# Nouvelles lames CGH array pour analyse pré- et post-natal

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# NGS-CGH

## Invest in the future without compromising quality

**NGS** SureSelect


**CGH** CGH Microarrays – processing reagents
SureScan Scanner

### Sample QC

TapeStation
Fragment Analyzer


### Software

**Alissa Software for CGH and NGS data interpretation**

### Automation

Magnis
Bravo

# GenetiSure Cyto Microarrays

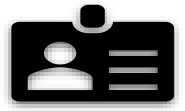
The perfect fit for cytogenetic and genetic labs using chromosomal microarrays to analyze constitutional samples

New design focused on clinically relevant and more recent databases:
ClinGen, ClinVar, OMIM, Development Disorder Genotype-Phenotype Database

Curated backbone with coverage on telomeres and PAR regions to improve coverage of clinically relevant regions and improve data interpretation

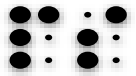
Design include only high quality probes, limiting GC biass, improving data quality and enabling more precise CN call, even for mosaic samples

Including spike-in probes for sample tracking



Free space on each array for easy customization to integrate laboratory specific gene/region of interest

Exon level coverage in genes usually tested by MLPA to improve test value and reduce need of secondary tests



Increased resolution for LOH down to 2.5Mb in autosomal chromosomes matching more updated guidelines from consortia

All

4x180K CGH

4x180K CGH+SNP

# Focused Microarrays for Pre- and Postnatal Clinical Research

## GenetiSure Cyto CGH Microarrays

| | Array type | AMADID | Array format | Total genes targeted * | Median probe spacing | | | | Free space | LOH resolution | Exon coverage |
|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | Target regions | Overall | Backbone | Telomere & PAR | | | |
| **GenetiSure Cyto 8x60K CGH** | CGH | 085590 | 8x60K | 3644 | 7.1 Kb | 50.6 Kb | 67.4 Kb | 31.8 Kb<br>13.5 Kb | ~500 | Na | No |
| **GenetiSure Cyto 4x180K CGH** | CGH | 085589 | 4x180K | 3619 | 3.5 Kb | 16.5 Kb | 19.8 Kb | 7.8 Kb<br>7.5 Kb | ~1500 | Na | Yes on 103 selected genes |
| **GenetiSure Cyto 4x180K CGH+SNP** | CGH+SNP | 085591 | 4x180K | 3644 | 7.3 Kb | 44.5 Kb | 57.1 Kb | 25.0 Kb<br>10.5 Kb | ~1500 | 2.5Mb across Autosomes | No |
| **GenetiSure Postnatal 2x400K CGH+SNP** | CGH+SNP | 078737 | 2x400K | 8106 | na | 9.5 Kb | na | na | na | 2.5Mb across Autosomes | 89% of genes 3 probes/exon |

* Genes are considered covered if they are targeted by 5 or more probes.
note: 4x180K CGH is targeting ''fewer'' than 8x60/4x180 SNP due to the large gene cutoff which is more aggressive due to the denser backbone.

# What's coming for SureSelect Human All Exon V8?

## Efficient design and best-in-class performance

### Same Comprehensive Content

Targets protein coding regions in major gene definition databases: CCDS, RefSeq, GENCODE

### Powered by Machine Learning

Machine learning-based probe selection & placement

### Made Using Improved Manufacturing Process

Probe printing targeting both forward/reverse strands

New probe recovery process

### Design Footprint Reduction

Significantly more efficient design that is more economical to sequence

**Current progress:**
**Design footprint <1.2**

### Uniformity Improvement

Deeper and more uniform coverage across all target regions

**Current progress:**
**Best-in-class 30x coverage and fold-80**

### Single-Exon CNV Calling

High resolution analysis of copy number variation (CNV) to increase diagnostic yield

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