

Prospective detection and differential diagnosis of cystic posterior fossa anomalies by assessing posterior brain at 11-14 weeks

Votino C, Fanelli F, Olivieri C, De Robertis V, Boito S, Persico N, Pilu G, Volpe P
Di Venere and Sarcone Hospitals, Bari, Italy

Objective

Abnormal appearance of the posterior brain at 11-14 weeks is a marker of cerebral anomalies; in fact an increased amount of fluid, particularly when the choroid plexus of the fourth ventricle is not visible and only two brain spaces instead of 3 are seen, may indicate the presence of cystic or cyst-like posterior fossa anomalies, such as Blake's pouch cyst (BPC) or Dandy-Walker malformation (DWM). The aim of this study was to assess the role of ultrasound in identifying cystic posterior fossa anomalies at 11-14 weeks.

Methods

A prospective cohort study of fetuses with cystic appearance of the posterior fossa at 11-14 weeks was performed. In all cases and in a control group of 40 normal fetuses, the brain stem – tentorium (BST) angle was also measured. The presence or absence of cystic posterior anomalies was determined at birth or at postmortem evaluation.

Results

In the period between 2014 and 2018, 32 fetuses with an increased brainstem-occipital bone (BSOB) distance and/or failure to visualize the choroid plexus of fourth ventricle (two brain spaces), were detected. Of these, 18 were terminated in the first trimester because of associated anomalies and were excluded due to unavailable autoptic findings. Of the remaining 14 fetuses, four were later diagnosed with a DWM, eight with a BPC and two with normal brain anatomy. Two brain spaces were seen in all cases with DWM and in 2/8 cases with BPC. Both BSOB measurement and BST angle were significantly altered in fetuses with DWM, BPC compared to controls ($p < .0001$). The BSOB z-scores of fetuses with DWM and BPC were consistently found to be + 3 and + 1.7 or more, respectively. The BST angle z-scores were consistently – 5 or less and – 0.1 or less respectively.

Conclusion

Our study confirms that sonography of the posterior brain at 11-14 weeks allows identifying cystic posterior fossa anomalies. A large BSOB predicts DWM or BPC. The presence of 2 brain spaces and a small BST angle are significantly correlated with the presence of DWM.