ThermoFisher SCIENTIFIC

CytoScan HT-CMA Suite

High-throughput assay with combined CNV and SNV detection

The Applied Biosystems™ CytoScan™ HT-CMA Suite is a complete cytogenetics microarray solution for research that includes an Applied Biosystems™ CytoScan™ HT-CMA Array Plate, reagent kit, and the simple, user-friendly Reproductive Health Analysis Software (RHAS). The data in RHAS can be automatically viewed in Applied Biosystems™ Chromosomal Analysis Suite (ChAS) software. The CytoScan HT-CMA Suite was designed to provide comprehensive coverage and high performance for detecting chromosomal aberrations in a broad range of sample types for prenatal, postnatal, and oncology research applications. Along with genome-wide copy number coverage, the Applied Biosystems™ CytoScan™ HT-CMA Assay Kit also includes a panel of single-gene variants relevant to prenatal and postnatal studies as well as oncology research applications. With a workflow utilizing the Applied Biosystems™ GeneTitan™ Multi-Channel (MC) Fast Scan Instrument for automated array processing, the CytoScan HT-CMA assay delivers an efficient high-throughput cytogenetic solution.

Highlights

- High analytical specificity, analytical sensitivity [1], and resolution [2] across the genome
- Comprehensive whole-genome coverage across RefSeq, OMIM®, and DECIPHER constitutional gene regions
- Forward-looking design covering not only the regions relevant today but also the ones that may become relevant in the future
- A hybrid dual design including trusted copy number probes and the power of high-density single-nucleotide polymorphisms (SNPs) for confident breakpoint determination, allelic confirmation of copy number changes [3], high-resolution detection of loss of heterozygosity (LOH) or absence of heterozygosity (AOH) [4], gene-level homozygosity mapping [5], parent-of-origin analysis [6], detection of mosaics, and detection of genomic contamination
- 1.1 million markers for copy number analysis, including 1 million SNP and 130,000 nonpolymorphic probes
- Enables consolidated testing with up to 178 relevant single-nucleotide variants (SNV) across 36 genes (*CFTR*, *DMD*, *FGFR3*, *HbA*, deafness-related, inborn errors of metabolism-related, and others) and a module for study of spinal muscular atrophy
- Exon-level coverage in select genes: *DMD*, *MECP2*, *MBD5*, *CFTR*, *HBB*, *MCOLN1*, and *NEB*
- Automated high-throughput design enhances cost effectiveness by increasing technician productivity and minimizing low hands-on time
- Batching of 96 samples per array plate with a 3-day turnaround time
- Robust proprietary manufacturing technology that produces highly reproducible arrays between batches, with no risk of probe dropout that occurs with bead array technology
- A robust manual or automated assay designed to save you time and money, reduce error, and deliver performance, results, and quality consistent with your research laboratory requirements
- User-friendly RHAS for cytogenetic and variant analysis enables visualization and summarization of chromosomal aberrations, and genotyping of specified variants; the ChAS software allows simple data analysis and generation of reports based on your specific requirements; the software adapts to the needs of any cytogenetics laboratory, from single analysis to database generation
- World-class support, from training and instrument maintenance to consulting and compliance, led by our multilingual team of technical specialists

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CytoScan HIT-CMA Array specifications

Markers used for copy number analysis	
Total number of markers	1,162,042
Number of nonpolymorphic markers	133,823
Number of SNP markers	1,028,219

Markers used for allele difference and B-allele frequencies (BAFs)	
Number of SNP markers	1,028,219

Performance specifications*	
Genome build used for development	hg38
Recommended mass of input gDNA	100 ng
Minimum resolution for losses	≥25 markers and 100 kb
Minimum resolution for gains	≥50 markers and 400 kb
Resolution for runs of homozygosity (ROH)	≥5 Mb
Mosaicism, limit of detection	≥20%

* Size of aberration (gain/loss/ROH/mosaicism)—the size of the segment call depends on the average marker spacing in the region. Best performance can be achieved in regions with higher marker coverage. Mosaicism detection may depend on the size of the altered segment and the type of aberration involved.

Marker distribution and spacing	
Number of autosomal markers	1,070,945
Number of pseudoautosomal markers	750
Number of pericentric markers	3,396
Number of subtelomeric markers	9,877
Number of intragenic markers	713,479
Number of intergenic markers	448,563
Average intragenic spacing (bp)	1,729
Average intergenic spacing (bp)	4,245
Average spacing (gene and non-gene backbone, bp)	2,700

Percentage of genes having ≥25 markers per 100 kb	
Clinical genes and gene regions (ClinGen, OMIM Morbid Map, and DECIPHER) (5,171)	92.6%
ClinGen (1,185)	99.7%
OMIM morbid genes (4,397)	95.7%
DECIPHER genes (1,949)	96.3%
RefSeq genes (21,784)	64.1%
Cancer genes (551)	85.1%

Ordering information

Product	Description	Cat. No.
CytoScan HT-CMA consumables and software		
CytoScan HT-CMA 96F Assay Kit	Arrays and reagents sufficient to process 96 samples	906025
Reproductive Health Research Analysis Suite (RHAS)	Available as a free download from thermofisher.com/chas	NA
Chromosome Analysis Suite (ChAS)	Available as a free download from thermofisher.com/chas	NA
CytoScan HT-CMA training products		
CytoScan HT-CMA 96F Assay Training Kit	Arrays, reagents, and control samples sufficient to process two runs of 96 samples	906027
CytoScan FAS On-Site Training	FAS-led on-site preparation and first week of training	000802
CytoScan FAS Assisted Training	FAS-led on-site preparation; customer completes training using self-paced tools	000803
Supporting products		
GeneTitan MC Fast Scan Instrument	Automated array-processing instrument required to hybridize, wash, stain, and scan arrays	00-0373
NIMBUS Target Preparation Instrument	Robotics workstation and laptop	00-0401

References

1. South ST et al. (2013) ACMG standards and guidelines for constitutional cytogenomic microarray analysis, including postnatal and prenatal applications: revision 2013. *Genet Med* 15(11):901–909.
2. Zimmerman E, Maron JL (2016) *FOXP2* gene deletion and infant feeding difficulties: a case report. *Cold Spring Harb Mol Case Studies* 2:a000547.
3. Rodriguez-Pascau L et al. (2012) Characterization of two deletions involving *NPC1* and flanking genes in Niemman-Pick type C disease patients. *Mol Genet Metab* 107(4):716–720.
4. Mason-Suares H et al. (2013) Density matters: comparison of array platforms for detection of copy number variation and copy-neutral abnormalities. *Genet Med* 15(9):706–712.
5. Mayer A et al. (2016) Homozygosity mapping and whole-genome sequencing reveals a deep intronic *PROM1* mutation causing cone-rod dystrophy by pseudoexon activation. *Eur J Hum Genet* 24(3):459–462.
6. Darcy D et al. (2015) Mosaic paternal genome-wide uniparental isodisomy with Down syndrome. *Am J Med Genet Part A* 167(10):2463–2469.

For additional instrument system configurations or individual system components to meet your needs, please contact your account manager.

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