Reference ranges for fetal nasal bone in screening for chromosomal abnormalities at 11-13 weeks

Parra-Cordero M, Sepulveda-Martínez A, Pedraza D, Muñoz H, Figueroa J, Rencoret G
1Fetal Medicine Unit, University of Chile Hospital and 2San Borja Arriaran Hospital, Santiago, Chile

Objective
Aims of this study were to establish nasal bone length (NBL) reference ranges throughout first trimester and to evaluate its value for screening for chromosomal defects in a Chilean population.

Methods
1) To determine NBL reference ranges for our population, the fetal profile was examined in 4,860 consecutive singleton pregnancies between 11+0 to 13+6 weeks gestation since January 2005 to January 2017 with a phenotypically normal newborn. The data was log transformed and fitted using a linear equation. 2) To determine the role of NBL as first trimester screening test for fetal aneuploidy, 6,004 pregnant women who underwent a routine ultrasound scan at 11+0 – 13+6 weeks gestation were included. Detection rate (DR), false positive rate (FPR) and likelihood ratio (LR) for absent nasal bone and hypoplastic nasal bone (defined as <2.5th percentile) were obtained.

Results
There was a positive correlation between NBL and gestational age (r=0.45; p < 0.001). Nomograms including the 2.5th, 5th, 50th and 95th percentiles were created for each gestational age. As expected, the NBL increased from 1.3 mm at 11 weeks to 2.1 mm at 14 weeks. In the group of patients who were scanned at 11+0-13+6 weeks, there were 36 cases with chromosomal abnormalities, being 13 trisomy 21 (1:450), 12 trisomy 18&13 and 6 Turner syndromes. The NB was absent and/or hypoplastic in 60 (1.1%) of the 5,526 chromosomally or phenotypically normal fetuses and in 10 (27.7%) of the 36 fetuses with an abnormal karyotype, being, therefore, the positive LR around 25. However, the vast majority of those hypoplastic NB were considered absent at the time of the assessment (9 out of 10), therefore, the detection rate, false positive and positive LR for absent NB was 25.0%, 0.6% and 41.7, respectively.

Conclusion
NBL is modified by ethnicity; therefore, establishing a normal reference range is essential for studying its role as a screening test for chromosomal abnormalities at different gestational ages. Absent NB performs better than hypoplastic NB in the assessment of chromosomal abnormalities during the first trimester of pregnancy.