Abstract ID: - 200

Abstract Topic: - Genetic counselling

Abstract Title: - Biological factors influencing Sex Chromosomal Aneuploidies results on NIPT

Presenting author name: - Angela Devanboo

Presenting author institute: - MedGenome Labs Ltd

Co-authors name: - Bhavana Nellore, Manasvi Vikku, Teja Vismaya, Shweta Mahalingam, Ehsan Ul Haq, Avinash Pradhan, E. Venkataswamy, Ramprasad V. L., Priya Kadam

Co-authors institute: - MedGenome Labs Ltd, MedGenome Labs Ltd, MedGenome Labs Ltd, MedGenome Labs Ltd, MedGenome Labs Ltd, MedGenome Labs Ltd, MedGenome Labs Ltd, MedGenome Labs Ltd, MedGenome Labs Ltd

Aims: - To understand the different biological factors that attributed to inconclusive Sex chromosomal aneuploidies (SCAs) results on NIPT.

Methods: - Retrospective analysis was performed on samples that were tested for NIPT in our laboratory and the results were recorded.

For inconclusive SCA results, further diagnostic testing along with appropriate genetic counselling and comprehensive ultrasound was recommended.

The outcomes of these samples were followed up and the results were recorded.

Results: - In about 0.2% of samples that were sent for NIPT, inconclusive SCA results were reported. Follow up information was obtained in 18% of the samples. In 47% of the cases, normal results were obtained on further testing. In about 35% of the cases, the inconclusive results could be attributed to history of vanishing twin. In about 12% of the cases, the fetus had a sex chromosomal abnormality (Monosomy X & Mosaic XXX). For the remaining 6%, the maternal biological background was confounding the NIPT result i.e. the pregnant woman was identified to have an SCA (XXX).

Conclusions: - This study highlights the importance of careful analysis when reporting SCA results on NIPT. This study also highlights the importance of appropriate pre-test and post-test genetic counselling in such scenarios.

Keywords: - In about 0.2% of samples that were sent for NIPT, inconclusive SCA results were reported. Follow up information was obtained in 18% of the samples. In 47% of the cases, normal results were obtained on further testing. In about 35% of the cases, the inconclusive results could be attributed to history of vanishing twin. In about 12% of the cases, the fetus had a sex chromosomal abnormality (Monosomy X & Mosaic XXX). For the remaining 6%, the maternal biological background was confounding the NIPT result i.e. the pregnant woman was identified to have an SCA (XXX).