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REVIEW

Cervical synostosis associated with Down's syndrome and Schneider syndrome. Clinical and medico-legal implications of an autopsy case☆

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Abstract This paper reviews the association of clinical symptoms and their clinical and forensic implications in a patient with Down's syndrome and congenital cervical synostosis who experienced a traumatic spinal cord injury. To date, no prevalent association between Down's syndrome and cervical synostosis has been reported in the literature. Given that both entities are prone to cause degenerative or traumatic cervical myelopathy, the combination of both conditions must be seen as a risk factor for spontaneous, and particularly traumatic, spinal cord damage. In these cases, radiological examination must be exhaustive, including MRI, given the possibility of spinal cord injury even after minimal trauma.

In the event of death, spinal cord autopsy is mandatory as it may reveal subclinical lesions, clarify the nature and extension of the spinal cord and skeletal injuries and help to establish an improved anatomo-clinical correlation.

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PALABRAS CLAVE
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Sinostosis cervical asociada a síndromes de Down y de Schneider. Implicaciones clínicas y médico-legales a propósito de un caso autópsico

Resumen Con ocasión de un caso de asociación entre síndrome de Down y sinostosis vertebral congénita que sufrió lesión medular traumática, se revisa la asociación de estos cuadros y sus implicaciones clínicas y forenses. La literatura no ha comunicado hasta ahora una asociación prevalente entre síndrome de Down y sinostosis cervicales. Ambos cuadros por separado propenden a la mielopatía cervical, bien de manera degenerativa, bien traumática. En consecuencia, la asociación de ambos debe verse como un factor de riesgo de daño medular o de manera espontánea o, sobre todo, después de traumatismos. En estos casos, la exploración radiológica debe ser exhaustiva incluyendo RMN ya que pueden existir lesiones medulares incluso ante traumatismos mínimos.

En caso de fallecimiento, la autopsia medular es obligada ya que puede revelar lesiones subclínicas, clarificar la naturaleza y extensión de las lesiones medulares y esqueléticas, así como ayudar a establecer una mejor correlación anatomoclínica.

Introduction

Central post-contusive myelopathy or Schneider syndrome was first described in 1954.[1] The typical presentation is that of a greater neurologic deficit in the upper limbs compared to the lower ones. In the original description, it was attributed to an anterior medullary contusion due to a herniated disc or other causes after a trauma.

Klippel–Feil syndrome (KFS) was described by these two authors in 1912.[2] Initially, the diagnostic criteria were clinical (short, stiff neck, and low posterior hairline). Later on, radiological requirements were added, with the main characteristic being the presence of congenital block vertebrae in the cervical spine.[3] It is known that the existence of vertebral synostosis between 2 or more spinal mobile segments promotes spinal cord injury over the course of a hyperextension, this greater propensity being attributed to adjacent hypermobile block segments.[4–6] This spinal cord injury may, in the cervical region, constitute Schneider syndrome.

Down's syndrome (DS) may predispose the development of degenerative cervical myelopathy,[7–9] with the changes increasing with age.

Being 2 conditions that independently tend towards degenerative or traumatic myelopathy, the association between cervical synostosis and DS may involve a high risk of neurologic injury, even in the face of minor trauma. This may present as primary or secondary injury, and is not always identified by routine tests.[10]

In this paper, we present a case of associated vertebral synostosis, DS and traumatic cervical myelopathy (Schneider syndrome). Clinical, radiological and autopsy data have been analysed and related literature reviewed, with the objective of considering the clinical and medico-legal issues posed by the possible coexistence of these 3 diseases.

As far as we know, no association between KFS and DS has been described in the literature.

Case report

On arrival at the hospital emergency department, the patient was conscious and presented with a lacerated contused wound to the scalp of approximately 4 cm without skull depression. Normal neck. Lacerated contused wound in superior maxillary region that communicates with oral cavity with loss of teeth.

The general examination revealed blood pressure of 56/33 mmHg, afebrile, conscious, reactive, agitated, nauseous, O_2 saturation of 94%. Nasal bone crepitus, periorbital haematoma in right eye and right forehead. Infranasal and upper lip lacerated contused wounds with loss of teeth in upper jaw.

Heart auscultation: sinus rhythm 90 bpm. No murmur. No jugular ingurgitation.

Lung auscultation: good ventilation in all lung regions. Pain on palpation in right inferior costal cartilage.

Erosion in left breast.

Abdominal auscultation: soft abdomen, depressible, erosion in abdominal wall at right flank level with haematoma in said area of approximately 7 × 9 cm in diameter. Erosion in pubic area.

Limbs: all 4 limbs moving. No focal neurologic deficit.

Additional emergency department tests:

- Blood and urine analysis and coagulation study: no significant abnormalities.
- Cranial CT scan with emergency intravenous contrast medium administration: no evidence of foci of intracranial haemorrhage or midline shift. Cortical and subcortical atrophy. No skull fracture lines observed. Paranasal sinuses with no warning signs. Deviated nasal septum. Nasal bone fracture.
- Cervical CT scan: no vertebral alignment disruption. Fusion of C5–C6 vertebral bodies. Spinous process