Journal of Forensic Radiology and Imaging 13 (2018) 23–26

Contents lists available at ScienceDirect

Journal of Forensic Radiology and Imaging

journal homepage: www.elsevier.com/locate/jofri

Case report

Subdural hematoma in an infant with alpha-1-antitrypsin deficiency and a rare primary intra-osseous vascular malformation

M.E.M. Vester[a,b,*], R.R. van Rijn[a,b], R.A. Bem[c], A. Maes[b,d], B. Kubat[b,e]

[a] Department of Radiology, Academic Medical Centre, Meibergdreef 9, 1105 AZ, Amsterdam, The Netherlands
[b] Department of Forensic Medicine, Netherlands Forensic Institute, Laan van Ypenburg 6, 2497 GB, The Hague, The Netherlands
[c] Department of Paediatrics, Academic Medical Centre, Meibergdreef 9, 1105 AZ, Amsterdam, The Netherlands
[d] Department of Pathology, Academic Medical Centre, Meibergdreef 9, 1105 AZ, Amsterdam, The Netherlands
[e] Department of Pathology, Medical University Centre Maastricht, P. Debyelaan 25, 6229 HX, Maastricht, The Netherlands

ARTICLE INFO

Keywords:
Child abuse
Subdural hematoma
Alpha 1-antitrypsin deficiency
Primary intraosseous vascular malformation
Forensic medicine

ABSTRACT

We present a unique case of a four-week-old boy who died of a severe hypoxic encephalopathy due to a subdural hematoma. A week before his demise, a small left sided parieto-occipital subdural hemorrhage was seen on both CT and MRI. On the right side a larger, fronto-parietal subdural hemorrhage was present with a prominently defined protuberance to the right hemisphere, causing a midline shift to the left. Subdural hematomas have a broad differential diagnosis, including abusive head trauma which can have far reaching consequences. In order to diagnose the underlying cause of the subdural hematoma, and subsequently confirm or refute the differential diagnosis, a medicolegal autopsy was ordered. The autopsy revealed a rare primary intraosseous vascular malformation of the skull, at the location of the subdural hematoma. This, in combination with the coagulation disorder due to alpha-1-antitrypsin deficiency, was considered to be the underlying cause of death.

1. Introduction

Accurate diagnosis of the cause of a subdural hematoma (SDH) can have far reaching medical, social, and legal consequences. This is because a SDH in young infants is more commonly seen as a result of abusive head trauma (AHT) than due to accidental causes or underlying diseases [1]. By correctly diagnosing SDH, particularly in cases where the origin is unknown, one can either assist in providing a safe environment for the infant or prevent false accusations and potentially erroneous convictions. We underline the importance of a broad differential diagnosis and close collaboration between clinicians and forensic pathologists by presenting a case report where AHT was initially considered, but a thorough medical and post-mortem forensic investigation resulted in the diagnosis of SDH as a result of a rare intra-osseous venous malformation.

2. Case report

A three-week-old boy was presented with vomiting and dehydration at a district general hospital. After admission, he rapidly developed epileptic seizures with impaired consciousness. The cause of which was diagnosed as SDH by ultrasonography. Furthermore, laboratory tests revealed a deficient coagulation (activated partial thromboplastin time (APTT) > 200 s [22–29 s], prothrombin time (PT) = PT > 180 s [9.7 – 11.9 s]), which was immediately corrected. After intubation and stabilization, he was transferred to a university hospital where a vitamin K deficiency, due to a suspected liver disorder, was diagnosed.

Given the SDH of unknown origin, AHT was part of the differential diagnosis and the child advocacy team of the university hospital was consulted. The boy was born at full term after an uncomplicated pregnancy to a G3P3 mother, into a socio-economic stable family. He had two healthy male siblings (respectively 4 and 5 years old) who had no significant social or medical histories. The boy received vitamin K supplementation, which consisted of 1 mg prescribed directly after birth, and then 150 μg daily from 8 days postpartum until the age of 12 weeks. One week after the birth, three small bruises on one of his shins were noted, with an otherwise normal physical examination. A full family history showed lung emphysema at a young age in one of his maternal grandparents and no abnormalities in his paternal family history. A full physical exam showed no bruises or other skin findings. A skeletal survey did not reveal any fractures or congenital abnormalities (Fig 1 A and B). Based on the results of these investigations by the child advocacy team, no risk factors for AHT were identified.

A head computed tomography (CT), performed at the university

Abbreviations: A1ATD, alpha-1-antitrypsin deficiency; AHT, abusive head trauma; CT, computed tomography; MRI, magnetic resonance imaging; SDH, subdural hematoma

* Corresponding author.
E-mail address: m.e.vester@amc.uva.nl (M.E.M. Vester).

https://doi.org/10.1016/j.jofri.2018.05.001
Received 12 March 2018; Received in revised form 2 May 2018; Accepted 2 May 2018
Available online 03 May 2018
2212-4780/

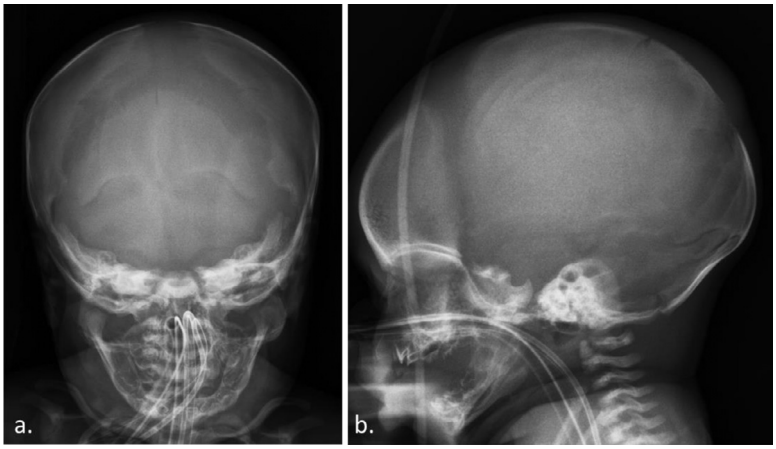

Fig. 1. a. AP and b. Lateral, conventional radiographs of a three-week-old boy showing widened sutures as a sign of increased intracranial pressure, along with bilateral, parietal accessory sutures. No other abnormalities are visible.

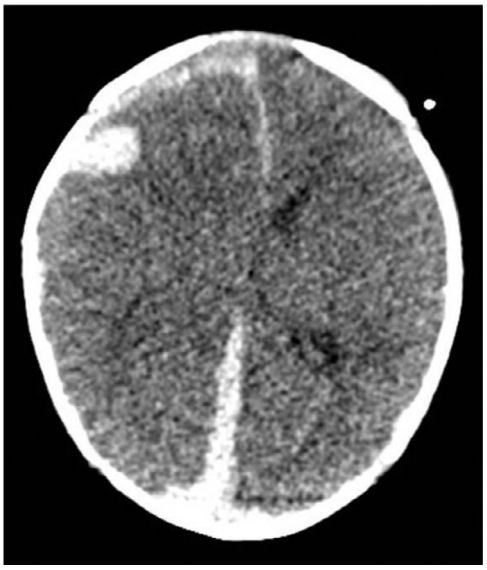

Fig. 2. Head CT of a three-week-old boy showing a right sided subdural hematoma with parenchymal hemorrhage causing a midline shift to the left.

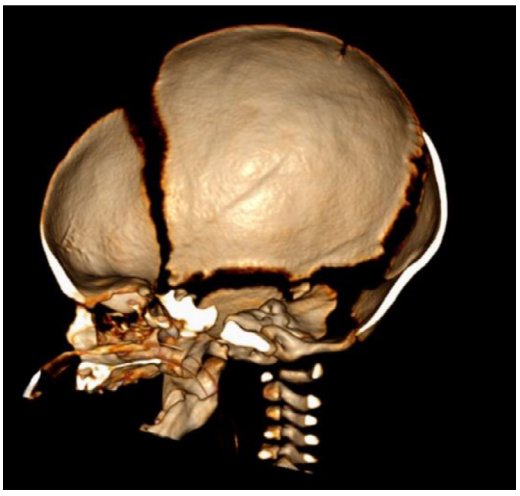

Fig. 3. 3D reconstruction of the right side of the head showing widening of the sutures. In retrospect, there is no demarcated lesion of the skull visible, as a possible sign of the vascular malformation.

hospital, revealed a 0.9 cm midline shift to the left due to a right-sided fronto-parietal SDH, extending along the falx and tentorium. There also was a frontally located right-sided focal parenchymal, possibly subarachnoid, hemorrhage (Fig. 2). As a result of an increased intracranial pressure, the sutures were widened (Fig. 3). On magnetic resonance imaging (MRI), bilateral diffusion restriction in the medial cerebral artery territory was seen (Fig. 4 A and B). There was no indication of an underlying vascular malformation at the site of the focal hemorrhage. On follow-up MRI the consecutive day, no changes were noted.

Based on the severe hypoxic encephalopathy and the widespread intracranial abnormalities, the treatment was, in light of the poor prognosis, discontinued. He died nine days after initial presentation. As the cause of death and reason for the subdural hematoma could not be explained, further investigations were required. Subsequently, a medicolegal autopsy at the Netherlands Forensic Institute was requested by the general prosecutor.

At external examination, jaundice, signs of medical intervention, and a 3 cm wide subcutaneous hemorrhage over the left side of the skull were noted. Autopsy showed 20 ml of subdural blood and blood clots on the right side of the skull, which resulted in an imprint on the brain tissue after removal (Fig. 5). Based on neuropathological analysis, the SDH was dated to be approximately one week old [2]. Cortical and some subcortical necrosis was present, indicating per-acute hypoxic encephalopathy. As part of the autopsy, a specimen radiograph of the skull was made, revealing a small but well demarcated lytic lesion of the right parietal bone (Fig. 6). Histology of this lesion showed the presence of an intra-osseous vascular malformation, with extra-osseous extension into the intracranial space (Fig. 7 A–C). This was ruled as a primary intra-osseous vascular malformation by the pathologist.

A small retinal hemorrhage, dating more than two days old, was seen in the right eye. The liver showed cholesteric damage with lobular bilirubinostasis and some local ductal bilirubinostasis. Furthermore, as a result of ventilation, some lung emphysema was found. Microbiology and metabolic tests were negative.

Based on autopsy, the cause of death was ruled to be encephalopathy, secondary to the SDH. Post-autopsy, immunochemistry results based on prior clinically obtained serum became available. These results revealed a homozygous ZZ phenotypic alpha-1-antitrypsin deficiency, an autosomal recessive disorder (A1ATD, OMIM #613490). A1ATD can lead to coagulation disorders and vascular malformations. The vascular malformation, especially in combination with the coagulation disorder, was considered to be the underlying cause of the SDH. Therefore, this case was ruled as a natural cause of death.