

so strong ($r = 0.62$, $p < 0.05$). The mean neonatal abdominal circumference was 34.71 centimeters and also did not correlate well with AC estimated with USG ($r = 0.54$, $p < 0.05$). The mean neonatal head circumference was 36.19 centimeters and did not correlate with HC ($r = 0.36$, NS). 11 patients already had Cesarean section, 14 patients presented gestational diabetes, 3 – pregestational diabetes, 4 – pregnancy induced hypertension, 6 – breech presentation.

Conclusion: Macrosomia as an indication for Cesarean section is quite often overdiagnosed and depends not only on objective estimation with USG scan, but on human factor and obstetric history.

P02.99

Assessment of fetal lung maturity in a non-invasive fashion

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Objective: We sought to test the hypothesis that several patterns of fetal breathing movements (FBMs) i.e., abdominal wall movements (AWm), thoracic wall movements (TWm), nasal fluid flow velocity waveforms (NFFVW) may be ascertained noninvasively by ultrasound and correlated with biochemical assessment of fetal lung maturity (FLM).

Material and Methods: We prospectively enrolled 208 high-risk pregnancies in which a complete US study of FBMs and FLM tests were performed. US-FLM was defined as the presence of regular NFFVW detected by pulsed Doppler and spectral analysis, or irregular NFFVW synchronous with TWm detected by M-mode. An US guided amniocentesis was performed in order to collect amniotic fluid (AF) and FLM was evaluated by L/S (lecithin/sphingomyelin) determination, presence phosphatidylglycerol (PG) and lamellar body (LBs) count. At the end of the study diagnostic accuracy of US-FLM was compared with that L/S/well as neonatal outcome.

Results: Diagnostic accuracy for US evaluation of FLM was as follow: sensitivity: 89.6%; specificity: 85.7%; PPV 92.8%; NPV: 80%. Diagnostic accuracy of FLM tests was as follow: sensitivity: 100%; specificity: 51.7%; PPV 100%; NPV: 50%.

Conclusion: Presence of regular NFFVW or irregular NFFVW and TWm correlate accurately with conventional FLM tests and predict neonatal outcome with acceptable accuracy. We suggest that this non-invasive procedure may be helpful to assess FLM in a noninvasive fashion, particularly under certain circumstances such as PPRM, logistic difficulties or patient refusal amniocentesis.

P03: FETAL CENTRAL NERVOUS SYSTEM

P03.01

Prenatal diagnosis of closed spinal dysraphism

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Objective: To observe ultrasound features seen in 14 fetuses with closed spinal dysraphism.

Methods: Clinical records from the spina bifida clinic at BC Children's Hospital were reviewed to identify 12 children born with radiological and/or surgical evidence of spinal lipoma. In all 12 cases, prenatal ultrasounds had been performed and images were available for retrospective review. 2 additional cases were identified prospectively on prenatal ultrasound and MRI with other forms of closed spinal dysraphism.

Results: In all 14 cases of closed spinal dysraphism, the posterior fossa was normal on prenatal sonography. There was no evidence

of the Chiari II malformation, and there were no cases with ventriculomegaly. In six children with spinal lipoma diagnosed after birth, no prenatal sonographic abnormalities were seen. In 8 out of 14 cases of closed dysraphism, prenatal sonographic abnormalities were identified, including: cystic and/or solid mass dorsal to the spine (5 cases); vertebral abnormalities (4 cases); foot deformities (2 cases); diastematomyelia (1 case); and abnormally low conus in one fetus with cloacal dysgenesis. MSAFP was negative in 3 cases and elevated in 1 case; AFAFP was elevated in 1 other case.

Conclusion: Eight out of 14 fetuses with closed dysraphism had prenatal sonographic findings of soft tissue masses, vertebral and/or foot abnormalities, or low conus. Spinal lipoma was detected antenatally in 6 out of 12 cases. None of the 14 cases showed any sonographic evidence of a Chiari II malformation, ventriculomegaly, or other intracranial abnormality. Due to the relative subtlety of the prenatal findings, the diagnosis of closed spinal dysraphism is considerably more difficult than it is with open dysraphism.

P03.02

The importance of genetic counselling in holoprosencephaly

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Background: Holoprosencephaly is the most common structural malformation of the forebrain with a prevalence of 1:8000 in the second trimester. In up to 40% of cases the fetus is aneuploid, commonly trisomy 13 and the recurrence risk is low. In euploid fetuses however, mutations in holoprosencephaly genes such as sonic hedgehog (SHH) are known to occur.

Objectives: To review management and determine the etiology in holoprosencephaly cases seen in our unit.

Methods: All cases of holoprosencephaly referred to our unit in the last 14 years were ascertained from computerized records. We examined FMU, neonatal, genetic and pathology records to determine pre- and postnatal management, outcome and genetic follow-up.

Results: 38 cases of holoprosencephaly presented in 38 women, of which 53% were lobar, and the remainder were lobar (24%) or semilobar (17%). Fetal karyotyping was performed at maternal request in 82% of cases ($n = 31$) and was abnormal in 52% of karyotypes (14 cases trisomy 13, 1 case ring chromosome 13 and 1 case trisomy 18). Parents were referred for genetic counselling in 9 of the 15 euploid fetuses. There was a recurrence of holoprosencephaly in 3 of the euploid cases, (20% of normal karyotypic fetuses). In one case, there were 5 affected pregnancies diagnosed at 20 weeks of gestation, and then at 12–14 weeks. The parental karyotypes were normal, but molecular analysis showed a mutation in the SHH gene. In two cases diagnosed at 22 and 24 weeks of gestation, there was one previously affected pregnancy, diagnosed at 27 weeks and birth respectively; a mutation was confirmed in the VAX2 gene in the former case.

Conclusions: In this series there was a 20% recurrence risk for parents with a fetus with holoprosencephaly and a normal karyotype. Genetic review for parental examination, MRI scanning and mutation analysis is important to accurately determine the recurrence risk. 1st-trimester ultrasound scanning is advised to detect recurrence early in gestation.