

Periventricular echogenicity can help diagnose Hb Bart's disease in cases of hydrops with unknown etiology

Periventricular echogenicity in fetuses with Hb Bart's related hydrops fetalis: a new sonographic marker

Sichitiu J, Vlachodimitropoulou E, Shannon P, Van Scheltema PA, Verweij EJT, Shinar S, Ryan G, Blaser S, Krishnan P, Van Mieghem T

Objective

- To explore the prevalence and etiology of a bright echogenic halo around the fetal lateral ventricles in Hb Bart's related hydrops

Methods

- Retrospective review of neurosonographic characteristics in 15 fetuses with genetically confirmed Hb Bart's related hydrops (2008-2022)
- Inclusion criteria: high-quality neurosonography data, excluding suboptimal imaging
- Maternal characteristics, pregnancy, and neonatal outcomes were collected
- Autopsy brain tissue samples re-evaluated by a neuropathologist
- Ultrasound and MRI examinations were reviewed

Results

- Bilateral periventricular echogenicity observed in all cases, persistent throughout gestation
- No other central nervous system anomalies noted
- MRI investigations (n=4): slight white matter T2 hyperintensity (n=3), mildly delayed gyration (n=1), no halo identified
- Intrauterine blood transfusions performed in 13 cases (87%)
- 12 (92%) cases were liveborn, 4 (33%) had early neonatal death
- Autopsy findings: occipital ependymal disruption and subependymal proliferation (2 cases), iron deposition (1 case)

Conclusion

- Periventricular echogenicity consistently observed in Hb Bart's hydrops, possibly indicating subependymal proliferation or iron deposition.

Figures

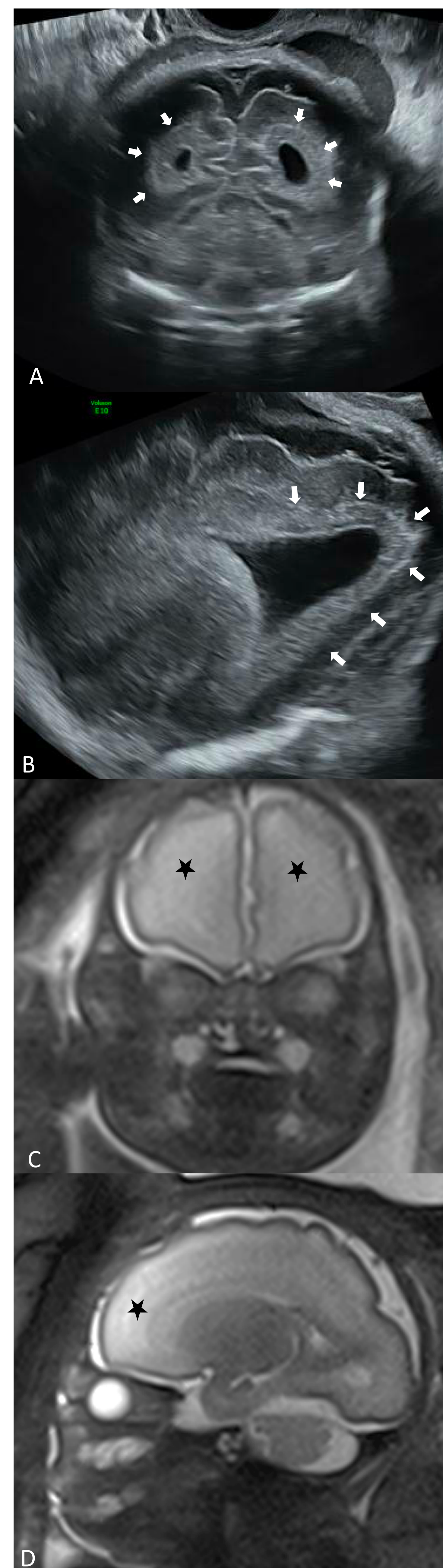


Figure 1. Bilateral periventricular echogenicity of the occipital and temporal horns (white arrowheads) at 31 weeks of gestation by ultrasound in (a) coronal plan, and (b) sagittal plane in a fetus who underwent in-utero transfusion for HbBart's. SSFSE-T2 weighted MRI (c) coronal and (d) sagittal images demonstrating diffusely increased white matter signal (black star) and exaggeration of layering at 30 weeks of gestation.