CytoScan 750K Accel Suite

Reliable and reproducible results with an accelerated 2-day workflow and improved coverage

The Applied Biosystems™ CytoScan™ 750K Accel Suite is an advanced cytogenetics microarray solution that includes the Applied Biosystems™ CytoScan™ 750K Accel Array, a reagent kit, the Applied Biosystems™ GeneChip™ System 3000 for array processing, and intuitive, user-friendly Applied Biosystems™ Chromosome Analysis Suite (ChAS) software. Research 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 750K Accel assay offers an accelerated 2-day workflow that may help you improve lab productivity by up to 100% compared to the CytoScan 750K assay.

The CytoScan 750K Accel Suite enables comprehensive whole-genome coverage and superb performance for detecting chromosomal aberrations in a broad range of sample types for constitutional and oncology research applications.

Highlights

- The CytoScan 750K Accel assay workflow can be completed in just 2 days.
- The assay input amount is 100 ng of genomic DNA, which is 50% less than some other commercially available chromosomal microarrays (CMAs).
- The CytoScan 750K Accel assay generates consistent target that is hybridized to the CytoScan 750K Accel Array to yield reproducible and reliable results.
- The reference model file includes challenging sample types to help enable the generation of higher-quality results. These sample types include buccal swabs, saliva, products of conception, amniotic fluid, and chorionic villus sampling (CVS).
- High specificity, sensitivity, dynamic range, and resolution across the genome.
- Improved coverage in more than 5,000 regions 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 B-allele frequency (BAF) and allele tracks. The high-density SNPs allow for confident breakpoint determination [1], independent allelic (or SNP) confirmation of copy number changes [2], high-resolution loss/absence of heterozygosity (LOH/AOH) analysis [3], gene-level homozygosity mapping [4], parent-of-origin analysis [5], enhanced detection of low-level mosaics [6], clonality determination [7], genomic contamination identification, and ploidy adjustments and detection [8].
- 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, identification of genomic contamination, trio consistency checking, and parent-of-origin analysis.
- 960,000 markers for copy number analysis, including 255,000 SNPs and 700,000 nonpolymorphic markers.
- Advanced, proprietary manufacturing technology that produces highly reproducible arrays between batches, with no risk of probe dropout that occurs with bead array technology.

- Intuitive software 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 research laboratory, 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 **CytoScan AIR**.
- World-class support, from training and instrument maintenance to consulting and compliance, led by our experienced, multilingual team of professionals.
- The CytoScan 750K Accel 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.

Accelerated CytoScan 750K Accel Array workflow from DNA digestion through CMA scanning

Day 1 ----------> Overnight --> Day 2

Digestion	Ligation	PCR amplification	Purification	Fragmentation	Labeling	Start of hybridization	Hybridization	Washing/ staining	Scanning
0.5 hr	0.5 hr	1.5 hr	1 hr	1 hr	1 hr	0.5 hr	16–18 hr	2.5 hr	0.25 hr*
		25 cycles							
		Break			Break				

* Dependent on number of cartridges.

CytoScan 750K Accel Array specifications

Markers used for copy number analysis	
Total number of markers	960,755
Number of nonpolymorphic markers	706,054
Number of SNP markers	254,701

Markers used for allele differences and B-allele frequencies (BAFs)	
Number of SNP markers	254,822

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 ROH	≥5 Mb
Mosaicism, limit of detection	≥20%

Marker distribution and spacing	
Number of autosomal markers	903,242
Number of pseudoautosomal markers	1,538
Number of intragenic markers	513,958
Number of intergenic markers	446,797
Average intragenic spacing (bp)	2,400
Average intergenic spacing (bp)	4,261
Average spacing (gene and nongene backbone, bp)	3,265

Percentage of genes having ≥25 markers/100 kb	
Clinical genes and regions (ClinGen, OMIM Morbid, and Decipher) (5,171)	89.2%
ClinGen (1,185)	92.8%
OMIM Morbid genes (4,397)	89.1%
Decipher genes (1,949)	91.0%
RefSeq genes (21,784)	78.8%

Customer support

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

Ordering information

Product	Description	Cat. No.
CytoScan 750K Accel Suite consumables		
CytoScan 750K Accel Array and Reagent Kit Bundle	Arrays and reagents sufficient for 24 reactions	952538
CytoScan 750K Accel Kit Plus 24	Arrays and reagents for 24 reactions with amplification kit	952540
CytoScan 750K Accel Kit Plus 96	Arrays and reagents for 96 reactions with amplification kit	952541
CytoScan 750K Accel Training Kit	Arrays and reagents sufficient for 24 reactions to perform assay training	952539
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	00-0079
GeneChip Hybridization Oven 645i	Single unit available for purchase separately from the GeneChip System 3000	00-0331

References

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2. 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.
3. Mason-Suares H (2013) Density matters: comparison of array platforms for detection of copy number variation and copy-neutral abnormalities. *Genet Med* 15(9):706–712.
4. 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.
5. Darcy D et al. (2015) Mosaic paternal genome-wide uniparental isodisomy with Down syndrome. *Am J Med Genet A* 167(10):2463–2469.
6. Oneda B et al. (2014) High-resolution chromosomal microarrays in prenatal diagnosis significantly increase diagnostic power. *Prenat Diagn* 34(6):525–533.
7. 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 Genet* 207(10–12):489–494.
8. 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. *J Hematop* 7(1):27–32.

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/750kaccel**