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KARNATAKA RADIOLOGY EDUCATION PROGRAM

CASE PRESENTATION

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HEREDITARY SPASTIC PARAPLEGIA

INTRODUCTION

- Hereditary spastic paraplegias (HSP) are a large group of genetic diseases characterized by progressive degeneration of the long tracts of the spinal cord, namely the corticospinal tracts and dorsal columns.
- Patients typically present with lower limb-predominant spasticity and weakness leading to gait abnormalities.
- Sensory deficits and urinary complaints are also frequently found
- In the realm of neurogenetics, HSP is perhaps the condition with the most striking genetic heterogeneity. It may segregate as an autosomal dominant, autosomal recessive or X-linked trait.
- There are now around 70 loci and 60 genes associated to different forms of HSP For the practicing clinician, this represents a diagnostic challenge and poses difficulty for proper therapeutic management as well as genetic counseling.

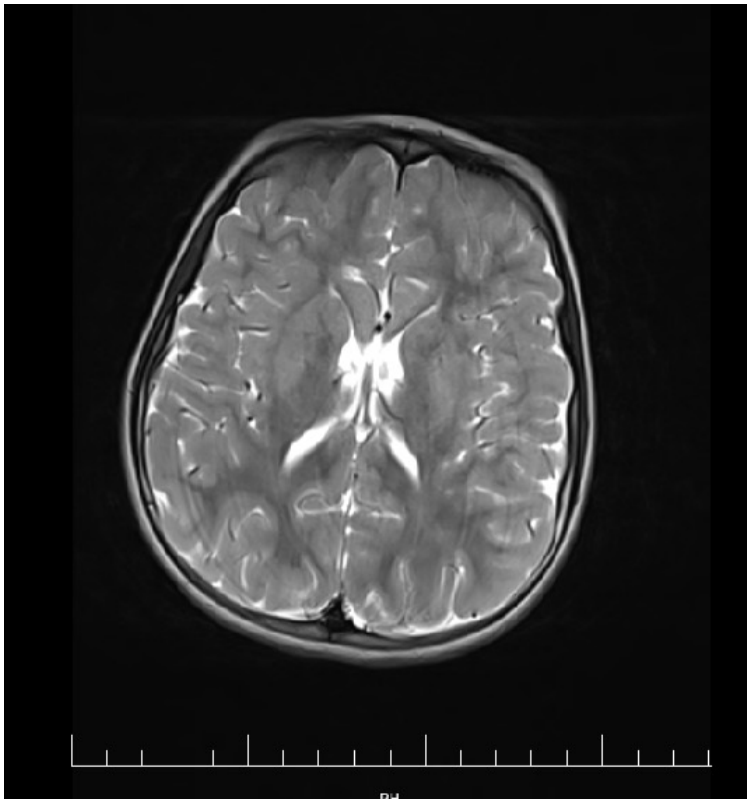
INTRODUCTION

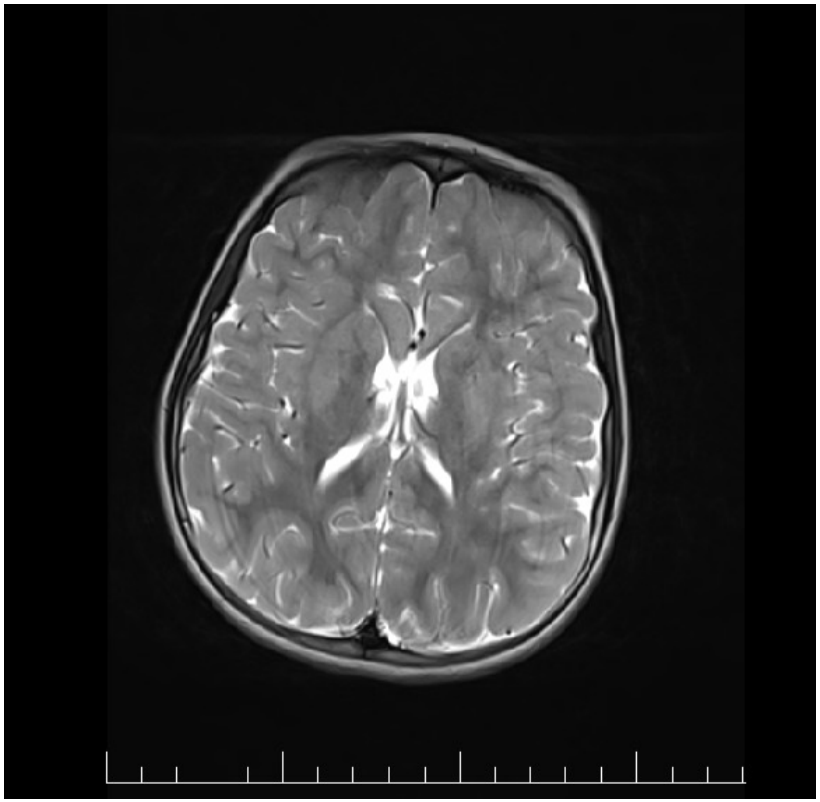
- Neuroimaging is a powerful tool that enables the structural and functional assessment of the central nervous system (CNS).
- In particular, advanced techniques of magnetic resonance imaging (MRI) are able to provide detailed microstructural and biochemical information of the CNS; they have proven useful to uncover abnormalities in closely related heredodegenerative diseases, such as spinocerebellar ataxias and amyotrophic lateral sclerosis
- In the context of HSPs, MRI may assist in the phenotypic characterization and therefore help in the genetic testing approach .
- More recently, some studies using advanced and quantitative techniques have shown microstructural white matter abnormalities in HSP, not detectable with routine MRI sequences
- These results brought novel insights into the pathophysiology of HSPs and raised the possibility of using MRI as a biomarker to track disease progression. In this review article, we will discuss the available data concerning neuroimaging in HSP.

- The precise diagnosis of HSPs, considering the great genetic variability with often similar phenotypes, is challenging.
- However, as with any neurological disease, a combination of detailed history, assessment of inheritance pattern, accompanying symptoms and physical examination is paramount to reduce the number of hypotheses and to optimize genetic testing
- In patients with spastic paraparesis, MRI is essential, initially to rule out usual causes of paraplegia such as compressive, inflammatory, infectious, or vascular myelopathies.
- Once the diagnosis of HSP is the most likely, neuroimaging may help in establishing the subtype.

CASE PRESENTATION

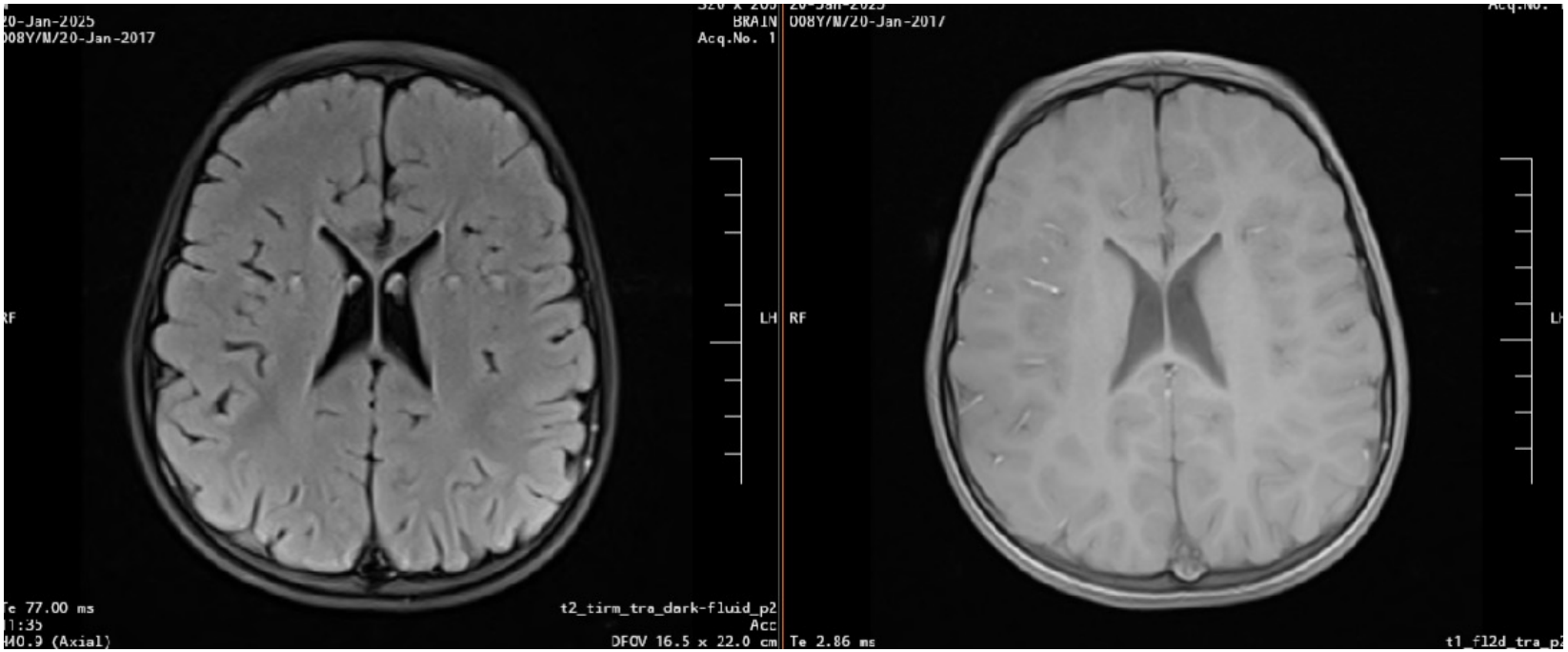
- A 8yr old male patient came with the complaints of difficulty in walking and weakness of bilateral lower limbs
- No history of trauma
- Birth and perinatal history – normal (FTVD)
- Patient was advised MRI of BRAIN (plain)





Axial T2W image shows high signal intensity extending a short distance from the anterior horns of the lateral ventricles,

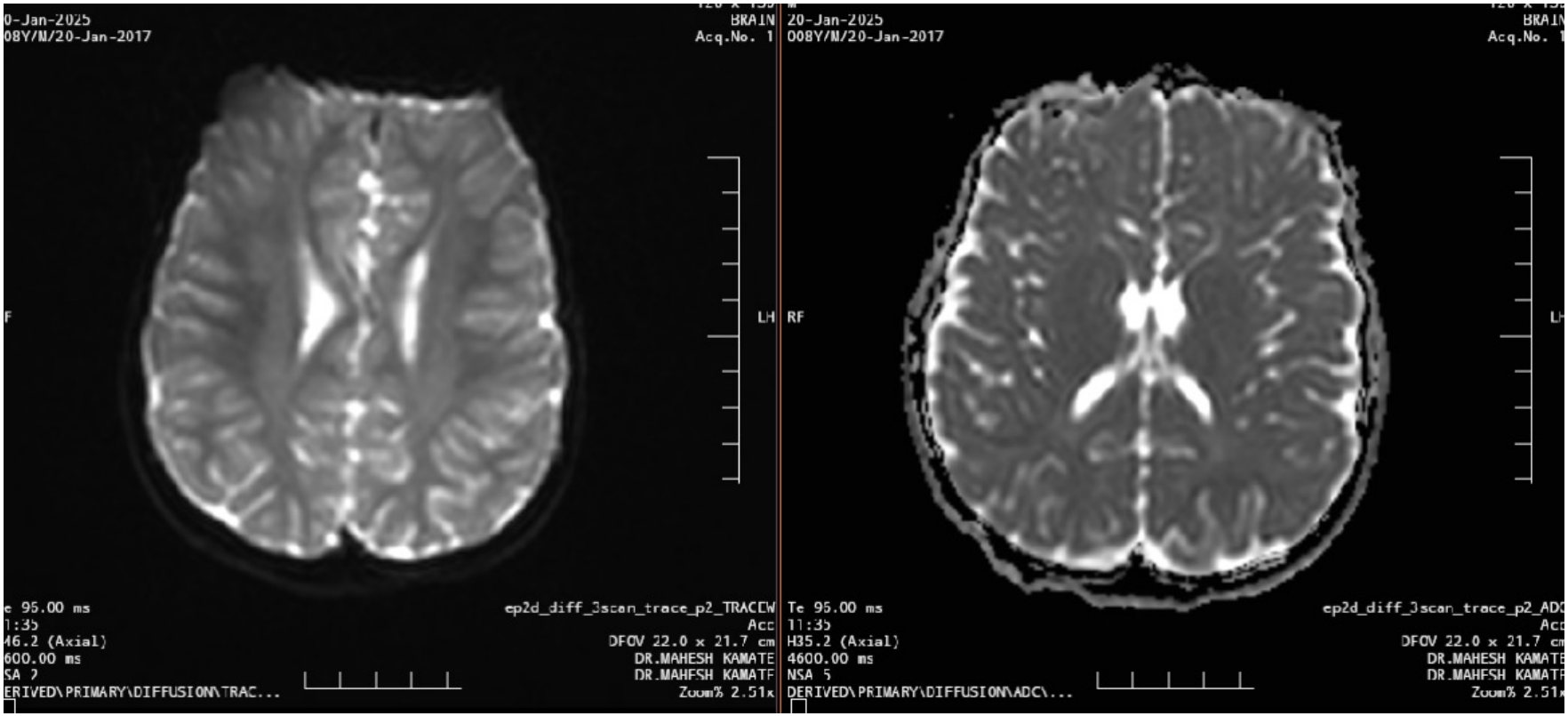
This appearance is the 'ears of the lynx' sign, named for the tufts of hair characteristically seen on the ears of these animals.



Axial FLAIR & T1W images showing with corresponding high & low signal intensities on the FLAIR & T1 weighted images

Thinning of corpus callosum noted.

Thinning of the corpus callosum is seen on the midline sagittal T1 & T2 weighted images.



DWI & ADC IMAGES SHOWING NO EVIDENCE OF DIFFUSION RESTRICTION.

- The ear of lynx sign has been described in patients with hereditary spastic paraplegia with thin corpus callosum (HSP-TCC) caused by mutations of the spatacin vesicle trafficking associated (SPG11) gene, coding spatacin.
- The sign may also be seen in SPG15, which is caused by a mutation in a different gene (zinc finger fyve domain-containing protein 26, ZFYVE26), encoding spasticin