SPASTIC LEGS, SLOWED MIND, AND THE LYNX SIGN: A DIAGNOSTIC TRIO IN SPG11

DR. A. SANTHOSH KUMAR, DR. MARIAN JUDE VIJAY, DR. VIGNESH ANBAZHAGAN,
DR. N. SHANMUGA SUNDARAM, DR. K. MUGUNDHAN,
INSTITUTE OF NEUROLOGY, MADRAS MEDICAL COLLEGE, CHENNAI

BACKGROUND & AIMS : Hereditary spastic paraplegia (HSP) refers to a diverse group of inherited neurodegenerative disorders marked by progressive lower limb spasticity and weakness, primarily due to corticospinal tract involvement. Among its autosomal recessive forms, HSP type 11 (SPG11) is the most prevalent, often presenting with cognitive impairment and characteristic MRI findings such as a thin corpus callosum. We present a case of genetically confirmed SPG11 to highlight its diagnostic features and clinical course.

MATERIALS & METHODS : A case report of a 17-year-old male with spastic paraparesis, supported by clinical evaluation, neuroimaging and genetic testing.

RESULTS : A 17-year-old adolescent boy, with poor scholastic performance since childhood, presented with a 4-year history of progressive lower limb weakness, accompanied by stiffness. Patient achieved all the milestones during childhood but had learning difficulties in schooling. The family history was unremarkable except for consanquinity. Neurologic examination revealed impaired attention, grade-3 spasticity and muscle weakness of grade 4 in both lower limbs, together with other pyramidal signs, such

as hyperreflexia and Babinski sign. The upper limbs were spared. The patient presented typical spastic gait, with toe walking and bilateral ankle contractures. No muscle atrophy was observed. Cerebellar function was normal.

Magnetic resonance imaging (MRI) of the brain showed T2-weighted/fluid-attenuated inversion recovery (FLAIR) periventricular white matter hyperintensities at the frontal horns of the lateral ventricles, so called “ears of the lynx” sign and the corpus callosum was visibly thinner in its genu, body and splenium. The cervical and thoracic spine MRI presented no abnormalities. Nerve conduction study was normal. The ophthalmic exam excluded any pathologies in the eyes. There were no significant changes in the laboratory tests. Neuropsychological assessment showed low IQ of 55 suggestive of mild intellectual disability. Genetic analysis by whole-exome sequencing with the next-generation sequencing (NGS) method was performed, indicating compound heterozygous variations in the patient’s SPG11 gene.

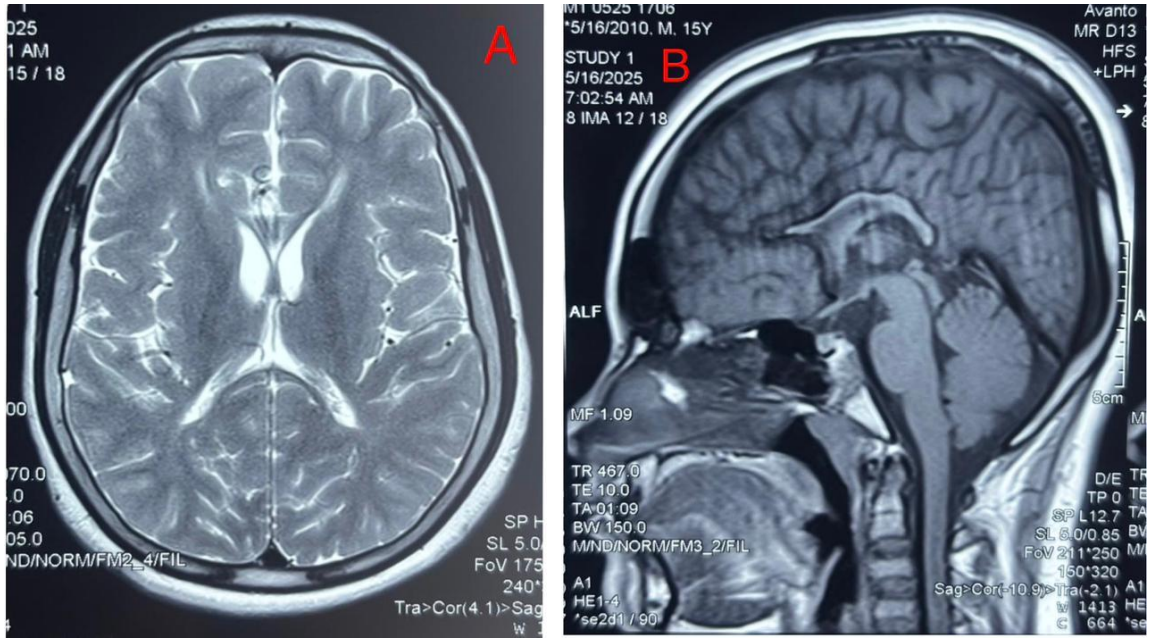

FIGURE.

A: Axial T2-weighted MRI image of the brain showing linear hyperintensity involving bilateral forceps minor and major.

B: Sagittal T1-weighted MRI image showing diffuse, severe thinning of the corpus callosum involving the genu, body, and splenium.

DISCUSSION : Hereditary spastic paraplegia type 11 is the most common autosomal recessive type of HSP, and accounts for up to 8% of all cases. The cardinal symptoms of HSP 11 include slowly progressive spastic paraparesis with sphincter disturbances. Additional clinical features are intellectual disability with learning difficulties in childhood and/or progressive cognitive retardation, peripheral neuropathy (axonal, motor or sensorimotor), cerebellar signs, parkinsonism and pseudobulbar involvement.

The clinical presentation in our patient is characteristic of SPG11-associated hereditary spastic paraplegia (SPG11-HSP). Cognitive impairment occurs in more than 90% of affected individuals, with a mean IQ of approximately 70.

Thinning of the corpus callosum (TCC) represents a highly specific MRI hallmark, observed in nearly all reported cases. White matter signal abnormalities are also common and are believed to reflect neuroaxonal degeneration secondary to impaired lysosomal and lipid metabolism.

The presence of the “ears of the lynx” sign in combination with corpus callosum thinning, although not pathognomonic, strongly supports SPG11 when observed alongside compatible clinical findings. Early genetic testing can facilitate timely diagnosis and guide prognosis, surveillance, and genetic counseling.

CONCLUSION : This case highlights the importance of integrating clinical, radiological, and genetic data for the accurate diagnosis of SPG11. Whole-exome sequencing is an essential diagnostic tool in suspected hereditary spastic paraplegias.