Agenesis of the septum pellucidum

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Accepted 16 December 2020

DESCRIPTION

We describe the case of a female infant born of a term pregnancy (38 weeks) with regular surveillance and course, despite the mother having class III obesity and gestational diabetes controlled with insulin. During pregnancy, the mother had three fetal ultrasounds, all with normal morphology and biophysical profiles. She was delivered by emergency caesarean section and needed resuscitation with respiratory support and admission to our neonatal intensive care unit at birth. The Apgar score was 4, 9, 9 at 1, 5 and 10 min, respectively. Blood gas analysis showed metabolic acidosis at 1 hour of life. She did not fulfil criteria for therapeutic hypothermia. She started treatment with ampicillin and gentamycin for clinical signs of sepsis on day 3, interrupting treatment after 5 days due to negative blood cultures. During the first week of life, she experienced paroxysmal events suggestive of seizure, which subsided without need for treatment. Neurological examination at the time exhibited hypotonia and cortical thumb sign. Transfontanellar ultrasound (TF) on the first day of life suggested the presence of agenesis of the septum pellucidum (ASP) (figure 1) without other changes. Brain MRI on day 11 (figure 2) confirmed ASP and small corticosubcortical haemorrhagic foci without other associated malformations. Electroencephalogram exhibited infrequent paroxysmal activity in temporoparietal regions with no record of electrographic or electroclinical crisis. She was discharged after 29 days without abnormalities on physical examination. Currently, at 9 months old, she presents normal growth and neurological development and there were no episodes suggestive of seizure nor other complications since discharge.

The septum pellucidum is part of the limbic system, formed during the eighth week of pregnancy. The absence of this structure is a rare condition (general prevalence of 2 to 3 in 100 000 individuals) and can be partial or complete, congenital or acquired.[1–3] It may occur isolated or associated with other conditions, such as agenesis of the corpus callosum,

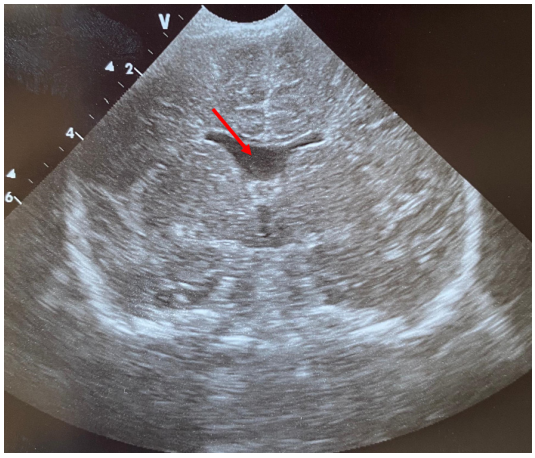

Figure 1 Transfontanellar ultrasound performed on the second day of life, in which the pellucid septum is absent (arrow).

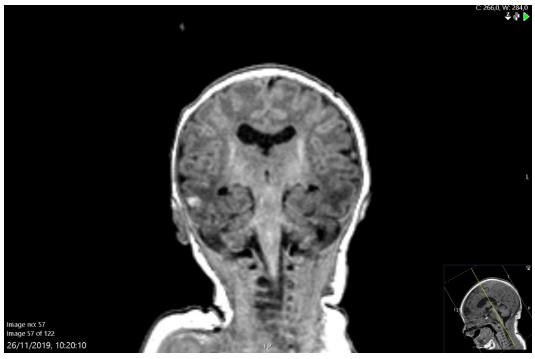

Figure 2 MRI on day 11 confirmed agenesis of the pellucid septum.

Patient's perspective

My experience with this problem was maddening. It was one of the worst experiences of my life, as I learnt about the malformation after a day and a half in labour and an emergency caesarean section. To make matters worse the baby was born with sepsis. I have to thank all the maternity hospital and intensive care unit personnel, who have always been very professional with me. Despite everything, she is a completely normal baby with a lot of energy.

Learning points

- Agenesis of septum pellucidum may arise in isolation or associated with other conditions such as agenesis of the corpus callosum, septo-optic dysplasia, Arnold-Chiari malformation, schizencephaly or holoprosencephaly.
- Prognosis depends on the clinical presentation and the associated anomalies, and information from fetal or postnatal MRI proves to be essential.
- Early recognition and regular follow-up are essential for the management of the condition.

To cite: Martins VL, Rodrigues J, Botelho T, *et al*. *BMJ Case Rep* 2021;**14**:e240716. doi:10.1136/bcr-2020-240716

septo-optic dysplasia, Arnold-Chiari malformation, schizencephaly or holoprosencephaly.[1 3–5] Prognosis depends on the clinical presentation and associated anomalies, and information from fetal or postnatal MRI proves to be essential. Published literature suggests mostly favourable developmental prognosis for isolated absent septum pellucidum.[4 5] Nevertheless, follow-up by Neurology, Endocrinology and Ophthalmology is recommended.[2] The described condition requires regular follow-up, in order to allow the anticipation and early recognition of signs or symptoms liable to intervention. Suspicion of ASP on prenatal or TF ultrasound must be excluded by prenatal or postnatal MRI, which are also essential to exclude other associated malformations. In our case, it was an incidental finding in a TF ultrasound performed due to concern for neonatal seizures and need for extensive resuscitation at birth. Although the exact aetiology of the small haemorrhagic lesions is unknown, we believe that they were probably related to hypoxic injury and not to the subject's absent septum pellucidum. Maternal biotype may have played a part for the fact that this finding was not evident in routine obstetric ultrasounds.

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Contributors All authors have participated in the drafting of the manuscript and have approved its final version. VLM and JR participated in the research of clinical data and wrote the text together. TB did the interview with the mother and gave information to the patient's mother to obtain patient consent. She also contributed to the revision of the discussion. RH reviewed the text and corrected errors and selected the images in the article.

Funding The authors have not declared a specific grant for this research from any funding agency in the public, commercial or not-for-profit sectors.

Competing interests None declared.

Patient consent for publication Parental/guardian consent obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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