

Evolutions des solutions et technologies Illumina au service de la génomique de demain

Nicolas Roserot

Clinical sales specialist RGH/GDT

26 Novembre 2025



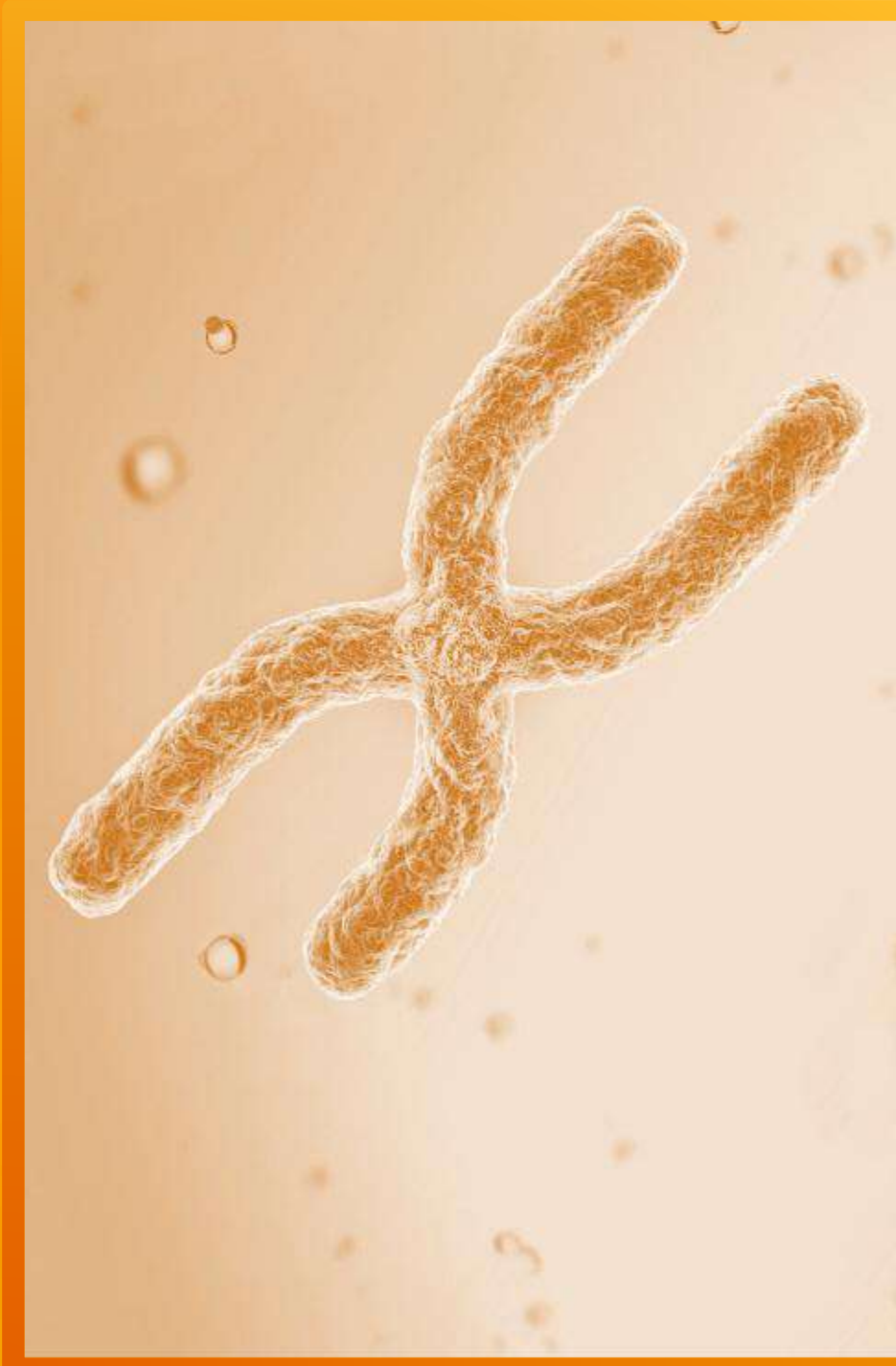
For Research Use Only. Not for use in diagnostic procedures.

| © 2025 Illumina, Inc. All rights reserved.



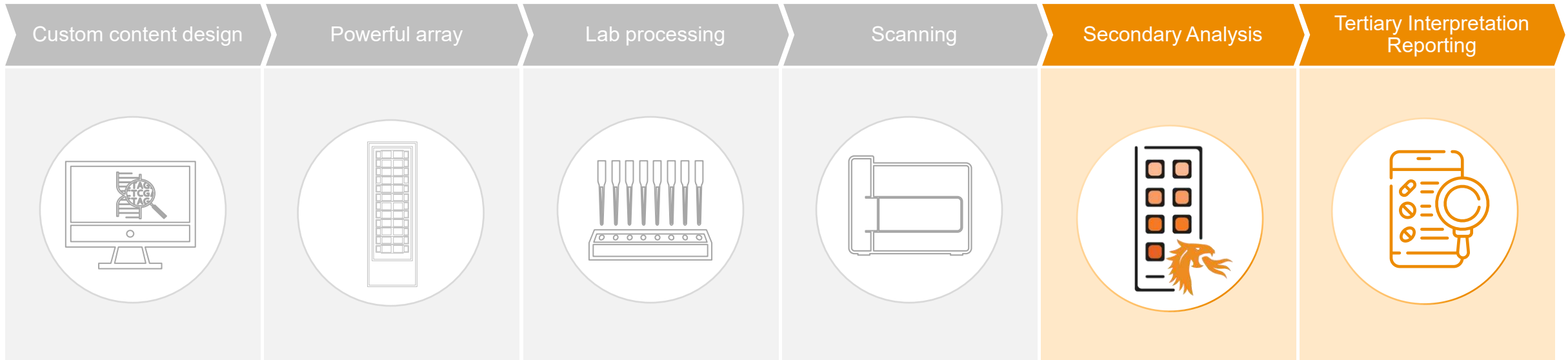
Infinium™ Arrays

Cytogenetics solution



Streamlined cytogenetic research workflow tailored to your needs

Versatility like never before



Supports all existing Illumina cytogenetics arrays:

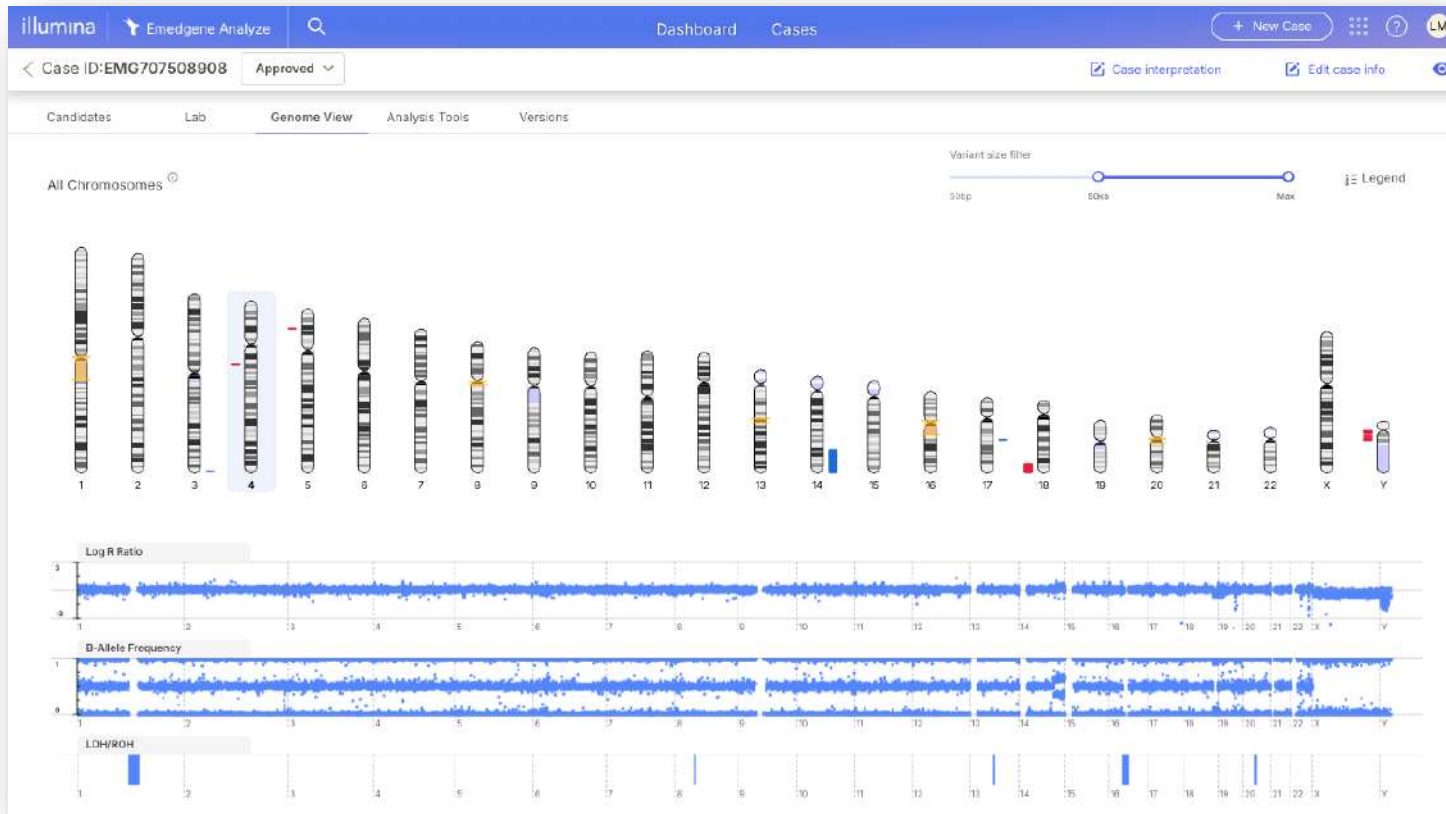
- **Global Screening Array** with Cytogenetics-24
- **Global Diversity Array** with Cytogenetics-8
- **CytoSNP-850K v1.4**

Data streams from iScan™ System to Illumina cloud platform

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

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

Chromosome-level and CNV visualization for easy data review



Visualize potential abnormality within the genome



Zoom in to any region of the genome for more detailed analysis



Override automated calls

Clear visualization for rapid data confirmation

Understand the significance of detected changes through curated annotations



Standard annotation databases for helpful data interpretation

- OMIM genes
- ClinGen dosage sensitivity
- gnomAD
- DGV
- Decipher



Customize annotations to leverage organizational knowledge

- Historic data from any VCF or FASTQ source
- User uploaded external databases

Quickly assess pathogenicity within standard cytogenetics visualization

INTRODUCING

BiInsight

Illumina's newest business, delivering deeper biological insights from large-scale multiomic data.

Unites Illumina's specialties—world-class sequencing, enterprise-ready software, and AI innovation.

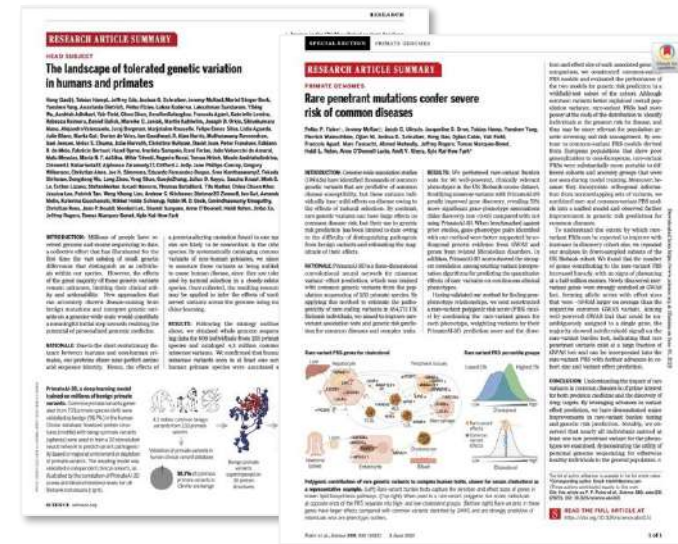
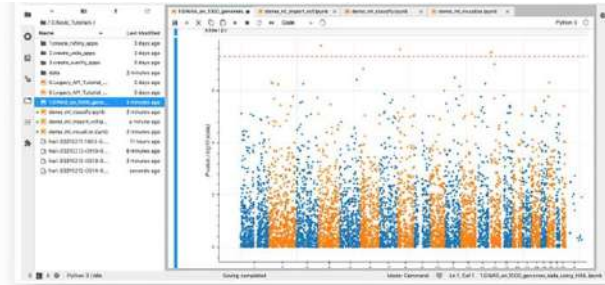
Created to accelerate discovery—delivering faster variant interpretation, pathway and similar-case insights, and prioritized drug targets to advance research, speed development, and improve future patient care.

Collaborative approach—working together to build richer data assets and develop AI capabilities that enhance analyses and continuously improve Illumina assays and bioinformatic products.



Rami Mehio
SVP, General Manager,
BiInsight, Illumina

Illumina's solutions for precision medicine, research, and discovery



1 Sequencing platform 2 Software ecosystem 3 AI & Perturb-seq

- Instrument
- Library prep
- Reagents

- DRAGEN secondary analysis
- Illumina Connected Analytics
- Emedgene
- Illumina Connected Insights
- Illumina Connected Multiomics

Leading AI algorithms and datasets for delivering new insights into human genetics at scale

Comprehensive genome analysis with DRAGEN



DRAGEN Germline



Multigenome mapping with Pangenome reference



Germline

Mito

Mosaic



SMA/HBA +15 other

PGx

HLA



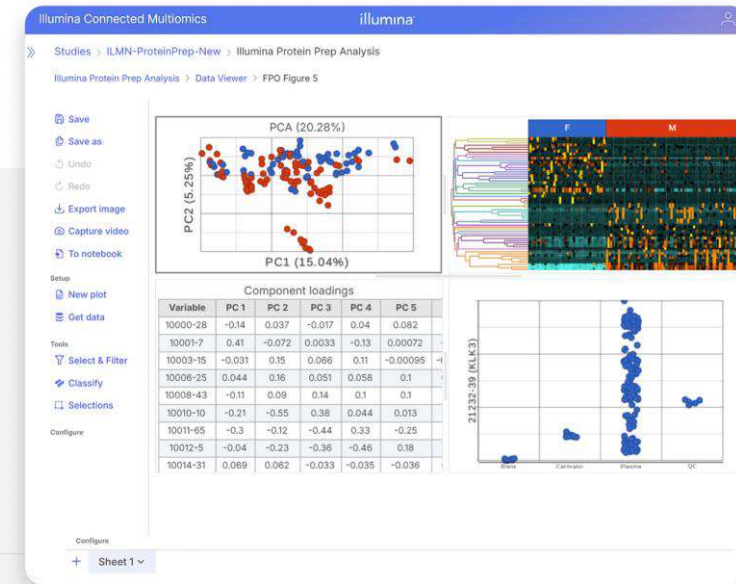
Comprehensive genome analysis and variant detection at scale using DRAGEN | Nature Biotechnology
<https://www.nature.com/articles/s41587-024-02382-1>

Beyond genomes: Multiomics

Add context to mechanism and disease biology



- Proteomics
- Transcriptomics
- Epigenomics
- Genomics
- Bulk, Single-cell, and Spatial



Illumina Constellation mapped read technology



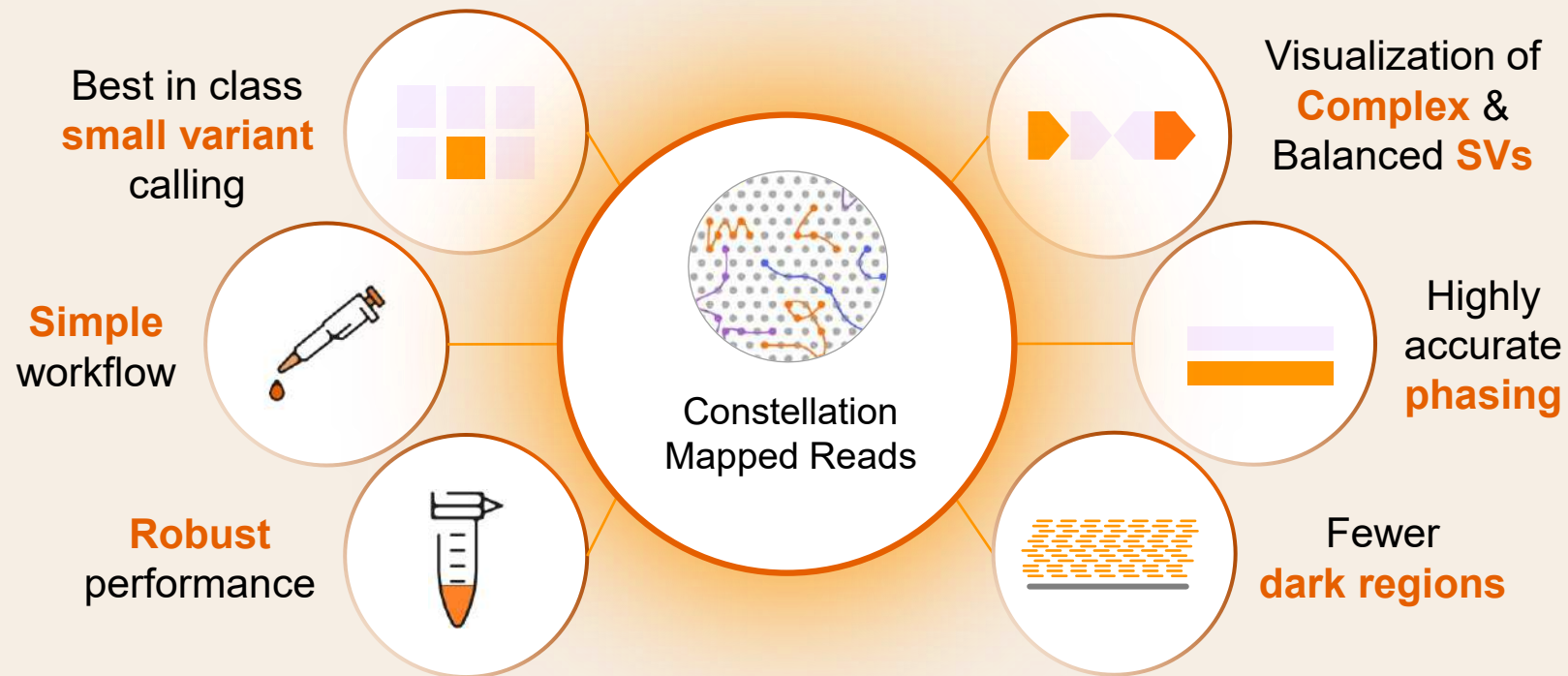
Illumina Constellation mapped read technology

Breakthrough technology advances WGS to reveal complex biology

Unprecedented
workflow simplification

AND

Our **most comprehensive**
genome yet



ASHG customer feedbacks

Webinar is online

Redefining the limits of what's possible:
Sequencing reimaged

On-demand webinar: ASHG 2025



Date & Time

15 Oct 2025

Presenter

Steve Barnard
Joe Devaney
Bekim Sadikovic

Topic

Software & informatics, Instruments

Watch Now



Constellation lab workflow

Run Set Up



- 350 ng DNA input
- 3 new reagents
- Basic lab tools
- ~10 minutes



Sequencing



- Standard NovaSeq™ X Plus
- Custom recipe
- 2x150 sequencing run
- ~24 hours



Analysis



- DRAGEN™ cloud analysis
- Standard outputs (FastQ, VCF)
- New output files

Genes with Mappability Issues

Spinal Muscular Atrophy

- *SMN1* and *SMN2* share 99% homology

Lynch Syndrome

- *PMS2* and *PMS2CL* share up to 98% homology

Chronic Granulomatous Disease

- *NCF1* and two pseudogenes (*NCF1B* and *NCF1C*) share 99% homology

Recessive Nonsyndromic Hearing Loss

- *STRC* and *STRCP1* share 99.6% homology

Repeat Expansions

Sample	Variant Details	Gene	Long Read	Constellation Repeat Counts
VAL_002	<i>DMPK</i> (11, >150)	<i>DMPK</i>	11, 254	11, 259
VAL_025	<i>FMR1</i> (30, 98, 102)	<i>FMR1</i>	30, 104	30, 61
VAL_046	<i>C9orf72</i> (6, >145)	<i>C9orf72</i>	6, 310	6, 250
VAL_047	<i>PHOX2B</i> (11, 31)	<i>PHOX2B</i>	20, 31	20, 31
VAL_074	<i>ATXN3</i> (19, 78)	<i>ATXN3</i>	16, 78	16, 76
VAL_108	<i>CNBP</i> (<26, 75>)	<i>CNBP</i>	15, 3466	15, 1300
VAL_114	<i>PABPN1</i> (10, 13)	<i>PABPN1</i>	10, 12	10, 12
VAL_153	<i>ATXN1</i> (30, 32)	<i>ATXN1</i>	31, 33	31, 33
VAL_276	<i>FMR</i> (39, >200)	<i>FMR1</i>	N/A	40, 660



For Research Use Only. Not for use in diagnostic procedures.

Summary of Constellation Results

99.5 % variant detection (4/366; Misses: *PKD1*, mtDNA deletion); Sample type agnostic

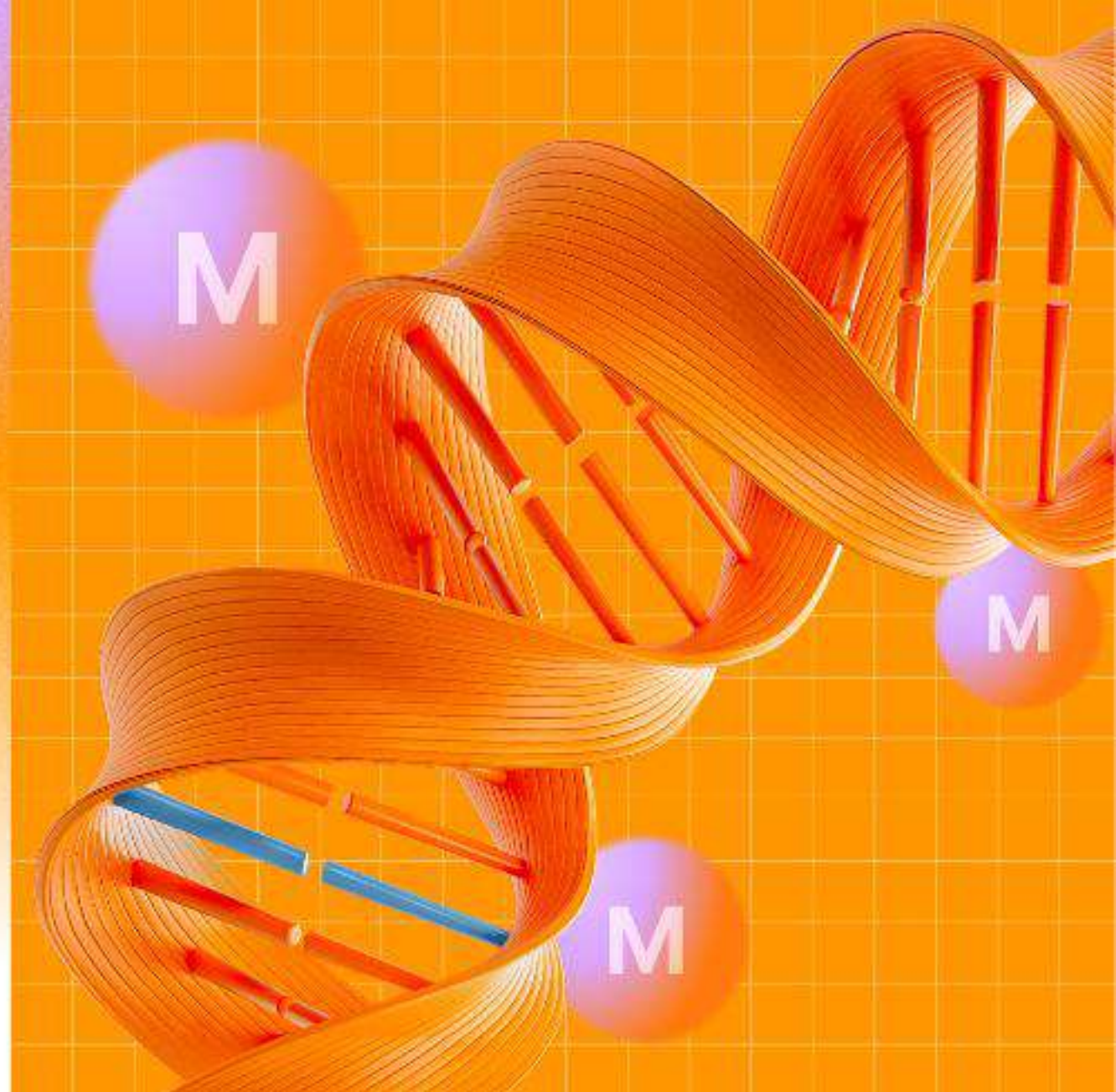
Reproducible and accurate results with numerous sample types including blood, buccal swabs, and prenatal samples

Called Mosaic Trisomy missed with long read method; Ability to call mtDNA heteroplasmy

Combines numerous lab methodologies into one assay; Reducing TAT

GeneDx

illumina 5-Base Solution now available





Illumina 5-base solution for methylation and variant detection

One easy assay. Dual insights.

Simultaneous methylation profiling and variant detection for whole genome and target enrichment

Library Prep

Prep in less than a day
Single-step enzymatic conversion

Sequencing

Greater mapping efficiency
Maximize sequencing data yield

Analysis

Expanded multiomic insights
Unparalleled genetic and epigenetic
variant detection accuracy