Characterization of Features of PHACE Syndrome on Fetal MRI and Natural History of Postnatal Management

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Background: PHACE syndrome (*p*osterior fossa defects, *h*emangioma, *a*rterial anomalies, *c*oarctation of the aorta and cardiac defects, *e*ye abnormalities) is a constellation of findings of unknown etiology. Diagnosis of this condition requires presence of large segmental hemangioma with at least one major criteria, including posterior fossa brain anomalies (such as Dandy-Walker anomalies) or other hypoplasia or dysplasia of the mid or hind brain. Early diagnosis of this condition is critical to minimizing long term effects of these congenital abnormalities.

Objective: Characterize fetal MRI findings that indicate the possibility of PHACE syndrome and implications of prenatal findings for postnatal management.

Methods: Here we report nine patients with ultimate diagnosis of PHACE syndrome who were found to have abnormal brain findings on prenatal anatomy ultrasounds, which were further characterized by fetal MRI. Patients were seen in our Center for Fetal Diagnosis and Treatment from 2007 to 2016. Electronic health records were reviewed to determine the additional work-up for PHACE syndrome, including postnatal imaging, management, and postnatal outcomes.

Results: Anatomy scans during pregnancy demonstrated posterior fossa anomalies. Fetal MRI studies were significant for unilateral cerebellar hypoplasia or cystic lesions in all patients. Other intracranial findings included asymmetric Meckel's cave and ventricular abnormalities. Postnatal diagnosis was confirmed by the presence of large facial segmental hemangiomas in all patients. Initial postnatal brain imaging was performed within the first several weeks of life, demonstrating evidence of unilateral hemangiomas ipsilateral to the cerebellar lesions. Children were treated with prednisone or propranolol within the first two months of life, maximizing potential of improved cosmetic outcome with early therapy. When available, repeat postnatal brain MRIs were reviewed, some demonstrating involution of previously seen hemangiomas. Multiple patients had other characteristics of PHACE syndrome, including cardiovascular and ocular abnormalities.

Conclusions: Unilateral cerebellar cystic or dysplastic lesions found on fetal MRI should raise suspicion for diagnosis of PHACE syndrome. This should prompt further evaluation *in utero,* including fetal echocardiogram, as presence of cardiac or aortic abnormalities strengthens the likelihood of this diagnosis. In addition, early postnatal evaluation by Dermatology, Cardiology, Neurology, and Ophthalmology are critical for effectively managing patients with PHACE syndrome.