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, the Applied Biosystems™ GeneChip™ System 3000 for array processing, and intuitive, user-friendly Applied Biosystems™ Chromosome Analysis Suite (ChAS) software. Laboratories may boost discovery yield and simplify variant interpretation with the Applied Biosystems™ CytoScan™ Automated Interpretation and Reporting (AIR) solution that unites ChAS software with Franklin (by Genoox), an end-to-end, artificial intelligence (AI)-driven research solution for automating genetic data analysis.

The CytoScan HD Suite provides comprehensive whole-genome coverage and advanced performance for detecting chromosomal aberrations. The CytoScan HD Suite supports various sample types for constitutional and oncology research applications, including blood, buccal swabs, saliva, uncultured or cultured cells, fresh and frozen tissue, chorionic villi, amniocytes, and products of conception (POC).

Highlights

- High specificity, sensitivity [1], dynamic range [2], and resolution [3] across the genome
- Exceptional coverage across entries in the 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 research applications but also in neurodevelopmental and stem cell research
- A robust and flexible manual or automated assay, designed to save time and money, reduce error, and deliver performance, results, and quality consistent with stringent laboratory requirements
- Intuitive software for cytogenetics and copy number analysis, ChAS software allows simple data analysis and generation of customized exports based on your specific requirements. ChAS software adapts to a variety of needs in cytogenetics laboratories, from single-sample analysis to database generation, and from constitutional tools to cancer algorithms
- Automated genetic analysis with the power of AI for quick and precise copy number variant interpretation with the CytoScan AIR solution

- World-class support, from training and instrument maintenance to consulting and compliance, led by our experienced, multilingual team of 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 markers	2,696,168
Number of nonpolymorphic markers	1,953,038
Number of SNP markers	743,130
Markers used for allele differences and BAFs	
Number of SNP markers	796,197
Performance specifications	
Genome build used for development	hg19
Recommended mass of input gDNA*	>250 ng
Minimum resolution for losses	≥25 markers and 25 kb
Minimum resolution for gains	≥50 markers and 50 kb
Resolution for ROH	≥3 Mb
Mosaicism, limit of detection	≥15%

* 250 ng is optimal, but users have reported success using as little as 10 ng of starting DNA.

Marker distribution and spacing	
Number of autosomal markers	2,491,598
Number of pseudoautosomal markers	4,620
Number of intragenic markers	1,449,255
Number of intergenic markers	1,246,913
Average intragenic spacing (bp)	851
Average intergenic spacing (bp)	1,527
Average spacing (gene and non-gene backbone, bp)	1,164
Percentage of genes having ≥25 markers/100 kb (in hg38)	
Clinical genes and regions (ClinGen, OMIM Morbid, and DECIPHER databases) (5,171)	98.4%
ClinGen (1,185)	99.0%
OMIM Morbid genes (4,397)	98.0%
Decipher genes (1,949)	99.0%
RefSeq genes (21,784)	94.0%

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, and field application scientists, can boost 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.

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
CytoScan HD Kit Plus 24	Arrays and reagents for 24 reactions with amplification kit	905824
CytoScan HD Kit Plus 96	Arrays and reagents for 96 reactions with amplification kit	905896
Analysis software		
Chromosome Analysis Suite (ChAS) Software	Available as a free download from **thermofisher.com/chas**	NA
CytoScan Automated Interpretation and Reporting (AIR) Tokens	24 tokens	00.1001
	96 tokens	00.1003
	384 tokens	00.1004
CytoScan training products		
CytoScan HD Training Kit	Arrays and reagents sufficient for 24 reactions for training purposes	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 System 3000	Includes: • GeneChip Scanner preassembled with AutoLoader • GeneChip Fluidics Station 450 • GeneChip Hybridization Oven 645i • Workstation with GeneChip Data Collection Software	00-0218
GeneChip Fluidics Station 450	Single station available to be purchased separately from the GeneChip System 3000	00-0079
GeneChip Hybridization Oven 645i	Single unit available to be purchased separately from the GeneChip System 3000	00-0331
NIMBUS Target Preparation Instrument	Robotics workstation and laptop	00-0401

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.

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 historical sample data and annotations for streamlined analysis
- Use application programming interfaces (APIs) to push and pull segment coordinates in and out of ChAS software
- Automatically generate a results file with no manual setup required

The new ChAS software 4.4: faster data analysis at your fingertips

- Seamless integration with Franklin (by Genoox) using the CytoScan AIR solution
- Whole-genome segmentation for large copy number aberrations on the Applied Biosystems™ CytoScan™ XON array
- Flag segments to bypass filter settings
- Display different LOH segment colors based on median copy number
- pHaplo and pTriplo scores
- ClinGen-curated regions in a recurrent/curated regions track
- Library files can be downloaded securely from the NetAffx™ server via https communication

CytoScan Automated Interpretation and Reporting (AIR) solution

Franklin (by Genoox) is an end-to-end, AI-driven research solution for genetic data analysis. With CytoScan AIR, users can combine the power of ChAS and Franklin to augment visualization of CNV gains, losses, and LOH with clinical research interpretation information.

- **Fast data interpretation and reporting**—results available in seconds so you can focus on discovery
- **Improved evidence support**—options for segment interpretation, including the most up-to-date American College of Medical Genetics (ACMG) classifications, phenotype matching, literature searches, and historical data
- **Customized reporting**—intuitive interface enables easy customization, reporting, evaluation, and sign-off
- **Evidence-based database development**—access to Franklin enables users to link evidence with observations for internal database management and expansion
- **Relevant insights**—more than 350,000 shared variant classifications and advanced findings from community-driven cytogenetics research

Request the CytoScan AIR **demo video**.

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