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CASE REPORT

Myotonic Dystrophy Mimicking Postpolio Syndrome in a Polio Survivor

ABSTRACT

Lim J-Y, Kim K-E, Choe G: Myotonic dystrophy mimicking postpolio syndrome in a polio survivor. Am J Phys Med Rehabil 2009;88:161–164.

We describe a 38-yr-old polio survivor with newly developed weakness from myotonic dystrophy. He suffered muscle atrophy and weakness in his legs as a result of poliomyelitis at the age of 3 yrs. After a stable interval of about 30 yrs, he felt new weakness and fatigue in his legs. Electromyography revealed generalized myotonic discharges, early recruitment, and findings of chronic denervation in his left leg. Genetic testing was consistent with myotonic dystrophy type 1. A biopsy from the right gastrocnemius revealed findings of both myotonic dystrophy and chronic denervation. This case report shows the importance of considering other uncommon conditions in the differential diagnoses of postpolio syndrome.

Key Words: Postpolio Syndrome, Myotonic Dystrophy, Differential Diagnosis

Late-onset neuromuscular symptoms in a previous polio survivor are known as postpolio syndrome. New muscle weakness, abnormal muscle fatigability, and muscle or joint pain start after a long interval of stable neurological function following an acute episode of paralytic poliomyelitis.[1,2] The diagnosis of postpolio syndrome is made on diagnostic criteria that are based on both clinical and electromyographic findings, but excluding other neurologic, medical, and orthopedic problems that could have similar manifestations is important.[2–4] A detailed examination should be carried out before making a diagnosis of postpolio syndrome. Several studies have reported other neuromuscular diseases manifesting similarly to postpolio syndrome, such as cervical polyradiculopathy[5] and spinal stenosis.[6] In those cases, the exact diagnosis and proper management were sometimes delayed because of the preconception that the symptoms were due to postpolio syndrome. In addition, some patients have chronic progressive coexisting problems leading to weakness, pain, and fatigue, such as myopathic disorders and other motor neuron diseases.[7–9] They could be diagnosed as having postpolio syndrome because of a lack of evidence for other causes, especially during the early stages of the chronic progressive disease. The early detection of correctable and treatable causes of late-onset weakness and pain may help to reduce the functional declines of polio survivors.

We had the opportunity to evaluate a polio survivor, whose symptoms were thought by others to be related to postpolio syndrome before we discovered that the patient also had myotonic dystrophy. To help physicians in making the

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differential diagnoses for postpolio syndrome, we present this case report as an example of another medical condition that can mimic postpolio syndrome.

CASE DESCRIPTION

A 38-yr-old man visited the electrodiagnostic laboratory of Seoul National University Bundang Hospital complaining of progressive weakness and fatigue in his legs along with low back pain. At the age of 3 yrs, he experienced a high fever for about a week. Subsequently, his left leg became weak and atrophied and he walked with a limp. He had been told that these were the sequelae of poliomyelitis. When he was 34 yrs old, he began to experience new weakness and fatigue in his legs with no sensory symptoms. He visited a local clinic and was told that the symptoms might have come from hip osteoarthritis or some other musculoskeletal problems. Six months later, he required a cane for walking, and eventually, at the age of 38, had to use bilateral forearm crutches to walk long distances. He was diagnosed with hypothyroidism 3 mos before visiting our hospital and received thyroid hormonal replacement therapy for 3 mos. His fatigue and heaviness in his legs improved slightly, but the symptoms and walking difficulty persisted. After spending 4 yrs visiting several local clinics for weakness and fatigue in his legs, he came to the orthopedic outpatient clinic of our hospital and was referred to our laboratory for electrodiagnostic examination.

His medical history included minor traffic accidents 5 and 8 yrs earlier that caused trivial low-back and cervical pain for a few months. The neurological examination revealed profound weakness in the left hip flexor and knee extensor (Medical Research Council grade 2), right hip flexor and knee extensor, and both ankle dorsiflexors (Medical Research Council grade 3). His upper extremity strength was relatively well maintained. Muscle atrophy was found in both thighs and calves, and it was more severe on the left side. The sensory examination was normal. His muscle tone and tendon reflexes had generally decreased, with no upper motor signs detected. He had mild scoliosis of the lumbar spine, with associated pelvic obliquity, which may have been at least a partial cause of his limp. Frontal balding, ptosis, and facial weakness were not noted. He had no complaints of dysarthria or dysphagia, but action myotonia with the warm-up phenomenon and percussion myotonia were observed in his hands.

The motor nerve conduction studies show that the compound muscle action potential of the left tibial nerve recorded in the abductor hallucis muscle had decreased to 5.1 mV compared with 14.8 mV for the right tibial compound muscle action potential. Other motor and sensory nerve conduction studies revealed no additional abnormal findings. Needle electromyography showed profound myotonic discharges in all of the examined muscles of the legs, lumbar paraspinals, and left arm. All of the muscles in the left leg showed high-amplitude long-duration polyphasic motor unit action potentials with reduced recruitment. Short-duration polyphasic motor unit action potentials with early recruitment were recorded in the left biceps brachii muscle.

He was admitted to our hospital for further workup and management. His serum creatine kinase was within the normal range. Thyroid function tests still showed mild hypothyroidism. Electrocardiography revealed no abnormal findings. A genetic study revealed abnormal expansion of the CPG repeat to 400 (normal range, <50) in the gene encoding dystrophia myotonica protein kinase on chromosome 19q13.3. The right gastrocnemius muscle was biopsied, and histologic examination revealed frequent nuclear internalization and nuclear chains, suggesting myotonic dystrophy (Fig. 1*A*). In addition, pyknotic nuclear clumps (clumps of hyperchromatic nuclei), commonly seen in longstanding denervation, as is seen in poliomyelitis, were found (Fig. 1*B*). Enzyme histochemical studies revealed large clusters of type 1 fiber with loss of the normal mosaic checkerboard pattern (Fig. 1*C*). The fiber-type grouping is associated with chronic denervation, and it probably reflects reinnervation by collateral sprouting from surviving motor nerve axons. No grouped atrophy or endomysial fibrosis was observed, which would be present if denervation had occurred without reinnervation.

After the diagnosis was made, we examined one of his younger brothers, who was 31 yrs old. He had no known history of poliomyelitis, a 10-yr history of seizures. He had recently complained of difficulty in climbing stairs and intermittent cramps of his hands and feet. The electrodiagnostic examinations and genetic study confirmed the diagnosis of myotonic dystrophy. A biopsy of the left vastus lateralis showed frequent nuclear internalization, and nuclear chains were found (Fig. 2*A*), whereas neither pyknotic nuclear clumps nor fiber-type grouping was noted (Fig. 2*B*).

DISCUSSION

Postpolio syndrome is now well known to physicians who examine and treat polio survivors. This can sometimes lead to a delay in making an accurate diagnosis of other pathologies manifesting symptoms similar to those of postpolio syndrome. Our case is an example of this type of delayed diagnosis.

Our patient had inherited myotonic dystrophy and had suffered poliomyelitis at the age of 3 yrs,

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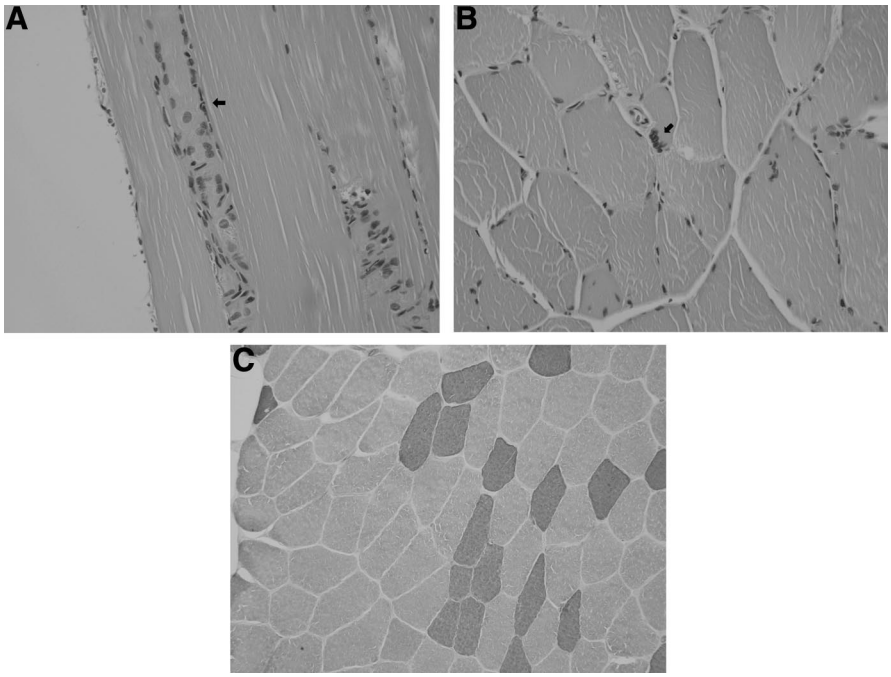

FIGURE 1 *Muscle biopsy showing findings of both myotonic dystrophy (*A*) and chronic denervation with motor unit reorganization and secondary fiber-type changes (*B, C*). A, frequent nuclear internalization and nuclear chains (*arrow*) (H&E, ×400); B, pyknotic nuclear clumps (*arrow*) (H&E, ×400); and C, fiber-type grouping with large clusters of type 1 fibers (pale fibers) (ATPase, pH 9.4, ×200).*

which left a sequelae of weakness and atrophy. The symptoms of myotonic dystrophy were not manifested until he was in his thirties, so the sequelae of poliomyelitis made an accurate diagnosis of myotonic dystrophy more difficult. He had lived with asymmetric muscle atrophy in his legs and had an associated spinal deformity (i.e., scoliosis). All these findings, along with the recent-onset progressive weakness, are typical symptoms of postpolio syndrome, so that other causes of his symptoms, such as myotonic dystrophy, were not considered.

The differential diagnosis of postpolio syndrome includes secondary sequelae as late effects related to the paralytic poliomyelitis itself. Most patients who experience postpolio functional deterioration have considerable orthopedic and neurologic impairments as a consequence of their polio that render them vulnerable to the development of new disabilities.[10] Degenerative changes or scoliosis owing to altered biomechanics from muscle weakness and the aging process, the normal loss of α-motor neurons with age, can also be included in the late-onset polio sequelae.[1,11] Cervical or lumbar radiculopathies and spinal stenosis are examples that have been reported previously.[5,6] Because we were concerned about these possibilities, our patient also underwent lumbosacral spine magnetic resonance imaging, which revealed no abnormal findings.

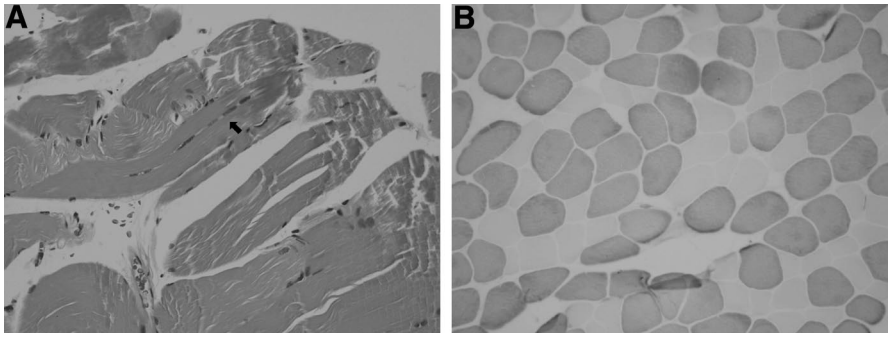

FIGURE 2 *Muscle biopsy of the patient's brother showing findings of myotonic dystrophy only.* A, *frequent nuclear internalization and nuclear chains without pyknotic nuclear clumps (*arrow*) (H&E, ×400).* B, *normal mosaic checkerboard pattern of type 1 fibers (*pale*) and type 2 fibers (*dark*) without fiber-type grouping (ATPase, pH 9.4, ×200).*

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The differential diagnosis of postpolio syndrome includes pathologies that can affect patients without a history of poliomyelitis. Studies have reported on inclusion body myopathy,[7,9] amyotrophic lateral sclerosis,[8] and spinal compression owing to cord tumor[12] mimicking postpolio syndrome, all of which are thought to be unrelated to the paralytic sequelae of poliomyelitis. Myotonic dystrophy can be added to these. Understanding that other diagnoses may mimic postpolio syndrome is important, because these may be treatable or controllable conditions, and a delayed diagnosis could be detrimental.

In the case reported here, we had no problem making the diagnosis of myotonic dystrophy, which was confirmed by genetic testing. However, because electrodiagnostic and pathologic evidence for chronic denervation existed in his leg muscles, as exhibited in a polio survivor, we questioned whether the progressive weakness was attributable to postpolio syndrome or myotonic dystrophy. His younger brother, who was diagnosed with myotonic dystrophy without any history of poliomyelitis, did not show any evidence of chronic denervation in contrast to our patient. The classic adult-onset myotonic dystrophy usually begins in midlife, at an age of 20–40 yrs.[13] Therefore, this type of weakness may have been strongly related to the symptom development and progression of myotonic dystrophy. These findings led us to confirm that our patient had suffered poliomyelitis and was experiencing residual sequelae, in addition to a new diagnosis of myotonic dystrophy. Because no diagnostic test for postpolio syndrome exists, considering and excluding other conditions that can produce similar symptoms are especially important. The concurrent diagnosis of poliomyelitis sequelae and myotonic dystrophy was made, and to the best of our knowledge, this type of case has not been previously reported.

CONCLUSION

The functional decline in middle-aged individuals who had acute paralytic poliomyelitis many years earlier in life is commonly recognized as postpolio syndrome. Other serious conditions can mimic its clinical findings, and myotonic dystrophy must be considered in the differential diagnosis when the clinical findings of postpolio syndrome are present.

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