Importance of isolated ultrasound markers at the 18 - 24 weeks’ scan in normally grown fetuses in the absence of structural defects

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The second trimester anomaly scan between 18 – 24 weeks is the most frequent scan performed in Obstetrics to look for fetal anomalies. Various markers have been described in the second trimester that increases the risk for aneuploidies. In the presence of an isolated marker, there is a dilemma if further invasive testing should be offered or not.

**Aim:** To assess the strength of isolated markers in the 2nd trimester in predicting fetal aneuploidies in structurally normal fetuses

**2T ultrasound Markers studied**

- Absent/hypoplastic nasal bone < 2.5mm
- Nuchal edema >6mm
- Ventriculomegaly = 10-15mm
- Echogenic intracardiac focus
- Hyperechogenic bowel
- Renal pelvic dilatation (5-10mm)
- Short femur/ humerus <5th centile
- Aberrant right subclavian artery

(Started studying on scan from July 2011-N=7579)

**Materials and methods:** Jan 2005 - Dec 2015
Scans performed by FMF certified operators; N=13467 singletons; structurally normal; known outcome wrt Karyotype
Exclusion: > 2 markers

**CONCLUSIONS:** Presence of an isolated marker, especially ARSA, nuchal edema and absence/ hypoplasia of the nasal bone are important markers even when isolated. LR for isolated markers are

- ARSA- 22.14; nuchal edema- 24.7; Hypopl /absent NB ~ 8.8; EIF-2.12

- Invasive testing can be offered to the couple if any of the above ISOLATED marker is present to confirm the karyotype

Search for associated defects and other markers along with confirmation of normal growth is very important as multiple markers >2 increases the risk of aneuploidies by atleast 10 times

In the absence of defects and markers in an otherwise normally grown fetus the risk is about 0.025%.