Follow up assessment of the “absent nasal bone” in the 2nd trimester - is there a benefit of waiting to perform an invasive test?
Reddy S, Dhengle S, Sahana R, Acharya V, Radhakrishnan P
Bangalore Fetal Medicine Centre, Bangalore, India

- Nasal bone (NB) is an important marker for chromosomal abnormalities in the first and second trimesters
- Addition of NB in the 1st trimester screening along with Nuchal Translucency (NT) and maternal serum biochemistry will increase the detection rate to 95%
- NB is absent in 60-70% of babies with Down Syndrome and 3% of chromosomally normal babies

AIM: To assess if the subsequent “normalisation” of the NB in the early 2nd trimester after detection of “absent NB” at the 11 - 13+6 weeks’ scan will reduce the need for invasive testing and can be considered as an option for some parents who are not keen on early invasive testing

Material and Methods
- Inclusion criteria -
  - Singleton pregnancies between Jan 2008 - Dec 2016
  - Scans performed by FMF certified operators
  - 1st & 2nd trimester scans performed in our unit
- Exclusion criteria -
  - Fetuses with structural abnormalities

RESULTS: Of the total 9485, NB was reported in 9212 fetuses (97.1%)
  - Absent/ Hypoplastic NB - 273 (2.96%)
  - Present - 8900 (96.6%)
  - Could not be assessed - 39 (0.4%), of which 31 (79.1%) had structural anomalies

CONCLUSIONS:
- “Absent NB” in the 1T in South Indian fetuses is an important marker for fetal defects and chromosomal anomalies
- “normalisation” of the nasal bone length in 2T does not exclude the increased risk for aneuploidy

Our study suggests that there is little benefit waiting for 2T assessment, as in both groups i.e. NB <5th centile and above the 5th centile, there is approximately 1:20 chance of a chromosomal anomaly anyway. Parents should be counselled regarding this lack of benefit versus the benefits of early diagnosis and termination following CVS.

REFERENCES: