Sodium channel myotonia (paramyotonia congenita): information for families

Myotonia is muscle stiffness that develops when the muscles do not relax after being squeezed. In myotonia, this stiffness may wear off after the muscles are exercised or 'warmed up'. However, if paramyotonia congenital, muscle stiffness is brought on by exercise. This is the opposite of the ‘warm up’ effect so is called ‘paradoxical’ or ‘paramyotonia’. This information sheet from Great Ormond Street Hospital (GOSH) explains about the symptoms and causes of paramyotonia congenital (also known as sodium channel myotonia) and how it can be managed.

Paramyotonia is more temperature-sensitive than classic myotonia. Muscle stiffness can be triggered by cold temperatures but also muscle weakness.

Muscle stiffness can last for many seconds, for instance, when getting up out of a chair or holding onto a doorknob. In classic myotonia, the muscles usually start to relax after 'warming up' but get stiff again if you stop moving. In paramyotonia, the stiffness gets worse as you carry on moving.

What causes paramyotonia?

Paramyotonia is a genetic condition, caused by a mutation (change) on a specific gene. Research has identified the affected gene as the \textit{SCN4A} gene. This is involved in making a protein that controls movement of sodium into the muscle cells.

The gene mutation can be passed on from parent to child but in many cases develops sporadically (out of the blue). If it is inherited, it is passed on in an autosomal dominant manner – this means that if one parent is affected half of their children will inherit the condition.

Paramyotonia congenital is a rare condition – around 1 in 100,000 people worldwide are thought to have the disorder.
What are the symptoms of paramyotonia?

The severity of symptoms can vary widely from person to person. The most common symptoms is being unable to relax the muscles after they have been squeezed, especially in a static position such as sitting, lying or standing still.

The symptoms of paramyotonia include stiffness, cramp or locking of muscles. Muscle stiffness may be a bit worse in cold temperatures or after periods of rest and inactivity. The muscles in the legs, feet and hands may feel like you have cramp and may be painful. However, there doesn’t seem to always be link between muscle pain and the degree of stiffness.

The muscles in any part of the body can be affected. Commonly the hands, arms, legs, tummy muscles, back, diaphragm and neck muscles can be affected. The muscles in the face, throat, eyes and even the tongue can also stiffen. When the tongue is affected, it becomes stiff which can make speaking difficult so the person may sound drunk. Sometimes eating cold food, such as ice cream, can trigger tongue stiffness.

Muscle stiffness can also be triggered by loud noises, illness, stress, some medications or foods.

How is paramyotonia diagnosed?

The doctor will take a careful description of what symptoms occur and when, along with a physical examination. A gene test from a blood sample is needed to make a definite diagnosis.

How can paramyotonia be treated?

The aim of treatment is to improve the muscle stiffness. A medication called mexiletine is the current treatment. Research showed that it improves stiffness and doesn’t cause too many side effects.

The most common side effect reported is heartburn or indigestion. Taking another medication to reduce the amount of acid produced by the stomach helps to make this manageable.

We can organise a prescription from our Pharmacy department if your local paediatrician (specialist children’s doctor) or family doctor (GP) cannot prescribe it.

Other forms of treatment are available – we will discuss these with you in your clinic appointment.

What is the outlook for children with paramyotonia?

Episodes of stiffness can usually be reduced in severity and frequency by taking medication. The vast majority of children and young people grow up living a near-normal lifestyle.

Further information and support

Please contact the Clinical Nurse Specialist in the Dubowitz Neuromuscular Centre at GOSH. Call 020 7405 9200 ext 1195 or email nmchan@gosh.nhs.uk.

Muscular Dystrophy UK is the main organisation offering support and advice to anyone affected by a neuromuscular disorder. Call their helpline on 0800 652 6352 or visit their website at www.musculardystrophyuk.org