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Neurology

# SPINOCEREBELLAR ATAXIA : A CASE REPORT

KEY WORDS:

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**ABSTRACT**

SCA1 is characterised by the development in the early or middle adult life of progressive cerebellar ataxia of trunk and limbs, impairment of equilibrium and gait, slowness of voluntry movements, scanning speech, nystagmus, dysarthria, dysphagia, rigidity. We report a case of genetically confirmed spinocerebellar ataxia type 1 (SCA1). The patient was a 35-year-old man who complained of gait instability and speech difficulties. Brain MRI showed marked atrophy of the cerebellum.

## INTRODUCTION

These are autosomal dominant group of hereditary neurodegenerative disorders characterized by cerebellar ataxia that may be associated with ophthalmoplegia, loss of vision, dysarthria, pyramidal and extrapyramidal signs, deep sensory loss, dementia or any combination of these abnormalities. Spinocerebellar ataxia is slowly progressive, which means that symptoms of the condition gradually worsen over a period of years.[1] Some types of SCA can progress more rapidly than others. Brain scans such as magnetic resonance imaging (MRI) and computerized tomography (CT) of affected persons often show shrinkage or atrophy of the cerebellum that becomes more noticeable as the disease progresses. Because there is an overlap of symptoms among the different types of spinocerebellar ataxia, genetic testing is needed to determine with certainty the type of SCA in an affected person. SCA 2, SCA 3, and SCA6 appear to be the most common and together account for nearly half of all families worldwide. SCA 2 is typically the most common among the SCAs in India, and the next most common SCA after SCA 3 worldwide.[2]

## CASE REPORT

A 35 years old non alcoholic male patient presented to us with 4 years history of very slowly progresssive dysarthia and gait imbalance during walking. He also complained of stiffness in his bilateral lower limbs .There was no abnormal posturing of hands or any other parts of body while carrying out activities of daily routine .There is no history of any motor or sensory deficit or incordination of upper limbs. his family history is significant and same type of illness is presented in his grandfather, father and sister.

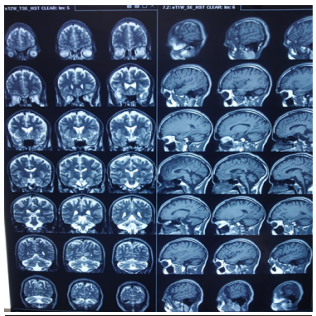
On examination vitals and general physical examination were normal. His higher mental function were preserved and cranial nerves were intact. On motor examination power was normal, tone was increased in lower and upper limbs. His deep tendon reflexes were brisk but plantar reflexes were bilaterally flexor. Sensory system examination was normal.

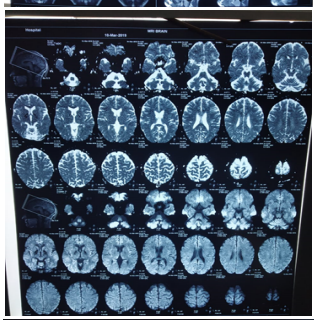
On cerebellar examination there was dysarthia. There was no nystagmus but a slight slowing of horizontal and vertical saccades were observed. There was marked incordination. Finger to finger, finger to nose, finger to doctor's finger, heel shin and romberg test were positive. Tandem walking test was also positive. There was marked gait ataxia during walking.

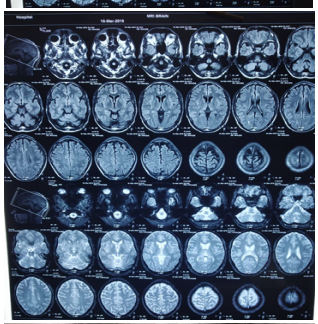
His routine investigations including hemogram, blood sugar, liver, renal, thyroid function tests and serum vitamin B12 level were all normal. He tested negative for antibodies to HIV, HCV, HbsAg.

Elecrocardiogram, x-ray chest and ultrasonography abdomen were also normal.

MRI brain revealed diffuse bilateral cerebellar atrophy.







Test Name : Spinocerebellar Ataxia PANEL (SCA1, 2, 3, 6, 7, 12,17)

Specimen : EDTA Whole Blood

Results

| Ataxia | CAG Repeats | Interpretation |
|---|---|---|
| SCA1 | 31 on Allele 1 and 53 on Allele 2 | Detected, Full Penetrance - Diagnosis of SCA1 confirmed |
| SCA2 | 22 on both the Alleles | Not Detected - Diagnosis of SCA2 excluded |
| MJD/SCA3 | 26 on both the Alleles | Not Detected - Diagnosis of SCA3 excluded |
| SCA6 | 11 on both the Alleles | Not Detected - Diagnosis of SCA6 excluded |
| SCA7 | 10 on both the Alleles | Not Detected - Diagnosis of SCA7 excluded |
| SCA12 | 10 on both the Alleles | Not Detected - Diagnosis of SCA12 excluded |
| SCA17 | 37 on Allele 1 and 39 on Allele 2 | Not Detected - Diagnosis of SCA17 excluded |

Method : PCR/ Fragment Analysis

## DISCUSSION

The phenotypic differences in symptoms between types of SCAs have been documented after cloning each of the SCA genes. Schols et al. reported differences in the characteristic findings between SCA1, SCA2, SCA3, and SCA6 patients.[3] The mean age of presentation according is in the fourth decade (34±14years).[4] The symptoms of our present patient were compared with those in their report and are listed. Although impaired optokinetic nystagmus and sensory disturbance were the typical abnormalities in SCA1, they were not observed in our case. Disease duration (4 years in our case vs. 7±4 years in typical reports), progression of gait deterioration (3 years vs. 6.0±4.2 years) were same in the present case as in typical SCA 1 case. SCA1 has a relatively high incidence of extrapyramidal features . Klockgether T et al. reported that the morphometrical study of brain MRI showed significant atrophy of the cerebellum and brain stem in three SCA mutations (SCA1, SCA2, SCA3) compared with a group of age- and sex-matched controls.[5] Comparison between the SCA groups showed that cerebellar and brainstem atrophy was more severe in SCA2 than in SCA1 and SCA3. Putaminal and caudate volume was reduced only in SCA3, but not in SCA1 and SCA2. However, a set of three morphological criteria defined by these investigators did not distinguish SCA1 from SCA2 and SCA3. Our case is consistent with their report.

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