MEDGENOME

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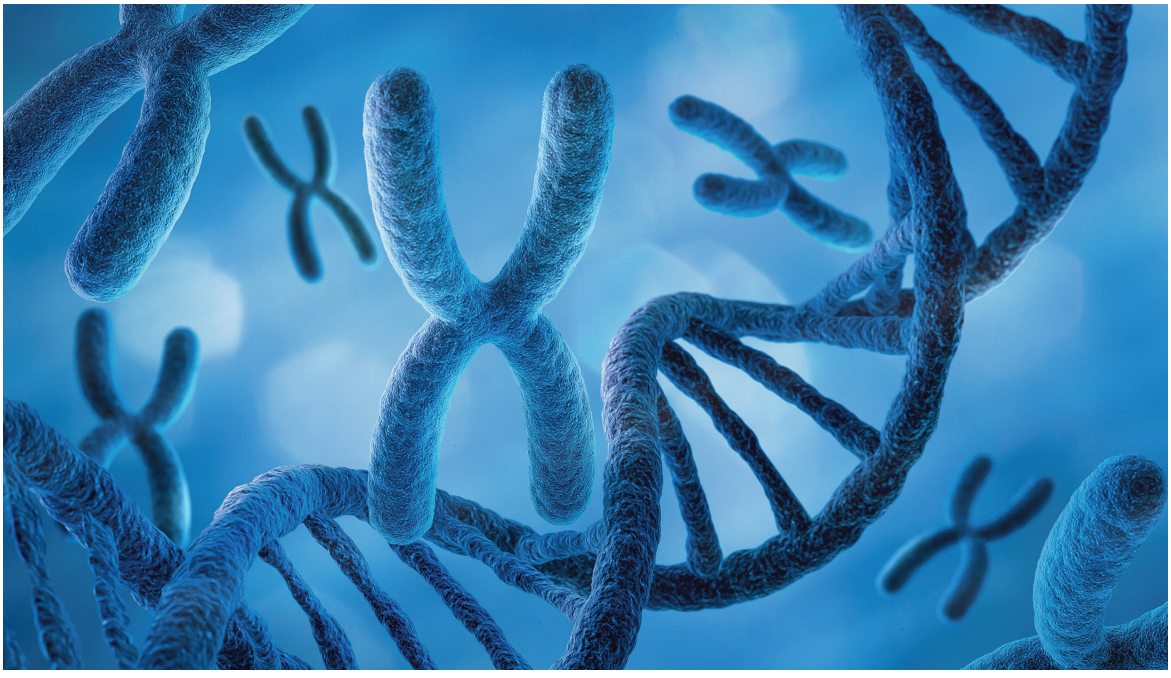
KaryoTrack

SNP array based powerful efficient test for genome-wide cytogenetic analysis



- Cost effective
- 700K Genome wide probes
- Suitable for both prenatal and postnatal samples
- Detects CNVs, ROHs, Mosaicism, UPD, Polyploidy

KaryoTrack features

- Provides a cost-effective solution for accurate detection of copy number variants (CNVs), loss of heterozygosity (LOH), mosaicism, uniparental disomy (UPD), and polyploidy.
- Targets coverage of more than 4800 cytogenetic related key genes across the genome
- Provides optimized content for a broad range of applications, ensuring high-quality and reproducible data.
- Contains ~700 K markers for high exonic coverage in regions of disease relevance, providing highly accurate copy number variation calls, and a high resolution of about 10kb for target regions.
- Uses inhouse build database (~15000 data) and research databases to support variant annotations and provides phenotype-associated variant ranking.
- Provides results that are in compliance with the current American College of Medical Genetics and Genomics (ACMG) guidelines.

Wider coverage of disease associated variants (Prenatal & Postnatal)

KaryoTrack has extensive coverage of phenotypes and diseases such as Neurological, Cancer, Cadiovascular, Endocrine & Metabolic, Hematological, Newborn and others (Figure 1a and Figure 1b).

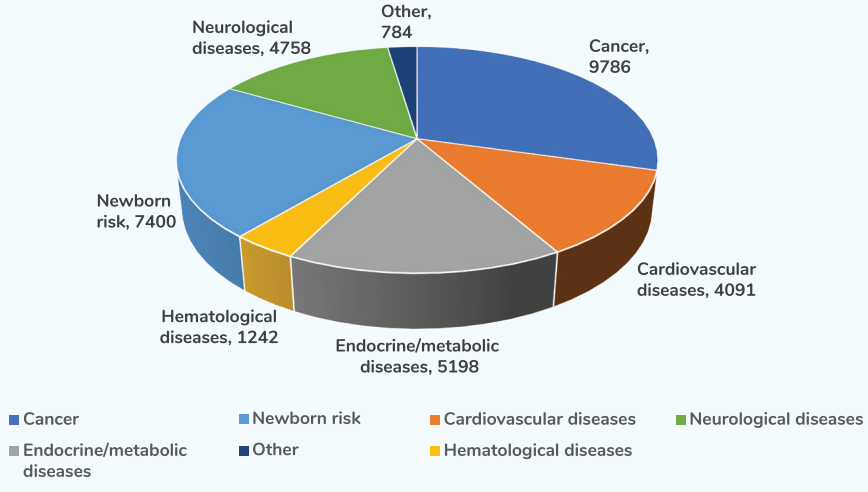


Figure 1a: Disease research content by category

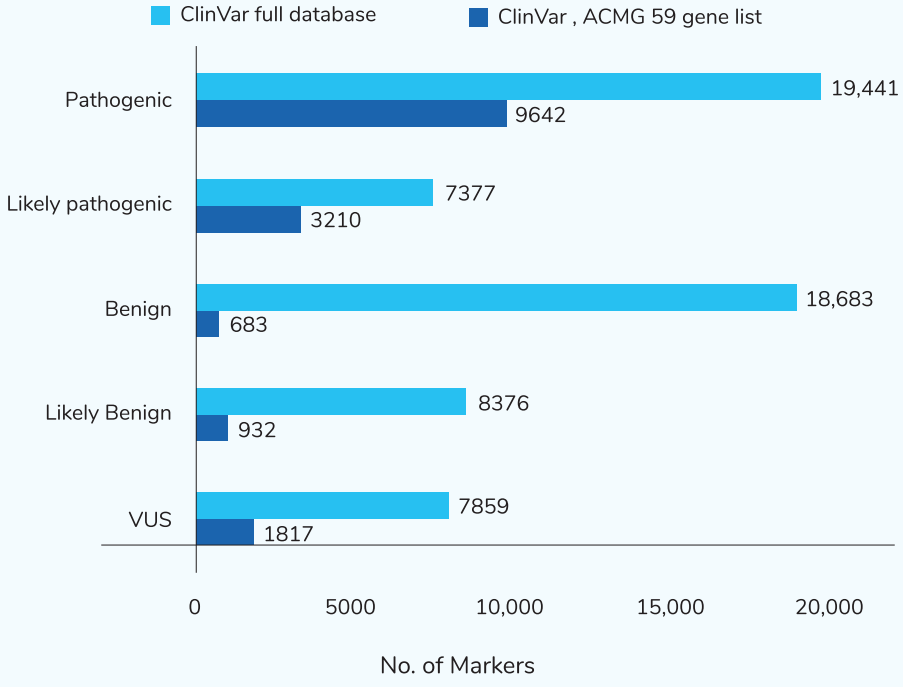


Figure 1b: Variants sorted by range of pathology classifications

Optimized spacing for Cytogenetic applications

KaryoTrack provides optimal coverage of key genes with spacing that supports CNV analysis. Cytogenetic application tiers include:

Application tier	Description	Genes in tier	Exons in tier	Average probe spacing	Median probes/exon	Exons with ≥1 probes	Exons with ≥3 probes
1	ClinGen pathogenic/likely pathogenic, haploinsufficient, and triploinsufficient	409	6214	1.25 kb	5	> 99%	> 99%
2	DDG2P Genes associated with cancer	1254	18,353	2.00 kb	4	> 99%	> 99%
3	Input from cytogenetics consortia Mendeliome Panel	2766	36,840	2.45 kb	3	> 99%	> 60%
4	OMIM Morbid Genes not otherwise tiered	456	5434	2.82 kb	3	> 99%	> 60%
	Total	4885	66,841				

a. DDG2P: developmental disorders gene to phenotype

KaryoTrack validation

KaryoTrack is an internally validated SNP array based test that has been validated using samples from various sources, including chorionic villus sampling (CVS), amniotic fluid (AF), cultured amniotic fluid, cord blood/cardiac blood products of conception tissue (POC), peripheral blood, direct DNA, and DNA isolated from cell lines or cultured cells.

KaryoTrack results have shown 100% concordance with other orthogonal method. This test can detect and report copy number variations (CNVs) from as few as a few exons (12.5 Kb) to whole chromosomal aberrations. The presence of SNP probes helps in the detection of regions of homozygosity, mosaicisms, uniparental disomy (UPD), and polyploidy.

Reports generated are in compliance with the current American College of Medical Genetics and Genomics (ACMG) guidelines.

Chipset	No. of markers	Detection of AOH/LOH	Resolution target	Spacing on tiers (Avg/Median)	% Tired exons well covered	Year Design
KaryoTrack	~700 K	<3 Mb	~10 Kb	~2 kb/~1 kb	~98%	2020

Test Details

Test Code	Test Name	Sample requirements	Sample Transportation	TAT
MGM2741	KaryoTrack	Chorionic Villus Sampling (CVS)/Amniotic Fluid (AF)/ Cultured Amniotic Fluid / Cord Blood / Cardiac Blood / Products of Conception Tissue (POC) / Peripheral Blood, Direct DNA / DNA Isolated from Cell Lines or Cultured Cells	Shipped at 2-8 °C	14 working days
MGM2758	KaryoTrack with Prenatal Cell Culture*	Amniotic Fluid (AF)		21 working days

*If required

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Talk to the Experts

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