SUBMITTED 11 FEB 23
REVISION REQ. 2 APR 23; REVISION RECD. 8 APR 23
ACCEPTED 19 APR 23
ONLINE-FIRST: MAY 2023
DOI: https://doi.org/10.18295/squmj.5.2023.024

Absent Septum Pellucidum

Search for other anomalies

Khadija Saleh,[1] Eiman Al-Ajmi,[2] *Amna Al Futaisi[3]

[1]Pediatrics Program, Oman Medical Specialty Board, Muscat, Oman; [2]Department of Radiology and Molecular Imaging, Sultan Qaboos University Hospital, Sultan Qaboos University, Muscat, Oman; [3]Department of Paediatrics, College of Medicine and Health Sciences, Sultan Qaboos University, Muscat, Oman
**Corresponding Author's e-mail: amnaf@squ.edu.om*

Introduction

A 6-week-old infant was referred to the paediatric neurology unit of a tertiary care hospital, in 2019 from a peripheral hospital for further evaluation. This referral was prompted by an antenatal scan that revealed an absent cavum septum pellucidum and a postnatal head ultrasound that suggested septo-optic dysplasia. Despite these findings, the infant appeared to be developing normally with normal muscle tone, strength and sucking reflex. The infant was born at full term via spontaneous vaginal delivery with a birth weight of 3 kg, head circumference of 34 cm, length of 52 cm and a good Apgar score; he had also received all the vaccinations recommended at birth. There was no history of maternal infection during pregnancy, medication intake, smoking or substance abuse. His parents were consanguineous, but there was no family history of genetic or neurological disorders.

An ophthalmologist examined the patient and found no optic nerve atrophy or other ocular abnormalities. Magnetic resonance imaging (MRI) of the brain at the age of thirty-three months revealed an isolated absence of the septum pellucidum (ASP) but no signs of septo-optic dysplasia, optic nerve hypoplasia or visible pituitary abnormalities (Figure 1). Furthermore, an endocrinological evaluation revealed no endocrine abnormalities. The

patient, who is currently 3 years old, is healthy with no obvious physical abnormalities and shows age-appropriate development; thus, was diagnosed with isolated ASP.

Informed consent was obtained from the parents for the publication of the images.

Comment

ASP is a rare developmental brain anomaly that can be partial or complete. It is often associated with congenital brain defects, such as holoprosencephaly, septo-optic dysplasia, schizencephaly and corpus callosum agenesis. Isolated ASP should be suspected in infants with normal findings on neurological examination but with an absent cavum septum pellucidum on antenatal scans.[1] It is challenging to differentiate between isolated ASP and septo-optic dysplasia *in utero* using current imaging techniques because it can be difficult to rule out the possibility of optic nerve hypoplasia and endocrine abnormalities.[2] Prenatal ultrasonography can detect cavum septum pellucidum between 18 and 37 weeks of gestation, or with a biparietal diameter of 44-88mm.The mean width of the cavum septum pellucidum is 5.3 +/- 1.7 mm with a range of 2 to 9 mm.[3] If the cavum septum pellucidum is not detected on antenatal scanning during this period, it may indicate abnormal brain development and necessitate further examination.[4]

Isolated ASP is uncommon, occurring in approximately 0.2–0.3 of every 10,000 people. However, the true incidence might be higher due to limitations in sonography such as skills of the operator, restricted field of view and visualization.[5] The prognosis of this condition is presently unknown, but it has been associated with undetectable pathological changes on ultrasound or MRI.[4]

Few cases of isolated septal agenesis have been associated with schizophrenia as it is considered part of the limbic system.[4] Other cases have been associated with speech delays or behavioural problems.[4] In a retrospective cohort study, antenatal genetic tests were performed in 30 foetuses with suspected isolated ASP, 2 of which had an abnormal result.[6] The majority of cases with a prenatal isolated ASP diagnosis have a favourable prognosis.[6] However, if other anomalies are detected, the clinical outcome is worse.[6] Screening for associated anomalies is mandatory for new-borns who are identified with ASP on antenatal ultrasound. An early understanding of whether ASP is an isolated anomaly or part of a syndrome enables the clinicians to provide appropriate counselling for families and to establish early rehabilitation and guidance for future management of such patients.

Authors' Contribution

KS did the initial writing of the manuscript. EA prepared the images and description and revised and edited the manuscript. AF was responsible for the idea of the manuscript as well as the revision and editing at all stages. All authors approved the final version of the manuscript.

References

1. Ben M'Barek I, Tassin M, Guët A, Simon I, Mairovitz V, Mandelbrot L, et al. Antenatal diagnosis of absence of septum pellucidum. Clin Case Rep. 2020 Feb 5;8(3):498-503. https://doi.org/10.1002/ccr3.2666
2. García-Arreza A, García-Díaz L, Fajardo M, Carreto P, Antiñolo G. Isolated absence of septum pellucidum: prenatal diagnosis and outcome. *Fetal Diagn Ther*. 2013;33(2):130-132. https://doi.org/10.1159/000338009
3. Falco P, Gabrielli S, Visentin A, Perolo A, Pilu G, Bovicelli L. Transabdominal sonography of the cavum septum pellucidum in normal fetuses in the second and third trimesters of pregnancy. *Ultrasound Obstet Gynecol*. 2000;16(6):549-553. https://doi.org/10.1046/j.1469-0705.2000.00244.x
4. Pilu G, Tani G, Carletti A, Malaigia S, Ghi T, Rizzo N. Difficult early sonographic diagnosis of absence of the fetal septum pellucidum. *Ultrasound Obstet Gynecol*. 2005;25(1):70-72. https://doi.org/10.1002/uog.1786
5. Hosseinzadeh K, Luo J, Borhani A, Hill L. Non-visualisation of cavum septi pellucidi: implication in prenatal diagnosis?. *Insights Imaging*. 2013;4(3):357-367. https://doi.org/10.1007/s13244-013-0244-x
6. Di Pasquo, E., Kuleva, M., Arthuis, C., Morganelli, G., Ormitti, F., Millischer, A. E., et al. Prenatal diagnosis and outcome of fetuses with isolated agenesis of septum pellucidum: cohort study and meta-analysis. *Ultrasound Obstet Gynecol*. 2022;59(2):153-161. https://doi.org/10.1002/uog.23759

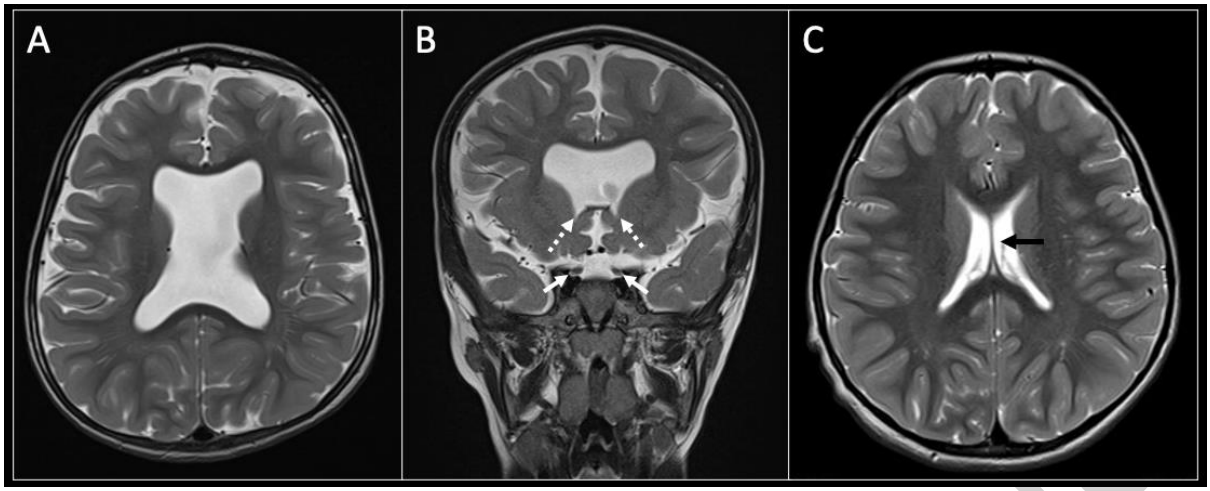

Figure 1. Magnetic resonance imaging of the brain done for the patient at age of thirty-three months shows absence of the septum pellucidum in an axial T2-weighted image (A). (B) Coronal T2-weighted image showing squaring of the frontal horns with inferior pointing (dashed arrows). The pre-chiasmatic segments of the optic nerves are bilaterally visible with normal size (arrows). (C) An axial T2-weighted image from another patient shows normal septum pellucidum (black arrow).