



## Early Onset Spinocerebellar Ataxia Type 2 from South India: A Case Report

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### ABSTRACT

*Spinocerebellar ataxia type 2 (SCA2) is a late-onset autosomal dominant ataxia characterized by progressive cerebellar ataxia, slow saccades, pyramidal findings and parkinsonism, caused by triplet CAG/CTG expansion in the ATX2 gene in chromosome 12. Age of onset is typically in the fourth decade with 10- to 15-year disease duration. We present a case of SCA2 with symptom onset within the first decade of life without extrapyramidal features or family history of ataxia. This case highlights the varied presentations of SCA's and the significance of genetic testing for ataxias.*

**Keywords-** SCA2, spinocerebellar ataxias, sporadic ataxia, CAG triplet repeat expansion

### INTRODUCTION

Spinocerebellar ataxia type 2 (SCA2) is an autosomal dominant ataxia characterized by progressive cerebellar ataxia, slow saccades, pyramidal findings and parkinsonism. The age of onset is in the fourth decade (34±14years).<sup>[1]</sup> All the patients affected with SCA2 have ATXN2 CAG trinucleotide repeat expansion. Most cases have 37 – 39 CAG repeats, with the expansions more than 200 in number being reported in the literature.<sup>[2]</sup>

### CASE REPORT

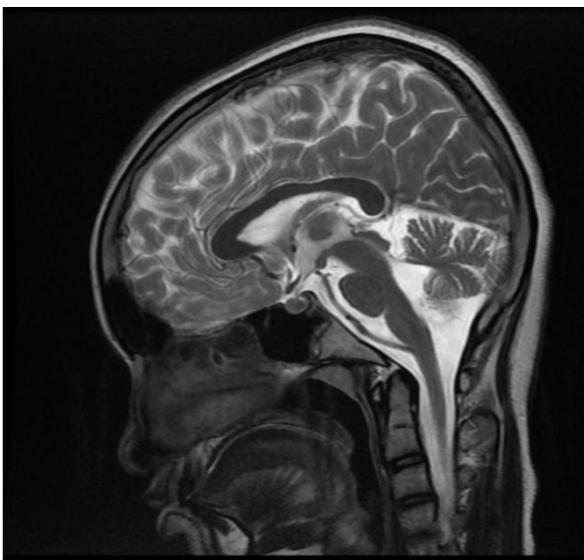
A 13-year-old school going girl resident of a coastal region of south India, presented with insidious onset

slowly progressive unsteadiness of gait and abnormality in gaze of three years duration. She had a tendency to reel and fall to sides while walking. She had an abnormal gaze when she was called from behind in that she would turn her head first and then her eyes. She was also noted to have loss of legibility of handwriting with macrographia. There was no history of tremor, chorea, dystonia, parkinsonian features or dementia. She was the first born of two siblings to non-consanguineous parents with normal birth and developmental history. Her past medical history was unremarkable. There was no history of similar illness in the family. There was no history of exposure to drugs, toxins or alcohol.

On examination she had normal vitals and general physical examination. Her higher mental functions were normal with a Folstein test MMSE score of 30/30. Her cranial nerve examination revealed significant slowing of saccades, normal pursuits with normal optic fundus and other cranial nerves. Motor system examination revealed normal bulk and tone with generalised hyporeflexic deep tendon reflexes. Plantar reflex was bilaterally extensor. All modalities of sensation were intact. There were bilateral cerebellar signs in upper and lower limbs with gait ataxia. Skull and spine were normal and other systems were normal.

Investigations revealed a normal routine blood examination and there was no evidence of acanthocytes on peripheral smear. Liver and renal function tests were normal. Thyroid function tests, chest x-ray and ECG were normal. Ophthalmological evaluation ruled out evidence of retinal pathology.

MRI brain revealed diffuse cerebellar atrophy and mild atrophy of cerebellar peduncles and pons (Fig1). There was no significant atrophy of brainstem structures.



**Fig. 1** T 2-weighted MRI showing prominent cerebellar atrophy

On DNA-PCR using fluorescent labelled primers for allele locus, and capillary electrophoresis analysis, triplet repeat size of 46 was obtained on one of the two alleles for SCA2, diagnosing the patient heterozygous for CAG repeat expansion.

## DISCUSSION

SCA2 is the commonest among autosomal dominant cerebellar ataxias in India.<sup>[3]</sup> The mean age of presentation according is in the fourth decade (34+14years).<sup>[1]</sup> The patient in the present case report had onset of symptom into her tenth year of life with a heterozygous triplet repeat number of 46. SCA2 has a relatively high incidence of extrapyramidal features and dementia. Dystonia and chorea are the prominent extrapyramidal features seen in more than one third of the cases.<sup>[4]</sup> Our patient lacked extrapyramidal features. This highlights the varied presentation of SCA2 and may question the proposed relation between triplet repeat number and symptoms. Environmental factors and allelic variations in CACNA1A are known to affect the age of onset, but the extend of variation is unknown.<sup>[5]</sup> Possibly environmental factors or other genetic modifiers previously described including CACNA1A play a more important role than was previously thought of.

In studies done on European patients, the yield of genetic testing was less when done in patients without a family history of ataxia (2 out of 842 patients).<sup>[6]</sup> In our case, there was a lack of family history of ataxia. This case maybe pointing that such data may not be applicable to the Indian population, and whether routine genetic testing is warranted in cases of cerebellar ataxic syndromes in Indian population given the higher incidence of SCA2, is debatable. Future studies are required in this direction to establish clear guidelines for genetic testing in ataxias.

## CONCLUSIONS

The clinical spectrum of inherited cerebellar ataxias continues to evolve, with unique clinical features being described periodically. Genetic testing plays an important role in diagnosing autosomal dominant cerebellar ataxias. There is a lack of proper guideline as to when genetic testing must be performed which is significant in developing nations, considering the higher cost of genetic testing.

**ACKNOWLEDGEMENT**

The authors thank the residents of Department of neurology, Govt.TD Medical College, Alappuzha for the support extended in preparation of manuscript.

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