P-140

Motor Function Gain Following Pyridostigmine in DNM2-Related Centronuclear Myopathy: A Case Report

Na Yoon Yoo[1], Seungbeen Hong[1]

[1]Department of Physical Medicine and Rehabilitation, National Health Insurance Service Ilsan Hospital

Background

Centronuclear myopathy is an inherited neuromuscular disorder characterized by marked weakness, hypotonia, external ophthalmoplegia, and respiratory failure. Patients with DNM2-related centronuclear myopathy may exhibit electrophysiological features consistent with neuromuscular junction (NMJ) dysfunction. Previous studies have explored the pathophysiological mechanisms linking centronuclear myopathy to NMJ abnormalities, and clinical observations have documented that treatment with pyridostigmine in affected individuals may lead to improvements in muscle strength and reductions in fatigability. Here, we share a case of an infant with genetically confirmed DNM2-related centronuclear myopathy who demonstrated significant improvement in motor performance following empirical pyridostigmine therapy.

Case Presentation

A female infant was born at 40+6 weeks of gestation by emergency cesarean section due to fetal distress, with a birth weight of 3100 g. Her Apgar scores were 5 at 1 minute and 5 at 5 minutes, and she was intubated and admitted to the NICU. At 1 week of age, she was referred to the Department of Physical Medicine and Rehabilitation for evaluation of a possible neuromuscular disorder.

Neurological examination revealed generalized hypotonia and weakness, and the Test of Infant Motor Performance (TIMP) yielded a raw score of 17, placing her below the 5th percentile (**Fig. 1, A**). Next-generation sequencing using a neuromuscular disorder panel identified a DNM2 mutation (c.1856C>T), a variant strongly suggestive of centronuclear myopathy.

Because neuromuscular junction dysfunction has been reported in association with DNM2 variants, a repetitive nerve stimulation test was performed but showed no decremental response. Nevertheless, empirical treatment with a cholinesterase inhibitor was initiated. Prior to therapy, a repeat TIMP assessment demonstrated a score of 18, essentially unchanged from baseline (**Fig. 1, B**). Pyridostigmine was started at 5 mg/kg/day and subsequently increased to 6.5 mg/kg/day. At 9 weeks of age, after 10 days of treatment, the TIMP score improved to 37, indicating newly acquired motor abilities (**Fig. 1, C**). At 11 weeks of age, following discharge, the TIMP score further increased to 43, confirming sustained improvement in motor performance (**Fig. 1, D**).

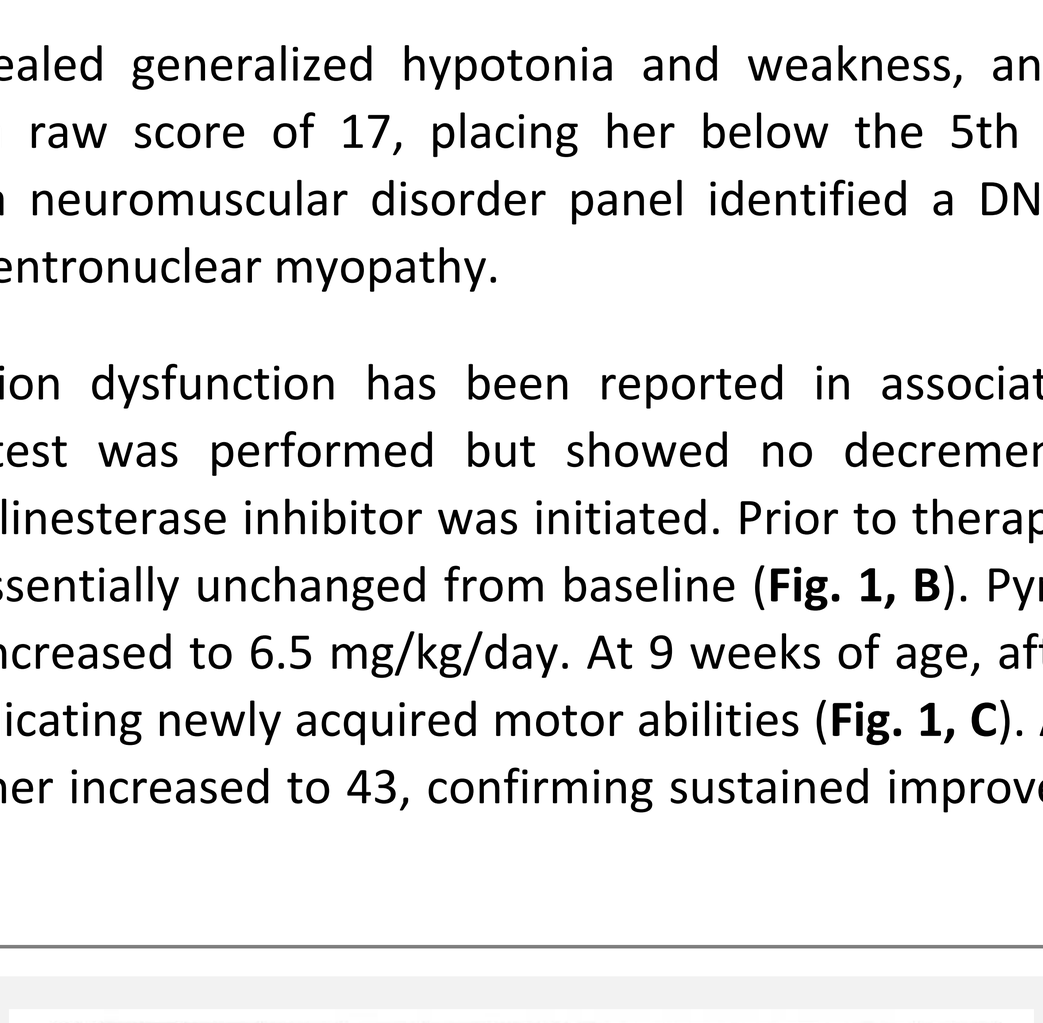

Fig. 1. TIMP raw scores. A: Initial assessment at 1 week of age; B: At 7 weeks of age, prior to cholinesterase inhibitor administration; C: At 9 weeks of age, after treatment with the maximum dose; D: At 11 weeks of age, following discharge.

Abbreviation: TIMP, Test of Infant Motor Performance.

Conclusion

This case illustrates a genetically confirmed centronuclear myopathy due to a DNM2 mutation in which empirical pyridostigmine therapy led to significant functional improvement, suggesting that cholinesterase inhibitors may provide clinical benefit even when neuromuscular junction dysfunction is not demonstrable on electrophysiological testing.