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Objectives

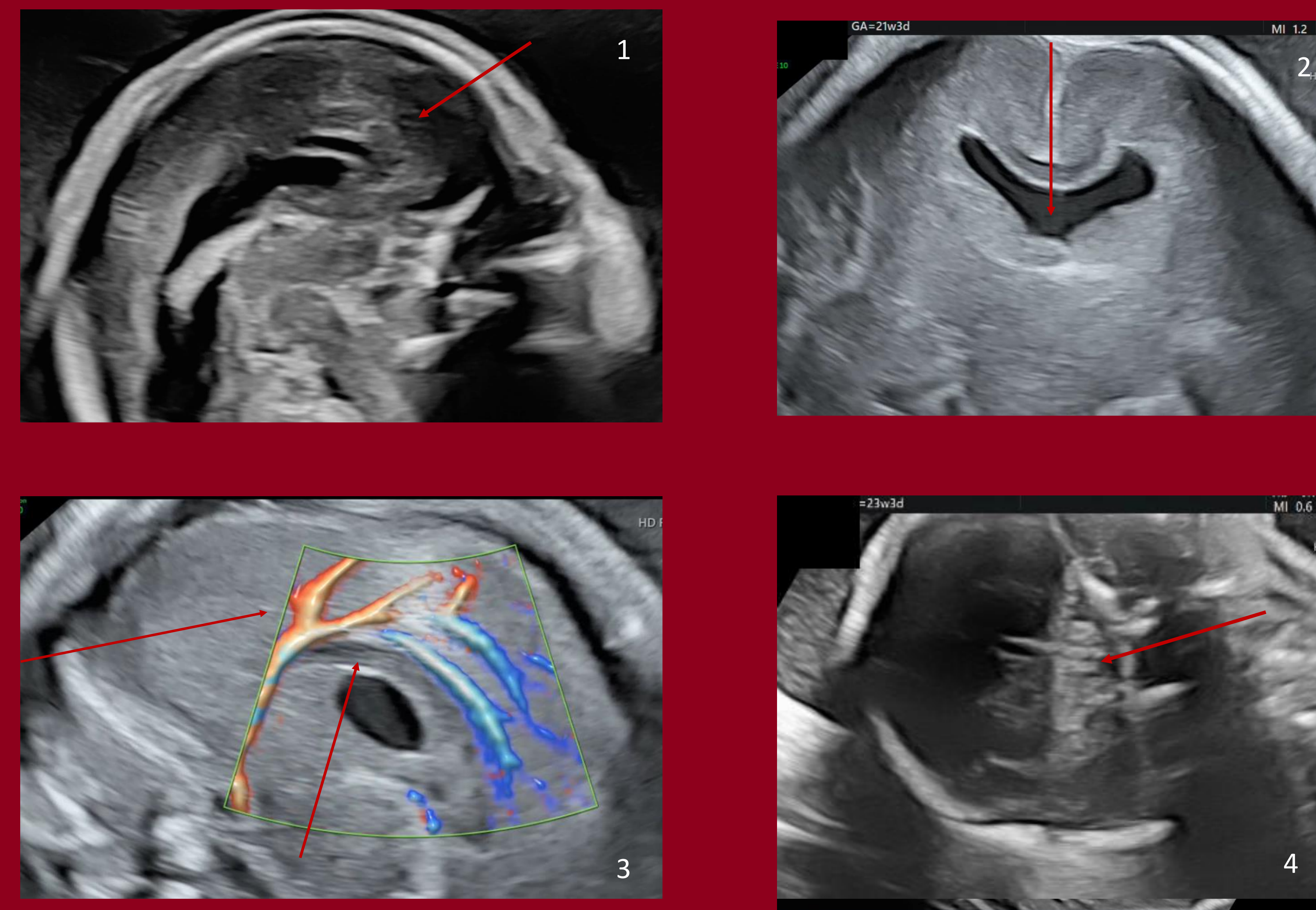
Agenesis of the septum pellucidum (ASP) refers to the incomplete formation of the thin membrane that separates the brain's lateral ventricles. It may occur on its own or alongside other brain abnormalities, often associated with conditions such as holoprosencephaly or corpus callosal agenesis. While isolated ASP generally carries a good prognosis, 14% of cases may involve additional anomalies that impact outcomes.

We present a case of isolated ASP that required a complex management approach. A 27-year-old (G2P1) pregnant woman underwent a second-trimester anomaly scan, which showed collapsed anterior horns and the absence of the cavum septum pellucidum. However, the corpus callosum, A. pericallosum, and optic chiasm were fully visualized and reduced the possibility of septo-optic dysplasia (SOD), that characterized by optic nerve hypoplasia, pituitary dysfunction, and midline brain malformations, including ASP.

First-trimester screening indicated low risks for T21, T18, and T13, though incomplete nasal bone ossification was noted. NIPT results were low risk, and the 16-week ultrasound showed no abnormalities.

The family chose to continue the pregnancy, and further evaluations—including amniocentesis and Chromosomal Microarray Analysis. MRI confirmed the absence of the septum pellucidum but did not provide details regarding the optic chiasm.

The pregnancy progressed without complications, and the baby was born at 38+1 weeks, weighing 2935 g, with an APGAR score of 9/9. At 12 months, the child had normal hearing, age-appropriate vision, and MRI confirmed isolated agenesis of the septum pellucidum.



Visualisation of collapsed anterior horns and absent cavum septum pellucidum (2), CC (3), CO (4), A. Pericallosa (3)
Distorted lamination in the frontal lobe (1)

Conclusion

A thorough understanding of fetal brain anatomy, paired with advanced ultrasound techniques, is crucial for detecting subtle structures and identifying associated anomalies. In fetuses with ASP, a detailed antenatal evaluation of the optic tract, optic nerve, and optic chiasm is essential for distinguishing SOD.