Spinal arterial malformation in a child with hereditary hemorrhagic telangiectasia

Case report


Department of Neurology and Neurosurgery, Royal Children’s Hospital, Brisbane, Australia

A case is reported of spinal aneurysm in a child with a family history of hereditary hemorrhagic telangiectasia causing spinal cord and cauda equina compression. The operative approach is discussed.

KEY WORDS • spinal aneurysm • spinal cord compression • hereditary hemorrhagic telangiectasia • myelography

Several hundred families have been reported with hereditary hemorrhagic telangiectasia (Rendu-Osler-Weber syndrome).6 Spontaneous hemorrhage from the nasal mucosa is the most common symptom and the most common visceral anomaly is arteriovenous malformation in the lung.7 Reports of lesions in the brain are few6 and a spinal malformation has been reported in only one case.3 Spinal vascular malformations are uncommon10,11 in the general population and the presence of an intrathecal aneurysm is perhaps unique.

Case Report

This 5-year-old boy had complained of leg cramps for 2 years and his parents noted that his right foot was “turning in.” During investigations for progressive muscle wasting in the lower limbs, a muscle biopsy and lumbar puncture were performed. The lumbar puncture produced uniformly blood-stained fluid. His paraparesis increased and bladder dysfunction occurred for the first time. On August 18, 1973, he was transferred to the Royal Children’s Hospital.

Family History. Enquiry revealed an extensive family history of bleeding from mucous membranes, and anemia (Fig. 1); a great-aunt had had large multiple pulmonary arteriovenous fistulas (Fig. 2). The patient’s father had multiple telangiectasia on his nose, lips, and tongue (Fig. 3).

Examination. The patient could elevate the right leg against gravity but there was no movement in the left. Knee and ankle reflexes were absent and the right plantar response was extensor. Superficial abdominal reflexes were present but the cremasteric reflexes were
absent. Vibration sense was impaired in the legs without other definite sensory loss. The bladder was distended. There was no spinal bruit or any skin lesion over the spine.

Plain spine films showed erosion of the pedicles of L-1 and L-2, with widened spinal canal from T-12 to L-1 and scalloping of the posterior borders of the vertebral bodies of T-12, L-1, and L-2. Myelography via the cisternal route demonstrated a filling defect at the lower border of L-2 (due to the aneurysm) and a partial block at the L-5 level presumably from the lower margin of the subarachnoid blood clot.

Operation. A laminectomy from T-12 to L-3 (inclusive) was performed. The laminae of L-1 and L-2 were very thin. A thin-walled, tense swelling (4 x 1.5 cm) was present under the attenuated expanded dura. Its upper pole was adherent to the conus medullaris and compressing the cauda equina (Fig. 4). Blood clot filled the subarachnoid space of the cauda equina. After evacuation of the subarachnoid blood, four large arteries were demonstrated entering the aneurysm caudally. During manipulation, the aneurysm ruptured. Profuse hemorrhage prevented adequate exposure of the feeding vessels without the risk of injury to the nervous tissue. Surgical extension of the rupture site in the aneurysm allowed control of hemorrhage by occlusion of the orifices of the emptying arteries with artery forceps from within the sac. Further dissection, clipping of abnormal vessels, and excision of the malformation from the conus and the cauda equina continued without difficulty.

Histological study showed a vascular malformation composed of laminated layers of hyalinated fibrous tissue. The vessels were arterial and there was no venous component. Four weeks postoperatively, the patient was able to walk unaided and had a satisfactory return of bladder function.

Discussion

Hereditary hemorrhagic telangiectasia is a not uncommon condition of protean manifestations inherited as an autosomal dominant. Central nervous system involvement is much less common than that of bleeding from mucous membranes, from bowel, or lung lesions. The present case of an arterial malformation in the lumbar spine of a child with a family history of hereditary hemorrhagic telangiectasia suggests that this may be an uncommon manifestation of the disease.

Arterial anomalies producing spinal cord compression may arise in the development of collateral circulations in association with congenital heart disease. The existence of spinal arterial malformations has been questioned and none has been reported by Russell and Rubinstein. Spinal arterial anomalies have been recorded by Wyburn-Mason (3 cases), Brasch (1 case) and Brion, et al., but these authors made no reference to a large aneurysm. An incidental finding of an aneurysm in the thoracic cord in a patient with syphilis was reported by Babonneix and Widiez.
Neurological symptoms of spinal vascular lesions may result from shunting of blood, catastrophic hemorrhage, or thrombosis. Symptoms include transient paresis, sensory disturbance, anal and bladder dysfunction, muscle wasting, and complete paraplegia. Spontaneous subarachnoid hemorrhage may occur from spinal lesions and should be considered when four-vessel angiography is normal. The investigation of cases with suspected spinal angiomata has recently been reviewed and it was noted that the hydrodynamic block often shown at myelography is sometimes due to an arachnoiditis and not always to the mass effect of the angioma. In the case here reported, myelographic block was due to the aneurysm and subarachnoid clot. Selective spinal arteriography has improved the localization, extent, and operability of spinal vascular abnormalities. Prior to this investigation, occasional cases were diagnosed by aortography or vertebral angiography.

The operative and histological findings in this case confirm the diagnosis of a spinal arterial aneurysm causing cauda equina compression. Surgical control of hemorrhage from within the sac has been commented upon. A further unusual feature is the family history of hereditary hemorrhagic telangiectasia.

References
2. Babonneix L, Widiez A: cited in ref. 11, p 46
4. Brach F: cited in ref. 11, p 46

Fig. 3. Multiple telangiectasia on the patient’s father’s lips and tongue.

Fig. 4. Operative photograph of spinal aneurysm at L1–2 level showing thin-walled aneurysm compressing the conus medullaris (top left) and cauda equina, a feeding artery running obliquely (top left), and blood clot under arachnoid membrane (right).

Address reprint requests to: Glen S. Merry, F.R.C.S., Ladhope, 131 Wickham Terrace, Brisbane 4000, Australia.