ThermoFisher
S C I E N T I F I C

CytoScan Optima Suite
Robust and streamlined analysis of prenatal and miscarriage samples

Approximately 60–70% of first-trimester miscarriages are caused by chromosomal abnormalities, including aneuploidy, triploidy, and uniparental disomy (UPD) [1,2]. Traditional cytogenetic analysis of these samples is frequently challenging due to high rates of culture failure and maternal contamination, increasing the turnaround time for the results [2]. The Applied Biosystems™ CytoScan™ Optima Suite has been designed with input from cytogeneticists worldwide and empirically optimized from the Applied Biosystems™ CytoScan™ Cytogenetics Suite. The Applied Biosystems™ CytoScan™ Optima Array has whole-genome coverage and increased probe coverage targeting 396 regions relevant to prenatal and perinatal research applications.

The CytoScan Optima Suite includes arrays, reagents, and easy-to-use Applied Biosystems™ Chromosome Analysis Suite (ChAS) software, for cost-effective and streamlined analysis of your prenatal and miscarriage products of conception (POC) samples. The CytoScan Optima Suite provides whole-genome coverage and higher resolution in key genetic regions to enable the detection of multiple types of chromosomal abnormalities (Figure 1).
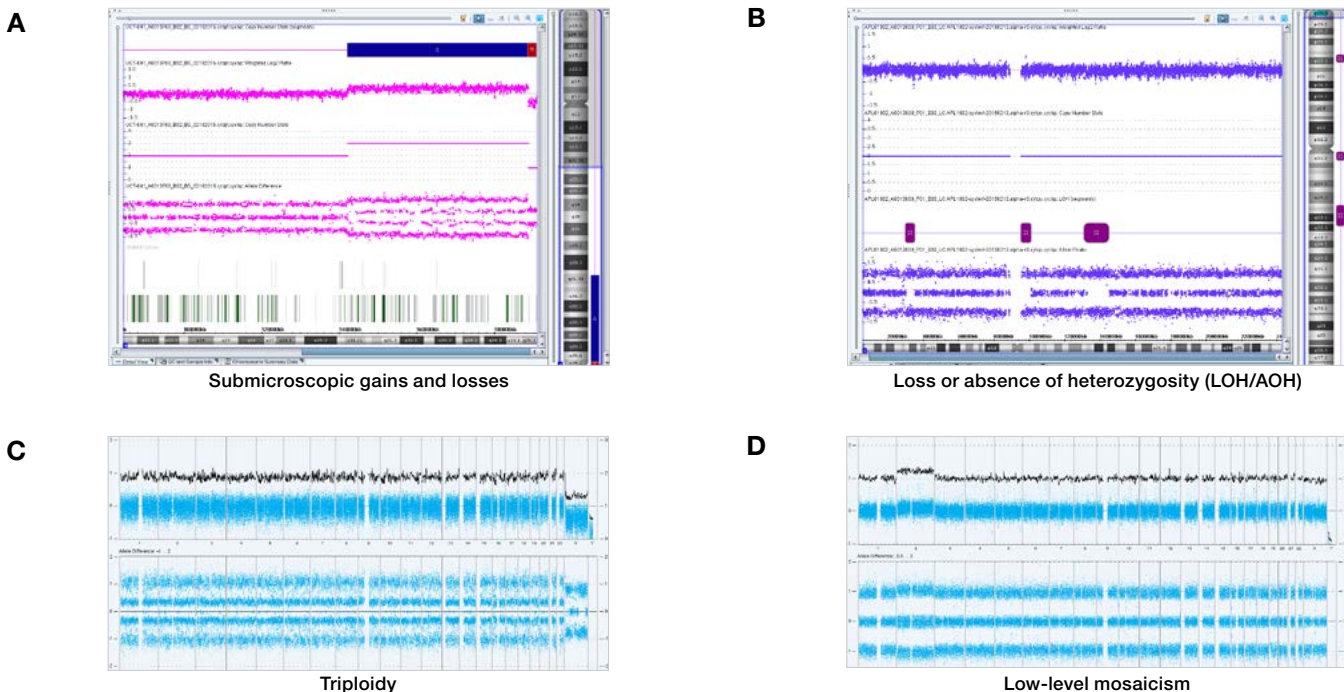
A

Submicroscopic gains and losses

B

Loss or absence of heterozygosity (LOH/AOH)

C

Triploidy

D

Low-level mosaicism

Figure 1. Detection of chromosomal abnormalities. (A) This POC sample shows a 46 Mb duplication in blue followed by a submicroscopic 2.2 Mb deletion in red. **(B)** This sample shows 3 confirmed purple LOH/AOH segments of different sizes and locations. **(C)** This whole-genome view shows 4 lines in the allelic track, concordant with a triploidy in this first-trimester POC sample. **(D)** An increase in the signal from chromosome 2 shows a ~20% mosaic trisomy in this chorionic villi (CVS) sample.

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Arrays and optimized reagents for a streamlined workflow

The Applied Biosystems™ CytoScan™ Optima Kit contains the arrays and all reagents required (including *Taq* DNA polymerase) to perform the CytoScan Optima Assay. Results can be obtained in as few as 2.5 days. The protocol also has stopping points to accommodate your schedule.

An intuitive and flexible workflow for accurate analysis

Chromosome Analysis Suite (ChAS) software enables you to easily view and summarize chromosomal aberrations across the genome (Figure 2). ChAS software also includes:

- A database for storing data
- Trio analysis tools
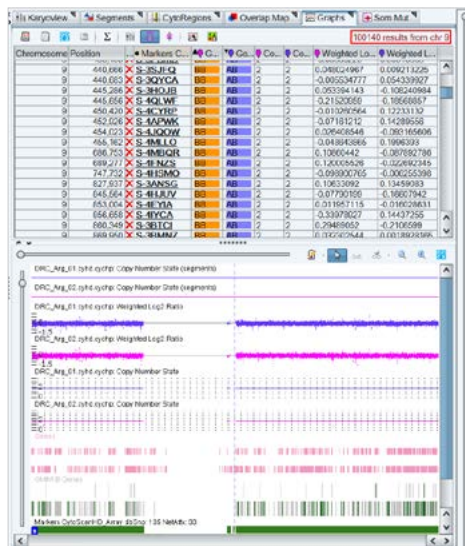- Flexible reporting tools
- Updated external annotations

Figure 2. Intuitive data analysis. ChAS software combines powerful data analysis with simple visualization and flexible reporting.

Specifications

The CytoScan Optima Array content has been empirically selected from the Applied Biosystems™ CytoScan™ HD Array and consists of a total of 315,608 features covering control, copy number (CN), and single-nucleotide polymorphism (SNP) probes. There are 18,018 CN and 148,450 SNP markers uniformly spaced over the genome, with enhanced interrogation of 396 regions of prenatal interest. Cumulatively, through the collection of SNPs and nonpolymorphic probes, the application provides the ability to support detection of CNVs, enables the elucidation of allelic imbalance, identifies abnormalities that are copy-number neutral such as AOH or LOH, and characterizes unbalanced translocation events in the samples of interest.

The CytoScan Optima Assay labels fragmented DNA with a DNA-labeling reagent (biotin transfer) and stains the labeled hybridized target with streptavidin-phycoerythrin (SAPE); phycoerythrin is the fluorophore.

The CytoScan Optima Suite offers:

- Minimum resolution of 1 Mb for losses, 2 Mb for gains, and 5 Mb for LOH/AOH
- Increased coverage density (25 markers/100 kb) in 396 empirically selected regions relevant to prenatal research
- A built-in reference file made of CVS, amniocytes, cultured cells, POC, and blood samples

CytoScan Optima Array specifications

Markers for copy number analysis	
Mosaicism	>20%
Input gDNA	250 ng*

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

References

1. Levy B et al. (2014) Genomic imbalance in products of conception: single-nucleotide polymorphism chromosomal microarray analysis. *Obstetrics and Gynecology* 124(2 Pt 1):202–209.
2. Wang BT et al. (2014) Abnormalities in spontaneous abortions detected by G-banding and chromosomal microarray analysis (CMA) at a national reference laboratory. *Molecular Cytogenetics* 7:33.

Ordering information

Product	Description	Cat. No.
CytoScan Optima Kit	Arrays and reagents sufficient for 24 reactions	902533
CytoScan Optima Training Kit	Arrays and reagents for 24 reactions, plus training materials	902534

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."

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

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