Tracing the Neurogenetic Roots of Ataxia: A Genetically Confirmed Case of ARSACS in Siblings from India

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INTRODUCTION

ARSACS is a rare autosomal recessive ataxia caused by SACS gene mutations. Though well-documented globally, genetically confirmed Indian cases are rare.
We present two affected siblings with typical clinical and radiological features, with genetic confirmation in one case.

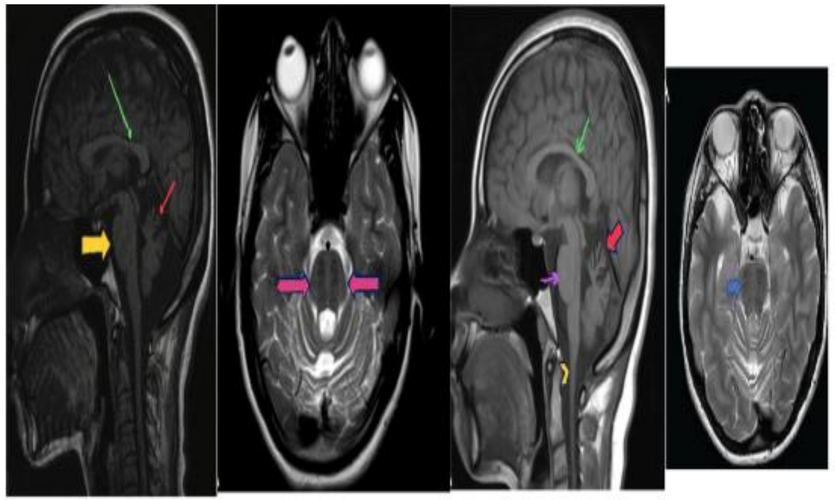
Cases

Case 1:A 29-year-old woman, born of third-degree consanguinity, presented with progressive gait unsteadiness, slurred speech, and lower limb spasticity since age 4. Exam showed distal limb wasting, spastic quadriparesis, sensory loss (vibration), cerebellar signs, and sensorineural hearing loss. NCS revealed motor-sensory axonopathy. MRI brain showed “striped pons” and superior vermian atroph

• . Genetic testing confirmed a likely pathogenic SACS gene deletion.

Case 2:

• Her 26-year-old sister had similar symptoms over 10 years—spastic ataxia, distal sensory neuropathy, and cerebellar signs without speech involvement. NCS showed axonal changes; MRI showed similar pontine striations and vermian atrophy. Genetic testing was declined. Based on clinical and radiological findings, ARSACS was considered.



DISCUSSION

ARSACS is a rare autosomal recessive disorder caused by SACS gene mutations, presenting with early-onset spasticity, ataxia, and neuropathy. MRI shows “striped pons” and vermian atrophy. NCS reveals sensorimotor axonopathy. Mean survival extends into the 4th to 5th decade, with gradual loss of ambulation but preserved cognition and fair quality of life.

CONCLUSION

“Classical MRI features (striped pons, superior vermian atrophy) along with clinical phenotype strongly supported the diagnosis of ARSACS, which was genetically confirmed in one sibling.”

REFERENCES

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