Hereditary Spastic Paraparesis Type 55: Case report

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INTRODUCTION

Hereditary spastic paraparesis is a large group of neurodegenerative diseases that result from primary retrograde dysfunction of the corticospinal system. C12orf65 gene mutations that cause hereditary spastic paraparesis type 55 have been associated with early-onset optic atrophy, progressive encephalomyopathy, peripheral neuropathy, and spastic paraparesis.

CASE

A 7-year-old boy born to healthy consanguineous parents was admitted with a decrease in visual acuity and gait disturbance. He was born 2800 g at term with cesarean section. He started walking at the age of 14 months. Visual impairment developed at three years, and gait disturbance after age five. He was receiving special education with the diagnosis of attention deficit and hyperactivity. His visual acuity was decreased (0.3 on the right, 0.2 on the left, +5.00/+5.00 with glasses), and optical disc was atrophied (Figure I). He had bilateral pes cavus, drop foot, and steppage gait. Deep tendon reflexes in the lower extremity were hyperactive.

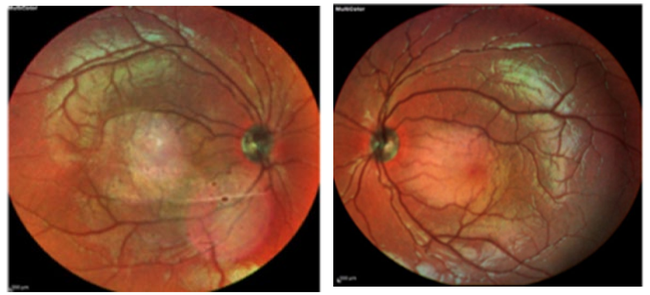

Figure I. Optic disc pallor, optic atrophy

The plantar reflex was flexor. Muscle strength was 5/5 in the upper extremity, 5/5 in the proximal lower extremity, 4/5 in the plantar flexion, and 2/5 in the dorsiflexion. There was atrophy in the distal muscle groups of the lower extremities (Figure II). Sensory examination was normal. Brain and spinal magnetic resonance imaging (MRI) and MR spectroscopy were normal, while electroneuromyography was compatible with polyneuropathy. Motor evoked potential showed signs of pyramidal pathway involvement. The patient who was found to have a homozygous frameshift mutation in the C12orf65 gene in whole-exome sequence analysis was diagnosed with hereditary spastic paraparesis type 55(SPG55) (Figure III). His parents were carriers of this mutation.

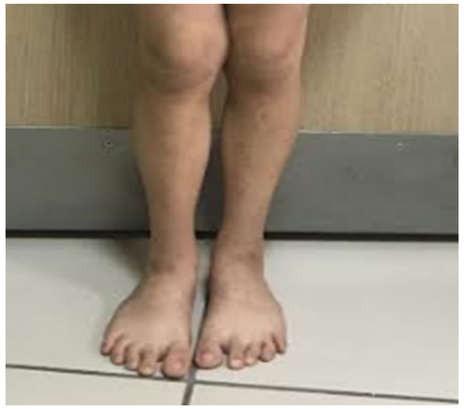

Figure II. Atrophy of distal lower extremity muscle groups and pes cavus

CONCLUSION

More than 80 genes have been associated with HSP, and it has wide genetic heterogeneity. C12orf65, which is the cause of SPG55, is one of them and is very rare. The most common clinical features in C12orf65 gene mutation are optic atrophy, peripheral neuropathy, and spastic paraparesis, as in our patient. C12orf65 mutation should be kept in mind in the differential diagnosis of spastic paraparesis and progressive acquired optic atrophy.

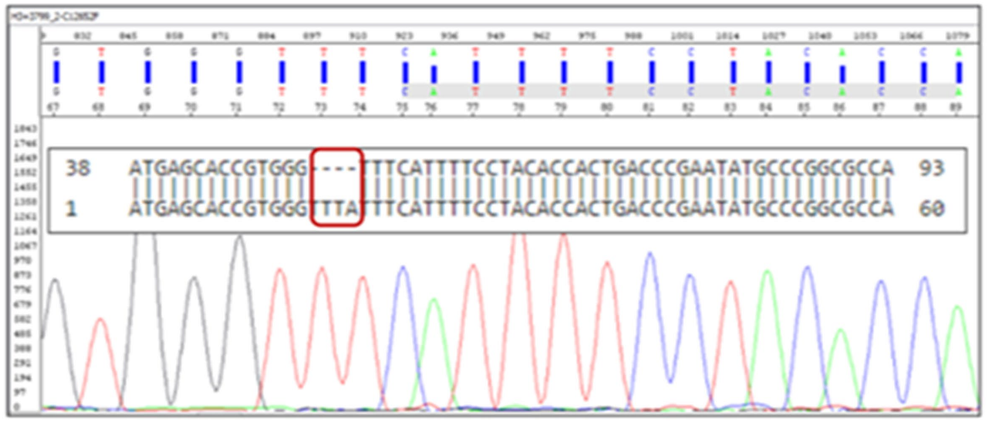

Figure III. Whole exome sequence analysis; C12orf65 gene c.15_18del (p.G5fsX) (NM_001143905.2)

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