Importance of pretest diagnostic test genetic counselling in cases with NIPS high risk for sex chromosomal aneuploidies

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Introduction -

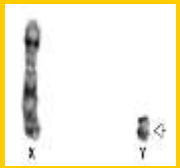
Traditional screening methods which included combination serum screening tests and ultrasound in the first and second trimester were used for detection of common fetal chromosomal aneuploidies like trisomy 21, 13 and 18. NIPS was introduced into clinical practice in late 2011. The option of screening for fetal sex chromosomal aneuploidies (SCA) is unique to NIPS and has not been available through traditional screening methods. f the screening test is positive for SCA it must be confirmed by diagnostic test.

Material and Method-

From January 2018 - April 2024 67 amniotic fluid samples with NIPS high risk for sex chromosome were received for Fluroscent Insitu hybridization (FISH) and chromosomal analysis. Samples were processed as per the standard protocol and reported as per ISCN 2020

Results-

38 cases were normal for FISH and karyotype. 22 had trisomy of sex chromosome. 4 cases had mosaic pattern. 1 case had rob(14;15) with trisomy of sex chromosome, 1 case had isochromosome of one of the sex chromosome and 1 case had deletion of one of the sex chromosome. High number of normal results could be due to higher rate of placental mosaicism for sex chromosome or due to maternal mosaicism for sex chromosome.



Discussion -

Often in prenatal genetic counselling sessions the couple is counseled for mostly the possibility of Trisomy 21 but not the possibility of SCA. Sex chromosomal abnormalities (SCA) are the most frequently occurring abnormality both in prenatal and postnatal cases. SCAs involve either duplication or deletion of complete X or Y chromosome or partial duplication, deletion or rearrangement of X or Y chromosome including inversions and translocations. The most common SCAs are 45,X, 47,XXX,47,XXY, 47,XYY and their mosaic forms. Pretest counselling for SCA high risk cases may be challenging as most individuals with SCA are not ascertained at birth due to lack of distinctive phenotypic features. A normal FISH report is reassuring to the couple. However during pretest genetic counselling the couple needs to be explained about the limitation of FISH test and presence of structural rearrangement of sex chromosome which can be detected only by karyotype.