

The following article “A Viewpoint about the Treatment of Wilson’s Disease” by Abdul Qayyum Rana, Abolfazl Avan, Iqra Aftab, Wasim Mansoor and Tjaard Ubbo Hoogenraad, published in the Canadian Journal of Neurological Sciences 2013 Jul;40(4):612-4, has been retracted by agreement between the authors and the Editor-in-Chief, Robert Chen. The reason for the retraction is that two of the authors, Dr. Avan and Dr. Hoogenraad, indicated that they do not agree with the content and the publication of the article.

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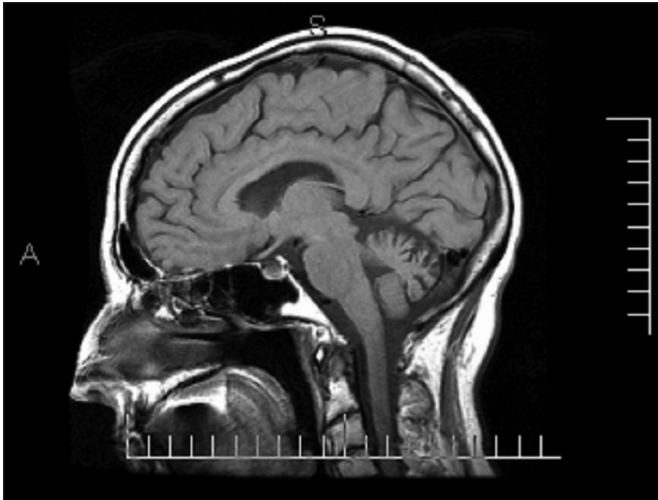
#### TO THE EDITOR

##### **Spinocerebellar Ataxia Presenting in the Eighth Decade of Life**

Spinocerebellar ataxias (SCA) are a heterogeneous group of autosomal dominant disorders characterized clinically by gait instability due to degeneration of the cerebellum and its connections.<sup>1</sup> Estimates of prevalence range from 3 per 100,000<sup>2</sup> to 19 per 100,000<sup>3</sup> population. There are at least 36 described subtypes<sup>4</sup> with SCA3 being the most common worldwide. The following report documents the onset of cerebellar ataxia at age 72 with a previously normal neurological history.

#### CASE REPORT

A 75-year-old woman was referred to the stroke clinic for dysarthria. Stroke was suspected because it was thought symptoms started one month earlier. She denied double vision or swallowing difficulties. In addition to dysarthria, she also reported a three-year history of slow and steadily progressive gait difficulty. Three years earlier she had a magnetic resonance imaging (MRI) of the cervical spine for this symptom. Cervical spondylosis was noted from the C3 through C7 levels. She was offered a cervical laminectomy which she declined. She had also been taking simvastatin at the time of initial symptoms; it was discontinued without abatement of symptom progression. Her medical history included hypertension, hyperlipidemia, colon cancer with right hemicolectomy, and osteoporosis. She denied a



**Figure:** T1 mid-sagittal MRI brain showing cerebellar atrophy.

prior history of migraines or episodic attacks of unsteadiness. Medications at the time of referral were nifedipine, colestipol, aspirin, multivitamins, calcium, and vitamin D. She did not use tobacco, alcohol, or illicit drugs. Her family history was significant for one of four sisters who developed gait difficulties and slurred speech after age 60 and a cousin who had gait disturbance in middle age and was clinically diagnosed with Friedrich ataxia. The neurological examination was notable for mild dysmetria with finger-to-nose and heel-to-shin testing, a wide-based gait, areflexia at the ankles with normal responses elsewhere, and diminished vibratory sensation in her toes. A brain MRI (Figure) showed cerebellar atrophy. Serologic testing was as follows: vitamin B12 358 pmol/L, folate 54 nmol/L, TSH 1.14 mIU/L, ESR 22 mm/h, cholesterol 5.5 mmol/L, triglycerides 1.5 mmol/L, HDL 1.7 mmol/L, LDL 3.2 mmol/L. An EMG showed no evidence of a large fiber neuropathy. She consented to genetic testing for spinocerebellar and Friedrich ataxia. There was a normal range of CAG repeats for the SCA1, SCA2, SCA3, and SCA7 genes. An abnormality of the SCA6 gene was detected with 13 and 21 repeats found. Friedrich ataxia mutation analysis was negative. She was diagnosed with spinocerebellar ataxia type 6 and referred to a neurogenetics clinic for further counseling and management.

## DISCUSSION

The above case description is notable for the age of symptom onset which was in the eighth decade of life. The patient was thought to have cervical spondylosis and cerebrovascular disease as a cause of symptoms most likely on the basis of her age. Family history and the progressive nature of symptoms suggested the alternative diagnosis. Compared with other spinocerebellar ataxias, SCA6 has an older average age of onset with a mean age in the fifth or sixth decade of life.<sup>5</sup> Previous reports of spinocerebellar ataxia have documented presentation in the eighth decade (up to age 74) with symptom onset in the seventh decade (up to age 65).<sup>1</sup>

The diagnosis of SCA6 is readily made on the basis of readily available testing. CAG expansions of 21 or more are considered

pathological, 19-20 intermediate, and 18 or fewer normal.<sup>1</sup> Mutation of CACNA1A, also observed in family hemiplegic migraine and episodic ataxia type 2, is the only known cause of SCA6. Besides cerebellar symptoms and signs, patients may also manifest dysarthria, bradykinesia, dystonia, hyperreflexia, diplopia, nystagmus, and peripheral neuropathy. Family history may not always be obvious as 10% of patients seemingly have a sporadic occurrence. The MRI findings consist only of cerebellar atrophy which may not always be severe, as in this patient. Pathological findings are typically limited to the cerebellum and include loss of Purkinje and granular cells. As a result of the restricted area of neurodegeneration, patients can typically live a normal lifespan despite symptoms. Treatment is supportive and includes genetic counseling, physical aids, and medications such as acetazolamide for episodic ataxia.

The diagnosis of spinocerebellar ataxia should be considered across the lifespan when a progressive gait disturbance occurs, even in patients who are in the eighth decade. Though no curative options exist at this time, a correct diagnosis prevents unnecessary additional testing and a definitive answer for the patient.

*Brian Silver*

*Rhode Island Hospital, Providence, Rhode Island, USA;*

*The Warren Alpert Medical School of Brown University, Providence, Rhode Island, USA*

*Email: bsilver@lifespan.org*

## REFERENCES

- Schols L, Bauer P, Schmidt T, Schulte T, Riess O. Autosomal dominant cerebellar ataxias: clinical features, genetics, and pathogenesis. *Lancet Neurol*. 2004 May;3(5):291-304.
- van de Warrenburg BP, Sinke RJ, Verschuuren-Bemelmans CC, et al. Spinocerebellar ataxias in the Netherlands: prevalence and age at onset variance analysis. *Neurology*. 2002 Mar 12;58(5):702-8.
- Shibata-Hamaguchi A, Ishida C, Iwasa K, Yamada M. Prevalence of spinocerebellar degenerations in the Hokuriku district in Japan. *Neuroepidemiology*. 2009;32(3):176-83.
- Ikeda Y, Ohta Y, Kobayashi H, et al. Clinical features of SCA36: a novel spinocerebellar ataxia with motor neuron involvement (Asidan). *Neurology*. 2012 Jul 24;79(4):333-41.
- Gomez CM, Thompson RM, Gammack JT, et al. Spinocerebellar ataxia type 6: gaze-evoked and vertical nystagmus, Purkinje cell degeneration, and variable age of onset. *Ann Neurol*. 1997 Dec;42(6):933-50.