Short report

Congenital spinal cord haemangioblastoma: another cause of spinal cord section syndrome in the newborn

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SUMMARY A newborn infant with negative perinatal history and characteristic clinical findings of upper cervical spinal cord section is described. Metrizamide myelography performed on the 7th and 22nd days of life was negative. Peroneal somatosensory evoked responses showed a conduction block at the cervical level. Necropsy revealed a haemangioblastoma extending from levels C1 to C5.

Since first described by Parrot in 1870,¹ the aetiology of spinal cord section syndrome (SCSS) in the neonate has been almost always associated with trauma during delivery.^{2,3} Better obstetric care and particularly the practice of prophylactic Caesarean sections in difficult cases with breech presentation has led to a decline in the number of birth injuries. However, the incidence of SCSS has not decreased proportionally, and factors other than birth trauma have proved to be responsible in some cases.^{4,5}

We present the necropsy findings of a spinal haemangioblastoma in a newborn with upper cervical SCSS, which to our knowledge has not been reported previously.

Case report

A 3,440 g newborn female was referred to us with the diagnosis of severe perinatal asphyxia. She was the product of a first, uncomplicated, 40-week pregnancy of a healthy young unrelated couple. There was no maternal history during pregnancy of viral infection, drug or alcohol ingestion, trauma or exposure to X-rays. Fetal movements were noticed by the mother from the 4th month of gestation. The amniotic membrane ruptured 10 hours prior to delivery; amniotic fluid was clear and no other signs of fetal distress were detected at that time. Delivery was vaginal with cephalic presentation and forceps extraction. Despite appropriate resuscitative procedures there was no spontaneous respiration and the patient required endotracheal intubation.

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On admission to the newborn intensive care unit, physical examination revealed marked hypotonia, absent deep tendon reflexes in the four limbs, and lack of spontaneous movement and respiration. No malformations or neurocutaneous stigmata were observed. No masses, liver or spleen enlargement were felt on palpation. There was no family history of neuromuscular disorders, retinal disease or tumours in the cerebellum or spinal cord. Chest radiography and routine laboratory tests were normal after a period of adjustment to mechanical ventilation. Cerebrospinal fluid contained 450 red blood cells with 0.97 g/l of protein and 0.73 g/l of glucose. Seven hours after admission she developed tension pneumothorax. After surgical drainage, persistence of fetal circulation syndrome was detected, requiring sedation and curare administration. Cerebral transfontanelle ultrasound study performed a few hours later was normal.

One week later, after curare and sedatives had been discontinued and the patient's circulatory complications had improved, neurological examination remained unchanged. Facial movements were normal. Sucking was present and strong. Pupil reactions and passive ocular movements were normal. No tongue fasciculations were noted. The patient consistently responded with a facial grimace or cry to pinpricks to the neck. She adopted a frog-like position and no movement of the trunk and limbs could be elicited even with sustained deep or superficial painful stimulation. Manual pressure on the bladder (Crede's manoeuvre) provoked dribbling of urine and decreased tone of anal sphincter. Myelogram followed by a CT scan with sagittal cuts showed no spinal cord pathology. Electromyography, nerve conduction velocities and muscle biopsy were normal.

Three weeks after birth, no changes were observed on neurological examination. Somatosensory evoked potentials obtained by stimulation of both medial nerves at the wrist showed normal Erb point response on both sides; the early cervical response was of low amplitude on the right and the remaining cervical and cortical responses were not obtain-

Fig (a) Tumour tissue invading the spinal cord. Note several anterior horn motor neurons on the left. (HE $\times 140$). (b) Section of the haemangioblastoma, composed of thin-walled capillaries, large endothelial cell nuclei and foamy cells. (HE $\times 555$).

able. A second myelogram revealed no spinal cord abnormality. The patient died at 6 weeks of age from cardio-respiratory complications.

Pathological findings

Post-mortem study revealed bacterial bronchopneumonia and sepsis as the cause of death. Macroscopic examination of the brain, brain stem and cerebellum was unremarkable. The spinal cord had a regular diameter at all levels. After sectioning, a striking yellow discoloration of the cervical cord with diminished consistency was observed. Transverse sections from different levels of the spinal cord were embedded in paraffin wax and stained with Haematoxylin Eosin (HE), Periodic Acid Schiff (PAS), Woelcke, Mallory, and Wilder's technique for reticulin fibres. In addition, frozen sections of the cervical level were stained with HE and Oil red O. Histology showed a tumour like vascular tissue, totally invading the cord at the upper cervical levels (C1–C2). The abnormal tissue surrounded and infiltrated the spinal structures irregularly as far as C5 (fig a). The neoplastic tissue was composed of capillaries with swollen endothelial cells. Vascular lumina were separated by abundant stromal foamy cells, which showed large numbers of sudanophilic lipid droplets on frozen sections (fig b). Wilder's

technique demonstrated an extensive network of reticulin fibres outlining the vascular architecture. The pathological diagnosis was congenital capillary haemangioblastoma of the spinal cord. No pathological manifestations of von Hippel-Lindau disease were found.

Discussion

Differential diagnosis of SCSS in the newborn includes neuromuscular diseases such as the infantile form of spinal muscular atrophy (Werdnig Hoffmann disease) infantile form of type II glycogenosis (Pompe's disease), cleft spinal cord, transversal myelitis and intra medullary tumours.⁶ Aetiology is difficult to assess since many patients with this syndrome die during the first hours of life, and post mortem study including the spinal cord is not always carried out.

Two different syndromes emerge from a review of more than 200 published cases:^{7–9} (1) cervico-thoracic section syndrome, generally associated with breech delivery and (2) upper cervical section syndrome, usually related to cephalic delivery with

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hyperextension of the neck. The former is characterised by flaccid weakness and areflexia in the lower limbs, with varying degrees of upper limb involvement and frequent Thorburn posture (arms held in strong abduction at the shoulders, with flexion at the elbows and wrists); sensory level is in the upper trunk. Sphincters are involved and there is paradoxical breathing. In the second syndrome, physical examination reveals flaccid tetraplegia with areflexia, lack of spontaneous respiration and neck pin-prick sensory level. Facial and extraocular movements are spared.

Upper cervical spinal cord section was diagnosed in our patient five days after birth, when curare was discontinued. Cephalic delivery, negative neuroradiologic findings and lack of perinatal trauma, made trauma an unlikely cause. However, sparing of the facial and extraocular muscles, absence of tongue fasciculations, normal nerve conduction velocities, normal muscle biopsy and the detection of a conduction block by somatosensory evoked responses ruled out other diagnoses.

Myelography is the method of choice in the diagnosis of spinal cord lesions. Magnetic resonance imaging¹⁰ could not be performed in our patient owing to her clinical condition. However, it was possible to obtain direct sagittal cuts on CT scan, a procedure which may be helpful and complementary after myelography in the newborn. Somatosensory evoked responses as in the case published by Bell,¹¹ were helpful in demonstrating a conduction block at cervical level, since neuroradiological studies were negative.

Congenital central nervous system tumours are rare and are mainly gliomas or teratomas, followed by meningeal tumours, neuroblastomas and medulloblastomas.¹² The finding of a congenital spinal capillary haemangioblastoma is exceptional.

This tumour is usually located in the cerebellum and occurs mainly in young and middle-aged adults. A spinal cord location is second in frequency followed by the medulla and some exceptional supratentorial cases.¹³⁻¹⁵ It can be associated with other lesions or tumours as in von Hippel-Lindau disease, and family incidence has been reported in a high percentage of cases.¹⁶⁻¹⁸

To our knowledge, congenital intracranial haemangioblastoma has only been reported once,¹⁹ and from all the published cases of spinal cord haemangioblastoma none was encountered in neonates.^{20 21}

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