Understanding Myotonia Congenita – the Patient's Perspective

Correcting the Myths and Misconceptions

I represent a group of myotonia congenita patients from all over the world who are dedicated to educating health care professionals about our very rare condition. Some of us lived with this perplexing condition well into adulthood before being diagnosed. The luckier ones were diagnosed early, and were therefore able to be understood by their families, physicians, friends and teachers. Some have no other affected family members, while others share it with parents, siblings, cousins and children.

The purpose of this letter is to clarify some misconceptions the medical community perpetuates about MC, causing both confusion and consternation for patients and their loved ones. It doesn't help that there are other conditions with similar names, for instance Myotonic Muscular Dystrophy or Becker's MD.

We address the two types of myotonia congenita caused by mutations in the skeletal chloride channel (CLCN1). The dominant form is Thomsen's Disease, the recessive form is Becker's Myotonia Congenita or Becker's Generalized Myotonia. Inappropriate coding has caused many of us to be denied access to health insurance.

The great majority of information disseminated by literature, websites and physicians states that this condition has no pain or significant handicap associated with it. We feel that it is a disservice to the sufferers of this condition to say this. As a matter of fact, while the channelopathy itself does not inflict pain, its manifestations most certainly do. And while the condition itself is not progressive or life-threatening, certain situations can cause it to become dangerous.

Some of the pain caused by this condition is actually quite severe, and patients whose pain has been dismissed, or attributed to psychological, psychosomatic or other dismissive reasons, are not being properly evaluated or acknowledged. When our muscles contract suddenly or unexpectedly (sneezing, stretching, stumbling, etc), they seize or spasm or lock so tightly that the pain can bring tears. And you are "stuck" that way until your muscles release you.

When we participate in physical activities, such as sports, dancing or gardening, we often feel very achy, weak and often shaky and unstable afterwards, and have increased difficulties with contractions and mobility. The after-effects of an adrenalin-stimulating incident can make our entire bodies ache like the flu, and our muscles may react to exercise by becoming exquisitely tender and sore. This can persist for days.

Many of us experience fairly significant fear of heights, open spaces, public places or public speaking, crossing the street, walking on uneven surfaces, and many other situations where our muscle contractions could seriously jeopardize our safety and well-being with just one wrong move.

It is a very real terror to know that at any time, if you let your guard down, you could be in a position where you lose the ability to control your own movement and can topple over like a bowling pin, unable to help yourself. Some even wear helmets to protect from head injuries.

You may consider the embarrassment of such occurrences – yes, it is traumatizing, and impacts the ways many of us choose to live our lives. But there is also the very real danger of tumbling down a flight of steps, off a horse, in front of a moving car, or off the edge of a lovely ocean bluff!

These are just a few of the sensations our "non-painful, non-dangerous" condition cause us, and we would very much appreciate updates to the general information found in medical books, journals, web-sites, conferences – and especially in the teaching done with residents in the fields of neurology, rheumatology, physiatry, orthopedics, and associated clinical and therapy fields.

You will find that we are extremely susceptible and vulnerable to cold, fear, stress and anxiety. We can experience frightening problems with swallowing and breathing. When our tongues become stiff, we are mistakenly thought to be drunk, or otherwise impaired. While our bodies look remarkably muscular, those muscles frequently betray us. As one patient puts it, "I may look like Mr. America, but you could knock me over with a feather."

We all grow up with a sickening dread of having to move quickly, as in fire drills or getting off a bus or train. Just as dreadful is being the object of attention or focus, for instance when we have to walk to the front of a classroom or give a report – you stand and hobble stiffly, your tongue gets stiff, your teacher tells you to speak more clearly and the students giggle while comparing you to Frankenstein. Many students become very introverted as a result of constant teasing.

The most dangerous potential of this condition is anesthesia reaction. We are all susceptible to the symptoms associated with malignant hyperthermia from certain muscle relaxants and anesthetics, as well as cardiac arrest from potassium buildup in the cells. It is imperative that patients be counseled to wear medical alert jewelry, and that anesthesia teams be consulted before surgery, even for family members who have no obvious symptoms of myotonia.

Medications need re-evaluation. Newer medications can be much more effective but patients are still often prescribed the older drugs listed in aging medical texts. For some who are unable to tolerate the side effects of medications used to treat myotonia, or prefer not to use them, a diabetic diet with restricted potassium can be quite effective. Foods which spike insulin levels, as well as potassium, and potassium-based preservatives and additives are particularly troublesome. This is similar to the reactions seen with hyperkalemic periodic paralysis, a sodium channel disorder.

Diagnosing myotonia congenita can be challenging. Physicians must be open to odd complaints or observations that once were dismissed as laziness, clumsiness, lack of motivation, attention-seeking or malingering. An EMG is usually the first step, but now, rather than painful and nearly obsolete muscle biopsies, we have readily available DNA testing, with approximately 80 mutations identified to date. We are asking for empathy and validation of our condition. We understand that it is not generally progressive or massively debilitating, but it is more than mildly inconvenient to live and deal with, and causes its victims very real pain, concern and disability.

We appreciate your kind and professional attention and concern, and hopefully your support. Many of us are willing to answer questions and give our personal histories to researchers, physicians and others interested in learning more "from the trenches".

With best regards,

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Representing The Myotonia Congenita Project, an international forum dedicated to the education and support of patients and families dealing with myotonia congenita, at www.MyotoniaCongenita.org.