

Defining “hypoplasia of the fetal nasal bone” as a marker for aneuploidies

Jain S, Acharya V, Shettikeri A, Sahana R, Radhakrishnan P
Bangalore Fetal Medicine Centre, Bengaluru, India

Objective

Hypoplastic nasal bone is an important marker of chromosomal abnormalities in the fetus. The objective of our study was to compare the detection rate of fetal aneuploidies by altering the cut-off for defining hypoplasia of the fetal nasal bone as 2.5th centile as opposed to the 5th centile.

Methods

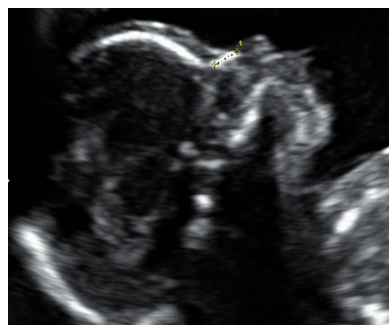
This is a retrospective study of prospectively collected data at a tertiary fetal care centre in South India. The study period was from January 2010 to December 2019. 12,694 singleton pregnancies examined during this time from 16 – 26 weeks gestational age, with known outcomes were included in the study. The nasal bone was measured when the fetus was in the mid-sagittal plane with angle of ultrasound beam to nasal bone at 45 - 135 degrees, as per the FMF protocol. The 2.5th and the 5th centiles for each gestational age was calculated. All scans were performed by FMF certified operators for the 18 – 24 weeks' scan. All examinations were recorded on Astraia fetal database software. Outcome of the pregnancy was obtained by telephonic conversation with the parents and examination of the delivery details in the hospital records. The aneuploidy detection rate with NBL < 2.5th centile and NBL < 5th centile was compared.

Results

There were 48/ 12,694 (0.3%) aneuploidies during this study period. 46 (95.8%) were major aneuploidies i. e. , trisomies, triploidies and monosomy. There were 2 (4.2%) balanced translocations, of which 1 fetus had the NBL below the 2.5th centile for the gestational age. The incidence of NBL < 5th centile was 628/ 12694 (4.9%) and < 2.5th centile was 332 (2.61%). There were 11 aneuploidies in the cohort with NBL < 5th centile, all of which were below the 2.5th centile as well. 10/ 11 aneuploidies were significant. 8 fetuses had Trisomy 21 and 2 had Trisomy 18.

Conclusion

Our study shows that the incidence of “hypoplastic nasal bone < 5th centile” is almost double than that defined by being < 2.5th centile for the gestational age. By reducing the percentile from 5th to the 2.5th centile, we were able to detect all significant aneuploidies. In addition, this would reduce the “false positive rate” of hypoplastic nasal bone significantly. In our study there would have been 24% fewer invasive tests to detect aneuploidies if 2.5th centile was considered. We propose to validate the use of the 2.5th centile as the cut-off to define “hypoplastic nasal bone” in larger studies for detection of fetal aneuploidies to reduce the need for invasive testing.



Aneuploidies	Total population (n = 12694)	NBL < 5 th centile (n = 628)	NBL < 2.5 th centile (n = 332)
No. of Aneuploidies	48	11	11
Major Aneuploidies	46	10 (T21 – 8, T18 - 2)	10 (T21 – 8, T18 - 2)
Balanced Translocations	2	1	1

Variable	NBL < 5 th centile	NBL < 2.5 th centile
Incidence	628 (4.9%)	332 (2.61%)
Detection rate	22.9%	22.9%
False Positive Rate	4.8%	2.5%