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A case report on spinocerebellar ataxia

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Abstract
Spinocerebellar Ataxia Type 1 (SCA1) is a progressive, autosomal dominant neurodegenerative disorder caused by CAG trinucleotide repeat expansion in the ATXN1 gene. It primarily affects the cerebellum, leading to gait ataxia, dysarthria, and impaired coordination, with gradual neurological deterioration. We report the case of a 23-year-old female born of second-degree consanguineous marriage who presented with insidious-onset, progressively worsening imbalance while walking for one year, along with slurring of speech for five months. Family history was suggestive of autosomal dominant inheritance. Clinical examination revealed hypotonia and cerebellar signs including dysdiadochokinesia and impaired tandem gait. Routine laboratory investigations were within normal limits. Magnetic resonance imaging of the brain demonstrated bilateral cerebellar atrophy. Molecular genetic testing confirmed CAG repeat expansion in the ATXN1 gene, establishing the diagnosis of SCA1. This case highlights the importance of early clinical recognition and confirmatory genetic testing in young patients with progressive cerebellar ataxia. Early diagnosis facilitates genetic counseling, anticipatory guidance, and management of secondary complications.

Keywords: Spinocerebellar Ataxia Type 1 (SCA1), ATXN1 gene, CAG repeat expansion, autosomal dominant inheritance, cerebellar atrophy, genetic counseling, progressive ataxia

Introduction

Spinocerebellar ataxia type 1 (SCA1) is a progressive, autosomal dominant neurodegenerative disorder within the heterogeneous group of hereditary spinocerebellar ataxias. It is characterized primarily by cerebellar ataxia with variable involvement of the brainstem, pyramidal tracts, and peripheral nervous system. SCA1 belongs to the class of polyglutamine (polyQ) expansion disorders and serves as a well-established model of CAG repeat-mediated neurodegeneration.

SCA1 results from an unstable CAG trinucleotide repeat expansion in the ATXN1 gene located on chromosome 6p23. Normal alleles typically contain 6-38 repeats, whereas pathogenic alleles generally harbor ≥39 repeats. The expansion encodes an elongated polyglutamine tract within the ataxin-1 protein, leading to a toxic gain-of-function. Disease severity and age at onset correlate inversely with repeat length. Genetic anticipation, characterized by progressively earlier onset in successive generations, is frequently observed, particularly with paternal transmission.

The global prevalence of autosomal dominant spinocerebellar ataxias is estimated at 1-5 per 100,000 population, with SCA1 accounting for approximately 5-15% of genetically confirmed cases. Geographic clustering has been reported in parts of Europe, Japan, and the Indian subcontinent, reflecting founder effects and population-specific genetic backgrounds. Both sexes are equally affected.

Clinically, SCA1 typically presents in early to mid-adulthood with gait instability, limb incoordination, dysarthria, and dysphagia. As the disease progresses, patients may develop slow saccadic eye movements, pyramidal signs, peripheral neuropathy, and cognitive impairment. The disease follows a relentlessly progressive course, with loss of independent ambulation over 10-15 years and significant morbidity related to bulbar dysfunction.

Pathophysiologically, mutant ataxin-1 undergoes misfolding and nuclear aggregation, disrupting transcriptional regulation and RNA processing. Abnormal interactions with transcriptional repressors, including Capicua, result in widespread gene expression changes. Selective vulnerability of cerebellar Purkinje cells leads to impaired cerebellar output and the cardinal manifestations of ataxia. Additional mechanisms such as mitochondrial dysfunction, oxidative stress, and impaired proteostasis further contribute to neuronal degeneration.

Understanding the molecular and cellular basis of SCA1 has provided critical insights into polyglutamine-mediated neurodegeneration and continues to inform emerging therapeutic strategies targeting transcriptional dysregulation and protein homeostasis.

Case Report

A 23-year-old woman, born of a second-degree consanguineous marriage, presented with progressive imbalance while walking for one year. The onset was insidious, initially manifesting as mild unsteadiness while walking on uneven surfaces. Over the subsequent months, the imbalance gradually worsened, leading to frequent swaying and difficulty performing tandem gait. The symptoms predominantly affected the lower limbs and were not associated with sensory loss, weakness, or visual disturbances.

Five months prior to presentation, she developed slurring of speech, described as slow and scanning in nature. There was no history of diplopia, seizures, altered sensorium, bowel or bladder dysfunction, or cognitive decline. There was no history suggestive of toxin exposure, alcohol abuse, or prior central nervous system infection.

Family History

A detailed three-generation pedigree revealed a significant history suggestive of autosomal dominant inheritance. The patient's father, aged 48 years, had a history of progressive gait ataxia beginning at 35 years of age, followed by dysarthria and dysphagia over the next decade. He is currently ambulatory with support.

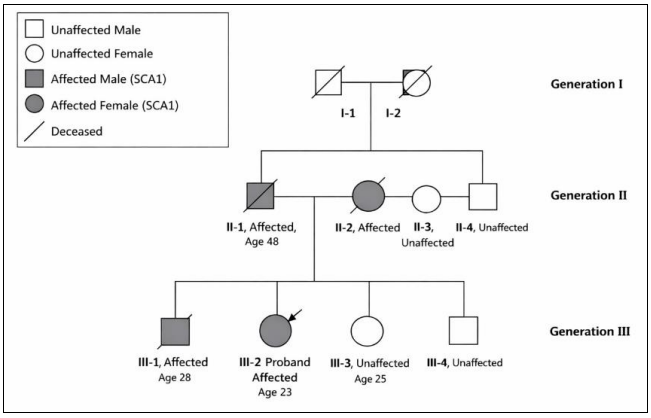
Among the patient's three siblings:

- The eldest brother (28 years) has experienced progressive imbalance since the age of 24 years and recently developed mild dysarthria.
- The second sibling, a 25-year-old sister, is asymptomatic.
- The youngest brother (19 years) is currently asymptomatic.

The paternal grandmother reportedly had progressive gait instability and became wheelchair-bound in her fifth decade, though no genetic diagnosis was established during her lifetime. There was no similar history on the maternal side.

The pedigree pattern strongly suggested autosomal dominant inheritance with possible genetic anticipation, as the age of onset in the proband appeared earlier than in the father.

Pedigree chart diagram



Examination

On general examination, vital parameters were within normal limits. Higher mental functions were intact.

Neurological examination revealed

- Hypotonia in bilateral lower limbs
- Normal muscle bulk and preserved power (5/5 in all muscle groups)
- Deep tendon reflexes were preserved; plantar responses were flexor bilaterally
- No sensory deficits

Cerebellar signs included

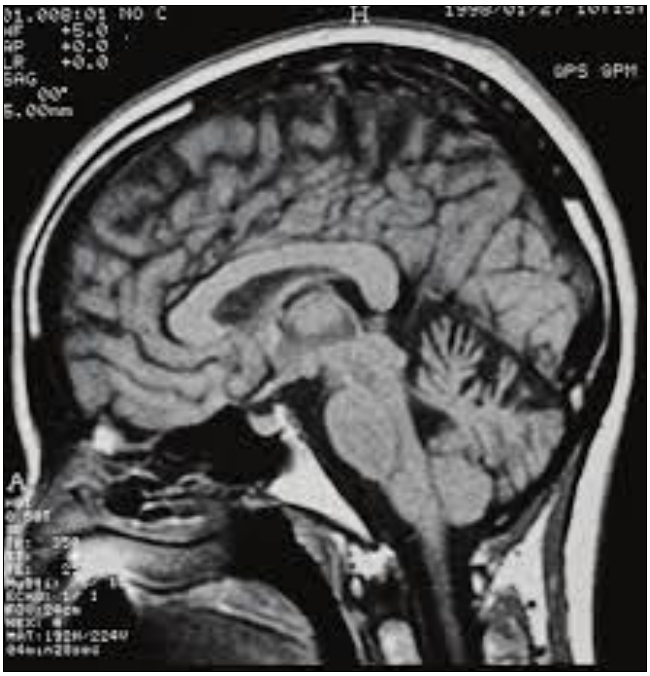
- Dysdiadochokinesia
- Impaired heel-shin coordination
- Intention tremor
- Broad-based gait with tandem gait swaying
- Scanning dysarthria

Ocular examination revealed mildly slow saccadic eye movements without nystagmus.

There were no extrapyramidal signs or evidence of peripheral neuropathy.

Investigations

Hemoglobin: 12 g/dL
RBC: 4.3 million/µL
Hematocrit: 37% Platelets: 3,42, 000/µL WBC: 8,150/µL
Serum Creatinine: 0.8 mg/dL Blood Urea: 32 mg/dL
Viral Markers: Negative
MRI Brain (Plain): Bilateral cerebellar atrophy



CAG Repeat Analysis in ATXN1 Gene

- **Allele 1:** 30 CAG repeats
- **Allele 2:** 47 CAG repeats

Discussion

Spinocerebellar ataxia type 1 is a progressive autosomal dominant cerebellar ataxia caused by CAG trinucleotide repeat expansion in the ATXN1 gene. The present case highlights early adult-onset ataxia with dysarthria and a strong multigenerational family history, consistent with autosomal dominant inheritance and probable genetic anticipation. The presence of hypotonia, dysdiadochokinesia, tandem gait impairment, and slow saccades supports predominant cerebellar and brainstem involvement, characteristic of SCA1.

Neuroimaging findings of cerebellar atrophy and molecular confirmation of pathogenic CAG repeat expansion establish the diagnosis definitively. The repeat length identified in this patient correlates with early onset disease, aligning with established genotype-phenotype associations.

This case underscores the importance of detailed pedigree analysis and early molecular testing in young individuals presenting with progressive ataxia. Timely diagnosis enables appropriate genetic counseling, risk assessment for first-degree relatives, and anticipatory management to reduce long-term morbidity associated with bulbar and gait dysfunction.

Conclusion

Spinocerebellar ataxia type 1 should be strongly suspected in young adults presenting with progressive cerebellar ataxia and a positive family history suggestive of autosomal dominant inheritance. This case illustrates the classical

clinical phenotype—gait ataxia, dysarthria, hypotonia, and cerebellar signs—supported by neuroimaging evidence of cerebellar atrophy and definitive molecular confirmation of CAG repeat expansion in the ATXN1 gene.

Early recognition and genetic confirmation are crucial for accurate diagnosis, prognostication, and genetic counseling of at-risk family members. Identification of pathogenic repeat expansion allows appropriate family screening and informed reproductive decisions. Although no disease-modifying therapy currently exists, timely multidisciplinary intervention can improve quality of life and reduce complications related to progressive motor and bulbar dysfunction.

This case emphasizes the importance of integrating clinical evaluation, pedigree analysis, and molecular diagnostics in the assessment of hereditary ataxias.

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