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Case Report

Atresia of the Aqueduct of Sylvius as a cause of congenital hydrocephalus ☆,☆☆

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ARTICLE INFO

Article history:
Received 12 February 2024
Revised 2 April 2024
Accepted 8 April 2024

Keywords:
Congenital hydrocephalus
Atresia of the sylvian aqueduct
Imaging
Brain MRI

ABSTRACT

Hydrocephalus is defined as an anomaly in the flow of cerebrospinal fluid, with multiple and varied etiologies, both acquired and congenital. The most dominant etiology in the congenital aspect is the stenosis or atresia of the Sylvian Aqueduct, whether isolated or associated with other malformations. We report a case of congenital hydrocephalus due to a stenosis of the Aqueduct of Sylvius in a 2 and a half-year-old child, which was unrecognized in the neonatal period, and the importance of imaging, especially MRI, in the rapid diagnosis of this pathology.

Introduction

Hydrocephalus is the most common brain malformation to be diagnosed in the antenatal period and can be managed and determine the etiology [1]. Hydrocephalus secondary to atresia or stenosis of the Sylvius aqueduct is the hydrocephalus that preserves the best neurological prognosis if surgical treatment is implemented the right time [1]. We report a case of congenital hydrocephalus due to a stenosis of the Aqueduct of Sylvius in a 2 and a half-year-old child, we highlight the importance of cerebral MRI.

Case presentation

A 33-hour-old female newborn was referred from another hospital to the University Hospital for management of suspected neonatal leukemia. Antenatal history included a mother with Type 1 diabetes on insulin therapy and hypertension under treatment. Delivery was by cesarean section for preeclampsia at 35 weeks of gestation, with antenatal corticosteroid therapy received. The infectious anamnesis was positive, with stained amniotic fluid. The birth weight was 3500 g. The newborn presented with a cutaneous hemorrhagic syndrome character-

☆ Competing Interests: The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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https://doi.org/10.1016/j.radcr.2024.04.023

ized by petechiae, bruises, and diffuse nodules on the trunk and back. Biological assessment showed thrombocytopenia at 61,000 elements/mm^3, leukocytes at 66,600 elements/mm^3 with 15% immature lymphoblasts, Hb at 15g/dL, and CRP at 11 mg/L. Antibiotic therapy with third-generation cephalosporin combined with aminoglycosides was initiated, leading to good clinical improvement and the disappearance of biological and clinical anomalies. TORCH serologies were negative. A transfontanellar ultrasound revealed a left frontal fluid collection, and radiological exploration was completed with a cerebral CT scan showing a left frontal collection communicating with the right frontal horn. The diagnosis of neonatal leukemia was rejected, and the baby was discharged with close follow-up and further exploration of the cerebral fluid collection. The baby was lost to follow-up. At the age of 2 and a half years, the mother returned for consultation due to her daughter's walking abnormalities. Clinical examination revealed a cranial perimeter above normal, a still open and wide non-fused anterior fontanelle, cerebellar syndrome, slightly brisk osteotendinous reflexes, normal muscle strength, and normal speech. There was convergent strabismus on the right side. The rest of the somatic examination was unremarkable. A brain MRI revealed a large left hemispheric fluid formation with broad continuity with the left lateral ventricle, suggestive of a large unilateral hydrocephalus. Both ventricles were otherwise enlarged, with a more moderate enlargement of the third ventricle, and a 4 mm-long atresia of the Sylvian aqueduct was identified. This collection caused a mass effect on the vault of the skull, which was bulging, and on the midline, which was slightly pushed to the right side (Figs. 1–4). Ophthalmological examination showed visual acuity of 9/10 on the right side and 4/10 on the left, esotropia of the right eye, a clear cornea and good anterior chamber, and an opalescent lens. On fundoscopy, there was a pale optic disc and diffuse chorioretinal atrophy. Given the state of cerebral engagement that the patient presented, surgical treatment could not be performed.

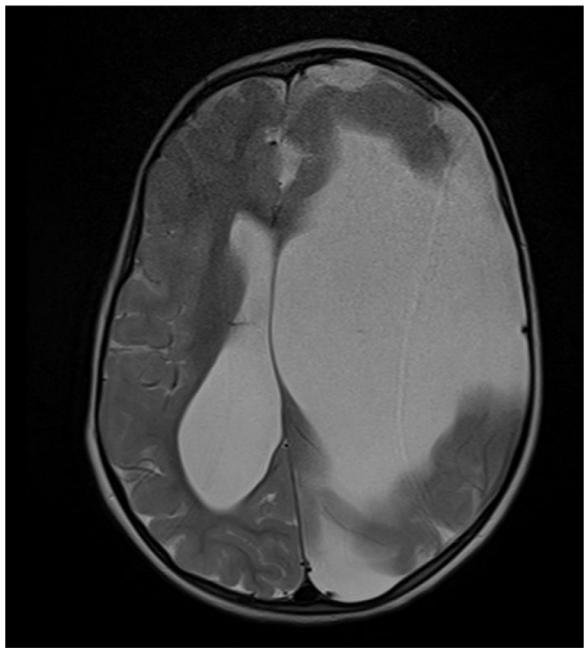

Fig. 2 – MRI brain demonstrates obstructive hydrocephalus with a mass effect in Axial T2-weighted image.

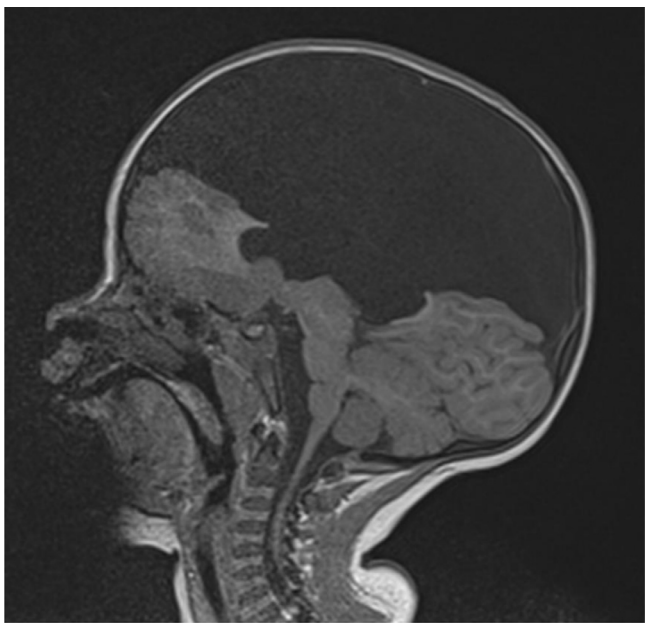

Fig. 1 – MRI brain demonstrates obstructive hydrocephalus secondary to Atresia of the Aqueduct of Sylvius in sagittal T1-weighted image.

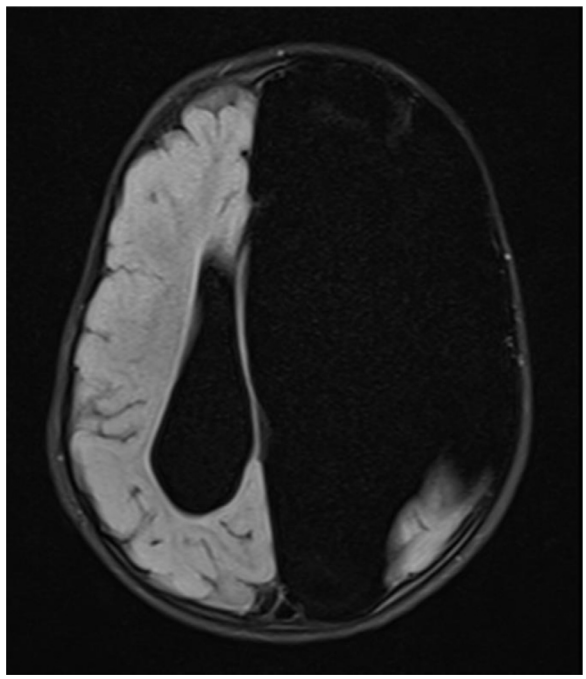

Fig. 3 – MRI brain demonstrates obstructive hydrocephalus with a mass effect in Axial T1-weighted image.

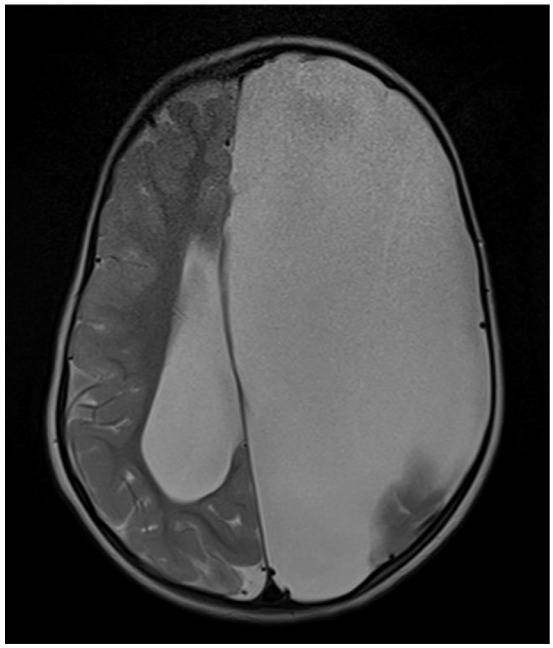

Fig. 4 – MRI brain demonstrates obstructive hydrocephalus with a mass effect in Axial T2 weighted image.

Discussion

The Sylvian aqueduct (SA) is the narrowest segment of the cerebrospinal fluid (CSF) flow pathway and the most likely site for intraventricular CSF obstruction [2]. Its etiology is mostly unknown [3]. It is responsible for 6% to 66% of cases of hydrocephalus in children and 5% to 49% in adults [3,4]. Its incidence varies from 0.5 to 1.0 per 1000 births, with a sibling recurrence risk of 1.0% to 4.5%. It can be inherited in an X-linked recessive pattern, as seen in Bickers-Adams-Edwards syndrome [4]. The frequency of clinical manifestations is seen either during the first year of life or at the beginning of adolescence [5]. Sometimes AS stenosis can be attributed to different causes, namely genetic, infectious, inflammatory, intraventricular hemorrhage related to prematurity, on which AS stenosis may develop, as was most likely the case of our patient [6]. Clinical manifestations are very diverse and rich in semiology, ranging in the acute phase from a simple increase in the cranial perimeter to delayed psychomotor and cognitive development, seizures, headaches, growth retardation, and sometimes urinary incontinence [2,7]. In the chronic stage, the symptoms are more severe: gait apraxia, standing difficulties, widened base of support, retropulsion, falls; head carried in slight lateral inclination, ataxia, gait disorders, pyramidal, and cerebellar syndrome from pressure on the white matter pathways surrounding the ventricles, increased reflexes, paroxysmal crises with hypertonicity of axial muscles and limbs in opisthotonus, spastic weakness of lower limbs with gait disturbances, akinetic mutism as the ultimate form of severe motor disturbance, neuropsychological tests detect a decline, with visuospatial or constructive difficulties associated with motor performance disorders; what is known as “the nonverbal learning disorder syndrome” Fletcher 1995 [7]. The examination of choice in front of this type of pathology remains the brain MRI either in the antenatal or postnatal period, for the diagnosis of a tri-ventricular hydrocephalus with a normal, non-dilated fourth ventricle. Especially in the sagittal section, this allows for better visualization of the stenosis as well as the presence of other cerebral malformations. On T1-weighted (WI) image sequences, the Sylvian aqueduct has the same signal intensity as CSF, whereas on T2 WI, a hypointense signal is seen as the result of CSF flow. This ’flow void’ extending down into the upper aspect of the fourth ventricle has been considered an important sign for diagnosing aqueductal patency [9]. cerebral MRI may precise topography of a possible obstruction; may show funneling superiorly of Sylvius aqueduct, distinguish the extent of obstructive hydrocephalus of the lateral and third ventricles with the fourth ventricle not dilated, transependymal resorption and signal abnormalities in the subependymal zone, images of leukoencephalopathy reflecting vascular processes more distant from the ventricles, monitorise the hydrocephalus during treatment [8]. Useful MRI protocols in AS are: sagittal T2 with the absence of flow-void signal intensity at the aqueductal level, obstructing web; on sagittal CISS and three-dimensional constructive interference in steady-state (3D-CISS): decreased aqueductal stroke volume; phase-contrast MR imaging: peak systolic velocity; cine cardiac-gated phase-contrast MRI: aqueductal CSF flow after aqueductoplasty with stenting [8]. Surgical treatment of hydrocephalus secondary to Sylvian aqueduct stenosis is possible by several surgical means, either ventriculoperitoneal shunt or endoscopic ventriculocisternostomy with or without stent placement [10–13]. Surgical treatment could not be performed on our patient due to the mass effect on the midline, which is pushed towards the right side, which is considered an element of poor clinical and therapeutic prognosis, which limits any means of surgical intervention. The frequency of progressing to a chronic picture is rare and serious, which leaves the attending physician unable to treat, the only way left is the monitoring and follow-up of the patient.

Conclusion

Hydrocephalus is a hard diagnosis announced to the couple, which remains delicate by its extensive damage assessment and its etiology. The stenosis of the Sylvius aqueduct is a rare etiology, but not to be ignored in front of a congenital hydrocephalus which can be managed immediately with fewer long-term sequelae due to imaging.

Patient consent

The authors declare that informed consent for publication was obtained from the patient’s parents.

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