LOCAL EXPERIENCE IN THE USE OF NON-INVASIVE PRENATAL TESTING (NIPT): A CASE SERIES OF DISCORDANT NIPT RESULTS AND SECOND TRIMESTER ANATOMY SCANS

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Study Background & Objective:

Non-invasive Prenatal Testing (NIPT) is a risk screening genetic test that analyzes fetal cell-free DNA (cfDNA) from maternal plasma. The test has been slowly introduced in genetics laboratories and institutions in the country and has proven its value in screening for common fetal aneuploidies. This paper describes our clinical experience and the possible pathways for NIPT use in the first two years of implementation of a local hospital-based NIPT program. Our objective is to present discordant cases found to have fetal structural anomalies on mid-trimester ultrasound and yet revealed a low risk NIPT or cfDNA screening result.

Method:

We reviewed all the individuals who underwent NIPT from December 2019 to January 2022 and correlated their NIPT results with ultrasound findings and neonatal outcomes.

Result:

Three cases out of twenty-five individuals were found to have fetal structural anomalies. These anomalies were intestinal dilatation with talipes equinovarus (Case 1) *(refer to Figure 1),* asymmetrical ventriculomegaly with corpus callosum agenesis (Case 2) *(refer to Figure 2)*, and complete atrioventricular septal defect with pulmonary valve atresia (Case 3) *(refer to Figure 3)*. In all three cases, NIPT or cfDNA screening results were negative, and neonatal karyotyping results were normal.

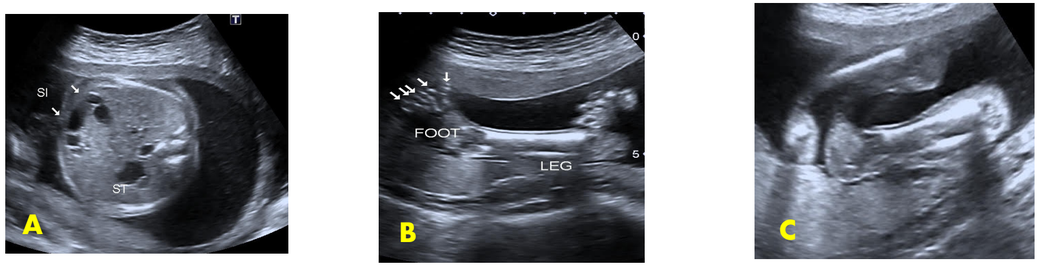

Figure 1A. Two-dimensional scan of the abdomen on axial view showing dilated small intestines (SI). **Figure 1B.** Two-dimensional image of talipes equinovarus showing the metatarsals and phalanges of the foot visualized in the same plane as tibia and fibula. **Figure 1C.** Bilateral adduction of the forefeet.

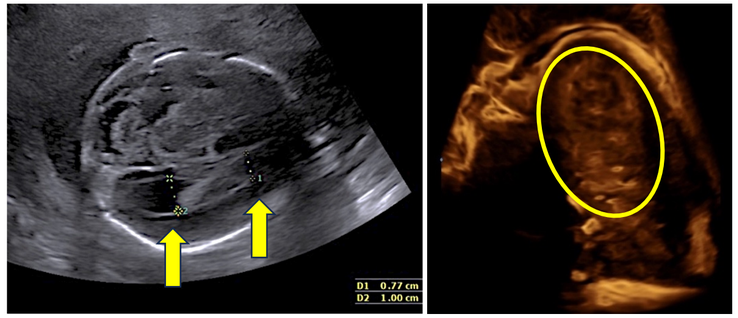

Figure 2A. Targeted fetal cranial ultrasound. Axial view of the fetal cranium showing asymmetrical mild ventriculomegaly at 23 weeks. **Figure 2B.** A non-visualized corpus callosum-cavum septum pellucidum complex on midsagittal view at 30 weeks.

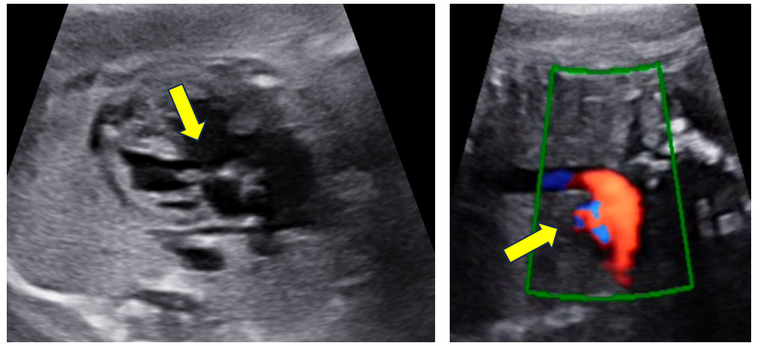

Figure 3A. Fetal echocardiography. Two-dimensional scan of the four-chamber view of the heart showing a complete atrioventricular septal defect, transitional type at 23 weeks. **Figure 4B.** On color Doppler, the flow across the ductus is reversed from the aortic arch (Ao) toward the pulmonary arteries.

Conclusion:

The discordant results of the cfDNA screening and second-trimester ultrasound findings suggest that there remains a considerable residual risk or probability of a genetic disorder following a negative cfDNA screening. Discussion of the concept of residual risk together with the benefits and limitations of the available genetic tests is an integral part of the pretest and posttest counseling for all patients. This report highlights the local clinical application of NIPT, an emerging technology for prenatal genetic screening in the country.

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