

Myotonic Disorders (non-dystrophic)

Myotonia Congenita (Thomsen's disease, Becker's Generalized Myotonia), Paramyotonia Congenita, Schwartz Jampel Syndrome

What is myotonia?

Myotonia is not a disease. It is a symptom of a number of different muscle disorders, and it affects a muscle's ability to relax after it has been used. For most people, a myotonic attack is a temporary, disabling stiffness in their muscle that occurs after some kind of voluntary movement. Not being able to relax a tight grip, and a difficulty getting up from a seated position, are two common forms of a myotonic attack.

What causes a myotonic disorder?

Besides Schwartz Jampel Syndrome – the origin of which is not known – myotonic disorders are usually caused by errors in the gene that helps to determine the structure of the body's skeletal muscle cells. In muscle cells, there are structures known as ion channels that allow the muscle to contract and then to relax after a contraction. When a muscle is affected by a myotonic disorder, these ion channels do not function properly. This dysfunction affects the ability of a muscle to relax after it has contracted. Until relaxation can properly occur, the muscles remain stiff and unusable, causing myotonic stiffness.

The length of time that a myotonic attack will last can range from seconds to minutes, and it can be anything from slightly uncomfortable to completely disabling. It usually occurs after a strenuous activity or an extended period of rest, but things like cold temperatures or loud, surprising noises can also trigger it.

The only areas of the body that are affected by myotonia are the skeletal, or voluntary muscles. This means that myotonia will only occur after conscious physical movements, but will not affect the heart or muscles in the digestive tract. Depending on the type and seriousness of the disorder, myotonia can affect anything from the legs, face, hands, hips, shoulders, feet, eyelids, to a person's ability to speak clearly.

Who can be affected by a myotonic disorder?

Myotonic disorders are genetic, so they can affect anyone. Both sexes are affected equally and anyone at any age can exhibit myotonic symptoms. Myotonic disorders are usually congenital, meaning that they begin to affect people in early childhood. Depending on the severity of the myotonic symptoms, however, the symptoms of one of these disorders can first be noticed as late as the teenage years or adulthood.

How are the myotonic disorders inherited?

There are two methods by which a myotonic disorder can be inherited: by an autosomal dominant inheritance pattern, or by an autosomal recessive inheritance pattern.

Autosomal Dominant Inheritance

Disorders that follow an autosomal dominant inheritance pattern usually appear in every generation, without skips. Autosomal refers to the fact that the genetic error can occur on any one of the chromosomes in each cell in the human body, except the two sex chromosomes. Dominant refers to the fact that it is necessary for only one parent to pass on the abnormal gene for the disorder. Thus, the disorder can be inherited from either parent, and each child of an affected parent has a 50% chance of inheriting the condition. The severity of the disorder and age of onset can vary from person to person. For more information about genetics and inheritance patterns, see the Muscular Dystrophy Canada information sheet titled *What is Genetics?*

Autosomal Recessive Inheritance

In autosomal recessive inheritance, the disorder usually appears in only one generation and there is typically no previous family history of the disorder. Both parents must be carriers of the faulty gene. A carrier has the faulty gene in his/her body, but usually shows no symptoms. Recessive refers to the fact that the child must inherit the altered gene from both parents in order to have the disease. Children of either sex can be affected. With each pregnancy there is a 25% chance the child will receive a faulty gene from both parents and, thus, will be affected with the disorder. If the child inherits a faulty gene from one parent, he/she will be a carrier of the disorder, but will probably show no symptoms. There is a 50% chance the child will be a carrier. There is a 25% chance that the child will not inherit the faulty gene from either parents, so he/she will not be affected or a carrier. For more information about genetics and inheritance patterns, see the MDC information sheet titled *What is Genetics?*

What are some of the myotonic disorders?

Myotonia Congenita:

Myotonia congenita, the most common non-progressive myotonic disorder and, is caused by a mutation in the muscle cell's chloride ion channels. It has no effect on a person's lifespan, and it has little effect on things like body structure or growth patterns. There are two forms of myotonia congenital depending on the type of inheritance.

The more common, and often more severe form of myotonia congenita is called Becker's generalized myotonia, and it is transmitted by an autosomal recessive inheritance pattern. Its onset is usually some time in late childhood or early adolescence but, depending on its severity, it may show up during early childhood. The symptoms can often become more severe for a few years after diagnosis, and will eventually become stable by a person's mid-to late twenties.

The autosomal dominant form is commonly called Thomsen's disease, after the Danish physician Dr. Asmus Julius Thomsen, who had the disease himself and documented its existence through his own family history. The symptoms of Thomsen's disease are generally less severe than those of Becker's, while the age of onset is usually sooner, with

symptoms becoming noticeable during early childhood and sometimes at birth. In some extremely mild cases, however, the symptoms can go unnoticed for many years before an official diagnosis is made.

The main symptom of both disorders is a general myotonia that is caused by voluntary movements. It is usually triggered by strenuous activities or it will follow a period of rest or prolonged muscle relaxation. The myotonia is often most severe in the legs, causing difficulty in walking, as well as occasional falling. The arms, shoulders, and head are also often affected, leading to difficulties in chewing, grasping objects tightly, and blinking. In very rare cases, those with the Becker form of myotonia congenita will also experience a disabling weakness following their attacks of myotonic stiffness. This weakness can last anywhere from a few seconds to a few hours, depending on the seriousness of the myotonic attack.

After an attack, both types of myotonia can eventually be overcome by continued use of the affected muscle. The stiffening will generally become worse for the first few contractions of the affected area, and then, after about five repetitions, the myotonic stiffness will cease, allowing the muscles to work normally until another prolonged period of rest. This phenomenon is known as “warm-up” and it gives those with myotonia the opportunity to be involved in some strenuous activities and sports, especially ones that rely on strength of movements rather than speed.

Although myotonia congenita does not severely affect the physical makeup of a person’s body, it does affect the size of certain muscles. Both Becker’s and Thomsen’s myotonia congenita can cause an unusual increase in the size of some skeletal muscles, especially in the areas of the legs and buttocks, but also in the arm, shoulder, and back muscles. This increase is usually referred to as muscle hypertrophy, and it will often give a person an athletic appearance in the affected muscles. Those with Becker’s myotonia are usually more severely hypertrophied than those with Thomsen’s.

Paramyotonia Congenita:

Paramyotonia congenita is a rare disorder of the sodium ion channel that is transmitted through an autosomal dominant inheritance pattern. It is not life threatening, and its severity usually remains stable throughout a person’s lifetime. The onset of this disorder generally occurs between birth and early childhood.

The primary symptom of paramyotonia congenita is a generalized myotonic stiffness that most seriously affects the hands and face, but also areas like the neck and arms. As in the other non-progressive myotonias, paramyotonia congenita is triggered by strenuous voluntary muscle

What about research?

There has been a great deal of progress in myotonia research during the past ten years. Much of the research has been focused on isolating the specific gene and ion channel defects that cause myotonic disorders, especially in the case of myotonia and paramyotonia congenita. Eventually, these findings should lead to more better ways of diagnosing the disorders, as well as research into drug treatments that do not cause such serious side effects as the existing ones.

activity, but it is also made worse by, and in some cases may only occur upon, exposure to cold temperatures. In most cases of cold induced myotonia, stiffness can be relieved upon exposure to warm temperatures.

Attacks of myotonic stiffness are also often accompanied by a disabling weakness in the affected area. This weakness may last longer than the myotonic stiffness itself, disabling the muscles for a few minutes to a few hours, with the weakness lasting longer for more severe myotonic attacks. Weakness, however, is not common to all cases of paramyotonia congenita. If there are no signs of it by adolescence, it is likely that there will be none at all. Unlike the other non-progressive myotonic disorders, paramyotonia congenita has no 'warm-up' affect in which myotonic stiffness is reduced by further muscle contraction. Instead, myotonic stiffness actually increases with continued movement of the affected area, disabling a person even further. This phenomenon is often referred to as paradoxical myotonia.

Schwartz Jampel Syndrome:

Schwartz Jampel syndrome is the most severe of the non-progressive myotonias. It is very rare and is transmitted through an autosomal recessive inheritance pattern. The age of onset is usually birth or shortly afterwards. The type and severity of symptoms are varied from case to case. One of major symptoms is a myotonic stiffness that is most pronounced in the face and thighs. There is a possibility that this myotonia will cause a tendency towards falling, speech difficulties, and a number of facial abnormalities. Like myotonia congenita, continued muscle use will often lead to a relief of myotonic stiffness, or a 'warm-up' effect. This 'warm-up', however, is only present to a minor degree and is in some cases completely absent.

Multiple skeletal deformities are also a prominent feature of Schwartz Jampel syndrome. These deformities often lead to some major growth problems that eventually lead to a shortened stature, as well as severe facial abnormalities, giving the face a distinct 'mask-like' look to it.

Other symptoms include hypertrophied (enlarged) thighs, atrophied (underdeveloped) shoulder and hip, severe continuous muscle movements (twitching), and, in some cases, mental retardation.

The origins of Schwartz Jampel syndrome are unknown. There is presently some question as to whether it is a disorder of the muscles like paramyotonia congenita and myotonia congenita; whether it is a disorder of the nervous system; or whether it is a combination of both.

How is a myotonic disorder diagnosed?

A doctor, outside of the laboratory, can do many of the diagnostic tests for myotonic disorders. One way is testing for myotonic reactions to strenuous movement, cold temperatures, percussion, or other stimuli. It is important, however, that these disorders are differentiated from other, more severe progressive disorders like myotonic dystrophy. To do this, doctors will often need a number of laboratory tests that will give more concrete results.

These will often include: an electromyogram (EMG) test to measure the electrical activity in the muscle; a blood and DNA analysis to look for a genetic mutation; or even a muscle biopsy, which will not provide a final diagnosis of a myotonic disorder, but is a good way of testing for many types of progressive muscle problems.

Is there any cure or treatment for a myotonic disorder?

There are no cures for any of the myotonic disorders, but most people learn to live comfortably with their symptoms without any kind of treatment. If myotonia becomes more severe, however, there are a number of drugs that have proved to be effective in relieving myotonic symptoms. The most widely used of these is mexiletine, but others such as quinine, procainamide, tegretol, and phenytoin have also been used with some success.

As with all drug treatments, the major drawback is always the side effects, which can be quite serious in themselves. Most people affected by myotonic stiffness usually find that experience is the best remedy. Once a person knows what types of situations will bring about myotonic attacks, then they will be more prepared and be better able to avoid them in the future.

How can I help?

Muscular Dystrophy Canada conducts year-round fund raising campaigns to support our diverse programs. Your gift will help the Association provide the dollars necessary to assist individuals living with neuromuscular disorders, and fund much needed medical research and educational information. Please make a gift through our National office or any Regional or Community Muscular Dystrophy Canada office.

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