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# A Rare Case of Hereditary Spastic Paraplegia: Case Report and Literature Review

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**Abstract** **Case Report**

Hereditary spastic paraplegia (HSP) is a rare neurodegenerative disorder presenting as lower limb spasticity having two clinical types and various modes of inheritance. A young male patient presented with bilateral lower limb weakness of one-year duration showing both sensory and motor abnormalities on examination. MRI of brain and spine done as part of workup revealed marked atrophy of anterior corpus callosum with abnormal signal intensity at tips of frontal horns of ventricles mimicking the appearance of the ear of a Eurasian lynx. No other abnormality was encountered in the brain or spinal cord in our case. Other imaging abnormalities which can be encountered in such a case include periventricular and central white matter changes as well as spinal cord atrophy. Pathologically this condition is recognised by degeneration of lateral and posterior columns of the spinal cord. The differential diagnosis includes structural, metabolic, neurodegenerative, motor and nutritional deficiency disorders.

**Keywords:** Hereditary Spastic Paraplegia, Ears of lynx, Thinning of the corpus callosum.

**Key messages:** Ears of lynx sign has several differentials and require corroborative clinical assessment. The presence of this sign in a patient having spastic paraplegia is diagnostic of Hereditary Spastic Paraplegia



## INTRODUCTION

Hereditary spastic paraplegia (HSP) is a rare group of familial neurodegenerative disorders characterized by progressive lower limb spasticity. Clinically of two types are "pure" when spastic paraplegia exists in isolation and "complicated" when other major clinical features such as mental retardation, epilepsy, optic atrophy, extrapyramidal symptoms, deafness, cerebellar signs, muscle wasting and ichthyosis are present. Genetically autosomal dominant, autosomal recessive, and X-linked recessive forms of inheritance are seen with both types with the latter being the most frequent mode of inheritance [Priya S *et al.*, 2016] [Coutinho P *et al*., 1999]. Genetic locus is commonly linked to chromosome 15q13-15[SPG11gene] which accounts for 41–77% of reported HSP [Coutinho P *et al*., 1999].

## CASE REPORT

A 26 years old male who, presented with insidious onset of bilateral lower limb weakness and spasticity for the last 1 year. The patient had suboptimal IQ in the range of mild mental retardation. No history of similar illness in the family. Examination findings included bilateral lower limb spasticity, bilateral extensor plantar reflex, and brisk reflexes of bilateral lower limbs in the motor examination. The sensory system revealed no bladder or bowel involvement with intact touch, pain and temperature sensations as well as proprioception. The rest of the neurologic examination was within the normal limits. MRI of the brain and spinal cord was performed as part of the imaging workup. The study revealed marked thinning of the anterior part of the corpus callosum depicting atrophy. In addition, there were triangular shape areas of altered signal intensity adjoining tips of frontal horns of lateral ventricles appearing hypointense on T1 and hyperintense on T2 and FLAIR sequences (Fig-1).

In the present case, the patient first visited our institute in March 2021 and is one supportive treatment for muscle spasms. The patient is adherent to treatment with no side effects to medications. The disease is showing a progressive worsening without any stabilization since then till the last patient visit in July 2021.

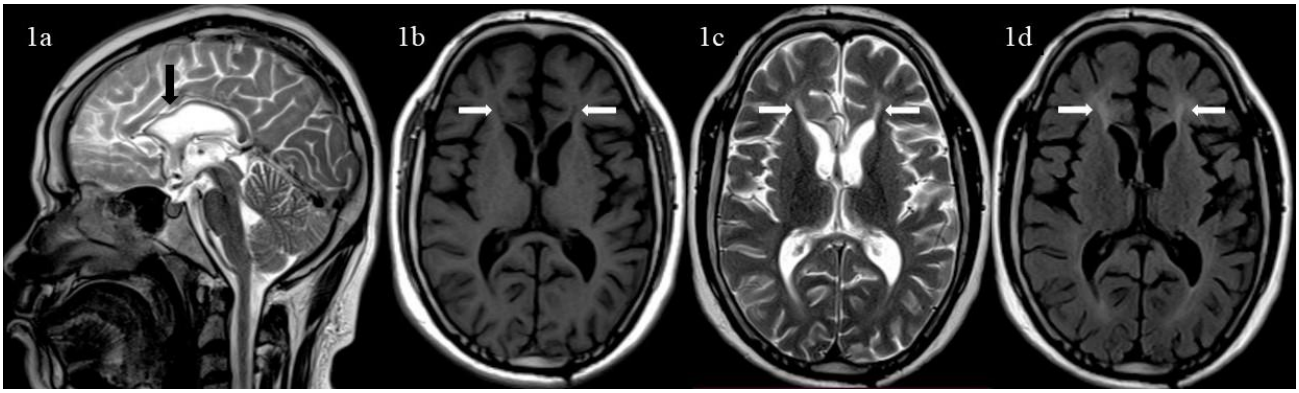


**Fig-1: T2 weighted sagittal MRI shows marked thinning of the corpus callosum (1a). T1 weighted axial MRI shows the hypointense signal at the tips of the frontal horns of the lateral ventricles having a triangular shape (1b), T2 (1c) and FLAIR (1d) axial MRI image at same level shows corresponding hyperintense signal.**

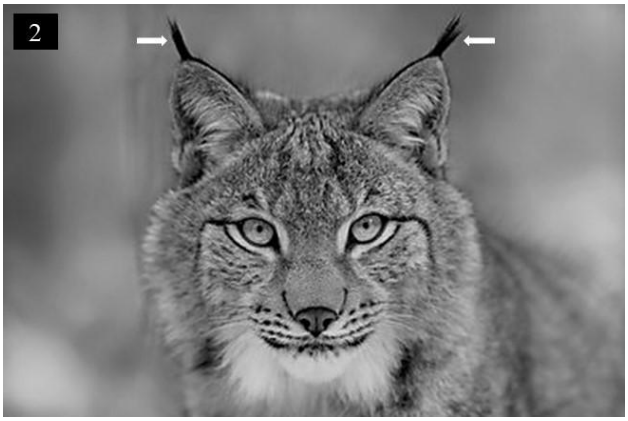


**Fig-2: Photograph of Eurasian lynx with a tuft of hairs at ear tips**

## DISCUSSION

HSP is a group of neurodegenerative disorders, genetically heterogeneous and progressive with time, characterized by multisystem degeneration. Patients show a spectrum of imaging abnormalities seen on MRI [3]. MRI may show nonspecific changes in many cases, of which the most frequently encountered are the brain and spinal cord atrophy along with white matter abnormalities in centrum semiovale and corona radiata [Reid E, 1999] [Okubo S 2000]. The Imaging findings vary among different phenotypes and in different stages of the disease, may even look normal altogether in many cases. Furthermore, the appearance is affected by a variable degree of axonal degeneration due to different degrees of genetic penetrance among families. Among the specific changes seen in a case of HSP include atrophy of corpus callosum, T2/FLAIR hyperintense signal in the posterior limb of the internal capsule and T2/FLAIR hyperintense signal in forceps minor of the corpus callosum. The latter named "Ear of Lynx Sign" (Fig-2) is mostly associated with a mutation in spatascin vesicle trafficking associated gene, leading to Spastic paraplegia type 11 (SPG 11). Our case turned out to be SPG 11 and demonstrated the same-named sign on MRI.

Grey matter changes are unlikely in pure HSP. In the complicated form, there can be a presence of grey matter atrophy with particular involvement of basal ganglia, which explains the phenotypic heterogeneity that frequently includes Parkinsonism and dystonia [Rezende TJ *et al.*, 2015]. These findings in isolation are not useful in diagnosis but when present along with spinal cord changes are highly suggestive of HSP. Additional reported findings include bilateral medial frontal atrophy, widening of interhemispheric fissure, and thalamic atrophy. Krabbe *et al*. reported significant atrophy of the upper spinal cord, especially D3 and D9 levels, in patients with HSP compared with the controls, which corresponded neuropathologically to degeneration of the lateral corticospinal tracts, uncrossed pyramidal tracts, and fasciculus gracilis (posterior columns of the spinal cord) from the lumbar level up to the upper cervical level [Krabbe K *et al*., 1997]. The neuroradiologic findings are not constant and vary among the different phenotypes and stages of HSP. As these are a rare group of disorders one must go through the proper history of the patient and keep in mind it as a differential. One needs to be familiar with some characteristic findings like – ear of lynx sign in axial plane MRI and thinning of the corpus callosum. More studies with multiple MRI parameters, increased sample size, and a spectrum of phenotypes will be needed for better understanding and defining radiological findings of this morbid disease.

The differential diagnosis of HSP is broad and includes the following disease categories:

- Motor neuron disease, particularly slowly progressive amyotrophic lateral sclerosis or primary lateral sclerosis. Amyotrophic lateral

sclerosis can mimic HSP when it affects the legs without significant amyotrophy or fasciculations.

- Structural abnormalities of the spinal cord such as tethered spinal cord syndrome and spinal cord compression.
- Leukodystrophies and demyelinating disorders, such as Progressive multiple sclerosis, Adrenomyeloneuropathy, Krabbe and Pelizaeus-Merzbacher disease and Metachromatic leukodystrophy.
- Neurologic impairments due to nutritional and metabolic deficiencies like Vitamin B12 and Copper deficiency, Methylenetetrahydrofolate reductase and Cobalamin C deficiency.
- Vascular malformations, most notably spinal dural arteriovenous fistulas, typically present after the fifth decade of life with progressive or, less often, fluctuating symptoms including weakness, sensory disturbances, gait abnormalities, sphincter dysfunction, and pain, sometimes exacerbated by exercise.
- Dopa-responsive dystonia, which typically begins in early childhood and is treatable with levodopa in relatively low doses.
- Hereditary ataxias with significant spasticity; can present with an HSP-like onset if the ataxia is confined to the spastic lower extremities. Examples include certain spinocerebellar ataxias, Friedreich ataxia.

There is no curative treatment for HSP and management lies in symptomatic treatment for motor and sensory changes and maintenance of posture and joint mobility. Pharmacological treatment includes drugs like baclofen and tizanidine and benzodiazepines for muscle spasms and anticholinergics for urinary bladder overactivity and incontinence. Prenatal genetic testing is possible for certain forms of HSP if the disease-causing mutation has been identified in an affected family member [Hedera P, 2000].

The pure form of HSP does not affect the lifespan of affected patients, although there can be a deleterious impact on the quality of life. Three types of progression have been described in the available literature a nonprogressive course, progressive worsening that stabilizes over time and inexorable decline.

**List of Abbreviations**

| Abbreviation | Definition |
|---|---|
| HSP | Hereditary spastic paraplegia |
| MRI | Magnetic Resonance Imaging |
| CC | Corpus Callosum |
| FLAIR | Fluid Attenuated Inversion Recovery |
| SPG | Spastic Paraplegia |

## ACKNOWLEDGEMENTS

Source of Image-2: Hulik T. Eurasian Lynx. Available from: https://www.shutterstock.com/ image-photo/eurasian-lynx-705339076

## REFERENCES

1. Priya, S., Siddique, N., Das, R., & Singh, A. (2016). Multiparametric 3T MRI evaluation of hereditary spastic paraplegia: A case report. *The Indian journal of radiology & imaging*, *26*(3), 328.
2. Coutinho, P., Barros, J., Zemmouri, R., Guimarães, J., Alves, C., Chorão, R., ... & Prud'homme, J. F. (1999). Clinical heterogeneity of autosomal recessive spastic paraplegias: analysis of 106 patients in 46 families. *Archives of neurology*, *56*(8), 943-949.
3. Hourani, R., El-Hajj, T., Barada, W. H., Hourani, M., & Yamout, B. I. (2009). MR imaging findings in autosomal recessive hereditary spastic paraplegia. *American journal of neuroradiology*, *30*(5), 936-940.
4. Reid, E. (1999). The hereditary spastic paraplegias. *J Neurol*, *246*(11); 995-1003.
5. Okubo, S., Ueda, M., Kamiya, T., Mizumura, S., Terashi, A., & Katayama, Y. (2000). Neurological and neuroradiological progression in hereditary spastic paraplegia with a thin corpus callosum. *Acta neurologica scandinavica*, *102*(3), 196-199.
6. Rezende, T. J., de Albuquerque, M., Lamas, G. M., Martinez, A. R., Campos, B. M., Casseb, R. F., & França Jr, M. C. (2015). Multimodal MRI-based study in patients with SPG4 mutations. *PLoS One*, *10*(2), e0117666.
7. Krabbe, K., Nielsen, J. E., Fallentin, E., Fenger, K., & Herning, M. (1997). MRI of autosomal dominant pure spastic paraplegia. *Neuroradiology*, *39*(10), 724-727.
8. Hedera P. Hereditary Spastic Paraplegia Overview. 2000 Aug 15 [Updated 2021 Feb 11]. In: Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2021..