SPECTRUM OF GENETICALLY CONFIRMED SPINOCEREBELLAR ATAXIA: A ONE-YEAR OBSERVATIONAL STUDY AT A TERTIARY CARE CENTER IN EASTERN INDIA

AIM

- This study aims to document the confirmed cases of SCA diagnosed at Tertiary care centre, AIIMS Patna over a one-year period and describe their clinical features.

MATERIAL

- A observational study was conducted in the Department of Neurology at AIIMS, Patna, over a one-year period from April 2024 to March 2025.

METHOD

- Inclusion criteria required genetically confirmed SCA patients.
- Demographic data, clinical presentation, family history, and subtype classification were collected and analyzed descriptively.

RESULTS

- During the study period, 3 patients presented with progressive cerebellar ataxia. Cases were genetically confirmed as Spinocerebellar Ataxia.
- The first was a patient with SCA2, with a positive family history involving his father.
- The second case was a 44-year-old male diagnosed with SCA1, with positive family history. His younger sister; aged 40 yr was also having similar illness and also diagnosed with SCA 1 being a third case.
- Both subgroups of SCA type 1 and type 2 were presented with gradually worsening gait ataxia, dysarthria, and impaired coordination. Genetic analysis confirmed trinucleotide repeat expansions consistent with their respective subtypes.

DISCUSSION

- The low number of confirmed cases reflects the rarity of SCAs, but may also indicate underdiagnosis due to limited access to genetic testing or late referrals.
- The presence of both familial and apparently sporadic cases underlines the clinical variability of SCAs and the importance of genetic confirmation in ataxia diagnosis.

CONCLUSION

- Over one year at AIIMS Patna, three genetically confirmed cases of SCA (2 SCA1 and 1SCA2) were identified.
- Strengthening genetic diagnostic services and increasing clinical awareness are essential for improving early detection and management of SCAs.