NIPT Report

Non-invasive pre-natal testing is a procedure for screening of maternal peripheral blood for fetal chromosomal aneuploidies at around 10 weeks to 20 weeks of gestation using lab procedures and computational methods. This is mainly done to reduce the number of invasive testing procedures like chorionic villi sampling and amniocentesis. For accurate detection of chromosomal aneuploidies, the maternal blood should have at least 4% of fetal DNA. Presently, many tools are being used to detect fetal chromosomal aneuploidies like RAPIDR, NIPTeR, NIPTmer, VeriSeq NIPT solution, SAGE prenatal screen and WisecondorX.

RAPIDR is an R based tool for detection of chromosomal aneuploidies but is proven to be less accurate for detecting trisomies in chromosome 13, 18 and 21 at higher sequencing depth.

NIPTmer is a k-mer based software which uses kmers for detection of chromosomal aneuploidies and no reference set is required. However, it has not proven to be accurate for sequencing depth of less than 15 Million reads per sample.

NIPTeR is an R based software for detection of chromosomal aneuploidies and it can give accurate results for sequencing depth as low as 5 million reads per sample. It gives both z-scores and NCV scores for robust results. The only disadvantage that NIPTeR has is that it doesn't give results for sex chromosomes.

VeriSeq NIPT solution by illumina is a paid software which detects all the chromosomal aneuploidies including rare autosomal trisomies and aneuploidies of sex chromosomes with >= 99.9% sensitivity and specificity.

Sage Prenatal screen by Yourgene Health takes the samples in a batch of 12 samples or 32 samples depending on the need of analysis and it can detect trisomies at a fetal fraction of >3.5%. It also provides sensitivity and specificity of 99.9%.

Finally, WisecondorX is a software which gives accurate results for samples with sequencing depth as low as 2.5 million reads per sample. It takes a reference set of pregnant females with both male and female samples and compare the sample to the reference set to provide z-scores of all the chromosomes including sex chromosomes.

For a Bioinformatics pipeline for NIPT, the detection of fetal fraction along with chromosomal aneuploidies is also very important. SeqFF and PREFACE are the tools which are reliable and provide accurate values of fetal fraction. SeqFF uses two different algorithms for detecting fetal fraction and takes the average of both as final value. For chromosomal aneuploidies, WisecondorX will be the best software because of availability of source code and accurate detection of chromosomal aneuploidies at a sequencing coverage of al low as 2.5M reads per sample. WisecondorX also provides the values for data noise and overall abnormality scores for accurate detection of results.