Brain sagging syndrome presenting with chorea

Eoin Mulroy, MB, BCh, BAO; James Caldwell, FRANZCR; Neil E. Anderson, FRACP; Barry Snow, FRACP

Patient 1 is a 42-year-old previously well man on no medications who presented to neurology clinic with 6 months of nonpositional chronic daily headache, hiccups, and behavior change. His wife reported impulsivity, disinhibition, and sexually inappropriate behavior. There was no family history of movement disorder or neurodegenerative disease. Six weeks prior to symptom onset, he had fallen 1 meter onto his back. Vital signs, bloodwork, and general examination were normal. He was disinhibited and inattentive and exhibited orofacial, tongue, and limb choreoathetosis, which worsened over 6 months of follow-up (video at Neurology.org/cp). MRI was consistent with brain sagging syndrome; basal ganglia were subtly deformed, but without signal disturbance (figure). CSF opening pressure was 18 cm H₂O. A spinal CSF leak could not be located with conventional MRI, CT myelography, or radionuclide cisternography. Oral methylprednisolone, continuous epidural saline infusion, theophylline, and 2 lumbar epidural blood patches yielded no sustained improvement. Fat saturated MRI revealed a probable CSF leak at T4, but CT-guided T4 epidural blood patch was unsuccessful. He awaits surgical exploration.

Patient 2 is a 64-year-old man with hyperlipidemia and previous C6/7 fusion who reported nine months of worsening dysarthria, dysphagia, hiccups, and orthostatic headaches. Examination revealed nasal speech. Gag reflex, tongue power, and other cranial nerves were normal. Choreoathetoid orofacial, tongue, and limb movements were evident (video). Imaging confirmed brain sagging and normal basal ganglia (figure). Lumbar puncture was not performed. Spinal MRI and CT myelography failed to detect a CSF leak. Two separate lumbar blood patches produced temporary improvement in speech and chorea, but this was not sustained.

DISCUSSION

The syndrome of CSF hypovolemia with brain sagging is a recently recognized entity with highly variable clinical presentations.^{1,2} The archetypal symptom is orthostatic headache, but even this is not invariably present or pathognomonic.^{1,2} Clinical presentations vary from completely asymptomatic or mild postural headaches to the other end of the spectrum with debilitating frontotemporal dementia, extrapyramidal syndromes, and coma.^{1,3-5} Imaging findings are equally fickle.¹ The classic pachymeningeal thickening and enhancement, subdural collections, and venous engorgement may be absent, the only clue in these cases being brain slumping manifested by descent of the brainstem and cerebellar tonsils, posterior fossa crowding, and flattening of the anterior pons. Most cases result from a spinal CSF leak, but localizing these is often difficult.^{1,2} Treatment with targeted blood patching, epidural fibrin glue injection, or surgical exploration, though potentially curative, is unsuccessful in a considerable minority.

Movement disorders are rare but recognized presenting symptoms of the syndrome.⁶ Chorea has been described in 4 patients.^{3,6} It may result from mechanical distortion or vasogenic edema of the basal ganglia and their connections.^{6,7}

Practical Implications

Brain sagging secondary to CSF hypovolemia should be considered in the differential diagnosis of patients presenting with chorea.

Video

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Departments of Neurology (EM, NEA, BS) and Neuroradiology (JC), Auckland Hospital, New Zealand.

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Correspondence to: EoinM@adhb.govt.nz

Figure Imaging

Sagittal T1-weighted MRI sections and axial T2-weighted MRI sections through the basal ganglia of patient 1 (A, C) and patient 2 (B, D). Sagittal imaging shows brain slumping with downward displacement of the brainstem and cerebellar tonsils, flattening of the anterior pons, and posterior fossa crowding. Axial imaging shows slight basal ganglia distortion without signal change in patient 1 (C) and normal anatomy in patient 2 (D).

Brain sagging may be causative in the strikingly similar movement disorders exhibited by these middle-aged men. Consistent with previous reports, both exhibited choreiform orofacial movements (as did 3 of the 4 previously described cases), pronounced tongue chorea, and to a lesser degree, finger and foot chorea. Chorea onset also coincided with that of other symptoms, and patient 2 had temporary improvement in chorea following both blood patches. Their movement disorders are illustrated in the video.

Clinicians should consider this diagnosis when evaluating patients with chorea, especially in the presence of other suggestive features such as orthostatic headache.

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AUTHOR CONTRIBUTIONS

Eoin Mulroy: manuscript concept, acquisition of data, manuscript preparation. James Caldwell: critical review and editing of manuscript, study supervision. Neil E. Anderson: critical review and editing of manuscript, study supervision. Barry Snow: critical review and editing of manuscript, study supervision.

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