

AN INTRACRANIAL ARTERIOVENOUS MALFORMATION AND PALATAL MYOCLONUS RELATED TO PSEUDOXANTHOMA ELASTICUM

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Abstract:

Pseudoxanthoma elasticum is a rare hereditary disorder of elastic tissue with central nervous system manifestations due to occlusive vascular disease and aneurysm formation. Here we report the first recorded case of an intracranial arteriovenous malformation (AVM) in a patient with pseudoxanthoma elasticum. The AVM, which was located in the pons, also had an unusual manifestation, namely palatal myoclonus. (Aust NZ J Med 1989; 19: 141-143.)

Key words: Pseudoxanthoma elasticum, palatal myoclonus, arteriovenous malformation.

Pseudoxanthoma elasticum (PXE) is a rare hereditary disorder of elastic tissue with protean manifestations including characteristic yellow papular skin lesions, retinal angioid streaks, and occlusive vascular disease. The reported neurological manifestations of PXE are cerebral infarction due to progressive vascular disease and subarachnoid hemorrhage due to aneurysm formation.^{1,2} Here we report the occurrence of an intracranial AVM and palatal myoclonus in a patient with PXE.

CASE REPORT

A 38-year-old female presented in 1987 with severe right sided headache, neck pain and photophobia. A dermatologist diagnosed PXE in 1962. In 1976 tingling suddenly developed in her left hand and over subsequent hours her left arm became numb, and she had difficulty using it. She also complained of being unsteady on her feet. Neurological examination in 1976 revealed horizontal and vertical nystagmus, reduced touch sensation in the left upper limb and ataxia of the right hand and left leg. A radionuclide brain scan, visual evoked potentials, and cerebrospinal fluid examination were normal. In 1977 she had a constant right sided headache and later that year had a severe right sided headache requiring admission to hospital. The numbness in her left upper limb had remained. She now had

intermittent claudication in both legs. Examination revealed neck stiffness, angioid streaks, a right partial internuclear ophthalmoplegia, a right Horner's syndrome, a positive Romberg's sign, and unsteadiness on tandem gait. A lumbar puncture showed uniformly blood-stained CSF. A vertebral angiogram demonstrated a pontine AVM (Figure 1) supplied by the right posterior cerebral artery, with a dilated vein draining into the great vein of Galen. The AVM was considered inoperable. In 1981 she had increasing difficulty with walking and clumsiness of her left arm, diplopia on looking to the left, and persistent right sided headache. Neurological examination showed a right partial internuclear ophthalmoplegia, upbeat nystagmus on upgaze, and right horizontal gaze-evoked nystagmus. There was a reduction in palatal elevation, weakness of left finger extension and ataxia in the left forearm and right leg. Light touch sensation was reduced in the left hand. The tendon reflexes were depressed and the plantar responses were flexor. Her gait was wide-based. In 1983 her problems were dysarthria, and increasing difficulty with walking with several falls. Neurological examination showed intention tremor in all limbs, dysarthria and other signs as listed above. She had a course of radiotherapy to the intracranial AVM in 1983. In 1984 she had episodic severe headaches which required admission to hospital on three occasions.

On examination in 1987 yellow papules measuring 3 mm in diameter and redundant skin folds were noted over the neck,

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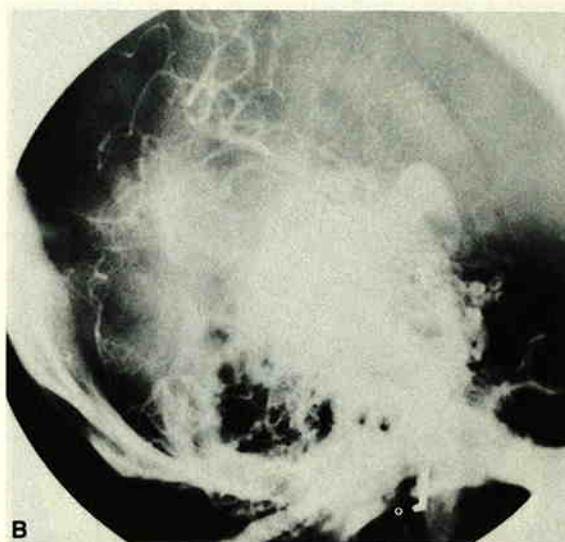
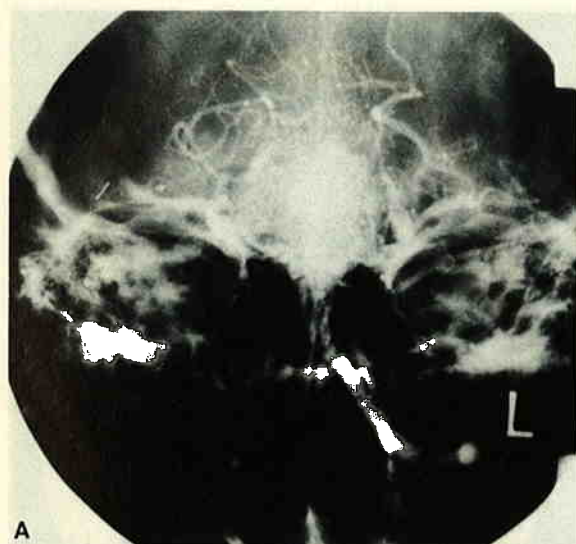


Figure 1 (A): Left vertebral angiogram showing arteriovenous malformation in anteroposterior view, Figure 1 (B): Left vertebral angiogram showing arteriovenous malformation in lateral view.

axillae and antecubital fossae. Titubation of the head was present. Her corrected visual acuity was 6/18 in the left eye and 6/12 in the right. Fundoscopy revealed normal optic discs, bilateral angioid streaks and hemorrhage at the left macula. There was a complex disorder of eye movements with a right internuclear ophthalmoplegia and disconjugate nystagmus with horizontal and vertical components. Light touch and pain sensation were reduced on the left side of the face. Examination of the mouth and neck showed bilateral palatal weakness with a normal gag reflex, palatal myoclonus at 2 Hz with slight deviation to the right, dysarthria, and involuntary vertical movements of the thyroid cartilage also at 2 Hz. The tone, power and deep tendon reflexes were equal and normal and the plantar responses were flexor. Postural instability, ataxia and dysidiadochokinesis of both upper limbs (worse on the left), involuntary movements of the left upper limb, and severe ataxia on heel-knee-shin testing were also present. She was unable to walk without support because of severe gait ataxia. Light touch sensation was reduced in the left arm. Head CT showed punctate calcification in the pontine region anterior to the fourth ventricle (Fig. 2A). Following contrast the region anterior to the fourth ventricle enhanced significantly (Fig. 2B and 2C). The cerebrospinal fluid was uniformly blood-stained. Polygraphic recordings demonstrated that the laryngeal and vertical eye movements were synchronous. Skin biopsy and review by a dermatologist confirmed pseudoxanthoma elasticum. The palatal myoclonus did not respond to carbamazepine, clonazepam or sodium valproate.

DISCUSSION

PXE is a rare inherited disorder of elastic tissue with diverse manifestations including angioid streaks, retinal hemorrhage and scarring, vascular disease of the limbs with claudication and diminished pulses, skin involvement with small yellow papules and redundant skin folds in the neck, axillae and groins, cardiac involvement and gastrointestinal hemorrhage.¹ Reported neurological manifestations include subarachnoid hemorrhage,²

multiple cerebral infarcts due to atheromatous changes in large and small arteries,³ carotid artery aneurysm,⁴ and anterior spinal artery aneurysm.⁵

This is the first reported case of an intracranial AVM in PXE. The pontine AVM had an unusual manifestation, namely palatal myoclonus. Palatal myoclonus and the associated involuntary movements involving the ocular, pharyngeal, laryngeal, diaphragmatic and limb muscles are due to a lesion of the pathway from the dentate nucleus through the superior cerebellar peduncle to the central tegmental tract and contralateral inferior olive.^{6,7} Causes of palatal myoclonus include syphilis,⁶ brain stem infarction,⁸⁻¹¹ multiple sclerosis,^{8,9} head trauma,^{8,9} Meige's syndrome,⁸ tumours including cerebellar astrocytoma,⁹ cerebellar degeneration,⁸ spinocerebellar degeneration,¹¹ Behçet's disease,¹¹ and possibly radiotherapy.¹²

We suggest that the palatal myoclonus in our patient is due to involvement of the central tegmental tracts by the pontine AVM. The pontine AVM also accounts for the other neurological signs. It is unclear whether the radiotherapy contributed to the development of the palatal myoclonus by damaging the pontine vasculature. The defect in elastic tissue in PXE may have predisposed to the development of the AVM. ■

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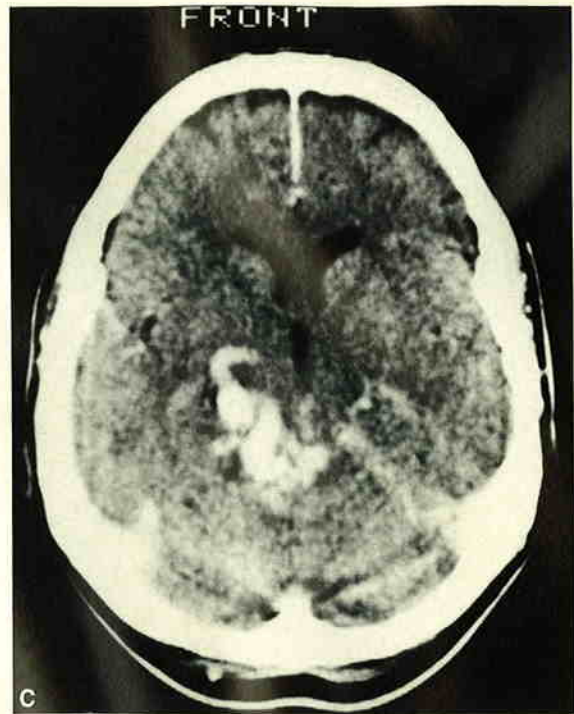
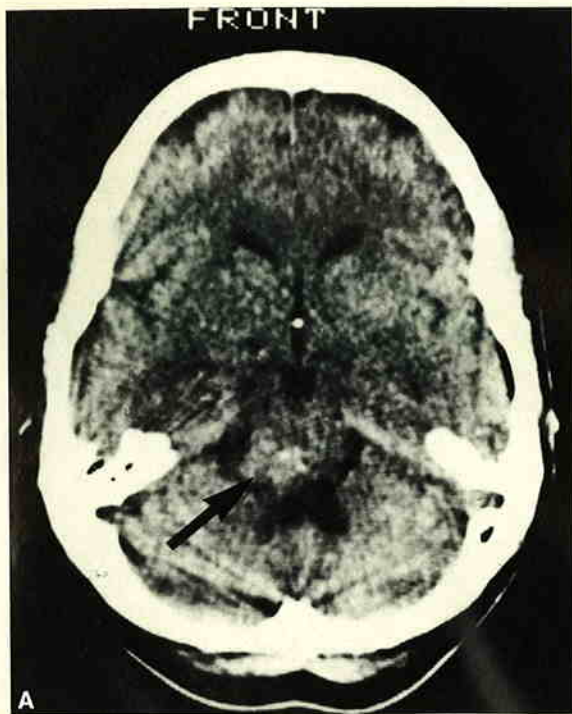


Figure 2 (A): CT without contrast, showing punctate calcification (arrow) in the region anterior to the fourth ventricle. Figure 2 (B): CT with contrast, showing enhancement in the region of the pons. Figure 2 (C): CT with contrast, showing dilated vessels draining the AVM.

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