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Can Naso Frontal Angle (NFA) predict outcome of fetuses with isolated "flat facial profile" at the 18 – 24 weeks' scan?

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Objective

The flat facial profile is a subjective appearance of fetal nasion forming an obtuse angle with the forehead. It is a "soft sonographic sign" which gives clues for various congenital syndromes and chromosomal aberrations. The aim of our study was to assess the accuracy of NFA to predict the antenatal and postnatal outcome in fetuses detected to have an isolated flat facial profile at the 18 – 24 weeks' anomaly scan.

Methods

This is a part of a retrospective audit of fetuses detected to have a "flat facial profile" at the 18 – 24 weeks' anomaly scan. The study period is from January 2012 to December 2021 and conducted at a tertiary referral center in South India. All scans were performed by FMF certified operators for the 18 – 24 weeks' anomaly scan. 75 fetuses with a "flat facial profile" were audited further for the presence or absence of the structural and chromosomal abnormalities. This included 4 fetuses where one of the dichorionic twin was affected. In 58 fetuses, stored 2D dicom images of the facial profile was available and the Naso-Frontal Angle (NFA) was measured by using the measuring tool provided on the medsynapse PACS system. Of these, 32 belonged to "isolated euploid" fetuses. The NFA was corelated to the outcome in these cases. The NFA is defined as the angle between the nasal and the frontal bones in the exact mid-sagittal plane of fetal facial profile. An NFA of 135 and 140 degrees was compared to assess the sensitivity of predicting a poor postnatal outcome in "isolated cases". Outcome of the pregnancy was obtained by telephonic interview of the parents.

Results

27/ 75 fetuses had associated structural and/ or chromosomal anomalies and were excluded for the purpose of the NFA study. One pregnancy is still ongoing. Outcome of the pregnancy was available in 45/ 48 isolated cases. In the 45 isolated cases with outcomes, 23/ 45 (51.1%) had a good outcome with respect to developmental milestones as appropriate for the age of the child. In the remaining 22 (48.9%) cases, there were 14 terminations (including one for a CNV on chromosome 7 and one for Trisomy 21), 5 miscarriages or intrauterine fetal demise, one neonatal death and two live births that have postnatal dysmorphic features suggestive of a genetic syndrome and are awaiting further evaluation. In the 32 isolated cases, fetal facial profile images were analyzed and the NFA was measured on the stored dicom images. When NFA cut off was set to 135 degrees, 11/ 14 (78.6%) had a poor outcome. When the NFA cut off was set to 140 degrees, 9/ 14 (64.3%) had a poor outcome. Taking 140 degrees, the sensitivity was 64.3%, positive LR was 1.93 (95% CI 0.90- 4.13) and negative LR was 0.54 (95% CI 0.25 – 1.16). Two additional cases could be detected by using 135 degrees with a sensitivity of 78.6% and positive likelihood ratio of 1.09 (95% CI 0.73 – 1.62) and a negative LR of 0.77 (95% CI 0.22 – 2.69). However, 7 additional cases with good outcome also had an NFA of 135 degrees, increasing the false positive by 38.8%.

Conclusion

The assessment of "flatness" of the fetal facial profile is still largely subjective. Hence, in the absence of associated anomalies, it is difficult to predict antenatal and postnatal outcomes. In such cases, parents choose to continue or terminate the pregnancy based on the 2D and/ or 3D appearances at the time of the anomaly scan. However, this is subject to change as the pregnancy advances and the facial bones grow. In our set up, parental preference to terminate was based on presence of associated anomalies and facial appearance, which is largely subjective. We attempted to use an objective measurement, i. e. , the NFA, albeit retrospectively to predict the known outcomes in the isolated group. An NFA of more than 135 and 140 degrees could predict the poor outcome in 78.6% and 64.2% isolated euploid cases. We recommend larger prospective studies to assess and compare the accuracy of these measurements on fetuses diagnosed to have a "flat facial profile" at the 18 – 24 weeks' anomaly scan.

FIGURE 1

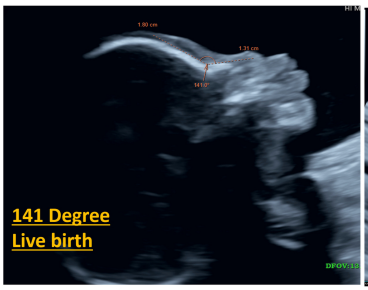

FIGURE 2

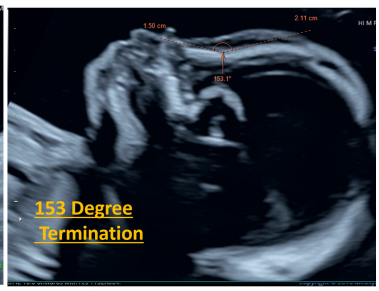

FIGURE 3

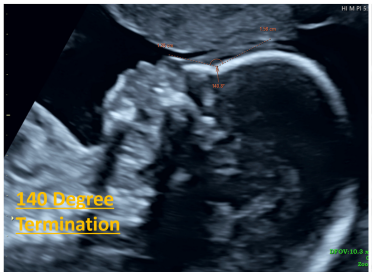

FIGURE 4