OC06.03
Fetal MRI findings in a cohort of 26 cases of prenatally diagnosed CHARGE syndrome

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Objectives: “CHARGE syndrome” (CS) is a multifaceted syndrome carrying a poor prognosis. Although the syndrome is well characterised in the postnatal period, the prenatal diagnosis is difficult and fetal anomalies that may raise the hypothesis of CS are not comprehensively described. This study was therefore undertaken to describe the anomalies picked up at MRI examination in our series of CHARGE syndrome.

Methods: This was a retrospective study of 26 fetuses that underwent MRI and had a confirmed diagnosis of CS, as proven by histopathological and/or neonatal examinations and/or presence of the CHD7 gene mutation. The sensitivity of the most important anatomical anomalies as based on fetal MRI, as compared to postnatal and fetopathological examinations (gold standard) were assessed.

Results: Among the 26 confirmed cases, CHD7 mutation was found in 20/26 cases (77%). The three most frequent MR anomalies confirmed histopathological and/or neonatal examinations were: arhinencephaly in 100% (26/26), dysplasia of the semicircular canals agenesis (SCA) in 100% (24/24) and posterior fossa anomalies in 100% (22/22). Our study also revealed short petrous bones with a particular triangular shape in 24/24 cases. This appears as another reliable sign for SCA, which has not been previously described in fetal MRI in patients with CS to our best knowledge. Other findings included external ear anomalies in 36% (9/25), cleft lip palate (9/9), ventriculomegaly (VMG) (6/6), Short Corpus Callosum (3/5), Ocular Asymmetry in 36.6% (4/11), choanal atresia in 13% (3/23), thymus anomalies in 85.7% (12/14), genital Anomalies in 50% (3/6), renal anomalies in 50% (4/8) and esophageal anomalies in 50% (1/2).

Conclusions: Our results highlight that fetal MRI may play a major role in the prenatal diagnosis of CS when suspected following referral ultrasonography. Our study highlights the excellent sensitivity of fetal MRI to diagnose the most frequent pathological findings and reveals that short petrous bones could also be easily used as a proxy for semicircular canals agenesis.

OC06.04
Application of prenatal ultrasound combined with fast MRI in the diagnosis of fetal agenesis of corpus callosum

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Objectives: To evaluate the diagnostic value of fetal neurosonography combined with fast intracranial magnetic resonance imaging (MRI) in fetal agenesis of corpus callosum (ACC). Malformations of the central nervous system (CNS) together with fetal heart defects are the most common congenital anomalies. The introduction of three-dimensional (3D) ultrasound for evaluating fetal brain structures has made it possible to diagnose pathological findings of the CNS with increasing accuracy.

Methods: Between January 2014 and December 2017, 117 fetuses, mean gestational age of 29.3 weeks (range 22-40 weeks), with mild ventriculomegaly and diaphragmatic hernia reduction or disappearance diagnosed at prenatual sonography were included in this study. All fetuses underwent intrauterine MRI according to the following protocol: images along the three orthogonal plane according to the longitudinal axis of the mother, and subsequently three orthogonal planes were acquired according to the fetal brain. Postnatal physical examination and diagnostic imaging were the standard of diagnosis. The sensitivity of each modality for detecting anomalies was calculated and compared by the McNemar test.

Results: In 117 fetuses, mean axial diameter of the lateral ventricle was 11.7 mm (range 10-17 mm), 69/117 fetuses (58.97%) showed normal morphology of the lateral ventricles, and the entire corpus callosum was visualised. In 48/117 fetuses (41.03%) were confirmed with ACC. 40 cases were diagnosed by detailed fetal neurosonography, 45 cases by intrauterine MRI and their sensitivity were 83.33% and 93.75% respectively. There was a statistically significant difference in the sensitivity for detecting anomalies between US and MRI (P <0.05). The sensitivity of combined diagnosis was 95.83%, with statistically significant difference to US alone (P <0.01).

Conclusions: Detailed fetal neurosonography combined with fast MRI can accurately diagnose fetal ACC, especially improve the detection rate of partial corpus callosum agenesis, and provide a reliable basis for clinical diagnosis.

OC06.05
Prenatal cerebellar growth trajectory as a novel biomarker for body composition of the newborn

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Objective: In our previous study, we showed a strong correlation between the periconceptional maternal body mass index (BMI) and growth trajectories of the embryonic and fetal cerebellum. We hypothesise that the growth trajectory of the fetal cerebellum is a determinant in the developmental origin of adiposity in the offspring. Here we investigate associations between the fetal trans cerebellar diameter (TCD) and neonatal body composition, and fetal thigh volume (TVol).

Methods: The study population is selected from the Rotterdam Periconception cohort conducted at the Erasmus MC. Two- and three-dimensional ultrasound scans are performed at 22, 26 and 32 weeks gestational age (GA). Air-displacement plethysmography is used to measure neonatal body composition at 42 weeks calculated from the last menstrual period. Cross-sectional analysis using linear regression and longitudinal analyses are performed using linear mixed models to estimate random intercept (RI) and random slope (RS) of the fetal TCD growth trajectories. Linear regression analysis of the RI and RS is used to investigate associations between TCD growth trajectories and body composition.

Results: 82 mother-child pairs are included for analysis. The cross-sectional analyses showed a positive association between TCD and fat free mass (FFM) at 22 (β=1.03; p<0.01), 26 (β=1.18; p<0.01) and 32 (β=0.76; p=0.01) weeks GA. Adjustment for maternal BMI, maternal age, fetal gender, parity, smoking and mode of conception shows comparable results. The longitudinal analysis shows a positive association between TCD growth trajectories and FFM (β=1.003; p=0.02). A positive association was shown between TCD and TVol at 26 (β=0.67; p<0.01) and 32 (β=1.221; p=0.02) weeks GA.

Conclusions: This data demonstrates that the fetal TCD is associated with TVol and neonatal FFM. Further research should elucidate whether the fetal TCD can be used as a novel marker to predict adiposity in offspring. Since TCD is a standard