

Difficult early sonographic diagnosis of absence of the fetal septum pellucidum

G. PILU*, G. TANI†, A. CARLETTI*, S. MALAIGIA*, T. GHI* and N. RIZZO*

Departments of *Obstetrics and Gynecology and †Radiology, Policlinico S. Orsola-Malpighi, University of Bologna, Bologna, Italy

KEYWORDS: congenital anomalies; fetus; magnetic resonance imaging; prenatal diagnosis; septo-optic dysplasia; ultrasound

ABSTRACT

Standard sonographic views of the brain obtained along the axial planes at mid-gestation failed to identify absence of the septum pellucidum in two consecutive fetuses. It was presumably the close proximity of the walls of the lateral ventricles that generated an artifact resembling a normal cavum septi pellucidi. In one of these fetuses, septo-optic dysplasia was confirmed by magnetic resonance demonstration of hypoplastic optic tracts. In the other fetus, isolated absence of the septum pellucidum was the final diagnosis. Standard axial scans are unreliable in predicting absence of the septum pellucidum in the mid-trimester fetus when the lateral ventricles are normal in size. Copyright © 2004 ISUOG. Published by John Wiley & Sons, Ltd.

CASE REPORTS

Case 1

A 17-year-old primigravid woman was referred for a second opinion at 22 weeks' gestation. She had undergone sonographic examinations in two different laboratories with conflicting results. In one center, absence of the fetal septum pellucidum had been diagnosed and septo-optic dysplasia had been suspected. In the other center, the intracranial anatomy was considered to be normal. Sonography performed with a standard axial approach demonstrated a seemingly normal cavum septi pellucidi. It was only when coronal planes were obtained that the frontal horns were found to be fused, with a flat roof and no interposed septum pellucidum. The remaining brain structures appeared normal, with a well-formed corpus callosum (Figure 1). The implications of the sonographic findings were discussed with the patient and the family. As the differential diagnosis included septo-optic dysplasia, magnetic resonance imaging was offered as well as funipuncture for evaluation of the fetal endocrine status^{1,2}. The patient declined the examination

and only returned for a follow-up scan at 30 weeks. At this time, the absence of the septum pellucidum could be easily confirmed both in the standard axial planes and in the coronal ones (Figure 2). Magnetic resonance imaging confirmed the absence of the septum pellucidum and also suggested optic tract hypoplasia (Figure 3)¹. The patient was informed that septo-optic dysplasia was the most likely diagnosis and eventually underwent late termination of pregnancy in another European country, where the anomaly was confirmed by a postmortem examination.

Case 2

A 29-year-old woman, gravida 2 para 0, was referred for a second opinion at 23 weeks' gestation on suspicion of absence of the septum pellucidum. The sonographic findings were identical to the ones described for Case 1. Axial scans suggested the presence of the septum pellucidum, while coronal scans clearly indicated that it was absent. The patient refused any further testing and underwent termination of pregnancy. Autopsy confirmed absence of the septum pellucidum with apparently normal optic chiasm and tracts. A final diagnosis of isolated absence of the septum pellucidum was made.

DISCUSSION

Absence of the septum pellucidum occurs as part of many cerebral anomalies³. In some cases, these are complex malformations that are rapidly identified sonographically, such as severe (alobar or semilobar) holoprosencephaly, agenesis of the corpus callosum and severe ventriculomegaly^{1,4}. Sonographic demonstration of isolated absence of the septum pellucidum with central fusion of the frontal horns is a clue to the diagnosis of septo-optic dysplasia (also referred to as De Morsier syndrome)^{1,4,5}. This condition is frequently associated with neurological and endocrine deficits⁶, including poor vision, deficiency of growth hormone

Correspondence to: Dr G. Pili, Clinica Ostetrica e Ginecologica, Policlinico S.Orsola-Malpighi, via Massarenti 13, 40138 Bologna, Italy (e-mail: pili@orsola-malpighi.med.unibo.it)

Accepted: 11 September 2004

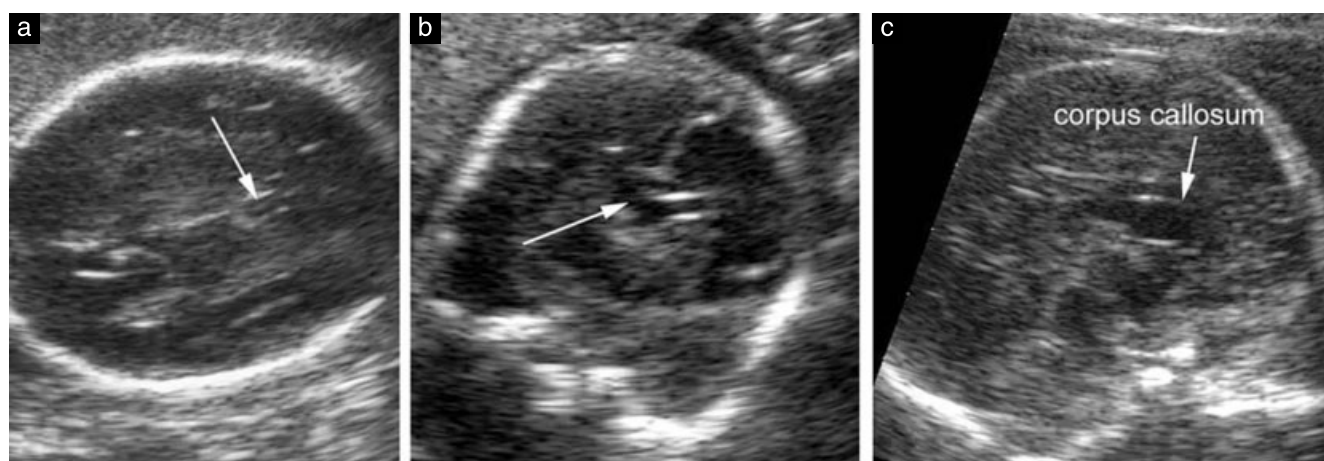


Figure 1 Sonographic images obtained at 22 weeks' gestation from Case 1: (a) a standard transventricular scan demonstrating a seemingly normal cavum septi pellucidum (arrow); (b) a coronal image obtained from the same patient demonstrating central fusion of the frontal horns with no interposed septum pellucidum (arrow); (c) a median scan demonstrating the presence of a well-formed corpus callosum.

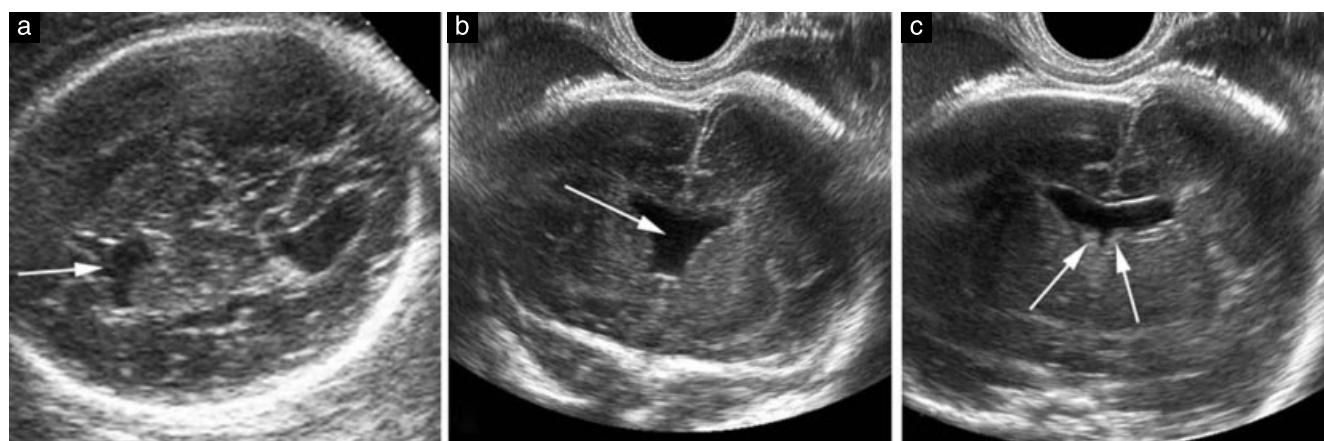


Figure 2 Sonographic images obtained at 30 weeks' gestation from Case 1. Both the axial (a) and the coronal (b) scans demonstrate the absence of the septum pellucidum (arrow); (c) coronal scan obtained posteriorly from (a) demonstrating the presence of two choroid plexuses (arrows).

and antidiuretic hormone, resulting in short stature and diabetes insipidus, respectively, and low levels of thyroid-stimulating hormone, luteinizing hormone and follicle-stimulating hormone. A subset of infants may present schizencephaly and other cortical plate disorders (septo-optic dysplasia plus)^{7,8} and these children are usually affected by severe neurodevelopmental retardation.

The leaves of the septum pellucidum and the interposed cavum can usually be well-visualized sonographically from at least 18 weeks⁹. Absence of the septum pellucidum is therefore theoretically amenable to diagnosis in the mid trimester. However, we have documented in two consecutive cases a similar artifact mimicking the presence of the septum pellucidum in the standard axial images of the fetal brain. We suggest that the artifact is due to the ultrasound beam crossing the walls of the frontal horns, which are normally in close proximity. This generates linear echoes that mimic the presence of septa within the ventricular cavity (Figure 4). This artifact was only present at mid-gestation, when the cavity of the fused frontal horns was small and

was scanned along the axial plane. It did not occur when the frontal horns were scanned with different angles of insonation, particularly in the coronal plane, or later in gestation, when the frontal horns had a larger cavity.

Our experience confirms that prenatal diagnosis of absence of the septum pellucidum can be made with ultrasound, and that a specific diagnosis of septo-optic dysplasia is possible by magnetic resonance demonstration of hypoplastic optic tracts¹. However, our cases also suggest that the ultrasound diagnosis is difficult at mid-gestation and may be impossible using a standard approach to the evaluation of the fetal brain.

We believe it is noteworthy that an anomaly such as septo-optic dysplasia, which is associated with significant clinical manifestations and is theoretically simple to demonstrate, may go undetected even by a well-performed basic sonographic examination of the fetal brain. In both our cases only non-routine views of the brain allowed the identification of the condition in the mid trimester.

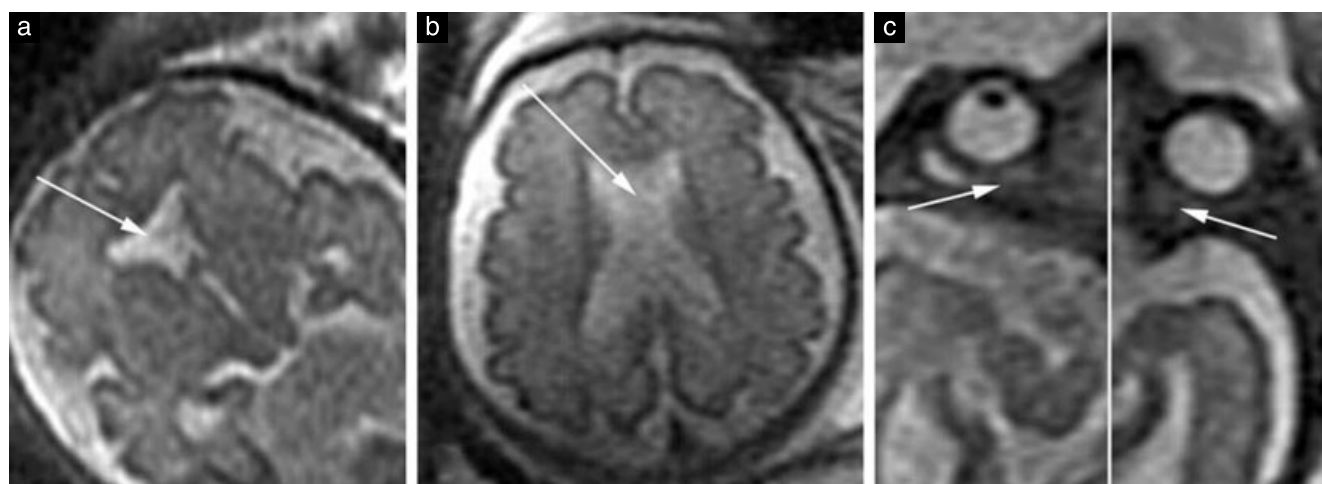


Figure 3 Magnetic resonance images obtained at 30 weeks' gestation from Case 1: brain views (a,b) demonstrating central fusion and squared roof of the frontal horns (arrows). In (c) very thin optic tracts, about 1 mm in diameter, are seen (arrows).

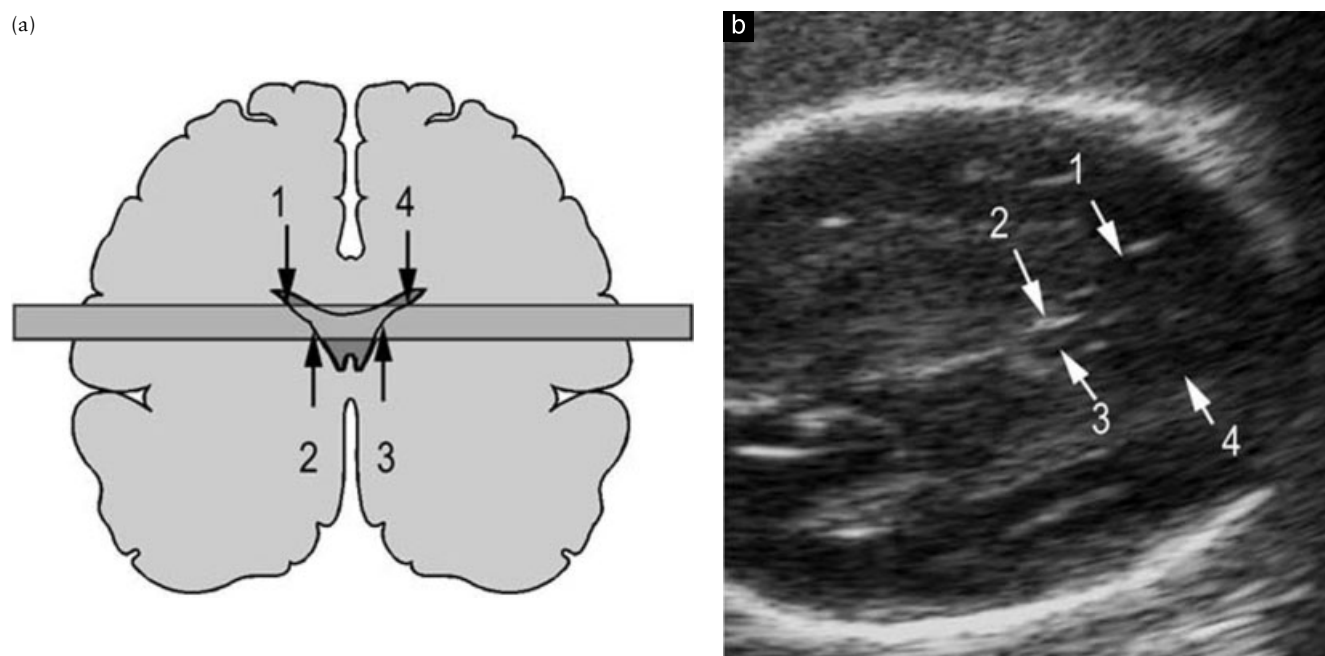


Figure 4 Schematic representation (a) and ultrasound image (b) showing how the artifact mimics the presence of the septum pellucidum. As the frontal horns have a small cavity, the ultrasound beam encounters the walls of the ventricles at different levels, causing multiple linear echoes (1,2,3,4).

REFERENCES

1. Lepinard C, Coutant R, Boussion F, Loisel D, Delorme B, Biquard F, Bonneau D, Guichet A, Descamps P. Prenatal diagnosis of absence of the septum pellucidum associated with septo-optic dysplasia. *Ultrasound Obstet Gynecol* 2005; **25**: 73–75.
2. Pilu G, Sandri F, Perolo A, Giangaspero F, Cocchi G, Salvioli GP, Bovicelli L. Prenatal diagnosis of lobar holoprosencephaly. *Ultrasound Obstet Gynecol* 1992; **2**: 88–94.
3. Barkovich AJ, Norman D. Absence of the septum pellucidum: a useful sign in the diagnosis of congenital brain malformations. *AJR Am J Roentgenol* 1989; **152**: 353–360.
4. Malinger G, Lev D, Kidron D, Heredia F, Herskovitz R, Lerman-Sagie T. Differential diagnosis in fetuses with absent septum pellucidum. *Ultrasound Obstet Gynecol* 2005; **25**: 42–49.
5. Pilu G, Sandri F, Cerisoli M, Alvisi C, Salvioli GP, Bovicelli L. Sonographic findings in septo-optic dysplasia in the fetus and newborn infant. *Am J Perinatol* 1990; **7**(4): 337–339.
6. Campbell CL. Septo-optic dysplasia: a literature review. *Optometry* 2003; **74**: 417–426.
7. Miller SP, Shevell MI, Patenaude Y, Poulin C, O'Gorman AM. Septo-optic dysplasia plus: a spectrum of malformations of cortical development. *Neurology* 2000; **54**: 1701–1703.
8. Sener RN. Septo-optic dysplasia associated with cerebral cortical dysplasia (cortico-septo-optic dysplasia). *J Neuroradiol* 1996; **23**: 245–247.
9. 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**: 549–553.