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A Rare Case of Pain Management in Pompe Disease

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Background: Pompe disease is an uncommon inherited disorder of glycogen metabolism and lysosomal storage caused by reduced alpha-glucosidase activity. The resultant accumulation of glycogen results in skeletal myopathy, which can be a painful condition. Muscles of the pelvic girdle and proximal lower limbs are usually preferentially involved.

Case Report: We present a case of long-standing axial low back pain in a patient with a known history of Pompe disease. She had failed management with physical therapy, multiple medications including nonsteroidal anti-inflammatory drugs, neuropathic medications, muscle relaxants, and opioids. Magnetic resonance imaging revealed marked fatty atrophy of paraspinal and psoas muscles, loss of lordosis, bilateral L4-L5 disc bulging with flattening of the ventral thecal sac margin and neural foraminal stenosis, and facet hypertrophy of T11-T12 resulting in posterior canal stenosis. Bilateral medial branch blocks of the L2-L5 levels with steroid and local anesthetic were performed twice for relief of axial back pain with good results.

Conclusion: Management of Pompe disease involves a multi-disciplinary approach that should aim to identify and alleviate pain symptoms. Medial branch blocks can be used successfully for the short-term treatment of axial back pain in Pompe disease.

Key words: Back pain, facet arthropathy, glycogen metabolism, medial branch block, muscle atrophy, myopathy, Pompe, radiofrequency ablation

BACKGROUND

Acid alpha-glucosidase deficiency (Pompe disease) is a rare inherited disorder of glycogen metabolism and lysosomal storage caused by reduced alpha-glucosidase activity. Accumulation of glycogen within lysosomes results in skeletal myopathy and progressive muscle weakness in the late-onset genotype. Pain is often a presenting feature, but treatment modalities for the management of axial back pain in Pompe disease are limited.

CASE PRESENTATION

A 55-year-old woman with Pompe disease reported a 10-year history of severe, progressive generalized pain and muscle weakness. Previous treatment modalities included physical therapy, ibuprofen, cyclobenzaprine, tramadol, clonazepam, tizanidine, oxycodone, gabapentin, and ketorolac injections, with no relief of symptoms. Magnetic resonance imaging (MRI) revealed marked fatty atrophy of the paraspinal and psoas muscles, loss of lordosis, bilateral L4-L5 disc bulging with flattening of the ventral thecal sac margin and neural foraminal stenosis, and facet hypertrophy of T11-T12 resulting in posterior canal stenosis. Bilateral medial branch blocks of L2-L5 with steroid were performed twice for relief of axial back pain with good results.

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DISCUSSION

Pompe disease is a rare metabolic myopathy caused by deficient or absent activity of the enzyme acid alpha-1,4-glucosidase required for lysosomal glycogen metabolism. It is inherited in an autosomal recessive pattern with marked allelic heterogeneity, with over 450 mutations known to affect enzyme activity levels that account for the variable phenotypes seen in Pompe disease (1). Late-onset acid alpha-glucosidase deficiency is characterized by reduced activity of the enzyme leading to tissue destruction, notably in skeletal muscle, cardiac muscle, and smooth muscle. The late-onset phenotype can manifest at any age, notably with progressive skeletal myopathy of variable severity. Oftentimes, pain is the only presenting feature of Pompe disease; it can interfere with general activities, mood, and quality of life (2). Patients are likely to report pain within the last 24 hours and an unsatisfactory treatment of their pain, and disease progression has been associated with more pain, greater fatigue, and poorer quality of life (2). While enzyme replacement therapy (ERT) has been instrumental in slowing disease progression and reducing mortality rates, it has been shown to be insufficient in the management of pain in patients with Pompe disease after 2 years of treatment, after which pain begins to worsen (3). Unfortunately, studies devoted to treatment modalities remain limited.

The mechanism of development of pain has been attributed to skeletal myopathy leading to muscle weakness and mechanical stress (4). Clinically, such patients present with features of proximal myopathy with muscles of the pelvic girdle and proximal lower limbs, more specifically paraspinal muscles, thigh adductors, and glutei muscles being preferentially involved (5). This often results in lumbar hyperlordosis, waddling gait, and Gower's maneuver. Fatty infiltration of erector spinae muscles has been demonstrated on MRI in patients with axial myopathy (6). A study of 30 patients that examined truncal muscle involvement via MRI or computed tomography in late-onset Pompe disease observed a selective pattern of muscle damage, with early involvement of the multifidus, followed by the obliquus internus abdominis and longissimus muscles (7).

In the case of our patient, marked fatty atrophy of the paraspinal and psoas musculature was noted on her imaging. Medial branch blocks are commonly used interventional pain modalities for the diagnosis and management of axial lower back pain. Given atrophy of the lumbar paraspinal muscles as evident on her MRI, we decided to perform a diagnostic and therapeutic medial branch block. The paraspinal muscles are comprised of 3 muscles: namely the iliocostalis laterally, longissimus in the middle, and the multifidus muscles, which are the largest, deepest and most medial layer. Multifidus muscles are innervated by the medial branches of the posterior divisions of the spinal nerves. Theoretically, blocking the lumbar medial branches should relieve the painful symptoms in cases where atrophic paraspinal muscles are the primary pain generator. As expected, this patient reported complete pain relief and return of functionality immediately following therapy up to 2 weeks after the first medial branch block. After 2 weeks, the patient reported sudden onset return of pain and eventual return to baseline, which may have been a result of excessive activity during the interval of pain relief. We repeated the bilateral L2-L5 medial branch blocks a second time, after which the patient reported pain relief and functional improvement for almost 2 months. Both times, a mixture of 80 mg of kenalog (40 mg/mL) and 6 mL of bupivacaine 0.25% was injected divided equally amongst all levels.

Typically, a successful response to 2 diagnostic lumbar medial branch blocks is followed by thermal radiofrequency ablation (RFA) to achieve pain relief and functional improvement for a longer period of time. Due consideration was given to the performance of RFA for this patient as well. However, we elected to not proceed with RFA due to concern for the possibility of postprocedure muscle weakness. In addition to innervating the facet joint, each medial branch also contains efferent fibers communicating motor activity to the adjacent multifidus muscle; this process may be interrupted by RFA (8). Loss of these nerves usually does not cause any significant loss of motor function in an otherwise normal patient. However, in this patient with preexisting skeletal myopathy we were concerned that possible loss of multifidus motor function post RFA may have worsened pain and weakness with the potential to result in further loss of stability. Hence, we decided to forego performing an RFA even with a positive response to diagnostic lumbar medial branch blocks.

Given the inability to clear neuronal glycogen in Pompe disease, there is continual toxic metabolic accumulation in tissues (2,9). Hence, a limitation to medial branch blocks for axial low back pain in these patients is that this technique may offer only temporary short-term therapeutic benefit. Nevertheless, pain symptoms in patients with late-onset alpha-glucosidase deficiency

remain prevalent, and research on the management of chronic pain in Pompe disease remains limited. Due to the progressive nature of the disease and considerable burden of illness, medial branch blocks may confer therapeutic benefit for the short-term management of chronic axial low back pain in Pompe disease.

CONCLUSION

Management of Pompe disease involves a multidisciplinary approach that should aim to identify and alleviate pain symptoms. Medial branch blocks can be used successfully for the short-term treatment of axial back pain in Pompe disease.

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