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, intracardiac foci, echogenic bowel, pyelectasis, 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: Choroid plexus cyst

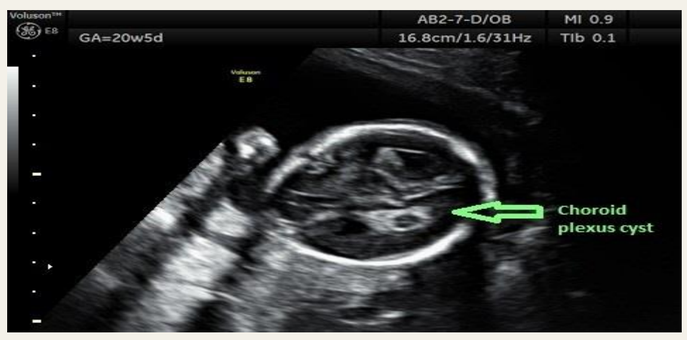

Figure1: Axial view of the fetal skull demonstrating a choroid plexus cyst at level of the posterior ventricle. (Green arrow)

Figure 2: Echogenic intracardiac focus

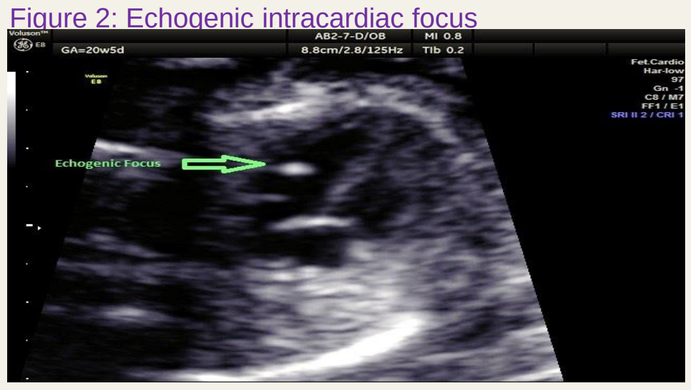

Figure 2: Four chamber view of the fetal heart containing an echogenic intracardiac focus in the left ventricular chamber (see arrow).

Figure 3: Echogenic bowel/ normal bowel

Figure 3: Longitudinal view of the fetal abdomen demonstrating an echogenic bowel (left) from Saha et al (2012) pg 759, labelled by author and normal appearing bowel (right).

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

Figure 4: Pyelectasis

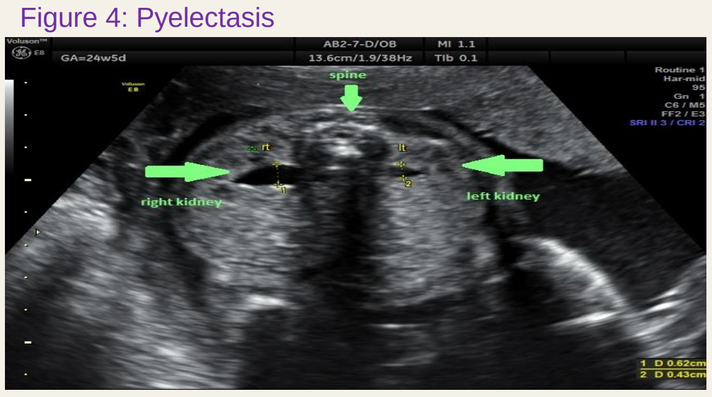

Figure 4: Shows a transverse view of the fetus at the level of the kidneys incorporating renal pelvis measurements in the anterior-posterior diameter .This image portrays a right sided pyelectasis of 6mm.

Figure 5: Single umbilical artery

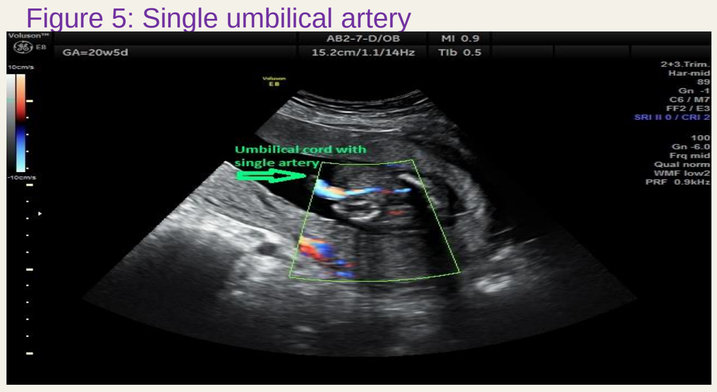

Figure 5: Portrays the umbilical cord insertion at the level of the bladder with single artery bifurcation and application of colour doppler.

Figure 6: Mild ventriculomegaly

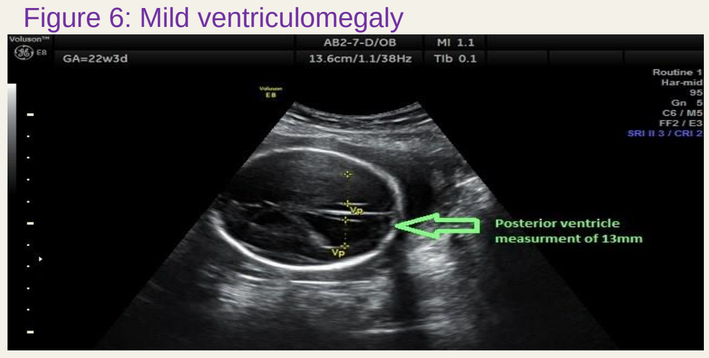

Figure 6: Demonstrates the posterior ventricles with mild ventriculomegaly of 13mms noted. Ventriculomegaly is described as cerebral ventricular measurement above 10mms (Agathokleous et al, 2013).

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 (Orzechowski and Berghella, 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.

Figure 7

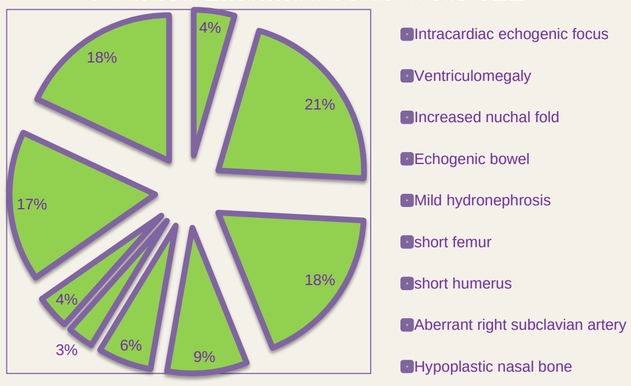

Figure 7: Graph summarising data from Agathokleous et al (2013).

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 (Zalel, 2013).

Figure 8

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

Although CVS and amniocentesis remain the definitive diagnostic tests in the detection of aneuploidy, recent advances in the field of non-invasive prenatal screening of maternal cell free DNA boast significant detection results without the added risks to the fetus. Analysis of cell free fetal DNA has detection rates of 99.2% for T21, 96.3% for T18, 91.0% for T13 and 93.0% for sex chromosome aneuploidy (Gil et al, 2015). Amniocentesis carries a 1% risk for miscarriage and should be carried out after 15 weeks gestation (RCOG, 2010).

In relation to ultrasound the inclusion of the nasal bone length as part of the detailed anomaly scan is also recommended, particularly in the diagnosis of T21. Zalel et al (2013) reviewed studies linking nasal bone hypoplasia (NBH) to aneuploidy in the second trimester. Findings suggested that NBH was present in 61.8% of fetuses with Down syndrome, 1.2% of normal fetuses and 3.3% with other chromosomal abnormalities, strengthening its significance as an additional soft marker.

Figure 9: Nasal bone

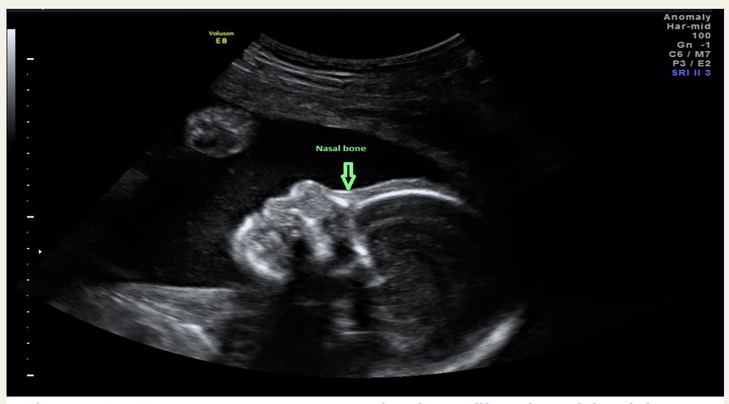

Figure 9: Demonstrates a sagittal profile view ideal for nasal bone measurement and assessment.

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 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.

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