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Cervical spondylotic myelopathy and Kennedy syndrome mimicking amyotrophic lateral sclerosis

A 62 year old male patient presented with a longstanding history of slowly progressive limb weakness, speech, and swallowing difficulties. In 1983 a diagnosis of amyotrophic lateral sclerosis had been made. At that time his physical examination showed tongue atrophy with fibrillations, proximal limb weakness, and brisk lower limb tendon reflexes. Electromyography showed abnormal "spontaneous activity" with fibrillations and positive sharp waves in muscles of all limbs. His further medical and family history was unremarkable.

In January 2000 neurological examination showed mild facial weakness, marked atrophy and fibrillations of the tongue, severe dysarthria and dysphagia, atrophy, and weakness of the shoulder girdle and arm muscles, and an unsteady and broad based gait. Apart from brisk knee jerks, deep tendon reflexes were absent and plantar responses were negative. Sensory testing was normal. General physical examination showed slight gynecomastia. Laboratory testing showed raised creatine kinase (305 U/l) and lactate dehydrogenase (195 U/l) concentrations. Needle EMG demonstrated positive sharp waves and fibrillation potentials and long duration polyphasic motor unit potentials with increased amplitudes in muscles of all limbs. By contrast, motor and sensory nerve conduction studies gave normal results.

As cervical myelopathy is an important differential diagnosis in patients with suspected motor neuron disease, cervical MRI was performed. As shown in figure 1, MRI disclosed marked cervical spondylosis with appreciable narrowing of the spinal canal between C3 and C6. In addition, T2 weighted images showed intramedullary changes with foci of high signal intensity at the level of C5 indicating myelopathy. Although these changes may readily explain the weakness in his upper limbs, the cause of bulbar symptoms and denervation in his lower limbs remained unclear.

The presence of slight bilateral gynecomastia prompted us to look for androgen receptor gene mutations, which cause X linked spinal bulbar muscular atrophy.¹ This disorder, also known as Kennedy syndrome, is caused by an unstable expansion of a CAG repeat in exon 1 of the androgen receptor gene (Xq11-12). The androgen receptor is highly expressed in motor neurons of the brain stem and spinal cord. The CAG repeat expansion is thought to confer a toxic gain of function to the androgen receptor protein resulting in irreversible damage of brain stem and spinal cord motor neurons. In addition, the impaired ability to transactivate androgen sensitive genes of the mutated receptor may account for endocrine features such as gynecomastia or testicular atrophy in spinal bulbar muscular atrophy.²⁻⁴

Genetic analysis in our patient showed one allele carrying an abnormally expanded CAG

Figure 1 Spondylotic ridging at the C3-C6 level causing spinal stenosis is shown in the sagittal T2 weighted MR image. Additionally increased signal intensity is seen within the spinal cord at the level of C5.

repeat (44; normal range 16-33) thus confirming the diagnosis of Kennedy syndrome.

The present patient with coexisting cervical spondylotic myelopathy and Kennedy syndrome highlights the diagnostic value of an intensified investigation including cervical MRI and androgen receptor gene analysis in patients with an unusual clinical presentation of motor neuron disease.

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Posturally evoked vomiting without nystagmus in a patient with Arnold-Chiari malformation

Arnold-Chiari malformation type I (ACM I) is a developmental anomaly of the rhombencephalon characterised by displacement of the cerebellar tonsils into the foramen magnum and elongation of the medulla. It usually presents in adult life with head

motion induced oscillopsia, ataxia, headaches, cervical pain, or Valsalva induced dizziness.¹

Various ocular motor abnormalities have been reported in patients with ACM I. Among these, downbeat nystagmus and periodic alternating nystagmus are the most common. Other often encountered ocular signs, such as gaze evoked nystagmus, rebound nystagmus, and impaired smooth pursuit, reflect cerebellar involvement.^{2,3}

Vertigo of vestibular origin, being peripheral or central, is usually accompanied by nystagmus and nausea, or vomiting, and is often influenced by head position.^{3,4} The entity of central positioning vomiting without, or little, vertigo and nystagmus (posturally evoked vomiting (PEV)) was first reported by Drachman *et al* and later recognised by Brandt and Baloh and Halmagyi.^{3,5}

Posturally evoked vomiting is generally poorly known as a warning symptom of a posterior fossa lesion and is often misinterpreted.⁵ Whereas it has been documented in patients with posterior fossa tumours, it has not been reported in patients with developmental abnormalities. We report on a patient with ACM I where PEV was the most prominent presenting symptom.

This 57 year old woman was seen in our vertigo clinic because of gait unsteadiness and postural vomiting. Her history included an aortic valve replacement for aortic insufficiency, nephrolithiasis, and peptic disease. She was treated with warfarin.

For years, she had dizziness and severe nausea while looking up. During the past months severe nausea and vomiting appeared when she tilted her head to either side and down. Lately, she had become unsteady. She also complained about left high pitched tinnitus and intermittent pain in the left shoulder.

On examination her eyes were properly aligned with a full range of movements. No primary or gaze evoked nystagmus was seen, with and without Frenzel's glasses. The saccadic eye performance was normal, but the smooth pursuit in both the horizontal and the vertical plane was saccadic. The vestibulo-ocular reflex, examined by a doll's eyes movement, head thrust test, and dynamic visual acuity test, was normal. Mild dysmetria on finger-nose testing and finger-finger testing was found bilaterally. The deep tendon reflexes were brisk in the upper and normal in the lower limbs. The plantar toe responses were flexor. Sensation was normal. The gait was atactic and the Romberg test negative.

On testing the eyes in the Dix-Hallpike position to either ear, as well as in the head down position, the patient reported severe nausea, and became pale and perspired. However, no nystagmus was seen either by direct observation, or with Frenzel's glasses.

The symptoms persisted with repetition of the positioning.

An electronystagmogram (ENG) documented saccadic eye tracking in the horizontal plane. The optokinetic nystagmus was asymmetric with little increment after increased speed velocity of the target. When supine and with her head turned to the left, nystagmus of 7°/s, beating to the left, was recorded. No nystagmus was recorded on Hallpike testing. The caloric test was within normal limits.