cmt United Kingdom

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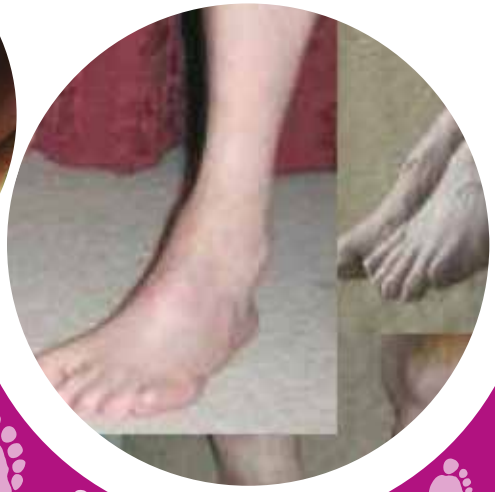
cmt:
a practical guide



cmt: a practical guide

Written and researched by Resonant Media Ltd

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Together we are stronger

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While every effort has been taken to make sure that the information in this book is accurate, neither the author nor CMT United Kingdom can be held responsible for incorrect entries. There are constant changes and developments in medicine and treatments available; we advise you to check with your health and social care professionals to make sure you receive the best treatment and care suitable for you.

Please help us keep this leaflet up to date and tell us if there are any mistakes, changes or new information that you think should be included in future editions of this book.

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Table of Contents

Preface

As chairman of CMT UK, I feel privileged to be given the opportunity of writing the preface to the new edition of such a valuable, highly-praised and informative book.

Charcot-Marie-Tooth Disease: A Practical Guide was first written some 14 years ago and updated and revised in 2007. Since then, our member database has increased to well over 3,000 and our enlarged website has attracted many visitors, garnering a considerable number of complimentary comments.

The Board is - more than ever - committed to supporting those affected by CMT, creating awareness among the general public and providing information to many others who work in the medical and clinical professions.

It is this commitment, together with the availability of an ever-growing pool of knowledge and results from recent research, which led to the decision to completely revise and re-issue the Practical Guide.

The success of our declared Strategic Plan to raise £2 million over the next five years will, to a large extent, depend upon the availability of up-to-date and easily assimilated information on the disease. Accordingly, the Board decided last year that this revision should proceed as quickly as possible.

The style and content have again been led by members of our National Support Groups – many of whom have direct experience of living with the condition and finding solutions to the problems it poses.

I am particularly grateful to our patron, Professor Mary Reilly, who has spent many hours collating a huge amount of medical knowledge and documenting it in a manner which is easily readable and uses a minimum of medical terminology (some will always be necessary!). Thanks also must go to John Isitt and his staff at Resonant Media who have produced a document which we believe is likely to be the definitive textbook on CMT for some time to come.

Richard Ouston
2014

Foreword

Charcot-Marie-Tooth Disease (CMT) is a condition that causes deterioration of the peripheral nerves controlling sensory information and muscle function in the feet, lower legs, hands and forearms. It can lead to foot problems such as high arches, foot-drop, muscle 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 very rarely life-threatening and does not usually affect life expectancy.

Coping with any medical condition can be daunting. This is especially true when there is little information available, even amongst doctors, as is the case with CMT. CMT is the most common inherited neurological condition, affecting one person in 2500 worldwide. Despite this, there is widespread ignorance about the condition and a diagnosis can be slow in coming, making life unnecessarily difficult for people with 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 CMT, to diagnosis, the latest treatments, and advice on coping and managing the condition day to day.

Professor Mary M Reilly MD FRCP FRCPI

Professor of Clinical Neurology and Consultant Neurologist
MRC Centre for Neuromuscular Disease
London

Chapter 1: What is CMT?

What is CMT - the basics

CMT is a genetic condition that damages peripheral nerves. These nerves are responsible for passing on commands from the brain to the muscles (motor nerves) and for passing information to the brain about sensations, such as pain, heat, cold, touch, importantly for balance - where your joints are in space (sensory nerves). When these are damaged, people are said to have a neuropathy.

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

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

Other key points about CMT are that the condition:

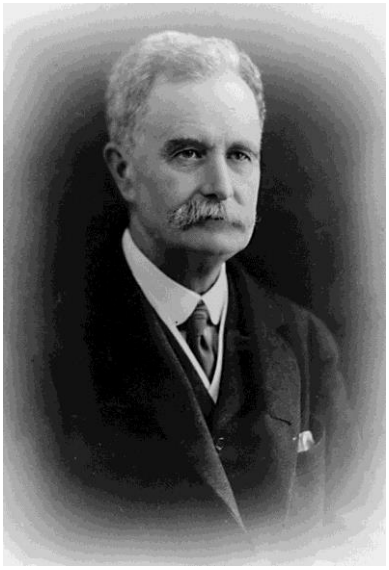
- is hardly ever life-threatening, but often becomes slowly worse over the years
- comes in many forms, some of which are much more severe than others
- can affect people very differently, even in the same family
- can cause the muscles in the foot, lower leg, hand and forearm to become wasted and weak
- can cause foot drop gait, foot bone abnormalities (e.g. 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)
- can be passed on from parent to child: the precise way it is passed on to subsequent generations depends on the genes involved
- affects all ethnic groups throughout the world
- is the focus of much research, bringing us close to answering the CMT enigma.

What's in a name?

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

Jean Martin Charcot

Pierre Marie



Howard Tooth

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 found in some medical literature.

The symptoms of CMT

The exact symptoms you experience may vary hugely from other people with CMT. This is because there are many different subtypes of CMT. Symptoms can also vary hugely, even within the same sub-type, for example between close family members.

While some subtypes have specific symptoms, some symptoms are common to most types of CMT. These include wasting, weakness and reduced sensation starting in the feet/legs and eventually involving the hands/arms.

Occasionally people develop no symptoms even though they carry the abnormal gene. Others may only get symptoms later in life, even in their 50s and 60s, despite having a type that 'normally' shows symptoms by the teenage years.

Early symptoms may include:

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

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 or do fiddly things such as doing up buttons. Weakness in the hands causes difficulty with grip, making it difficult to 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
- hearing difficulties - usually mild and coming on later in life

People with CMT can develop more severe symptoms, though this is rare for those with the commonest type. These can include:

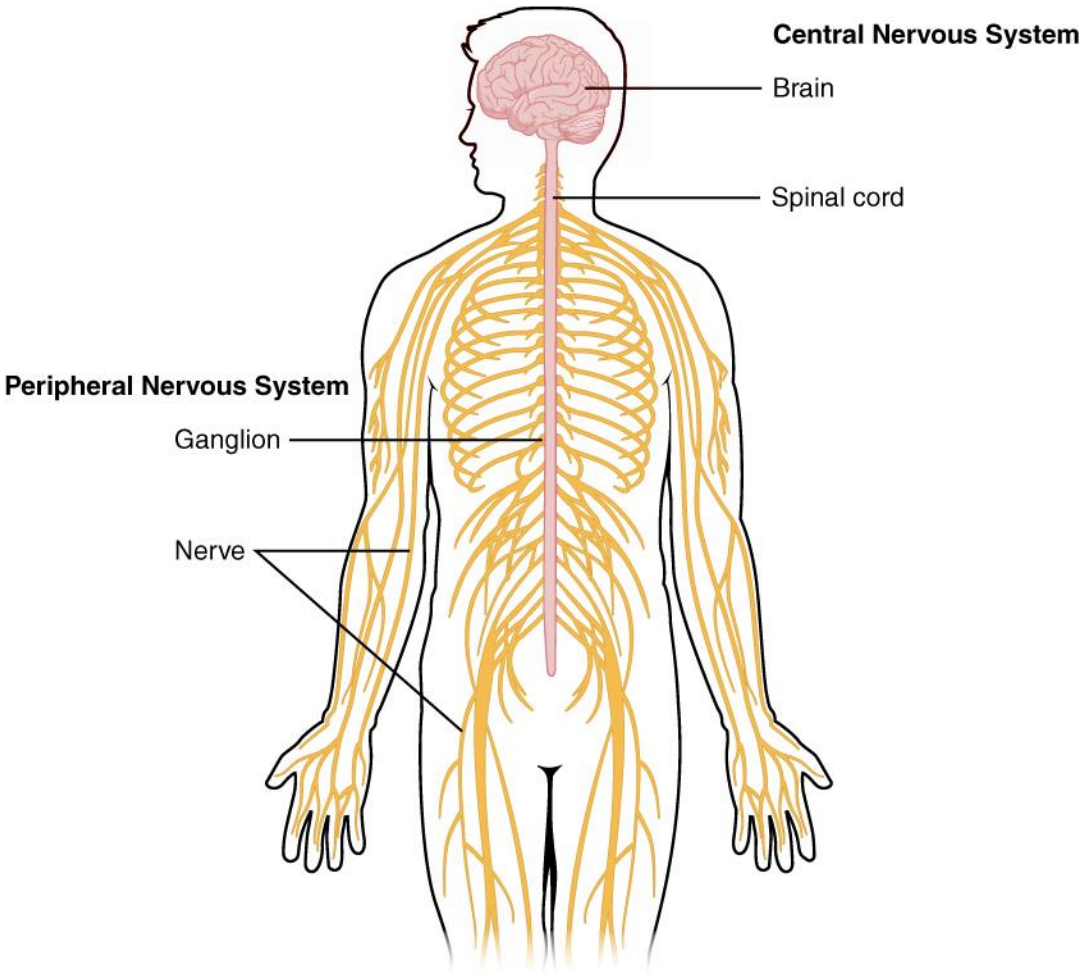
- a severe curve of the spine (scoliosis)
- speech and swallowing difficulties
- some difficulty breathing, particularly at night (sleep apnoea)
- voice difficulties making speech quieter

In some rare forms of CMT a wheelchair may be needed, particularly later in life.

What causes CMT?

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

In order for you to move with speed and precision, messages must 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.

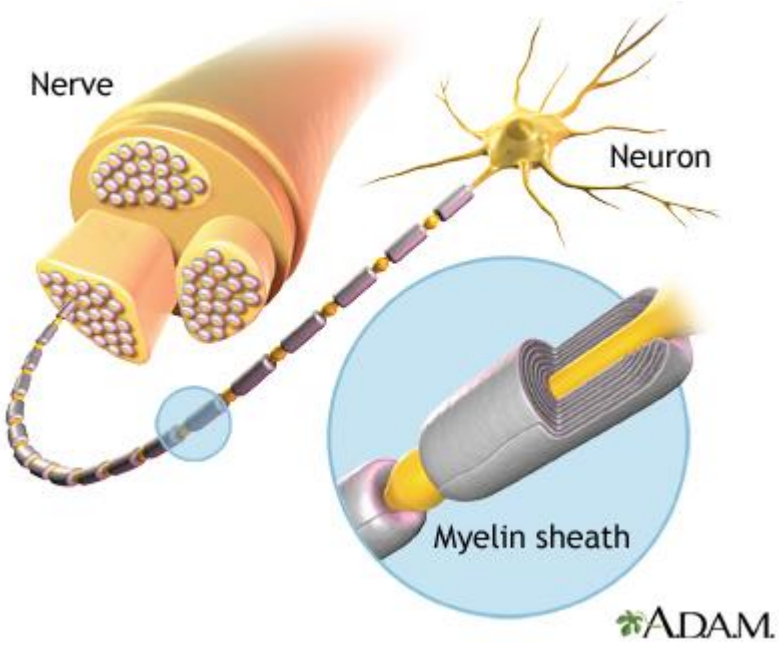


The nerves in your arms and legs, called peripheral nerves, can be compared to electrical cables. The central 'wire' is known as the axon and the 'plastic outer' is called the myelin sheath.

Axons transmit the electrical signals to and from the brain, and the 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: if the signal is slowed down, the axon is eventually damaged as well.

Damage to the axon (whether the problem started in the axon or initially in the myelin) 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 of November 2014, 80+ 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.

For more on the genetic causes of CMT see Chapter 2, Genes: what they mean for you.

The different types of CMT

There are two main types of CMT:

Demyelinating (CMT1) – this form of CMT affects the myelin sheath that insulates the central axon.

Axonal (CMT2) – this form of CMT affects the central axon.

Sometimes, when it is difficult to know if the problem is mainly in the myelin (CMT1) or in the axon (CMT2), it is called intermediate CMT. Depending on the type of genes involved, it may be known as DI-CMT or CMT4.

You may also have heard of other types of CMT, including CMT3, or Dejerine-Sottas Disease. This terminology is not only confusing but is also constantly changing as new genes are identified. 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. The basic division into CMT1 and CMT2 has stood the test of time.

Demyelinating CMT (CMT1)
CMT1 is the most common form of CMT, (six out of ten people with the condition will have CMT1A) affecting the nerve's insulating myelin sheath. People with CMT1 will usually notice symptoms developing in childhood or adolescence, typically between the ages of five and 15.

Some forms of CMT1 have an earlier age of onset and these forms used to be called CMT3 in the old classification, but are now more commonly referred to as severe CMT1. Two of these forms are sometimes referred to as Dejerine Sottas Disease (DSD) and Congenital Hypomyelinating Neuropathy (CHN) but these are both forms of CMT1.

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, but CMT2 can start earlier or later and may appear in the first five years of life.

CMTX1
CMTX1 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 in late childhood or adolescence. The nerve conduction studies

look like CMT1. For more on nerve conduction tests see Diagnosis p xx Women may be completely unaffected - in which case they tend to be known as 'carriers' (they can pass this gene onto their children) - or they may be affected but usually not as severely as males. The results of their nerve conduction studies usually look like CMT2.

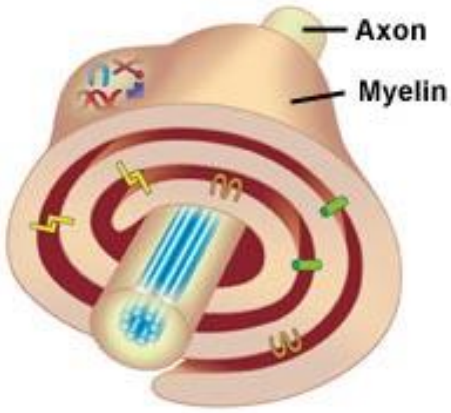
DI-CMT
DI stands for 'dominant intermediate'. This form of CMT is rare and usually has similar symptoms to the common versions of CMT1 and CMT2. It is called intermediate because both the myelin sheath and the axon are damaged equally.

For both CMT1 and CMT2 there are many different subtypes and you may hear about CMT1A or CMT2A, for example. These are all different genetic variations of the main types.

For more on the different types of CMT and the genetic background to each, see 23, Genes: what they mean for you p xx.

CMT – the axon and myelin sheath

Some more information about how the myelin sheath and the axon work together may be helpful at this point.



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 damage to the myelin sheath continues, the axon is damaged as the myelin breaks down. 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 the symptoms of CMT. In fact you can live

with slow nerves for decades with no symptoms or signs 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 a medical history, examination and nerve conduction tests can usually tell you whether you have CMT1 or CMT2. An accurate diagnosis is only possible with a genetic test, but not all the genes that cause the different types of CMT have been found, though there have recently been huge advances in the technology used to look for new genes.

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 the type of CMT you have, but rather on your particular needs. This is likely to change in the future as new treatments are likely to be gene-specific which means each treatment will only benefit people with certain genetic types of CMT.

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

- It can help give you a better idea of the course of your CMT in the future. Specific genes are linked to a spectrum of probable symptoms and indicate how the condition is likely to change over the years. For example, teenagers trying to make a career choice could get a better understanding of how CMT will affect them in 10 or 20 years' time.

- It can prevent you having unnecessary tests, such as nerve biopsies or lumbar puncture, to rule out other possible causes of neuropathy.

- It can help you understand how likely it is that any children you have will inherit CMT. For more on this, see Chapter 3, Genes: what they mean for you.

- If medical trials to test treatments for CMT take place in the UK, it is likely that only people with certain genetic causes of CMT will be able to take part (e.g. the recent trial of vitamin C in the UK was only for people with CMT1A).

You can get a genetic test by asking your GP to refer you to a neurologist or a neurogenetics clinic.

Of course, some people may not want a genetic diagnosis, perhaps because there are no treatments currently available specifically for any genetic type of CMT. It is your choice and this should be respected by the medical practitioners involved in your diagnosis and treatment.

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. Problems usually start in the feet as the nerves to the feet are the longest in your body. They can then affect the hands.

> **Muscle wasting** (loss of muscle mass) and weakness (loss of muscle power), usually first noticed in your feet and later in your 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.
>
> **Loss of sensation** again usually starting in your feet and later in your hands, although this is often not noticed until it is severe or has caused skin problems.

Muscle wasting and weakness

As the muscles become wasted and weak two problems arise in addition to the weakness:

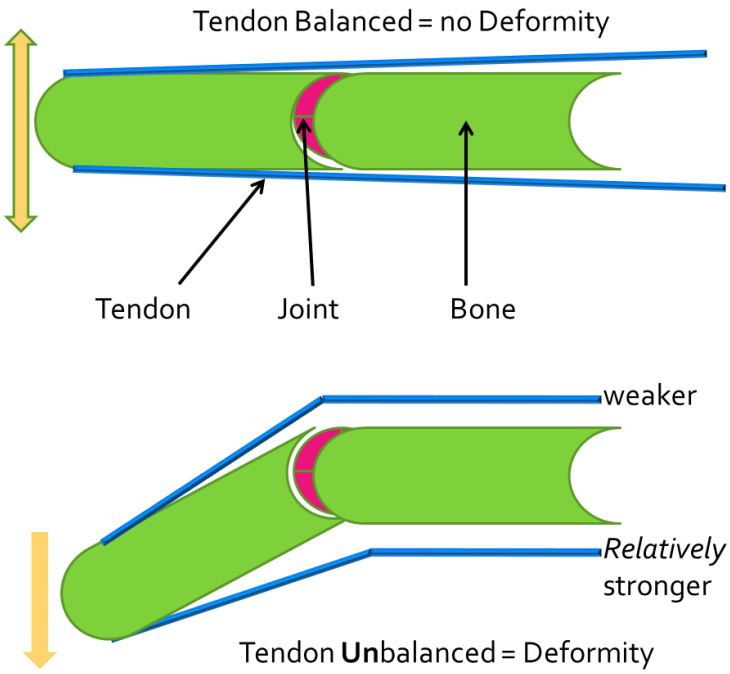
- contractures
- strain on other muscle groups

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 body-builder'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.

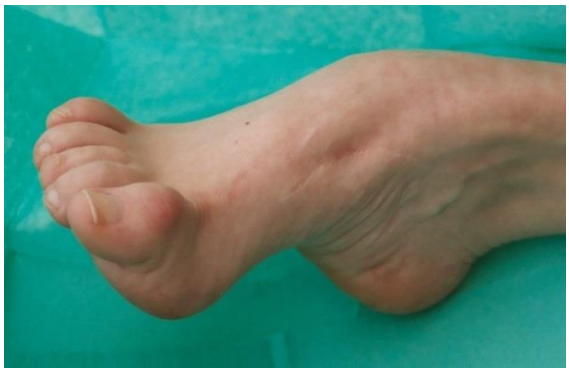
Muscles have a natural tendency to contract and tighten, only being stretched by their opposing muscle. Problems arise when a muscle weakens: the opposing muscle that continues to function is not stretched by this weak muscle, allowing the stronger muscle to become tighter and shorter. This mismatch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

One of the most important goals 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. For more on this see Chapter 3, Managing your CMT: Stretching, exercise and physiotherapy.



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

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

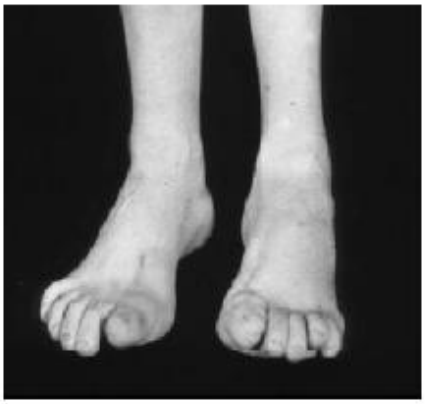


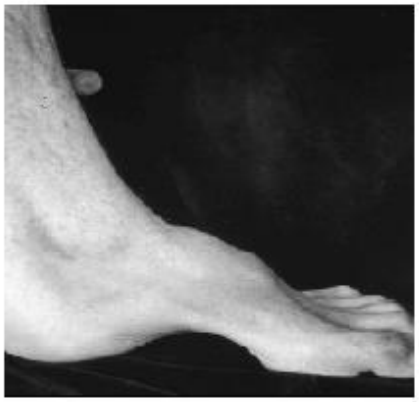
In CMT the most common changes are found in the foot and ankle, due to wasting and weakness of the shin muscles. The foot is mainly controlled by the shin muscles - at the front of your lower leg - which pull the foot up, and the calf muscles - large muscles at the back of your lower leg - which pull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

Because of weakness in the shin muscles, people develop foot drop as it becomes harder for the

shin muscles to pull up the foot. 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 known as heel varus.

At the same time, the Achilles tendon at the back of the foot and the 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. One example is the increased strain on the back during walking caused by weak legs, which frequently leads to back pain in people with CMT.

Loss of sensation

Because CMT damages the sensory nerves, many people with the condition 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 complete numbness, it can make people with CMT less likely to notice problems such as splinters, cuts to the feet or accidental burns – for example hot cups burning the hand or hot water bottles scalding the feet.

As well as loss of feeling (numbness), damage to the sensory nerves affects your awareness of your joint position and the 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 partly 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: many people with CMT have far poorer balance with eyes shut or in the dark. You may hear this referred to as lack of 'proprioception' – or knowing where your body is in space.

Cold feet are a common complaint of people with CMT. This is probably due to a combination of factors including lack of muscle bulk, lack of movement and sensory loss. Although blood vessels in the feet are not damaged in CMT, people often experience leg swelling caused by weak muscles being unable to properly pump up the blood.

Many people report a worsening of symptoms with cold weather, such as difficulties in doing buttons and zips or turning keys. We do not fully understand why this happens, but it may be related to slowing of nerve signals in low temperatures.

Summary of problems due to muscle wasting/weakness and sensory loss

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

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.

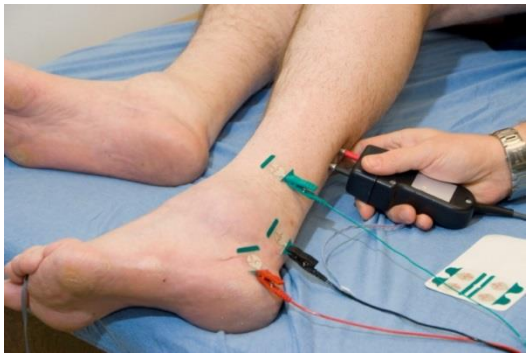
Steps to a diagnosis

The neurologist will follow a number of steps to confirm whether you have the condition or not. The order of these steps may vary and some people will receive a diagnosis without needing to go through the whole process. Technology is changing rapidly, so the steps below should be seen as a guide rather than a blueprint.

1. Your doctor will take a standard patient history, including a family history. He or she will ask 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 noted: these may be seen or felt through the skin.

3. If there is an established family history of CMT where a family member has already received a confirmed genetic diagnosis, then clinical assessment alone may be all that is needed to make a diagnosis. Sometimes you will be offered a genetic test for the most common form of CMT (type 1A). This might be the case where there is clear family history with typical signs of CMT, or where there are a long waiting lists for electro-diagnostic tests.



The next stage may be electro-diagnostic tests. These consist of two parts:

Nerve conduction tests. Electrodes are placed on your skin over your peripheral motor or sensory nerves, usually on the legs These electrodes will give you a mild electrical shock, which may be uncomfortable - or occasionally painful. 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.

Electromyography (EMG). A needle electrode is inserted through your skin to measure the electrical activity of the muscles in the legs.

Electrodiagnostic tests help doctors to distinguish between different types of CMT. If there is a test result for another member of your family with CMT, it may not be necessary for you to have an electrodiagnostic test.

In the UK, the next stage of diagnosis will be a genetic test to try to confirm 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.

In very rare circumstances, when there is doubt about the precise diagnosis, nerve biopsy may be advised. This involves removing a small piece of peripheral nerve through a cut in the skin, usually from the calf, under local anaesthetic. The nerve is then examined in a laboratory for any signs of abnormalities.

For more on genes and genetic testing, see Chapter 2, Genes: what they mean for you.

What should happen now?

Once you have been diagnosed, certain things should happen.

You may find it useful to print out this list and talk it through with your neurologist and GP to make sure that you get the care you deserve.

- You should receive a copy of your clinic letter. Ideally this will be phrased in such a way that you can show it to other family members. This will help them understand your diagnosis and why it would be a good idea for them to see a specialist too. For more on telling people about CMT, see Chapter 3, Managing your CMT: Talking about CMT
- Appropriate follow-up arrangements should be offered to you.
- Find out as much as you can about CMT. A good source of information is the CMT United Kingdom website (cmt.org.uk). For more on reliable sources of health information see Chapter 5, Organisations that can help you.
- Get a referral to see a physiotherapist with a good understanding of neuromuscular conditions and also to see an orthotist (a specialist in the use of mobility devices) if necessary.

- Find out what to do if you discover other, new symptoms and how to check whether or not these symptoms are connected to CMT.

As part of your follow-up you should be given information on:

- CMT United Kingdom
- Disability Living allowance for a child under 16, or Personal Independence Payments for adults between 16 and 65.
- Mobility issues, especially the legal requirement to inform the DVLA if a diagnosis of CMT is confirmed.
- Where to find assistance and support with schooling, college, etc. including extra time in exams, for example.
- Occupational help, if necessary.

Unfortunately, in the real world, your diagnosis may be given to you by someone who does not know much about CMT. You may be told that CMT is a progressive condition, with no cure and no treatments. This is unhelpful and quite wrong, although it is true that there is no cure at the present time.

CMT can be well managed and well treated. But you may need to knock some heads together to get the care you deserve. If you are struggling to get the treatment you need, contact CMT United Kingdom.

Reacting to your diagnosis

Everyone reacts differently when told they or their child has CMT. Nobody – not your doctor, partner, family or friends – can tell you what you should or should not be feeling or how you should react.

The important thing to remember is that an 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 of the condition for years before discovering the underlying cause. Learning that there is a name for what you have been experiencing can help you to understand and come to terms with your symptoms. Some people call this 'closure' – a sense of relief on diagnosis that the problems you have been living with for years are not symptoms of something more serious.

You may find it helpful to follow these five steps.

Take the time you need
Do not rush into 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 all the time you need to make decisions can help you to:

- feel less anxious and stressed
- avoid depression
- cope better with CMT
- feel more in control

Get the support you need
When you are ready, talk with your family and friends. Some people find it helpful to chat to other people with CMT: they are going through the same thing as you and 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. Visit the charity's Facebook page (www.facebook.com/cmtuk) to talk to other people coping with the condition.

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

For more on how specialist healthcare professionals can help you, see Chapter 3, How to Manage your CMT: getting the right treatment.

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 decisions about their own healthcare.

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. For more on good sources of evidence-based health information, see Chapter 3, How to Manage your CMT: getting the right information p xx.

Ten questions to ask your doctor after diagnosis

1. What is the technical name for my specific condition, and what does it mean in plain English?
2. What is my outlook for the future (prognosis)?
3. Will I need any additional tests, and if so, what kind and when? When can I get a genetic test? What about testing my children?
4. What are my treatment options and how soon do I need to make any decisions about treatment?
5. What are the pros and cons of my treatment options (including risks of not having the treatment)?
6. When can I see a specialist physiotherapist who understands neurological conditions?
7. Is there a current research study (clinical trial) that I could benefit from?
8. What changes – if any – 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?

Myths and facts

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 is closely related to CMT but it differs from CMT as affects only 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.

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, most commonly caused by diabetes, leading to a damaged and swollen joint. People with CMT may also suffer from Charcot's joint, especially if their sensory nerves are mainly affected.

Muscular dystrophy
It is a common mistake to think of CMT as a muscular condition. Despite muscle weakness in the legs and arms, CMT does not directly affect the muscle, unlike muscular dystrophy (MD), which is caused by a specific protein being missing in the muscle fibres. CMT is a neuropathic condition, meaning that it damages the nerves. This damage to the nerves then leads to weakness in the muscles, because of the lack of stimulation by the nerves.

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 affects only 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. In MS, no single cause has been identified: there appears to be some genetic link, but it seems that MS is triggered by something like a virus or an allergy.

What is affected in CMT?

It is easy to be confused about which parts of the body are affected by CMT. The list below should make things clearer.

Yes

Eyes: sometimes in CMT2A but not usually significantly

Ears –rarely, in some very rare types only. [Remember deafness is common in the general population anyhow]

Inner ear (balance) – rarely. Most balance issues are related to problems in the feet, rather than the inner ear.

Swallowing – not usually affected

Vocal cords – vocal cord paralysis is a rare complication of severe forms of CMT.

Tongue/Facial muscles – Tongue atrophy and facial weakness are occasionally seen in some forms of CMT

Breathing – diaphragm weakness is a definite and documented, but rare, complication of CMT. The phrenic nerve, which activates the diaphragm can become affected, and can lead to breathing difficulties, especially when lying flat. However, these problems are rarely severe, and can be managed. They tend to affect people who are severely affected by CMT, especially those with upper arm weakness. Sleep Apnoea can also be associated with CMT An excellent video explaining the mechanics of breathing, and how it can be affected by CMT, made by CMTA, can be found at http://bit.ly/cmtbreathing

Hands – very common

Back/spine problems / scoliosis – can be caused by walking badly. Scoliosis seems to be more common in people with CMT but rarely needs corrective surgery.

Hips – wear and tear issues may cause 'arthritis-type' problems, necessitating hip replacements. Hip dysplasia also seems to be slightly more common in children with CMT.

Knees – again, wear and tear causes problems with knees. Dislocations seem more common.

Feet and ankles – very common.

Definitely not affected by CMT

Brain

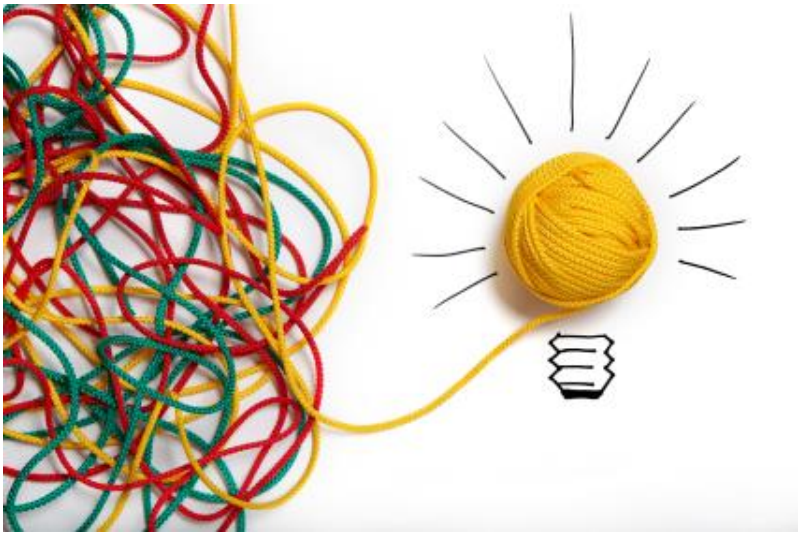
Heart

In CMT, muscles undergo atrophy and the heart is, of course, an important muscle. In addition, nerves are involved in the electric wiring of the heart. Nonetheless, while cardiac problems are a major concern in muscle disorders, cardiac involvement does not occur in CMT. This is why we can confidently state that life expectancy in classical CMT does not differ from the general population.

Other internal organs such as stomach, bladder and bowel (or no evidence of being affected)

Erectile issues

Uterus



Other conditions closely related to CMT

CMT is a motor (wasting and weakness) and sensory (reduced sensation) neuropathy (neuropathy is a term for any disease affecting the nerves). It is closely related to two other rarer inherited neuropathies:

- the hereditary sensory neuropathies (HSN), also called hereditary sensory and autonomic neuropathies (HSAN), that only or mainly involve sensory nerves
- the distal hereditary motor neuropathies (HMN), in which there is only motor involvement.

These three disorders (CMT, HSN and HMN) are often collectively termed CMT and related disorders.

Hereditary Neuropathy with Liability to Pressure Palsy (HNPP) is a condition closely related to CMT where people are at risk of developing damage to their nerves at points of pressure e.g. elbows and knees. It is related genetically to the common form of CMT, CMT1A. For more on this, see Chapter 2, Genes: what they mean for you.

Hereditary Sensory Neuropathy (HSN)

HSN (also called Hereditary Sensory and Autonomic Neuropathy or HSAN) is much rarer than CMT.

Although there have been 15 genes identified as causing HSN, most of the causative genes have not yet been identified. This means many people with this condition do not have an accurate genetic diagnosis.

People with HSN have much more sensory involvement than motor involvement. As a result, they generally develop problems with reduced feeling in their feet first rather than wasting and weakness.

In some forms there is no wasting and weakness: in other forms it comes later than the reduced sensation. The sensory loss is usually more severe than in CMT and people frequently develop complications due to lack of sensation.

These complications include ulcers which may become infected. If not treated quickly, skin infections can spread to the bones (osteomyelitis). When this happens there is a risk that part of the bone may need to be surgically removed (amputation). It is very important that people with HSN are educated on how to carefully look after and protect their skin.

In some rare forms of HSN, the autonomic nervous system can be involved. The autonomic nervous system is responsible for the automatic control of certain functions including blood pressure, heart rate and sweating. If this is impaired, fluctuations in blood pressure or lack of sweating (anhidrosis) may develop.

Distal Hereditary Motor Neuropathy (HMN)

Distal HMN is also much rarer than CMT. Symptoms are similar, except there is no reduced sensation. When examined, people with Distal HMN show normal sensation: nerve conduction studies of the sensory nerves are also normal.

So far, 18 genes have been identified as causing distal HMN but most have yet to be identified. As a result, most people with distal HMN do not get an accurate genetic diagnosis.

People with HMN usually present in a very similar way to those with CMT, in that they develop difficulties in walking due to foot drop caused by wasting and weakness of their muscles. As with CMT, people can develop symptoms at any age but it is common to see the first symptoms between 10 and 30 years of age. In some rare forms of distal HMN, wasting and weakness in the hands develops long before the feet are affected. Some people with HMN only ever have symptoms in their hands.

Hereditary Neuropathy with Liability to Pressure Palsy (HNPP)

Although once thought to be an entirely different condition to CMT, HNPP is now known to be genetically very similar. 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 copies of the gene.

The symptoms of HNPP, however, are usually quite different. Most people get occasional attacks of numbness with or without weakness (palsy) involving parts of the arms or legs. These attacks are usually transient, which means they last only a short time, although this can vary from minutes to weeks depending on how severe the attack is.

The reason the condition is called Hereditary Neuropathy with Liability to Pressure Palsy is that an attack is usually triggered by pressure on an individual nerve. Certain nerves just under the skin are more vulnerable to these attacks as it is easy for them to come under pressure. An example is the ulnar nerve at the elbow. It is very common for people who work at a desk to rest their elbows on the surface for long periods. This can cause a

transient ulnar pressure palsy where people get numbness in the 4th and 5th fingers and some weakness of the hand muscles.

Another commonly-affected nerve is the peroneal nerve near the knee. This is vulnerable to pressure if people sit with their knees crossed for prolonged periods. When this nerve is damaged it causes a transient foot drop with numbness.

As well as attacks like these, people with HNPP also often have frequent minor attacks of numbness lasting seconds or minutes e.g. numbness in their hands if they sleep awkwardly at night or if they hold a mobile phone too long. Occasionally people who experience multiple attacks can suffer permanent damage to their nerves with some permanent weakness and reduced sensation, most commonly in the hands and feet. Other permanent symptoms from damaged sensory nerves include pins and needles, which can be painful.

Activities that may bring on palsy include:

- crossing legs at the knee, sitting with legs crossed, sitting tailor-style
- leaning on elbows
- sitting slightly askew in a chair
- sitting 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 such as a handbag, suitcase or – especially – a loaded plastic grocery bag
- 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 - even one inch heels - can make toes numb
- painting too long (holding brush or roller) or painting above head
- walking for more than an hour
- riding a bicycle or motor bike can cause hand symptoms
- certain contact sports
- specific sporting activities e.g. parachuting or bungee jumping
- carrying a heavy rucksack
- lifting weights
- using a computer mouse
- 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, managing the primary symptoms can prevent secondary complications. It is very important for people with HNPP to be educated - usually by a specialist neurophysiotherapist - about how to avoid excess pressure on their nerves.

If you are undergoing surgery and likely to be in one position for a long time under anaesthetic, tell the anaesthetist about your condition. Similarly, you should tell your midwife if you have HNPP to avoid being in the same position too long during labour.

Other helpful measures include:

- pain management - medication and counselling
- occupational therapy - work and home adaptations, e.g. kitchen devices, proper mattresses and pressure relieving devices
- orthotic devices - to help with mobility difficulty caused by foot problems
- wrist and arm splints/support
- physical therapy – to help maintain mobility. Ideally, find a physiotherapist with a particular interest in neuromuscular conditions.

Surgery to relieve pressure can sometimes – but not always – be useful in HNPP. Each case should be assessed individually, ideally by someone with expertise in this condition.

Much of the information in Chapter 3, Managing your CMT, may be helpful for people with HNPP.

How common is HNPP?

Many people with HNPP remain undiagnosed so it is difficult to assess how common it is: studies suggest it affects between seven and 16 per 100,000 people.

For more information about HNPP contact www.hnpp.org

Chapter 2: Genes: what they mean for you

Genes: What they mean for you

The human body is made up of billions of cells, possibly as many as 10,000 trillion or 10,000,000,000,000,000. They range in size from less than 20microns wide - about two-hundredths of a millimetre - to over a metre, in the case of the filaments of nerve cells.

How your cells grow and function is determined 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. People should have two copies of the PMP-22 gene, one each inherited from their mother and father. In people with CMT1A one copy of the gene PMP-22 is doubled up (duplicated) so that people have three copies of this gene in total. In another related condition, HNPP, one copy of the PMP-22 gene is lost (deleted) so that people have only one copy of this gene. Because the balance between the products of this gene and other genes important in the myelin sheath is disturbed, the myelin sheath does not work properly, leading to the symptoms of CMT.

Passing on your 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 46 chromosomes in 23 pairs.

In contrast, the sperm and egg cells each only have a single set of chromosomes. When a sperm cell fertilises an egg, the two sets 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. Not all faulty genes will have an effect. It all depends on the inheritance patterns of that single gene.

Inheritance patterns

Dominant (autosomal dominant)

Some faulty genes are dominant, which means that you only need one faulty gene to have CMT. In effect, the dominant gene over-rules 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.

Dominant inheritance (AD) (Most types of CMT1 and CMT2)

- One parent with CMT = one in two chance child will have CMT whether male or female

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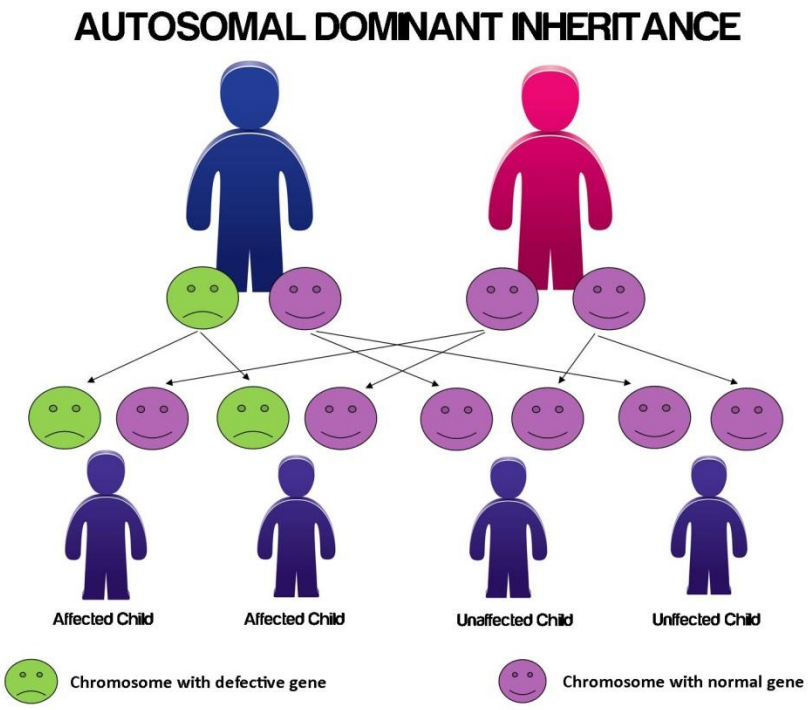


Dominant [Affected + Unaffected]

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

Autosomal Recessive (Some types of CMT1 and CMT2)

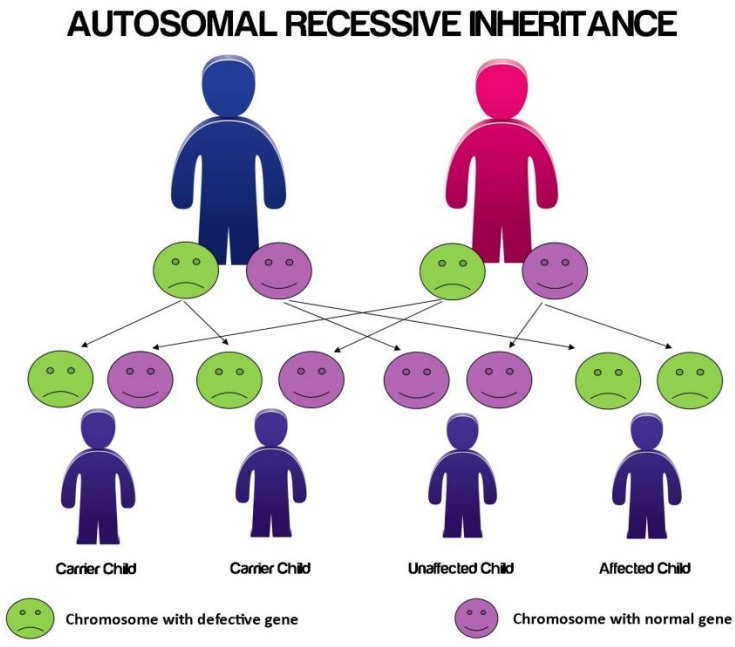
If a faulty gene is recessive it means that you need two copies of the same faulty gene (one from each parent) before you could develop CMT.

If you have only one faulty recessive gene you are known as a carrier. You will not develop 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.

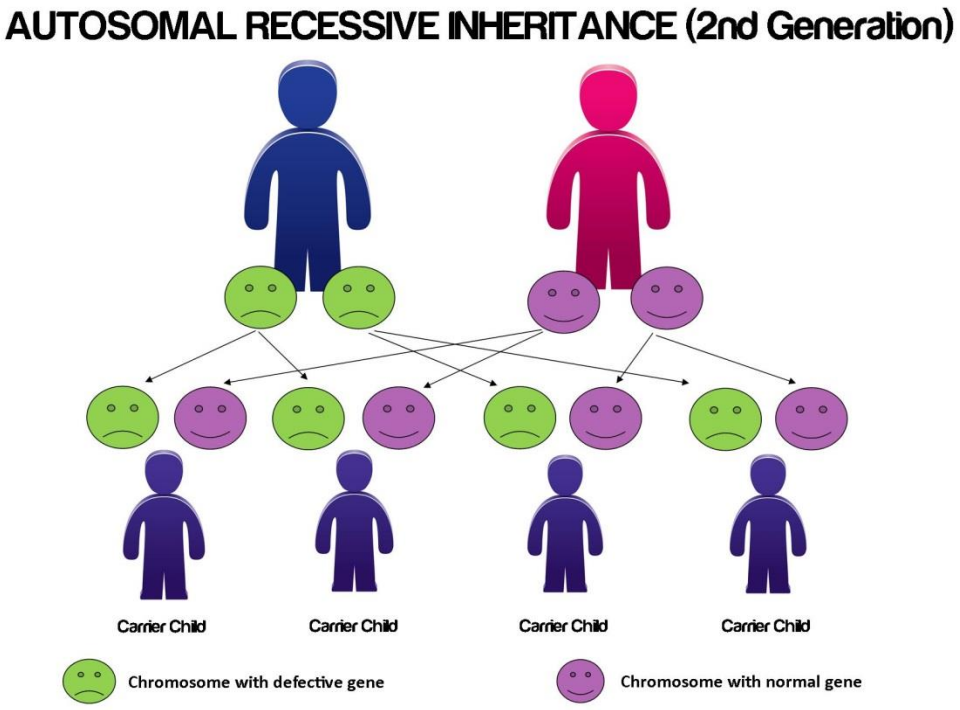
Some forms of both CMT1 and CMT2 are recessive.

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 (i.e. they each only have one copy), 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).



Both parents are carriers = one in four chance of having a child with CMT and a 50:50 chance of child being a carrier



One parent has CMT and one parent neither affected nor a carrier = no children have CMT, but all children will be carriers

X-linked (sex-linked)

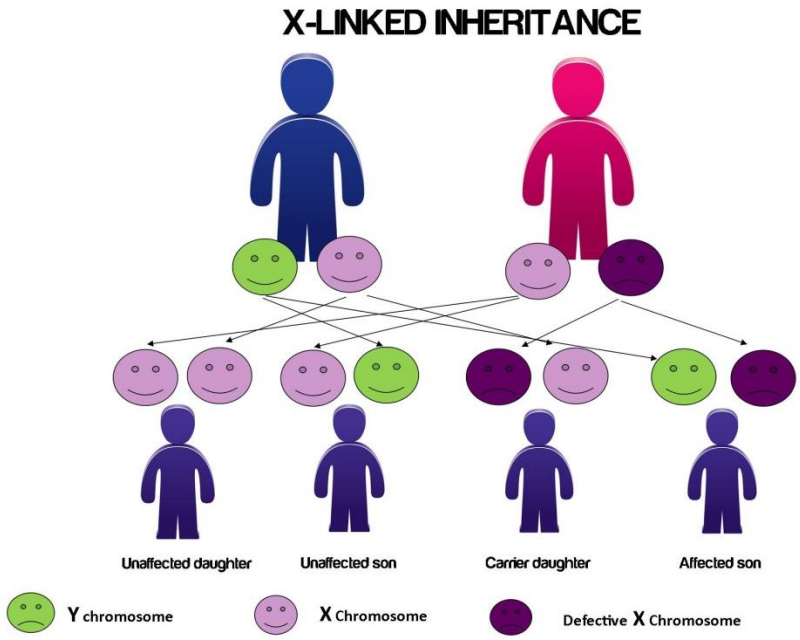
Of your 23 pairs of chromosomes, the 23rd pair is known as the pair of sex chromosomes, X and Y. If you inherit two X chromosomes you become a woman and if you inherit an X and a Y you become a man.

Four faulty genes responsible for CMT have been discovered on the X chromosome. Men who get X-linked CMT are often more severely affected than women.

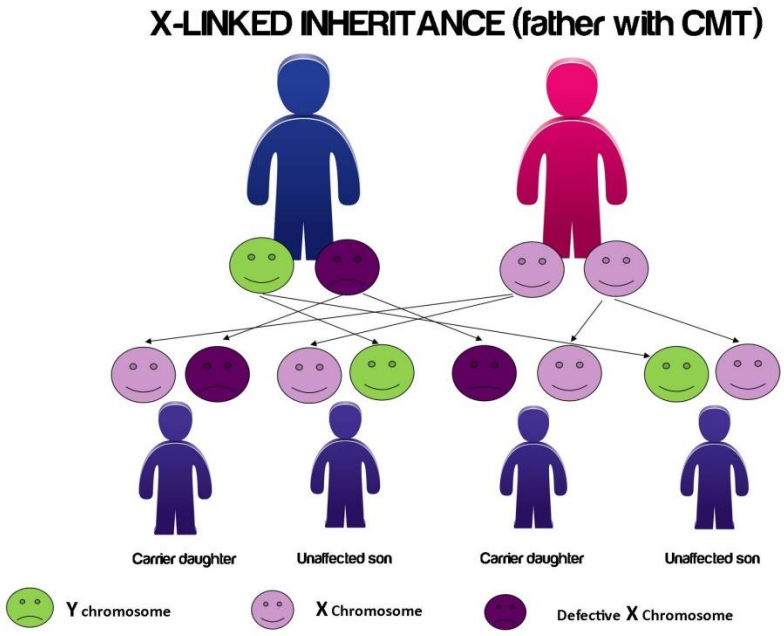
X-linked inheritance (CMTX)

- Mother has faulty gene and father unaffected = 50:50 chance that children will have CMT (males more severely than females, who are either usually mildly affected or unaffected)

- Father has CMT and mother unaffected = all daughters will inherit the faulty gene (and are usually either mildly affected or unaffected), but sons will be neither affected nor carriers



X-linked mother (who would usually have mild CMT) + unaffected father



X-linked affected father + non-carrier mother

'De novo' mutation

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

About one in 20 people with CMT1A have a new genetic mutation, but these are much more commonly seen with other types of CMT, for example CMT2A.

Sporadic CMT

The term sporadic is used when someone with CMT apparently has no family history (i.e. no other family members are affected). There are a number of possible reasons for this:

- There are no other family members available to be examined to see if they may be affected or the family is very small, making it less likely that someone else has CMT.
- There has been a 'de novo' mutation as explained above.
- In the case of somebody with an autosomal recessive form of CMT, they may be the only child with CMT and their parents will both be disease-free as they are carriers.

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 either carriers of, or affected by 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)

Classification of CMT, HSN and distal HMN

It is beyond the scope of this book to describe all the 80+ causative genes behind CMT, HSN and distal HMN. The next section aims to describe briefly the current understanding of the genetic classification of the commoner types of the condition.

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.

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. Some forms of CMT1 have an earlier age of onset and these forms used to be called HMSN3 in the old classification but are now more commonly referred to as severe CMT1. Two of these forms are sometimes referred to as Dejerine Sottas disease (DSD) and Congenital Hypomyelinating Neuropathy (CHN) but these are both forms of CMT1. Autosomal dominant CMT1 (AD CMT1) is much more common than autosomal recessive CMT1 (AR CMT1, more commonly called CMT4). Generally, people with AR CMT1 / CMT4 have an earlier onset and more severe form of CMT.

AD CMT1

- **CMT1A:** The Chromosome 17 duplication containing the peripheral myelin protein 22 gene (PMP-22), is responsible for six out of ten cases of CMT1 in the UK. CMT1A is the 'classical' form of CMT. People usually develop symptoms in the first 20 years of life, have a very slowly progressive condition and a normal lifespan. A deletion of the same part of chromosome 17 causes hereditary neuropathy with liability to pressure palsies (HNPP). Mutations in PMP-22 can also cause a more severe form of CMT1 (DSD and CHN in old literature).
- **CMT1B:** Mutations in MPZ (myelin protein zero) are responsible for CMT1B. This form of CMT is the second commonest form of AD CMT1. It can be just like 'classical' CMT but onset is sometimes earlier and the condition more severe. Mutations in MPZ can also cause a late-onset mild form of CMT2.

AR CMT1

- **CMT4C:** Mutations in SH3TC2 cause the commonest form of autosomal recessive CMT1 in the UK. This form of CMT is usually more severe than 'classical' CMT and scoliosis is commonly an early problem.

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 but CMT2 can start earlier or later. Like CMT1, autosomal dominant CMT2 (AD CMT2) is much more common than autosomal recessive CMT2 (AR CMT2), which is very rare.

AD CMT2

- **CMT2A:** Mutations in Mitofusin 2 (MFN2) are responsible for about one in five cases of AD CMT2. This usually presents in a more severe form than the 'classical' CMT, commonly in the first decade of life. People with CMT2A can be severely affected - with some needing wheelchairs - while others are more mildly affected. There is therefore a wide spectrum of disease severity in CMT2A.

AR CMT2

- The gene mutations that cause AR CMT2 are all very rare

X-Linked CMT

CMTX is a variation of CMT1 and CMT2. It is called 'X' because the genes that cause this form of CMT are 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.

CMTX1

CMTX1: Mutations in GJB1 (the protein from this gene is connexin 32) cause the commonest form of x-linked CMT. This is the second-commonest form of CMT, affecting about 10% of all people with the condition. This form usually affects males more severely than females. Males usually present in the first decade of life and on average have a more severe form of CMT than the 'classical' form. They still have a normal lifespan and rarely need a wheelchair. Their nerve conduction studies look like CMT1. Females may have no symptoms (but often have subtle signs when examined) or they may be mildly affected. The nerve conduction studies in females look like CMT2.

DI-CMT

DI stands for 'dominant intermediate'. This form of CMT is rare and usually has similar symptoms to the common versions of AD CMT1 and AD 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 CMT2A, for example. These are all different genetic variations of the main types.

Classification of HSN

The HSNs are much rarer than CMT. They are characterised by mainly involving the sensory fibres and in the UK are more commonly autosomal dominant than autosomal recessive. Involvement of the autonomic nervous system is particularly seen in some of the AR forms. There is one form of AD HSN, AD HSN1A, which is seen more frequently in the UK than anywhere else in the world as there is a 'founder effect'. This means all patients seen to date have inherited the gene from a 'founder' who can be traced back to more than a 100 years ago. HSN1A is due to mutations in the SPTCL1 gene. This usually presents in the second decade of life with damage to the feet (ulcers) due to lack of sensation. The sensory involvement is always worse than the motor, but after about 20 years there is usually motor involvement too, with weakness which can be severe in the feet and the hands.

Classification of distal HMN

Distal HMN is also much rarer than CMT but is very similar to CMT except there is no reduced sensation. So far, 18 genes have been identified as causing distal HMN, but as most of the genes have yet to be identified, most people with distal HMN do not get an accurate genetic diagnosis. AD distal HMN is more common in the UK than AR distal HMN: the commonest gene to cause AD distal HMN in the UK is the HSPB1 gene which causes HMN 2B. Patients usually present in a very similar way to those with CMT in that they develop difficulties in walking due to foot drop caused by wasting and weakness of their muscles. What is different is that people usually develop wasting and weakness of their calf muscles first, with difficulty standing on their toes before their heels. Like CMT, symptoms can develop at any age but it is common to get the first symptoms between 10 and 30 years of age. In some rare forms of distal HMN, people develop the wasting and weakness in their hands long before their feet and sometimes they only ever have symptoms in their hands.

Genetic testing

Of the 22,000 genes in the human body, 80+ have been identified as causes of CMT as of November 2014. The first causative gene for CMT - CMT1A secondary to the chromosome 17 duplication - was identified in 1991, but in the last three years there have been huge advances in the technology to look for new genes. The new techniques currently being used, called Next Generation Sequencing (NGS) mean that new genes are being identified much more quickly than previously - currently at a rate of about one new gene a month.

We now know about 65% of the causative genes for all types of CMT, HSN and distal HMN. This means that around two thirds of people with CMT in the UK can get an accurate genetic diagnosis. This figure is the same in the US and in other parts of Europe and is expected to increase in the next few years as new genes are identified with NGS. By 2020 all the causative genes are likely to have been identified.

Up until 2014, only four genes were commonly tested in most clinics in the UK but with NGS this has completely changed and it is now much easier to check all the genes in one go. Two laboratories now offer this in the UK. Testing vast numbers of genes is no longer the challenge it used to be. What is now more challenging is working out if the mutations identified are the real cause of the CMT in an individual patient. This is particularly difficult with the rare genes. For this reason, doctors often ask if they can see other members of your family (both with and without CMT) to trace the gene.

Testing for the genes that cause CMT is usually done in one of four circumstances:

Diagnostic: Somebody has symptoms and signs of CMT and wants to know which gene is causing their CMT i.e. the genetic cause.

Pre-symptomatic: This is where a person with no symptoms and signs of CMT but who has a relative with a confirmed genetic diagnosis of CMT wants to know if they have the same causative genetic mutation as their relative so that they can get information about their likelihood of developing the condition. This is not a common request in CMT as there is no treatment. If treatments become available, an early diagnosis will become more important.

Ante-natal diagnosis: This is where a pregnant women who either has CMT herself or whose partner has it, screens the pregnancy (usually at about 12 weeks by CVS testing) to see if the foetus carries the mutated gene.

Pre-implantation diagnosis: This is where the sperm and the egg of parents (one of whom has CMT) are fertilised outside the womb (by IVF). The resulting embryos are then tested to see if they carry the mutated gene and only the embryos without the mutated gene are implanted into the mother's womb.

Where can you get advice?

Genetic testing for the underlying cause of CMT in different families is changing. Testing for the commonest cause - the duplication of chromosome 17 - can be done very readily. Testing several other genes can be arranged through the 22 regional genetic centres (find a list of centres on the NHS website) throughout the country (and the Republic of Ireland). It can also be done in specialised neuropathy clinics. Your neurologist or your family doctor can make the appropriate referral for you and any relatives who are concerned.

Testing for less common causes of CMT is improving rapidly but may require referral to a specialist research laboratory. Much of the CMT research is done in the neurogenetics unit at The Institute of Neurology.

To make the most of any genetic findings you need to have your results interpreted by your consultant neurologist or by a 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.

Planning a family: how developments in genetics can play a part

From the twelfth week of pregnancy onwards it is possible to test a foetus for those forms of CMT for which the causative gene is known. Investigations of affected family members may be needed before the tests are done. Unless you are considering ending the pregnancy if the foetus is affected, there may be little advantage in discovering at this stage whether or not the child will have the gene that causes CMT, because the test itself carries a risk of miscarriage. Tests in pregnancy may improve dramatically over the next few years and be available earlier in pregnancy and without risk of miscarriage.

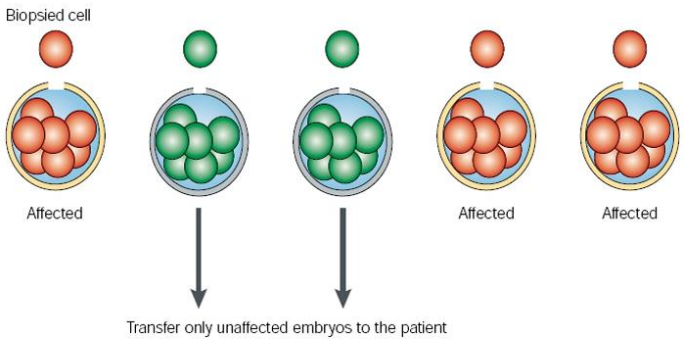
Pre-implantation genetic diagnosis (PGD)

PGD is a method of screening embryos before they are implanted into the womb during in-vitro fertilisation (IVF). After the embryo is created in the laboratory from the eggs and sperm of a couple, it is then tested for the relevant genetic disorder – in this case CMT. One or two unaffected embryos are then transferred into the womb, in the hope that a pregnancy will occur.

PGD is a complex procedure involving time and emotional and physical commitment, but it does avoid the need for termination of an affected pregnancy. This is the main reason given by those who request PGD.

Couples will first have an appointment with a PGD Genetic Counsellor to discuss the procedure. For CMT, laboratory work is needed to prepare the genetic test that will be used on the embryos. Couples will also need a pre-treatment appointment with an IVF doctor in the Assisted Conception Unit (ACU). Preparation for a PGD cycle takes some time and after an initial discussion it is likely to be around eight to nine months before a treatment cycle can start, sometimes even longer.

As PGD involves using IVF, the success rate is relatively low compared with the chance of conceiving naturally. Approximately one in three cycles of PGD will result in a baby. If a couple get as far as having embryos transferred, then there is approximately a one in two chance of success.



You will need a referral by a geneticist or genetic counsellor to a PGD clinic for an initial discussion appointment. This will give you the opportunity to ask questions and find out more about PGD before reaching your decision. NHS funding

for PGD may or may not be available.

Each separate type of CMT must be licenced for PGD separately by the Human Fertilisation and Embryology Authorities, and currently licences are in place for only a small number of genetic types. Therefore, when considering PGD, bear in mind that it may take a considerable time to gain the appropriate licence for your type.

Questions and answers

Will knowing the precise genetic cause of my CMT 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 suggest 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 to present in the first year or two of life, whereas others cause it to present at 20 or 30. Your consultant neurologist will be able to tell you more about this.

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

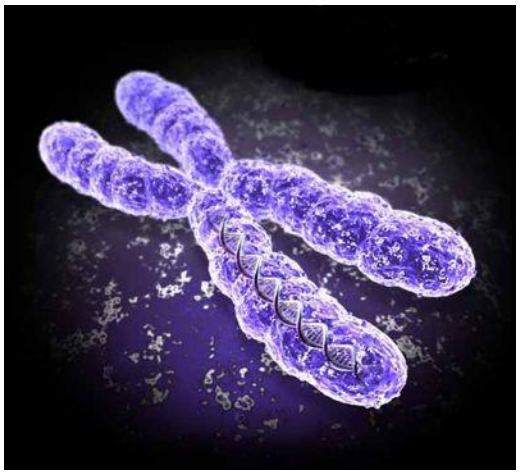
This is a matter of debate. Most doctors do not recommended genetic tests on children who have no signs of CMT. Children can make their own decision as to whether they want testing when they are 18. If they do develop possible signs or symptoms of CMT during childhood, they should have a genetic test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

However, many people with CMT believe that parents should be able to ask for genetic testing to be carried out on their children – a case of forewarned is forearmed. CMT United Kingdom's position on this sensitive issue is that asymptomatic testing (i.e. 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.

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 each of the 10,000 trillion cells which form the human body behaves.

Apart from the sex cells (sperm and egg) each human cell has two copies of each of the 23 types of human chromosome. This amounts to 46 single chromosomes in each cell nucleus. We inherit a single full set of 23 chromosomes from each of our parents.



Twenty-two of our chromosomes are known as autosomal and these are the same in boys and girls. The twenty third pair is our pair of sex chromosomes, the X and the Y chromosomes. If we have two Xs we become female and if we 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. This newly-fertilised egg cell then contains the full 23 pairs of chromosomes. 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 may be completely unaffected, in which case you are known as a carrier. If you are affected, this is usually more mildly than in a male. This variability occurs because women use only one of their two X chromosomes in any one cell: which X is used in each patch of tissue is decided early in development and varies randomly. The two copies of the CMT-X gene may be employed about equally (in which case the woman will be affected more mildly than a male) but sometimes the affected copy is used predominantly and then she may be affected much as a man might be.

Chapter 3: How to manage your CMT

How to manage your CMT

Living with a disability can be difficult, particularly a long-term condition like CMT, which may change over time. You may find that at different times in your life you face different problems or you may find that a task you have been dealing with perfectly well for years becomes more awkward.

However CMT affects you, somebody has probably dealt with that particular problem before and there is invariably an answer, technique, or method for tackling it so that you can live life to the full. It may take time to find the right solution for you, but do ask.

Remember, you are not alone and there are people who can offer support when you need it. Apart from your friends and family, many people with CMT are willing to share their experiences and methods of overcoming the difficulties you may face. As well as CMT United Kingdom, there are a number of other useful voluntary organisations, not forgetting healthcare professionals. You'll find a list of helpful organisations in Chapter 5.

What you can do

You are the person who best knows how your CMT affects your body on a day-to-day basis. If you don't take responsibility, no one else will. For more on this, see Coping with CMT,.

Professionals such as doctors and social workers, are there to offer specialised advice and support on issues like treatments, drugs and benefits, but they will only be with you for short periods. The rest of the time your wellbeing, both physical and mental, is in your hands. You understand how symptoms affect you, both mentally and physically, which makes you 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
- help prevent further complications
- communicate effectively with professionals
- become more realistic about the impact of your disease on you and your family
- lead a fuller life

Remember that getting the right information about CMT is important, helping you to make informed decisions for yourself.

If you want to learn how to take a more active role in your general health and wellbeing, as well as managing your CMT, the NHS Expert Patient Programme may suit you.

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. It is your GP's role to look after your general health, help you deal with some of your symptoms and refer you on to other specialists when necessary. These may include:

- neurologist
- physiotherapist (with an interest in neuromuscular conditions)
- orthotist
- chiropodist
- occupational therapist
- orthopaedic surgeon
- rehabilitation medicine physician
- podiatrist

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. For more on staying active - including daily stretches and gentle exercises – see Stretching, exercise and physiotherapy.

A healthy weight

Being overweight or obese is bad for anybody's health whether they have CMT or not. For people with CMT, carrying extra weight can make matters worse because it:

- 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 maintain a healthy weight is to keep active and eat healthily. For more on this see Healthy eating.

Alcohol

Alcohol was removed from the list of neurotoxic drugs in July 2004. While moderate drinking does not usually cause ill effects to those with CMT, alcohol does affect balance and coordination, which may already be compromised in people with the condition. Heavy drinking or getting drunk is not advisable because it can damage your nerves and this effect is likely to be exaggerated in people with CMT. If you have questions about alcohol and your health, see your GP.

For more on sensible drinking, see 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.

Chilblains

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

- the normal heat generated by muscle activity is missing due to loss of muscle bulk and lack of movement
- CMT can affect the autonomic nerves which control the blood vessels and therefore blood flow. This is also why the skin looks patchy and discoloured

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

The most comprehensive source of high quality information on CMT is CMT United Kingdom's website. The internet is awash with information on almost every ailment imaginable, but not all of it is reliable and some is positively dangerous. For more on trustworthy websites see Chapter 5, Organisations that can help you.

Ultimately it is up to you to decide whether to trust a source of information or not.

- If the claims seem to be too good to be true, they probably are.
- Beware of websites that ask you to hand over large amounts of cash.
- Beware of websites advising you to stop the medication prescribed by your doctor. Never do this without first consulting your doctor.
- Is the website British-based? 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.

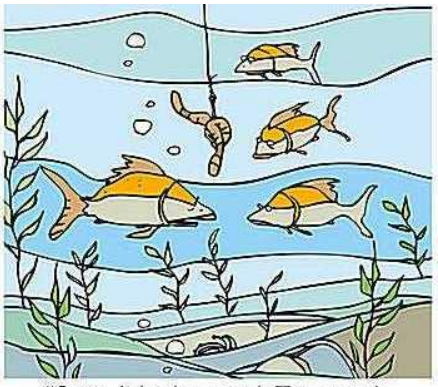
Beware of adverts and false claims

If you come across a website that claims a '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. Beware of:

- Phrases such as 'scientific breakthrough', 'miracle cure', 'exclusive product', 'secret formula' or 'ancient ingredient'.
- Claims that the product can treat a wide range of problems.

- Anything that uses 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.
- Money back guarantees – especially if you are sending the money abroad or to a PO Box.
- Adverts or websites that fail to list the company's name, postal address or other contact information.

Make sure the information is evidence-based and discuss your options with your healthcare team. Evidence-based means that the information has been reviewed in light of the latest scientific findings in medical journals.



"Sure, it looks good. Too good.
Trust me, there's always a catch."

Treating your CMT

In a nutshell

At present, there is no specific treatment or cure for the underlying genetic cause of CMT. Nor are there any drugs to stop or reverse the damage caused to the peripheral nerves. However, the situation is not as bleak as it sounds because a number of treatments are available. These can slow the development or ease some of the secondary complications linked to CMT, greatly improving your quality of life.

Look for the least invasive way to treat your problems using a combination of the following:

Self-management

- stretching and exercise
- healthy eating

Rehabilitation and Therapies

- physiotherapy
- orthotics
- occupational therapy
- podiatry
- rehabilitation medicine
- psychology

The aim of rehabilitation is to help you make the most of your potential, often focusing on specific goals that you want to achieve. Rehabilitation is best done by a team containing some or all of the professionals above. If there is a consultant in rehabilitation medicine who knows about CMT in your area, he or she may run a service with a multidisciplinary team. The consultant will help with medical management of problems such as pain and fatigue, as well as using his or her specialist knowledge of treatments such as orthotics, psychological therapies and physiotherapy to work with you for the best outcome.

Specialist medical expertise

- A consultant neurologist or a rehabilitation medicine physician with an interest in neuromuscular conditions in partnership with your GP should oversee your treatment.

The aim of all of these treatment options is to:

- **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.
- **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 (e.g. wheelchairs) may bring some relief.

Getting the right treatment

Treatments exist for many of the complications of CMT, but at present there are no disease-modifying therapies to either halt the progression or cure CMT. However, many aspects of the condition can be managed for most people.

Current treatments include physiotherapy, orthotics, occupational therapy, pain and fatigue management and - when indicated - orthopaedic surgery, speech therapy and respiratory support. It is important to be aware of these treatments because people are sometimes told by their GPs that there is no treatment for CMT and therefore no point in referring them to specialists such as neurologists, paediatricians, orthopaedic or surgeons, to name a few.

Your CMT should be treated by people who know about the condition. This means your treatment should be overseen by a consultant neurologist (or paediatric neurologist) with a specialist interest in peripheral neuropathy (which includes CMT) or a rehabilitation medicine physician with an interest in neuromuscular conditions. The specialist should work in partnership with your GP.

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 difficult to see the right neurologist as there are just not enough doctors with this type of knowledge. There are less than 50 neurologists in the UK with particular speciality in peripheral nerves. However, there are 550 to 600 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 progression of your CMT.

specialist paediatrician – a paediatrician (ideally a paediatric neurologist) 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 – a specialist 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. They can be especially helpful in helping overcome work-related issues.

podiatrist - sometimes called a chiropodist - specialists in foot care.

orthopaedic surgeon – a surgeon who specialises in bone and joint surgery. Preferably you should see one who specialises in foot and ankle problems.

rehabilitation specialist – a doctor who works to get you back on your feet following a health setback.

neuromuscular nurse specialist – increasingly, neuromuscular clinics in the UK have specialised nurses attached to them. These nurses are usually contactable by email, text or phone outside normal clinic times and provide a wealth of information and help for patients.

Information is available from CMT United Kingdom or the British Peripheral Nerve Society, a society for neurologists who are interested in neuropathies. Ask your GP to refer you to your nearest specialist. Unfortunately, the waiting list for these specialists may be very long.

Getting the most out of a consultation

A visit to the doctor can be stressful. 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 out 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 consider taking a trusted friend or relative: they can take notes and remind you of questions you wanted to ask. It can be helpful to bring 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. This is why a list of questions and key points can be useful.

Get information

Feel free 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. You should receive a copy of your clinic letter from your specialist. Make sure you have a contact number for the secretary or nurse specialist in case you have any queries.

Consent for procedures

Before having treatment, particularly surgery, you will be asked for your consent (permission). Before giving this you should ask about:

- the percentage success – and failure – rates
- what will happen during the treatment
- why you need the treatment or procedure
- what could go wrong
- whether there are 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.

CMT and medication

In theory, some medication may affect people with CMT more than the general population. However, the evidence is slight or non-existent. The only drug for which there is evidence of risk of acute worsening of CMT1A and probably HNPP is **vincristine**. This is a drug used in chemotherapy (treatment for tumours) and should be avoided in people with CMT1A confirmed by genetic testing.

It is a good idea to make sure that any doctor who is about to treat you knows that you have CMT and understands what it is.

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

You can check the list of drugs that **may** have a detrimental effect on you at www.cmt.org.uk

To put the possible risk into context: alcohol and illegal recreational drugs are much more likely than prescription medication to have a harmful effect on people with CMT.

Complementary and alternative treatments

Some people swear by complementary or alternative treatments. However, there is no evidence that alternative treatments are effective in CMT.

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. The foot is a complicated part of the anatomy: 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

Before undergoing surgery:

Make sure that the surgeon doing your operation knows all about CMT. Ask the surgeon how many similar cases he or she has treated before. Ideally, go to an orthopaedic surgeon specialising 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.

Ask for a copy of your clinical letter to be sent to you and to your referring doctor. Better still, ask for a letter to be sent to you outlining in lay terms the benefits of surgery, its consequences (i.e. time in hospital, time in plaster, time off work etc.) and the risks.

Find out how likely the operation is to succeed – and the chances of failure - as well as the success rates of the individual surgeon who will be operating on you.

Ask how long you will take to recover. Will you be able to get around or work during this time? 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.
This kind of surgery can be extremely successful - giving a much better long term outcome than bony surgery - and needs less revision in the future. However, ideally it must be done before deformities become fixed – in other words, whilst the foot can still be put into the correct position manually.

Osteotomy – cutting the bone and repositioning it. Usually the bone is fixed in its new position with plates, screws and other devices. Sometimes in cases with drop foot, the drop foot can be improved or overcome by tendon transfers or osteotomy.

Triple arthrodesis – stabilising the hind foot joint by stiffening (welding) three joints together (arthrodesis is another term for 'fusion'), preventing movement in two directions (left-right and ankle tilt). A triple arthrodesis is only carried out once the foot has stopped growing – usually no earlier than 12 in girls and 14 in boys – and sometimes later. This surgery is rarely carried out, unless there really is no choice, due to the risk of developing arthritis in other ankle joints later in life.

Plantalar fusion – triple fusion combined with ankle fusion, for the most severe cases of inturned ankle and feet, fuses the ankle as well as the triple joints. It leads to a very stiff ankle region, with loss of drop-lift of the ankle as well as the left-right and ankle tilt of the triple fusion. On the plus side, it gives a straighter and more stable ankle in cases where recurrence is otherwise likely.

Foot problems most commonly considered for surgery include:

Claw toes
If clawing of the toes is the only problem being addressed by surgery, 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 to prevent recurrence. Operations on claw toes are often done at the same time as other procedures such as osteotomy and arch correction.

Turned-in heel (heel varus)
If turning in of the heel is not adequately corrected by orthotics, 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.

High arched foot (cavus foot/pes cavus)
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 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.

Surgical techniques

Operations to correct the position of your foot include:

- Calcaneal osteotomy – the heel bone can be shifted to bring your heel back under your leg and the position fixed with a screw or plates and screws.
- First metatarsal osteotomy – the bone leading to your big toe can be shifted and repositioned.

Operations to rebalance the pull of the muscles in order to prevent the deformity returning include:

- Peroneal tenodesis – re-positioning and strengthening of the peroneal muscles (which turn your foot outwards)
- Tibialis posterior transfer – one of the muscles in your lower leg (called the tibialis posterior), which causes the foot to turn inwards (and cause deformity) is transferred to the outside of the foot to assist the weak muscles that turn the foot outwards

Surgery may also include one or more of the following: soft tissue releases; other tendon transfers; other bone procedures; and joint fusions.

These procedures should result in:

- a stable foot in a neutral position
- improved function/mobility
- less pain
- improved walking – able to walk with fewer aids and orthotics (insoles)
- better muscle balance
- decreased callosities (hard skin)/pressure areas
- maintenance/improvement of range of movement

Full recovery may take up to twelve months

Bear in mind that:

The recovery time from surgery is often very long. Doctors tend to refer to recovery time from surgery as being the time in plaster – around eight weeks. In fact, it can take much longer to achieve decreased pain and return of function – a year or two for complete recovery from aching and swelling in some cases.

You may well need help with everyday tasks following the operation. It is very important to rest and keep your foot elevated.

People having major surgery requiring casts or splints will normally be prescribed some kind of blood thinning drugs, as recommended by NICE (National Institute for Health and Care Excellence). People with reduced mobility – such as that caused by CMT – may be at particular risk of developing blood clots in the leg or lung, which are potentially very serious complications. Before you leave hospital, make sure you know how long you need to be on the prescribed drugs and that you are fully trained in administering them: some of them have to be injected by you or a relative.

The physiotherapists in the hospital may expect you to be able to ambulate with crutches or a Zimmer in the 'normal' fashion, by hopping – possibly before they sanction your discharge. You may well not be able to do this, due to the weakness in your arms and wrists, so do not be afraid of standing up for yourself, and explaining clearly why this is unsafe or difficult for you to do. You will need to have made a plan as to how you will manage in your home if you cannot get around using crutches. Do you have a wheelchair or can you borrow one in the short term? Can you get in and out of a wheelchair safely, or will you need a transfer board? Is there room to get around your home in a wheelchair? Can you get in the front door? How will you get to the toilet or upstairs to bed? It is far better to plan all these things well in advance of the surgery, so that you can inform the staff that all is organised. Your community occupational therapist can help you get any equipment you require before the surgery takes place. Otherwise, it is unlikely that any equipment could be put in place quickly enough before you are discharged.

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 telling the anaesthetist about your condition well in advance, he or she can work out with you what type of pain relief is best for you. The anaesthetist may also want to check your respiratory function before the operation. As an extra precaution you will probably be asked to stay in hospital longer than usual after the operation, which is why people with CMT should not be admitted for 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 the anaesthetic lasting longer and having 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. However, spinal or epidural anaesthesia have been reported as successful techniques.

If general anaesthesia is used, non-depolarising muscle relaxants (as opposed to 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 wears off or can be reversed. Depolarising muscle relaxants should be avoided but inhalational anaesthetics are safe.

It is extremely important that your surgeon and anaesthetist are informed if you have HNPP as being in one position for a long time under anaesthesia must be avoided.

Help yourself

Questions to ask before surgery:

- Do the anaesthetist and the doctor carrying out the operation – usually a surgeon – and their team understand that you have CMT? It is vital that the anaesthetist knows that you have a neuro-muscular condition.
- Do they all understand what CMT is?
- Are they aware of any drugs that should not be used on people with CMT? Ask CMT United Kingdom to provide you with the list of drugs that should be avoided.

- Have you told them about all the drugs and treatments you are taking, including any over-the-counter treatments (e.g. cough mixtures) and any complementary or alternative remedies?
- Have they discussed with you all the possible options - including no treatment - and their benefits, consequences and risks (including being no better or possibly even worse-off after surgery)? This is known as 'informed consent': 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 fully understand.
- Has a full assessment been planned prior to surgery of your mobility, nursing and other needs? This is normally done by a physiotherapist or occupational therapist at a pre-admission clinic.
- Has your healthcare team discussed with you the risks of thrombosis, and how it will be managed?

The Royal National Orthopaedic Hospital patient information guides *Preparing for Foot and Ankle Surgery* and *Post-Operative Advice Following Pes Cavus* contain useful information and advice. For further information see http://www.rnoh.nhs.uk/.

Feet

You and your feet

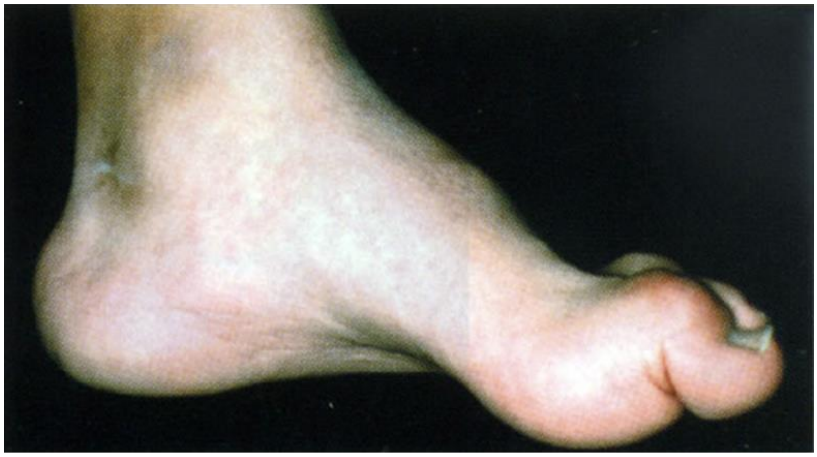
Although easily overlooked, the human foot is a complex and marvellous piece of engineering that needs proper care. Each foot is made up of 26 bones bound together by ligaments, supported by muscles and supplied with blood and nerves. The feet are usually the first area to be affected by CMT and need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

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. If not prevented or treated early this can lead to a higher risk of tripping and sprained ankles as well as strain on the knees, hips and lower back.

Measures to ease or slow down the onset of problems

regular exercise – swimming is particularly good
physiotherapy assessment and stretches/strengthening exercises as advised
orthoses – if considered beneficial by your physiotherapist or podiatrist

Treatments for existing problems

orthoses
physiotherapy
stretching under supervision
surgery

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 (e.g. blisters, cuts, splinters), which can lead to:
sores
ulcers

Measures to ease or slow down the onset of problems

properly fitting shoes
daily foot checks
podiatry
orthoses

Treatment for existing problems

wound dressings and drug treatments, either from your GP or in hospital.
podiatry
orthoses

Other common problems

swelling of the feet
extreme cold feet
slow healing

These symptoms are due to the neuropathy causing limited movement and sensory loss and are not usually due to a blood supply problem.

Some people notice that the healing process after injury is not as good as it should be, mainly due to sensory loss. This makes ulcers more likely if damage to the feet goes unnoticed. Foot care management, including daily checks, is extremely important.

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 and never put feet on a direct heat source
massage and 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

Daily foot care: key points

The all-important day-to-day care is down to you. The basic rules are:

- 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
- dry your skin carefully – don't rub hard with a towel
- check daily for cuts, broken skin or blisters – after a bath or shower for example
- do not cut corns, calluses or ingrown toenails – see your doctor or podiatrist
- try to avoid bruises, burns, cuts, cracks, chilblains and frostbite. If you get any of these seek professional advice

- any problems with a mole on the foot should be checked by your doctor or podiatrist immediately

- avoid exposure to cold and damp

- seek immediate professional advice if you ever get an ulcer or sore on the foot or leg

If you are somebody who experiences loss of feeling or numbness in your feet or legs:

- see a podiatrist at least once a year or at more frequent regular intervals

- check your feet daily for cuts, splinters, abrasions or blisters. If they are not healing properly, 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 shoes, shake them out to get rid of any pebbles and then check the insides with your hands to feel for any rough spots.

- if you have Hereditary Sensory Neuropathy (HSN) - a subtype of CMT with more sensory loss and a susceptibility to developing ulcers - be extra careful with your feet and hands. Pain and temperature perception are greatly affected in HSN and you may develop complications such as ulcers which can become infected

The right shoes

- wear shoes that are comfortable, offer good support, fit well and don't rub

- when you get a new pair of shoes, break them in gradually. Start with an hour on the first day, two hours on the second day then increasing until you can wear them all day

Podiatry (chiropody)

Podiatrists and chiropodists are specialists in foot care who diagnose, treat and manage a variety of foot conditions. They can assess your nerve function, circulation and foot function and give you information on foot care.

Podiatrists can treat hard skin to prevent it breaking open and treat 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 interchangeably. 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:

On the NHS through a referral by your GP – remember to stress lack of sensation or numbness to the GP when asking for the referral, as this increases the likelihood of getting an appointment. Many NHS podiatrists are mainly concerned with diabetic patients, as the neuropathy they can experience is serious. However, CMT neuropathy can be just as severe, if not monitored and treated adequately.

Privately. You can find a private podiatrist through the Health Professions Council.

Ideally, everyone with CMT should see a podiatrist once every six months for a check-up.

Hands

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

- muscle wasting, causing lack of hand strength and grip
- loss of sensation
- reduced dexterity

Muscle wasting and weakness

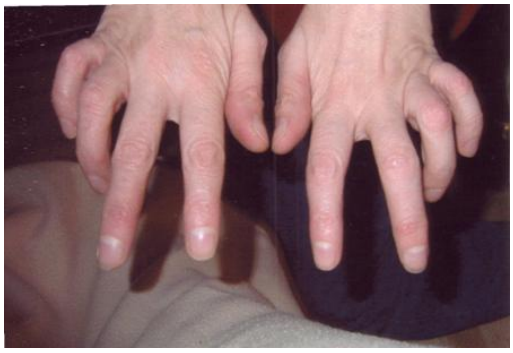
Usually the small muscles in the hand that straighten the fingers are affected first, resulting in the classic 'CMT hand', with hollows where there should be bulk, for example at the base of the thumb. At the same time, people may notice that fine movements become more difficult, affecting writing, fastening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

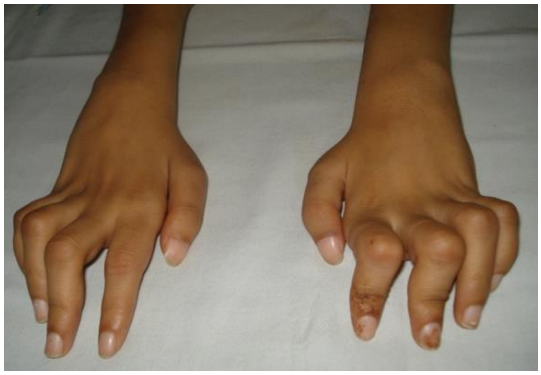
Common secondary problems

claw fingers
strain and tightness in shoulder and neck - as muscles in the upper arm try to compensate for loss of hand strength.

Measures to treat or slow down the onset of problems

stretching. See Stretching, exercise and physiotherapy
orthoses: to either stretch or help with grip movements. See Orthotics
occupational therapy. See Occupational therapy
physiotherapy See Stretching, exercise and physiotherapy
hand surgery





Orthotics

Orthotics is a specialist care area involving the provision of supports of various kinds for the feet and – less commonly – the hands. These supports are known as orthoses.

These include:

- splinting
- supports
- braces
- insoles
- special footwear

A prescribed orthosis is designed to correct your alignment to the ideal position. Where this is not possible, the orthosis will accommodate your position and provide the necessary support to stabilise your limb(s). This will help reduce the secondary complications associated with CMT.

Historically, orthoses have been cumbersome, large, unattractive and, often quite uncomfortable to wear. However with advances in technology they can be substantially lighter, lower profile and permit movement. All of this can increase your mobility and substantially improve your quality of life.

An orthotist is an allied healthcare professional whom has met the requirements set by the Health and Care Professions Council (HCPC) to become UK state registered. You can check the registration status of your orthotist at www.hcpc.org.uk.

A referral to the orthotic service is normally made by your GP or hospital consultant. An orthotist will first complete a clinical assessment and analyse your walking pattern before deciding on an appropriate prescription to reach your mobility potential. This ideally should involve the extended multi-disciplinary team (MDT), namely a neurological physiotherapist, occupational therapists and in some cases a rehabilitation/ neurology consultant.

If you have difficulty distinguishing between some of the terms in this section, Paul Charlton, CMT specialist orthotist offers the following:

> *"The specialist working in the field of orthotics - who may fit an orthosis from a selection of orthoses - is known as an orthotist."*

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. Different orthoses often give different degrees of control and it may take time, discussion and experimentation to find the appropriate degree of control for you. 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 you have not been offered an orthosis, you may have to push for it.

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.

The orthotist should recognise that everyone with CMT has different needs and prescribe the right orthosis for you. As you will find from a quick internet search, there is a large range of different orthoses available, each 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. It is recommended that physiotherapy sessions are provided when you have a new orthosis to help you adapt to a new walking pattern.

Make sure that the orthotist also understands any problems that you have with hand function. It is no use being given wonderful orthotics if you cannot do up the straps or fastenings independently: this is just as important to you as any other aspect of the orthotic. It can also be a challenge getting shoes that work with the orthotic. Do not allow your orthotist to fob you off with an orthotic that you **know** will not fit into any shoes.

New orthoses

Orthoses can take some getting used to and simple modifications can make all the difference to whether a new appliance is comfortable or not. A compromise may need to be made between what is best and what is comfortable and tolerable. Work with your orthotist to get the best solution.

When you remove your orthosis, it may leave red marks on your skin. These should disappear within 30 minutes. If redness persists for longer, stop using the orthosis and consult your orthotist. Orthoses should never cause breaks in your skin.

Do not also be afraid of making a nuisance of yourself at the orthotics clinic to make sure that the new orthotic is absolutely as you require it to be. It can take considerable patience

to get them to a wearable condition. Clinic staff may not tell you that orthotics can be lined for comfort, or have holes drilled in them for airflow (although this might damage the strength of the orthotic). Most orthoses can be adapted and changed far more than clinic staff may be willing to admit!

Your orthotist will advise on 'wearing in' your new orthosis. Generally, the orthosis should initially be worn for short periods, increasing wearing time as it becomes clear that the fit and function are correct. This is particularly important as many people with CMT have reduced peripheral sensation.

While you may prefer 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.

Common orthoses

There is a vast range of different types of orthoses – coming in different materials, colour and size – so if the pictures and descriptions below 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.

There are two main ways an orthosis can help with walking.

- swing phase - if your toes aren't up or held up, you may trip as your leg swings through the air when you step forward
- stance phase - as you balance with one foot on the ground as your other leg swings forward

If you cannot stand still, it suggests you have stance phase problems which can be helped considerably with more rigid devices. By fixing your ankle in an optimum position, you are unable to wobble on it and can then balance better.

Some of the orthoses you might hear about include:

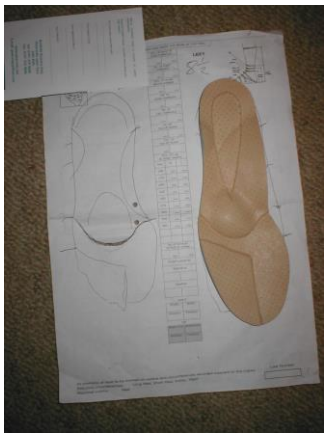
- FO - Foot Orthosis (insoles)
- FFO - Functional Foot Orthosis (insoles)
- SMO Supra-malleolar Orthosis (ankle brace)
- AFO - Ankle Foot Orthosis
- DAFO - Dynamic Ankle Foot Orthosis
- SAFO - Silicone Ankle Foot Orthosis
- Hinged AFO - Jointed Ankle Foot Orthosis
- KO - Knee Orthosis
- KAFO - Knee Ankle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

Insoles

Insoles correct and stabilise your feet by altering their position or accommodating the existing shape, so bringing pain relief. Insoles may have wedges, referred to as posts, to realign the feet and ankles so as to correct the weight-bearing line. When it is not possible to correct the feet, the existing foot shape must be captured and 'offloaded' to evenly distribute pressures, providing comfortable weight-bearing.

For example, if you have a high-arched foot with a tendency to lean to the outside, you would be given an insole with wedging to the outside to shift your weightline more to the inside. This would reduce the strain to the outer foot muscles which would otherwise be overloaded and prone to inflammation. The goal is to maintain your foot in a neutral alignment to ensure your muscles and tendons are in a comfortable mid-range position.

If your foot does not conform to the shape of standard footwear lasts, you may be prescribed specialist footwear. There is a large selection of footwear styles available. Your orthotist will offer you a catalogue of different styles, colour and fastenings. Specialist footwear can also have reinforcement in key areas to provide support where needed. Many shoes can be adapted to add raises, wedges, reinforcements etc. as required.

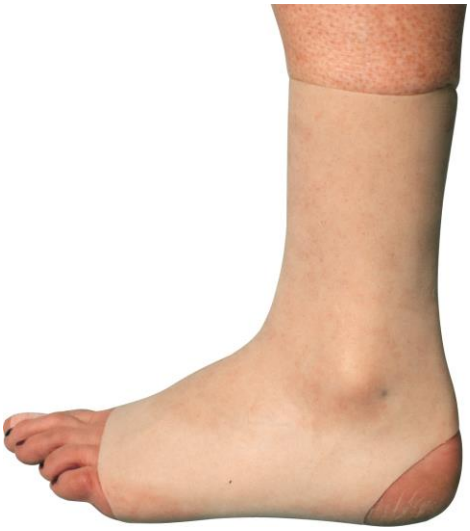


A dynamic insole which supports the dynamic arches of the foot and creates a more stable base. Useful in a mobile foot but can be uncomfortable and intolerable on a rigid foot.

Ankle braces

Ankle braces may offer some support to prevent too much movement if your 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.



A silicon AFO, manufactured by Dorset Orthopaedic Ltd

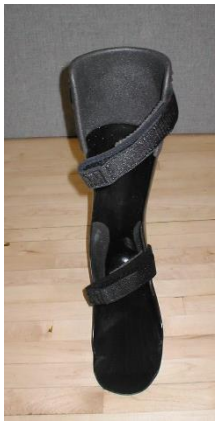
Ankle foot orthoses (AFOs)

AFOs come under the foot, and part way up the back of the calf, usually strapping at the ankle. These can be bought off the shelf, though people with CMT usually need a custom-made device.

Calf-length AFOs may be made of plastic which can be flexible and very helpful in countering foot drop. Alternatively, a more rigid design can help provide improved alignment of your joints. This not only helps feet and ankles but has an effect on knees, hips and general posture.

You may be prescribed conventional below-knee callipers when direct pressure is uncomfortable or painful or if there is swelling. Generally, these offer less correction than plastic AFOs as they cannot provide corrective forces as accurately.

In general, made-to-measure AFOs fit better as they have been designed to match the exact shape of your limb(s).



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, they are more effective than the ankle braces for controlling footdrop

Dynamic Walk

Knee ankle foot orthoses (KAFOs)

Knee ankle foot orthoses (KAFOs) or full-length callipers are used to stabilise non-functional hip and knee muscles. These generally have a knee lock so the knee can be held straight for standing and walking but released for bending for sitting. They are usually combined with the designs for ankle support mentioned previously, and can be metal or plastic.

More recently, there have been innovations in KAFOs design allowing for stance phase control during walking. This means the knee joint can be automatically locked and unlocked during walking, depending on the position of the weight line. Having a flexed knee during swing phase removes the need for secondary compensations to achieve clearance and reduces energy expenditure.

Materials
Traditionally, leather, plastic and metal have been used in orthotic devices, but carbon fibre is now more readily available. It is lighter in weight, thinner and more rigid than other materials. The increased rigidity allows for reduced trim lines without compromising structural integrity.

Orthoses for hands

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

- function (e.g. helping you hold a pen)
- maintaining good position of the hand at rest, especially at night
- providing a stretch to specific hand muscles as part of a hand programme

An occupational therapist, rather than an orthotist, will usually provide orthoses for your hands.

Occupational therapy

An occupational therapist can help people with CMT by providing:

- environmental assessments – at school, work, home
- equipment recommendations
- fatigue management
- career advice
- workplace assessments

There is an old saying that a physiotherapist will teach you to walk, but an occupational therapist will teach you to dance! Occupational therapists work with people of all ages, including children, - and with a range of difficulties - to help them achieve their full potential.

OTs are people-centred and their goal is to promote and enable independence. They will assess how well you cope with activities of daily living (ADLs), listen to your needs concerning personal care, leisure, work, study, travel and household management and advise on options for you. Their assessment may involve breaking down the activities you find hard into their component parts.

For example, if you have CMT you may struggle with everyday activities like getting dressed, opening food packets or holding a pen to write. Your OT will work with you to find solutions to these problems to help you remain independent. Solutions may come in the form of trying some adaptive equipment to compensate for your difficulty, or by working on activities to help maintain strength in certain muscle groups.

OTs are also skilled in making splints for hands. People with CMT may develop slightly clawed fingers and experience some muscle wasting in their hands. A hand splint will help to keep your hand in a good position in order to minimise pain and muscle contractures.

How an OT can help

At various stages of CMT an OT may be able to offer expertise in areas such as:

- individualised fatigue management programmes to understand the nature of your particular fatigue within your daily life
- how to more effectively prioritise and manage your time to achieve the things you want to do
- strategies to improve sleep and good quality rest
- relaxation as a coping strategy - for example as a stress or pain management technique

- ergonomic information about effective joint protection and energy conservation strategies
- hand-care techniques including provision of hand exercise programmes, fabrication of custom made hand splints to aid daily tasks, pain management and hand positioning
- adaptive equipment from small aids to major adaptations for helping you at home or in your workplace
- signposting and referring on to agencies to help with the cost of purchasing daily living aids and adaptations
- information on employment legislation and your rights within the workplace
- graded return-to-work and remaining-in-work programmes

How to get an OT appointment

People with CMT often find it most helpful to see an OT if their physical ability changes and they find it harder to do things that they had previously been able to manage with little effort, such as opening a jar.

OTs work in various settings including community teams, social services and hospitals. The health professionals involved in your care, including doctors, nurses and therapists, can refer you to an occupational therapist if this is required. You may also be able to self-refer to some therapy services – so it is always worth giving your local social services a call. They will explain the correct process for your area. You can also see an OT privately if you are willing to pay. Discuss your concerns with your GP or medical team to identify the services you need.

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 you have difficulty paying, your OT may be able to advise you on seeking financial help.

Just a few items your OT may be able to help you with.

Stretching, exercise, and physiotherapy

Exercise plays a very important part in managing CMT and helping to prevent or ease the complications of the condition. In fact, exercise is more important for people with CMT than for those without the condition.

This is because:

- exercise, combined with a healthy diet, is the main way of controlling body weight. Being overweight or obese puts extra pressure on already weakened muscles and joints
- it 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

Which type for me?

There is no fixed rule about which exercises or activities best suit people with CMT. Do not let your worries about your CMT restrict you from trying different things. Just bear in mind the following points:

- 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)
- talk to your physiotherapist or gym instructor about what would suit you
- if one form of exercise is not right for you, try another (e.g. if walking doesn't work, try cycling)

Exercise does not have to mean going to the gym. The important thing is to make physical activity part of your normal routine. Getting off the bus one stop early, climbing the stairs instead of taking the lift or standing on the escalator, cycling to the station, walking to the shops or school are all good ways of reaching the 30-minute recommended daily minimum.

What kind of exercise?

'Aerobic' exercise is good because it works your heart and lungs. Walking, swimming and cycling for example all help to increase endurance, reduce fatigue, improve mood and improve your ability to do day-to-day activities.

Exercise that strengthens your posture muscles is important to help with your sense of balance - for example, yoga, Pilates and Tai Chi. These forms of exercise will also train your balance responses so you are less likely to fall.

Stretching

Daily stretches are vital for people with CMT to keep hands, feet and ankles flexible. They help 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.

The large number of joints in the human hand enables it 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 will slow down the development of hand deformities. It is important that you get into the habit of doing these stretches **before** deformity and stiffness develop.

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 when brushing your teeth at the bathroom sink or standing at a work

surface while waiting for the kettle to boil. Sinks and work surfaces are an ideal height for support.

Orthoses and stretching

Orthoses can play an important role in helping to maintain flexibility and joint range and in preventing the muscles tightening and shortening. This can be by means of insoles which may be thicker on one side 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 the 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. For more on orthoses see Orthotics p xx.

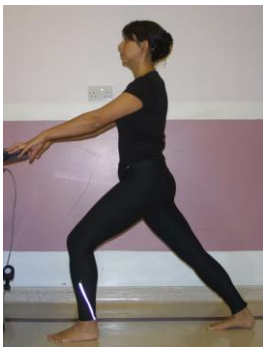
Daily stretches and exercises for people with CMT

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

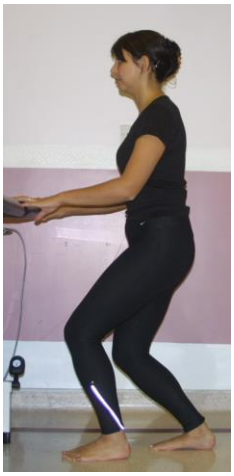
Calf stretch:



- 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

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, and may be felt lower down the back of the leg
- Hold still for 20 to 30 seconds. Repeat three times and then swap legs

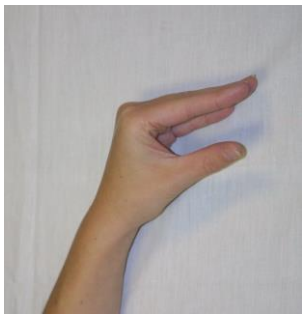


Hip stretch:

- 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

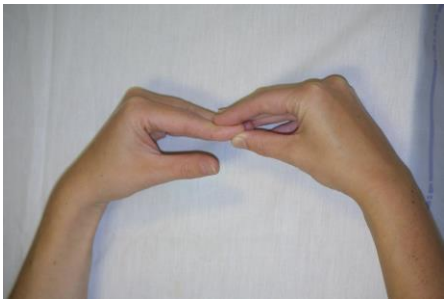
Hand stretch:

- 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 over the back of the hand, **NOT** pain
- Hold for 20 to 30 seconds. Repeat three times and then swap hands



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

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

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

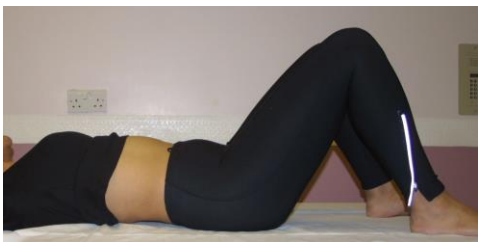
Upper legs:

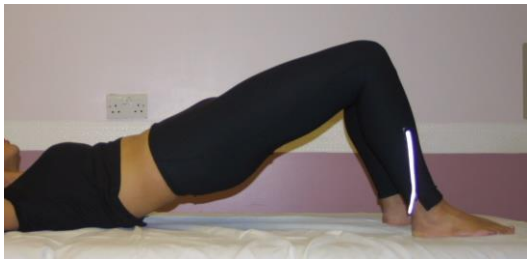
- Sit on the edge of your bed or a dining chair
- Keep feet hip width apart. Keep your arms by side
- Stand up fully then slowly sit down
- Repeat the movement for two sets of 8-12 repetitions

your

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 for two sets of 8-12 repetitions
- 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

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, keep this position and turn your head right and left. Practise for one to two minutes
- If this is easy, extend your arms and rotate them around to the right and left. Practise for one to two minute

Before exercise

Before you start to do any new exercise, 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 ones 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 – 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, both physical and mental.

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. This decline in physical activity has less to do with the 'modern world' and more to do with our own attitudes. As a society, we have become lazier - less inclined to do activities that take effort, preferring to drive a car or stand on an escalator.

Regular physical activity:

- reduces the overall risk of premature death by up to a third
- cuts the risk of coronary heart disease, stroke and type 2 diabetes by up to half
- combined with a balanced diet, is the best way of keeping to a healthy weight
- reduces the risk of osteoporosis
- helps treat mild to moderate depression
- helps you feel better, beat stress and get a good night's sleep
- helps keep mental faculties sharp, particularly into old age
- reduces the risk of falling and of being seriously injured in older people

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 the benefits of low to moderate intensity exercise far outweigh the risks, with no increase in weakness. You can reduce the risk of harm 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, 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 build up your upper arm strength through weight training, you would not expect to lose this effect due to 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 in trying to strengthen extremely weak muscles. However, physiotherapists recommend general exercise as well as (or instead of) weight training, plus 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 was some argument over whether people with CMT should do any weight training. 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 – talk to a gym instructor or physiotherapist to help you begin 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 wellbeing benefits gained from exercise only last a short time. If you stop exercising you will quickly begin to lose the health benefits. This is one reason why you need to exercise regularly. Try doing little and often rather than a lot occasionally.

The general recommendation is that we all need at least 30 minutes of moderate intensity aerobic activity five times or more a week – for example swimming or fast walking. That doesn't mean there is no benefit from doing less. If you can't achieve that much, try to increase your level of activity over time. Even chair based exercises can help.

If possible, you should also do activities on two or more days a week that work the major muscle groups – legs, hips, back, abdomen, chest, shoulders and arms.

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

6	Very, very light
7	
8	
9	Very light
10	
11	Fairly light
12	
13	Somewhat hard
14	
15	Hard
16	
17	Very hard
18	
19	Very, very hard
20	

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.

Children and young people aged five to 18 should do at least 60 minutes of physical activity over the course of every day, which should include both moderate-intensity activity (e.g. playground games and cycling) and vigorous-intensity activity (e.g. sprinting and football). They should also do three 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 an additional focus on strengthening and balance exercises. As we get older, we slowly lose muscle and our ability to balance declines. Studies have shown that regular physical activity reduces the risk of falling and of being seriously injured.

I find walking tough. What would you advise?
If you find walking difficult, then you may prefer swimming or- if you can't swim - water-based exercise. This is because water-based exercises take the pressure off your joints. Talk to your local leisure centre about activities appropriate for you. You could also try cycling, either on a normal bicycle or - if your balance is poor - on a static exercise bike.

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 early enough so that it can be treated and, possibly, prevented – for example, any stress you may be putting on other joints like your knees or hips. This can help prevent secondary weakness causing other problems.

They will also help you devise ways to manage your condition yourself on a day-to-day basis, for example: a suitable exercise programme; aids and strategies to keep you moving well; knowing when to seek help.

You should be able to get a referral to a physiotherapist from your GP or consultant neurologist. If you would like to see a physiotherapist privately, contact the Chartered Society of Physiotherapy for the name of a physiotherapist near you. Ask to see a neurological physiotherapist or a neuromuscular specialist physiotherapist.

Balance

The main muscles that affect our balance when standing are the calf muscles. They hold the ankle joint still and stable. If your calf muscles are weak, you may find that you have problems standing still and move your knees a lot to keep stable.

When walking, the main problem that leads to falls is tripping over dropped feet. Foot drop happens when the muscles at the front of the shin are weak.

In some people with CMT, the muscle that runs down the outside of the calf (peroneus longus) is weak. It is responsible for pulling your foot out and if it weakens, your foot can 'turn in' at the ankle. 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), balance problems and also pain and calluses.

If weak ankles are combined with some loss of sensation in the foot, this makes balance much harder. This is because as well as depending on the stability of the ankle to keep balanced, the body also relies on sensory information from the foot. Without this sensory information, it takes the body longer to recognise that you are about to topple over, slowing your reaction to losing your balance.

Poor balance can lead to trips and falls which in turn can cause ankle sprains and other falls-related injuries.

There are various ways to reduce the chance of falls including:

- orthoses
- occupational therapy (including handholds, assessment of your house and walking aids)
- balance training
- surgery

Orthoses to support the ankles and improve balance

Some people feel more balanced and can walk better if the arches of their feet are propped up, and/or the feet are re-aligned with wedges. There are light braces that can reduce the risk of tripping if you have foot drop.

If you are unable to stand still because of calf weakness, it may be appropriate to try rigid ankle foot orthoses to support and stabilise your weak ankles. Consider 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 look of these orthoses as they are bulky and often require bigger shoes. However, it is worth considering what they do to the appearance of walking: this can often be far more normal and have a bigger impact than the appearance of the orthoses themselves.

By addressing problems at the ankle, it is often possible to improve alignment and improve your strength at the knees, hips and trunk. This is an area where it can be very useful to combine use of orthoses with physiotherapy. For more on orthoses see Orthotics.

Falls

CMT can increase the chance of your tripping and falling. If you are inactive because you are recovering from a fall, or are nervous about falling again, you will 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. There's a risk of a vicious cycle developing: 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.

Most falls in people with CMT occur in their own homes. This may be because people pay less attention to threats to balance whilst at home. It may also be because people remove their supportive shoes and braces.

Think about your footwear and try to clear any trip hazards from your home. Taking particular care on uneven ground can all help you avoid falls.

The falls prevention action plan

Take regular exercise, even if this is only a short walk, to keep muscles as strong as possible and joints supple

Fit easy-grip handrails on both sides of the stairs. Avoid/minimise climbing stairs if it makes you feel unsafe
Keep stairs and living areas well lit. Keep a torch by the bed

Try to keep furniture or clutter encroaching on circulation spaces to a minimum, giving you plenty of room to manoeuvre sticks, crutches or walkers

Never leave objects which you can trip over, on stairs or in walking areas. Avoid flexes and cables crossing walking areas

Use non-slip rubber mats in the bath/shower. Fit a handrail near the bath/toilet. Avoid small rugs in the bathroom

Replace worn rugs and carpets. Nail or tape down the edge of rugs to avoid slips and trips

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

If you must climb, use proper steps

Get up from chairs/beds slowly. Blood pressure falls as you get up and you may feel dizzy if you do it too quickly

Avoid poorly fitting shoes or slippers (talk to a podiatrist)

Have regular eye tests. It is now possible in many areas to have an eye test in your own home

Avoid clothes which may trip you, such as trailing nightdresses or long tracksuit bottoms

Don't rush to answer the telephone. Warn friends that it may take you longer to reach the telephone, and if you have an answering machine, you can probably extend the number of rings before it cuts in. Have an extension socket fitted upstairs, or if you have one, carry a mobile or cordless phone with you

If prescribed medication is making you feel dizzy, keep taking it, but consult yourGP

Keep rock salt/grit handy to put on external paths in cold weather

It may also be worth having a plan for what you would do if you do fall, especially if you can't get up unaided. You may no longer be able to kneel, but can you pull yourself up onto a low chair? If that doesn't work, how will you raise the alarm, if you're on your own?

Consider the following:

Subscribe to a call alarm service. There are many of them (usually run by, or registered with your local authority). They'll provide you with a call button that you wear at all times. To summon help, you simply press the button.

If that is a step too far, consider purchasing a couple of very basic mobile phones – you can get them from places like Carphone Warehouse or Amazon for around £10? Providers like GiffGaff offer cheap call plans or pay-as-you-go calling. Position one phone by the toilet, perhaps, or in the kitchen. At the very least, have a landline phone low enough to reach if you are on the floor.

Having raised the alarm, how are you going to let in the rescuers, whether they are members of the ambulance service (which can be called out for these sorts of emergencies)

or a neighbour? A keysafe is a good option. This is a small cast-iron box containing your key that is secured in an inconspicuous place near your main entrance. The box is opened with a pin number: these are low tech devices that need no electricity. You give the pin number to rescuers or friends/family/carers as required. Any good disability gadgets store or website should stock them - or ask your occupational therapist to help you get one.

Whilst it's scary having a plan in your mind, it's better to be prepared. It may never be needed.

A Keysafe with cover

Pain

Many people with CMT experience pain at some point in their lives – sometimes briefly, sometimes long-term. Generally, 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 different from the person next door. For example, evidence shows that women feel pain differently from men.

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

> Pain due to tissue damage. It may be caused by stresses and strains on your body due to CMT, especially on the bones, joints, tendons and ligaments. This is called musculoskeletal pain, or sometimes 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.
>
> Neuropathic pain, in other words pain 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
- medication
- specialist pain clinics

If you have lived with your pain for a long time you may find that various psychological, social and behavioural training methods and counselling can help you manage your pain and generally function better. Also, physical therapy and exercise can help control pain. For more on this, see Stretching, exercise and physiotherapy, p xx.

Drugs for pain

Everyone responds to drugs differently: if one doesn't work, another may. Remember to take special care if you have another medical condition or take other drugs (including over-the-counter and complementary treatments). If in doubt, check with your GP or pharmacist.

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

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

Non-steroidal anti-inflammatory drugs (NSAIDs)
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 person in ten suffers 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. Drinking plenty of water and eating foods with a high fibre content may prevent constipation
- Some people suffer from drowsiness, nausea and vomiting
- Opioids can be addictive so could give withdrawal symptoms when stopping. This is less likely with weaker opioids like codeine, but can still occur
- Some people find that they have to take higher and higher doses of opiates to get the same level of pain relief, although 'tolerance' – as this is called - is not common

Anti-epilepsy drugs
Anti-epileptic drugs such as gabapentin and pregabalin are used for treating pain arising from some peripheral nerve disorders, either diseases or injuries. In most cases pain in CMT is due to mechanical problems. However in a sub-set of patients it will be neuropathic. Your neurologist can assess if the pain is likely to be neuropathic, in which case these drugs may be recommended.

Antidepressants
Certain antidepressants, in particular a type called tricyclic antidepressants (TCAs) can be extremely helpful in the management of long-term pain. They can give you a dry mouth and can make you drowsy, or constipated but these symptoms usually disappear after a short time.

Other treatments for pain

Other 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)

Pain clinics

There are around 300 pain clinics in the UK. Most are in hospitals and have teams of staff from different medical areas, including occupational therapists, psychologists, doctors, nurses and physiotherapists. They all work together to help people with pain.

Pain clinics vary but usually offer a variety of treatments aimed at relieving long term pain, such as painkilling drugs; injections; hypnotherapy and acupuncture.

You will need to be referred to a pain clinic by your GP or hospital consultant.

You can find your nearest pain clinic by putting your postcode into http://www.nationalpainaudit.org/

Fatigue

We all feel exhausted sometimes, but fatigue due to a condition like CMT can really interfere with everyday tasks and normal life.

There are many definitions of fatigue. Some relate to normal fatigue when we have done too much. For example: *Fatigue is when we have exercised to the point where the task feels more effortful (MacIntosh and Rassier 2002) and our muscles produce less force (Gandevia 2001).*

There are also definitions that match the more general fatigue that people with CMT describe: *Fatigue is an overwhelming sense of tiredness, lack of energy and feeling of exhaustion (Krupp and Pollina 1996).*

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 maintain our stamina and generally stay fit: cutting it out will lead to under-activity.

The causes of fatigue are not fully understood but it is a real symptom that the majority of people with CMT experience.

People with CMT report that fatigue can be triggered by:

- too much activity: overdoing it
- too little activity: where people become lethargic
- stress
- concentrating for long periods
- day to day coping with the limitations that living with CMT presents

Measures to treat 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

Exercise and fatigue

Surprisingly, exercise can help keep fatigue at bay by increasing your stamina, strength and flexibility. 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 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
- 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.

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

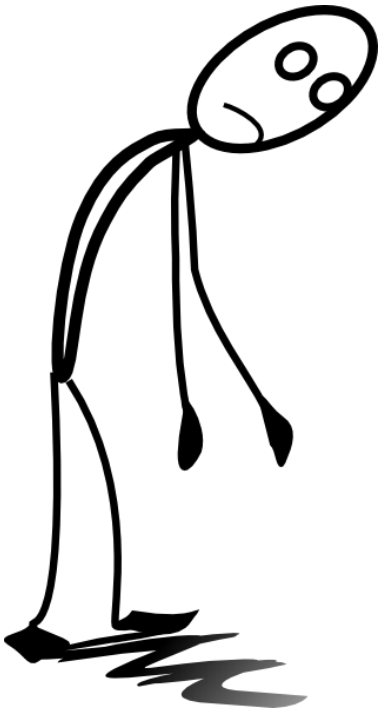
For more on this, see Healthy eating.

Tips on beating fatigue

The following tips should help you save your energy when you are tired. They are only a guide and you may not find that all of them are suitable for you. Strike a balance between staying physically active when you feel more energetic and saving energy when you are more fatigued. For more specific advice on how to conserve energy, consult an occupational therapist.

Arrange the world to suit you
Keep frequently used items where they are easy to reach. Try duplicating household supplies in different areas, e.g. 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. They are easier to use.



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 – make it harder to get around.

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.

Cut out needless effort

Sit rather than stand whenever possible as it takes up a quarter less energy. Try sitting while in the shower, or when you are 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. (Better still invest in a dishwasher!)

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.

Take your time – rushing around really does use more energy.

Think about using some kind of mobility device – a wheelchair (especially if you have someone to push you) or a scooter for longer distances, shopping outings or the like. This is NOT about giving up walking, but about conserving energy for other, more interesting, tasks.

Shopmobility Schemes are located in most large shopping centres – use them!

Plan ahead

Gather all the supplies you need for a task before starting.

Phone the shops before you go to make sure they have everything in stock.

Schedule breaks, making sure you rest **before** getting tired. Try setting an alarm to go off after 30 minutes to remind you. Have a break for 15 minutes and then start again.

Keep a diary to try and identify what activities exhaust you.

Prioritise

Cut out tasks that are not important.

Talk to your occupational therapist to see if any help can be organised for you.

Delegate

Delegate tasks to other family members, or friends – this is not giving up or giving in, but using resources available to you.

A good night's sleep

A good night's sleep can help you restore your energy levels, cope with pain and rest your joints. Resolving fatigue isn't just about getting a good night's sleep, but adequate rest is important in enabling you to get on with your day.

Work out how to get the best night's sleep possible by finding what works for you. It could be painkillers, a hot bath or a complementary remedy. If you feel listless and fed up in the daytime it can be tempting to sleep, but spending too long in bed can mean that you do not wake up feeling refreshed.

A comfortable and peaceful environment is also conducive to a good night's sleep. If the room is too hot or too cold, this could disrupt how well you sleep. A pillow should give you the right cushioning to support head and neck properly. According to the British Sleep Council, a pillow should hold your head in the same position to your shoulders and spine as if you were standing with the correct posture. Your choice of bedding can also have a bearing – heavy bedclothes resting on your feet can be uncomfortable or even painful.

Learning to relax before you go to bed can help. Some people find reading a book or listening to music helpful. Relaxation exercises may work for those who find it harder to switch off. Well-known relaxation methods include deep breathing, progressive relaxation (tensing and relaxing different muscles in turn), meditation and imagery.

Coping with CMT

Most information on CMT describes the physical effects of the condition or focuses on 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, so ongoing psychological ill health, such as stress, can directly undermine your physical health.

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

Because CMT changes and develops over time, you may have to adapt the way you manage and cope with the condition. You will need to be flexible in the way you deal, not only with the changes in the condition itself, but also with the resultant changes in your life, including work, family and home. Many people experience a wide range of feelings, and it's worth developing strategies to deal with both physical and emotional challenges.

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. The impact of chronic illness or disability can produce challenges at work, home or in social settings and these situations can contribute to stress or anxiety.

A little 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). However, persistent stress has been shown to be bad for both our mental and physical health – dampening our body's natural defences. Ongoing negative stress is a contributing factor to depression and should, be tackled early.

Common causes of stress include:

- long-term illness
- uncertainty about the future
- the unpredictability of CMT or other

condition
- disability
- lack of control
- financial difficulties

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. Here are some suggestions:

- 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. Even virtual friends whom you find on social media can help immensely, especially if they share your problems. Find CMT United Kingdom on Facebook
- 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 – tobacco, alcohol and drinks like coffee, tea and cola are all stimulants. Rather than calming you down, they 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. 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. Avoid caffeine and alcohol in the afternoon and evening and do not watch TV just before bedtime. For more on this, see Fatigue.
- Rest – if you are ill, do not carry on regardless
- 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 why. It may help you identify and deal with the root cause of the stress
- Manage your time – plan ahead and take things one at a time. It can be helpful to plan time 'buffers' so that you have space to deal with unexpected events
- Be realistic – in the words of the Serenity Prayer, learn "serenity to accept the things I cannot change, the courage to change the things I can, and the wisdom to know the difference"

Depression

While CMT itself does not cause depression, if the problems caused by the condition are not successfully managed, the ongoing strain and stress may make you more susceptible to mental illness, including depression.

It's not uncommon to feel 'down' at times or to experience dips in motivation and energy levels. This shouldn't be confused with more serious symptoms of clinical depression or anxiety. Depression is a recognised illness and treatment is available.

In the words of the Depression Alliance, if you are depressed you have an illness which means that "intense feelings 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 may include (with thanks to the Depression Alliance):

- feeling tired and loss of energy – bear in mind that CMT will already make you feel more tired than 'normal'
- 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.

Contact the Depression Alliance for more information on depression. See Chapter 6, Organisations that can help you

Self-help

Acceptance
Accepting the fact that you have CMT and that it is likely to have an impact on your life can be challenging but it is an essential step to developing your physical and emotional wellbeing. Only through acceptance can you start to take responsibility for your health and put everything into perspective.

Taking control
Although it may seem that you've lost control of some parts of your life, it is important to take control of others. The following are examples of how you may want to take more control.

- Knowledge – generally, the more understanding you have of a condition, the better you feel and respond to treatment. Do be aware, however, that there are huge variations among people with CMT (even within the same family) when it comes to symptoms. There is still no way to predict how CMT may progress or impact your life so don't make assumptions based on the experience of others.
- Planning – CMT may diminish your energy levels or restrict your mobility so planning ahead becomes more vital. Taking control can involve thinking about logistical issues such as physical access to buildings and transport, and allowing more time to do things and get about.
- Perspective - there's no denying the difficulties associated with CMT, but focusing on limitations only contributes to frustration and stress. Rather than dwelling on what you CAN'T do, try to focus on what you CAN. Positive thinking can make a surprising difference.
- Problem solving – By getting proper support and by breaking a task down into its individual components you may be able to find a way to do things you thought were impossible.

Contact with others
Talking to others at home, work or leisure is a valuable way to enlist support, explore solutions and boost your morale. Share any concerns you have with:

- 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 ways to manage it.
- CMT United Kingdom – talk to others like you on our Facebook page, visit one of our local groups or call us for a chat. You will be able to share experiences and learn methods of overcoming any difficulties you may face.

Talking about your CMT

This section highlights the importance of talking to various people about your CMT. These may include:

- family and friends
- employers and colleagues
- your children
- government agencies and financial institutions
- educational establishments
- health professionals
- support groups

When and how you tell people about your CMT is largely up to you, although they may initiate the conversation with questions, sometimes when you first meet.

Usually, people do not want a detailed account of your medical history. You're likely to adapt your responses depending on the background of the person you're talking to and your relationship with them. It can help to have a few phrases ready for different circumstances. Be prepared to educate: people are likely to know nothing about CMT and what it means for you, so it may be helpful to have some facts about the condition at your fingertips.

For official business it can be helpful to have a brochure such as those produced by CMT UK 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 talking to. You may find it nerve-racking because it feels like a violation of privacy. Alternatively, you may feel quite relaxed and prefer to be open and direct.

As with any medical condition, it can be tough deciding who to tell, as well as when and how to tell them. Each situation is different so it's impossible to generalise about the benefits of sharing your concerns with others. However, it's worth remembering that old adage 'a problem shared is a problem halved.'

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 frequently suffice. For example, for questions such as, 'Why do you walk like that?', or 'Why do you wear those shoes/orthoses', you could try saying: 'I have weak ankles because of a problem with the nerves to my legs. My nerves fire inefficiently which makes my muscles weaker.'

A life 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 people at work

You're not required by law to tell your employer that you have CMT unless it poses a health and safety risk. It's worth checking what, if anything, your contract requires you to disclose.

Telling your employer that you have CMT may seem daunting, but there are many advantages. Firstly, even if your condition doesn't affect your work now, you may need time off for medical appointments or may need assistance in future.

Talking to colleagues may also be useful. On an individual level, it can help others to understand the challenges that you're dealing with and make it easier for you to request help, if necessary. On a broader level, talking openly about chronic illness and disability is also a way of tackling the stigma that many people experience regarding both physical and mental health issues.

Telling your adult friends and family

Whoever you are telling, their reactions can be unpredictable, and possibly quite emotional. They could be very upset, even angry: on the other hand, they may seem entirely uncaring. They may ask questions that seem intrusive or even offensive – 'How did you catch it?' 'Is it contagious?' - so steel yourself and be prepared.

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 is a chance that they have inherited the condition too. For more on this, see Chapter 2, Genes: what they mean for you.

Telling children that you have CMT

It may help to bear these points in mind when considering whether to tell a child that you or a close family member has CMT:

From an early age children usually know when something is wrong or different. Receiving an explanation in simple and direct terms that they can share with others, if they choose, can prevent them from being isolated or excluded. Children also sense when adults are being less than honest, so in order to maintain trust and empathy, it's best not to fudge the truth
Very young children may be relieved to be told that your CMT is not their fault ('If I'd be been good', etc.)

Children have different levels of understanding and will need information in a language they can relate to. Their GP or 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. Invite them to ask questions and be prepared to keep addressing the same questions in different ways. If they seem reticent or unwilling to talk, respect their need for time and space to digest the information

Because children may need time to process their understanding, they may not seem overly curious or interested when you first raise the subject. It's quite likely that questions will emerge some time later, maybe days or weeks after initial discussion

Telling a child that he or she has CMT

'To tell or not to tell' is a perennial question and there's no correct answer. Probably the best advice is to follow your heart and your intuition within the parameters of your family and your child's ability to understand the information.

Children with CMT may well be aware at an 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 work out very quickly - from the reaction of others and the fact that they may not be able to do the same activities as their friends - that there is something different about them.

There is a strong case for giving your child knowledge appropriate to her/his maturity so that he/she can answer the same kinds of questions you are asked. Ability to deal with the reactions of others is a prime way of building confidence and self-esteem. As soon as children enter school, they are under scrutiny by their peers. Any perceived difference (freckles, smaller or taller, red hair, etc.) can give rise to questions or in worse situations, bullying.

Help your child to understand what support is available in school such as talking to peer buddies or form teachers. Encourage them to talk openly to you about what happens in school, both in the classroom and in the playground.

If you think it appropriate – for example, if your child is shy, uses a wheelchair, or may need help or understanding in non-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, it may add to a sense of insecurity and even inferiority. Children can be very tactless and even cruel. A child who 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 in childhood can lead to low self-esteem throughout adult life.
Honesty, a sense of humour and plenty of encouragement will help children come to terms with their physical difficulties and give them confidence.

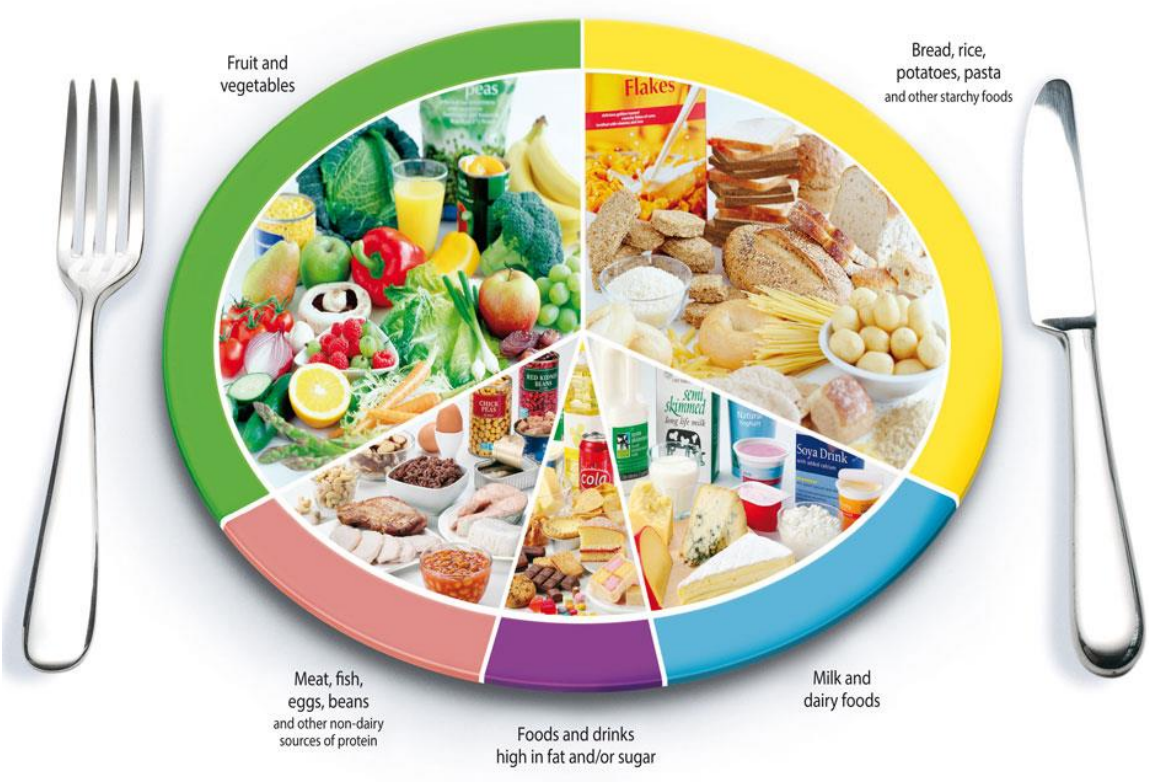
Dealing with your own responses
It is quite normal and common for parents, when faced with their child's diagnosis, to go through similar emotions to those they would have if they had been diagnosed with the condition themselves, including denial. In metaphorical terms your 'ideal' child has 'died' and you must realign your hopes and dreams with the child you have been given. For more on this, see Chapter 6, Appendix 1, Models of adjustment to disability. Your child will probably not star as an athlete or ballerina but how important is this to you? Choices can 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 is obviously inherited. This can be difficult 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, parents and those with the condition will find that these challenging periods of doubt, confusion and guilt will pass.

Healthy eating

Getting the Balance Right

The eatwell plate



The eatwell plate demonstrates the ideal balance of foods to have over a day. It splits food into 5 groups: bread, rice, potatoes, pasta and starchy foods; milk and dairy food; fruit and vegetables; meat, fish, eggs, beans and non-dairy protein; and food and drinks high in fat and/or fat and sugar. Each group has a role within our diet and provides different nutrients.

Bread, rice, potatoes, pasta and starchy Foods

These foods should count for a third of your daily intake, so try to have one at each meal; for example cereal or toast at breakfast, bread/potatoes/pasta/couscous at lunch and evening meal.

Starchy foods provide slow-release energy and help fill you up as well as supplying essential minerals and vitamins. Try to choose wholemeal products when you are able - the fibre they contain keeps you fuller for longer, helps regulate bowels and can lower cholesterol.

Most the 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. Therefore, cutting out carbohydrates can aid weight loss. However, these diets can have side-effects and are difficult to maintain. A healthier way to lose weight is to opt for lower-fat products to go with your starchy foods.

Fruit and vegetables

Fruit and vegetables are an important source of fibre, minerals and vitamins. Aim for five portions a day, but try to vary it throughout the week to ensure a wide range of nutrients. The more portions you have the better the protection against cardiovascular disease and some forms of cancer.

Fresh, frozen, tinned, dried and juiced all count. A 'portion' could be one of the following:

- one glass of fruit juice
- three tablespoons of vegetables – raw, cooked, frozen or canned
- one dessert bowl of salad or stewed fruit
- one medium-sized fruit, - apple, orange, banana
- two small fruits – plums, apricots, kiwis
- a small handful of grapes or berries
- one heaped tablespoon of dried fruit

Milk and dairy Foods

Milk, cheese and yogurt are the foods included in these group. They are a good source of protein, calcium and other minerals.

Aim for two to three portions a day. A portion is a small matchbox of cheese, 200ml (a small glass) of milk and 125g of yogurt (a typical small pot).

Dairy foods can be high in fat, particularly in saturated fat (the type that increases the risk of cholesterol deposits in your arteries). Choose low-fat versions, e.g. semi-skimmed milk, half-fat cheese and low-fat, low-sugar yogurt, to help you keep to a healthy weight and reduce your fat intake.

Meat, fish, eggs, beans and other non-dairy protein

This food group is the main source of protein in the diet and also provides vitamins and minerals, e.g. iron. Beans and pulses also provide fibre. Try to have two helpings of foods from this group 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.

Some meat is very high in fat. Choose leaner meat and methods that reduce the fat content, e.g. grilling, rather than those that increase the fat content, e.g. frying.

Foods and drinks high in fat and/or sugar

Cakes, biscuits, chocolate, crisps, fried foods, cream, fizzy drinks, butter, oil, sweets and pastries are all high in fat, sugar or both. Because these foods are often high in calories and provide little other nutrition, they are often known as "empty calories".

It is important to choose a variety of foods from the first four groups every day to get a wide range of nutrients. Remember, the fifth group foods should stay as treats.

Keeping to a healthy weight

Dealing with too much weight

Being overweight or obese increases the risk of cardiovascular disease, some cancers and type 2 diabetes as well as decreasing mobility, increasing joint pain and breathlessness. The following factors all contribute to being overweight:

- reduced mobility/lack of exercise
- fatigue
- convenience foods
- eating for comfort or from boredom
- reduced muscle mass (muscle uses more calories than fat)

Losing weight or preventing weight gain can be difficult and sadly there is no magic solution. Here are some ideas to help you reduce the number of calories you eat and use more by exercise.

- Eat a healthy balanced diet. Using the eatwell plate, reduce the foods high in fat and sugar and increase your intake of fruit and vegetables
- Reduce your portion sizes
- Exercise as you are able. See Stretching, exercise and physiotherapy, p xx
- Plan your meals
- Do not go to the shops when hungry. Take a shopping list – and keep to it!
- Avoid having tempting foods at home

- Choose healthy and low calorie snacks such as fruit, vegetables, plain cereal bars and plain popcorn (do not add salt)
- Read labels carefully. If they are traffic-light based, choose products that are mainly green and yellow

If you're having trouble with keeping weight off, you'll find useful information at the British Dietetic Association's Weightwise website at http://www.bdaweightwise.com/

Being underweight

Being underweight should be avoided as much as being overweight. Remember, it is not always easy to recognise that you are underweight and weight loss often occurs over a number of months or years.

Reasons for weight loss include:

- difficulty eating and drinking
- difficulty in preparing meals
- using more calories than usual, for example during periods of shortness of breath when your body is working harder

Some of the problems associated with being underweight include:

- becoming physically weaker (fatigue)
- being less able to fight infections
- increased risk of wounds
- being less mobile
- reduced ability of the heart and muscles involved in breathing to work properly
- depression

If you are underweight you should be referred to a registered dietician - you can ask your GP or any doctor you see to do this. To help gain weight or prevent weight loss there are a couple of changes you can make while waiting for your appointment:

- use full-fat and sugar products
- try to eat little and often - aim for two snacks and three small meals
- choose drinks that provide calories - milkshakes, smoothies, milky hot chocolate, coffee or malt drinks
- add extra calories (fortify) to meals and snacks - e.g. cream/cheese to soup, stews or sauces; butter or cheese sauce to vegetables and cream/ice cream/custard to puddings
- increase the section of foods high in fat and sugar on the eatwell plate in your diet

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 may be harmful. People with CMT should be particularly cautious of mega doses of vitamins A, B6 (Pyridoxine) and D. (A mega dose 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 dietician before taking regular supplements. This is particularly important if you are thinking of taking a combination of supplements.

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

Alcohol

Alcohol is a toxin (poison) and can damage muscle and nerves. If you are generally healthy, not on certain medications and drink sensibly, then alcohol should not harm you.

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, i.e. 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.

Practical Issues

It's often the simple day to day tasks that can make CMT feel like the end of the world is upon you, particularly when they become difficult or tiring. But there is help available. It's all about knowing where to find it – and that's what this section is for. For more on specific websites, organisations, stores and resources, see Chapter 5, Organisations that can help you.

Daily Living

Because your balance and proprioception (the awareness of where your body is in space) are impaired due to poor sensory feedback, make sure your home is lit well, with nightlights in strategic spots. Fit any dark areas in and around your home with lights on automatic sensors. Be especially careful if the ground is wet or slippery. If you must venture out in snow or ice, buy grips to attach to your shoes.

Personal Care/Dressing/Bathing
There are many gadgets to help you with dressing, bathing and toileting. You can easily buy smaller gadgets from specialist stores, both in the high street and online. These can make a big difference. For example, a long handled shoe horn not only helps with shoes but can be used to hook clothes to pull them up. Make life easier for yourself – think Velcro instead of buttons, t-shirts instead of shirts.

For larger items, such as gadgets to get you in and out of the bath, advice on replacing your bath with a shower, or toilet raisers and hand rails, always seek advice from an occupational therapist. Not only will they help locate the right article for you, but they may find ways to assist with the cost, which can be substantial.

If you need assistance or support to live an independent life, ask for an assessment by your local authority. If they agree that you require support, you can receive Direct Payments (also known as Personal Budgets) to allow you to purchase your own care services. These are not social security benefits and will not affect your entitlement to benefits. If you would rather not have the hassle of employing and sorting out your own care, the local authority is obliged to provide the services you need once you have been assessed as requiring care.

In the Kitchen
Cooking can present challenges – for example dealing with hot and heavy pans. There are a huge range of gadgets than can help, from simple jar openers to specialist pans designed to be easy to carry. Many are available from supermarkets as well as specialist stores. Online stores such as Amazon or Lakeland are also full of useful gadgets. For a few of the many suppliers out there, see Chapter 5, Organisations that can help you, p xx.

Don't forget, if sensation in your hands is impaired, you are more at risk of burning yourself without realising it. Make sure you protect your hands well, and be extra careful.

Adapting your home

Always get the advice of an Occupational Therapist before embarking on major adaptions to your home, to make sure that you're doing the right things and that there aren't funds available to help with the cost.

- **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 to your home are necessary, an application will be made for the grant to your local Council. Grants can only be applied for by an occupational therapist. Typical work can include: improving access into the bathroom, living room or bedroom; providing or adapting suitable (or additional) bathroom facilities; alterations to the kitchen to make it accessible or larger; improving general access to a property, or providing a stair lift. 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. Grants are also means tested, and subject to various other conditions – check the latest information on www.gov.uk.

Value Added Tax

If you have CMT, you won't be charged VAT on products designed or adapted for your own personal or domestic use, as long as the product qualifies. Your supplier can advise you, but products designed or adapted for a disability usually qualify. For example, certain types of:

- adjustable beds
- stair lifts
- wheelchairs
- medical appliances to help with severe injuries
- alarms
- motor vehicles - or the leasing of a Motability vehicle
- building work like ramps, widening doors, installing a lift or toilet

Your disability must also qualify for VAT relief. For VAT purposes, you're disabled or have a long-term illness if:

- you have a physical or mental impairment that affects your ability to carry out everyday activities – like CMT
- you have a condition that's treated as chronic sickness, like diabetes

You'll need to confirm in writing that you meet these conditions. The supplier may give you a form for this.

Getting around outside the home

Again, there is a huge range of walking sticks, crutches, walkers, wheelchairs (both manual and powered) and scooters to choose from. Your physiotherapist will advise you on the best types of sticks and crutches – go for ones with easy hand grips (ergonomic grips) as they will be less difficult to hold. Hiking poles are often a good solution.

Wheelchairs

Wheelchairs are often most helpful outside your home for covering longer distances and for crossing rougher terrain. Understandably, many people are reluctant to use a wheelchair, feeling that they are 'giving up'. Look upon it as a way of taking back some freedom of movement when out and about, rather than any sort of failure on your part. They are just another 'gadget', when all's said and done.

Wheelchairs are available through the NHS (not social services). 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. An assessment should take into account your physical needs as well as your social ones, plus the environment in which you live and work. Search wheelchair services at www.nhs.uk — the page includes a link to all wheelchair service centres.

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 and not at every wheelchair service).

Budget issues mean you will often be offered the cheapest wheelchair suitable, but the assessors should take into consideration all circumstances. For example, a lighter version should be considered if you have arm weakness which would make it difficult to use a heavy chair independently or if you or your carer could not lift a heavier chair into a car.

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 or Personal Independence Payments, you may be able to buy a wheelchair on preferential terms through Motability. You can get help choosing the right wheelchair for you from the Disabled Living Foundation and the Disabled Living Centres Council.

It is probably worth doing some research into what kinds of chair might suit you before heading to the local wheelchair services – there are many stores on the high street which sell wheelchairs of all varieties, and most are very willing to let you 'try before you buy'. Then, at least, you can ensure that you are fully informed about the options.

The Shopmobility Scheme is also ideal for excursions around your local shopping centre. Most large centres have a Shopmobility scheme operating, and can loan you a scooter or a powered wheelchair for the duration of your visit. Check your local directory listings for where they are located.

Driving
You will find masses of information for disabled people on the legalities and practicalities of driving on the www.gov.uk website. Also, check out Disabled Motoring UK www.disabledmotoring.org for information on everything from insurance, to blue badges, to the driving test.

If you qualify for the highest rate of the mobility component of Disability Living Allowance, or the enhanced mobility component of Personal Independence Payments, then you will also qualify for:

Vehicle Excise Duty (Road Tax) exemption. Ask the Benefits Agency to provide you with an Exemption Certificate, and present this to your local Post Office (which usually deals with Road Tax applications) when taxing your car, together with your vehicle's registration document, insurance and MOT. They will make the necessary changes, and provide you with a receipt to prove that your car is taxed. Remember, no tax disk is issued anymore.

The Motability Scheme. If you are getting the higher rate of the Mobility Component of the DLA or the enhanced rate of Mobility Component of Personal Independence Payment (or the War Pensioners' Mobility Supplement) you may be able to get help on preferential terms through Motability with leasing a suitable car, including wheelchair-accessible vehicles. There is also limited financial assistance available for advance payments or adaptations. www.motability.co.uk

There are other advantages to having those benefits, like free entry to the congestion zone in London (for tax-exempt cars), free use of certain toll bridges etc.

If you are having difficulty driving, and feel that your driving ability should be reassessed, contact the Forum of Mobility Centres. This is a network of 17 independent organisations covering England, Scotland, Wales and Northern Ireland offering professional, high quality information, advice and assessment to people who have a medical condition or are recovering from an accident or injury which may affect their ability to drive, get into or out of a motor vehicle. There is a central contact freephone number for details of your nearest centre (0800 5593636) or go to www.mobility-centres.org.uk

There are a wide range of adaptations available for your car. Rica (www.rica.org.uk formerly Ricability) is a good place to start your research. It has excellent, impartial guides on everything from getting in and out of a car, how to lift a wheelchair in and out, and driving adaptions.

Driving and CMT: 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, must fill in form CN1 and give written consent to DVLA for them to contact your doctor for information concerning your medical condition. On the www.gov.uk website, CMT is classified under the 'peripheral neuropathy' section and the form can be downloaded from there. Failure to inform DVLA is a criminal offence that may be subject to a fine of up to £1000.

Many people with CMT are considered safe to drive, both now and in the future and are given a full 'till 70' driving licence, although it is common to be issued with a three year licence when first declaring the condition. 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. The taking of certain drugs may also have an impact on your ability to drive.

Insurance companies will take the view that insurance cover is invalid if a medical condition has not been declared to themselves and DVLA.

Blue Badge

The Blue Badge scheme provides help with parking for 'people with severe walking difficulties who travel either as drivers or passengers'. It is recognised, with variations, throughout the European Union, and you may even find that your badge is also usable in other countries like the USA, Canada and Australia – see if you can find out the situation before you go abroad. If in doubt, take the badge with you anyhow – you might be lucky!

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:

- exemptions in some cases from limits on parking times (very rare outside 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, among them certain city centres (including some London boroughs), some airports and private roads.

You may qualify for a Blue Badge if you:

- get the higher rate of the Mobility Component of the DLA (or get the War Pensioners Mobility Supplement), enhanced Mobility PIP, or points or more on the "Moving Around" criteria of PIP
- 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 contact your local social services department, or search for more information on www.gov.uk However, each local council appears to have its own criteria, and if you don't qualify due to receiving PIP or DLA, it is increasingly difficult to get a Blue Badge in some areas. Don't be afraid to challenge the decision, and be prepared to back up your case with letters from your consultant - and anyone else you can think of.

Disabled Persons Railcard

A Disabled Persons Railcard is valid for 12 months (or 3 years, at higher cost) and will give you up to a third off the cost of rail travel.

To qualify you must receive at least one of the following:

- Attendance Allowance
- Disability Living Allowance/Personal Independence Payments (in the Higher Rate for help with getting around, or in the Higher or Middle Rate for help with personal care)
- Severe Disablement Allowance
- War Pensioner's Mobility Supplement.
- War or Service Disablement Pension for 80 per cent or more disability.
- or you are buying or leasing a vehicle through the 'Motability' scheme.

Children aged five to 15 may also be eligible for a Disabled Persons Railcard, if they meet at least one of the required criteria. While they pay the normal child's fare, an adult can travel with them at the discounted rate of one third off the adult fare.

There is a cost involved in purchasing a card, but if you use the train frequently, it will save you money in the long run.

If you need help travelling by train, for example getting from your car to the train, contact National Rail Enquiries 24 hours in advance, especially if you are travelling with a scooter or wheelchair, as there is limited space on most trains.

Many stations have toilets accessible to wheelchair users that can be opened using the National Key Scheme key, available from RADAR.

Visit the Disabled Persons Railcard website www.disabledpersons-railcard.co.uk or call 0845 605 0525 for more information.

Employment and the Law

It's against the law for employers to discriminate against you because of a disability. The Equality Act 2010 (which replaced the Disability Discrimination Acts of 1995 and 2005) protects you and covers areas including:

- application forms
- interview arrangements
- aptitude or proficiency tests
- job offers
- terms of employment, including pay
- promotion, transfer and training opportunities
- dismissal or redundancy
- discipline and grievances

Your workplace
An employer has to make 'reasonable adjustments' to avoid you being at a disadvantage compared to non-disabled people in the workplace. For example, adjusting your working hours or providing you with a special piece of equipment to help you do the job.

This could include:

- a phased return to work – e.g. working flexible hours or part-time
- time off for medical treatment or counselling
- giving another employee tasks you can't easily do
- providing practical aids and technical equipment for you

Recruitment
An employer who's recruiting staff may make limited enquiries about your health or disability.

You can only be asked about your health or disability:

- to help decide if you can carry out a task that is an essential part of the work

- to help find out if you can take part in an interview
- to help decide if the interviewers need to make reasonable adjustments for you in a selection process
- to help monitoring
- if they want to increase the number of disabled people they employ
- if they need to know for the purposes of national security checks

You may be asked whether you have a health condition or disability on an application form or in an interview. You need to think about whether the question is one that is allowed to be asked at that stage of recruitment.

Redundancy and retirement
You can't be chosen for redundancy just because you're disabled. The selection process for redundancy must be fair and balanced for all employees. Your employer can't discriminate against you because of your disability - you're protected by the Equality Act 2010. They must also keep your job open for you and can't put pressure on you to resign just because you've become disabled.

Time off work
If you're an employee and can't work because of your disability, you may be able to get Statutory Sick Pay (SSP). Some employers have their own sick pay scheme instead. If you still can't work after 28 weeks, or you can't get Statutory Sick Pay, you can apply for Employment and Support Allowance. Time off from work should not be recorded as an 'absence from work' if you're waiting for your employer to put reasonable adjustments in place.

Benefits

The benefits system in the UK is in a constant state of change and revolution, making it impossible to go into any detail in this book.

For the latest information on benefits, go to www.gov.uk and search disability benefits.

Briefly –

- if you have major issues with daily living or mobility, you may be able to apply for Disability Living Allowance (up to aged 16) or Personal Independence Payments (16-65) or Attendance allowance (over 65). All these benefits require huge form filling and may include in-person assessments
- if you can no longer work – apply for Employment and Support Allowance
- if you are a carer, you may qualify for Carers Allowance

- if you have a child who qualifies for the higher rate of DLA, you should get increased Child Tax Credits

For impartial advice (but not personal advice) and detailed guides on applying, check out www.benefitsandwork.co.uk

Benefits and Work
Guides you can trust

Holidays and Leisure

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 are accessible to your needs.

Visit the Open Britain website for a huge range of ideas and places to visit.
www.openbritain.net

Travelling Overseas
It is extremely important to check in advance and make your needs very clear when travelling overseas, as the scope for things going wrong is great, and there is likely to be less legal recourse. One way can be to book through a disability specialist travel agency or tour operator. Tourism for All www.tourismforall.org.uk is a great place to start your research.

If you are booking a flight and need assistance at the airport or on board the plane, specify this when making the reservation, or not later than 48 hours before departure. Travelling by air is not covered by the Equality Act 2010. However, the Act does apply to the use of services in the UK, like booking systems and airport facilities including shops and check-in.

Since July 2007, it has been illegal for an airline, travel agent or tour operator to refuse a booking on the grounds of disability, or to refuse to allow a disabled person to board an aircraft when they have a valid ticket and reservation. This applies to any flight leaving an airport in the European Union, and also to flights on European airlines arriving in the EU. The law also covers people with reduced mobility, including those with a temporary mobility problem. In rare circumstances these rights may not apply - for example, where there are legitimate safety or technical reasons why a disabled person cannot board an aircraft. In such cases, you must be told the reasons and offered a reasonable alternative.

New rights for disabled passengers were introduced in July 2008, designed to ensure a seamless level of service at airports and on board aircraft.

Don't forget, you MUST declare CMT when buying travel insurance. There are a number of specialist and mainstream insurers who will cover people with pre-existing conditions.

Leisure

There are a number of organisations dedicated to making sports and leisure more accessible for people with disabilities. For more on this, see Chapter 5, Organisations that can help you.

The sports covered include – among many others - water-skiing, horse riding and scuba diving. Ultimately, you are the best judge of 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 less active moments, there are numerous arts-based organisations that can help you enjoy culture, even if you do have difficulty getting around.

Gardeners should seek out the many companies and organisations that can help with specially- designed tools and techniques. The charity Thrive offers advice and information, including an informative website www.carryongardening.org.uk

It is not possible to cover all leisure activities here, but local organisations of people with disabilities may be able to 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.

Chapter 4: CMT and your family

CMT and your family

Like any medical condition, CMT can have an impact on family life. In this section we deal with some of the issues than can arise if you or a member of your family has CMT.

Relationships

If you want to talk to someone about personal or sexual relations there are various options including:

- your GP
- specialist voluntary organisations that help people with family planning, relationship or sexual issues. Relate, www.relate.org.uk, offers a counselling service for adult couples (whether married or not) who are having relationship difficulties.

Having a baby

Generally speaking, there is no reason why you shouldn't have children of your own. However, there are issues you need to consider.

If you or your partner has CMT, you have up to a one in two chance of passing the condition onto your child. For more detail on this see Chapter 3, Genes: what they mean for you.

In rare cases, pregnancy and labour can cause your symptoms to worsen. When you become pregnant, your antenatal team needs to be fully aware of the implications of CMT

Of course, making the decision to have what could be a disabled child is a huge one, and needs careful thought. You'll need to discuss this with your partner – do they want to take this risk? How you feel about your own CMT, and the impact it has on your life will make a huge difference to which way you go. Remember, there **is** no right or wrong choice here – it's entirely up to you. For more on the emotional issues, see Chapter 3, How to Manage your CMT: Coping with CMT. Also, talk to others via Facebook, social media, or CMT United Kingdom, or ask for a consultation with a genetic counsellor to get the information that you need to make an informed decision. For more on practical issues, see Chapter 2, Genes: what they mean for you: Planning a family.

Pregnancy

Women with CMT frequently ask about whether pregnancy will affect their CMT or whether CMT will affect their pregnancy.

Some women report a worsening of symptoms during pregnancy, more unsteadiness or more gait instability due to the increased weight. If you already have walking difficulties before pregnancy, they may be worse in late pregnancy. However, there is no clear evidence that pregnancy can affect CMT.

Women with CMT do not seem to have a greater risk of complications during pregnancy and delivery. You may suffer from fatigue, which can be significant during a long labour. There is no evidence that you might need a particular type of delivery because you have CMT, but you should inform the midwife and/or the obstetrician about your condition.

Childbirth

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 explaining the condition to them. Give them 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.

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 you already have hand weakness, you may have problems after you have given birth, for example when handling your baby or supporting it while breastfeeding.

Organisations such as the Disabled Parents Network (www.disabledparentsnetwork.org.uk) can provide advice and support about the huge range of adaptive equipment is available. Above all, though, don't worry – you will find a way – people with CMT always do. And remember, there is no right or wrong way of doing things, despite what some people may say – what is right for you is all that matters.

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. For more information, contact your local council.

Education

Most children with neuromuscular conditions (like CMT) 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.

You should 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.

Things to consider

The earlier you address any issues that may affect your child's education and overall school experience, the better.

Children who find it hard to write due to weakness, fatigue, cramp, associated joint laxity should have a handwriting assessment. Young children may benefit from aids such as a pencil grips, a writing slope or an iPad. As they progress through primary school a laptop and keyboard skills tuition, may be useful. Later, they may need additional time for rest breaks in exams.

The school should be made aware if your child is more severely affected by CMT, to ensure that they can safely access all areas of curriculum. This could include:

- assistance dressing/undressing and in PE
- help with a school lunch tray
- an adult on hand in case of falls in break time
- help with practical lessons e.g. lifting saucepans in food technology, using equipment in DT
- ability to leave lessons early in senior school to avoid crowds on stairs - or use of lift
- provision of a locker to avoid carrying too many books in a rucksack, which should always be carried on both shoulders!
- a second set of textbooks (if needed) at home to avoid carrying books to and from school
- transport to and from school if using public transport is problematic. Only certain pupils will qualify

The best source of information on the statutory obligations of schools and local authorities is Contact a Family, (www.cafamily.org.uk) a national charity supporting parents of disabled children (with any kind of condition). They have resources explaining the latest legislation, which changed as of Autumn 2014. The rules are also different for England, Scotland, Wales

and Northern Ireland, so you will also need to take that into consideration. Contact a Family also operates a free helpline that will help you with these changes to the system.

Help 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 people with CMT or their carers include:

Carers Allowance – may be available to you if are you looking after someone who is severely disabled, a disabled child or an elderly person
Council Tax Reduction – some carers and disabled people can get reductions in Council Tax depending on their level of income
Personal Independence Payment – this has replaced the Disability Living Allowance for new adult applicants
Disability Living Allowance - may be available for a child who has difficulties walking or needs more looking after than a child of the same age who doesn't have a disability

You can find out more about benefits and how to claim them at www.gov.uk.

If you are a Carer

For guidance and advice on a wide range of things to consider if you are a carer, go to Carers UK (www.carersuk.org). The website has information on your legal rights under the latest legislation, benefits, assessments and much more. They also provide peer support for carers and campaign to improve the lives of carers.

Parents as carers

Parents may also come under the category of 'carer'. 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. Disabled Parents Network and Contact a Family can be particularly helpful.

Chapter 5: Organisations that can help you

Health Issues

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 0800 652 6316; emailing info@cmtuk.org.uk or writing to CMT United Kingdom, 98 Broadway, Southbourne, Bournemouth, BH6 4EH.

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, 98 Broadway, Southbourne, Bournemouth, BH6 4EH
Tel: 0800 652 6316; Web: www.cmt.org.uk; Email: info@cmtuk.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.
Web: www.csp.org.uk. Enquiry Service 020 7306 6666;

Depression Alliance - http://www.depressionalliance.org/

Muscular Dystrophy UK – provides practical, medical and emotional support to people with all forms of muscular dystrophy, including advocacy and has a wealth of excellent information and publications that are relevant to people with CMT.

Muscular Dystrophy UK, 61A Great Suffolk Street, London, SE1 0BU
Information and support Line - Freephone 0800 652 6352
Tel: 020 7803 4800; Fax: 020 7401 3495 (open 9.00am-5.00pm Monday – Friday)
Web: www.musculardystrophyuk.org ; Email: info@musculardystrophyuk.org.

Patient.co.uk – is an online resource encompassing pretty much every health condition you can think of, including conditions, support groups, and lots more
Web: www.patient.co.uk

NHS Choices - www.nhs.uk

HealthUnlocked is a patient-friendly forum linked to NHS Choices - there is a CMT community - find it here - https://HealthUnlocked.com/cmtuk

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.
Pain Concern, Unit 1-3, 62-66 Newcraighall Road, Fort Kinnaird, Edinburgh, EH15 3HS
Tel: 0300 1230789 (10am – 4pm weekdays) Email: info@painconcern.org.uk.
Web: www.painconcern.org.uk

Pain Toolkit - a great website advocating self-help for people with persistent pain.
www.paintoolkit.org

Royal College of Anaesthetists – mainly a professional body, but with some excellent free information on having anaesthetic.
Tel: 020 7092 1500; Web: www.rcoa.ac.uk/patients-and-relatives ; Email: info@rcoa.ac.uk

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.
IOCP, 150 Lord Street, Southport, Merseyside, PR9 0NP
Tel: 01704 546141 Web: www.iocp.org.uk

British Orthopaedic Foot Ankle Society – has a section to help you find a specialist foot/ankle surgeon
Web: www.bofas.org.uk

Other support groups such as the MS Society, Spinal Muscular Atrophy UK, the Motor Neurone Support Group and others sometimes have great general information resources that are applicable to people with CMT. Don't read their medical information, as it'll confuse you, but do look at their other resources.

General Disability Information, rights, benefits and employment

Benefits and Work - a news and information company producing highly accurate and detailed guides to applying for various disability benefits. Individual membership costs £19.95 per year, but members of CMT UK can access their guides for free.
www.benefitsandwork.co.uk

Disability Rights UK – for information on benefits and your rights you can't get much better than Disability Rights UK . They also publish the 'Disability Rights Handbook' referred to as the 'bible' for disability issues.
Disability Rights UK, 12 City Forum, 250 City Road, London, EC1V 8AF
Tel: 020 7250 3222 (office number); Web: www.disabilityrightsuk.org;
Email: enquiries@disabilityrightsuk.org

Disability Rights UK run a number of Freephone helplines on a variety of topics:
Disabled Students Helpline: 0800 328 505
Equality Advisory Support Service (taken over from the Equality and Human Rights Commission Helpline): 0800 444205
Independent Living Advice Line (NOT benefits): 0300 5551525

Gov.uk – the website that brings together all the government services into one place.
Web: www.gov.uk

DIAL Network – an independent network of local disability information and advice services run by and for disabled people. Now run by Scope
Telephone: 01302 310123 to find your local branch
Web: www.scope.org.uk/dial

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.

Equality Advisory and Support Service – now taken over the helpline services from the Equality and Human Rights Commission
Tel: 0808 800 0082
Web: www.equalityadvisoryservice.com
Web: www.equalityhumanrights.com

Turn2us – helps people access the money available to them through benefits, grants and other financial help
Web: www.turn2us.org.uk

Daily Living and Mobility Issues etc

For all government related mobility enquiries, including how to contact the DVLA regarding your CMT, go to www.gov.uk

For the specific page to download the form for the DVLA – go here - https://www.gov.uk/peripheral-neuropathy-and-driving

Foundations – national coordinating body for home improvements agencies in England.
Tel: 01457 891909. Web: www.foundations.uk.com.
For Wales: Care and Repair Cymru. Tel: 029 2057 6286; www.careandrepair.org.uk
For Scotland: Care and Repair Forum (Scottish Homes). Tel: 0141 221 9879;
web: www.careandrepairscotland.co.uk
For Northern Ireland - Fold Housing Association. Tel: 02890 428314 Web: www.foldgroup.co.uk

AA – breakdown cover, insurance and more – gives priority to breakdowns for disabled people
Tel: 0800 262050; Web: www.theaa.com

Disabled Motoring UK – offers support and information on all issues to do with disabled driving.
DMUK, Ashwellthorpe, Norwich, NR16 1EX
Tel: 01508 489449; Web: www.disabledmotoring.org

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 9am-5pm Monday – Friday);
Web: www.mobility-centres.org.uk; Email: mobility@rcht.cornwall.nhs.uk

Motability – the government funded charity that can, if you are on the higher level of the Mobility Component of Personal Independence Payments or Disability Living Allowance, help you buy or loan a car or an electric wheelchair.
Tel: 0845 60 762 60; Web: www.motability.co.uk.

National Rail Enquiries - for information on travel, including help travelling.
Tel: 08457 484950 www.nationalrail.co.uk

RAC – breakdown cover, insurance and more – gives priority to breakdowns for disabled people
Tel: 0800 029029; Web: www.rac.co.uk

Rica - produce independent, impartial guides to pretty much every major purchase and gadget that you may require - www.rica.org.uk

Disabled Persons Railcard – for Disabled Person's Railcard giving 1/3 off rail travel. Also includes information on planning rail travel and who to contact for assistance.
Disabled Persons Railcard Office, PO Box 11631, Laurencekirk, AB30 9AA
Tel: 0845 605 0525 Email: disability@atoc.org
www.disabledpersons-railcard.co.uk

Shopmobility – find your local scheme through www.yell.com or your local directory.

Good online stores for daily living tools, etc

DO NOT spend potentially thousands of pounds on wheelchairs, stair lifts, bath lifts and the like without getting professional advice and assessment - your OT can help.

CMT United Kingdom doesn't recommend or endorse any of these companies.

But for smaller goods:

Nottingham Rehab Supplies provide a huge range of disability aids and mobility equipment. Our disability equipment is of the highest quality and caters for everyone. www.nrs-uk.co.uk

The Key Safe Company offers a range of key safe solutions that will ensure and reinforce your security priorities. https://keysafe.co.uk

Betterlife Healthcare - for a huge range of gadgets and equipment. Good prices for car-friendly scooters. www.betterlifehealthcare.com

Co-operative Independent Living - offers a wide range of mobility aids and daily living aids designed to make life a bit easier.
Tel: 0800 622 6001 Web: www.co-operativeindependentliving.co.uk

Disabled Living Foundation – is a national charity providing impartial advice, information and advice on independent living. They have a number of downloadable factsheets, a huge directory of suppliers of relevant equipment and lots more.
Web: www.dlf.org.uk; Email: advice@dlf.org.uk

Living made easy is a website full of clear practical advice on daily living equipment (includes a separate section for disabled children) – www.livingmadeeasy.org.uk

Telecare

Telecare is the term used for call assist-type services. Generally speaking, each local authority has its own preferred service, so contact your local social services for information. The Telecare Association can give you a place to start your research. www.telecare.org.uk

www.nhs.uk/planners/yourhealth/pages/telecare.aspx also gives impartial information.

Organisations for holidays, accommodation and leisure

Tourism For All UK – contains everything you want to know about accessible tourism.
TFA, 7A Pixel Mill, 44 Appleby Road, Kendal, Cumbria, LA9 6ES
Tel: 0845 124 9971 Email: info@tourismforall.org.uk
Web: www.tourismforall.org.uk

Family Holiday Association – gives money towards holidays and transport, but your social worker needs to apply on your behalf.
Family Holiday Association, 3 Gainsford Street, London, SE1 2NE
Tel: 020 3117 0650; Web: www.fhaonline.org.uk.

Family Fund Trust – gives grants to families raising disabled and seriously ill children aged 17 and under.
Family Fund, 4 Alpha Court, Monks Cross Drive, York, YO32 9WN
Tel: 08449 744 099 (Mon-Fri 9.00-5.00); Email: info@familyfund.org.uk
Web: www.familyfund.org.uk

Vitalise – respite for carers and holidays for people with severe disabilities at their centres in the UK and abroad.
Tel: 0845 345 1972 (open 9.00am-5.00pm Monday – Friday)
Web: www.vitalise.org.uk. Email: info@vitalise.org.uk

There are a vast number of disability leisure and sports organisations, far too many to list here. Google what you'd like to do, and it'll almost certainly point you in the right direction.

Travel Insurance

This can sometimes be a challenge for people with pre-existing conditions, but don't consider going abroad without adequate insurance.

Some good companies are below, CMT United Kingdom doesn't recommend or endorse any one in particular. It is your responsibility to disclose ALL your medical conditions.

www.insureandgo.com has been used by a number of CMTers in the past, and is cheap!

www.allcleartravel.co.uk operate a comparing services that can be useful, as does www.moneysupermarket.co.uk and www.medicaltravelcompared.co.uk

Organisations for individual personal issues

Genetic Alliance UK – lots of information about genetics, NHS genetics services and genetic ante-natal options including pre-implantation genetic diagnosis.
Genetic Alliance UK, Unit 4D, Leroy House, 436 Essex Road, London, N1 3QP
Tel: 0207 704 3141 Email: contactus@geneticalliance.org.uk
Web: www.geneticalliance.org.uk

Regard – information and support for gays and lesbians with a disability.
BM Regard, London, WC1N 3XX
Tel: 08444 431277 Email: secretary@regard.org.uk
Web: www.regard.org.uk

Relate – counselling for adult couples (whether married or not) who have having relationship difficulties. Tel: 0300 100 1234; Web: www.relate.org.uk

Organisations for Families and Carers

Carers UK – an organization giving support and information for people in a caring role
Carers UK, 20 Great Dover Street, London SE1 4LX
tel: 020 7378 4999 Freephone: 0808 808 7777
website: www.carersuk.org

Carers Wales
River House
Ynys Bridge Court
Cardiff CF15 9SS
t: 029 2081 1370

Carers Scotland
The Cottage
21 Pearce Street
Glasgow G51 3UT
t: 0141 445 3070

Carers Northern Ireland
58 Howard Street
Belfast BT1 6JP
t: 02890 439 843

Contact a Family – by bringing together families with children with disabilities they offer support, advice and shared experiences. Lots of information and advice on benefits, educational issues and much more.
Contact A Family, 209-211 City Road, London, EC1V 1JN
Tel: 0808 8083555 (open 9.30am to 5pm, weekdays) Email: Helpline@cafamily.org.uk
Web: www.cafamily.org.uk

Council for Disabled Children – is the umbrella body for the disabled children's sector in England, seeking to make a difference to the lives of disabled children with special education needs. Lots of resources and links to other organisations for parents.
CDC, 8 Wakley Street, London, EC1V 7QE www.councilfordisabledchildren.org.uk
Tel: 0207 843 1900 Email: cdc@ncb.orguk

Disabled Parents Network – support and information for parents who are also disabled.
DPN, Poynters House, Poynters Road, Dunstable, Bedfordshire, LU5 4TP
Tel: 0300 3300639; Email: information@disabledparentsnetwork.org.uk
Web: www.disabledparentsnetwork.org.uk

Disability, Pregnancy and Parenthood – the national information charity on disability and parenthood
Disability, Pregnancy and Parenthood, 336 Brixton Road, London, SW9 7AA
Tel: 0800 0184730; Email: info@dppi.org.uk
Web: www.dppi.org.uk

Fair Play for Children – making the world fit for children – mostly about child safeguarding
Fair Play for Children, 32 Longford Road, Bognor Regis, PO21 1AG
Tel: 0843 289 2638 Email: fpfc@fairplayforchildren.net
Web: www.fairplayforchildren.org

Scope – no longer just for people with Cerebral Palsy, there is a huge amount of information here for families with a disabled child
Scope, 6 Market Road, London, N7 9PW
Helpline: 0808 800 3333 Email: response@scope.org.uk
www.scope.co.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.

Chapter 6: Appendices

Appendix 1 - Models of adjustment to disability

Since World War Two, considerable attention has been paid to the psychosocial adaptation and adjustment to chronic illness and disability. Livneh (1984) proposed a five-stage linear model:

Stage 1: "Initial impact"—shock, anxiety.
Stage 2: "Defence mobilization"—bargaining, denial.
Stage 3: "Recognition"—mourning, depression, internalised anger.
Stage 4: "Retaliation/rebellion"—anger, aggression.
Stage 5: "Reintegration"—acknowledgment, acceptance, adjustment.

This model bears a striking similarity to Elizabeth Kübler-Ross' famous "five stages of grief" (1969): **Denial and Isolation; Anger; Bargaining; Depression; Acceptance.** However, psychologists nowadays recognise that a person's response to disease, death or any kind of loss does not follow a predictable, linear pattern. It is instead a highly idiosyncratic process. Someone with CMT will likely experience all of these "stages" at different times and with different intensities—sometimes all in the same day!

The process of adjusting to and coming to terms with progressive physical disability in particular—as opposed to sudden disability through an accident or amputation, for example—is an ongoing process of gradually mourning the loss of many things: mobility; functionality; certain previously-fulfilling activities; a former identity; able-bodied relationships with others, work, leisure, and the world; a "normal" lifestyle; an imagined non-disabled future. At each stage on the CMT journey, a process of adjustment takes place, and the sense of one's self and one's life shifts once again. There are positives as well as negatives.

In a later review, Livneh and Parker (2005) identified further models for adjustment to disability, culminating in the "chaos and complexity model," in which adaptation to disability is "nonlinear, unpredictable, and discontinuous . . . Such an approach recognizes the complexity, uncertainty, transformation, and ever-evolving dynamics of the human spirit, especially as it seeks to transcend the constraining barriers imposed by chronic illness and disability" (2005: 26).

In other words, your response to having CMT is yours and yours alone. No one has the right to tell you how you "should" be feeling about it. The life of each person with CMT is unique. The same applies to your response to discovering that a child or other close relative has the condition.

Whatever your feelings, there's no need to suffer in silence. Make sure you get whatever help and emotional support you can, whether from family, friends, colleagues or professionals such as doctors, physiotherapists, counsellors, online support groups and, of course, CMT UK.

Appendix 2 - Treatment development in CMT

Understanding of CMT is increasing rapidly every year, with more and more genes being identified that have an effect on our nerves. As of 2014 there are more than 80 causative genes identified for CMT. Studying these genes has allowed researchers to understand more about the way faulty genes cause a neuropathy. With this greater understanding comes an increasing hope that effective treatments for CMT may become available.

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. This is already important as there have been drug trials performed in CMT1A and to enter these trials patients needed a confirmed genetic diagnosis. It is anticipated this will apply to all future trials.

Challenges in developing therapies for CMT

One of the main challenges in developing therapies for CMT is the increasing number of causative genes identified.

As in many other northern European and US populations, CMT1A is the commonest form of CMT in the UK accounting for about 60% of people with CMT. CMTX1, caused by mutations in the gap junction protein beta-1 gene (GJB1, which encodes for the protein connexin 32) is the second commonest cause, accounting for about 10% of cases. The remaining genes affect the other 30% of the population and there are more genes continually being identified. This means that the other forms of CMT are rare with many affecting just a few families and in some cases just a single family. This poses challenges for therapy development, especially for the rare forms.

The second major challenge in developing treatments for CMT is that fortunately CMT (especially CMT1A) usually progresses very slowly disease and does not usually affect life expectancy. This means that any treatments developed have to be very safe, as serious side effects or long-term adverse effects would be unacceptable. As most forms of CMT start in childhood new treatments are likely to have to be started in children so would need not to interfere with their normal development.

Disease-modifying therapy development in CMT

Disease-modifying therapies refer to drug therapies or genetic therapies that are developed to specifically treat the roots of CMT in contrast to therapies like physiotherapy or orthopaedic therapies that treat the symptoms and complications of CMT. There are two main areas in which research to develop disease-modifying therapies are being directed.

Over the last two decades since the first causative gene for CMT was identified in 1991, the major research effort in CMT has being directed at understanding how genes in which there is a mutation cause CMT and specifically how they cause the neuropathy which characterizes CMT.

This research is largely done in models of the disease rather than in people themselves. It involves initially understanding the normal function of the protein (which is coded for by the gene) in normal nerves. This is often unknown when the gene is identified as causing CMT. Once the normal function of the protein is worked out, researchers try to identify how a mutated protein can cause nerves not to work properly and hence cause a neuropathy. This is often very difficult to work out.

Finally, once this is known, research is directed at trying to correct the defect in the mutant gene or protein to try to treat the neuropathy. All of these processes take considerable time.

Once a mechanism of treatment is identified then researchers need to develop drug or genetic therapies that can correct the defect and this is followed by extensive safety studies of the treatment to make sure it can be used in humans. For example in CMT1A, although the causative genetic defect (the chromosome 17 duplication) was identified in 1991, the first treatment trials of vitamin C were not started until 2006 meaning the steps above took 15 years.

The second area where research is being directed is to make sure that the patient population is trial-ready. There is no point in developing a therapy if the mechanisms are not in place to do a trial. Being trial-ready involves many processes including:

- studying the natural history (how a disease develops over time) of the different forms of CMT in order to understand what to measure to show a treatment works in a trial
- developing tools to measure the changes identified over time and making sure whatever tools are developed are sensitive enough to measure change in a short period e.g. over two years (the typical duration of a drug trial)
- training researchers in multiple centres to be able to use these tools
- training researchers to work with people who have CMT to make sure plans for drug trials are appropriate and acceptable to the participants

Specific approaches to therapy development in CMT

There are three broad approaches to developing therapies, all of which are currently being pursued.

Treatments that correct the underlying genetic defect.

> These kinds of treatments are being developed and trialled in many genetic conditions. The rationale behind these treatments is not specific to any genetic disorder but can be adapted to specific diseases: the current trials of anti-sense treatment for Duchenne's muscular dystrophy are an example.
>
> The aim of anti-sense treatment is to fool a person's cell into producing a normal or near normal protein by altering the way the defective DNA is decoded. There are many other genetic therapies like this in development.
>
> The challenge for CMT is three-fold. Firstly, we do not yet fully understand the risks of these type of treatments: some involve using viruses to deliver the genetic treatments, which have their own potential risks. In CMT which progresses very slowly, we would need to have absolutely safe therapies to justify using them. This is not to say that we should not be looking carefully at potential genetic therapies like these - and many groups in the world are - but safety is of paramount importance in a slowly progressive condition.
>
> The second - and major - challenge, is that these treatments usually need to be given to the tissues in the body where the defect is, which in CMT are the peripheral nerves. Our peripheral nerves are protected from the circulating blood by what is called the blood/nerve barrier. This means drugs which are either taken by mouth or by intravenous injection are present in high concentration in circulating blood but are not efficiently delivered into the peripheral nerves. Current research is trying to develop ways of penetrating the blood nerve/ barrier.
>
> The third challenge is that with over 80 causative genes identified for CMT to date, each gene would need to have its own gene-specific therapy developed and this may not be feasible.

The second way to develop therapies for CMT is where most progress has occurred to date. This is based on understanding the pathogenesis of CMT. What this means is that once a

causative gene is identified for CMT - e.g. the chromosome 17 duplication for CMT1A - research is done to understand how the genetic defect causes the disease.

With CMT1A, this research has shown that a major part of the problem is having too much PMP22 protein because of the extra copy of the PMP22 gene. Laboratory studies then showed that reducing the amount of the PMP22 protein helped laboratory models of CMT1A and hence compounds were identified that could reduce the amount of PMP22. One of these compounds was vitamin C (ascorbic acid).

There have now been two large international trials of people with CMT1A taking vitamin C for two years, (including the UK / Italian trial) both of which were negative i.e. there was no evidence that vitamin C slowed down the progression of CMT1A. This does not mean that reducing the amount of PMP22 is not the right approach - just that vitamin C may not be the best way to do so.

Currently other compounds that can reduce the amount of PMP22 are being explored at laboratory level. An exciting new development is the ability to do what is called high throughput screening of known drugs. This is a mechanism where if you identify the pathway by which the mutated gene causes CMT, you can then screen multiple drugs that are already in use for other conditions to see whether any of them can work on this pathway. This method is currently be used to screen for therapies for CMT1A .

The third approach to developing treatments for CMT is based on finding therapies that are not specific to a particular gene but that help damaged nerves repair.

This is rather like treating high blood pressure. Many people have high blood pressure. Doctors often do not know the exact reason in an individual person why the blood pressure is raised but they have very good drugs for reducing the blood pressure.

There are many causes of neuropathy as well as the genetic causes but there have been no drugs developed to date that can repair damaged nerves regardless of the cause. Nerve growth factors, which we know are needed for nerve development, have been trialled in diabetic neuropathy and also in a limited number of people with CMT. To date these are too toxic to use but research is ongoing in this area.

One of the most exciting developments from the identification of so many causative genes for CMT and the understanding of their pathogenesis is that we are beginning to discover common pathways in nerves involving many different genes. For example, the pathways involved in axonal transport - i.e. how nutrients and essential building blocks for nerves are transported down from the nerve bodies in the spinal

cord to the ends of the nerves and how the waste products from nerve endings are transported back to the cell bodies - are gradually being understood.

Many of the genes identified especially for CMT2 and HMN are involved in axonal transport and developing drugs to help axonal transport may help many different forms of CMT. Similarly, other pathways are being identified that involve more than one gene including the CMT1 genes. This pathway approach to developing therapies shows great potential.

Appendix 3 - Assistive technology and adaptive technology

Assistive technology is basically any item, product or system that can be used to help challenged folks operate in a better way; **adaptive technology** is often defined as the electronic or computer subset of assistive technology.

What follows is the beginnings of an ever expanding list of things to think about:

Computer hardware

Portable or not
Fixed desktops are prevalent in the workplace, but portable laptops have adequate processing power for most people and can obviously go where you need them. But, that comes with the challenge of carrying, holding and opening them.

Go into a shop to test the suitability of the machine, don't simply buy one on the Internet that you can't physically test. Laptops become lighter, which makes them easier to carry, but make sure that the clasp is manageable and for safety sake, purchase a carrying bag with a strap that you can put over your shoulder for example.

Seriously consider whether a full-blown laptop is required, tablet computers with touchscreens can provide virtually everything you need on a daily basis. They are price competitive, simple to use, easy to carry and come in a variety of sizes from the pocket to a sheet of paper. Think about how to charge it using inductive technology such as QI or specially adapted cables (see below) and get a protective cover for accidental knocks and drops.

Logging on
Difficulty in hitting start keys (e.g. control, alt, and delete simultaneously) can be eased by using fingerprint recognition logon available on many laptops.

Low-pressure keyboards
Laptop keyboards are typically low-pressure impact devices, which helps with fatigue on the hands whilst typing. Similar external keyboards are available for desktops and also as additional items for the laptop - this has the added advantage of being full-size as compared to the typical laptop and consequently easier to use with less errors.

Consider wireless peripherals (keyboards and mice) where the Bluetooth connector is small and can be left permanently plugged in.

Oversize touchpads
Large, external touchpads offer bigger buttons alongside a bigger area to move over. These can be typically programmed to do specific tasks as well

Cables
Make the USB cable bigger and more manageable by taping foam pipe insulation around the end.

Banks
Be wary of ATM machines, theft reduction devices on the card reader can make it very challenging to get the card out once it's in.

Computer software

Speech recognition
Software that will allow you to reduce the number of keystrokes is readily available now, ensure that you have a machine that meets the specification because the processing requirement can be high. Don't expect it to work from day one, but you'll find that it continuously learns your speech patterns and vocabulary and after a few weeks, it's very usable. That profile can become one of your most valuable assets so ensure that you back it up regularly.

It's also possible to use the speech recognition to click on buttons on applications and websites, albeit somewhat tiresome, but it does work!

Communication devices

Computer headsets
Decent headsets, including those with noise reduction if you're in a busy office help you get the best out of any speech recognition software that you use.

Mobile telephones
Mobile telephones have now become smart with touchscreens that don't require small key depressions. If you have trouble holding the device then fit it with a rubber sleeve for protection and grip and add a strap that you can even put around your neck or wrist.

Most operating systems now also provide voice recognition to save on typing, but don't expect them to be as accurate as a computer-based one with a headset.

Mobility

E-trikes
Mobility and distance travelled by pedal can be improved with three wheels and the inclusion of an assistive electric motor. Just remember that you will need to take the battery off to charge it and they can be weighty, some models allow on bike charging, but you have to be able to get the power to where they're parked.

E-scooters
Stable four-wheel scooters with lead acid batteries can give you up to 25 miles of travel, some batteries can be taken on board aircraft in the hold and travel free of charge, as would a wheelchair.

Cars
A Motability specialist dealer can advise you on the things to look out for on a vehicle, such as height of seeds, heaviness of steering, et cetera but also think about electronic gadgets like proximity switches so that you don't fumble about with keys. Consider using the strap of your lanyard with the e-key on to open the door by looping it round the handle.

Home automation

Door locks
Save yourself the effort of trying to hold and turn keys by fitting electronic door locks with proximity card readers or dongles. They are secure, means that the door is more likely to be locked then if you have to do it manually and with an easy way of keeping track of the numerous keys issued.

Light switches
Passive infrared detectors (PIR) can be fitted to save you fumbling for lights in the dark et cetera. They can also become cost-effective in hallways, because they will switch off after a given amount of time.

Alongside fitting normal fire detection devices. Also consider putting in emergency lighting, giving you a better chance of balance when walking in difficult conditions.

Alerts
Once simply limited to a pushbutton device around your neck, this whole area is expanding rapidly into fall detection, tied in smoke and heat detectors alerting a specialist team or nominated individuals.

If you are concerned about someone then you may want to consider some of the low-cost security monitoring options, including cameras across the Internet.
Kettles

Avoid the danger of lifting a kilo of boiling water by using single cup kettles where the vessel is put under a spout, hitting a button then boils the water and delivers it safely. It's also very energy efficient and only needs refilling periodically!

Heating system controls
Instead of trying to manually rotate valves on radiators, you can consider automated ones either on an individual basis based on time or a centralised one running off a computer system that can be controlled remotely. These are being marketed in the UK now by the big energy companies.

Window blinds
Once again, the ability to open and close window blinds can be automated and driven by remote control, either individually or from a central computer system.

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 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 (dominant) 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).
Chromosome	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
Contracture	A shortening of a muscle or tendon, which prevents 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. 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.
Fatigue	Physical or mental exhaustion due to exertion. Fatigue (tiredness) is a normal response to activity, but in a condition like CMT it can lead to someone needing more and more rest; even after very little activity.
Foot drop	Weakness in the shin muscle at the 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 and leading to a higher risk of tripping and sprains.
Genes	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.
Geneticist	Someone who studies genes.
Genotype	A person's genetic (**genes**) makeup.
Heel varus	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.
Heredity	Passing on genes and therefore certain traits, including a condition like CMT, from parents to children.
Hereditary neuropathy with liability to pressure palsies (HNPP)	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).
High arches	Also known as pes cavus or cavus foot, people with CMT often develop very high arches, which can be painful to walk on. This happens because the muscles and ligaments in the foot tighten (see **contracture**), pulling the ends of the foot closer.
Hindfoot varus	See **heel varus**
HNNP	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 fluid surrounding 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 nerve fibres (**axon**).

Nerve	Cordlike bundles of fibre that conduct 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 and efficiently messages are being carried by your nerves. They measure both the size and the speed of the message.
Nervous system	The system that controls all our brain and limb functions. It includes the central nervous system (brain and spinal cord) and the peripheral nervous system (nerves in the limbs that come from and go to the spinal cord).
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 cord into the arms and legs and those radiating back from the arms and legs into the spinal cord, 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 nerves in 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 usually become worse over time, often very slowly over years, if not decades, with little or no change from young adulthood to late adulthood. This can vary with different types of CMT.

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**

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Introducing CMT United Kingdom

CMT United Kingdom is the UK's national support group for people with Charcot-Marie-Tooth disease also known as Hereditary Motor and Sensory Neuropathy. The organisation was formed in 1986, and is now in contact with over 3000 individuals in the UK and around the world, and with several hundred interested professionals.

We provide support by means of a magazine, ComMenT, produced three times per year, plus an e-newsletter, sent out monthly; a comprehensive website with resources exclusively for members; a popular Facebook page; leaflets, resources and of course, this book; and of course by personal contact – either by phone, email or letter. We also host a Conference each year, and there are also a number of local groups around the UK, who can provide that bit of extra personal support.

We have a small office in Bournemouth, Dorset and the organisation is run by the Board of Trustees, aided by three staff.

We charge a small subscription for our services and receive no statutory funding from the UK government and are entirely reliant on fundraising and donations from our wonderful members and supporters.

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