Chloride channel myotonia: information for families

Myotonia happens when there is delayed relaxation after muscle contraction through activity or exercise. This can show up as stiffness, cramp or locking of a variety of muscles. This information sheet from Great Ormond Street Hospital (GOSH) explains about chloride channel myotonia (also known as myotonia congenita), what causes it and how it can be treated.

The muscles affected by chloride channel myotonia include those in the hands, arms, legs, abdomen, back, diaphragm, neck, face, throat, eyes and even the tongue.

There are two forms of chloride channel myotonia: Becker disease and Thomsen disease – they can vary from each other in terms of severity and age of onset.

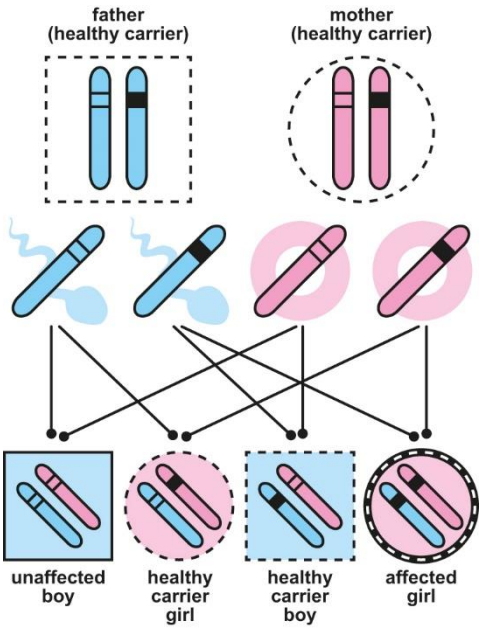
What causes chloride channel myotonia?

Every cell in the body has a thin membrane wall that lets a mixture of water, sodium and chloride in and out through 'channels' that act a bit like gates. How these channels work is controlled by our genes.

Chloride channel myotonia is a genetic disease, caused by a fault on the CLCN1 gene, which produces a protein that controls the amount of chloride flowing in and out of the muscle cells.

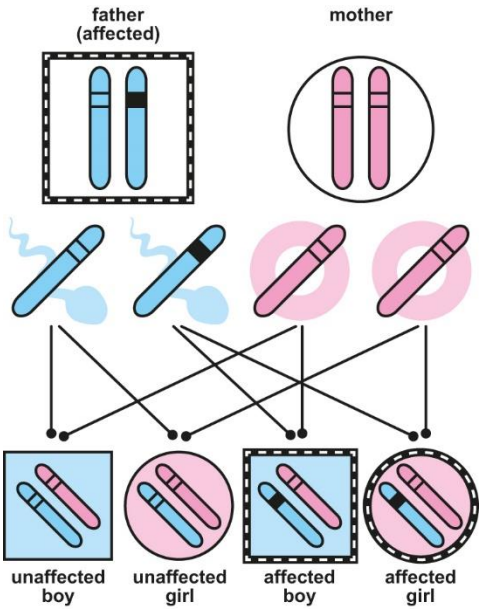
The faulty gene can be passed on from parent to child. The two forms of chloride channel myotonia are passed on in different ways.

Becker disease is passed on when both parents carry a faulty gene – this is called autosomal recessive inheritance. There is a 25 per cent (1 in 4) chance of a child having the condition, a 25 per cent chance they will be completely unaffected and a 50 per cent (1 in 2) chance that a child inherits one faulty gene and one normal gene so is a 'carrier' of the condition. The chances are the same with each pregnancy.



In **Thomsen disease**, if one parent has the faulty gene, there is a 50 per cent (1 in 2) chance that it will be passed on to their child causing the condition and a 50 per cent chance that it will not.

This is called autosomal dominant inheritance. This chance is the same for every pregnancy.



In some cases, the faulty gene can occur sporadically (out of the blue) in a child so is not inherited from either parent.

Who is affected by chloride channel myotonia?

Becker disease usually affects older children and can be more severe than Thomsen disease, especially in boys. Both conditions are estimated to affect 1 in 100,000 people worldwide but is more common in northern Scandinavians.

What are the symptoms of chloride channel myotonia?

While the severity of stiffness can vary widely from person to person, the most common and obvious symptom is the inability to relax muscles after contraction, especially after being in a static position such as sitting, lying or standing still.

The precise effects of the stiffness will vary depending on the muscle that is stiff. For instance, if the muscles in the hands lock, they will be stuck in one position so will affect specific tasks carried out by the hands. If the tongue is affected, this can impact on speech and swallow. Stiffness may feel like cramp if the legs, feet and hands are affected. Stiffness in the legs can cause falls.

This stiffness may last for many seconds, for instance, when getting up out of a chair or holding a door handle to open a door. Usually the muscles begin to relax and work more freely after they are flexed, warmed up and stretched. However, if the activity is paused or stopped, the muscles may need to be 'warmed up' again.

Stiffness may be worse in cold temperatures during the winter or after periods of rest but usually improves with repeated actions (warming up).

Other factors which trigger stiffness can include:

- sudden loud noises
- intense exercise
- stress
- certain foods or medications
- other illness

How is chloride channel myotonia diagnosed?

The doctor will take a careful description of what symptoms occur and when, along with a physical examination which may show the doctor your child has myotonia. A gene test on a blood sample is needed to make a definite diagnosis.

How is chloride channel myotonia treated?

The aim of treatment is reduce the frequency and severity of attacks of stiffness. This is done with a combination of medication alongside dietary and activity changes.

Medication

The main medication prescribed is called mexiletine hydrochloride. At GOSH this is supplied

under the brand name Namuscula® in the form of capsules.

The most common side effect is heartburn. This can be managed by taking Namuscla® with food or sometimes giving another medicine alongside it that reduces the amount of acid produced in the stomach is required. Sleeping problems and wobbliness have also been reported.

Other treatment options may be suitable for your child – the Clinical Nurse Specialist will discuss these with you in clinic.

What is the outlook for children and young people with chloride channel myotonia?

Episodes of stiffness can usually be reduced in severity and frequency by making simple lifestyle changes and 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