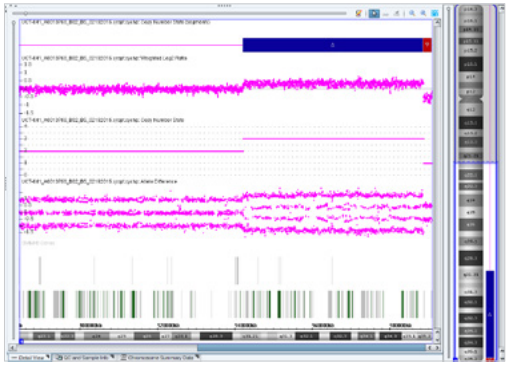
# CytoScan Optima Suite

## 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 for prenatal and perinatal research applications.

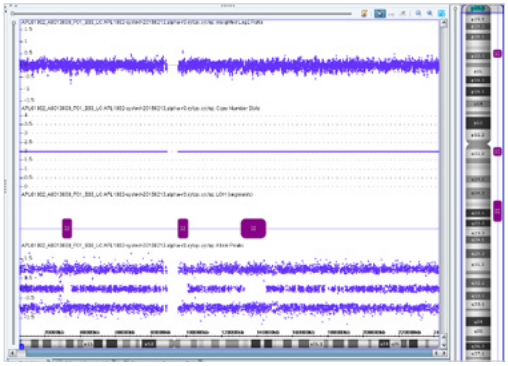
The CytoScan Optima Suite includes arrays, reagents, and easy-to-use data analysis software for cost-effective and streamlined analysis of your prenatal and miscarriage products of conception (POC) samples. The CytoScan Optima Suite provides whole-genome coverage and higher resolution in key genetic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

A



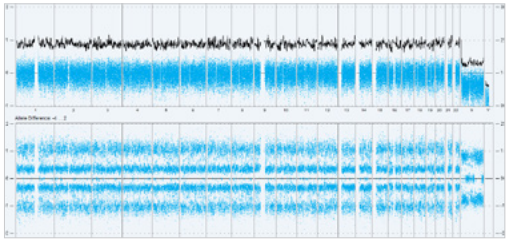
Whole-chromosome aneuploidies and submicroscopic gains and losses

B



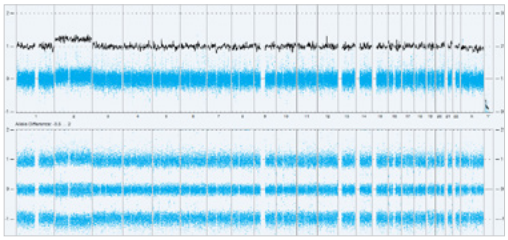
Loss or absence of heterozygosity (LOH/AOH)

C



Triploidy and maternal cell contamination

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.

ThermoFisher SCIENTIFIC

applied biosystems

## Arrays and optimized reagents for a streamlined workflow

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

## An intuitive and flexible workflow for accurate analysis

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

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

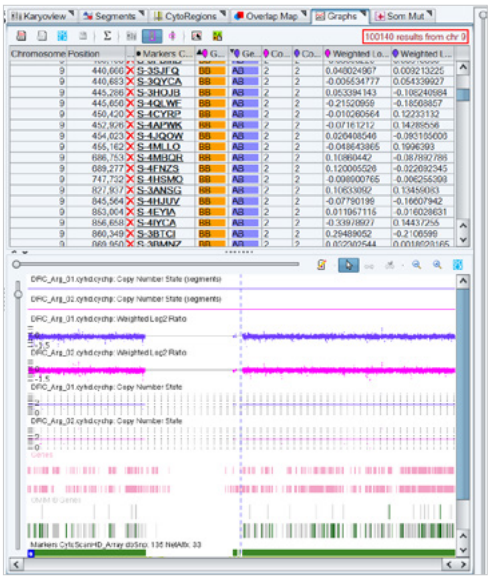


**Figure 2. Intuitive data analysis.** ChAS combines powerful data analysis with simple visualization and flexible reporting.

## 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 Suite offers

- A 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 for prenatal research
- A built-in reference file made of CVS, amniocytes, cultured cells, POC, and blood samples

### 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 |

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

ThermoFisher SCIENTIFIC