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Illumina Cytogenetics Solutions

Accuracy when it matters most

M-GL-03524 v2.0

Partner of choice for **all** your cytogenetics needs.

Since its inception, Illumina has been committed to developing the most powerful tools in support of biomedical researchers worldwide.

Our portfolio includes an integrated cytogenetics solution offering comprehensive genomic coverage, high-throughput automation, and intuitive analysis software.

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Why is cytogenetic analysis important?

Cytogenetic analysis is the examination of chromosomes to determine chromosome abnormalities such as aneuploidy and structural abnormalities.

Irrespective of the specific application, both clinical researchers and patients benefit from technology offering speed, accuracy, and scalability.

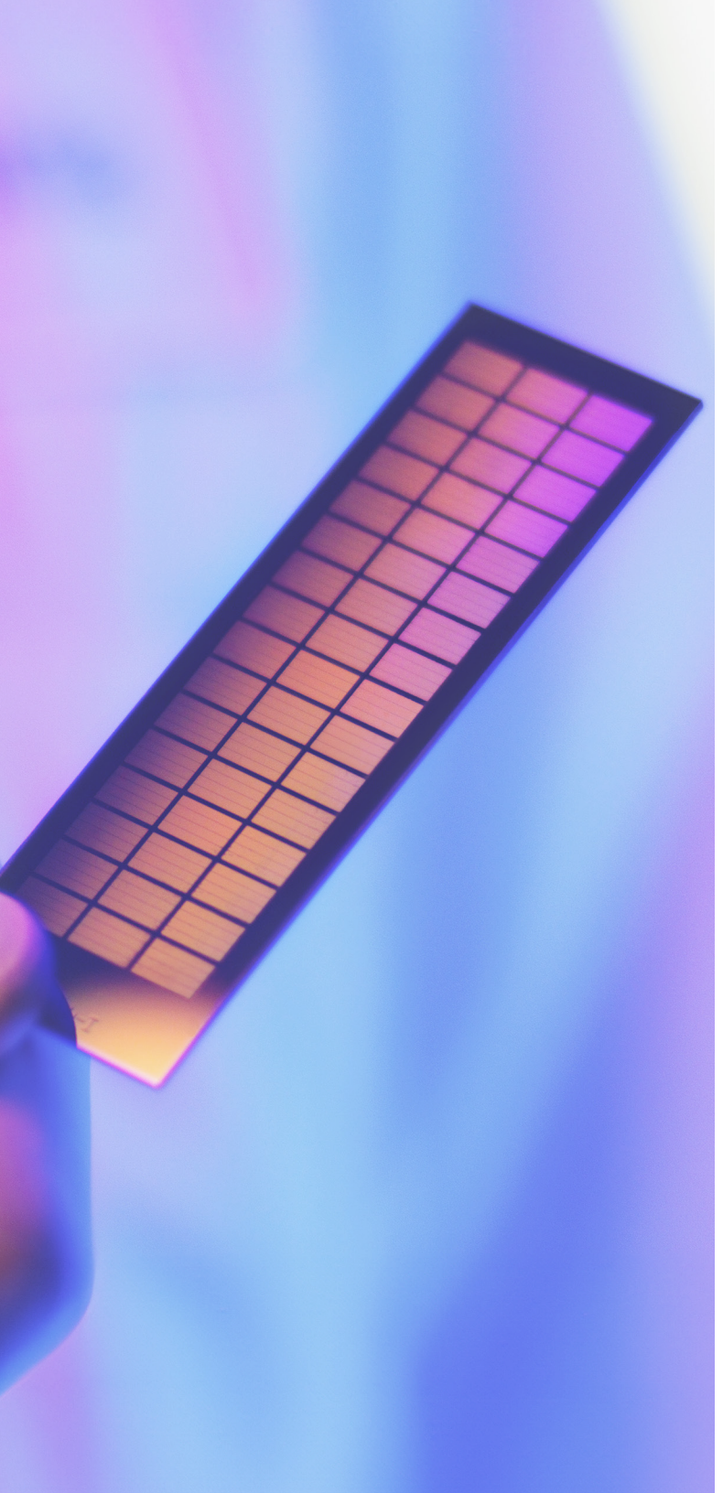
PRENATAL TESTING

CANCER DIAGNOSIS AND PROGNOSIS

FERTILITY ISSUES

PERSONALIZED TREATMENT

DIAGNOSIS OF GENETIC DISORDERS



Genomic technology offers detection of...

Structural variants

- Insertions & deletions
- Copy Number Variations (CNVs)
- Balanced translocations or inversions
- Unbalanced translocations

Mosaicism

Loss of Heterozygosity (LOH)

Uniparental Disomy (UPD)

Small variants

- Single Nucleotide Variations (SNVs)
- Small insertions & deletions

Table 1: Genomic Technologies for Cytogenetics

Parameter	FISH	Karyotyping	Arrays	NGS (large gene panel)	NGS (whole genome)
Whole-Genome View	No	Yes	Yes	No	Yes
Resolution	> 50 kb	> 5 Mb	< 1 kb	1 base pair	1 base pair
Aneuploidy	Yes	Yes	Yes	Yes	Yes
Unbalanced Translocation	Yes, if known	Yes, if large	Yes	Yes[a]	Yes
Balanced Translocation or Inversion	Yes, if known	Yes	No	No	Yes
Mosaicism	Yes	Yes	Yes, if 20% of cells present	Yes	Yes
Polyploidy	Yes (indirect)	Yes	Yes (SNP arrays only)	Yes	Yes
UPD	No	No	Yes (SNP arrays only)	Yes[b]	Yes
Copy-Neutral LOH	No	No	Yes (SNP arrays only)	No	Yes
SNVs	No	No	No	Yes	Yes
Gene Fusions	Yes	No	Yes, if unbalanced No, if balanced	Yes	Yes

Using arrays and NGS to complement traditional methods, cytogeneticists can obtain a comprehensive view of genetic abnormalities, both large and small.[2]

a. Yes, if branch points are targeted

b. Yes, if both parents are analyzed

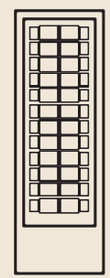
A workflow tailored to your needs.

Versatility like never before.

Custom content design

Infinium arrays offer optional addition of custom markers, ensuring your most critical questions are answered

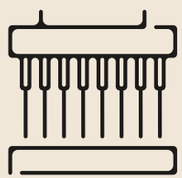
Powerful array



Supports all existing Illumina cytogenetics arrays:

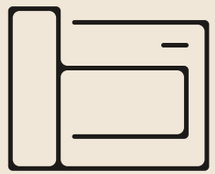
- Global Screening Array with Cytogenetics-24
- Global Diversity Array with Cytogenetics-8

Lab processing



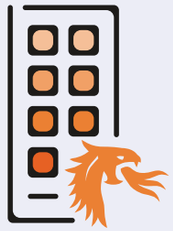
Up to **24 samples on a single BeadChip** supports improved turnaround time and reduced hands-on time compared with other solutions on the market

Scanning



Data streams from iScan™System to Illumina cloud platform

Secondary analysis



DRAGEN™ Array genome-wide CNV calling algorithm detects duplications, deletions and loss of heterozygosity based on array data

Tertiary analysis Reporting

Emedgene™ provides AI-supported variant prioritization, visualization, interpretation and research reporting for cytogenetics

Superior coverage. Superior throughput. It's your choice.

	Infinium Global Diversity Array with Cytogenetics-8	Infinium Global Screening Array with Cytogenetics-24
Total markers	~ 1.8 M	~ 700 k
Samples per BeadChip	8	24
Targeted cyto coverage	>4800 key genes	>4800 key genes
Probe spacing (kb) - Mean/Median	~3.9kb / 2.3kb	~1.1kb / 0.63kb
Resolution (kb) - Targeted/Backbone	~10kb / 25kb	~5kb / <20kb
Workflow chemistry	LCG	HTS
Throughput	1728 samples/week	5760 samples/week
Hands-on time	65 minutes for automated workflow	65 minutes for automated workflow
Input DNA	200 ng	200 ng
Sample types	Blood, FFPE tissue, Buccal swabs, Saliva	Blood, FFPE tissue, Buccal swabs, Saliva

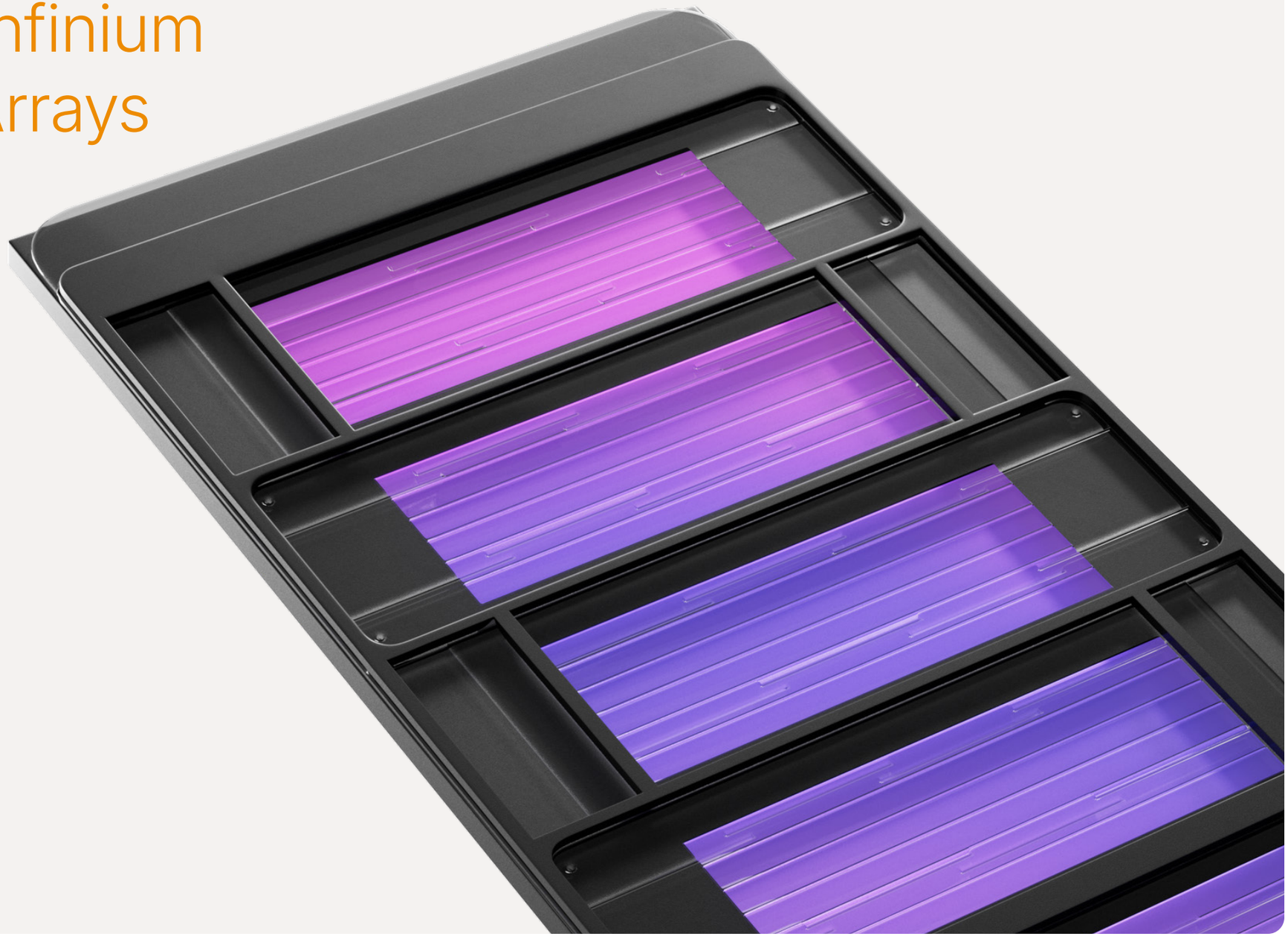
What our customers are saying about our arrays:

"...the cost was not quite an order of magnitude less, but it was a lot less per sample than alternative options. **We've essentially been able to justify hiring two additional techs to work in the lab because of the projected cost savings** we're going to have using this array, compared to our old platform."

- Benjamin Darbro, MD, PhD; University of Iowa

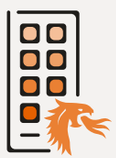
Hallmarks of Infinium Cytogenetic Arrays

- Exon-focused design strategy with new booster content
- Built on proven Illumina Infinium technology
- Genome-wide coverage across high-value regions of known disease association



Accuracy and efficiency you can count on.

DRAGEN Array secondary analysis for cytogenetics



Genome-wide CNV calling algorithm to detect duplications, deletions and absence of heterozygosity (AOH) regions

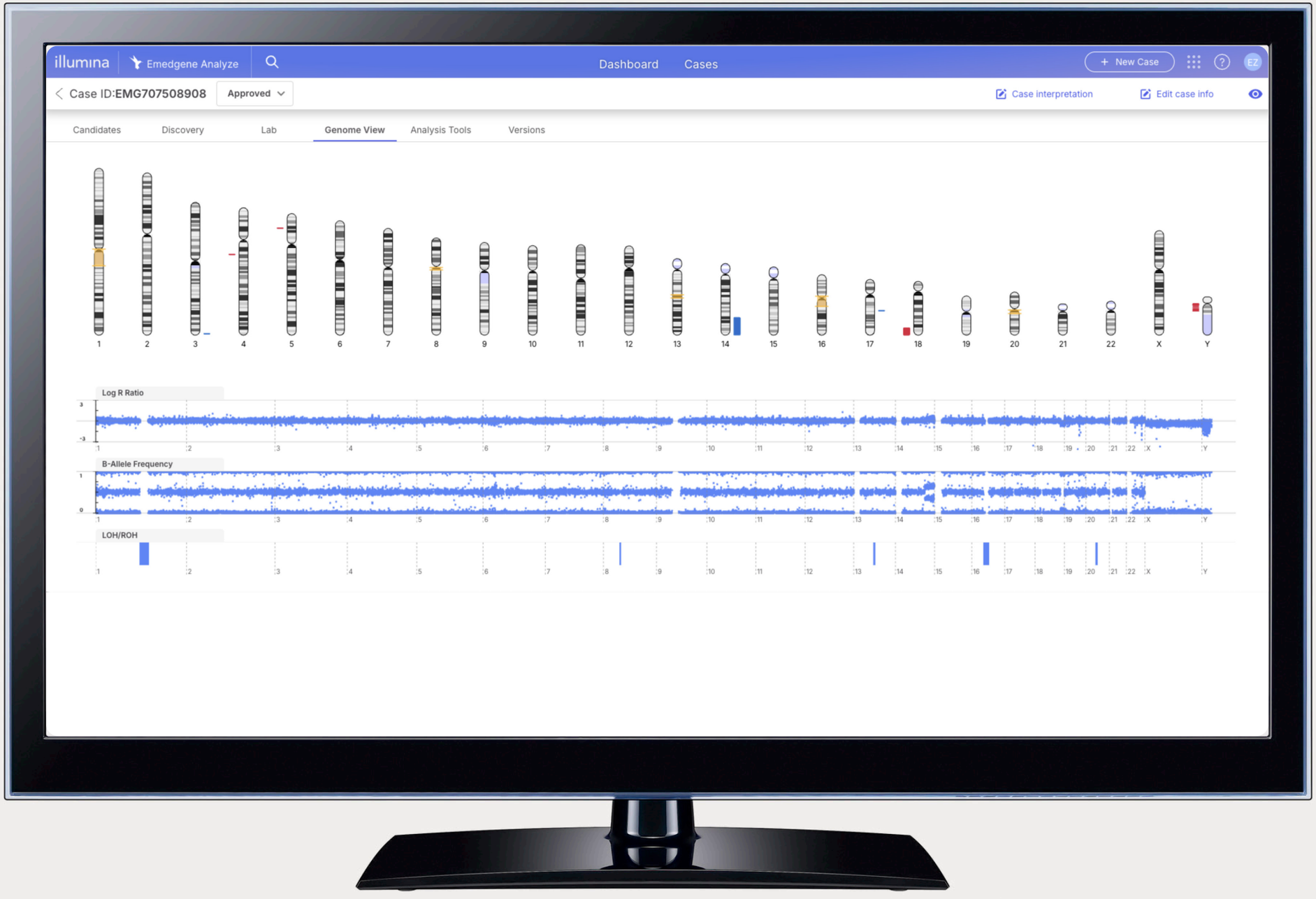
- Single analysis software supporting all Illumina cytogenetics arrays
- CNV output format in VCF for standardized reporting
- Bedgraph files for Log R Ratio and B-allele Frequency to use in further visualization

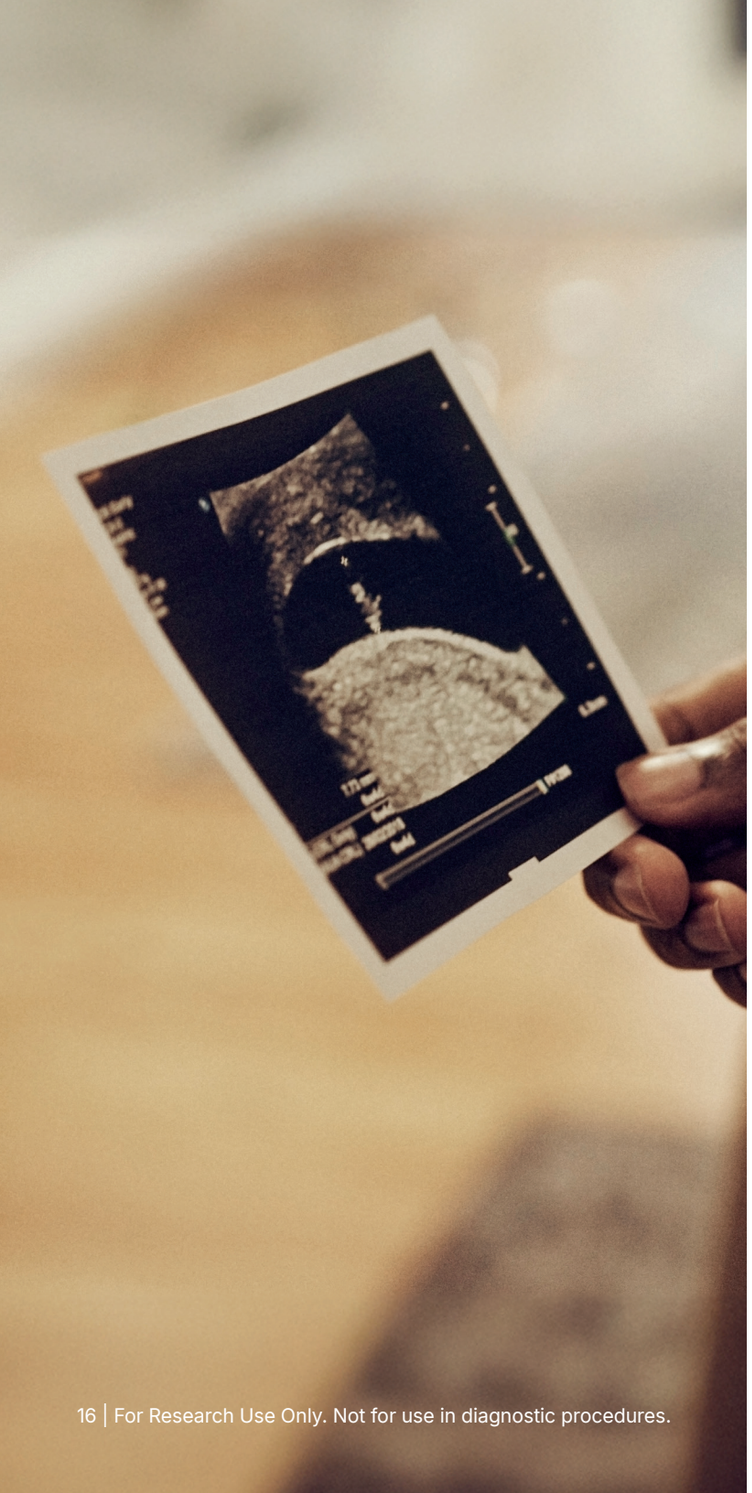
Deployed via **DRAGEN Array Local and Cloud**

- Local deployment for research applications
- Cloud deployment with simple BaseSpace™ Sequence Hub (BSSH) user interface for the cytogenetic CNV/LOH calling pipeline

Streamline cytogenetics analysis to solve the genomic analysis bottleneck

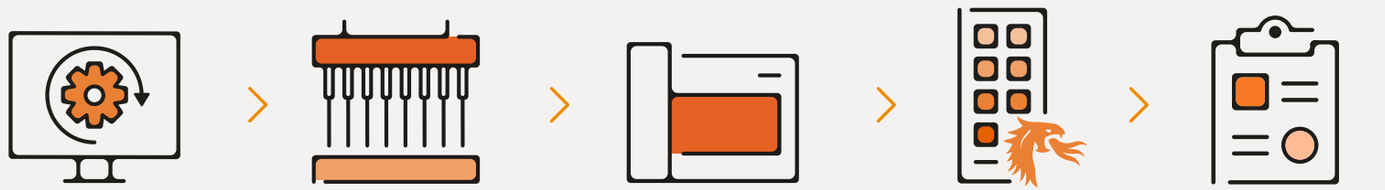
- Single software to review array or sequencing data
- Genome-wide, chromosome-level, and CNV visualizations for easy data review
- Leverage a wealth of annotation and historical data tracks to determine call significance
- Explainable AI (XAI) shortlists CNVs with evidence to streamline the data review process
- Automated ACMG classification and curation-to-reporting flows
- Cloud-based deployment for easy setup and on-boarding


illumina
Emedgene Analyze
Dashboard
Cases
+ New Case
EZ
Case ID:EMG707508908
Approved
Case interpretation
Edit case info
Candidates
Discovery
Lab
Genome View
Analysis Tools
Versions
1 2 3 4 5 6 7 8 9 10 11 12 13 14 15 16 17 18 19 20 21 22 X Y
Log R Ratio
B-Allele Frequency
LOH/ROH



The whole is greater than the sum of its parts.

Streamlined, complete solution for efficient cytogenetic analysis



Optional custom content design

Lab processing

Scanning

DRAGEN Array secondary analysis

Emedgene tertiary analysis reporting

Product		Catalog no
Arrays		
	Infinium Global Screening Array with Cytogenetics-24 v1.0 (24 samples)	20122862
	Infinium Global Screening Array with Cytogenetics-24 v1.0 (48 samples)	20066469
	Infinium Global Screening Array with Cytogenetics-24 v1.0 (288 samples)	20066470
	Infinium Global Screening Array with Cytogenetics-24 v1.0 (1152 samples)	20066471
	Infinium Global Diversity Array with Cytogenetics-8 v1.0 (8 samples)	20122861
	Infinium Global Diversity Array with Cytogenetics-8 v1.0 (16 samples)	20066507
	Infinium Global Diversity Array with Cytogenetics-8 v1.0 (48 samples)	20066508
	Infinium Global Diversity Array with Cytogenetics-8 v1.0 (96 samples)	20066509
	Infinium Global Diversity Array with Cytogenetics-8 v1.0 (384 samples)	20066510
Software		
	Emedgene - Annual Support and Maintenance – Array	20136545
	Emedgene – Array Sample	20136549
	Illumina Connected Analytics (ICA) Basic annual Subscription	20044874
	Illumina Analytics – 1 iCredit	20042038
Hardware & Automation		
	iScan System	11291093
	Infinium Automated Pipetting System with ILASS	20051293
	Infinium Amplification System	20064466

Learn more:

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