4781: Assessment of the nasal bone at 11^{+0} to 13^{+6} weeks gestation can improve the detection of aneuploidies even when there is a degree of uncertainty

Chandra N, Savaskar S, Acharya V, Shettikeri A, Radhakrishnan P, Bangalore Fetal Medicine Centre, India

BANGALORE FETAL MEDICINE CENTRE

Introduction: Absent nasal bone is a useful marker for chromosomal aneuploidy. This is a study with large sample size of structurally normal fetuses with ANB in the South Indian population in the first trimester. It shows the importance of assessing the NB and also compares the incidence of aneuploidies in not only fetuses with ANB but also when there is an uncertainty about the presence of the NB with and without associated markers .

Objectives:

- To assess the incidence of absent nasal bone(ANB), equivocal/uncertain nasal bone (ENB) and normal/present nasal bone(PNB) in the first trimester (1T) in South Indian population.
- To assess the prevalence of aneuploidies in each group ANB, ENB, PNB and compare them.
- To assess the increase in prevalence of aneuploidies when there were associated markers in each group.

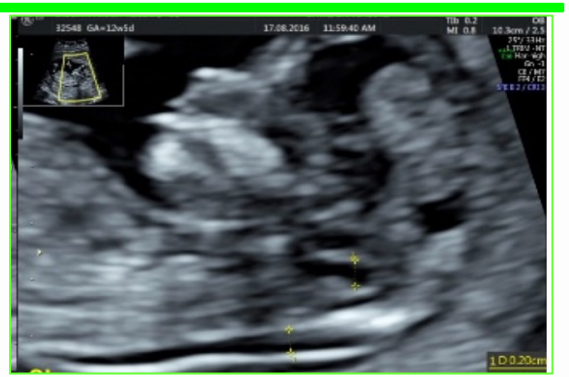


Fig 1: (a) ANB in 1T

Methodology:

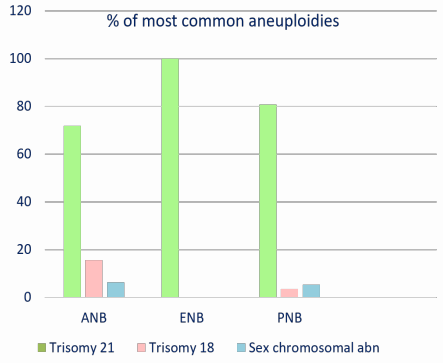
- Retrospective comparative study of prospectively collected data from a single tertiary fetal care referral centre during Jan 2005 to Dec 2022
- 13,903 , structurally normal fetuses who underwent assessment of nasal bone in 1T between 11^{+0} – 13^{+6} weeks with known outcomes were included. ANB and PNB were defined as per the FMF protocol All scans were performed by FMF certified operators 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, fetuses with major structural anomalies and unknown karyotype/follow up were excluded from the study

Results

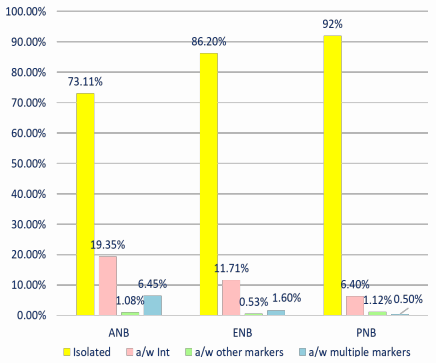
Incidence of ANB, ENB, PNB and aneuploidies overall

Total fetuses (13903)	Incidence of fetuses in each group	Aneuploidies = 94/13903 (0.67%)
ANB	372/13903 (2.7%)	32/372 (8.61%)
ENB	188/13903 (1.35%)	5/188 (2.66%)
PNB	13343/13903 (95.9%)	57/13343 (0.43%)

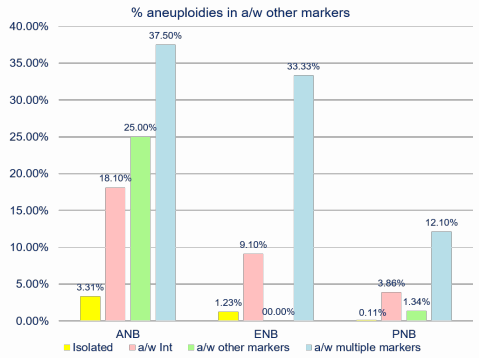
Specific aneuploidies in each group



Association of each group with other markers



Increase in aneuploidies in association with other markers



Conclusions:

- Overall incidence of ANB in the South Indian population in the 1T in structurally normal fetuses is 2.7%
- Uncertainty regarding absent/ present NB occurs in 1.35%
- Higher incidence of chromosomal anomalies in both groups
- Significantly increased in the presence of iNT and further increased with a greater number of markers
- Most frequently associated aneuploidy with isolated ANB/ ENB remains T21

Reference: Absent nasal bone at 11–14 weeks of gestation and chromosomal defects S. CICERO, D. LONGO, G. REMBOUSKOS, C. SACCHINI and K. H. NICOLAIDES Harris Birthright Research Centre for Fetal Medicine, King's College Hospital Medical School, London, UK

Higher incidence of chromosomal anomalies seen in both groups (ANB- 3.31% and ENB-1.23% respectively, which is 30 and 11 times higher than the "present NB" group)

Correspondence:
drprathima@bangalorefetalmedicine.com