

EP06.86

Prenatal diagnosis of complex cerebral malformation with multisystem involvement: an Aicardi spectrum phenotype in a male fetus

Cardoso, I., Vicente, A.P., Lopes, D., Ladeira, G., Sá, R., Nunes, F.

24 years old primigravida, with unremarkable personal and family history, referred at 20+4 weeks' gestation.

- US at 18+4 weeks - mild ventricular asymmetry incomplete visualization of the corpus callosum, and cardiac ventricular disproportion
- US at 25+5 weeks - unilateral right ventriculomegaly and persistent suspicion of callosal agenesis, extracranial findings including left renal pyelectasis, bowel dilation, and ambiguous genitalia.
- Fetal brain MRI at 29 weeks - a complex malformation of cortical development, including asymmetric microcephaly, agenesis of telencephalic commissures, thalamic fusion, and abnormal sulcation suggestive of polymicrogyria. Additional findings - cerebellar dysgenesis, ventriculomegaly, and left microphthalmia.
- Fetal growth remained appropriate.
- Fetal echocardiography was normal and chromosomal microarray analysis revealed a normal male genomic profile.

Termination of pregnancy and fetopathological examination was performed.

The combination of severe central nervous system anomalies, ocular involvement, and normal genomic testing raised suspicion of an Aicardi spectrum disorder. However, the presence of a male fetus and atypical extracranial findings (anal imperforation and type IIIA colonic atresia) suggest an Aicardi-like phenotype or broader disorder of neuronal migration.

Conclusion

This case highlights the pivotal role of advanced ultrasound, neurosonography and fetal MRI in the prenatal characterization of complex brain malformations and underscores the importance of integrated imaging and postmortem correlation.

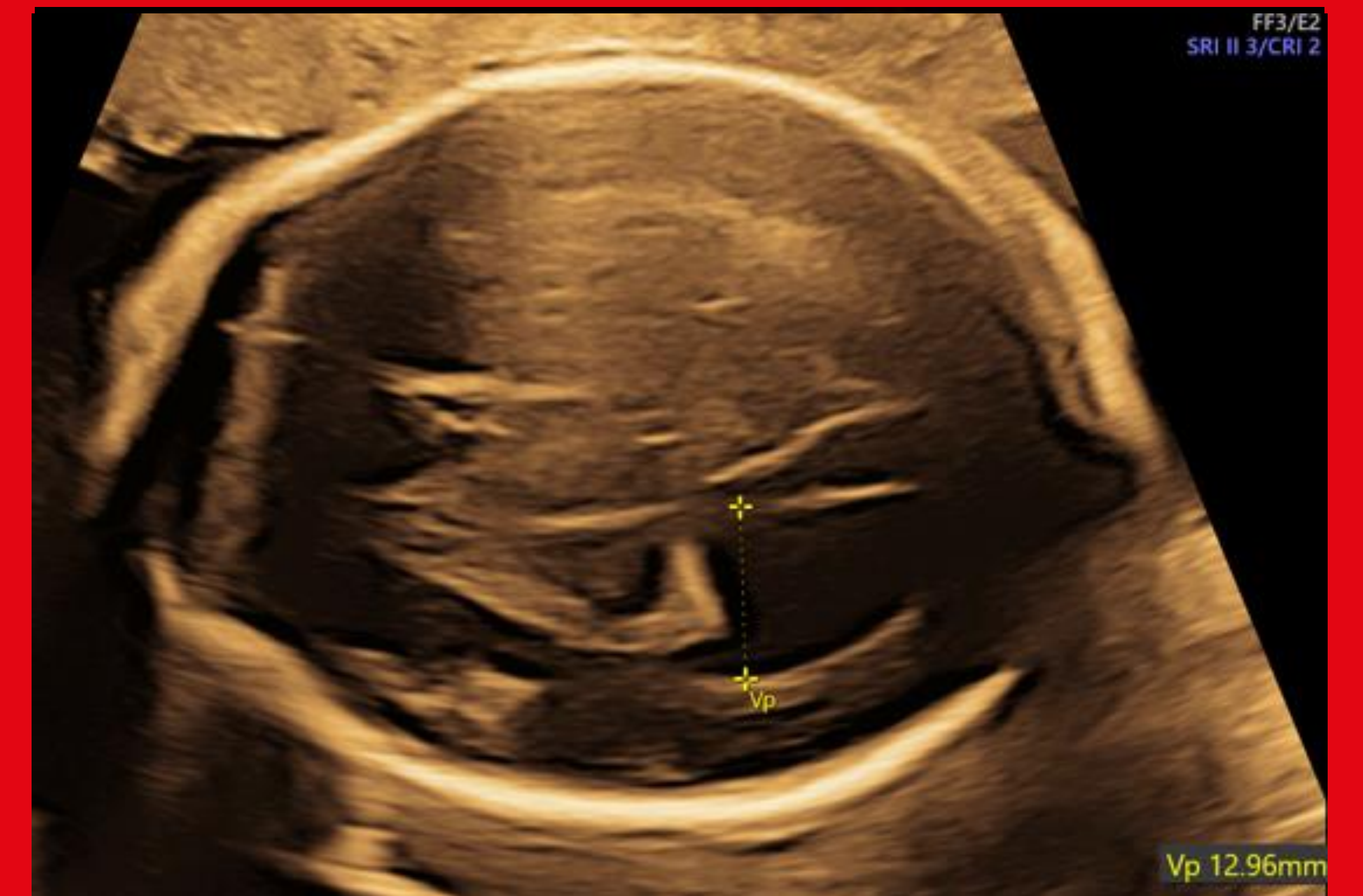


Figure 1. Ventricular asymmetry on 25w5d US

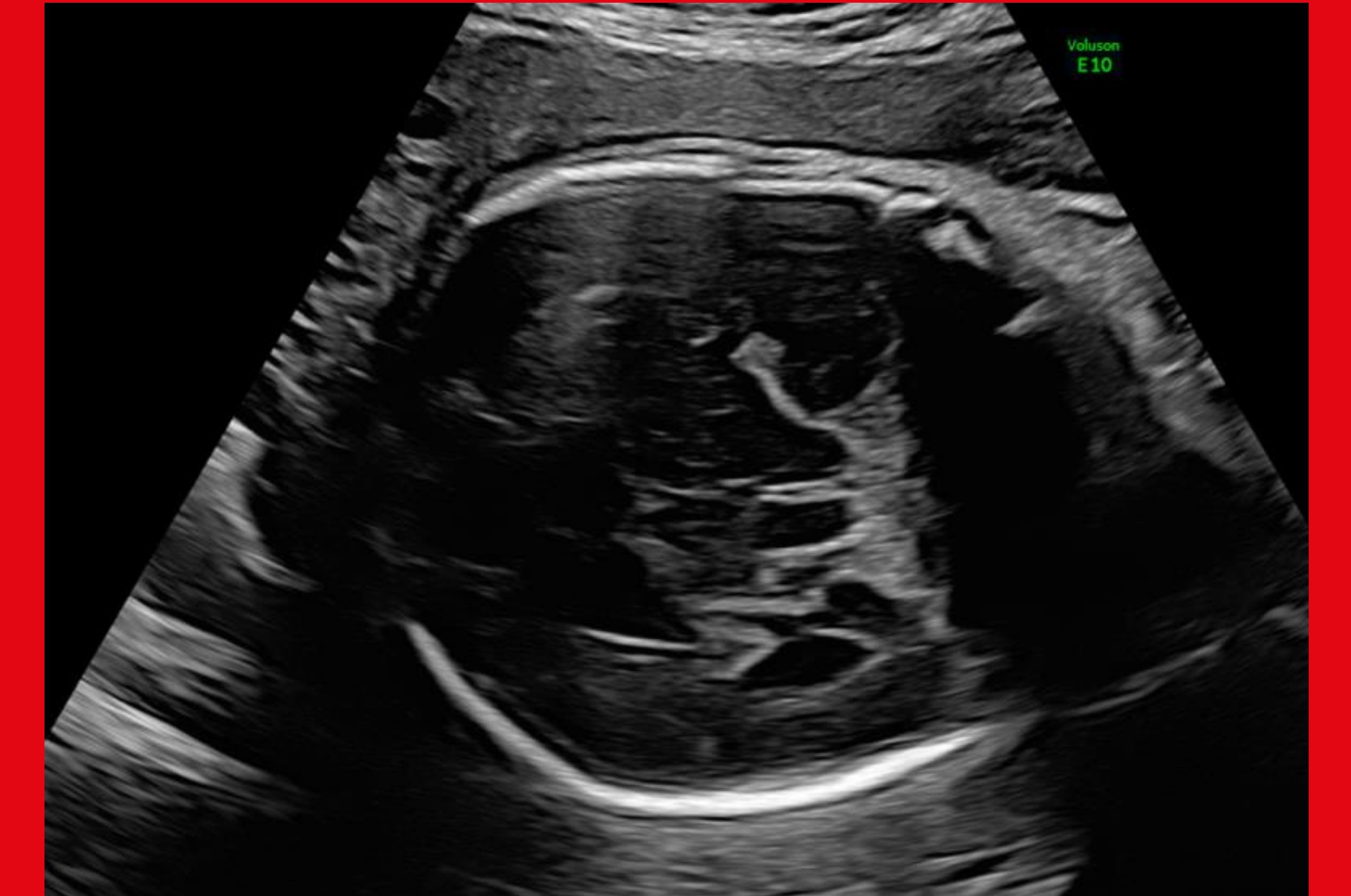


Figure 2. Ventricular asymmetry on 31w5d US



Figure 3. Fetal MRI at 29w