

decided to follow up the pregnancy during two weeks and perform the cordocentesis in this time, karyotype was normal. At 26 weeks we found progressive changes in fetal anatomy: heart herniation became larger, left lobe of liver began to protrude, the absence of the sternum and cartilaginous parts of the ribs was confirmed. Patient was offered pregnancy termination because of the impossibility of surgical correction. Radiography, CT and morphology of abortus confirmed ultrasound diagnosis of isolated absence of the sternum and cartilaginous parts of the ribs.

P02.39

Nuchal translucency in screening for congenital heart defects in chromosomally normal fetuses

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Objective: Assess the accuracy of the nuchal translucency (NT) measurement between 11 weeks and 13 weeks and 6 days of gestation as a sonographic marker to screen for congenital heart defects (CHD).

Methods: Multicentric retrospective study, analyzing single pregnancies from euploid fetuses. NT measurement was performed in the first trimester, when fetuses had from 45 to 84 mm of crown-rump length (CRL), according to the criteria established by the Fetal Medicine Foundation. Different cut-off point were used to define enlarged NT (95th and 99th centiles, 2.5, 3.0 and 3.5 mm). Cardiac anomalies were evaluated either by echocardiography or by clinical examination during the first month of life.

Results: 3,664 pregnancies were analyzed and 20 newborns had CHD diagnosed until the first month of life (prevalence of 0.55%). The median NT of the fetuses with CHD was 1.70 mm and 1.60 mm for fetuses without CHD. No significant difference was found (Mann-Whitney test, $p > 0.05$). The sensitivity of NT in detection of CHD varied from 15 to 20%, with a range of false positive probability from 86.4 to 97.9%, depending on the cut-off point used. However, the odds ratio was high, when compared to the classic indications of echocardiography, ranging from 4.7 to 33.7 according to the cut-off point.

Conclusion: In spite of the low sensitivity of the test, enlarged NT is an important risk factor for CHD and should be used in prenatal screening for CHD.

P02.40

The influence of maternal age on the association of an isolated fetal intracardiac echogenic focus and fetal aneuploidy

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Background: Fetal intracardiac echogenic foci (IEF) have frequently been reported to be associated with fetal aneuploidy. The overall incidence of IEF ranges from 0.17% to 20% varying with background risk. Data on this association remains inconclusive, while an association was shown in initial studies, recent studies differentiating between different risk groups could not reproduce those findings in low risk populations.

Objective: To evaluate the incidence of IEF and its association with fetal aneuploidy in patients referred to our prenatal department for 2nd-trimester genetic sonogram.

Method: We performed a retrospective chart analysis of all patients with an IEF at the prenatal department of the University Hospital, Mainz from 1999-2005.

Results: An IEF was found in 89 of a total of 10527 screened patients (0.85%). 23 out of those 89 patients opted for invasive diagnostics

(26%). An isolated IEF was found in 73 cases (82%), the majority being located in the left ventricle (73%). 7 fetuses were found to have aneuploidy (trisomy 13, 18 and 21) (0.079%). In 6 out of the 7 fetuses maternal age was over 35 years (86%). Out of those 7 fetuses, 3 had an isolated IEF, in all those cases, maternal age was over 35 years. A first-trimester screening had been done in 2 out of those 7 fetuses and was normal.

Conclusion: Our collective has a relatively low incidence of IEF indicating a low risk background population. In the case of fetal aneuploidy we found the majority of patients to have at least one other risk factor, the most common being maternal age over 35 years (> 90%). In those patients with fetuses with aneuploidy and who were older than 35 years we found an isolated IEF in 50% (3 out of 6). There was however, no case of aneuploidy in our women with IEF who were younger than 35 years. According to our data there is no medical indication for invasive diagnostics in case of an isolated IEF in a patient younger than 35 years coming from our low risk population.

P02.41

Isolated right pulmonary agenesis in a twin gestation – prenatal diagnosis and postnatal follow-up

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Isolated pulmonary agenesis is a rare congenital anomaly and most reported cases were diagnosed postnatally as an incidental finding. Only few cases of prenatal diagnosis were previously reported and data is lacking in regard to the postnatal outcome of these cases. We report on a 33 year old G3P1 patient with a dichorionic diamniotic twins that presented to our ultrasound unit for a detailed anatomy scan at twenty weeks of gestation. One of the twins, twin B, was found to have dextrocardia and complete absence of the right side pulmonary tissue and vessels. No other anomalies including cardiac anomalies were identified. At that stage the patient continued her care in a different center where she delivered in a Cesarean section at 37 weeks of gestation. The affected twin weight in delivery was 2250gr and the other was 2760gr. Postnatal MRI confirmed the diagnosis of isolated pulmonary agenesis and a normal karyotype was found for both twins. FISH for 22q11 deletion was negative. Currently, the infant is four months old, in good health and adequate weight and met all developmental milestones. To our knowledge this is the second report on prenatal diagnosis of isolated pulmonary agenesis with postnatal follow up. As in the previously reported case our case supports good prognosis for these infants.

P02.42

Abstract withdrawn

P02.43

Congenital diaphragmatic hernia diagnosed in the first trimester

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We present a case of left-sided diaphragmatic hernia diagnosed at 12 weeks of gestation and review the literature on first-trimester diagnosis of the defect. A 28-year old caucasian woman presented at Alexandra Maternity Hospital for her routine 11–14 weeks' ultrasound scan at 12 weeks of gestation. Her medical history was unremarkable and she reported one previous first-trimester miscarriage. The fetus had a crown-rump-length of 60 mm and nuchal translucency of 1.4 mm. Examination of the fetal anatomy according to our protocol showed the stomach at the left side of the thorax, mediastinal shift and displacement of the heart.