GENETIC,PHENOTYPIC AND RADIOLOGICAL DIVERSITY IN HEREDITARY SPASTIC PRAPARESIS

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AIM

- To describe a case series of patients with HSP, evaluating their clinical features, family history, radiological findings and genetic testing results.
- To identify disease spectrum and address diagnostic challenges.

METHODOLOGY

- A retrospective analysis of patients having HSP phenotype with genetically proven mutation at a tertiary care centre in western India.

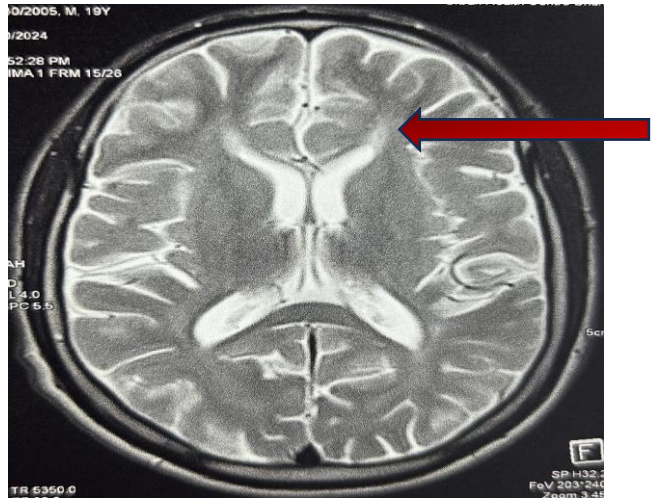
RESULTS

- Total 7 patients with spastic paraparesis with or without additional neurological or non-neurological features.

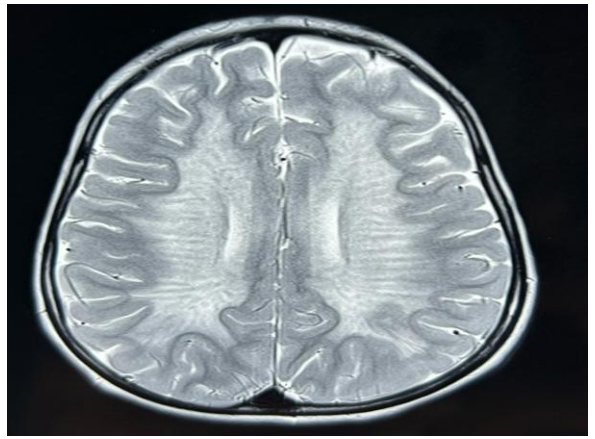
- Most common inheritance pattern - autosomal recessive
- Mutations -SPG 11, KIF5A, SPAST, SPG7, ZYFEV26 (SPG15), NFU1.

- Two patients – abnormal MRI Brain,
- SPG11 mutation classic **"EAR OF LYNX"** sign
- SPG15 mutation – **"TIGROID Pattern"**

EAR OF LYNX



TIGROID PATTERN



RESULTS

MUTATION	ADDITIONAL CLINICAL FEATURE
KIF5A	HAMMER TOES , HALLUX VALGUS
ZYFVE26	COGNITIVE IMPAIRMENT
NFU1	CONGENITAL NYSTAGMUS

MUATATION	MRI FINDING
SPG -7	DORSAL CORD THINNING
ZYFVE26	TIGROID PATTERN
SPG-11	EAR OF LYNX

DISCUSSION

- Our study had 50 % patients with autosomal recessive inheritance, in contrast to autosomal dominant as per literature.
- Average age of presentation was 13 yr in our study.
- Unique finding was“ Tigroid pattern” on MRI brain with SPG15 mutation, not been reported earlier in literature.
- Tigroid pattern in other conditions e.g. metachromatic leukodystrophy, Krabbe disease, Pelizaeus-Merzbacher disease, ARSACS.

CONCLUSION

- HSP is common disease even in adolescent age group.
- Imaging and additional clinical features can aid in diagnosis of HSP.