Hemiparesis and Absent MRI Findings in Freidreich's Ataxia

Sir,

Freidreich’s Degenerative Ataxia (FRDA) is the most common hereditary ataxia among Caucasians, with a prevalence of 1 in 40,000.\(^1\,2\) As an autosomal recessive disorder, FRDA is caused by a mutation in the Frataxin (FXN) gene located on chromosome 9. The FXN gene encodes for frataxin - a 210 amino acid mitochondrial protein that has been implicated with numerous metabolic processes.

This patient was a previously healthy 19-year-old, right-handed female who presented with difficulty of maintaining her balance while walking. Early development was unremarkable, and walking began between 12 - 15 months. She reported noticing some throbbing in her calves at the age of 14 years. Slowly, her symptoms got worse and she decided to seek medical attention. There were no indications of cardiac murmurs, no problems with the lungs, no signs of hepatosplenomegaly and no abnormal facial features. Her ECG and echocardiogram showed no signs of abnormality. Upon presentation, she felt unbalanced and unsteady when walking - particularly when changing direction. She reported tightness in her legs and weakness in the left side of her body. She was not on prior medications, had no relevant allergies and no significant past medical or surgical history. She was a non-smoker and did not consume any alcohol. She was a student who lived with her parents. There was no consanguinity, additional family history of neurological conditions, or additional symptoms of ataxia.

Upon examination, she displayed spastic dysarthria. Motor testing showed weakness on her left side, which was pyramidal in distribution and graded at 4/5, but was not accompanied by unilateral fine motor impairment. Her deep tendon reflexes were absent, while her plantar responses were flexor on both sides. She had very mild dysmetria upon finger-nose-finger testing and heel-knee-shin testing. Sensory testing revealed a decreased sensitivity to pin pricks on to her mid-calves and wrists, decreased vibration on toes, although joint position sense was normal. She had a wide-based gait which did not show asymmetrical signs, and was unable to perform tandem gait. The rest of the neurological and systemic examination was unremarkable.

Contrast enhanced MRI of the spine and brain were atypical, showing no signs of spinal cord compression, stroke, or brain and cerebellar atrophy. Genetic testing was positive for a mutation of FXN. This patient displayed a GAA repeat size between 123 and 500 counts. In normal populations, the GAA repeat range should be no greater than anything between 6 and 36. Typical findings of FRDA are displayed in Figure 2 and Figure 3. Contrary to these findings, the case we are presenting exhibits moderately severe symptoms of FRDA in the absence of atrophy and other associated MRI abnormalities as shown in Figure 1.

Hemiparesis has been exhibited in the lacunar syndrome of ataxic hemiparesis, where patients have small infarcts in the pons and the right frontal subcortical white matter.\(^3\) Interestingly, our patient displayed mild left hemiparesis, which is not typically found in patients with FRDA. More so, this case is particularly unique as no damage or atrophy in the nervous system can account for the symptoms of FRDA and hemiparesis. From these observations, we suggest that further

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**Figure 1:** Our patient is a 19-year-old woman with FRDA who first presented with symptoms at the age of 14. This is her sagittal T1-weighted MR image showing the absence of atrophy at the level of the cerebellum and spinal cord.

**Figure 2:** Thirteen-year-old boy with FRDA who could walk with bilateral support six years post-onset. Sagittal T1-weighted (500/30, four excitations) SE MR image shows thinned cervical spinal cord and medulla oblongata (adapted with permission from Mascalchi et al., 1994).

**Figure 3:** Eighteen-year-old woman with FRDA who was confined to a wheelchair seven years following onset. Sagittal T1-weighted (500/30, four excitations) SE MR image shows scoliosis of cervical spine with abnormal posterior position of spinal cord within spinal canal (adapted with permission from Mascalchi et al., 1994).
investigation is needed into the etiology of these findings. Furthermore, it is recommended that clinicians look for hemiparesis in their patients. Clinicians should be active in reporting any new symptoms to the scientific arena for further examination.

REFERENCES


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