4743: Second trimester follow up of fetuses detected to have "Absent nasal bone" in the first trimester.

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Introduction: Absent nasal bone is a useful marker for chromosomal aneuploidy. The association of absence or hypoplasia of fetal nasal bone (AHNB) in the first or second trimester scans, with increased risk for trisomy 21 has been shown in many studies. However, with advancing gestation the nasal bone is more frequently visible in trisomy 21 fetuses, although it is frequently hypoplastic as compared to normal fetuses. Inclusion of nasal bone in screening along with other parameters has been known to increase the performance of screening tests.

Objectives:

- To evaluate fetuses detected to have absent nasal bone (ANB) in the 1T
- Follow up in the 2T with respect to the nasal bone length (NBL)
- To assess the relationship with thus far undetected aneuploidies and structural anomalies

Methodology:

- Retrospective comparative study of prospectively collected data from a single tertiary fetal care referral centre during Jan 2005 to Dec 2022
- 13,992 singleton pregnancies who underwent first trimester (1T) screening between 11^{+0} – 13^{+6} weeks with known follow ups were included. All scans were performed by FMF certified operators as per FMF criteria and were documented on Astraia fetal database software
- Outcomes were obtained by telephonic interview and examination of delivery details in hospital records
- Multi fetal pregnancies and fetuses with major structural anomalies were excluded from the study

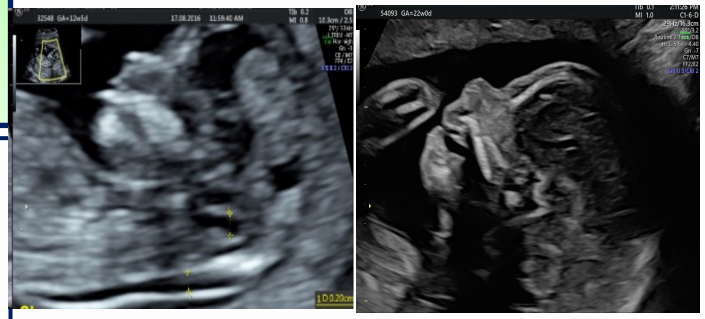


Fig 1: (a) ANB in 1T; (b) Follow up in 2T

Results

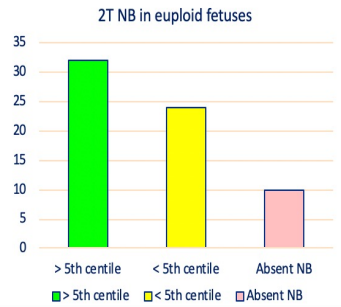
ANB in 1T = 372/13,903 (2.7%).
Isolated ANB = 272

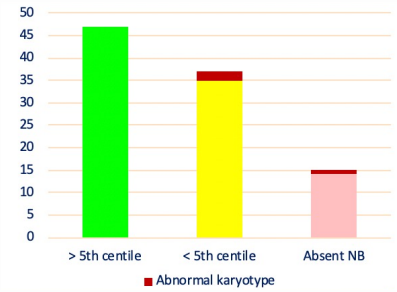
66/115 euploid fetuses followed up in 2T

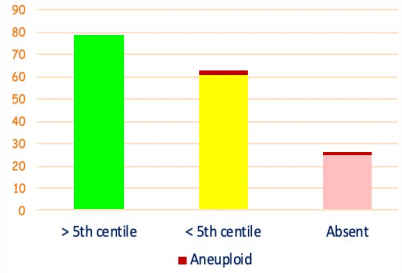
Of 141 fetuses who were to follow up in 2T, 99 followed up

Available 2T follow up of all fetuses with ANB

ToP without testing = 10
Invasive/ cfDNA = 121
No testing in 1T = 141
Of 121,
Normal karyotype – 115 (95.04%)
Abnormal karyotype – 6 (4.95%)







11/ 246 (4.47%) euploid fetuses were found to have structural abnormalities in second trimester in comparison to 14/11,757 (0.11%) euploid fetuses with present nasal bone

Conclusions:

- ANB is an important marker for aneuploidy.
- About 50% of these fetuses will continue to have hypoplastic/ ANB in the second trimester and invasive testing should be 're-offered' to these parents
- Detailed anatomical survey of the fetus is recommended, preferably by transvaginal scan
- Invasive testing should be offered to all with absent nasal bone the 1T
- We need further prospective studies to assess if invasive testing can be deferred when the NB "normalizes" in the 2T

Reference: Cusick W, Provenzano J, Sullivan CA, Gallousis FM, Rodis JF. Fetal nasal bone length in euploid and aneuploid fetuses between 11- and 20-weeks' gestation: a prospective study. J Ultrasound Med. 2004 Oct;23(10):1327-33. doi: 10.7863/jum.2004.23.10.1327. PMID: 15448323.

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