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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 (Li et al, 2014). Improvements in ultrasound resolution now mean that subtle anatomic variations related to "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, intracardiac focus, echogenic bowel, pylolecstasis, shortened femur, single umbilical artery and mild ventriculomegaly. 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: Echogenic bowel/ normal bowel

Figure 4: Pylectasis

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 pylectasis >4mm as a soft marker for Down syndrome showed a notable positive and negative likelihood ratio of 2.76 respectively (Orzechowski et al, 2013). In turn a more extended study carried out by Agathokleous et al (2013) looked 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.

Positive Likelihood ratios for T21

Figure 8: Image of aneuploidy from births injury Justice.org.

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 (Zelei, 2013).

Conclusion
Currently in Ireland there is no standardized policy for second trimester aneuploidy screening. The detailed anomaly scan is usually the first screening tool applied to determine structural normality, exposing abnormal anatomic variants as possible warning signs. Referral for follow up and further investigation often poses dilemmas due to the lack of a set classification critique and a universal specific policy for management. Overall with proper awareness of the specific soft markers and follow up care advancements, sonographers can strive to improve client care in this much debated field of sonography.

References