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Case Report

Unraveling HSP: A Magnetic Resonance Imaging Case Report

Niveditha Srinivasan[1], Rajoo Ramachandran[2], Harini Gnanavel[2], R Rajeswaran[3]

[1]Final Year Resident, Department of Radiology and Imaging Sciences, Sri Ramachandra Medical College and Research Institue, Chennai, Tamil Nadu, India, [2]Professor, Department of Radiology and Imaging Sciences, Sri Ramachandra Medical College and Research Institue, Chennai, Tamil Nadu, India, [3]Head of Department and Professor, Department of Radiology and Imaging Sciences, Sri Ramachandra Medical College and Research Institue, Chennai, Tamil Nadu, India

Abstract

Background: Hereditary spastic paraplegia (HSP) presents a clinical challenge due to its varied phenotype and genotype. Magnetic resonance imaging (MRI) plays a crucial role in diagnosis, aiding in distinguishing it from other similar conditions.

Clinical Description: We report a case of a 21-year-old female presenting with progressive gait difficulties and speech impairment over 11 years, with typical features of HSP. Clinical examination revealed lower limb spasticity and normal muscle bulk.

Imaging Findings: MRI demonstrated diffuse thinning of the corpus callosum, cone-shaped hyperintensities adjacent to the frontal horns of the lateral ventricles, and mild cervical spinal cord atrophy. These findings align with established HSP imaging characteristics, corroborating clinical suspicion.

Conclusion: MRI serves as a valuable tool in diagnosing HSP, showcasing distinctive features such as corpus callosum thinning and spinal cord atrophy. Recognition of these imaging hallmarks aids in early diagnosis and facilitates genetic testing, enabling tailored management strategies for affected individuals.

Key words: Hereditary spastic paraplegia, corticospinal tract, posterior column of spinal cord.

INTRODUCTION

Hereditary spastic paraplegia (HSP) is a group of genetic disorders characterized by progressive weakness and stiffness (spasticity) in the lower limbs. It results from degeneration of the motor neurons in the spinal cord, leading to difficulty with walking and balance. HSP varies in severity, age of onset, and symptoms depending on the specific gene involved. While some forms of HSP are isolated to lower limb symptoms, others can affect additional systems, leading to more complex forms of the disease. Radiological diagnosis plays a crucial role in evaluating the extent of neurodegeneration and ruling out other potential causes of spasticity. Magnetic resonance imaging (MRI) is the imaging modality of choice for HSP, focusing on the brain and spinal cord. Advanced MRI techniques, such as diffusion tensor imaging (DTI), can further assess microstructural changes in the corticospinal tracts. These radiological features, combined with clinical and genetic evaluations, help in confirming the diagnosis of HSP. There is currently no cure, and treatment focuses on managing symptoms and improving quality of life.

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CASE REPORT

Clinical History

A 21-year-old female with a normal developmental history who was born into non-consanguineous parenthood and with no other comorbidities complained of speech difficulties and a gradual beginning of progressive gait problems that had been present for 11 years. There was no record of prior complaints of a similar nature in the parents or first- or second-degree relatives.

Corresponding Author: Dr. Rajoo Ramachandran, Department of Radiology and Imaging Sciences, Sri Ramachandra Medical College and Research Institue, Chennai, Tamil Nadu, India.

On Examination

Muscle bulk was normal with spasticity of bilateral lower limbs.

The power in the lower limbs was normal.

Sensory system examination was normal.

No abnormalities were detected on hematological investigations.

For further investigation, the patient was subjected to MRI:

3 T MRI Brain (Figure 1a) in sagittal T1 W image shows diffuse thinning of the rostrum, genu, and anterior two-thirds of the body of corpus callosum;

Axial T2 W sequence (Figure 1b) of the brain show symmetrical cone-shaped hyperintensities adjacent to the frontal horn of bilateral lateral ventricles.

Sagittal T2 W cervical spine (Figure 2) shows mild thinning of the cervical spinal cord suggestive of spinal cord atrophy.

DISCUSSION

With approximately 50 gene products and roughly 71 different loci, hereditary spastic paraplegia, also referred to as Strümpell-Lorrain syndrome, is categorized under inherited Mendelian conditions. Its distinctive characteristic is the gradual spasticity and weakening of the lower limbs. All forms of HSP have an estimated prevalence that varies from 1:100,000 to 10:100,000.

Degeneration of the corticospinal tract resulting in damage to the pyramidal tract is the characteristic clinical manifestation. Hereditary spastic paraplegias have been divided into two categories: pure HSP (pHSP) and complicated HSP (cHSP), based on the clinical phenotype and the Harding criteria. The pyramidal motor system is the only one affected by pHSP symptoms. Conversely, various neurologic domains, including peripheral nerve systems and the cognitive, cerebellar, or extrapyramidal domains, may be involved in cHSP.

The type of inheritance (autosomal recessive or X-linked) has recently been used to categorize hereditary spastic paraplegias. The specific chromosomal gene or locus causing the paralysis is designated as "SPG" (spastic gait) and is followed by a progressive number.

Around 25% are asymptomatic and hence careful clinical evaluation of families is included in genetic studies. If symptomatic the most common presenting symptom is gait disturbance with other symptoms, such as stiffness of legs and urinary disturbance. Since the age at which the disease manifests with symptoms can vary from birth to 80 years, there is genetic heterogeneity and thus there is difficulty in ascertaining an exact onset time. Upon clinical examination, the primary abnormalities include spasticity (mainly in the hamstrings, quadriceps, and ankles) that causes a classic gait disturbance that shows walking on tiptoe and circumduction, hyperreflexia and extensor plantar responses and weakness in a pyramidal distribution of the lower limbs.[1] One of the main characteristics of HSP is the noticeable difference between mild muscle weakness and severe spasticity. One or two decades later, the majority of those affected become debilitated.

Clinical diagnosis of HSP is challenging since spastic paraparesis can be caused by a long list of conditions. Hence possible acquired and genetic causes of spastic paraparesis, need to be ruled out with laboratory tests and neuroimaging studies. On Diffusion tensor (DT) MR imaging, patients with pHSP and cHSP show severe damage to the motor white matter, including the body of the corpus callosum, primary motor and pre-motor cortices, and cerebral peduncles.[2] Harding described the clinical criteria for HSP diagnosis, but the genetic analysis is the gold standard confirmatory test.[3]

Nonetheless, MRI is a helpful tool for early diagnosis and rule out other conditions that present with a similar clinical picture, such as infections like neurosyphilis and HIV, leukodystrophies, compressive neuropathies, motor neuron diseases, such as amyotrophic lateral sclerosis, spinocerebellar ataxias, and inherited or acquired metabolic disorders.[3]

The MRI findings of HSP include hyperintense T2 signal of the posterior limb of the internal capsule and mild brain and spinal cord (most affected being the cervical sections of the cord) atrophy. Other findings, such as widening of interhemispheric fissure, medial frontal lobe atrophy, and reduced thalamus size can also be seen. It is rare to get gray matter involvement in pure HSP, but basal ganglia can be involved in complicated cases, which leads to heterogeneous phenotype.[4]

On MR imaging SPG11, SPG 15 presents with the classic imaging feature of "ear-of-the-lynx sign". It is seen as hyperintensity on T2-FLAIR-weighted and hypointensity on T1-weighted images at the forceps minor of the corpus callosum. Usually, there is associated corpus callosal thinning as presented in our case. The sensitivity and specificity are as high as 94 and 97%, respectively.[5] This feature is highly suggestive of the disease even in the absence of significant clinical features.

Other MRI features include a thinning of the corpus callosum, enlargement of the lateral ventricles, and symmetrical white matter lesions. HSP complicated by corpus callosum thinning is characterized by features, such as progressive spastic paraplegia and dementia developing at an earlier age along and other atypical symptoms, such as extra-pyramidal symptoms, seizures, spastic quadriparesis, atrophy of the muscles, sensory deficits and cerebellar ataxia.[6]

Single-voxel proton MR spectroscopy in patients of HSP shows decreased levels of *N*-acetylaspartate (NAA), choline (Cho), and creatine (Cr), with increased levels of myo-inositol.[7]

N-acetylaspartate is a marker of neurons and thus its decrease denotes neuroaxonal tissue loss. Choline being a marker of membrane synthesis and myelin turnover, if reduced, denotes altered myelination. Creatine is a marker of metabolism and is reduced in areas of cell death. Astrocytic proliferation is denoted by Myo-inositol levels. These metabolic changes may occur in HSP and indicate progressive neuroaxonal loss and astrocytic proliferation.[7]

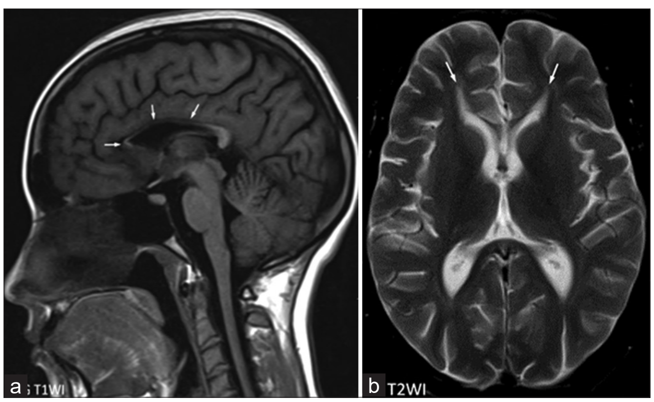

Figure 1: 3 T MRI Brain: (a) Sagittal T1 W image shows diffuse thinning of the rostrum, genu, and anterior two-thirds of the body of the corpus callosum. (b) Axial T2-W section of the brain shows symmetrical cone-shaped hyperintensities adjacent to the frontal horn of bilateral lateral ventricles

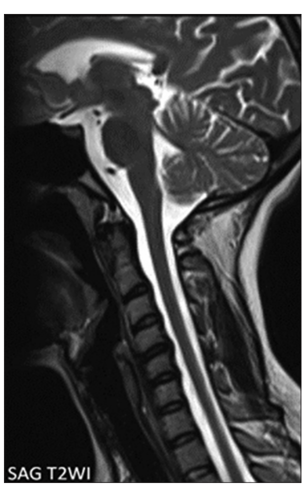

Figure 2: Sagittal T2-W image of the cervical spine shows mild thinning of the cervical spinal cord suggestive of spinal cord atrophy

In the periventricular white matter, DTI shows higher mean diffusivity with lower fractional anisotropy, which is consistent with myelinated axonal injury. Reduced blood flow and hypometabolism in the thalamus and fronto-temporo-parietal cortex can be seen using SPECT and FDG-PET.[7]

Though the prognosis is different between families, life expectancy is normal in hereditary spastic paraplegia. It has been concluded from various studies that in early-onset HSP (<35 years), there is slow disease progression with most patients being ambulant, except a few who develop motor weakness in later stages of life. However, in late-onset HSP (>35 years), the patients can have rapid disease progression and can get confined to a wheelchair in the late 70s.[1]

CONCLUSION

HSP is a heterogenous group of neurodegenerative disorders with variable and non-specific imaging findings. The ear of the lynx sign, atrophy of the corpus callosum, high T2 signal intensity involving the posterior limb of the internal capsule, and spinal cord atrophy are among the reliable and firmly established radiological findings. These imaging findings may vary based on the stage of the disease and the different phenotypes identified on genetic screening. Hence the clinician should be aware of the varied imaging features of HSP which aides in early diagnosis, a clue to the involved gene and hence deciding on further course of management.

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