Microarrays

CytoScan Dx Assay

Whole-genome analysis with a US FDA–cleared and EU IVDR–compliant assay

Introduction

The Applied Biosystems™ CytoScan™ Dx Assay is a whole-genome diagnostic test to aid physicians in identifying the underlying genetic cause of developmental delay, intellectual disability, congenital anomalies, or dysmorphic features in children. **The CytoScan Dx Assay is cleared by the US Food and Drug Administration (FDA) and compliant with the EU *In Vitro* Diagnostic Regulation (IVDR) 2017/746.**

The complete Applied Biosystems™ CytoScan™ Dx Cytogenetics Suite is an FDA-cleared, IVDR-compliant sample-to-insight solution that includes the Applied Biosystems™ CytoScan™ Dx Array, a reagent kit, the Applied Biosystems™ GeneChip™ System 3000Dx platform for array processing, and user-friendly Applied Biosystems™ Chromosomal Analysis Suite (ChAS) Dx Software.

Highlights

- A robust and flexible manual assay, designed to save time and money, reduce error, and deliver performance, results, and quality consistent with stringent laboratory requirements
- High specificity, sensitivity, dynamic range, and resolution across the genome
- Exceptional coverage across entries in the OMIM® database, RefSeq, ClinGen, and DECIPHER/DDD constitutional regions
- 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, independent allelic (or SNP) confirmation of copy number changes, high-resolution loss/absence of heterozygosity (LOH/AOH), gene-level homozygosity mapping, parent-of-origin analysis, enhanced detection of low-level mosaics, clonality, genomic contamination, and ploidy adjustments and detection.
- High-density SNPs with >99% genotype accuracy enable visualization of low-level mosaicism, absence of heterozygosity (AOH) and uniparental disomy (UPD) detection, copy number change confirmation, triploidy detection, allelic imbalance pattern visualization, genomic contamination identification, trio consistency checking, and parent-of-origin analysis
- 2.69 million markers for copy number analysis, including 743,318 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
- User-friendly software for cytogenetics and copy number analysis, **ChAS Dx Software**, allows simple data analysis and generation of customized exports based on your specific requirements. ChAS Dx software has an intuitive graphical interface for streamlined analysis workflows, ISCN array nomenclature, and links to databases* to support data analysis workflows.

* Links in ChAS Dx Software to external databases such as Database of Genomic Variants (DGV) have not been evaluated or curated by Thermo Fisher Scientific.

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CytoScan Dx Array specifications

Markers for copy number analysis	
Total number of markers	2,696,564
Number of nonpolymorphic markers	1,953,246
Number of SNP markers	743,318
Markers used for allele differences and BAFs	
Number of SNP markers	796,461
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	≥20%

Marker distribution and spacing	
Number of autosomal markers	2,491,919
Number of pseudoautosomal markers	4,624
Number of intragenic markers	1,535,333
Number of intergenic markers	1,161,231
Average intragenic spacing (bp)	916
Average intergenic spacing (bp)	1,365
Average spacing (gene and nongene backbone, bp)	1,079
Percentage of genes having ≥25 markers/100 kb (hg19)	
Clinical genes and regions (ClinGen, OMIM Morbid Map, and DECIPHER) (5,148)	98.4%
ClinGen (1,172)	99.3%
OMIM morbid genes (4,388)	98.4%
DECIPHER genes (1,946)	99.0%
RefSeq genes (20,846)	96.7%

Intended use

The CytoScan Dx Assay is a qualitative assay intended for the postnatal detection of copy number variations (CNV) in genomic DNA obtained from peripheral whole blood in patients referred for chromosomal testing based on clinical presentation. The CytoScan Dx Assay is intended for the detection of CNVs associated with developmental delay, intellectual disability, congenital anomalies, or dysmorphic features. Assay results are intended to be used in conjunction with other clinical and diagnostic findings, consistent with professional standards of practice, including confirmation by alternative methods, parental evaluation, clinical genetic evaluation, and counseling, as appropriate. The CytoScan Dx Assay is limited to healthcare professionals trained on the assay and is to be performed in a laboratory environment as a nonautomated assay. Interpretation of assay results is intended to be performed only by healthcare professionals, board certified in clinical cytogenetics or molecular genetics.

This device is not intended to be used for standalone diagnostic purposes, preimplantation or prenatal testing or screening, or population screening, or for the detection of, or screening for, acquired or somatic genetic aberrations.

Ordering information

Product	Description	Cat. No.
CytoScan Dx Assay Kit	1 kit	902420
CytoScan Dx Assay Training Kit	1 kit	902450
GeneChip System 3000Dx v.2	Includes: • GeneChip Scanner 3000Dx v.2 with preassembled GeneChip AutoLoader Dx • GeneChip Fluidics Station 450Dx v.2 • Workstation with GeneChip Data Collection Dx Software	00-0334
GeneChip Fluidics Station 450Dx v.2	Single station available to be purchased separately from the GeneChip System 3000Dx v.2	00-0335

Learn more at **thermofisher.com/microarrayivd**

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