Muscular Dystrophy New Zealand

Myotonia Congentia (Becker & Thomsen Disease)

What is Myotonia Congenita Myotonia congenita is a disorder that affects muscles used for movement (skeletal muscles) and is characterised by muscles failing to relax normally after contracting which can be severe enough to affect normal daily activities. Failure to relax causes periods of tense muscles which is described as myotonia.

It is estimated to affect 1 in 100,000 people worldwide. This condition is more common in northern Scandinavia, where it occurs in approximately 1 in 10,000 people.

Myotonia congenita symptoms typically begin to be experienced in childhood. Although myotonia can affect any skeletal muscles, including muscles of the face and tongue, it occurs most often in the legs. Myotonia causes muscle stiffness that can interfere with movement. In some people the stiffness is very mild, while in other cases it may be severe enough to interfere with walking, running, and other activities of daily life.It is not generally accompanied by pain. These muscle problems are particularly noticeable during movement following a period of rest. Difficulties can be found when making sudden movements which can result in a fall.

Many affected individuals find that repeated movements can temporarily alleviate their muscle stiffness, a phenomenon known as the warm-up effect. Muscle enlargement may also be experienced as part of the condition. This often occurs and affects the calves, thighs, shoulders and forearms, sometimes in quite a pronounced way.

Skeletal Muscles are the muscles used for movement. It is one of three major muscle types, the others being cardiac muscle and smooth muscle. Most skeletal muscles are attached to bones by bundles of collagen fibers known as tendons.

Varieties of Myotonia congenita

The two major types of myotonia congenita are both genetically inherited conditions and known as Thomsen disease and Becker disease. These conditions are distinguished by the severity of their symptoms and their patterns of inheritance. Becker disease usually appears later in childhood than Thomsen disease and causes more severe muscle stiffness, particularly in males. People with Becker disease often experience temporary attacks of muscle weakness, particularly in the arms and hands, brought on by movement after periods of rest. They may also develop mild,

permanent muscle weakness over time. Becker disease is inherited in an autosomal recessive manner – see the following information box.

Autosomal Recessive Inheritance
An autosome is any of the chromosomes that is not a sex (X or Y) chromosome. When a disease or genetic trait is recessively inherited two copies of the gene are required (i.e one copy from each parent) for the disease or genetic trait to be expressed. In each pregnancy there is a 25% chance that the child will have
the condition.

Thomsen Disease can be noticed in early childhood, often in infancy when the baby is seen to keep their eyes closed for longer then expected when crying. This myotonia is experienced as mild and does not progress. The muscle weakness seen in people with Becker disease is not present. Thomsen Disease is inherited in an autosomal dominant manner – see the following information box.

Autosomal Dominant Inheritance
An autosome is any of the chromosomes that is not a sex (X or Y) chromosome. When a disease or genetic trait is dominantly inherited only one copy of the gene (i.e one copy from one parent) is required for that disease or genetic trait to be expressed. In each pregnancy there is a 50% chance that the child will be affected.

Causes of Myotonia congenita

Mutations in the CLCN1 gene cause both types of myotonia congenita.

The CLCN1 gene provides instructions for making a protein that is critical for the normal function of skeletal muscle cells. For the body to move normally, skeletal muscles must tense (contract) and relax in a coordinated way. Muscle contraction and relaxation are controlled by the flow of charged atoms (ions) into and out of muscle cells.

Specifically, the protein produced from the CLCN1 gene forms a channel that controls the flow of negatively charged chlorine atoms (chloride ions) into these cells. The main function of this channel is to stabilize the cells' electrical charge, which prevents muscles from contracting abnormally.

Mutations in the CLCN1 gene alter the usual structure or function of chloride channels. The altered channels cannot properly manage the ions flow, and a reduction in the amount of chloride ions being able to enter the muscle cell triggers the prolonged muscle contractions, and this causes the myotonia.

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Diagnosis of Myotonia congenita

Myotonia congenita is diagnosed clinically by the presence of episodes of myotonia beginning in early childhood, alleviation of stiffness by brief exercise, myotonic contraction elicited by percussion of muscles, electromyography revealing myotonic bursts, elevated serum creatine kinase concentration, and family history consistent with autosomal dominant or autosomal recessive inheritance.

Clinical diagnosis can be confirmed by testing for mutations in the CLCN1 gene and is detected in approximately 95% of cases.

Management of Myotonia congenita Muscle stiffness may respond to sodium channel blockers such as mexiletine (currently the medication with best documented effect), carbamazepine, or phenytoin. Beneficial effects have also been reported with quinine, dantrolene, and acetazolamide. Although many individuals manage their condition with repeated movements and avoiding triggers like the cold, fatigue and emotional stress as much as possible.

Agents/circumstances to avoid: in general, anesthesia should be used with caution. Depolarizing muscle relaxants (e.g., suxamethonium), adrenaline, beta-adrenergic agonists, propranolol, and colchicine may aggravate myotonia.

Genetic counseling is available to families who have had a diagnosis of myotonia congenita. This service provides information, helps families understand inheritance patterns and what this means in their family. They can also explain reproductive options available enabling people to make more informed family-planning decisions. Because individuals with myotonia congenita may be at increased risk for adverse anesthesia-related events, testing of atrisk individuals during childhood to clarify their genetic status is appropriate.

Support

The MDA Fieldworkers are available for support. They have in-depth knowledge of a range of neuromuscular conditions, and will have a better understanding of your needs and challenges. Have a chat over the phone or they can come to you for a kanohi ki te kanohi/face-to-face visit. They may have some real practical suggestions that have worked for others to offer as well. This service is offered free of charge to MDA members and is funded through donations and grants. Contact your local MDA Branch to be put in contact with your fieldworker.

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The MDA Support Network allows people with similar circumstances or challenges to come together to share their experiences and provide each other with emotional and moral support in addition to practical advice and information. By bringing together people with common experiences, support networks can provide an invaluable addition to medical care. The MDA of New Zealand Support Network currently has over 700 members throughout New Zealand who want to be in touch with others livings with neuromuscular conditions. Please see the MDA website www.mda.org.nz for contact details and more information that you might find relevant for you and your whanau.

Education

There is no reason why individuals with Myotonia congenita should be disadvantaged in terms of receiving full education. For more information, request the Education Pack available from the MDA.

Employment

Disability should not hinder employment possibilities. Any individual has the right to equal pay and equal rights for employment. For more information contact the Employment Relations infoline on 0800 800 863 or visit www.ers.dol.govt.nz.

The government promotes equal employment opportunities in private sector and can be contacted on (09) 525 3023 or visit www.eeotrust.org.nz . Workbridge provides a professional employment service for people with all types of disabilities and administers support funding on behalf of Work and Income. Contact on 0508 858 858 or visit www.workbridge.co.nz.

More Information: Muscular Dystrophy Association can be contacted for further information, assistance, advice, support and referrals, on 0800 800 337 or by e-mail at info@mda.org.nz.

Sourced from

http://ghr.nlm.nih.gov/condition/myotonia-congenita
http://www.musculardystrophyuk.org/app/uploads/2015/02/myotonias.pdf
https://en.wikipedia.org/wiki/Skeletal_muscle
http://www.ncbi.nlm.nih.gov/books/NBK1355/