Quantitative MRI analyses of regional brain growth and cerebral sulcal development in living fetuses with isolated ventriculomegaly and Down syndrome.

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OBJECTIVES: Fetuses with isolated cerebral ventriculomegaly (IVM) and Down syndrome (DS) share common neuroimaging (fetal sonographic and magnetic resonance imaging (MRI)) features of mild to moderate ventriculomegaly despite distinct neurodevelopment differences after birth. We hypothesized that fetuses with IVM and DS have distinct brain development features that can be detected with non-invasive, quantitative fetal (qf) MRI.

METHODS: Pregnant women who had fetuses diagnosed with IVM (n=13, 26.1±4.8 week of gestation, mean±SD) or DS (n=10, 29.1±4.2) were recruited prospectively. Healthy pregnant women carrying fetuses without apparent medical conditions were recruited as controls (n=11, 25.1±5.3). IVM was diagnosed when fetal MRI showed ventriculomegaly without associated anomalies. Fetuses with DS were recruited when either cell-free DNA screens were positive or karyotype/chromosomal microarrays were diagnostic. Conventional fetal MR images were processed for post-acquisition qfMRI analyses, including regional volumetric analyses and Similarity Index based sulcal pattern matching/similarity analyses. Statistical analysis was performed using non-linear regression model to compare fitted to regional growth curves between condition and two-sample t-test, with significance set at 0.05.

RESULTS: In fetuses with DS, fitted growth curves of cerebellar hemispheres and
subcortical parenchyma (white matter and basal ganglia) showed smaller growth curves compared to controls. In contrast, fetuses with IVM had significantly larger growth curves in cortical plate and subcortical parenchyma compared to the controls. In the sulcal pattern analysis, fetuses with DS had altered absolute cerebral sulcal positions compared to the control in the second and third trimester. Surface analysis of IVM is ongoing.

CONCLUSIONS: qfMRI detected distinct regional growth curves in the second and third trimester fetuses with IVM and DS. Fetuses with DS have distinct sulcal development compared to controls.

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