Absent/Hypoplastic Nasal Bone as a Soft Marker and its Genetic Association

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Abstract:

Introduction: Absent/hypoplastic nasal bone (AHNB) is recognized as an important soft marker on prenatal ultrasound, particularly during the first and second trimesters. The nasal bone, usually visualized by 11–14 weeks of gestation, is absent or shortened in a proportion of chromosomally abnormal fetuses. Hypoplasia or non-visualization of the nasal bone is strongly associated with trisomy 21, with studies reporting its presence in 60–70% of affected fetuses. It is also observed, though less frequently, in trisomy 18, trisomy 13, and other aneuploidies.

Aim: To evaluate the outcomes of fetuses diagnosed with absent/hypoplastic nasal bone

Method: This was a prospective, observational study from September 2024 to June 2025. It included all fetuses who were diagnosed or referred to us with AHNB from 16 weeks onwards. Amniocentesis was offered to all women, and the sample was sent for QFPCR and microarray. All the women were followed up for neonatal outcome.

Result: A total of 70 patients were detected with AHNB who underwent invasive testing. Out of 70, 23 patients had an isolated AHNB, 30 patients had AHNB with other soft markers, and 17 patients had AHNB with an increased biochemical risk.

Out of 70 cases, 6 came positive for Trisomy 21, among them 3 had AHNB on USG, 1 had AHNB with VSD, and 2 had AHNB with increased biochemical risk. 4 cases came positive for different genetic syndromes on CMA.

Conclusion: As a soft marker, AHNB is not only associated with aneuploidy lie Trisomy 21 but, when combined with other markers or abnormal biochemical screening results, substantially increases

the risk for other genetic conditions as well. The advent of non-invasive prenatal testing (NIPT) and chromosomal microarray analysis has further refined risk stratification in such cases. Genetic counseling is essential to balance the implications of this marker with other clinical parameters. Thus, AHNB serves as a valuable sonographic sign, enhancing the detection of chromosomal abnormalities while emphasizing the importance of integrating ultrasound findings with advanced genetic screening.