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The role of fetal MRI for evaluating brain pathology in fetuses with abnormal chromosomal microarray analysis

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Objective

Chromosomal microarray analysis (CMA) is rapidly increasing for fetal genetic screening, resulting in an increasing demand for fetal MRI in search of brain abnormalities. Our objective was to assess the value of fetal brain MRI (fbMRI) over detailed fetal sonography for detection of brain pathology in fetuses with CMA abnormalities.

Methods

Records of pregnant women referred to fbMRI due to CMA abnormalities were retrospectively reviewed for imaging findings on fbMRI and brain ultrasonography and for CMA results. CMA findings were classified as either high probability (i. e. associated with known clinical syndrome) or variant of unknown significance (VOUS). Patients with abnormal karyotype were excluded from our study.

Results

Fifty fbMRI studies were performed following abnormal CMA results between 2012-2018 (1.5% of all studies). In 5 fetuses (10%), novel findings were found on fbMRI, i. e. white matter T2-hyperintensities (3), dysplastic corpus callosum (1), facial abnormalities (1) and delayed gyration pattern (1). Of these, only one CMA was classified as high probability and 4 were classified as VOUS. In 13 fetuses (26%), fbMRI findings matched known sonography findings, i. e. dysplastic corpus callosum, enlarged cisterna magna, asymmetric ventricular size or mild ventriculomegaly. In 32 fetuses (64%), fbMRI was found to be normal. None of the fetuses showed cortical migration or organization abnormalities.

Conclusion

FbMRI confirmed known sonographic findings and discovered novel findings in fetuses with CMA abnormalities. The most common novel findings were non-specific white matter signal hyperintensities. Further studies are needed to quantify and correlate these imaging findings with long term outcomes.