Fetal Diagnosis of Posterior Fossa Abnormalities is Enhanced by Fetal MRI

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BACKGROUND: Posterior fossa anomalies can be diagnostic dilemmas during the fetal period. The prognosis for different diagnoses of the posterior fossa varies widely, and outcomes range from normal to severe neurologic disability.

OBJECTIVE: Our aims were to determine whether fetal MRI and neurology consultation lead to alternate diagnoses for maternal-fetal dyads referred to a fetal neurology program due to concern for a fetal posterior fossa anomaly and to determine how often the postnatal evaluation differed from fetal diagnosis.

METHODS: This is a retrospective study of cases referred to the Fetal Medicine Institute at Children’s National, Washington, DC, from January 2012 to June 2018. The following referral diagnoses were included: Dandy-Walker continuum (DWC), cerebellar hypoplasia (CH), vermis hypoplasia (VH), Blake’s pouch cyst (BPC), mega cisterna magna (MCM), or “other” posterior fossa anomaly (PF). A pediatric neuroradiologist reviewed all fetal and postnatal imaging studies blinded to initial clinical report.

RESULTS: In total, 187 cases were referred for fetal posterior fossa anomalies, underwent fetal MRI, and had a neurology consultation. Fetal MRI and neurology consultation resulted in a change in fetal prognosis in 124 (66%) cases; 37 (20%) had a normal brain, 62 (33%) had better prognosis, 24 (13%) had a worse prognosis, and 64 (34%) had no change in prognosis. The most common referral diagnosis was DWC (81, 43%), but only 20 (25%) patients had DWC by fetal MRI; 10 (12%) had a normal posterior fossa; 49 (60%) had a diagnosis with a better prognosis; and 2 (2%) had a diagnosis with a worse prognosis. Postnatal diagnosis was obtained for 54/130 (41%) live-born infants. Of 13 cases of fetal BPC, 7 (54%) remained stable, 2 (15%) normalized, and 4 (30%) had a different diagnosis on postnatal imaging. Of 6 cases of CH, only 1 had a change in prognosis postnatally. Seven cases of fetal DWC had postnatal imaging; 5 (71%) remained stable; 1 (14%) had Joubert syndrome; and 1 (14%) had a BPC. Of 7 cases of fetal MCM, 2 (29%) normalized, 4 (57%) remained stable, and 1 (14%) had VH postnataally. Lastly, 13 cases of VH had postnatal imaging; 7 (54%) remained stable, 3 (23%) normalized, 1 (8%) had BPC, and 2 (15%) had more severe findings.

CONCLUSIONS: Fetal MRI and neurology consultation improved the accuracy of fetal diagnosis of posterior fossa anomalies over that provided by prenatal US only, particularly when there was concern for DWC. Additional information or change from referral diagnosis may have important repercussions on pregnancy management and infant neurologic care.