Hereditary Spastic Paraplegia: Role of MRI

Sir,

Hereditary spastic paraplegia (HSP), also known as Strumpell-Lorain syndrome, is a group of hereditary disorders characterised by progressive lower limb weakness, spasticity and exaggerated reflexes.[1] HSP can clinically present as "uncomplicated or pure," showing pyramidal signs leading to lower-limb spasticity, and as "complicated" when associated with ataxia, dementia, extrapyramidal dysfunctions, mental retardation, visual or hearing impairment, adrenal insufficiency, and ichthyosis.[2]

However, currently classifications are based on genetic linkage and mode of inheritance. Hence HSP can be X-linked, autosomal recessive or autosomal dominant. About 70% of HSP cases demonstrate dominant inheritance, while 20% show a recessive pattern.[3] HSP is rare with a global prevalence of 1.8:100000 for autosomal recessive and dominant types.[4] To the best of our knowledge, there is no evidence of any reported case in Pakistan evaluating radiological patterns of HSP.

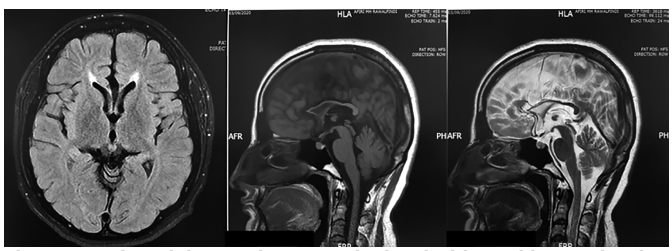

Figure 1: Left: Axial FLAIR image at the level of frontal horns showing periventricular hyper intensities giving ear of the Lynx sign. Centre and Right: Sagittal T1W and T2W images showing marked thinning of the genu and body of corpus callosum.

A 24-year young woman presented with gradual onset progressive asymmetric weakness in both legs for 3 years associated with stiffness in her legs. The patient could walk with support but required help for activities of daily living. On examination, she had upper motor neuron signs in both legs. MRI brain was performed for diagnostic evaluation. Plain sagittal T1/T2W images revealed marked thinning of the genu and anterior part of body of corpus callosum, while rostrum and splenium were unremarkable. Periventricular cap like hyperintensities were noted on axial T2W/FLAIR sequences along frontal horns of both lateral ventricles showing classical ear-of-the-lynx sign (Figure 1). Persistent cavum septum pellucidum and cavum vergae were also seen (normal anatomical variants). MRI of cervicodorsal spine was also done, which was normal. Genetic testing was not done as facility was not available. The patient was started on multi-vitamins and pregabalin, improved clinically and was followed up in outpatient department.

Correct diagnosis of HSP is important because patients can develop long term complications like muscle contractures, back-/knee pain, stress and depression with significant effects on quality of life. Genetic testing for confirmation of specific gene mutation requires better laboratory facilities, is expensive and not routinely available in our region. MRI can significantly aid in diagnosis by showing features like mild brain/cerebellar and spinal atrophic changes predominantly in motor areas and pericentral gyri, T2/FLAIR hyperintensity in posterior limb of internal capsule, unspecific white matter lesions and thinning of corpus callosum.[3] Ears-of-the-lynx sign on MRI can be highly specific for most common genetic subtypes of HSP with a thin corpus callosum (HSP-TCC). When this sign is seen, there is a high chance of genetic mutation more likely associated with SPG11 or SPG15, even if there is no family history.[5]

A delayed diagnosis of HSP may lead to complications with reduced quality of life. MRI can play a significant role in identifying this rare group of genetic disorders early and improving patient's clinical outcome as imaging features are fairly specific.

CONFLICT OF INTEREST:
The authors declared no conflict of interest.

AUTHORS' CONTRIBUTION:
ARP: Conception or design of the work, acquisition, analysis, interpretation of work, radiological diagnosis and drafting the work.
MS: Radiological diagnosis, drafting the work and revising it critically for important intellectual content; and final approval of the version to be published.
KH: Initial clinical diagnosis, drafting the work and revising it critically for important intellectual content; and final approval of the version to be published.

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