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Abstract Topic:- Molecular and cytogenetic diagnostics

Abstract Title:- KaryoSeq- an innovative whole genome sequencing to diagnose prenatal and newborn conditions.

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Aims:-KaryoSeq is a new genetic test that utilizes whole genome sequencing for faster and accurate detection

of aneuploidies of all chromosomes and Copy Number Variations, detection of

chromosomal abnormalities including trisomy such as trisomy-21, trisomy-18, trisomy-13, Turner's syndrome, abnormalities including, Klinefelter syndrome, Jacob's syndrome, triple X syndrome, and microdeletions DiGeorge/velocardiofacial syndrome (DG/VCFS), larger than 1 Mb, that may not be easily identified through microscopic examination alone. KaryoSeq has significant advantages over traditional cytogenetic and molecular techniques. KaryoSeq- cutting-edge innovation to redefine the genetic testing landscape. Leveraging Next-Generation Sequencing (NGS), aim to significant milestone in the field of prenatal and newborn genetic testing in this part of the world, empowering clinicians with accurate and comprehensive genetic information.

Methods:- KaryoSeq is a low-pass whole-genome sequencing-based test, where CNVs and aneuploidies are detected based on low-pass whole-genome sequence data. Libraries are prepared using DNA isolated from the DNA sample type Amniotic Fluid, Chorionic villus biopsy, Product of conception, Direct fetal DNA, peripheral blood. The data generated is analyzed using a bioinformatics tool on the human reference genome (GRCh37/hg19). Interpretation is based on current American College of Medical Genetics and Genomics (ACMG) guidelines.

Results:- In this study retrospective review involves data from 2023, Total 95 cases performed in MedGenome premises, in 67 cases no Copy number variant detected, in 28 cases detected Aneuploidies, larger CNVs, numerical Abnormalities. such as trisomy-21, trisomy-18, trisomy-22, trisomy-15, Turner's syndrome, Jacob's syndrome and accurately detected abnormalities including pathogenic and uncertain significance classified microdeletions, microduplication genetic variation within the genomes > 1Mb.

Conclusions:- KaryoSeq seamlessly complements existing assays, synergistically enhancing diagnostic

capabilities. Remarkably, when combined with karyotyping, it has demonstrated an impressive fivefold increase in diagnostic yield. empowering the identification of aneuploidies by targeting CNVs ranging from 1MB to the highest resolution achievable through karyotyping, its integration with exome sequencing offers a comprehensive perspective on CNV detection. KaryoSeq uncovers elusive variations often missed by conventional techniques.

Keywords:- KaryoSeq, CNV, NGS, Low Pass