a frequency of 1/1000 to 1/2500 livebirths with a highly variable phenotypic expression. Genetic diagnosis by sequencing of the three main genes (PTPN11, SOS1, RAF1 and RIT 1) is possible in only 30–60% of cases. Thus, looking for sonographic abnormalities and dysmorphology remain important for the diagnosis of Noonan syndrome.

Aim of the study was ro descr he new sonographic signs suggesting diagnosis of Noonan syndrome.

Methods: In two tertiary centers, from a retrospective series of 8 fetuses with a Noonan syndrome, two sonographic signs were described: typical aspect of ears and heart defects especially valve stenosis.

Results: The 8 fetuses had an increased nuchal translucency with a normal karyotype. All fetuses were carriers of low-set ears, especially with a fleshy and projected forward lower lobe. Four of 8 fetuses were carriers of a heart defect including two pulmonary stenosis and one sub aortic stenosis. A mutation confirming Noonan syndrome was identified in 7 fetuses out of 8.

Conclusions: Prenatal diagnosis of Noonan Syndrome cannot always be confirmed genetically but can be strongly suspected prenatally in the presence of typical sonographic signs.

Supporting information can be found in the online version of this abstract

EP12.02

Extending the window for genetic screening sonograms: needs assessment for an ultrasound-based Down's syndrome risk model up to 24 weeks gestation

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Objectives: 30 years have passed since Down's syndrome (DS) screening was first proposed using sonographic markers in the 2nd trimester (Lockwood, 1987). Multiple markers have since been studied, however, the original gestational age window of ~15-20 weeks persisted in many of the studies. A recent meta-analysis (Agathokleous, 2013) offers both positive and negative likelihood ratios (LR), allowing for calculation of DS risk depending on presence or absence of 9 different markers. We developed a Sonographic Assessment Software program (DSASP), initially for the 15–20 week window (BPD 28.0-50.0 mm) based upon results of Agathokleous, Locatelli (2000), Nyberg (1995), and others. The function of DSASP is to calculate a final, integrated LR automatically. We sought to determine the impact of extending the gestational age window for DSASP for the genetic screening sonogram on our referral population.

Methods: Analysing ultrasound data for five years (2011–2015), images of 2nd trimester detailed fetal anatomy sonograms with BPD 28.0 to 50.0 mm and 50.1 to 60.0 mm were assessed. Cases with severe cerebral ventriculomegaly or anencephaly were excluded from the analysis.

Results: Of 3,128 cases, 2399 were performed at BPD 28.0–50.0 mm, 729 (23.3%) were performed at BPD 50.1–60.0 mm. We revised our DSASP software to accommodate values appropriate for the extended gestational window.

Conclusions: Using a cut-off of 50.0 mm for fetal BPD (~20 weeks) for the DS genetic sonogram would prohibit its use in 23% of our referral population to a U.S. inner city perinatal center. Based upon these data, we re-modeled the DSASP to accommodate a gestational age window that extends to a BPD of 60.0 mm (~24 4/7 weeks). We present our revised DSASP, a tool which can quickly calculate a final "combined" likelihood ratio for DS and, furthermore, encompasses a later gestational age window that will be highly useful in similar patient populations.

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Comparison of pregnancy outcomes in fetuses with increased nuchal translucency according to the sonographic features

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Objectives: We aimed to analyse the frequency and type of abnormal karyotype in fetuses with nuchal translucency (NT) ≥ 3.5 mm according to the sonographic features.

Methods: We retrospectively reviewed the medical records and ultrasonography of pregnant women whose fetuses were diagnosed with NT ≥ 3.5 mm at 11 + 0–13 + 6 gestational weeks and underwent karyotyping from 2009 to 2013 at Cheil General Hospital, Seoul, Republic of Korea. We divided study population into three groups according to sonographic findings; simple increased nuchal translucency (INT), septated cystic hygroma (SCH) and INT with additional abnormalities. We analysed the frequency and types of abnormal karyotypes of three groups with regard to the thickness of nuchal translucency.

Results: Among 165 pregnancies with NT ≥ 3.5 mm, 94 (57%) had simple INT, 56 (34%) had septated cystic hygroma, and 15 (9%) had INT with other anomalies. Chromosomal abnormalities were confirmed 4 (4%) fetuses in simple INT, 21 (37%) in SCH and 10 (67%) in INT with other anomalies. Among the 97 fetuses with NT between 3.5 to 4.4 mm, 3 cases were diagnosed with chromosomal anomalies in 77 fetuses with simple INT, 5 cases in 15 SCH, and 4 cases in 5 INT with other anomalies. Among the 26 fetuses with NT between 4.5 and 5.4, no fetus had chromosomal abnormality in 12 simple INT, 2 fetuses in 11 SCH and one case in 3 INT with other anomalies. In the 42 fetuses with NT greater than 5.5 mm, 1 case in 5 simple INT, 14 in 30 SCH and 5 in 7 INT with other anomalies.

Conclusions: As the thickness of nuchal translucency increases, the proportion of SCH increases. Even though nuchal translucency is equally measured, the frequency of chromosomal abnormality is significantly higher in fetuses with SCH compared to simple INT.

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MOM of prefrontal space ratio in Iranian euploid fetuses

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Objectives: To evaluate the MOM of prefrontal space ratio in 16–24 weeks euploid Iranian fetuses.

Methods: In a prospective observational study utilising 2D images of 16–24 weeks euploid fetal faces that were recorded during prenatal ultrasound examinations at the Perinatology Department of three university based Hospitals of Tehran University of Medical Sciences, 200 euploid fetuses between 16and 24 weeks gestation were examined. A line was drawn between the leading edge of the mandible and the maxilla and extended in front of the forehead. The ratio of the skin (d1) to the distance between the skin and the point where the MM line was intercepted (d2) was calculated (d2/d1). MOM of PFSR in Iranian euploid fetuses and their relationship with gestational age was calculated.