

Muscular Dystrophy Ireland

Charcot-Marie-Tooth Information Day Saturday 27th September 2008

Muscular Dystrophy Ireland's first ever information day on Charcot-Marie-Tooth, also known as the Hereditary Motor and Sensory Neuropathies, was held on Saturday 27th September 2008. It was an exceptionally well attended day and we were pleased to have a range of high profile speakers with expertise in this condition. The following is a report on the day.

CMT UK

Karin Rogers is Vice Chairperson of CMT UK, the support organisation in the UK for people with CMT. While based in the UK, this organisation is open to anyone, and Irish people are welcome to join and receive information from them. There are currently around 30 Irish members of CMT UK. Members receive a newsletter every three months – CoMmenT – and are invited to attend their annual conference. Among other activities, there is an annual young people's weekend for 11-18 year olds, and Karin is involved in organising this. If anyone would like to receive information from CMT UK or is interested in joining, their website is www.cmt.org.uk

What is CMT / HMSN?

Dr. Mary Reilly, Consultant Neurologist from the Institute of Neurology and Neurosurgery, London, gave an overview of CMT. She explained that the term CMT is a collective term for a range of conditions. HMSN – the hereditary motor and sensory neuropathies, is another name to describe the range of conditions. This heterogeneity can make it difficult to come up with a definitive diagnosis, and to research potential new treatments.



CMT is a relatively common condition. It is estimated to affect around 1 in every 2500 people, but in reality it is probably more than this. In Northern Europe, around 90% of people with CMT have an autosomal dominant or x-linked form.

There are around 50 different genetic mutations that can lead to a diagnosis of CMT. CMT type 1a is the most common, but CMT type 2 may be more common than originally thought.

CMT type 1 is a demyelinating neuropathy, meaning that the myelin sheath is affected – the part that insulates and nourishes the nerve's axon. Type 2 is an axonal neuropathy, meaning that the axon is directly affected.

To identify the type of CMT a person has, electrical studies, family history and clinical observation are used to indicate what type of CMT and therefore what gene or set of genes to look for.

In the most common form of CMT type 1a, the abnormal gene is found on chromosome 17, and the protein which is affected is called PMP-22 (peripheral myelin protein 22). Affected people have an extra copy of the gene – 3 copies instead of 2. Usually this is inherited in an autosomal dominant way, meaning that it came from one affected parent.

The second most common form of CMT type 1 is CMT 1X, where there is a mutation of the connexin 32 gene on the X chromosome, one of the chromosomes that determines the sex of a child (females are XX, males are XY). In this type, males inherit the condition from their mothers, who are carriers.

In certain cases of CMT types 1 and 2, the inheritance is autosomal recessive, meaning that a person has to inherit two abnormal copies of the gene in order to have the condition. Many people with a recessive form do not have affected relatives as each child only has a 1 in 4 chance of being affected and families now tend to be smaller. Rarely, CMT can arise due to a new mutation, where neither of their parents had the condition or carried it.

Dr. Reilly's team continues to work on identifying the genes involved in the rare forms of CMT and to try to understand their pathogenesis (the step by step development and chain of events that led up to a condition).

Physiotherapy

Dr. Gita Ramdharry from St. George's, London, spoke about physiotherapy for people with CMT.



In CMT, the calf muscles are weakened and this affects balance when standing. Walking can also be more tiring because of the weakened ankle muscles. The push from the calf muscle does not come, so the thigh lifts up higher to compensate, and this leads to a high stepping gait and a possible toe strike. Joint pain can also affect walking.

The goal of the physiotherapist is to assess the current problems; prevent or minimise specific symptoms; optimise function; promote self management and provide advice and ongoing assessment.

Exercise is important to increase strength and endurance; control weight; increase the ability to do day to day activities; reduce fatigue and improve mood. The types of exercise include fitness training, stretching, muscle strengthening and balance training.

Fitness training, for example swimming, is good at improving overall fitness levels, leading to a reduced effort in day to day tasks and reduced fatigue. The general advice would be to exercise at a low to moderate activity (about 5-6 out of 10 for effort) and **DO NOT EXERCISE TO EXHAUSTION**. Pace yourself. Muscle soreness should not last beyond 48 hours. You also have to consider the impact of balance and joint pain.

Stretching is important to reduce muscle shortening, prevent or reduce deformity and reduce the effort by muscles working in normal ranges. You can stretch actively yourself, by using orthotics or soft tissue massage. Advice for stretching would be to identify the muscles which need stretching; stretch daily, holding for 30 seconds and take care if you have reduced feeling or changes to joint position. Stretching should not be painful.

There are also benefits to muscle strengthening, such as reversing disuse weakness, reducing effort and fatigue, and increasing activation and bulk. If you don't use it, you lose it. However, you should seek advice regarding which muscles to strengthen and in which position. You should also be cautious with very weak muscles. If you are using weights, use only a low to moderate effort (5-6 out of 10) and increase repetitions rather than weight as you progress.

Balance training is good at increasing stability when standing and walking. It is important to challenge your balance safely however, either with a physiotherapist, someone else or something to support you. Other ways to assist balance include the use of orthotics and walking aids.

If in doubt, ask a physiotherapist.

Surgery

Mr. Michael Stephens, Consultant Orthopaedic Surgeon in Cappagh and the Central Remedial Clinic uses the term HMSN (hereditary motor and sensory neuropathy) as opposed to CMT. He was involved in producing an article called "Foot Deformities in Children with HMSN", by Wines, Chen, Lynch and Stephens in 2005.



Approximately 65% of people with HMSN have HMSN type 1 and of these, around 75% have HMSN type 1a. It is thought that there could be some people diagnosed with cerebral palsy who actually have HMSN type 5.

In his research, 52 children with HMSN were studied to check for deformity of the feet. It was found that in type 1, it is more common to have a high arch. In type 2 however, a flat foot is more common. In other types then, the high arch is more common again.

The first method of management is to treat with physiotherapy and orthotics. These must work in tandem. There is no point using orthotics without a stretching programme. When conservative treatment no longer works, then surgery is considered.

A foot deformity can be fixed or flexible. A flexible deformity is potentially correctible. With a fixed deformity, it cannot be corrected, but surgery can be used to accommodate it and provide more comfort.

With operative treatment, there is no rigid set of guidelines. Options that preserve motion are preferred over fusions. The surgeon must also look at all problems as a package. For example, they would not operate on only one area (e.g. the toe) without looking at other areas (e.g. shortening the outer bone of the foot) in order to bring the foot in line.

Bone alignment would be corrected first and then soft tissue can be addressed, e.g. tendon release, elongation / recession or transfer. Sometimes the Achilles tendon has to be addressed but lengthening the tendon can weaken it. The problem with the tendon can be primarily due to the high arch and so could be addressed when the high arch is dealt with.

It is better to change the foot when it is flexible, as deformities become more rigid with time and surgery is then more complicated.

Orthotics

Ms. Donna Fisher from Cappagh Hospital and the CRC spoke about the use of orthotics.



An orthosis is an external aid which provides support, corrects mobile deformities and maintains functional position.

There is a multidisciplinary approach in the CRC, with assessments by the neurologist, physiotherapist, orthopaedic consultant and orthotist. The orthotic assessment would look at the level of mobility, risk of damage to joints, pain

and lack of stability. What they prescribe then, will depend on the level of the joint affected (foot / ankle / knee / hip / spine), the number of joints affected, the severity of the problem and the needs of the person with HMSN / CMT.

Foot orthotics include OTS insoles, functional foot orthoses and custom made moulded insoles. People can now buy insoles and orthotics from a range of places, but going to an orthotist will ensure that you get the best one for you. Every foot is different, so the orthotist would first take a mould of the foot to ensure a good fit. The orthotist also has to think about footwear, as insoles need to be able to fit into shoes. Special footwear would be a last resort, as they are generally not fashionable and so people are reluctant to wear them. An adaptation can be done to the outside of the shoe, but this is not what everyone wants.

If insoles or footwear cannot correct the problem, ankle – foot orthotics (AFOs) can be used. These can be rigid, flexible or jointed, depending on the individual case.

Silicone AFOs are available but they are not used a lot in the CRC. They are very expensive, especially for children who will grow out of them. There is also not a lot of evidence to show that they work any better. However, they may work for a select group of adults.



Dr. Gita Ramdharry answers a question, watched by Donna Fisher, Mr. Michael Stephens and Karen Pickering (MDI Information Officer / Chair for the day)

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