Hypotonia or Spasticity?
A 2-year-old Male with Hereditary Spastic Paraplegia. A Case Report

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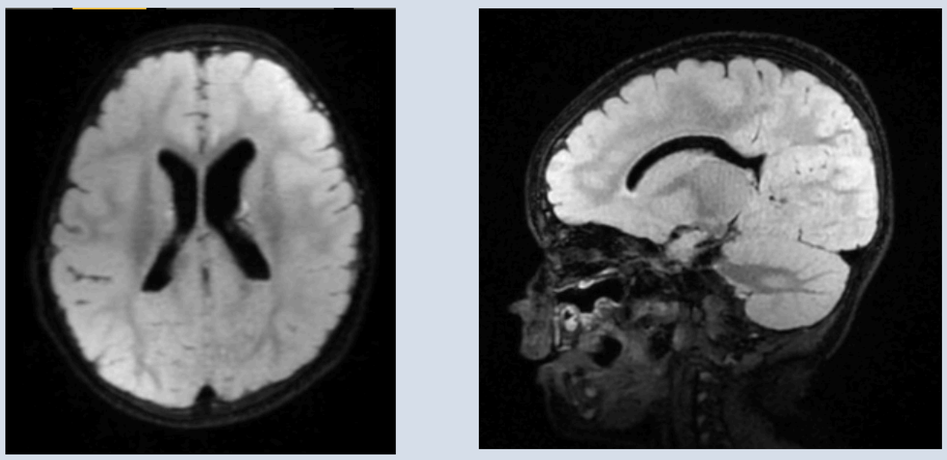
INTRODUCTION

Hereditary spastic paraplegia 50 (SPG50) is an autosomal recessive neurodegenerative disorder that presents with global developmental delay, impaired speech, intellectual disability, microcephaly, seizures, and progressive motor symptoms. Early onset hypotonia evolves into hypertonia and spasticity. The majority of children become non-ambulatory and oftentimes wheelchair bound[1-2].

Figure 1. MRI of the brain (A) Axial view T2 FLAIR image (B) Sagittal view, T2 image



CASE PRESENTATION

A 2-year-old boy with hypotonia, seizure disorder, ectopic left kidney, and oropharyngeal dysphagia presented for assessment of gait. His developmental history was remarkable for global delay with developmental regression at 12 months of age. A brain MRI showed nonspecific abnormalities including bilateral cerebral white matter volume loss with ex vacuo enlargement of the lateral ventricles, and hypoplasia of the corpus callosum (figure 1).

Initial physical exam was significant for microcephalia, synophrys, and left more than right valgus foot deformity. Patient had full passive range of motion with no spasticity. He was able to grasp objects using both hands as well as reach for objects, and transition from laying to sitting, but not from sitting to standing. He understood but did not speak. He was ambulating with assist, and gait pattern was notable for bilateral toe walking. Extensive genetic workup including whole exome sequencing, identified compound heterozygous pathogenic mutations in the *AP4M1* gene, consistent with a diagnosis of SPG50. Patient received early intervention therapies including physical, occupational, and speech therapy. At 2-years-old, patient received bilateral solid ankle foot orthoses to help with walking; and at 3-years-old, after developing spasticity, received botulinum toxin injection.

DISCUSSION

Many of the initial clinical manifestations of SPG50 are nonspecific and may resemble other disorders characterized by spasticity and developmental delay such as cerebral palsy. Our patient presented in the hypotonic phase of the disease and was diagnosed early. At this time there is no known cure for this disease, however AP4B1 gene therapy is under investigation.

REFERENCES

1. *Fink JK. Hereditary spastic paraplegia: clinico-pathologic features and emerging molecular mechanisms. Acta Neuropathol. 2013;126(3):307-328.*
2. *Ebrahimi-Fakhari D, Behne R, Davies AK, et al. AP-4-Associated Hereditary Spastic Paraplegia. GeneReviews. 2018 Dec 13.*

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