FetalSeq assay:

Analysis of Products of Conception (POC) on Next Generation Sequencing

Clinical Indication:
Intra uterine death

Negative
(No clinically significant Copy Number Variants detected)

**Genetic test results are reported based on the recommendations of American College of Medical Genetics.*

Result	**Seq(hg19)(1-22)x2,(XY)x1** **Normal autosome and male sex chromosome complements.**

Sample Description:
Sample quality is optimum for the test. Maternal cell contamination was not ruled out due to non avalibility of mother blood.

DNA Storage:
The provided sample's DNA was extracted and stored at SN Genelab, Surat for further genetic testings.

Karyoview:

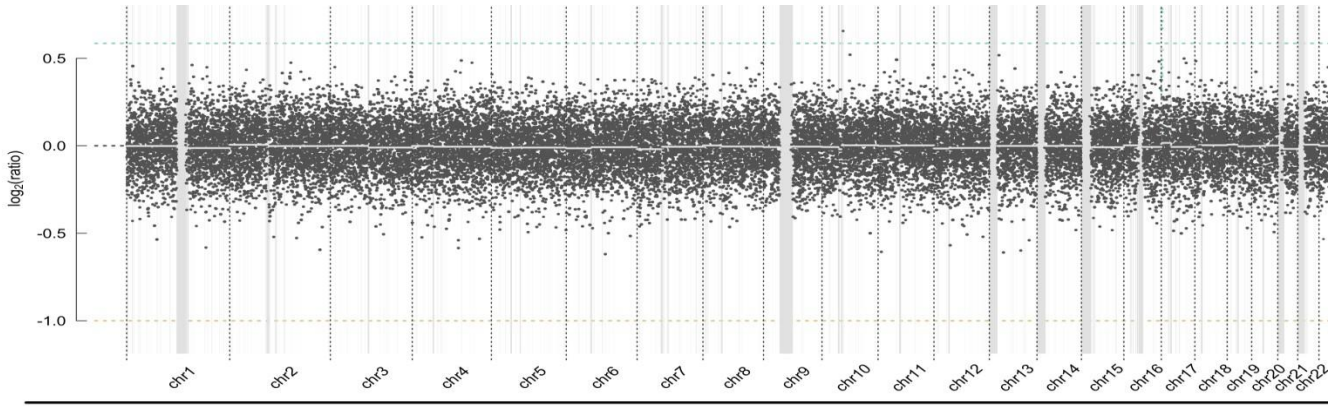

Fig 1: Genomic View of Duplication/Deletion in the analysed DNA sample.

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Indication :

Test Methodology:

- FetalSeq assay is designed for identification of copy number variants (All chromosome Aneuploidies, Microdeletions and Microduplications) through low pass whole genome sequencing using Illumina NovaSeq 6000 next generation sequencing.
- Genomic DNA extracted from Products of Conception (POC)/Cord Blood or any other standard source is used for quality check followed by library preparation.
- Prepared library were checked for optimal amplification and target size.
- QC passed library was sequenced using Illumina NovaSeq 6000.
- Data was analyzed using bioinformatics pipelines (reference genome: Hg19) to identify genome wisecopy number variants.

Limitations:

- FetalSeq is limited to detection of gain or loss of genomic material. It does not detects low level mosaicism (<20%), balanced translocations, inversions or point mutations that may be responsible for the clinical phenotype.
- This assay can detect a minimum resolution of 1 MB for losses & 1 MB for gains.
- This assay has increased coverage density (25 markers/100 kb) in 396 empirically selected regions relevant for prenatal research.

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Indication :

Variant classification as per ACMG guidelines:	
Variant	A change in a gene. This could be disease causing (pathogenic) or not disease causing (benign).
Benign	A variant which is known not to be responsible for disease has been detected. Generally, no further action is warranted on such variants when detected.
Likely Benign	A variant which is very unlikely to contribute to the development of disease however, the scientific evidence is currently insufficient to prove this conclusively. Additional evidence is expected to confirm this assertion of Pathogenicity.
Pathogenic	A disease-causing variation in a gene which can explain the patient's symptoms has been detected. This usually means that a suspected disorder for which testing had been requested has been confirmed.
Likely Pathogenic	A variant which is very likely to contribute to the development of disease however, the scientific evidence is currently insufficient to prove this conclusively. Additional evidence is expected to confirm this assertion of pathogenicity.
Variant of Uncertain Significance	A variant has been detected, but it is difficult to classify it as either pathogenic (disease causing) or benign (non-disease causing) based on current available scientific evidence. Further testing of the patient or family members as recommended by your clinician may be needed. It is probable that their significance can be assessed only with time, subject to availability of scientific evidence.

References:

1. Richards S, Aziz N, Bale S, et al. Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology, Genetics in Medicine, 2015 May;17(5):405-24

2. 4. McKenna, A., et al., The Genome Analysis Toolkit: a MapReduce framework for analyzing next-generation DNA sequencing data. Genome Res, 2010. 20(9): p. 1297-303

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