A simple case of torticollis leads to the diagnosis of an infantile desmoplastic astrocytoma

Gilis N.[1], Bex A.[2], Bex V.[1], Tebache M.[3], Michotte A.[4], Piette C.[5], Vaessen S.[5], Defresne A.[6] & Lückers O.[1]

(1) Department of Neurosurgery, CHR Liège, Liège, Belgium
(2) Faculty of Medicine, CHU Liège, Liège, Belgium
(3) Department of Radiology, CHR Liège, Liège, Belgium
(4) Department of Pathology, UZ Brussel, Vrije Universiteit Brussel, Jette, Belgium
(5) University Department of Pediatrics, CHR Liège, Liège, Belgium
(6) Department of Anaesthesia and Intensive Care Medicine, CHU University Hospital of Liège and CHR Citadelle, Liège, Belgium

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Introduction

The pediatric cerebral oncologic pathology requires a precise diagnosis. Surgeons might be outside their comfort zone to diagnose rare pathology such as desmoplastic astrocytomas.

Case Report

We report the case of a 12-month child presenting motor regression (impossibility of standing straight and holding her head anymore) associated with a stiff neck and a right sided torticollis for the past three weeks. The use of her right arm has always been better than the left since birth. She showed a recent increase in head circumference. There is no history of traumatism, no fever, no vomiting, nor strabismus.

CT-scanner (Figures 1 & 2) and MRI (Figure 3) showed a huge supratentorial mass (10x10x8cm) involving the right frontal lobe and the lateral gyrus, there was also a small node in the right inferior parietal gyrus. This multilocular tumor is compounded by a central cystic (no enhancement after iv contrast) and a peripheral solid portion, which seems attached to the underlying dura and enhanced by Gadolinium injection. Vasogenic edema was also found. There was a mass effect and a subfalcine herniation of the cystic portion.

We performed surgery, gross total resection was not recommended due to the local extension to the lateral sulcus and gyrus involving the arteries. We could not risk a postoperative hemiplegia. Microscopic examination demonstrated a heterogeneous mass presenting alternatively high and low mitotic activity without neuronal component. The ATRX gene was positive: diagnosis of desmoplastic infantile astrocytoma (DIA) is made.

A radio-clinical follow-up will take place every 6 months. As of today, there is no regrowth of the remaining lesion.

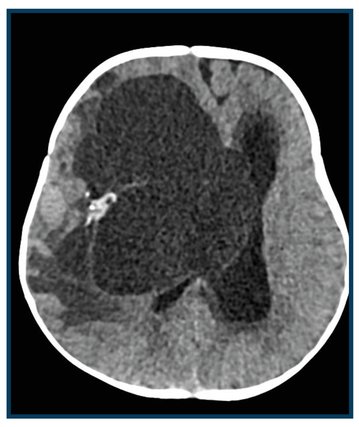

Figure 1: CT-scan without contrast

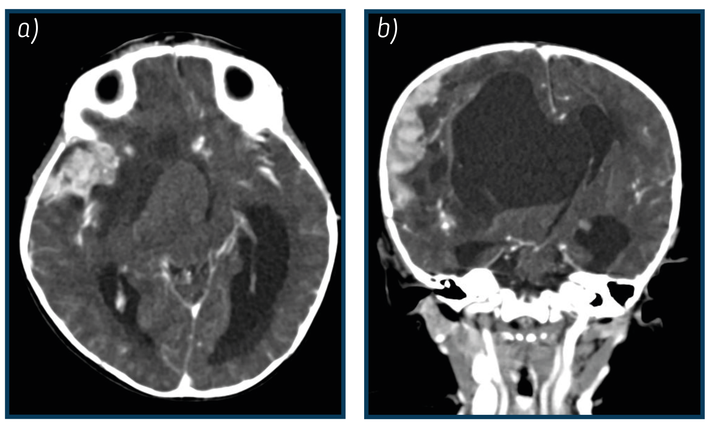

Figure 2: CT-scan with contrast

a) axial b) coronal views

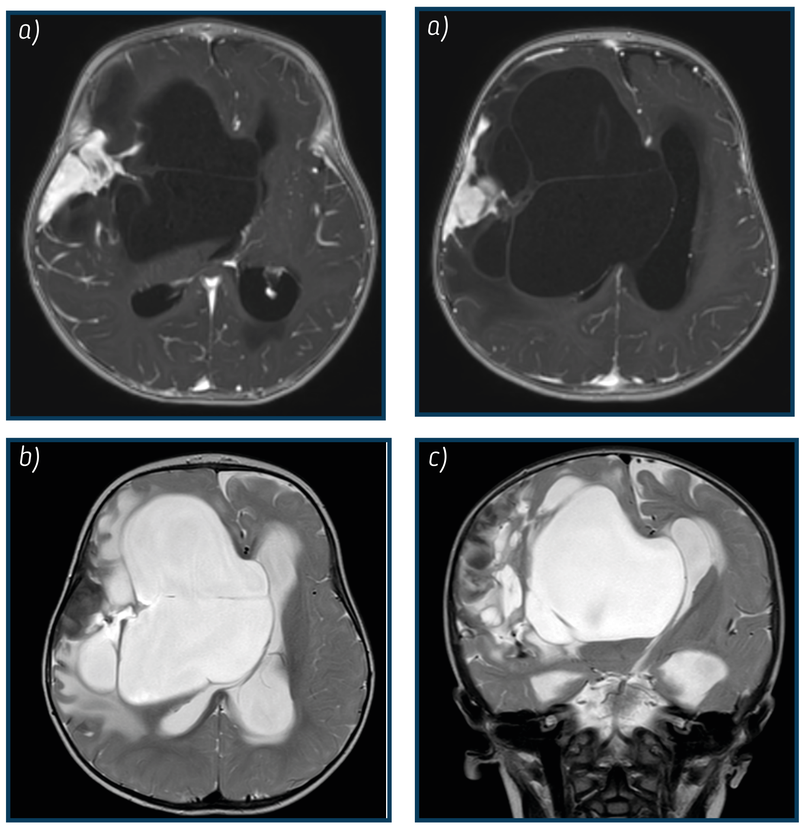

Figure 3: MRI-scan

a) axial T1 GADO b) axial T2 c) coronal T2 views

Discussion

Because of their similarities, desmoplastic infantile ganglioglioma (DIG) and desmoplastic infantile astrocytoma (DIA) regrouped under the same entity according to the 2016 WHO classification. They are benign tumours (WHO grade I).

DIA are mostly seen in infants (< 2 y.o.), clinical presentation is often related to the raised intracranial pressure. In our case we highlight three warning signs for clinicians:

(1) recent increase of the head circumference (large for age);
(2) torticollis;
(3) laterality of symptoms.

Differential diagnosis includes for each warning sign (1) hydrocephalus, (2) vision disorder and muscular problem, (3) stroke or cerebral palsy.

Radiological findings are pathognomonic showing a solid peripheral portion invading and thickening the meninges associated with a central cystic part.

Surgery remains the gold standard treatment.

Conclusion

Desmoplastic astrocytoma is a rare condition: the diagnosis can be challenging. It should be evoked when it is discovered in a child with a large supratentorial lesion made of a central cystic part and a peripheral solid component.