CASE DIAGNOSIS: Pain, Muscle Spasm and Myotonia Congenita in a 23 Year Old Male: A Case Report

CASE DESCRIPTION: A 23-year-old male with multiple episodes of worsening lower extremity weakness underwent acute inpatient rehabilitation after admission to acute care for inability to walk and severe pain. The patient had profusely elevated creatinine kinase. Initial evoked potentials revealed motor unit action potentials of reduced amplitude consistent with myopathy. Naege EMG/NCV of the lower extremity revealed elevated repetitive myotonic discharges and spontaneous abnormal potentials consistent with muscle spasm. After performing an extensive genetic marker analysis which was positive for a gene variant sequence, we confirmed the diagnosis of Myotonia Congenita.

DISCUSSION: The patient presented with severe muscle spasm, hypokinesia and atrophy of his back and lower extremities rendering him nearly immobile. We initiated treatment with oral baclofen and titrated up the dose. We utilized tiotidine initially which was replaced by cyclobenzaprine as an alternative agent. Aggressive treatment with antispasmodic medication in conjunction with the use of applied heat, therapeutic ambient temperature and aquatherapy successfully improved functional outcome. Initial Functional Independence Measure (FIM) on admission was moderate to maximum assistance for mobility and lower extremity activities of daily living. Discharge FIM score was modified independent. The patient returned home with a walker and lower extremity orthoses. A month after discharge, the patient was able to ambulate, drive and return to work without assistive devices. His muscle spasm became less painful allowing for weaning of cyclobenzaprine. He remained on oral baclofen.

CONCLUSIONS: This case of Myotonia Congenita presented a special challenge with respect to acute inpatient rehabilitation due to the patient’s severe pain related to muscle spasm on initial presentation. Patients with Myotonia Congenita require an aggressive multimodal approach utilizing heat and antispasmodic medication to return them to their previous level of productivity and independence.

Introduction

- Myotonia congenita caused by mutations in the skeletal muscle ion channel gene (CLCN1) on chromosome 7 responsible for shutting off electrical excitation causing muscle fiber membrane hypersensitivity (Figure 1).
- Prolonged muscle contractions often with severe spasm is hallmark of myotonia congenita.
- Symptoms include delayed relaxation of the muscles after voluntary contraction along with hypokinesia, weakness, pain, and cramping.
- PRO Found to be prolonged forward flexion contractures enhanced by cold and inactivity, and relieved by repetitive movement in a phenomenon known as the “warm-up effect.”
- Stress hormones make myotonia worse with associated functional decline during stress syndromes associated with systemic illness such as pneumonia and infections.
- Becker and Becker types distinguished by severity and patterns of inheritance.
- Becker disease appears later in childhood and causes more severe myotonia, muscle stiffness and pain and is inherited in an autosomal recessive pattern.
- Becker disease has temporary attacks of muscle weakness, often in arms and hands, brought on by movement after periods of rest with most persistent muscle weakness.
- Permanent muscle weakness is not seen in Thomsen disease which is inherited in autosomal dominant pattern such that an affected person usually has one parent with the condition.
- As more individual mutations that cause myotonia congenita are identified, broad disease classifications are becoming less widely used.
- High phenotypic variability and severity of symptoms can vary greatly between individuals with over 80 different mutations that can cause the disorder, each with a specific phenotype.
- Forms of myotonia congenita is estimated to affect 1 in 100,000 people worldwide.

Discussion/Conclusion

The patient’s severe muscle spasm, hypokinesia and atrophy of his back and lower extremities rendered him nearly immobile and hindered active participation in his rehabilitation. We initiated treatment with oral baclofen and titrated up the dose while weaning down to an alternating regimen of hydrocodone and tramadol. We utilized tiotidine initially, but it was poorly tolerated and eventually replaced by cyclobenzaprine as an alternative agent to baclofen. Given that cold exacerbated the disease phenotype, applied heat before starting and progressive exercise with low weights resulted in a “warm-up effect” that improved mobility. Additionally, the ambient temperature of the patient’s room was increased after providing education on the deleterious effects of cold temperature as the patient had a poor history of heat intolerance. The patient was able to discharge on high dosage of baclofen and hydrocodone.

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Figure 1. Describes the sites of the mutations in the skeletal muscle ion channel (CLCN1) channel. Circles represent dominant myotonia congenita (Becker) and squares recessive myotonia congenita (Becker). (1)

Figure 2. Shows myotonic discharges on needle EMG. Note: Sweep 50 μv/div, sensitivity 10 μv/pinh.(2)

Figure 3. Shows cramp discharges on needle EMG. Note: Sweep 500 μv/div, sensitivity 10 μv/pinh. (3)

Figure 4. Functional grade made for the patient from admission to discharge. Note: From acute inpatient rehabilitation as compared to the respective national benchmark. (4)

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