Myelomeningocele Diagnosis

It is diagnosed with prenatal blood work, amniocentesis, physical exam, and ultrasound. An MRI is sometimes needed to get more information and guide care.

Abnormal intracranial findings are often detected at mid-trimester ultrasound (US) in fetuses with Myelomeningocele (MMC). It is unclear whether these findings constitute a spectrum of the disease or are an independent finding, which should contraindicate fetal surgery.

Objective: To ascertain the spectrum and frequency of US detected cranial findings in MMC fetuses.

Search strategy: MEDLINE, Embase, Web of Science and CENTRAL search, January 2000-June 2020.

Selection criteria: Study reporting incidence of cranial US findings in consecutive cases of second trimester MMC fetuses.

Data collection and analysis: Publication quality was assessed by NOS and modified NOS. Meta-analysis could not be performed due to high clinical diversity and study heterogeneity.

Results: 14 cranial US findings were reported in 15 studies. Findings in classic Chiari II malformation (CIIM) spectrum included posterior fossa funnelling (96%), small transcerebellar diameter (82-96%), 'banana' sign (50-100%), beaked tectum (65%) and 'lemon' sign (53-100%). Additional cranial findings were small BPD and HC (<5th centile; 53% and 71%, respectively), ventriculomegaly (45-89%), abnormal pointed-shape of the occipital horn (77-78%), thinning of the posterior cerebrum, perinodular heterotopia (11%), abnormal gyration (3%), corpus callosum disorders (60%) and midline interhemispheric cyst (42%).

Kunpalin et al. identified 14 cranial findings by second trimester US in MMC fetuses. The relatively high incidence of these findings and their unclear prognostic significance might not contraindicate fetal surgery in the case of normal fetal genetic testing. Some cranial findings however may independently impact postnatal outcome. Long-term detailed follow up is required to investigate this [1].

1)

Kunpalin Y, Richter J, Mufti N, et al. Cranial findings detected by second trimester ultrasound in fetuses with myelomeningocele: a systematic review [published online ahead of print, 2020 Sep 14]. BJOG. 2020;10.1111/1471-0528.16496. doi:10.1111/1471-0528.16496