CytoScan HD Suite

Optimized for cytogenetics research

The Applied Biosystems™ CytoScan™ HD Suite is a complete cytogenetics microarray solution that includes Applied Biosystems™ CytoScan™ HD Arrays, a reagent kit, and intuitive, user-friendly Applied Biosystems™ Chromosome Analysis Suite (ChAS) software. The CytoScan HD Suite was designed to provide the most comprehensive whole-genome coverage and highest performance for detecting chromosomal aberrations in a broad range of sample types for constitutional, cancer, stem cell, and neurodevelopmental applications. The CytoScan HD Suite supports various sample types for analysis of cancer and constitutional cytogenetic research, including blood, bone marrow, buccal swabs, saliva, fresh and frozen tissues, cultured and uncultured cells, amniocytes and products of conception (POC), and fresh as well as formalin-fixed, paraffin-embedded, unarchived specimens.

Highlights

- High specificity, sensitivity [1], dynamic range [2], and resolution [3] across the genome
- Superior coverage across entries in OMIM® database, RefSeq, ClinGen, DECIPHER/DDD constitutional regions, and the COSMIC Cancer Gene Census (CGC)
- Forward-looking design, with dense probe coverage of regions known to be relevant today as well as regions that may become relevant in the future
- The hybrid, dual-probe design includes both copy number probes empirically selected for performance and SNPs chosen for their high minor-allele frequency to exhibit the best separation of allele tracks. The high-density SNPs allow for confident breakpoint determination [4], independent allelic (or SNP) confirmation of copy number changes [5], high-resolution loss/absence of heterozygosity (LOH/AOH) [6], gene-level homozygosity mapping [7], parent-of-origin analysis [8], enhanced detection of low-level mosaics [9], clonality [10], genomic contamination, and ploidy adjustments and detection [11]
- High-density SNPs with >99% genotype accuracy enable visualization of low-level mosaicism, absence of heterozygosity (AOH) and acquired UPD (aUPD) detection, copy number change confirmation, triploidy detection, allelic imbalance pattern visualization, genomic contamination identification, trio consistency checking, and parent-of-origin analysis
- 2.67 million markers for copy number analysis, including 750,000 SNPs and 1.9 million nonpolymorphic probes
- Advanced, proprietary manufacturing technology that produces highly reproducible arrays between batches, with no risk of probe dropout that occurs with bead array technology
- Proven technology, extensively cited, with more than 250 publications per year not only in constitutional and cancer applications but also in neurodevelopmental and stem cell research
- A robust and flexible manual or automated assay, designed to save you time and money, reduce error, and deliver performance, results, and quality consistent with your laboratory requirements
- Advanced software tailored for cytogenetics and copy number analysis, ChAS software allows simple data analysis and generation of customized exports based on your specific requirements; the software adapts to the needs of any cytogenetics laboratory, from single sample analysis to database generation, and from constitutional tools to cancer algorithms
- World-class support, from training and instrument maintenance to consulting and compliance, led by our experienced team of multilingual professionals
- The CytoScan HD assay labels fragmented DNA with a DNA-labeling reagent (biotin transfer) and then stains the labeled hybridized target with streptavidin-phycoerythrin (SAPE); phycoerythrin is the fluorophore

CytoScan HD Array specifications

Markers for copy number analysis	
Total number of probes	6,876,796
Number of nonpolymorphic markers	1,953,246
Number of SNP markers	743,304
Total number of SNP markers suitable for genotyping	749,157
Genome build	hg19
Autosomal markers	2,491,915
Pseudoautosomal markers	4,624
Intragenic markers	1,410,535
Intergenic markers	1,286,015
Mosaicism	>15%
Input gDNA	250 ng*
Minimum resolution for losses	25 kb
Minimum resolution for gains	50 kb
Resolution for LOH/AOH	3 Mb

* Customers have reported success using as little as 10 ng of starting DNA.

Average marker spacing (base pairs)	
Intragenic (within all the genes below)	880
Intergenic (nongene backbone)	1,737
Overall (gene and nongene backbone)	1,148
Percentage of genes covered (25 markers/100 kb)	
ClinGen (formerly ICCG and ISCA) (3,483)	100%
Cancer genes (526)	100%
OMIM Morbid genes (3,561)	100%
X chromosome OMIM Morbid genes (177)	100%
RefSeq genes (36,121)	98%
DDD [12] (1,309)	98%

Customer support

With our comprehensive onboarding service and support offerings for ChAS software, the team of experienced professionals, including technical sales specialists, field service engineers, field application scientists, and clinical application consultants, ensures your confidence with the advanced features of typical workflows. Learn more from our service and support **brochure**.

References

1. South ST et al. (2013) ACMG Standards and Guidelines for constitutional cytogenomic microarray analysis, including postnatal and prenatal applications: revision 2013. *Genetics in Medicine* 15(11):901–909.
2. Ambros I et al. (2014) Ultra-high density SNParray in neuroblastoma molecular diagnostics. *Frontiers in Oncology* 4:202.
3. Zimmerman E, Maron JL (2016) *FOXP2* gene deletion and infant feeding difficulties: a case report. *Cold Spring Harbor Molecular Case Studies* 2:a000547.
4. Rodriguez-Pascau L et al. (2012) Characterization of two deletions involving *NPC1* and flanking genes in Niemman-Pick type C disease patients. *Molecular Genetics and Metabolism* 107(4):716–720.
5. Chen W et al. (2013) Identification of chromosomal copy number variations and novel candidate loci in hereditary nonpolyposis colorectal cancer with mismatch repair proficiency. *Genomics* 102(1):27–34.
6. Mason-Suares H (2013) Density matters: comparison of array platforms for detection of copy number variation and copy-neutral abnormalities. *Genetics in Medicine* 15(9):706–712.
7. Mayer A et al. (2016) Homozygosity mapping and whole-genome sequencing reveals a deep intronic *PROM1* mutation causing cone-rod dystrophy by pseudoexon activation. *European Journal of Human Genetics* 24(3):459–462.
8. Darcy D et al. (2015) Mosaic paternal genome-wide uniparental isodisomy with Down syndrome. *American Journal of Medical Genetics Part A* 167(10):2463–2469.
9. Oneda B et al. (2014) High-resolution chromosomal microarrays in prenatal diagnosis significantly increase diagnostic power. *Prenatal Diagnosis* 34(6):525–533.
10. Jiangchuan T et al. (2014) Concurrence of B-lymphoblastic leukemia and myeloproliferative neoplasm with copy neutral loss of heterozygosity at chromosome 1p harboring a *MPL* W515S mutation. *Cancer Genetics* 207(10–12):489–494.
11. Choi S et al. (2014) Near-haploid B lymphoblastic leukemia with an apparent hyperdiploid karyotype: the critical role of SNP analysis in establishing proper diagnosis. *Journal of Hematopathology* 7(1):27–32.
12. Fitzgerald TW et al. (2015) Large-scale discovery of novel genetic causes of developmental disorders. *Nature* 519(7542):223–228.

Ordering information

Product	Description	Cat. No.
CytoScan HD Suite consumables		
CytoScan HD Array and Reagent Kit Bundle	Arrays and reagents sufficient for 24 reactions	901835
Chromosome Analysis Suite (ChAS) software	Available as a free download from **thermofisher.com/chas**	NA
CytoScan HD Kit Plus 24	Includes: • CytoScan HD arrays and reagents for 24 reactions • CytoScan Amplification Kit for 96 reactions	905824
CytoScan HD Kit Plus 96	Includes: • CytoScan HD arrays and reagents enough for 96 reactions • CytoScan Amplification Kit for 96 reactions	905896
CytoScan training products		
CytoScan HD Training Kit	Arrays and reagents sufficient for 24 reactions, plus training materials	901834
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		
GeneChip 3000 7G with Workstation and AutoLoader	Includes: • GeneChip Scanner 3000 7G with AutoLoader • n2D Handheld Barcode Reader • GeneChip Fluidics Station 450 • GeneChip Hybridization Oven 645 • Computer workstation with instrument control software	00-0218
GeneChip System 3000Dx v.2*	Includes: • GeneChip Scanner 3000Dx v.2 with AutoLoaderDx • GeneChip Fluidics Station 450Dx v.2 • Workstation with Affymetrix Molecular Diagnostics Software	00-0334
GeneChip Hybridization Oven 645		00-0331
NIMBUS Target Preparation Instrument	Robotics workstation and laptop	00-0401

* Recommended: GeneChip Hybridization Oven 645.

Chromosome Analysis Suite (ChAS) software

Leading genetic data analysis software that continues to evolve along with the needs of your laboratory

ChAS provides an intuitive and flexible suite of software for cytogenetic analysis that enables you to view and summarize chromosomal aberrations across the genome. Chromosomal aberrations may include copy number gain or loss, mosaicism, and loss of heterozygosity (LOH).

ChAS software is available to customers for free. Boost your confidence with advanced genetic data analysis now.

To request a demo, visit **thermofisher.com/chasdemo**

Key features of ChAS software

- Analyze copy number, mosaicism, and LOH segment data at different levels of resolution
- Automatically prioritize segment data using ACMG-inspired scoring
- Customize and load your own annotations and regions for focused analysis
- Store, query, and display historic sample data and annotations for streamlined analysis
- Directly access NCBI, UCSC Genome Browser, DECIPHER, ClinVar, ClinGen, Ensembl, and OMIM® databases and others
- Export user-selected data in formats like browser extensible data (BED), Applied Biosystems™ Affymetrix™ extensible data (AED), and variant call format (VCF) files

The new ChAS software 4.3: Faster data analysis at your fingertips

- A new Mosaic Segmentation Algorithm
- Additional data types supported in VCF
- APIs to push and pull segment coordinates in and out of ChAS software
- Support for multiple input/output folders for Automatic Cel Analysis
- Include QC metrics plus Frag QC from Automatic Cel Analysis in QC history file
- Additional annotation track to complete the OMIM morbidity map

ChAS software training videos

We offer on-demand training videos for you to compare your analysis pipeline and see new and enhanced features you may want to incorporate.

For training modules, visit **thermofisher.com/chastraining**

"I have been using ChAS software to analyze and interpret cytogenomic microarrays in our cytogenetics lab and find it invaluable in my daily work as a lab director. I appreciate the many tools for CNV interpretation that are available in the software, yet it is user-friendly and easy to navigate. I have been impressed with the improved mosaic segment detection in the newest version that has allowed us to identify low-level mosaic CNV that were not observed previously."

Ferrin C. Wheeler, PhD
Medical Director, Cytogenetics Laboratory
Associate Director, Molecular Diagnostics and Clinical Genomics Laboratory
Vanderbilt University Medical Center

Find out more at **thermofisher.com/microarrays**