CytoScan 750K Suite

Coverage without compromise

The Applied Biosystems™ CytoScan™ 750K Suite is a complete cytogenetics microarray solution that includes Applied Biosystems™ CytoScan™ 750K Arrays, a reagent kit and Applied Biosystems™ Chromosome Analysis Suite (ChAS) software. The CytoScan 750K Suite was designed to provide the most comprehensive 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 750K Suite supports various sample types for analysis of constitutional cytogenetic research, including blood, bone marrow, buccal swabs, saliva, fresh and frozen tissues, direct/cultured cells, amniocytes and products of conception (POC), and fresh as well as formalin-fixed, paraffin-embedded, unarchived specimens.

Highlights

- High specificity, sensitivity [1], and resolution [2] across the genome
- Comprehensive whole-genome coverage across entries in OMIM® database, RefSeq, ClinGen, DECIPHER/DDD constitutional regions, and the COSMIC Cancer Gene Census (CGC)
- 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 not only the best of copy number probes but also the power of high-density SNPs for confident breakpoint determination [3], allelic confirmation of copy number changes [4], high-resolution loss/absence of heterozygosity (LOH/AOH) [5], gene-level homozygosity mapping [6], parent-of-origin analysis [7], enhanced detection of low-level mosaics [8], clonality [9], genomic contamination, and ploidy adjustments and detection [10]
- 750,000 markers for copy number analysis, including 200,000 SNPs and 550,000 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
- User-friendly software tailored for cytogenetics and copy number analysis, ChAS software allows for 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 technical professionals
- The CytoScan 750K 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

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CytoScan 750K Array specifications

Total number of probes on the microarray	
Total number of probes	1,768,397
Markers used for copy number analysis	
Total number of markers	750,344
Number of nonpolymorphic markers	549,916
Number of SNP markers	200,428
Markers used for allele difference and BAFs	
Number of SNP markers	200,550
Performance specifications	
Genome build used for development	hg19
Recommended mass of input gDNA*	250 ng
Minimum resolution for losses	≥25 markers and 200 kb
Minimum resolution for gains	≥50 markers and 400 kb
Resolution for ROH**	5 Mb
Mosaicism, limit of detection	≥20%

* 250 ng is optimal, but users have reported success using as little as 10 ng of input DNA.
** ROH: runs of homozygosity.

Marker distribution and spacing	
Number of autosomal markers	702,222
Number of psuedoautosomal markers	994
Number of intragenic markers	386,011
Number of intergenic markers	364,333
Average intragenic spacing (bp)	3,195
Average intergenic spacing (bp)	5,526
Average spacing (gene and non-gene backbone, bp)	4,181
Percentage of genes having ≥25 markers/100 kb (using hg38)	
Clinical genes and regions (ClinGen, OMIM Morbid, and DECIPHER) (5,171)	82.0%
ClinGen (1,185)	88.3%
OMIM Morbid genes (4,397)	82.0%
DECIPHER genes (1,949)	83.7%
RefSeq genes (21,784)	73.2%

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. Zimmerman E, Maron JL (2016) *FOXP2* gene deletion and infant feeding difficulties: a case report. *Cold Spring Harbor Molecular Case Studies* 2:a000547.
3. Kim KB et al. (2014) Prenatal diagnosis of a 7q21.13q22.1 deletion detected using high-resolution microarray. *Obstetrics & Gynecological Science* 57(4):318–324.
4. Liu WQ et al. (2015) Genetic evaluation of copy number variations, loss of heterozygosity, and single-nucleotide variant levels in human embryonic stem cells with or without skewed X chromosome inactivation. *Stem Cells and Development* 24(15):1779–1792.
5. 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.
6. 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.
7. 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.
8. Oneda B et al. (2014) High-resolution chromosomal microarrays in prenatal diagnosis significantly increase diagnostic power. *Prenatal Diagnosis* 34(6):525–533.
9. Sudesh P et al. (2015) Mosaic 22q11.2 deletion and tetralogy of Fallot with absent pulmonary valve. *World Journal for Pediatric & Congenital Heart Surgery* 6(2):342–345.
10. 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.
11. 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 750K Suite consumables		
CytoScan 750K Array and Reagent Kit Bundle	Arrays and reagents sufficient for 24 reactions	901859
CytoScan 750K Kit Plus 24 (Available outside US/Canada only)	Includes: • CytoScan 750K arrays and reagents for 24 reactions • CytoScan Amplification Kit for 96 reactions	905924
CytoScan 750K Kit Plus 96 (Available outside US/Canada only)	Includes: • CytoScan 750K arrays and reagents for 96 reactions • CytoScan Amplification Kit for 96 reactions	905996
CytoScan 750K Training Kit	Arrays and reagents sufficient for 24 reactions, plus training materials	901860
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
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 for purchase separately from the GeneChip System 3000	00-0334
GeneChip Hybridization Oven 645i	Single unit available for purchase separately from the GeneChip System 3000	00-0331

Chromosome Analysis Suite (ChAS) software

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

ChAS is 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

Enhance your genetic data analysis with the new ChAS software 4.5

- ChAS CEL Uploader installed in the workstation
- Option to change the default administrator password for increased security
- Left–right scroll button within the “Detail View” for easier scrolling
- Better navigation and tracking of OMIM genes, including disorder-causing genes with a phenotype map key value of three

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 historic 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 cytogenetic research

View the CytoScan AIR **demo video**.

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