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Association of single umbilical artery with aneuploidy, congenital malformations and perinatal outcome: Findings in 49,698 unselected pregnant women

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Objectives: To determine the possible association between single umbilical artery (SUA) in the second trimester and the incidence of fetal aneuploidy (T21,13&18) and other malformations. To report the incidence of associated perinatal mortality.
Methods: Colour flow imaging undertaken between20–23 weeks of gestation during routine scan in 49,698 cases in two tertiary referral centres between the period of 2000–2007. Referred cases for second opinion have been excluded as well as Fetuses diagnosed with single umbilical artery and no other anomalies or markers (isolated SUA). Retrospective analysis of associated aneuploidy, malformations and intrauterine deaths (IUD).
Results: SUA was diagnosed in 219/49698 (0.44%). Among the 219 fetuses with SUA, 156 (71.2%) had no associated markers or anomalies and 63 (28.2%) had associated findings. 31/63 (49.2%) fetuses had only soft markers (one or two). 32/63 (50.8%) had associated anomalies (at least one structure defect). 8/63 (12.7%) fetuses had aneuploidy (4 cases of T18, 3 cases of T13, one case with unbalanced translocation 18-Y).
55/63 (82.5%) fetuses had structure anomalies of which 13/63 (20.6%) had multiple anomalies (> 3 anomalies).14/63 (22.2%) had cardiac anomalies, 8/63 (12.7%) had CNS anomalies, 6/63 (9.5%) had renal anomalies (3cases of hydronephrosis, 2cases of renal agenesis and one case of multicystic dysplastic kidney). 4/63 (6.3%) had cleft palate, 4.8% had skeletal anomalies, 3.2% had NTD, and 2/63 (3.2%) had diaphragmatic hernia.
Perinatal mortality rate reported in 14/63 (22.2%). 10 fetuses had intrauterine death while 4 fetuses were terminated.
Conclusion: This case series of unselected population indicate high association of aneuploidy in particular T18 and T13 and many other structure anomalies. No case of T21 was detected. We conclude that second trimester sonographic detection of SUA and other findings is not associated with increased risk of T21.

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Complicated gastroschisis - can prenatal ultrasound predict outcome?

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Objectives: Fetal gastroschisis is a well described prenatal and postnatal finding. Generally counseling of prospective parents is optimistic and stresses a good outcome. There is increasing concern in the pediatric literature regarding poor outcome in infants diagnosed with *complicated* gastroschisis. This includes infants with bowel atresias, perforations, malrotations and obstructions that lead to much poorer outcome and significant long term morbidity.
Our goal was to correlate prenatal ultrasound findings with neonatal outcomes in an attempt to distinguish complicated and uncomplicated gastroschisis prenatally.
Methods: Our study is a retrospective case series evaluating 83 cases of fetal gastroschisis diagnosed and treated both pre- as well as postnatally at two hospitals associated with our institution from 2001 to 2007. Data was collected by chart review.
Results: Of 83 infants with gastroschisis, 64 (77%) were classified as uncomplicated, 19 (23%) as complicated. There was no difference in both groups in gestational age at delivery (36.1 vs. 35.3 weeks), mode of delivery (54% cesarean section) and male to female ratio. While there were significant differences in multiple postnatal findings, eg days on TPN (30 vs. 86 days, $P < 0.0001$), ventilator dependency (9 vs 15 days, $P = 0.01$), age at discharge from the NICU (40 vs 100, $P < 0.0001$), we were unable to show significant difference in prenatal ultrasound characteristics. When fetal bowel appearance was described there was a trend toward greater bowel dilation in fetuses that presented with complicated gastroschisis postnatally (47% complicated vs 28% uncomplicated). Defect size, presence of peristalsis and measurement of bowel dilation were inconsistently documented.
Conclusions: The diagnosis of complicated gastroschisis remains a postnatal one; future studies focusing on ultrasound characteristics of fetal bowel appearance may aid in the prenatal differentiation of complicated and uncomplicated gastroschisis.

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Prenatal diagnosis of omphalocele with normal karyotype: A real antenatal dilemma

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Objectives: To analyze the main characteristics and outcome of euploid fetuses with omphalocele (OC).
Methods: Retrospective study from 1990 to 2007. Inclusion criteria: prenatal diagnosis of OC, normal karyotype, ongoing pregnancies > 22 weeks, complete follow-up. Prenatal and postnatal data were obtained from our database and clinical charts. Postnatal follow-up for at least 3 months was available for all the survivors.
Results: 103 OC were diagnosed prenatally and 28 met the inclusion criteria (27%). Mean gestational age at diagnosis was 22 weeks. Extracorporeal liver (ECL) was observed in 11 (39%). The OC was an isolated finding in 23 (82%) (group A) and additional major structural anomalies were found in 5 (18%) (group B). Postnatal exams revealed a genetic syndrome in 9 patients (32%, 8 with Beckwith-Wiedemann syndrome (BW) and 1 with Sphrintzen-Goldberg syndrome) and an associated malformation in 1 (4%, imperforate anus with recto-vaginal fistula). The overall survival rate was 82%. This rate was higher in group A (96%, 22/23) than in group B (20%, 1/5). There were 2 stillbirths (both in group B) and 3 neonatal deaths (1 in group A). Among syndromic patients, most (6/9) had sequelae due to the syndrome and one of them died in the neonatal period. When comparing second-trimester findings in fetuses with and without genetic syndrome, no differences were observed in the rate of ECL, or in the mean size of the OC, nor in the rate of associated malformations. Similarly, we found no differences in late-onset findings such as the rate of polyhydramnios or in the newborns' weight.
The final rate of OC in euploid fetuses actually isolated was 50% (14/28).
Conclusions: Prognosis of euploid fetuses with OC remains controversial. In our experience, many of these patients have associated conditions that are not totally manifest in fetal life, therefore making their prenatal detection very difficult. This information must be taken into account at the time of prenatal counselling.

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Management and outcome of isolated abdominal calcifications

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