CMT disabled? No, not unless the condition has started to have an affect on your ability to carry out normal day to day activities.

Specifically the Act states: “if you have a progressive condition and it will substantially affect your ability to carry out normal day to day activities in the future, you will be regarded as having an impairment which has a substantial adverse effect from the moment the condition has some effect on your ability to carry out normal day to day activities.”

For more information contact the Disability Rights Commission on 08457 622 633.

Disability Living Allowance

Currently there are about four million people claiming Disability Living Allowance (DLA). The allowance looks at the help you need, rather than the condition you have, and has two strands to it:

1. Help with personal care.
2. Help with your mobility out of the house on a daily basis.

The benefit is tax-free, and not income related and is for people disabled before the age of 65.

To get the benefit you will need to fill in an extensive questionnaire to work out what care or mobility needs you may have, or you can choose to have a medical examination.

The benefit cannot be backdated – so get any application formally dated when it is sent out as there is usually a six week turn round period. If your claim is successful, then the benefit will be paid from the date on the form.

Other benefits

Benefits for disabled people are constantly changing so it is impossible to give a full list. Contact the Benefit Enquiry Line (a helpline for people with disabilities, carers and representatives) (BEL) on 0800 88 22 00, textphone 0800 24 33 55 for an up-to-date advice on benefits and information available. However main benefits, apart from the DLA, include:

- Attendance Allowance – a weekly benefit for people aged 65 or over that need a lot of help with personal care because of illness or disability.
- Child Tax Credit – has replaced the child-related payments in the Disabled Person’s Tax Credit.
- Working Tax Credit – a tax credit to top up the earnings of working people and will replace the Disabled Person’s Tax Credit.
Charcot-Marie-Tooth: A Practical Guide

- Incapacity Benefit – gives working age people a replacement income when they become sick or disabled and stop working or looking for work. You get it through a medical certification followed by a medical test known as the Personal Capability Assessment.
- Value Added Tax (VAT) – there are a wide range of VAT relief for things bought by people who are disabled. You can find out more information in VAT Notice 701/7 VAT Reliefs for People with Disabilities – available from your local VAT Business Advice Centre (in your phone book under “Customs & Excise”).

For more information about the support, help and benefits that may be available to you, visit the government’s www.Direct.Gov.uk site. Clearly presented, it is packed full of information.

You can also find a lot of very helpful information on benefits and the problems associated with claiming them at www.benefitsandwork.co.uk. To access the full website you need to pay, but even the free information is very useful as it written from the user’s point of view, rather than the government’s.

Holidays, accommodation and leisure

Holidays

If you are planning to get away, either in the UK or abroad, you may want to check that the accommodation, travel options and destination in general is accessible to your needs.

“As far as socialising and leisure goes I’ve never been one for sport and as far holidays are concerned I’ve been on cruises, toured Eastern Australia and been on safari in Zimbabwe. If I can get there I will. I’m finding tour companies are more aware of what’s needed and you do find access to places is easier.” Margaret

In the UK there is the National Accessible Standards for tourist accommodation, giving accommodation up to three different ratings:

1. Mobility – how accessible the accommodation is for people with mobility impairment.
2. Hearing level – whether it meets the needs of people with a hearing impairment.
3. Visual level – whether it meets the needs of people with a visual impairment.

If you are travelling abroad you will need to do some more research to make sure that your destination is going to meet your needs.

Whether you are travelling in the UK or further a field Tourism for All is probably the best place to start. Although they are a membership organisation, they offer information, advice and a booking service for destinations in the UK and abroad for all comers.
Alternatively, use the Visit Britain (the new name for the Tourist Council) website. They have a search function that allows you to find accommodation using the National Accessible Standards rating.

“I first met my husband on the Olympic squad 9 years ago and we said five years ago that we’d go travelling. We’ve both taken the view that we’ve got to enjoy life to the full and enjoy things that maybe other disabled people don’t – not for the praise but to do as much as we can.” Lisa

Leisure

There are a number of organisations that are specially geared up to make sports and leisure more accessible to people with disabilities. The sports covered include water-skiing, horse riding and scuba diving to name but a few.

Ultimately, you are the best guide to what you can or can’t do. If it hurts, stop. Remember, if ever in doubt about whether you should take part, talk to your GP, physiotherapist or the instructor at the sports facility.

For a more sedate approach to leisure time, there are numerous art-based organisations about to help you enjoy culture, even if you do have difficulty getting around.

Or, if you are looking for ways to stay in the garden there are companies that can help you with specially designed tools and techniques. One of them, the charity Thrive, offers a range of advice and information, including a website called carryongardening.org.uk.

It is just not possible to go through all leisure activities here, but your local organisation of people with disabilities may help you with the activity that gives you most pleasure.

You can also contact your local authority social services department and leisure department to see what is available. Many swimming pools and sport centres have special facilities and organised sessions especially designed for people with a disability.

Sexual relationships

If you want to talk to someone about personal or sexual relations there are a number of places you can turn to, including:

- your GP
- specialist voluntary organisations that help people with family planning, relationship or sexual issues.

Relate, offers a counselling service for adult couples (whether married or not) who are having relationship difficulties.
Having a baby

In the vast majority of cases there will be no reason why you shouldn’t decide to have children of your own. However, there are three issues you need to think about:

1. If you or your partner has CMT then you have up to a one in two chance of passing the condition onto your child. (Read more about inheritance in the chapter, Genes: what they mean for you.)
2. Pregnancy and labour rarely may cause your symptoms to worsen.
3. As a woman having a child your antenatal team needs to be fully aware of the implications of CMT.

Possible problems in pregnancy

Pregnancy can be hard work for any woman due to the extra strain on the body. For the majority of women with CMT there should be no specific problems. Difficulties you may face include:

- if you already have walking difficulties before the pregnancy, they may be worse in late pregnancy
- if you already have hand weakness then there may be problems after you have given birth, for example handling the child or breastfeeding.

CMT and your antenatal team

As with other healthcare professionals, most staff on the labour ward will not be familiar with CMT, so you may have to spend some time telling them. Get them to read some of the leaflets available from CMT United Kingdom.

Most women with CMT will have no problems using the usual pain relievers – including gas (entonox), pethidine or vaginal injections – available in labour. However you should make sure that your midwife and doctors understand that CMT is a neurological condition. See the section on anaesthetics in the chapter, Treating your CMT.

Caesarean sections

Some women with CMT choose to have a caesarean section. Again this is a decision for you to make with your healthcare team depending on your own personal situation.

If, for whatever reason, you decide not to have your own children, you may be interested in fostering or adopting a child. There is a huge need for carers nationally. If you’re interested, contact your local council or one of the organisations listed in here.
**Education**

Most children with neuromuscular conditions can be fully included at their local mainstream school, and will receive the best education there, enabling them to reach their full potential. Getting it right is a rewarding experience for all involved and will greatly enrich any school and community.

The Muscular Dystrophy Campaign has produced “Inclusive Education for Children with Muscular Dystrophy and other Neuromuscular Conditions. Guidance for primary and secondary schools” to support teachers educational advisers and parents.

It addresses the challenges that arise when children with a neuromuscular condition attend mainstream school. It includes practical advice on a range of issues such as adaptations to the physical environment, staff support, school policies and health concerns as well as how to develop a close working partnership with parents and professionals.

An understanding of the specific needs of each child, and how these will change, is essential. This guidance provides detailed information on the effects of a neuromuscular condition and a structured approach to forward planning.

The guidance includes:

- Information on the range of neuromuscular conditions affecting children
- A checklist for teachers/educational advisers on the issues, strategies/approaches and the resources available, broken down by Key Stages
- Chapters on the roles and responsibilities of staff and professionals, the emotional and psychological issues which may affect the child, health issues, environmental considerations and equipment, educational policies, extended schools provision and transition planning
- Resource materials for teachers, parents and pupils (including booklets for pupils with a neuromuscular condition and materials for their classmates) in a format that can be photocopied
- Useful contacts and bibliography.

To contact the Muscular Dystrophy Campaign call 020 7720 8055, or email info@muscular-dystrophy.org. Or visit www.muscular-dystrophy.org.

Also, see Appendix 3 for a summary of the current guidance on Education, with thanks to Contact A Family.
Genes: what they mean to you

The human body is made up of billions of cells (some estimates put the number at 10,000 trillion – or 10,000,000,000,000,000) that come together to make every part of your body. They range in size from less than 20 microns wide (that is about two-hundredths of a millimetre) to over a metre, in the case of the filaments of your nerve cells.

Your cells are directed on how to grow and function by your genes, the biological equivalent of a sheet of instructions or recipe. The instructions in your genes are followed to the letter. If there is a “mistake” in the genes, then the cells will be given the wrong instructions again and again resulting in faulty cell growth and behaviour.

For example, the gene peripheral myelin protein 22 (PMP-22) is one of the genes responsible for the insulating myelin sheath around your peripheral nerves. In people with CMT1A the gene PMP-22 is doubled up. As a result the myelin sheath does not work properly leading to the symptoms related to CMT.

“The more I’ve dug I’ve realised that an uncle had the condition – but it came out a different way – his feet were badly deformed, and I’ve subsequently found out a second cousin was also affected.” Carolyn

Passing on our genes and CMT

Humans have about 22,000 genes. These are grouped together on 23 paired chromosomes: 23 single chromosomes from your mother and 23 from your father. And, apart from the sex cells – sperm and unfertilised eggs – every single human cell has a full copy of these 23 paired chromosomes.

In contrast, the sperm and egg cells only have a single strand of chromosomes. When a sperm cell fertilises an egg the two strands combine, forming a new cell, with the double set. This cell then multiplies, creating a new individual with 10,000 trillion cells, each one containing a copy of the original 23 pairs of chromosomes.

In this way genes, including faulty ones (mutations), are passed on down through the generations. But not all faulty genes will have an effect. It all depends on the inheritance patterns of that single gene.

Inheritance patterns

1. **Dominant** (autosomal dominant)

   Some faulty genes are dominant, which means that you only need one faulty gene to cause CMT. In effect the dominant gene overwrites the instructions from the ‘good’ gene.

   The most common forms of CMT in the UK are dominant, including most forms of CMT1 and CMT2.
If you have a dominant form of CMT, you have a 50:50 chance of passing on the faulty gene to any children. If they inherit it, they will usually develop the symptoms of CMT.

2. **Recessive** (autosomal recessive)

If a faulty gene is recessive it means that you need two copies of the same faulty gene (one from each parent) before you could develop CMT.

If you only have one faulty recessive gene you are known as a carrier. You will not get CMT, but you have a 50:50 chance of passing on your faulty gene to your children. If they inherit it, they will only get CMT if they inherit a second faulty gene from the other parent.

Some forms of both CMT1 and CMT2 are recessive.

If your CMT is caused by two faulty recessive genes, you will definitely pass on one of your faulty genes to your children. But unless they inherit the same faulty gene from the other parent, they will only be a carrier.

If both parents are carriers for the same recessive gene, their children have a one in four chance of developing CMT (by inheriting two copies of the gene) and a 50 per cent chance of being carriers (by inheriting one copy of the gene).

3. **X-linked**

Of our 23 pairs of chromosomes, the 23rd pair is known as the sex chromosomes – either an X or a Y. If you inherit two X chromosomes you become a woman and if you inherit an X and a Y you become a man.

One faulty gene responsible for CMT has been discovered on the X chromosome. Men who get X-linked CMT are often more severely affected than women.

4. **“De novo” mutation**

Genes can change (mutate) “out of the blue”. If this happens then with future generations will be exposed to the risk of inheriting the changed gene in just the same way as described above.

About one in 10 people with CMT1A have a new genetic mutation.

5. **Sporadic CMT**

The term sporadic is used when someone with CMT has apparently no family history (ie no other family members affected). This can be due to either a “de novo” mutation as explained above or because there are no other family members around to examine to see if they may be affected.

“There was no one else in the family that had it. [The doctors] took me right through my family background. My mother’s family was the largest, but there was nothing. My father’s family was small but most of them had died. It was a bit of a mystery.” Rose
Summary of inheritance
You can inherit CMT in one of the following ways:
- if one parent has a dominant form of CMT
- if both parents are carriers of, or have, a recessive form of CMT
- if your mother has X-linked CMT
- if you are female and your father has X-linked CMT
- by a new mutation of the gene. ("de novo" mutation)

“It’s gone from generation to generation in the family; I remember aunties had sticks and callipers. It tended to run on the female side, my father was one of 16 children and was the only one of the males to have it and to pass it on. At least 13 of the others were affected. We just got on with it, it was never discussed. I just knew I walked funny. At the end of the day it was like having red hair – it was just there.” Ian

The odds of passing on CMT

Dominant inheritance (AD) (Most types of CMT1 and CMT2)
- One parent with CMT = one in two chance child will have CMT
- Both parents have same type of CMT = 75% risk of children having CMT

Recessive inheritance (AR) (Some types of CMT1 and CMT2)
- Both parents are carriers = one in four chance of having a child with CMT and a 50:50 chance of child being a carrier.
- Both parents have CMT = all children will have CMT.
- One parent has CMT and other is a carrier = 50:50 chance that each child will have CMT and 50:50 chance that each child will be a carrier.
- One parent has CMT and one parent unaffected = no children have CMT, but all children will be carriers

X-linked inheritance (CMT1X)
- Mother has faulty gene and father unaffected = 50:50 chance that children will have CMT (males more severely than females)
- Father has CMT and mother unaffected = all daughters will be affected, but sons will be unaffected.
Dominant [Affected + Unaffected]

With each pregnancy, this couple has a 50:50 chance of having a child with CMT, whatever the child’s sex.

Dominant [Affected + Affected]

With each pregnancy, there is 75% risk of children having CMT.
Recessive [Carrier + Carrier]

With each pregnancy, this couple has a 25% chance of having an unaffected child (of either sex), a 50% chance of having a carrier child, and a 25% chance of having an affected child (of either sex).

Recessive [Carrier + With CMT]

With each pregnancy, this couple has a 50% chance of having an affected child and a 50% chance of having a carrier child.
X-linked mother + unaffected father

X-linked affected father + non-carrier mother
Genetic testing

Of the 22,000 genes in the human body, as of February 2006 21 had been identified as causes of CMT. But every year around another five genes are being discovered.

Of these, only one – duplication of chromosome 17 – can be tested throughout the UK. And another four can be tested in specialist centres. A number of other CMT genes are tested but these are done on a research basis. Much of the CMT research is done in the neurogenetics unit at The Institute of Neurology.

- Chromosome 17 duplication (containing the peripheral myelin protein 22 gene (PMP-22)) – responsible for seven out of ten cases of CMT1 in the UK and very occasional cases of Dejerine Sottas disease (DSD) or Congenital Hypomyelinating Neuropathy (CHN). A deletion of the same part of chromosome 17 causes hereditary neuropathy with liability to pressure palsies (HNPP)
- Connexin 32 (CX32) – found on the X sex chromosome, it is responsible for CMTX.
- P0 (myelin protein zero) – responsible for CMT1B, CMT2 and some forms of DSD and CHN.
- PMP-22 (mutations) – responsible for CMT1, some cases of DSD and CHN and rare cases of HNPP.
- Mitofusin 2 (MFN2) – responsible for about one in five cases of AD CMT2

CMT is rapidly advancing in terms of the understanding of the genes and, therefore, the cause and the potential for treatments.

Where can you get advice?

The various tests can be carried out and genetic advice given at 22 regional genetic centres throughout the country (and the Republic of Ireland). Your neurologist or your family doctor can make the appropriate referral for you and any of your relatives who are worried.

To make the most of any genetic findings you need to have your results interpreted by your consultant neurologist or geneticist with an interest in neurology. They will be able to find out the implications for your particular diagnosis and give a spectrum of likely symptoms and how the condition is likely to change over the years.

(See the list of Regional Genetics Services at the end of this chapter)
Classification of CMT

The following is a summary of the current understanding of the genetic classification of CMT.

The key point to remember is that 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.

The rest of the various classifications are all variations of these two main groups and, currently, will have no impact on the management of the condition.

<table>
<thead>
<tr>
<th>Clinical type</th>
<th>Inheritance</th>
<th>Locus / Gene</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. Demyelinating (CMT 1)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>CMT 1A</td>
<td>AD</td>
<td>Duplication 17p11.2-12 / PMP-22</td>
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<td>17p11.2-12 / Point mutation PMP-22</td>
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<tr>
<td>CMT 1B</td>
<td>AD</td>
<td>1q22-q23 / Point mutation Po</td>
</tr>
<tr>
<td>CMT 1C</td>
<td>AD</td>
<td>16p13.1 - p12.3 / SIMPLE / LITAF</td>
</tr>
<tr>
<td>CMT 1D</td>
<td>AD</td>
<td>10q21-q22 / Point mutation EGR2</td>
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<tr>
<td>Charcot-Marie-Tooth type 1 x-linked (CMT X)</td>
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</tr>
<tr>
<td>CMT X</td>
<td>X-linked</td>
<td>Xq13.1 / Point mutation Cx32</td>
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<tr>
<td>Dejerine-Sottas disease (HMSN III)</td>
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<tr>
<td>DSD A</td>
<td>AD (AR)</td>
<td>17p11.2-12 / Point mutation PMP-22</td>
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<tr>
<td>DSD B</td>
<td>AD (AR)</td>
<td>1q22-q23 / Point mutation Po</td>
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<tr>
<td>DSD C</td>
<td>AD</td>
<td>10q21-q22 / Point mutation EGR2</td>
</tr>
</tbody>
</table>
Congenital hypomyelinating neuropathy (CHN)

CHN A  AD  17p11.2-12 / Point mutation PMP-22
CHN B  AD  1q22-q23 / Point mutation Po
CHN C  AD (AR)  10q21-q22 / Point mutation EGR2

Hereditary neuropathy with liability to pressure palsies (HNPP)

HNPP A  AD  Deletion 17p11.2 / PMP-22
17p11.2-12 / Point mutation PMP-22

Charcot-Marie-Tooth type 1 autosomal recessive (CMT1 AR)

CMT1 ARA  (CMT4A)  AR  8q13 - 21.1/ GDAP1
CMT1 ARB1  (CMT4B1)  AR  11q22 / MTMR2
CMT1 ARB2  (CMT4B2)  AR  11p15 / MTMR13
CMT1 ARC  (CMT4C)  AR  5q23-q33 / KIAA1985
CMT1 ARD  (CMT4D / HMSNL)  AR  8q24 / NDRG1
CMT1 ARE  (CCFDN)  AR  18q
CMT1 ARF  (CMT4F)  AR  19q13.1-13.3 / Periaxin
CMT1 ARG  (HMSNR)  AR  10q22-q23

2. Axonal (CMT 2)

Charcot-Marie-Tooth type 2 autosomal dominant (CMT 2 / HMSN II)

CMT 2A  AD  1p35 – p36 / KIF1Bb
AD  GTPase mitofusin 2
CMT 2B  AD  3q13 – q22 / RAB7
CMT 2C  AD  12q23 – q24
CMT 2D  AD  7p14 / GARS
CMT 2E  AD  8p21 / NF-L
CMT 2F  AD  7q11-q21 / HSP 27
CMT 2G  AD  12q12-q13.3
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<th></th>
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<th>Chromosome Range</th>
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<td>CMT 2L</td>
<td>AD</td>
<td>12q24 / HSP 22</td>
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<tr>
<td>CMT 2</td>
<td>AD</td>
<td>1q22-q23 / Point mutation Po</td>
</tr>
<tr>
<td>CMT 2 (HMSNP)</td>
<td>AD</td>
<td>3q13.1</td>
</tr>
</tbody>
</table>

**Charcot-Marie-Tooth type 2 x-linked (CMT 2X)**

| CMT 2X                | X-linked   | Xq24 – q26             |

**Charcot-Marie-Tooth type 2 autosomal recessive (CMT 2 AR)**

| CMT2 AR               | AR         | 1q21.2 – 21.3 / LMNA   |
| CMT2 AR               | AR         | 19q13.1                |
| CMT2 AR               | AR         | 8q21 / GDAP1           |

3. **Dominant Intermediate CMT (DI-CMT)**

| DI-CMTA               | AD         | 10q24.1-q25.1          |
| DI-CMTB               | AD         | Dynamin 2              |
| DI-CMTC               | AD         | 1p34-p35               |
Questions and answers

Knowing the precise genetic cause of my CMT, will that help me understand how my CMT is likely to affect me in the years to come?

How you experience CMT is likely to be specific to you – people within the same family can have very different symptoms. But, certain genes give a spectrum of likely symptoms and how the condition is likely to change over the years. So even though there is a spectrum within a family, there is a bigger difference between genes.

There are certain genes that cause CMT in the first year or two of life, whereas other ones cause it at 20 or 30. Your consultant neurologist will be able to tell you more about this.

Can CMT be identified in the unborn child tested for in pregnancy?

It is possible to test for those forms of CMT for which the causative gene is known in early pregnancy. Although these tests on the unborn child can be done from the tenth week of pregnancy onwards, investigations of some family members may be needed before the tests are done. But, unless you are absolutely certain that an affected foetus would be aborted, there is no real advantage in discovering at this stage if the child will have the gene that causes CMT.

I’ve just had a child and would like to find out whether they have CMT. Is it possible to have a genetic test?

This is a matter of some debate. Most doctors do not recommended genetic tests on children who have no sign of CMT. The children can make their own decision as to whether they want testing when they are 18. If they do develop signs or symptoms of CMT during childhood then they can have a genetic test in the same way as adults.

But a lot of people with CMT believe that parents should be able to have genetic testing done – a case of forewarned is forearmed. CMT United Kingdom’s position on this sensitive issue is that asymptomatic testing (ie before there are any noticeable symptoms) ought to be available to any parent who requests it – after suitable genetic counselling. It is possible that very early diagnosis could lead to earlier interventions (like physiotherapy or orthotics), ultimately having a positive long-term effect.

“Both my children were tested before their first birthday – a positive result for my daughter was instrumental in getting her early physiotherapy and intervention – which might have a positive long term effect.” Karen
What is a chromosome?

A chromosome is the biological equivalent of an enormous volume of instructions or recipes. (Each individual recipe or instruction is called a gene.) Our chromosomes are responsible for the way that the 10,000 trillion cells that form the human body behaves.

Apart from the sex cells (sperm and egg) each human cell has 46 single chromosomes paired together. We get a single set of 23 chromosomes from each of our parents.

Twenty two of our chromosomes are known as autosomal. The twenty third pair is our sex chromosomes. Made up of either an X or a Y, if we have two Xs we become female and if have an X and a Y we become male.

If we have children, one of our sex cells (a sperm cell or an egg cell) merges with our partner’s sex cell. The chromosomes in each cell combine into 23 pairs and form one new cell. This cell divides and multiplies and, eventually, forms a new human being.

Can women get “X-linked” CMT?

If you are a woman and carry the X-linked gene you can either be completely unaffected, in which case you are known as a ‘carriers’. You may be affected but this is usually milder than males. This variation is because female mammals have a problem with gene dosage. Each adult woman when she was an embryo randomly suppressed one or other of her X chromosomes in each cell in her body. If she carried the variant gene on the X chromosome, then it may be that this X chromosome is active in most cells in the relevant tissue, eg nerve cells, leading to neuropathy.
Regional Genetics Centres

North of Scotland Regional Genetics Service
Department Of Medical Genetics, Medical School, Foresterhill, Aberdeen AB25 2ZD
Tel: 01224 552120 Fax: 01224 559390

Northern Ireland Regional Genetics Centre
A Floor, Belfast City Hospital, Lisburn Road, Belfast BT9 7AB
Tel: 028 9026 3874 Fax: 028 9023 6911

West Midlands Regional Genetics Services
Birmingham Women’s Hospital, Edgbaston, Birmingham B15 2TG

South Western Regional Genetics Service – Bristol, Bath, Somerset and Gloucestershire
St Michael’s Hospital, St Michael’s Hill, Bristol BS2 8DT
Tel: 0117 9285652

East Anglian Medical Genetics Service
Addenbrooke’s NHS Trust, Box 134, Hills Road Cambridge CB2 2QQ
Tel: 01223 586828

Medical Genetics Services for Wales
Institute Of Medical Genetics, University Hospital Of Wales, Heath Park, Cardiff CF14 4XW
Tel: 02920 744028/744036 Fax: 02920 747603

Republic of Ireland Genetics Service
National Centre for Medical Genetics, Our Lady’s Hospital for Sick Children, Crumlin, Dublin 12,
Republic of Ireland
Tel: (01) 409 6737 (General Information) Fax: (01) 456 0953

Dundee - Human Genetics
Pathology Department, Ninewells Hospital, Dundee DD1 9SY

South East Scotland Clinical Genetics Service
Western General Hospital, Crewe Road, Edinburgh EH14 1JF

South Western Regional Genetics Service - Devon and Cornwall
Clinical Genetics Department, Royal Devon & Exeter Hospital (Heavitree), Gladstone Road, Exeter
EX1 2ED
Tel: (01392) 40 5726 Fax: (01392) 40 5739
“Working to support those who are affected by Charcot-Marie-Tooth Disease, also known as Hereditary Motor and Sensory Neuropathy or Peroneal Muscular Atrophy”
Northern Genetics Service
Institute of Human Genetics, International Centre for Life, Central Parkway, Newcastle upon Tyne
NE1 3BZ
Tel: 0191 2418600 Fax No: 0191 2418799

Nottingham Centre For Medical Genetics
H-Block, City Hospital NHS Trust Hucknall Road Nottingham. NG5 1PB
Tel: 0115 962 7728 Fax: 0115 962 8042

Oxford Regional Genetics Service
The Churchill, Old Road, Oxford. OX3 7LJ

Sheffield Regional Genetics Services
Sheffield Children’s NHS Trust Western Bank Sheffield. S10 2TH

Wessex Clinical Genetics Service
Princess Anne Hospital Southampton. SO16 5YA
“Working to support those who are affected by Charcot-Marie-Tooth Disease, also known as Hereditary Motor and Sensory Neuropathy or Peroneal Muscular Atrophy”
I don’t have CMT

For carers

If you are caring for someone with a disability or illness you may be able to get some support and help in your own right. This could include benefits, information, money towards a holiday, general support or even a trained carer to help you out or give you a break.

Some of the state benefits that are available to carers include:

- Carers Allowance – may be available to you if you are looking after someone who is severely disabled, a disabled child or an elderly person.
- Council Tax Benefit – some carers and disabled people can get reductions in Council Tax depending on their level of income.

You can find out more about what benefits are available to you by contacting your local Citizen’s Advice Bureau or calling the Benefits Enquiry Line for Disabled People or your local social services offices.

For parents

Being a parent of a child with CMT can be challenging, particularly if you have CMT yourself. You may have concerns about the progression of your child’s disability or you may be worried about how your child is going to get on in school.

There are networks of parents who can offer support, advice and, in some cases, grants, and the Disabled Parent’s Network and Contact a Family can be particularly helpful.

Education

You will need to discuss your child’s CMT and any resulting disability with her or his teachers to ensure the school is aware of your child’s needs. A fine line exists between expecting too much from a child and being overprotective, both for well-intentioned parents and teachers alike. (It’s worth noting that the recent Special Educational Needs and Disability Act 2001 gives parents and pupils extra rights to be included in mainstream schools.)

An excellent source of information on all aspects of the maze of rules and regulations surrounding education is the Muscular Dystrophy Campaign’s comprehensive publication, “Inclusive Education for Children with Muscular Dystrophy and other Neuromuscular Conditions. Guidance for primary and secondary schools”. Apart from the postage it’s free.
Holidays, activities and taking part

There is no blanket reason why your child shouldn’t be able to take part in activities with her or his friends. If in doubt, apart from CMT United Kingdom, you may find that Phab may be able to help. Phab is a charity dedicated to getting people to mix together – whether they have a disability or not – and organises holidays and activities across the country.

Further help

As a parent you may also come under the category of “carer” so do read about the support available to carers.

You may also find that the Disability Living Allowance (DLA) is available for your child.
Personal stories

What impact CMT will have on your life will depend on your individual circumstances. It is impossible to predict if your symptoms will be severe or non-existent. But however CMT affects you, reading how others have been affected and have coped with the condition may help give you ideas of how to get the most out of life.

If you would like to chat to others with CMT, why not log on to CMT United Kingdom’s online Forum (www.cmt.org.uk). Although anyone can read the discussions – only CMT United Kingdom members can contribute directly.

The opinions expressed here are personal ones and may not reflect the views of CMT United Kingdom. Some names may have been changed to protect people’s privacy.

Lisa’s story

Lisa is 44 years old and is a former Team GB Para-Olympic athlete.

My father had CMT so when it was noticed that I wasn’t walking right the doctor said I was copying dad. Dad had callipers. Some doctors said it was polio some that it was CMT.

I always knew there was something different I had problems walking, running and climbing trees. CMT was confirmed in 1983. Both dad and I went through tests and it was genetically confirmed.

School and after

When I started primary school I had half callipers. I think I had one case of bullying at school but one of my friends sorted him out – that was in Secondary. That school had four flights of stairs and I was in full length callipers by this point. In the summer I used to go home white with the energy I’d used. They were only 30 minute lessons so you had to keep moving.

I always liked swimming. We used to have sports days and we had a sponsored swim one year on how many lengths you could do. No one knew I could swim and I came second in class. I think that kind of changed things. When the girls had games I used to get an hours swim.

When I left school I went to college and they were better with people with physical disabilities. And I had a lot of opportunity to swim.

After college I worked with medical social workers for six months but it was full time and up stairs and I just couldn’t cope so I got signed off sick. When I was 18 or 19 I began using a wheelchair as my back kept giving way with the full length callipers.

After that I took up a sporting career. I trained and went to the Olympics in 1984 in New York in the swimming and field events. I came back with gold in the field event and bronze in swimming. I was 22.
Recent developments

The balance is getting a bit dodgy now and the area around the neck takes all the strain. My physio gives me a good stretch.

I don’t think my GP had seen anyone else with the condition. I know more than them.

I’m now a full time powerchair user. It was my decision and it enabled me to do more things because I have the energy. So what if I use a wheelchair – least it stops my feet getting tired!

One thing I hate is people leaning on my chair; it’s a real bug bear.

My husband is also in a wheelchair. We often get funny looks when we’re rolling along side by side.

Travel and marriage

I first met my husband on the Olympic squad 9 years ago and we said five years ago that we’d go travelling. We’ve both taken the view that we’ve got to enjoy life to the full and enjoy things that maybe other disabled people don’t – not for the praise but to do as much as we can.

Five years ago we went around the world in 62 days. Fifteen aeroplanes – Manchester, Frankfurt, Dubai, Singapore, Australia, New Zealand, Tahiti where we got married, America – New Orleans for Christmas Day and New Year’s Eve in Florida. South Africa is next in February or March 2006 – on a Safari.

We try to do it all.

Margaret’s story

Margaret is a 68 years old. Until retirement she was a social worker. She now uses a wheelchair.

The early years of CMT

I would imagine that it was noted that something wasn’t right at a very early age. Unfortunately I came from a family where disability and illness were very dirty words. I used to fall and would sprain my ankle, but it was put down to me being a clumsy child.

I was no good at games. My reflexes in my knees and ankles weren’t there and I had a high stepping gait.

It was when I was running for the bus at 14 and I broke my ankle that the physiotherapist said that I had no reflexes and that I was basically living with a disability that no one wanted to acknowledge. My father had something – a mild floppy foot.

At 17 when my ankle really hadn’t improved I saw someone, I think it was a neurologist, but my mother and father refused to tell me anything that was said. This was in the early to mid 1950s when people didn’t discuss things. I felt very angry that people wouldn’t tell me. In the late 60s, one of my mother’s sisters actually said it was all in my mind – even after I’d had quite a lot of surgery. My mother’s family were so healthy they couldn’t accept someone in the family had anything wrong.
Finally, a diagnosis

In 1960 I trained as a hospital social worker and got a job in Glasgow in the orthopaedic department. I was watched by the orthopaedic surgeon I was under as a patient who was very good at suggesting things that could be done to help. I then started to have surgery to have my toes straightened.

In 1967 I had two major operations to fuse my ankles to correct drop foot syndrome which helped to lessen the strain on my back.

In the 1960s I was told that I had a rare form of Muscular Dystrophy. But finally, when I came to Cardiff in 1981, I was told it was Charcot Marie Tooth (CMT). When I asked about the original diagnosis they said, “Oh we knew what you had, but thought you would understand Muscular Dystrophy better”.

Progression and wheelchairs

Gradually, as the years have gone on, my CMT has got progressively worse and I’ve had more surgery.

Like a lot of progressive conditions you can go through a period of rapid progression and then a period remission. It’s strange how it sometimes hits you and you realise you’ve got worse – but whether it’s some form of denial I don’t know. I realised about four or five years ago that I was having problems tying my shoelaces and it dawned on me it had probably been that way for a while.

I’ve now had the wheelchair about 10 to 12 years. I never envisioned I might reach that stage. I’m in the wheelchair only when I got out. If I go to the shops I use it, mainly because people don’t tend to notice sticks. I always seem to find in a crowd that everyone seems to walk the opposite way to me and it’s a constant battle against people. If on my own I can get quite panic stricken. I now do most of my shopping mail order, but then I never was a big shopper.

As far as socialising and leisure goes I’ve never been one for sport and as far holidays are concerned I’ve been on cruises, toured eastern Australia and been on safari in Zimbabwe. If I can get there I will. I’m finding tour companies are more aware of what’s needed and you do find access to places is easier.

Children

I have no children. I was told when I was 28 that I must never have children because I have a progressive medical condition that I could pass on. It was an unfortunate attitude of the last century. I was in a relationship at the time and when I told him he ran away. I thought every man was like that which is, of course, unfair. But in 1981 I got married, but by that time it was too late for kids.

Seeing doctors

In Glasgow I always saw my neurologist right away, perhaps because I worked in the hospital. Down here [in Wales] there was no regular follow up – it was only when I decided that I wasn’t having this and complained that I got into a six to 12 month regular appointment system. Now I’m usually seen once a year but I can phone and make an appointment if necessary.
But I know the system and so many people don’t. There just isn’t the money going into CMT as it’s not a life threatening condition. I’d like to see that change.

If I do go to see [a doctor] and they ask me if they can bring in students, because as they say, “We don’t get to see many like you”, I say yes. At least then the students have a chance to see someone with the condition and they might remember one day when they see it again.

Coping

The way I had coped with the condition was that I went on as normal. I think I had to cope on my own because of my parent’s attitude. I think this was why I went away to university and then made sure I went away to work. I was extremely fortunate in the head of department I had, I not only had physical support but also emotional.

The 60s and early 70s were very bad and I was off work a lot with depression and on high doses of tranquillizers. I was never referred to a psychiatrist. It was with the help of friends and colleagues that I could simply try to come to terms with something that was progressive.

I know my CMT is going to get worse – since my spinal op in May my balance has been poor, I’ve just got to adapt accordingly. Whether it’ll be easy or not, I don’t know.

I’ve got sclerosis of the spine, a floppy gait and my hands are gradually getting worse. Trying to do buttons is almost impossible now and if I didn’t have a husband it would all be Velcro.

Most people accept me for what I am and they don’t make concessions and I don’t expect any. If I do find someone who is difficult I just think you don’t have to respect their opinions.

Rose’s story

*Rose is 58. She first went to her GP in 1994, but was only diagnosed in 1997 with CMT type 2.*

I had problems for quite a long time before I was diagnosed and even then it came as a sort of, “Didn’t they tell you what you had?” That was in 1997.

There was no one else in the family that had it. The doctors took me right through my family background. My mother’s family was the largest, but there was nothing. My father’s family was small but most of them had died. It was a bit of a mystery.

However, certain things pointed to other people in my family having had it. When I was born the midwife said, “Oh she’s got feet just like Mrs Griffiths” who was my aunt. She certainly had a problem walking.

There wasn’t any issue about walking when I was a child, but I was certainly taken to see a foot specialist when nine or 10, when I began to get some deformity in my toes. The person I saw was rather unpleasant and accused my mother of not getting shoes that fitted properly – but we always got our feet measured. My mother’s reduct was that my brother’s feet were alright. When I was diagnosed I asked her, she was in her seventies by this time, if she remembered it, she said yes, and I told her what the real problem was. She cried – all that time later, she cried.
I noticed I couldn’t wear high heels – I was trying to walk and I couldn’t understand why I couldn’t. But you rationalise it – you say, “Oh it must be because I twisted my ankle so many times”. But of course twisted ankles are another symptom. This was the late 80s and I was in my early forties.

**Diagnosis**

I was starting to get numbness – not total numbness – like having an injection at the dentist. I was also getting weak – not able to grip things – not able to maintain pressure. I kept the problem with my hands to myself: I was anxious about it and wondered what was going on. People did say “You’re walking a little oddly, are you ok?” I was also getting very tired.

In September 1994 I went to the GP, who ruled out MS but said it was something neurological and referred me to a neurologist who I saw in January 1995. When I went into his office, from across the desk he recognised that my hands were wasting. By the end of the first visit I knew I had peripheral neuropathy, but to make sure they put me in for panoply of tests.

I was told the causes of peripheral neuropathy were many and varied and sometimes they don’t find exactly what the cause is. They had me back every 12 months for a recall appointment.

About a year to 18 months later I saw a senior registrar who said I should see the genetics department. They were extremely thorough. On the second visit my sister happened to be visiting so I rang and said would it help if she came along. When they did a simple neurological reflex test they found that she had no reflexes in her feet and ankles. But she has had no problems and can stand on her toes and trip the light fantastic.

At the time, I was terrified; I didn’t know what the future held and didn’t know how quickly I was going to get worse. It seems such an individual condition.

**Work**

My CMT seemed to progress quite quickly. I had a career as a dental hygienist but I noticed I was dropping things and you can’t do that in that job, because I was putting patients at risk from sharp instruments – and then there was the issue of insurance.

I finished working in 1996. I had to retire. It was a huge, huge blow. You don’t envisage being forced to take retirement like that. I was sitting and chatting to my GP one day and she asked if I was upset to give up work. I just dissolved into tears. I’d been bottling it up and she unplugged it.

It was much better though after leaving work. I was used to seeing people by appointment and living by the clock. It took a while to stop thinking, “I must do this, I must do that”. I was able to work at my own speed and had much less stress especially knowing there was no potential for me to harm anyone.

It was an amazing release, but it took an enormous psychological toll. Whatever we say to the contrary we define ourselves by work – you go to a party and someone says, “What do you do?” When this is taken away it’s difficult to negotiate socially. I found the best way of dealing with it was to be totally upfront about the condition.

If I’d have been in my thirties when I was diagnosed I would have taken a complete change in my career – I could have been a teacher like my sister and wouldn’t have had to give up work.
Coping

Soon after I left work I needed braces on my leg to walk any distance. And I have now got splints on my hands which I use if I get weak.

If I’m in a crowd of people all jostling it can be quite terrifying and I do panic in those situations. Wearing leg splints do prop you up and give you a bit more confidence.

I still feel extremely unsure of what the future may hold and old age.

I can cope with not walking but the possible loss of hands and arms I’m finding very challenging. It makes you dependent on others. If I was told 10 years ago that I’d be where I am now I would have been horrified, but you cope.

The condition tends to go in fits and starts. Sometimes I get frightening symptoms – my right hand started to go numb and lasted eight or nine hours – when that sort of thing happens it never quite returns to what it was before.

Getting medical care

When my neurologist retired, his list was taken over by another who felt he didn’t need to see me on an annual basis. But he has arranged for me to have open access – so if I’m anxious about a new development I can see him without having to be re-referred.

I see my GP every six months, who is wonderful. And I have a regular annual appointment with a physiotherapist.

I’m very obedient in doing my exercises. I swim a lot – I was a good swimmer before this happened and I feel completely free in the water.

After talking a lot to others I would say my medical care has been very good – at least it’s been ongoing which is more than a lot of people get. Especially physiotherapy – very few people seem to get physiotherapy care and open access to a neurologist.

Children and family

My children are adopted because of an entirely separate issue. However, and this is my own very personal view, if I had known that I had CMT and was thinking about having a child I wouldn’t have done it, because I wouldn’t have taken the chance of passing it on.

My family has been great – I’ve got three grandchildren now and they’re very used to granny having an extra set of legs (my SAFOs). But I find it very, very sad that I can’t lift them up. My kids are terrific they say, “Don’t worry mum you’re just going wobbly round the edges”.

My husband has always been extremely supportive but he’s very active and I hate the idea of him retiring into inactivity – a beige old age. We made the decision to spend some money and he’s going white water rafting with my brother. I’m going out to join them at the end of their adventure.

I guess old people say they don’t want to be a burden and that’s it exactly. And anyway I still want to do exciting things with my life too.
Carolyn’s story

Carolyn is 48 and her parents spotted something was wrong when she was a toddler.

My parents knew there was something wrong when I started to walk. My walking was slow, and by the time I was five or six I had an awkward gait and could not run as fast as other children.

My father and his mother had similar walking problems, although doctors who saw me as a child dismissed any connection. My mother had never discussed my father’s awkward walk with him, and my grandmother’s explanation for her lack of mobility and misshapen feet was that she had fallen under a pony and trap as a girl. In recent years I’ve realised that other family members were also affected by CMT, although their symptoms were not quite the same as in my immediate family.

As a child I walked “like a duck” and tended to trip easily. My parents were told it could be flat feet, knock knees, or even polio, despite the fact I’d been vaccinated. From the age of around 9 to 11, I attended physiotherapy sessions at a local hospital. I was also given arch supports, and later a leg iron for my weakest leg. I later discarded all these, as they did not seem to help much.

Diagnoses

At the age of 12 I was referred to a consultant – I presume a neurologist. He told my parents I had “Peroneal Muscular Atrophy”, and confirmed it was a hereditary condition. The general feeling was that we’d just have to live with it.

In my early 20s I went to see my GP to ask what the chances were of passing on my condition to children. He explained that there was a 50 per cent chance. He also assured me that there was no reason to suppose I would get any worse over the years.

It was only when I was married that I started to query what Peroneal Muscular Atrophy was. I had already decided (and told my husband-to-be) that I might not want children, but I hadn’t entirely ruled it out, and wanted all the information I could find on my disability. I found a medical encyclopaedia in the library and discovered that I had Charcot Marie Tooth disease – and it was a progressive disorder! This did not match what my family doctor had told me previously, but rather than question his knowledge, I simply re-defined the meaning of the word “progressive”. And so the process of denial began.

When I was at primary school, I had been able to flex my toes and lift my ankles. I lost this ability in my teens, but decided that it was my fault for not doing my exercises regularly. By my 20s, I could no longer skip with a jump-rope or run for a bus, but I put that down to simply not being a child any more. In my 30s I started to trip more often, and resorted to wearing ankle boots to prevent foot-drop. I also found I needed a handrail to climb stairs. Now I’m in my 40s, I avoid steps and stairs, and even find kerbs difficult to climb. I wear a silicone ankle-foot orthosis (SAFO), but still feel very wobbly unless I use a stick for support. It is only recently that my hands have given me bother – I do not have a strong grip, and in cold weather they don’t work properly at all!

Through learning about CMT through CMT United Kingdom, I now accept that I will probably get less mobile. I am still amazed that it took me and my family over 30 years to simply find out what was wrong with us – especially when I found out that CMT was first identified in the 1800s!
Medical Input

I don’t have regular contact with either a neurologist or a physiotherapist. About ten years ago, when I could no longer deny the progression of my CMT, I saw a neurologist who merely confirmed the diagnoses, and suggested I might visit the surgical appliance department. Later still, I asked for electro-conductivity tests, to establish what type of CMT I had (Type 2). This was mostly for my brother, who appears to be unaffected by the disease, and wanted to start a family.

A few years ago I asked my GP to refer me to a physiotherapist for “maintenance” exercises, and was granted six sessions during which I was given a regime to follow. I would like to have regular contact with a medical professional, but local resources are limited, and I am reminded that, “There’s not a lot we can do for you, you know”. I try to help myself by going swimming and walking on the treadmill at the gym.

As for orthotics, when I finally asked for help, I was offered bog-standard rigid splints, which were too painful to wear. They then gave me a calliper for my worst foot, which did the job up to a point. When I heard about the new SAFOs, I asked the hospital for some, and by a fluke (a locum consultant!), they agreed. However, they’ve said this will never happen again, and so I funded my next pair myself.

Getting About

I now use a stick for balance when walking outside, and trekking poles when in the countryside. In the past ten years I’ve had two serious falls, in which I tore ligaments in each knee in turn, and am terrified of it happening again. Sometimes I put off going out because of this.

After working in design, retail, and admin jobs, I ended up working in a centre for people with physical disabilities. This was long before I considered myself disabled. I was originally a handicraft instructor, which was fine for me, but then the job changed to include manual handling of disabled people – which of course I cannot do. My employees have been very understanding. Working with disabled people has helped me to face my fears, and also gives me real empathy with the centre users.

Fatigue

One of the biggest impacts of CMT is the constant tiredness. The centre where I work has no steps, but I do get tired because I am constantly on the move. If I get the chance to do a sit-down job, I find I am less weary when I go home. I would like to either change to a more sedentary job, or cut down on my hours – or both. At the moment I am hanging on, working full-time, because less hours means less money. I have asked if I would be entitled to any financial help if I worked part-time, but I do not meet the criteria.

I feel as though having CMT has limited my social capacity. Throughout childhood I endured teasing and name calling – although I also had some good friends, and did not generally regard myself as “different”. As a teenager my disability knocked my confidence with the opposite sex, and I was horribly conscious of my skinny legs. Middle-age has brought self-acceptance and confidence – as it does for many, disabled or not. Although I cannot wear the clothes or shoes I would like, because of The Feet, I treat myself to good haircuts and enjoy doing my make-up.
Looking back with hindsight, I wish I had done more, such as travel and exploring the countryside on foot. I do occasionally get angry, downhearted or depressed – but that can be caused by many factors, not just CMT. I joined CMT United Kingdom about 10 years ago. It’s been a comfort and an encouragement – the conferences are like a family reunion because of our “genetic” connection to each other.

**Ian’s story**

*Ian is 48. Despite having CMT since aged five, he was only told what the condition was called a few years ago. He recently retired from local government, where he was a senior manager, due to progression of the condition. He now uses sticks when out.*

According to my mother my CMT was diagnosed when I was about five, following chicken pox. My sister and brother also have CMT and the diagnosis follows a similar pattern.

It’s been passed on from generation to generation in the family. My father was one of 13 children (eight female and five male). I’m pretty certain his mother had CMT. To my recollection four aunts were affected although to a youngster the fact they had walking aids (calipers, sticks) made them no different. In fact I did a lot of running round for them to get a bit of pocket money!

Of the four females, two started families. One had three children and two generations later CMT has not resurfaced. The other had one son who has CMT but that hasn’t stopped him from becoming a very successful businessman. As far as I’m aware his two sons and their offspring have escaped CMT.

**Growing up**

With regards to a disability running in the family some saw it as a stigma (a legacy from Victorian upbringing I’m afraid). Most of us just got on with it and was rarely discussed. Personally I just knew I walked funny but that never stopped me carrying out a normal life and I was even OK at sport except for cross country which no one liked anyway so it was a good excuse to get out of it.

Obviously you got teased and called disrespectful names but it was rarely in malice and just reflected the ignorance of that and preceding eras. Kids will be kids. Although my Father rarely discussed it I know from talking to him in later years he felt hurt when others made comments about his kids.

My sister, brother and I used to have regular consultations with a child specialist who recommended various remedies including minor operations for both my brother and sister. That must have worked for my sister because as a child she wore calipers but they were abandoned I think before she reached her teens. For some reason I escaped the operating table and was just issued with corrective footwear.

During all this time I wasn’t aware what my condition was called nor was I particularly interested as it never affected my working or social life. However my sister was a bit more curious. And it is through her that I learned its name: CMT.

At 16 I applied for a job at Warwickshire County Council and that started my long Local Government career.
Work

I’ve worked 32 years and CMT never affected my work situation until the last couple of years.

I’d always worked in council buildings which are generally accessible and I regularly walked around Nuneaton Town Centre every lunchtime. However about three years ago I was transferred to another post based at the Council Depot which was about a mile away from Town. I’m convinced that started my downwards spiral as it restricted my mobility.

There was no doubt my job was stressful but I’ve always got on with life. However my health, and in particularly blood pressure and cholesterol levels were giving me concern and I was getting more immobile. But apart from a few trusted close friends and work colleagues I kept these matters under wraps. That is, until one day when everything came to a head when, after suffering chest pains, I was rushed into hospital and kept in for observation.

To me this was a reality check as due to high blood pressure I could have had a stroke, which would have ended my mobility full stop. I knew I would have to be realistic and accept that for once CMT was central to this state of affairs. I also decided to do some research and found CMT United Kingdom’s website extremely useful as I pieced together the picture.

Eventually, I retired early on ill health grounds. What it has taught me is that

- You have to accept that CMT is progressive and as you get older it is more likely to affect you. However try to adapt as changes take place and use your rights at work
- As well as the physical side of CMT there is the mental side.
- Be honest with everyone: I have retired with my head held high and with everyone’s good wishes because they now know about CMT.
- CMT does not prevent anyone working and at very high standard.
- Keep focused and positive.

I’m hoping these experiences, whilst quite painful on recall, can be shared and lead to a better understanding of CMT by all parties.

Getting around

Three years ago I could walk around town now I can only walk a few yards. What’s changed in last few years is the fear factor. If something happens I panic which I’ve never experienced before. I have this fear that I will not find somewhere to rest. Even crossing a road frightens me. My brother has expressed the same views and he’s younger than me!

I live on my own and I try to do everything for myself as I value my independence. Now I’m retired I can relax and ironically the issues of high blood pressure and cholesterol seems to have gone on the back burner and, for the first time in over three years, the nurse gave me a clean bill of health. However I have taken the experiences of the last couple of years into perspective.

About six months ago joined the gym and now go the Midland Sports Centre for the Disabled. Initially the sight of all those exercise machines frightened the life out of me. But once you’ve worked out a programme it’s surprising what you can do and how much confidence it can give you. Walking still frightens the life out of me; however once I start on a recumbent bike there is no stopping me.
Most of the facilities can accommodate disabled people. The exercise is also good because it gets your heart pumping. Surprisingly this also stimulates you mentally and once I get going I don’t think about the CMT.

The Sports Centre also has a pool and after a gap of 20 years I can still swim – this is excellent as you feel so confident in water. You can also use the Health facilities (sauna and steam room) so there is no excuse for being a bit overweight and blame it on lack or exercise!

I try to get about as best I can and occasionally use a walking stick if there is a crowd. However if I’m upright too long I get severe back pain. My biggest motivation is that I can still use my legs and keep positive that will remain the case for many years to come. I think if you lose your independence and motivation that is the time to pack in.

Despite being retired at the ‘grand age of 48’ I don’t know where the time goes. I look back on last year where every minute was focused on what the future would bring and what further impact that would have on CMT. Now that is confined to the annals of history and I look on this as a new phase in my life. In the last few months I’ve had a lot of work done to the house to make things easier and I am always helping others as the vast knowledge I have acquired over the last few years will definitely not go to waste.

I look at retiring with sadness, that I’ve finished, but with relief in other ways.
Glossary

Words in **bold**, indicate that the term has its own entry in the glossary and that additional information can be found there.

**Atrophy**
Wasting and weakening of a part of the body, such as a muscle or organ. People with CMT often find that the muscles in their lower legs and feet often start to waste first.

**Autosomal dominant/recessive**
The term for a single faulty **gene** that causes a health problem, even if its paired gene is normal. If both paired genes need to be faulty before a problem occurs, it is known as recessive. An autosome is one of the 22 out 23 paired **chromosomes** that are not sex chromosomes.

**Axon**
The **nerve** fibre that passes on electrical signals between the brain and other parts of the body. Axons can be over a metre long and are usually insulated by a **myelin** sheath.

**Cavus foot**
See **high arches**

**Cell**
The basic unit of all living things. The human body is made up of billions of cells (some estimates put the number at 10,000 trillion – or 10,000,000,000,000,000).

**Chromosomes**
A threadlike structure, found in every **cell** of the body, that carries the **genes**. If a single gene can be compared to a single recipe or instruction for cell growth and behaviour, then a chromosome can be compared to a huge book of body instructions/recipes.

In every cell there are 23 paired chromosomes; 23 from the mother and 23 from the father.

Of these 23, the 23rd pair is known as the sex chromosomes, as they determine your gender. If you have two ‘X’ chromosomes, you will be a woman; and an ‘X’ and ‘Y’ chromosome, a man.

**Claw toes**
Sometimes also called a hammertoe, it is a description of a toe that is locked (**contracture**) into a clenched, claw like, position. It is a common symptom of CMT. Seek advice from a physiotherapist or orthotist on how to prevent and treat it.

**Congenital (disorder)**
A condition or illness that is present at birth
### Charcot-Marie-Tooth: A Practical Guide

<table>
<thead>
<tr>
<th>Term</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Contracture</td>
<td>A shortening of a muscle or tendon, which then stops the associated joint from moving freely. Contractures happen when one of a pair of muscles that controls a joint – such as an ankle or toe – weakens (atrophies) allowing the opposing muscle to tighten and shorten, pulling the joint out of shape (deformity). At first a contracture causes a ‘flexible’ deformity which may be helped by exercise, stretching and orthoses. But if not treated, the joint will lock (fuse) and the problem becomes known as a ‘fixed’ deformity. At this point, surgery may be the only effective treatment option.</td>
</tr>
<tr>
<td>Fatigue</td>
<td>Physical or mental exhaustion due to exertion. Fatigue (tiredness) is a normal response to activity, but through problems, such as CMT, it can lead to someone needing more and more rest; even after very little activity.</td>
</tr>
<tr>
<td>Foot drop</td>
<td>Weakness in the shin muscle (front of your leg below your knee) makes it harder to pull your foot up. Increasingly, the foot will drop downwards, making it more likely to drag on the floor, leading to a higher risk of tripping and sprains.</td>
</tr>
<tr>
<td>Genes</td>
<td>The biological equivalent of a sheet of instructions, or recipe, that give directions to the cells in our body on how to grow and function.</td>
</tr>
<tr>
<td>Geneticist</td>
<td>Someone who studies genes.</td>
</tr>
<tr>
<td>Genotype</td>
<td>A person’s genetic (genes) makeup.</td>
</tr>
<tr>
<td>Heel varus</td>
<td>Also known as hindfoot varus, is a condition where the heel turns in, so that, when viewed from behind it appears that the person is walking on the outside edge of the foot. It can cause instability and balance problems.</td>
</tr>
<tr>
<td>Hereditary</td>
<td>Passing on genes and therefore certain traits, including a condition like CMT, from parents to children.</td>
</tr>
<tr>
<td>Hereditary neuropathy with liability to pressure palsies (HNPP)</td>
<td>A condition that is similar to CMT1A both genetically and because both conditions cause problems in the insulating myelin sheath because of a defect in the PMP22 gene (in HNPP there is one too few and in CMT1A there is one too many). Geneticists now classify HNPP as a form of CMT1.</td>
</tr>
<tr>
<td>High arches</td>
<td>Also known as pes cavus or cavus foot, people with CMT often develop very high arches, which can be painful to walk on. It is caused because the muscles and ligaments in the foot tighten (see contracture), pulling the ends of the foot closer.</td>
</tr>
</tbody>
</table>
Hindfoot varus  
See heel varus

HNPP  
See hereditary neuropathy with liability to pressure palsies

Ligament  
A sheet or band of tough, fibrous tissue connecting bones or cartilages at a joint or supporting an organ.

Lumbar puncture  
Inserting a fine hollow needle into the lower part of the spinal cord to either withdraw some fluid (for tests) or to inject drugs.

Motor nerve  
Your nerves can be compared to electrical cables, passing on information and messages between your brain and the rest of your body. Some nerves pass on commands from your brain to your muscles, instructing the muscles to contract or relax. These nerves are known as motor nerves. (Sensory nerves pass on information about sensation, such as pain, heat and cold, from your extremities to your brain.)

Myelin (sheath)  
A tissue that insulates and nourishes the certain nerve fibres (axon).

Nerve  
Cordlike bundles of fibre that conducts sensory or motor messages between the brain and spinal cord and another part of the body. Nerves can be compared to electrical wires, carrying electrical signals.

Nerve biopsy  
When a nerve, or a part of a nerve, is removed to be studied in a laboratory.

Nerve conduction tests  
Tests to find out how quickly messages are being carried by your nerves.

Nervous system  
The system of nerves that pass on commands and sensory information between the brain and other parts of the body.

Neurology  
The study of the anatomy, physiology and diseases of the nervous system.

Neuropathy  
A condition that damages the peripheral nerves (the nerves radiating out from the spinal into the arms and the legs), usually leading to muscle weakness and some loss of sensation.

Orthopaedic surgeon  
A surgeon who specialises in bone surgery.

Palsy  
Another word for paralysis.

Peripheral nervous system  
All parts of the nervous system outside of the central nervous system (brain and the spinal column), including the arms and legs.
**Pes cavus**
See [high arches](#).

**Progressive**
Getting worse over time. The term does not define how much worse or over what time span. The problems linked with CMT do usually become worse over time, but usually very slowly over the years, if not decades, often with little or no change from young adulthood to late adulthood.

**Sensory nerves**
Your nerves can be compared to electrical cables, passing on information and messages between your brain and the rest of your body. Sensory nerves pass on information about sensation, such as pain, heat and cold, from your extremities to your brain. (Your **motor nerves** pass on commands from your brain to your muscles, instructing the muscles to contract or relax.

**Varus heel**
See [heel varus](#).
Organisations that you may find helpful

This is not an exhaustive list, but should provide you with a good starting point to find the information and support you’re looking for. If you have any suggestions for other organisations that ought to be added, please let us know by calling us on 0870 7744314; emailing secretary@cmt.org.uk or writing to CMT United Kingdom, PO Box 5089, Christchurch BH23 7ZX. Thank you.

Generally helpful sources of help and information

- **CMT United Kingdom** – the national charity of and for people with CMT. It offers information, publications and support to people with CMT, their friends, family and carers.
  
  CMT United Kingdom, PO Box 5089, Christchurch, BH23 7ZX;
  
  Tel: 0870 7744314; Web: www.cmt.org.uk; Email: secretary@cmt.org.uk

- **Disability Alliance** – for information on benefits and your rights you can’t get much better than the Disability Alliance. They also publish (for £16.50) the ‘Disability Rights Handbook’ referred to as the ‘bible’ for disability issues.
  
  Disability Alliance, Universal House, 88-94 Wentworth Street, London E1 7SA;
  
  Tel: 020 7247 8776; Minicom: 020 7247 8776 (open 10.00am-4.00pm Monday-Friday);
  
  Web: www.disabilityalliance.org.

- **DirectGov** – the website that is supposed to bring together all the government services into one place.
  
  Web: www.direct.gov.uk

- **Disabled Living Foundation** – offers a great deal of information equipment, basically has five roles:
  
  1. Information on disability equipment;
  2. Factsheets on buying the right equipment for you;
  3. Contacts database of useful information;
  4. Helpline for queries about equipment for individual living.
  5. A permanent showroom (not for sales) so you can try equipment out.

  Disabled Living Foundation, 380-384 Harrow Road, London W9 2HU;
  
  Tel: 0845 130 9177; Textphone 020 7432 8009 (10.00am-4.00pm Monday-Friday).
  
  Web: www.dlf.org.uk; Email: advice@dlf.org.uk

- **DIAL (Disability Information Advice Line)** – run by people with disabilities and can put you in touch with your local DIAL branch to give you advice on benefits available to you locally.
  
  Tel: 01302 310 123; Textphone: 01302 310 123 (please use voiceannouncer); (open 9.00am-5.00pm Monday – Thursday, 9.00am-4.00pm Fridays)
  
  Web: www.dialuk.org.uk; Email: enquiries@dialuk.org.uk

- **Muscular Dystrophy Campaign** – provides practical, medical and emotional support to people with all forms of muscular dystrophy and has a wealth of excellent information and publications that are relevant to people with CMT.
  
  Contact them at, Muscular Dystrophy Campaign, 7-11 Prescott Place, London, SW4 6BS;
  
  Tel: 020 7720 8055; Fax: 020 7498 0670 (open 9.00am-5.00pm Monday – Friday)
  
  Web: www.muscular-dystrophy.org; Email: info@muscular-dystrophy.org.
Organisations for physical and mental well being

- **BackCare** – the charity for healthier backs with lots of information available. 16 Elmtree Road, Teddington, Middlesex, TW11 8ST; Tel: 020 8977 5474; (open 9.00am-4.30pm Monday – Thursday). Helpline: 0870 950 0275 (Helpline Hours: 9.00am – 12pm Monday, Tuesday & Friday, 12.00-2.30pm & 7.30pm-9.00pm Wednesday, 1.00pm-4pm Thursday) Web: www.backcare.org.uk; Email: info@backcare.org.uk

- **Benefit Enquiry Line for Disabled People and their Carers** – the official government helpline on what benefits are available, including getting applications for the Disability Living Allowance (make sure they formally date stamp the application before sending it out to you). Tel: 0800 882200 (Open: 8.30am-6.30pm Monday-Friday; 9.00am-1.00pm on Saturday).

- **British Dietetic Association** – mainly a professional body for dietitians it offers some information to the general public and can help you find a private dietitian. Tel: 0121 200 8080; Web: www.bda.uk.com; Email: info@bda.uk.com.

- **Chartered Society of Physiotherapists** – mainly a professional body, but some limited information for the general public. They can help you find a private physio in your area. Only web contacts. Web: www.csp.org.uk.

- **Department of Health Publications line** – provides the excellent and free ‘A Practical Guide for Disabled People or Carers’ (Ref: 29614). Tel: 08701 555455.

- **Depression Alliance** – a UK charity that works to relieve and to prevent depression by providing information, support and understanding to those who are affected by it via our publications and extensive range of services for people affected by depression. Depression Alliance, 212 Spitfire Studios, 63 - 71 Collier Street, London N1 9BE. Tel: 0845 123 23 20; Web: www.depressionalliance.org.

- **Dietitians Unlimited** – website to help people find a private dietitian (in association with the BDA). Web: www.dietitiansunlimited.co.uk.

- **Disability Alliance** – for information on benefits and your rights you can’t get much better than the Disability Alliance. They also publish (for £16.50) the ‘Disability Rights Handbook’ referred to as the ‘bible’ for disability issues. Disability Alliance, Universal House, 88-94 Wentworth Street, London E1 7SA; Tel: 020 7247 8776; Minicom: 020 7247 8776 (open 10.00am-4.00pm Monday-Friday); Web: www.disabilityalliance.org.

- **Expert Patients Programme** – Training and support for people with a long-term medical condition. Tel: 0845 606 6040; www.expertpatients.nhs.uk. If you have difficulty contacting them through these central details, try your local Primary Care Trust.

- **FRANK** – direct information about recreational and illegal drugs, the law, their dangers and why and how people use them. It has a confidential helpline and email
Charcot-Marie-Tooth: A Practical Guide

facility to check out specific information.
Tel: 0800 77 66 00; Web: www.talktofrank.com; Email: frank@talktofrank.com.

- **Health Professions Council** – the independent, UK-wide health regulator. Their job is to protect the health and wellbeing of people who use the services of the 13 types of health professionals registered with them, including podiatrists, dietitians, occupational therapists, orthotists and physiotherapists. Write to: Health Professions Council, Park House, 184 Kennington Park Road, London SE11 4BU.
Tel: 020 7582 0866. Web: www.hpc-uk.org

- **NHS Direct** – advice and information from trained NHS nurses.
Tel: 0845 4647. Web: www.nhsdirect.nhs.uk.

- **Pain Concern** – information and support for people who suffer from pain. They offer a Listening Ear helpline - a chance to talk to another pain sufferer.
Tel: 01620 822572 (9.00am-5.00pm Monday - Friday)
Web: www.painconcern.org.uk; Email: info@painconcern.org.uk.

- **Royal College of Anaesthetists** – mainly a professional body, but with some excellent free information on having anaesthetic. They have a whole website targeted at people worried about having an anaesthetic.
Tel: 020 7813 1900; Web: www.rcoa.ac.uk; Email: info@rcoa.ac.uk

- **Royal College of Psychiatrists** – the professional body for psychiatrists, but which also produces information for the general public on mental health, including a leaflet on sleeping well.
Tel: 020 7235 2351. Email: rcpsych@rcpsych.ac.uk. Web: www.rcpsych.ac.uk

- **TOAST (The Obesity Awareness & Solutions Trust)** – information, support and education around the issues of obesity. TOAST, The Latton Bush Centre, Southern Way, Harlow, Essex, CM18 7BL;
Tel: 01279 866010; Helpline: 0845 045 0225 (open 10.00am-4.00pm Monday – Friday).
Web: www.toast-uk.org.uk; Email: enquiries@toast-uk.org.uk

- **Weightwise** – website from the British Dietetic Association (see above) on healthy eating for a healthy weight. Web: www.bdaweighwise.com.

**Organisations for foot management**

- **British Association of Prosthetists and Orthotists** – the professional body for orthotists, unfortunately it does not have any general information for the general public, but can help you find a private orthotist.
Tel: 0845 166 8490; Web: www.bapo.org; Email: admin@bapo.com.

- **Institute of Chiropodists and Podiatrists** – has some general information for the general public on good foot care and orthoses on its website and can help you find a private chiropodist in your area. More importantly, their public relations officer is happy to field calls from the general public – send your questions to Fred Beaumont, 46 The Gardens, Whitley Bay, Tyne-and-Wear, NE25 8BQ.
Tel: 0191 297 0464. Web: www.inst-chiropodist.org.uk.
- **Chartered Society of Physiotherapists** – mainly a professional body, but some limited information for the general public. They can help you find a private physio in your area. Only web contacts. Web: www.csp.org.uk.

- **College of Occupational Therapists** – some general information, but the Association recommends that you contact your GP to find an OT, unless you are able to go private.
  Tel: 020 7357 6480; Web: www.cot.org.uk

- **British Orthopaedic Foot Surgery Society** – has some general information on foot surgery on its website, but does not accept requests for information. Web: www.bofss.org.uk.

### Organisations for aids and adaptations

- **Assist UK** – free and ethical advice and information about products that can help you stay independent.
  Tel: 0161 834 1044. Web: www.assist-uk.org; Email: general.info@assist-uk.org

- **Benefit Enquiry Line for Disabled People and their Carers** – the official government helpline on what benefits are available, including getting applications for the Disability Living Allowance (make sure they formally date stamp the application before sending it out to you).
  Tel: 0800 882200 (Open: 8.30am-6.30pm Monday-Friday; 9.00am-1.00pm on Saturday).

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  5. A permanent showroom (not for sales) so you can try equipment out.
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  Tel: 0845 130 9177; Textphone 020 7432 8009 (10.00am-4.00pm Monday-Friday).
  Web: www.dlf.org.uk; Email: advice@dlf.org.uk

- **Foundations** – national coordinating body for home improvements agencies in England.
  For Wales: Care and Repair Cymru. Tel: 029 2057 6286;
  For Scotland: Care and Repair Forum (Scottish Homes). Tel: 0141 221 9879

- **Leonard Cheshire Foundation** – a provider of services to people with disabilities.
  Tel: 020 7802 8200; Web: www.leonard-cheshire.org; Email: info@london.leonard-cheshire.org.uk
• **Motability** – the government funded charity that can, if you are on the higher level of the Mobility Component of the Disability Living Allowance, help you buy or loan a car or an electric wheelchair. Tel: 0845 4564566; Minicom: 020 7654 4224; Web: www.motability.co.uk.

• **National Centre for Independent Living** – information and advice on all aspects of independent living. 4th Floor, Hampton House, 20 Albert Embankment, London, SE1 7TJ; Tel: 0207 587 1663; Textphone: 0207 587 1177; (open 9.30am-5.30pm); Web: www.ncil.org.uk; Email: ncil@ncil.org.uk.

• **Occupational Therapists in Independent Practice** – a website directory of state registered OTs who can give you independent advice or provide private treatment. Tel: 0800 389 4873. Web: www.otip.co.uk.

• **Social Services** – contact your local authority.

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**Organisations to keep you mobile**

• **AA** – along with the RAC it is one of the leading motoring organisations in the UK. Has a disability helpline and discounts for Blue Badger holders. The AA operates a priority response for drivers who breakdown and feel vulnerable, for whatever reason. Tel: 0800 262050; Minicom: 0800 328 2810; Emergency messaging SMS service: 07900 444999; Web: www.theaa.com.

• **Department of Health Publications line** – provides the excellent and free ‘A Practical Guide for Disabled People or Carers’ (Ref: 29614). Tel: 08701 555455.

• **Department for Transport Mobility and Inclusion Unit** – information on the Blue Badge (including use in the EU); Road Tax exemption and travelling on public transport. They can also provide you with a Blue Badge ‘User Guide’ which includes a map of central London. Tel: 020 7944 8300 (open 8.30am-5.30pm Monday – Friday); Web: www.mobility-unit.dft.gov.uk.

• **Mobilise** – formed by the merger of the Disabled Drivers Association and the Disabled Drivers Motor Club. It offers support and information on all issues to do with disabled driving. Tel: 0870 770 3333; Web: www.dda.org.uk; Email: hq@dda.org.uk

• **Disabled Drivers Association** – see Mobilise

• **Disabled Drivers Motor Club** – see Mobilise

• **DVLA (Drivers and Vehicle Licensing Agency)** – People with CMT need to use form B1 to inform the DVLA. Tel: 0870 600 0301; Web: www.dvla.gov.uk.

• **Forum Of Mobility Centres** – for advice about your driving, the forum will direct you to the centre nearest to where you live. Tel: 0800 559 3636 (open 8.30am-5.30pm Monday – Friday); Web: www.mobility-centres.org.uk; Email: enquiries@mobility-centres.org.uk
Motability – the government funded charity that can, if you are on the higher level of the Mobility Component of the Disability Living Allowance, help you buy or loan a car or an electric wheelchair.
Tel: 0845 4564566; Minicom: 020 7654 4224; Web: www.motability.co.uk.

National Association of Blue Badge Holders – a charity that works on behalf of people who need or have a blue badge. The National Association of Blue Badge Holders, 18 Spruce Drive, Brandon, Suffolk IP27 0UU;
Tel: 01842 812459; Email: admin@bluebadge.org; Web: www.bluebadge.org.

National Rail Enquiries - for information on travel, including help travelling.
Tel: 08457 484950. Most individual train operators also have services for the disabled traveller. For example, Virgin Trains Journey Care: Tel: 08457 44 33 66; Textphone: 08457 44 33 67 (open 8am to 10pm daily).

RAC – along with the AA the Royal Automobile Association is one of the leading motoring organisations in the UK. Like the AA, the RAC operate a system whereby if you tell them of a disability, they will come out quicker.
Tel: 020 8917 2500; Web: www.rac.co.uk.

RADAR – Royal Association for Disability and Rehabilitation. Provides a range of services including the National Key Scheme.
Tel: 020 7250 3222; Minicom: 020 7250 4119; Web: www.radar.org.uk.

Railcard Helpline – for Disabled Person’s Railcard.
Tel: 0191 218 8103 (open 9.00am-5.00pm Monday – Friday)

Transport for London – 0845 900 1234 (Minicom: 020 7649 9123) or write to Congestion Charging, PO Box 2985, Coventry CV7 8ZR. Web: www.cclondon.com.

Tripscope – reliable transport advice and information, free of charge and nationally, for disabled and elderly travellers, including local, long-distance or foreign holidays. Equipment available for hire.
Tel: 0845 7585641; Web: www.tripscope.org.uk; Email: enquiries@tripscope.org.uk

Organisations for Disability, employment and benefits

Benefit Enquiry Line for Disabled People and their Carers – the official government helpline on what benefits are available, including getting applications for the Disability Living Allowance (make sure they formally date stamp the application before sending it out to you).
Tel: 0800 882200 (Open: 8.30am-6.30pm Monday-Friday; 9.00am-1.00pm on Saturday).

British Council of Disabled People – the umbrella organisation for all disabled organisations, it offers support and information and can put you in contact with an organisation in your area.
Tel: 01332 295551; Minicom: 01332 295581; Web: www.bcodp.org.uk; Email: general@bcodp.org.uk.
• **Department of Health Publications line** – provides the excellent and free ‘A Practical Guide for Disabled People or Carers’ (Ref: 29614).
  Tel: 08701 555455.

• **Disability Rights Commission** – it provides a helpline and information on the Disability Discrimination Act and advice on your personal situation.
  Tel: 08457 622 633. Web: www.drc-gb.org.

• **DIAL (Disability Information Advice Line)** – run by people with disabilities and can put you in touch with your local DIAL branch to give you advice on benefits available to you locally.
  Tel: 01302 310 123; Textphone: 01302 310 123 (please use voice announcer); (open 9:00am-5.00pm Monday – Thursday, 9.00am-4.00pm Fridays) Web: www.dialuk.org.uk;
  Email: enquiries@dialuk.org.uk

• **Citizens Advice Bureaux** – with a network of local offices throughout the UK this is one of the best places to find out what is available to you in your local area. Find your nearest Bureau in your phone book. Web: www.adviceguide.org.uk.

• **Disability Alliance** – for information on benefits and your rights you can’t get much better than the Disability Alliance. They also publish (for £16.50) the ‘Disability Rights Handbook’ referred to as the ‘bible’ for disability issues. Disability Alliance, Universal House, 88-94 Wentworth Street, London E1 7SA;
  Tel: 020 7247 8776; Minicom: 020 7247 8776 (open 10.00am-4.00pm Monday-Friday);
  Web: www.disabilityalliance.org.

• **GLAD (Greater London Action on Disability)** – for people in London this is one of the leading sources of information and support.
  Tel: 020 7022 1890;  Web: www.glad.org.uk; Email: office@glad.org.uk

• **Jobability** – run by the Leonard Cheshire Foundation and totaljobs.com this is the main disability jobs website. Web: www.jobability.com.

• **JobCentre Plus** – find your local JobCentre Plus in the phone book or use their website.
  Web: www.jobcentreplus.gov.uk.

**Organisations for holidays, accommodation and leisure**

**Holidays and accommodation**

• **Tourism For All UK** – c/o Vitalise, Shap Road Industrial; Estate, Kendal, Cumbria, LA9 6NZ;
  Tel: 0845 1249971 (info); 0845 1249973 (reservations); Web: www.tourismforall.org.uk;
  Email: info@tourismforall.org.uk

• **Visit Britain** – the British Tourist board.
  Tel: 020 8563 3379;  Web: www.visitbritain.com; Email: blvcinfo@visitbritain.org.

• **Visit Scotland** – the Scottish Tourist board. They produce a brochure ‘Accessible Scotland – a guide to accessible to accommodation and visitor attractions for people with physical disabilities.
  Tel: 0845 22 55 121; Web: www.visitscotland.com.
• **Social Services** – may be able to help you with holidays and transport.

• **Family Holiday Association** – gives money towards holidays and transport, but your social worker needs to apply on your behalf.
  Tel: 020 7436 3304; Web: www.fhaonline.org.uk.

• **Family Fund Trust** – gives grants for holidays for severely disabled children.
  Tel: 0845 1304542 (Mon-Fri 9.00-5.00); Web: www.familyfundtrust.org.uk.

• **Handicapped Aid Trust** – helps towards the cost of helpers of severely disabled people over the age of 17 who need a holiday.
  Tel/fax: 01253 780011.

• **Phab** - a national charity dedicated to promoting and encouraging the coming together, on equal terms, of disabled and non-disabled people to achieve an integrated and inclusive society. Organises holidays and activities.
  Tel: 020 8667 9443 (open 10.00am-4.00pm Monday – Friday); Web: www.phabengland.org.uk; Email: info@phabengland.org.uk.

• **Vitalise** – respite for carers and holidays for people with severe disabilities at their centres in the UK and abroad.
  Tel: 020 7017 3420 (open 9.00am-5.00pm Monday – Friday)
  Web: www.vitalise.org.uk; Email: info@vitalise.org.uk

**Leisure**

**Sport**

• **Disability Sport Events** – Tel: 0161 953 2499; Web: www.disabilitysport.org.uk.
  Email: info@dse.org.uk

• **Kidsactive** – adventure play for disabled children.
  Tel: 020 7738 9688; Web: www.kidsactive.org.uk.

• **Scottish Disability Sport** – Tel: 0131 317 1130;
  Web: www.scottishdisabilitysport.com

• **Sports Council for Wales** – Tel: 029 2030 0500; Web: www.sports-council-wales.co.uk.

• **British Canoe Union** – Tel: 0115 9821 100

• **British Ski Club for the Disabled** – Tel: 01747 828515; Web: www.bscd.org.uk.

• **British Disabled Water-ski Association** – Tel: 01784 483664;
  Web: www.bdwsa.org.uk.

• **Riding for the Disabled** – Tel: 024 7669 6510; Web: www.riding-for-disabled.org.uk.

**Art**

• **Arts Council of England** – offers details of organisations involved in arts and disability issues.
  Tel: 020 7973 6517; Textphone: 020 7973 6564; Web: www.artscouncil.org.uk
• **Artsline** – free information and advice for disabled people on arts and entertainment in London.
  Tel/textphone: 020 7388 2227; Fax: 020 7383 2653.  Web: www.artsline.org.uk; Email: admin@artsline.org.uk

• **Arts Council of Wales** – Tel: 029 2037 6500; Textphone: 029 2039 0027; Web: www.artswales.org.uk.

• **Scottish Arts Council** – Tel: 0845 6036000; Web: www.scottisharts.org.uk

• **Shape** – runs workshops, projects, events and courses.
  Tel: 020 7619 6160;  Textphone: 020 7619 6161; Web: www.shapearts.org.uk.

**Gardening**

• **Thrive** – gives training courses, advice and information on gardening as a therapy as well as advice for disabled gardeners.
  Tel: 0118 988 5688;  Web: www.thrive.org.uk; Email: info@thrive.org.uk.

**Organisations for individual personal issues**

• **Disabled Parents Network** – support and information for parents who are also disabled.
  Tel: 08702 410 450; Textphone: 0800 018 9949;  Web: www.disabledparentsnetwork.org.uk; Email: information@disabledparentsnetwork.org.uk

• **Disability, Pregnancy and Parenthood International** – Tel: 0800 0184730; Textphone: 0800 0189949; Web: www.dppi.org.uk.

• **Family Planning Association** – all forms of information and advice on contraception and sexual health, including local support.  Web: www.fpa.org.uk
  England - Tel: 0207 837 5432;
  Scotland - Tel: 0141 948 1179;
  Northern Ireland - Tel: 028 9022 2603;
  Wales Tel: 029 2064 4034.

• **Outsiders** – a nationwide self-help, community where people meet up and practice socialising. They have recently taken over the helpline of the defunct Association to Aid the Sexual and Personal Relationships of People with a Disability (SPOD).
  Sex and disability helpline: 0707 499 3527; Web: www.outsiders.org.uk.

• **Regard** – information and support for gays and lesbians with a disability.  Web: www.regard.org.uk; Email: secretary@regard.org.uk.

• **Relate** – counselling for adult couples (whether married or not) who have having relationship difficulties.  Relate has nearly 100 centres throughout England and Wales.
  Tel: 0845 4561310; Web: www.relate.org.uk

• **British Association of Adoption and Fostering** – Tel: 020 7593 2000; Web: www.baaf.org.uk.
• **Fostering Network** – Tel: 020 7261 1884; Web: www.thefostering.net.

### Organisations for carers

• **Benefit Enquiry Line for Disabled People and their Carers** – the official government helpline on what benefits are available, including getting applications for the Disability Living Allowance (make sure they formally date stamp the application before sending it out to you).
  
  Tel: 0800 882200 (Open: 8.30am-6.30pm Monday-Friday; 9.00am-1.00pm on Saturday).

• **Carers Online** – produced by Carers UK (formerly Carers National Association) provides carers and those supporting them with local and national information. Web: www.carersuk.org

• **Citizen’s Advice Bureau** – find your local office in your phone book or through the national website of Citizen’s Advice Bureau: www.citizensadvice.org.uk

• **Crossroads: Caring for Carers** – a national network that provides trained care workers to come to the home to give a carer under strain some support.
  
  Tel: 01788 573653; Web: www.crossroads.org.uk.

• **Family Fund Trust** – gives grants for holidays for severely disabled children.
  
  Tel: 0845 1304542 (Mon-Fri 9.00-5.00); Web: www.familyfundtrust.org.uk.

• **The Princess Royal Trust for Carers** – provides information and support, centrally and through its local groups, for all unpaid carers. 142 Minories, London EC3N 1LB;
  
  Tel: 020 7480 7788; Web: www.carers.org; Publications email: info@carers.org; General enquiries: help@carers.org.

• **Vitalise** – respite for carers and holidays for people with severe disabilities at their centres in the UK and abroad.
  
  Tel: 020 7017 3420 (open 9.00am-5.00pm Monday – Friday)
  
  Web: www.vitalise.org.uk; Email: info@vitalise.org.uk

• **Social Services** – contact your local social services to find out what support is available to you.

### Organisations for parents

• **Children’s Society** – runs projects throughout England and Wales and gives information on benefits and will help you with advocacy.
  
  Tel: 0845 3001128; Web: www.childrenssociety.org.uk

• **Contact a Family** – by bringing together families with children with disabilities they offer support, advice and shared experiences.
  
  Tel: 0808 8083555 (open 10.00am-4.00pm Monday – Friday);
  
  Web: www.cafamily.org.uk.
• **Council for Disabled Children** – acts as an umbrella organisation of many organisations concerned with helping children with a disability.
  Tel: 020 7843 1900; Web: www.ncb.org.uk.

• **Disabled Parents Network** – see under Individual Personal Issues above.

• **Muscular Dystrophy Campaigns** – see their excellent ‘Inclusive Education’
  Tel: 0207 720 8055; Web: www.muscular-dystrophy.org; Email: info@muscular-dystrophy.org

• **Parents for Inclusion** – parents helping parents so their disabled children can learn, make friends and have a voice in ordinary schools and throughout life.
  Tel: 0800 652 3145; Web: www.parentsforinclusion.org.
  Email: info@parentsforinclusion.org.

• **Phab** - a national charity dedicated to promoting and encouraging the coming together, on equal terms, of disabled and non-disabled people to achieve an integrated and inclusive society. Organises holidays and activities.
  Tel: 020 8667 9443 (open 10.00am-4.00pm Monday – Friday);
  Web: www.phabengland.org.uk; Email: info@phabengland.org.uk.

• **Social Services** - your local social services should be able to provide help for you and your child, including home visits, advice, information on local nurseries, childminders or playgroups, respite care and loan of equipment and play materials.
“Working to support those who are affected by Charcot-Marie-Tooth Disease, also known as Hereditary Motor and Sensory Neuropathy or Peroneal Muscular Atrophy”
Appendix 1

The Kubler-Ross stages of mourning

In 1969 Elizabeth Kubler-Ross wrote “On Death and Dying”. From her research, Kubler-Ross saw a pattern emerging that she expressed in the way of stages. These stages begin when the person is first aware of a terminal illness. While Kubler-Ross believed this to be universal, there is quite a bit of room for individual variation. Not everyone goes through each stage and the order may be different for each person.

Although her focus was on how people reacted to a diagnosis to a terminal illness, it has been noted that people go through similar stages when they learn that they have a long-term illness or disability. You may find that you experience some, or all, of these stages at different points in your life with CMT, as well as at the point of diagnosis.

Hope is an important aspect of all stages. A person’s hope can help them through difficult times.

1. **Denial and Isolation:**
   Felt by almost all everyone in some form. It is usually a temporary shock response to bad news. People can slip back into this stage when there are new developments or the person feels they can no longer cope.

2. **Anger:**
   Although common, people have different ways of expressing it, including:
   - “Why me?” often associated with the feeling that others are more deserving.
   - Envy of others: Other people don’t seem to care; they are enjoying life while I’m unwell. Others aren’t dying.
   - Projected on the environment: Anger towards doctors, nurses, and families.

3. **Bargaining:**
   A brief stage, hard to study because it is often between the individual and God.
   - If God didn’t respond to anger, maybe being “good” will work.
   - Attempts to postpone: “If only I could live to see . . .”
4. **Depression:**

Mourning for losses

- Reactive depression (past losses): loss of job, hobbies, mobility.
- Preparatory depression (losses yet to come): dependence on family, etc.

5. **Acceptance:**

This is not a “happy” stage. It takes a while to reach this stage and a person who fights until the end will not reach it. It consists resigning yourself to your diagnosis.
Appendix 2

HNPP (hereditary neuropathy with liability to pressure palsy)

Also known as HLPP (hereditary neuropathy with liability to pressure palsy), HNPP used to be thought to be an entirely different condition to CMT. It is now understood that HNPP is genetically very similar; in fact HNPP is now classified as a subtype of CMT1.

HNPP is an inherited nerve disease which affects the peripheral motor (muscle) and sensory nerves. Specifically HNPP leads to damage of the myelin sheath that insulates and nourishes the nerve fibre, known as the axon.

In a similar way to CMT, HNPP can affect people quite differently. However, usually the symptoms of a HNPP are not noticed until the second decade of life, although onset can vary from birth to old age. Some people with HNPP are only very mildly affected.

HNPP is known as a progressive condition, which means that it usually slowly gets worse over time.

Pressure palsy

The main hallmark of HNPP is pressure palsy. Palsy is an archaic word for paralysis; in HNPP it refers to a tingling, pins and needles like feeling and numbness, which often leads to muscles weakness and loss of function of a particular muscle.

Most people, with or without HNPP, will get some experience of palsy if they apply any prolonged pressure to a nerve. For example, sitting cross legged for any length of time can lead to pins and needles. However, for most people these odd sensations usually clear up within a matter of minutes. For people with HNPP the numbness and muscle weakness can go on for minutes to several weeks or months.

Other common symptoms linked to HNPP:

In addition to palsy, people with HNPP often experience the following problems:

- Foot drop – the foot muscle weakness/palsy causes the foot to drag.
- Ankle sprains and difficulty walking.
- Weakness of grip and loss of hand function can lead to difficulty with fine hand control, such as writing.
- Loss of overall strength.

As HNPP gets worse, it is not unusual for the following symptoms to occur:

- More generalised leg and arm weakness, including neck and shoulder problems. As with CMT, secondary muscle sprains and joint problems can develop.
- Tiredness and fatigue is common, which may lead to sleeping problems.
- Pain is frequently reported, despite many doctors declaring that pressure palsies are painless.
Common problems that bring on palsy

The following list includes some activities that may well bring on palsy:

- Crossing legs at the knee leaning on elbows sitting with legs crossed, or sitting tailor style.
- Sitting slightly askew in a chair.
- Sitting on something, such as a, chair, step, or stool, with legs out so there is pressure on the back of the thigh.
- Sitting in one position too long without readjusting.
- Kneeling.
- Gardening.
- Carrying anything by its handle (purse, suitcase, camera case). Loaded plastic grocery bags are among the worst.
- Using scissors.
- Knitting.
- Working with hand power tools.
- Holding the telephone in one position too long.
- Tying shoes too tight or tight shoe straps.
- High heels can make toes numb (even one inch heels).
- Painting too long (holding brush or roller) or painting above head.
- Walking too long (more than an hour).

If your HNPP progresses, you may find the following are likely to bring on palsy:

- Lifting weights.
- Using a mouse at the computer.
- Typing.
- Any activity on hands and knees.

Treatment and management

As with CMT, there is no specific treatment or cure for the underlying cause of HNPP. However, just as with CMT, it is possible to manage the primary symptoms so as to try and avoid secondary complications. These include:

- Pain Management - medication, and sometimes counselling.
- Occupational Therapy - work and home adaptations, eg kitchen devices, and also pressure relieving devices.
- Orthotic devices - to help with mobility difficulty caused by foot problems which can be helped with AFOs. These can include wrist and arm splints/support.
- Physical therapy – to help maintain mobility. A therapist with a particular interest in neuromuscular conditions would be ideal.
- Surgery - surgery is sometimes offered for nerve entrapment release. It is not generally
Genetic causes of HNPP

The same mechanism that causes the genetic fault in CMT1A is also responsible for HNPP. Both conditions are due to a problem with the gene peripheral myelin protein 22 (PMP-22). In CMT1A the gene is duplicated, but in HNPP it is deleted causing an under-expression of PMP-22. In other words, in HNPP, there is only one gene in the relevant pair, rather than the “normal” two, or three in CMT Type 1a.

Both HNPP and CMT1A lead to damage in the myelin sheath that nourishes and insulates the central nerve fibre called the axon. This is known as demyelination.

Because of the generalised demyelinating neuropathy, HNPP families have occasionally been diagnosed as having CMT, and only since the advent of genetic testing, has the cause been identified as HNPP.

How common is HNPP

As with CMT1a, HNPP is autosomal dominant. This means there is an equal chance of both males and females inheriting the condition, and a 50:50 risk for each child. The incidence is around 2 to 5 in 100,000 of the population – however the rate is thought to be 15 to 20 in 100,000, ie similar to that of CMT Type1a. Many people are undiagnosed or misdiagnosed.

For more information about HNPP contact http://www.mjleonard.pwp.blueyonder.co.uk/hnpp/ or www.hnpp.org
Appendix 3

Education

As a parent of a child with special educational needs, education may be one of your key concerns. Parents naturally have worries and anxieties about schooling, including whether special schools or mainstream education are the best options.

The Special Educational Needs (SEN) and Disability Act 2001 sets out changes to education for children with SEN in England and Wales. It also brings access to education within the remit of the Disability Discrimination Act, making it unlawful for education providers to discriminate against disabled pupils, students and adult learners. The Special Educational Needs and Disability (Northern Ireland) Order 2005 has strengthened SEN legislation to schools and the further and higher education sectors for the first time.

Education in England, Wales and Northern Ireland

If your child has special educational needs (SEN) s/he may need extra help and support to get the most out of the education system. For those with special educational needs which cannot be met within the early years settings or school’s own resources, the Local Education Authority (LEA) or Library and Education Board in Northern Ireland, may carry out an assessment to help them decide whether further action is required. The appropriate authority must consider an assessment unless it thinks it is unnecessary or if your child has had an assessment in the last 6 months. As a result a document may be issued, known as a Statement of Special Educational Needs, which sets out how they should be met. The whole process, from considering whether an assessment is necessary, to producing a final Statement should take no longer than 26 weeks. In Northern Ireland, the timescale is 18 weeks.

England and Wales each has their own Code of Practice for Special Educational Needs, as does Northern Ireland, which is also governed by separate legislation.

Statements in England and Wales

The following stages of assessment are set out in the relevant Special Educational Needs Code of Practice which details the key principles for identifying and assessing SEN.

Educational Assessments for children under 2

If your child has a particular condition or major health problem which has caused concern from an early age, they may be referred to the LEA for an assessment. This assessment does not need to follow the statutory procedures that are applicable for children over 2.

Educational Assessments for children under compulsory school age and over 2

**Early Years Action** - this happens when it is felt a child needs additional support to those provided as part of the usual curriculum on offer. This Action will be interventions agreed by the SENCO (Special Educational Needs Co-ordinator) and child’s teacher in consultation with the child’s parents.
Early Years Action Plus - this stage is when outside support services or more specialist advice is sought to aid a child’s development.

Requests for a statutory assessment - for some children the stages above are not sufficient to meet their additional needs. It is at this stage that a request for a statutory assessment may be made. This request can be made by you as a parent, or it may be from one of the professionals who have contact with your child.

Educational Assessments for Children attending school

Some children will have had their special educational needs identified before attending school - for others this is not the case. The stages are similar to a child attending an early years setting.

School Action - this is when a teacher or SENCO identifies a child with SEN who needs additional or different support to those offered in the general curriculum.

School Action Plus - This results in the request for help from external services or specialist help to meet a child’s needs.

Request for a Statutory Assessment - Where it is felt the child still has significant needs which cannot be met by the actions of previous stages, the school may request a statutory assessment of the child. Again parents also have the right to ask for a statutory assessment of their child.

LEAs must notify parents whether or not they have decided to assess. The Local Education Authority should give you 29 days notice before the assessment begins. This period is to allow parents to respond with information and views to be included as part of the assessment. The assessment usually includes reports from school, an Educational Psychologist, a doctor or health professional and parents.

Contact a Family publishes two factsheets "Special Educational Needs - England" and "Special Educational Needs - Wales" as well as a Disability and Education Pack for England (0.4Mb) which can be downloaded in Adobe Acrobat pdf version 5 format from their website, www.cafamily.org

If a parent is unhappy with a child’s statement, and is unable to resolve the issue with the LEA, then they may wish to appeal. The Special Educational Needs and Disability Tribunal (SENDIST) produce two leaflets entitled: “Special Educational Needs - How to Appeal” and “Disability Discrimination in Schools - How to make a Claim”, as well as a video called: “Right to be Heard”. The Special Educational Needs Tribunal for Wales (SENTW) also produce versions of these two leaflets applicable to provision in Wales.

SENDIST, Procession House, 55 Ludgate Hill, London EC4M 7SW Tel: 0870 241 2555 SEN Helpline Tel: 0870 606 5750 Discrimination Helpline e-mail: tribunalqueries@sendist.gsi.gov.uk Web: http://www.sendist.gov.uk

SENTW, Unit 32, Ddole Road Enterprise Park, Llandrindod Wells LD1 6PF Tel: (01597) 829800 Helpline e-mail: tribunalenquiries@wales.gsi.gov.uk Web: http://www.sentw.gov.uk

Many Local Education Authorities in England have a Parent Partnership Scheme, contact the LEA or Contact a Family helpline for details. In Wales, every County has a similar scheme, which is provided by an independent voluntary organisation. Contact the LEA or the Contact a Family office in Wales.
Leaving School

In England and Wales, during year 9 (or when your child is 14) the annual review of the statement must include the drawing up of a Transition Plan to prepare for post-16 education. If you live in England this review must involve the Connexions Service which offers guidance to young people in the transition to adult life. If you live in Wales, it is Careers Wales which needs to be involved.

Understandably, many parents are concerned about their child’s future after leaving full-time education.

There are however, many colleges of further education that are able to accept disabled students on their mainstream courses. There are also special courses which aim to ease the transition from school to further education or employment. Some courses focus on helping young people towards independent living, including training on home management, personal relationships and use of leisure time.

The Transition Plan should include a comprehensive review of a young person’s needs as they approach adulthood. This should go beyond education and training needs. Parents, young people, their advocates and a cross-section of professional staff should be involved in the process. The Transition Plan must be reviewed annually. Contact a Family produces a factsheet called “Transition in England and Wales.”

Education in Scotland

The educational system in Scotland is currently undergoing major change. The existing special educational needs system will be replaced by the new Education (Additional Support for Learning) (Scotland) Act 2004, due to come into effect on 14th November 2005.

The new Act introduces the wider concept of Additional Support Needs (ASN) and extends the right of extra help to more pupils. Education authorities have a new duty to identify and support all children who would benefit from extra help with their learning. This could be because of any form of disability or if they have social, emotional or behavioural problems, are particularly gifted, English is their second language, are being bullied or have experienced bereavement.

Pupils with enduring, complex or multiple needs and who require services that the education authority cannot provide on their own, e.g. therapy services, will be entitled to a Co-ordinated Support Plan (CSP). The Co-ordinated Support Plan is intended to ensure better co-ordination of services from education, health and social work. Parents will be able to take disputes about Co-ordinated Support Plans to a new tribunal system.

There will be a period of transition to the new system for children and young people who have a Record of Needs immediately prior to the start of the new Act. The education authority will have up to 2 years to decide, following assessment, whether children with a Record of Needs require a Co-ordinated Support Plan. During this time the existing level of support must be maintained. Where it is decided that a Co-ordinated Support Plan is not required, the new Act ensures that current provision is preserved as a minimum for a further 2 years. For some children and young people with a Record of Needs this could mean that the level of support they get in the existing system will be preserved for up to 4 years from the date the Act is commenced.
The new legislation comes into effect on 14th November 2005. Until then current arrangements remain in force.

Further information is available from:

**Enquire, The Scottish Advice Service for Additional Support for Learning** Tel: 0845 123 23 03 Web: http://www.enquire.org.uk

or from Contact a Family Scotland, tel: 0131 475 2608

Many thanks to Contact a Family for allowing us to reproduce this information from their leaflet “When your child has additional needs”, which is available to download from their website, www.cafamily.org

The information is correct at the time of publication, but in view of the constant changes in education legislation, we would ask you to check that the rules are applicable in your area. You can either contact your Local Education Authority or Contact A Family.

**Contact A Family**

Across the UK, a child is diagnosed with a severe disability every 25 minutes and over 98% of disabled children are cared for at home by a parent or other family member who didn’t “apply for the job” but who has quickly had to become an expert.

When parents find out that their child is disabled they feel isolated and alone because usually they don’t know anyone else facing the same problems. They want contact with another family who’ve been through a similar experience and they want information about their child’s condition.

Contact a Family is the only UK-wide charity providing advice, information and support to the parents of all disabled children - no matter what their health condition. We also enable parents to get in contact with other families, both on a local and national basis. Each year we reach at least 250,000 families.

“I have received more information about my child’s condition from Contact a Family in the past week than I have from anyone else in the last 15 years.” Parent

We offer a range of services to support families including a freephone helpline and a number of offices, staff and volunteers around the UK. We also provide information to professionals and developmental advice to support groups. Most of our information materials can be found on this website, others will need to be ordered. We also work to influence services and have campaigns that aim to improve the quality of life for families with disabled children.

**Contact a Family, 209-211 City Road, London EC1V 1JN** Tel: 020 7608 8700 Fax: 020 7608 8701 Helpline 0808 808 3555 or Textphone 0808 808 3556 Freephone for parents and families (10am-4pm, Mon-Fri) e-mail: info@cafamily.org.uk

Registered Charity Number 284912

“Working to support those who are affected by Charcot-Marie-Tooth Disease, also known as Hereditary Motor and Sensory Neuropathy or Peroneal Muscular Atrophy” 157
Appendix 4

Forum of Mobility Centres - List of All Centres

Belfast
Disability Action, Portside Business Park, 189 Airport Road, Belfast, BT3 9ED
Tel: 028 9029 7880, Fax: 028 9020 7881 Email, mobilitycentre@disabilityaction.org

Birmingham
Regional Driving Assessment Centre, West Heath Hospital, Rednal Road, Birmingham, B28 8HR
Tel: 0121 627 8228, Fax: 0121 627 8629 Email: paula.beagan@southbirminghampct.nhs.uk

Bristol
Mobility Service of the Disabled Living Centre (West of England), The Vassall Centre, Gill Avenue, Fishponds, Bristol, BS16 2QQ
Tel: 0117 965 9353, Fax: 0117 965 3652 Email: mobserv@dlsbristol.org

Bodelwyddan
North Wales Mobility and Driving Assessment Service, Disability Resources Centre, Glan Clwyd Hospital, Bodelwyddan, Denbighshire, LL18 5UJ
Tel: 01745 584858, Fax: 01745 535042 Email: alexbarr@btconnect.com

Cardiff
South Wales Mobility and Driving Assessment Service, Rookwood Hospital, Fairwater Road, Llandaff, Cardiff, CF5 2YN
Tel: 029 2055 5130, Fax: 029 2055 5130 Email: helen@wddac.co.uk

Carshalton
Queen Elizabeth’s Foundation Mobility Centre, Damson Way, Fountain Drive, Carshalton, Surrey, SM5 4NR
Tel: 0208 770 1151, Fax: 0208 770 1211 Email: info@mobility-qe.org Web: www.qefd.org/mobilitycentre/
Crowthorne
Mobility Advice and Vehicle Information Service (MAVIS), Crowthorne Business Estate, Old Wokingham Road, Crowthorne, Berkshire, RG45 6XD
Tel: 01344 661000, Fax: 01344 661066  Email: mavis@dft.gsi.gov.uk  Web: www.dft.gove.uk/access/mavis

Derby
DrivAbility (Derby Regional Mobility Centre), Kingsway Hospital, Kingsway, Derby, DE22 3LZ
Tel: 01332 371929, Fax: 01332 382377  Email: driving@derbyhospitals.nhs.uk  Web: www.drmc.uk.com

Edinburgh
Scottish Driving Assessment Service, Astley Ainslie Hospital, 133 Grange Loan, Edinburgh, EH9 2HL
Tel: 0131 537 9192, Fax: 0131 537 9193  Email: marlene.mackenzie@lpct.scot.nhs.uk

Leeds
The William Merritt Disabled Living Centre and Mobility Service, St Mary’s Hospital, Green Hill Road, Armley, Leeds, LS12 3QE
Tel: 0113 305 5288, Fax: 0113 231 9291  Email: mobility.service@nhs.net  Web: www.williammerrittleeds.org

Maidstone
DART Driving Assessment and Advice Centre, Cobtree Ward, Preston Hall Hospital, London Road, Aylesford, Kent, ME20 7NJ
Tel: 01622 795719, Fax: 01622 795720  Email: janicestannard@nhs.net

Newcastle upon Tyne
The Mobility Centre, Regional Neurological Rehabilitation Centre, Hunters Road, Newcastle upon Tyne, NE2 4NR
Tel: 0191 219 5694, Fax: 0191 219 5693  Email: mobilitycentrenewcastle@nap.nhs.uk  Web: nap.nhs.uk/snrs

Oxford
Oxford Mobility Centre, c/o Regional Driving Assessment Centre, West Heath Hospital, Rednal Road, Birmingham, B38 8HR
Tel: 0121 627 8228, Fax: 0121 627 8629  Email: paula.beagan@southbirminghampct.nhs.uk
Charcot-Marie-Tooth: A Practical Guide

Thetford
Kilverstone Mobility Assessment Centre, 2 Napier Place, Thetford, Norfolk, IP24 3RL
Tel: 01842 753029, Fax: 01842 755950 Email: mail@kmacmobil.org.uk
Web: www.kmacmobil.org.uk

Truro
Cornwall Mobility Centre, Servicing the South West Peninsula, Tehidy House, Royal Cornwall Hospital, Truro, Cornwall, TR1 3LJ
Tel: 01872 254920, Fax: 01872 254921 Email: enquiries@cornwallmobilitycentre.co.uk
Web: www.cornwallmobilitycentre.co.uk

Welwyn Garden City
Hertfordshire Action on Disability Mobility Centre, The Woodlands Centre, The Commons, Welwyn Garden City, Hertfordshire, AL7 3DD
Tel: 01707 324581, Fax: 01707 371297 Email: driving@hadnet.co.uk Web: www.hadnet.org.uk

Wigan
Wrightington Mobility Centre, Wrightington Hospital, Hall Lane, Appley Bridge, Wigan, Lancashire, WN6 9EP
Tel: 01257 256409, Fax: 01257 256538 Email: mobility.centre@alwpct.nhs.uk

List from the Forum of Mobility Centres website - www.mobility-centres.org.uk
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  - Margaret Read
  - Ian Oakley
  - Howard Cohen
  - Wendy Bailey
  - Karin Fletcher
  - Susan Salzberg

- Dr Henry Houlden, Senior Lecturer in Neurology, Institute of Neurology, London, who kindly reviewed the entire book.

- CMT United Kingdom’s board of trustees

General medical and genetic advice on CMT

- Dr Mary Reilly, Consultant Neurologist and Honorary Senior Lecturer, Centre for Neuromuscular Disease and Dept. of Molecular Neurosciences, National Hospital for Neurology and Neurosurgery and Institute of Neurology, London.

- Dr David Hilton-Jones, Clinical Director of the Oxford MDC Muscle & Nerve Centre and consultant neurologist at the Radcliffe Infirmary and honorary senior lecturer in the University of Oxford.

- Mary Davis, senior geneticist, University College London Hospital.

Exercise and stretching

- Gita Ramdharry, Research Physiotherapist at the Institute of Neurology, University College Hospital.

- Liz Dewar, Senior Physiotherapist at The National Hospital for Neurology and Neurosurgery, Queen Square, London.
Orthotics

- Paul Charlton, Senior Orthotist at Peacocks Medical Group and Vice Chairman of the UK members section of the International Society for Prosthetics and Orthotics.

Occupational Therapy

- Pip Wilford, Clinical Specialist Occupational Therapist, National Hospital for Neurology and Neurosurgery, London.

Healthy eating

- Erin Dooley, Senior Neurosciences Dietitian at University Hospital Birmingham NHS Trust.

Surgery and anaesthetics

- Mr Dishan Singh, Consultant Orthopaedic Surgeon, Foot and Ankle Unit, Royal National Orthopaedic Hospital, Stanmore, Middlesex.
- Dr Michael Hetreed, Consultant Anaesthetist, Royal National Orthopaedic Hospital, Stanmore, Middlesex.

Podiatry

- Richard Leigh, Chief Podiatrist, University College Hospital, London.
Your Notes