

CNSeq

Advanced Chromosomal Analysis with Low-pass whole genome sequencing







CNSeq: Low-pass Whole Genome Sequencing

CNSeq revolutionizes chromosomal analysis. Leveraging NGS technology, it detects Copy Number Variants (CNVs) with unmatched accuracy compared to traditional cytogenetic methods.

The key to CNSeq's power lies in its strategic use of low-pass whole genome sequencing, reading each base a few times. This focuses on identifying large-scale CNVs uniformly across the genome for faster analysis and potentially deeper insights into chromosomal abnormalities.

It empowers clinicians with the ability to explore insights in detail, enabling a comprehensive grasp of structural variations in the genome with unbiased CNV calling across the genome.





Key Features



Increased CNV callers for improved detection reliability



Validated for both prenatal & pediatric settings



Advanced pipeline adjusts for maternal cell contamination in calculating mosaicism and aneuploidies

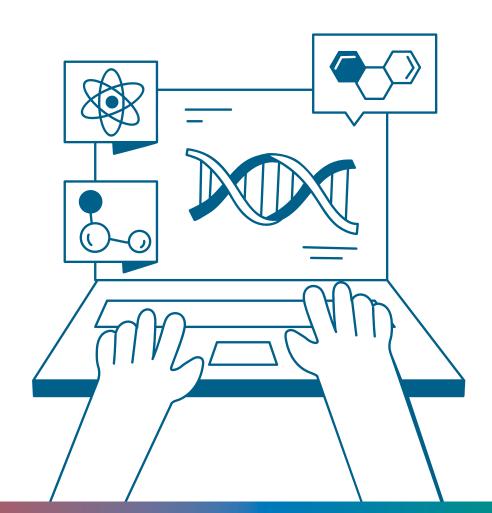




Sample Requirement
Amniotic Fluid, CVS, POC, PVB & gDNA

Going beyond traditional array with CNSeq

Technique	Karyotype	Prenatal CMA	750K	CNSeq	CNSeqHD
Coverage	>5Mb	Probe-based	Probe-based	Complete genome coverage	Complete genome coverage
Resolution	>5Mb	>500Kb	>100Kb	500Kb	>50Kb
DNA quantity requirement	Cell Based	250 ng	250 ng	50 ng	50 ng
Low-level Moscaism sensitivity	10%	>20%	>20%	>20%	>20%
Abnormalities detected	Aneuploidies, larger CNVs, Structural Abnormalities, Balanced Translocation	Aneuploidies, CNVs, UPDs, Polyploidy, AOH/LOH	Aneuploidies, CNVs, UPDs, Polyploidy, AOH	All Aneuploidies, CNVs & Structural Variants	All Aneuploidies, CNVs & Structural Variants





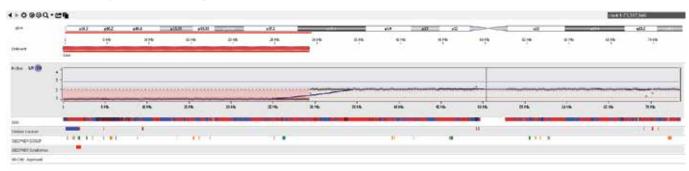
CNSeq & CNSeqHD: Validation Data

				Detected Events - CNVs & Aneuploidies (bp)					
Coriell Controls	10 - 100K	100 - 250K	250 - 500K	500K - 1M	1M - 5M	5M - 30M	>30M	Whole Chr	
CNSeq									
NA50324						• 🛦			
NA21699				•		A	•		
NA04375						A			
NA04409				A		•			
NA03623							••		
NA11385					A				
CNSeqHD									
NA21886		••••		A		A			
NA04409			•	A		•			
NA12878		****							
PCE-2	A								
PCE-3	•								
NA21699		A		•		A			

● Gain; ■ Mosaic Gain; ▲ Loss; ◆ Polymorphic event undetected; ■ Borderline gain event undetected

Unique CNVs picked up by CNSeq

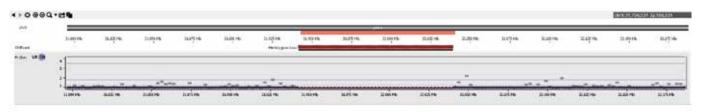
Wolf-Hirschhorn Syndrome - Pathogenic loss





Unique CNVs picked up by CNSeqHD

Congenital Heart Defect - 10kb Deletion Pathogenic Copy Number Loss



Proprietary Platforms



Additional Testing Options

- Exome Plus: Exome Sequencing
- MaatriSeq: NIPS
- AneuXpress: Quantitative Fluorescent PCR
- Sanger Based Testing

- Couple Carrier Screening
- Whole Genome Sequencing
- Preimplantation Genetic Testing:
 PGT Aneuploidy



+000,08 Genetic Tests Reported

500+ Projects Executed for Genomics Majors Globally Presence in 20+ Countries **SCAN TO KNOW MORE**







Reference Sdn Bhd (1278003-P) Wisma MEDINICS, No.2, Jalan Astaka U8/88B, Seksyen U8, Bukit Jelutong, 40150 Shah Alam, Selangor, Malaysia.

