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Image of Interest

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Spinocerebellar Ataxia Type 2 (SCA2)

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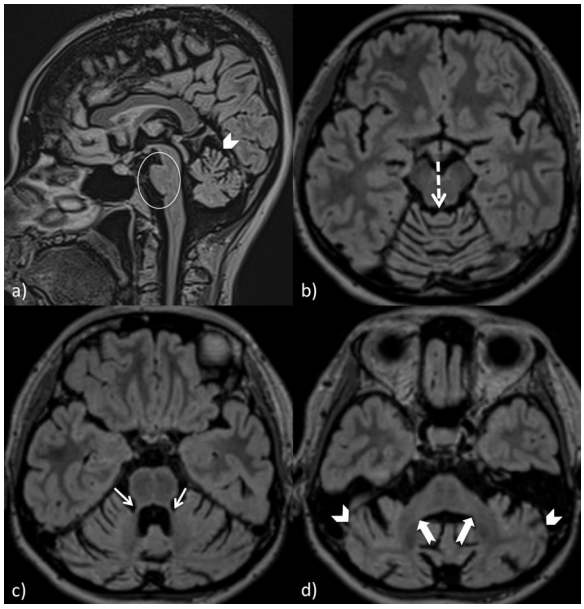

A 36-year-old Malay woman presented with a two-year history of progressively worsening unsteady gait and imbalance, first noticed by her colleagues. She remained independent in activities of daily living and required no mobility aid. There was no prior history of trauma, chronic illness, or drug use. Neurological examination revealed cerebellar signs bilaterally, mild left gaze-evoked nystagmus, and a broad-based gait. Power and reflexes were normal. A brain magnetic resonance imaging (MRI) revealed changes consistent—pontocerebellar atrophy (**Figure 1**). No abnormal parenchymal signal or lesion was noted in the brainstem or cerebellum. Genetic testing showed 38 CAG (Cytosine-Adenine-Guanine) repeats in the ATXN2 gene, confirming the diagnosis.

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The MRI (**Figure 1—refer legend**) showed marked atrophy of the bilateral cerebellar hemispheres, cerebellar vermis, superior and middle cerebellar peduncles, and the pons – pontocerebellar atrophy. The clinical and radiological features suggested spinocerebellar ataxia (SCA). The genetic testing confirmed the diagnosis SCA type 2 (38 CAG repeats in ATXN2 gene). Her family history revealed an older sister previously diagnosed with spinocerebellar degeneration. A pedigree chart was constructed, supporting an autosomal dominant inheritance pattern (**Figure 2**).

SCAs are a genetically and clinically heterogeneous group of neurodegenerative disorders primarily affecting the cerebellum and its associated pathways. SCA2, one of the more common subtypes in Southeast Asia, is caused by CAG trinucleotide repeat expansion in the ATXN2 gene. The disease typically presents in the third or fourth decade of life with progressive cerebellar dysfunction — as was seen in this patient. While the early symptoms are predominantly motor (ataxia, dysmetria, gait instability), additional features like dysarthria, peripheral neuropathy, and saccadic slowing may emerge with disease progression.[1]

MRI plays a pivotal role in the diagnostic workup. Although neuroradiological features are not pathognomonic, certain findings—such as pontocerebellar atrophy and preserved supratentorial structures—are strongly suggestive of polyglutamine SCAs. In SCA2, the superior cerebellar peduncles and pons are commonly affected early,[2] and this characteristic pattern, coupled with the clinical findings, can help prioritise genetic testing in resource-limited settings. MRI also provides a baseline for disease monitoring and assess disease progression.[3]

Figure 1: MRI Multi Planar Reconstruction in (a) sagittal and (b, c, d) axial views at different levels. There is marked atrophy of bilateral cerebellar hemispheres (arrowheads), vermis (dashed arrow), superior (white arrow) and middle (thick arrow) cerebellar peduncles and pons (circled).

Abbreviation

MRI	Magnetic resonance imaging
CAG	Cytosine-Adenine-Guanine
SCA	Spinocerebellar ataxia
ATXN2	Ataxin 2

Declarations

Conflict of interests

The authors declare no conflict of interests.

Patient Consent

Patient consent has been obtained.

Acknowledgement

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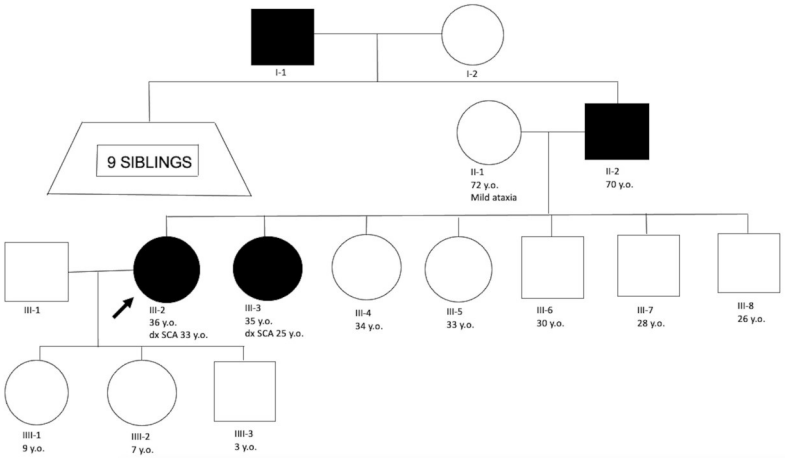

Figure 2: Schematic pedigree chart of the patient's family. There was at least one individual affected at each generation, with the index case (black circle with black arrow) being the third generation as seen in the pedigree. However, not all individuals were affected, which raise the possibility of incomplete penetrance of this hereditary disorder. Black: affected family member, white: unaffected family member.