

An unusual cause of clumsiness: spinocerebellar ataxia type 2

From her mid-40s, this patient had progressive balance problems and falls over 1 year. Precision hand movements were clumsy and speech was slurred. The patient's father had 'funny gait', the paternal grandmother unexplained falls, and the elder brother speech problems. Examination showed ataxic gait, dysarthria, slow saccadic eye movements, limb ataxia with dysdiadochokinesis, relatively preserved power, and diminished tendon reflexes. Magnetic resonance brain imaging (*Figure 1*) showed profound cerebellar atrophy.

Diagnostic genetic testing for autosomal dominant cerebellar ataxias (many forms are described) was undertaken because of the family history of possible inherited disorder.

Ms V Todaro, Visiting Clinical Erasmus Student, Walton Centre for Neurology and Neurosurgery, Liverpool

Dr AJ Larner, Consultant Neurologist, Walton Centre for Neurology and Neurosurgery, Liverpool L9 7LJ

Correspondence to: Dr AJ Larner
(a.larner@thewaltoncentre.nhs.uk)

This showed CAG trinucleotide repeat expansion (approximately 40 copies, normal <32) in the ataxin-2 gene on chromosome 12q24.12, establishing the diagnosis of spinocerebellar ataxia type 2.

Spinocerebellar ataxia type 2 is characterized by gait ataxia, dysarthria, action tremor, slow saccades and decreased reflexes, as a result of abnormal accumulation of mutant ataxin-2 protein in intracellular

inclusions (Antenora et al, 2017; Velázquez-Pérez et al, 2017). **BJHM**

Antenora A, Rinaldi C, Roca A et al. The multiple faces of spinocerebellar ataxia type 2. *Ann Clin Transl Neurol.* 2017 Sep;4(9):687–695. <https://doi.org/10.1002/acn3.437>

Velázquez-Pérez LC, Rodríguez-Labrada R, Fernández-Ruiz J. Spinocerebellar ataxia type 2: clinicogenetic aspects, mechanistic insights, and management approaches. *Front Neurol.* 2017 Sep 11;8:472. <https://doi.org/10.3389/fneur.2017.00472>

Figure 1. a. Axial T2-weighted and **(b)** sagittal fluid-attenuation inversion recovery magnetic resonance brain imaging showing marked cerebellar atrophy, with normal appearing white matter on axial T2-weighted image.

