Myotonia Congenita and Pain

As moderator of a large international support group for non-dystrophic myotonias, I often hear from members whose doctors tell them that any musculoskeletal pain they are experiencing is caused by the myotonia. There are two main reasons I have seen for this misconception.

The first is that most doctors have never seen a patient with myotonia congenita, so they either check their neurology textbooks (often outdated) or do a quick search online to try to get some information. Unfortunately, when using the search term “myotonia” the majority of articles will be related to myotonic dystrophy (DM), not myotonia congenita (MC). When they read about the symptoms, which can include significant nerve involvement, they assume this applies to MC. They often attribute cardiac abnormalities or GI issues to MC for the same reason.

The second reason it is often misunderstood is because they assume myotonia congenita is a neuromuscular disease, therefore it can involve nerves and nerve inflammation. Actually, myotonia congenita is a chloride ion channel disorder that only affects the cells in skeletal muscles. When the muscle contracts, the positively charged sodium and potassium ions have to be neutralized by chloride ions in order for the muscle to relax. Normally this happens smoothly and there is no awareness of the depolarization/repolarization cycle as we voluntarily move our muscles. But in a person with MC, this process can take ten times longer, and that can become very noticeable. In some cases, such as with Becker’s recessive MC, the person can seize up for several seconds and even fall. This is the cause of the falling seen with “fainting goats.”

Myotonia congenita is the inability of the muscle to maintain that cycle of contraction/relaxation at a normal pace. That’s all. It can affect any part of the body related to skeletal muscle including eyes, diaphragm, throat, upper esophagus and even tongue. The larger white fiber muscle groups such as calves, quads, abs and biceps tend to be the most affected. But hands are also quite problematic since it’s difficult to release a grip quickly when opening doors, shaking hands or setting down an object.

Myotonia congenita does not affect smooth muscles like the GI tract, uterus, bladder, or kidneys. Because the upper third of the esophagus is skeletal muscle, it can cause the tendency to choke or to develop esophageal pouches. Sphincter muscles in general are not affected, but the one exception seems to be the anal sphincter which over time morphs to contain some skeletal muscle. Myotonic discharges have been seen in that sphincter and could contribute to some spasms.

The skeletal muscle pain with myotonia is caused by microtears to the muscle when exerting force on a muscle that has not relaxed (repolarized) from the current contraction. We especially see this when people are self-conscious about climbing stairs and force their leg to take the next step when it hasn’t released from the previous contraction. The effect is very similar to body builders who purposefully “rip” muscles in order to increase bulk. As the adaptive fibers form to deal with the extra stress, this causes hypertrophy of the muscle. The degree of hypertrophy seen in someone with myotonia will depend on their genetic makeup including distribution of white and red fiber muscles, glycogen storage, electrolyte balances, and the degree of inhibition of the ion channel caused by their particular mutation or mutations.

Having a mutation associated with myotonia does not in itself cause pain. The majority of patients with documented mutations do not experience pain, only stiffness. We can often control the tearing of the muscles and resulting soreness and cramping by awareness of our movements and pausing to give a

contracted muscle group time to relax. However, that is not always convenient or even possible. This pain should be equivalent to what you would see in people who went to the gym and overextended themselves. In a normal person, this soreness would usually heal in a few days. But because we are confronted with this isometric force daily, every time we get out of a chair and start to walk or climb stairs, the pain from the muscle damage can be quite persistent, but it is not permanent and it is not caused by nerve-related issues.

If a person with myotonia congenita has symptoms of spine pain or inflammation, or if there is ongoing widespread muscle pain, it should then be assumed that there is a co-existing condition. We are just as likely as anyone else to develop back and joint pain or neuropathy. Unfortunately, when patients mention these symptoms to their doctors, they are often told it's just the myotonia and no further investigation is carried out. As a result, many suffer needlessly when the other conditions could be successfully treated.

The most common reasons I have seen for pain other than muscle soreness are herniated or bulging discs, spinal stenosis, degenerative disc disease, peripheral neuropathy and autoimmune conditions. Some of our members also have Ehlers-Danlos Syndrome or a mitochondrial disease. These are totally unrelated to myotonia congenita. The preferred pharmaceutical treatment for MC is either sodium channel blockers like mexiletine or flecainide, or the anti-seizure meds like carbamazepine and lamotrigine. These reduce the number of positively charged ions in a muscle cell so less chloride ions are needed to repolarize the cell. Doctors often prescribe muscle relaxants which are not effective unless the person has an additional condition which benefits from them.

When treating pain in someone with myotonia, NSAIDs can actually worsen the stiffness. This is because they tend to increase serum potassium, so moderation is the key. Local anesthetics used for nerve blocks need to be without vasoconstrictors. Epinephrine will greatly worsen myotonia temporarily. Corticosteroids will temporarily improve myotonia because of the increased excretion of potassium, but they are discouraged as a regular treatment unless needed to control autoimmune conditions because of the long-term side effects. Magnesium has been shown to be very beneficial in relieving myotonia as well as nerve pain. The forms that seem to be most effective are magnesium glycinate or citrate.

In conclusion, it is essential that a healthcare provider differentiate between the muscle soreness and cramping normally seen with myotonia in skeletal muscles, and pain from other conditions. Myotonia congenita is not considered a disabling or life-shortening condition. If someone is so severely affected they are not able to work or participate in normal daily activities, then either another genetic or acquired condition is contributing, they are on medications that are further inhibiting the chloride channel, or they are being exposed to environmental factors such as pesticides and herbicides that are affecting the function of the enzyme (see contraindicated medications list). Diet and supplements high in potassium may also worsen myotonia, as will a high sugar intake, stress (from increased adrenaline output) and intense exercise (increased insulin).

I hope this is helpful in determining whether additional testing should be undertaken to diagnose possible co-existing conditions.

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