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Sonographic soft markers in the second trimester: Subtle indicators or significant findings?

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**Abstract**

Advances in ultrasound technology over recent times, mean that the once controversial area of aneuploidy detection is becoming a popular topic for sonographers worldwide. Improved resolutions from high end machines mean that very subtle anatomic variants may have a part to play in the detection of chromosomal abnormalities. When combined with laboratory testing and risk assessment tools, early detection of these soft markers can provide a rationale for the diagnosis and management of fetal chromosomal defects.

**Introduction**

The second trimester detailed anomaly scan remains a highly effective screening tool for the assessment of structural normality and recognition of fetal abnormalities (Liau et al, 2014). Improvements in ultrasound resolution now mean that subtle anatomic variations referred to as “soft markers” are more detectable, often leaving practitioners in a dilemma regarding referral for further testing and follow up care pathways. The National Institute for Health and Care Excellence recommend early screening for aneuploidy and that all women are offered a detailed anomaly scan between 18 weeks and 20±6 weeks (NICE, 2012).

The literature is abundant with information defining these soft markers and the inclusion criteria varies throughout institutions; however the majority of sources have a classification list comprising of: choroid plexus cyst, intraacardiac foci, echogenic bowel, pyelectasis, shortened femur, single umbilical artery and mild ventriculomalegy. This poster highlights the ultrasound image analysis of the most relevant soft markers and discusses the recent developments in the field.

**Ultrasound Findings**

**Figure 1:** Axial view of the fetal skull demonstrating a choroid plexus cyst at level of the posterior ventricle. (Green arrow)

**Figure 2:** Echogenic intracardiac focus

**Figure 3:** Longitudinal view of the fetal abdomen demonstrating an echogenic bowel (left) from Saha et al (2012) pg 795, labelled by author and normal appearing (right).

The gain is reduced to assist confirmation of echogenicity which is similar to bone (Saha et al, 2012).

**Figure 4:** Pyelectasis

**Figure 5:** Single umbilical artery

**Figure 6:** Mild ventriculomegaly

**Figure 7:** Graph summarising data from Agathokleous et al (2013).

**Discussion**

On review of the current literature the significance of specific soft markers has changed considerably over the years. In relation to single umbilical artery as an isolated finding, Voskamp et al (2013) undertook a large systematic review and meta-analysis and found no evidence that fetuses with isolated single umbilical artery have an increased risk of aneuploidy compared to a 10% risk mentioned in past studies.

On the other hand the diagnosis of an isolated fetal pyelectasis >4mm as a soft marker for Down syndrome showed a notable positive and negative likelihood ratio of 2.78 respectively (Orechowich and Berghella, 2013). In turn a more extended study carried out by Agathokleous et al (2013) looks at the various different soft markers and their correlation with Down syndrome. The positive likelihood ratios are shown in the chart below to demonstrate the significance of each individual marker.

Statistics showed that ventriculomegaly, increased nuchal fold, aberrant right subclavian artery and hypoplastic nasal bone yielded the highest positive likelihood results.

**Follow up and further investigations**

Referral for further management will depend on institutional guidelines and client preference, however the majority of settings advocate further investigations if two or more anatomic variations are noted on ultrasound. The number of soft markers detected can have a significant bearing on the risk for aneuploidy. Finding one soft marker determines a risk of 2%, it rises to 11% in the presence of two softmarkers and up to 66% with five and in turn 92% with eight (Zielot, 2013).

**References**