PHASE GENOMICS

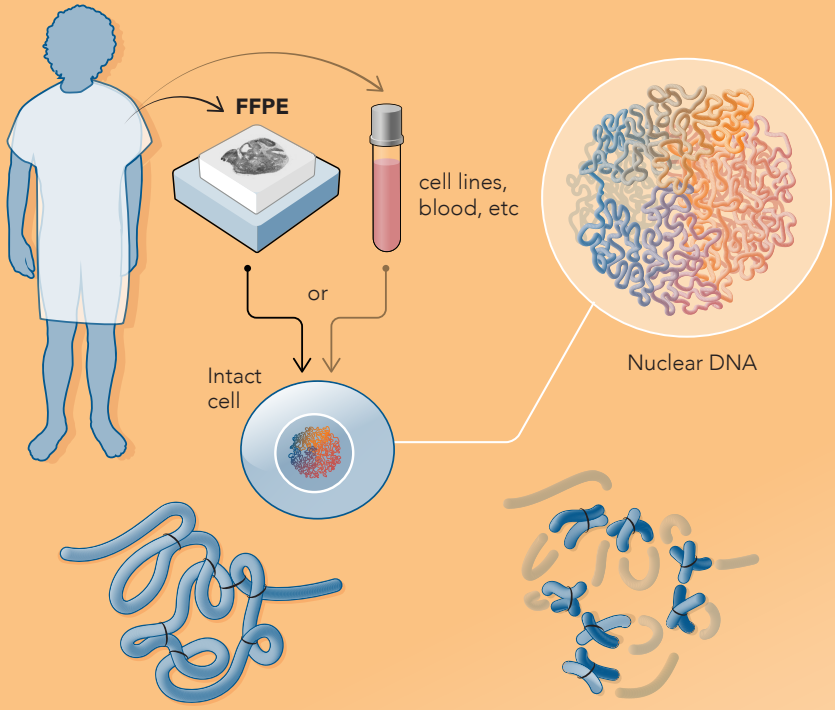
CytoTerra™ Platform

Reveal the Topography of the Genetic Map

The CytoTerra™ Platform combines the genome-wide structural variation detection power of traditional cytogenetics with the molecular-level precision of chromosomal microarrays and FISH in a single, cost-effective assay.

Key features and benefits:

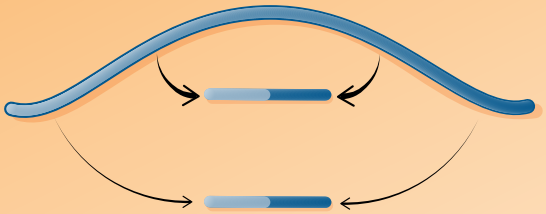
- Genome-wide detection of chromosomal abnormalities in a single, NGS-based assay
- Complements conventional cytogenetics methods, such as CMA and FISH
- Actionable results in standard nomenclature
- Scalable, fast, and cost-effective. No dedicated instrumentation required.



How it works

Chromatin is packaged into 3D structures that retain a relationship between genomic and physical distance (i.e. sequences that are closer on the same chromosome are also closer in physical space).

This method exploits the relationship between linkage and proximity to enable the reconstruction of chromosome structure.

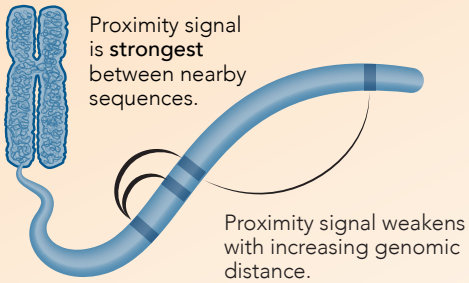


1 DNA is crosslinked *in vivo* to fix DNA sequences inside the nucleus. Crosslinking traps sequence interactions across the entire genome and between different chromosomes.

2 Crosslinked DNA junctions are fragmented, ligated, and converted into a sequencing library.

3 Junctions between physically linked loci are sequenced and mapped back to the reference genome. Each read represents a single, physical connection between two points on the genome. Collecting millions of these data points provides a statistical measure of the distance between any two positions along the genome.

Reconstructing the Genome



Measuring the physical proximity between all loci in the genome creates a genomic "map" unique to each sample, enabling the detection of a wide range of chromosomal abnormalities.

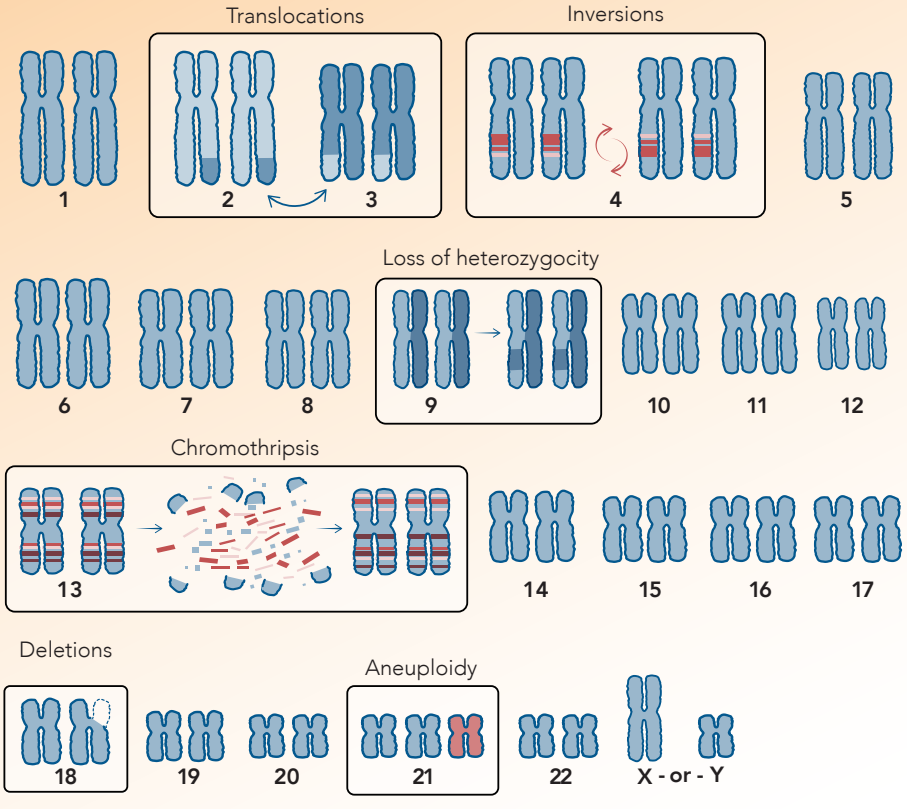


Illustration by Tolpa Studios

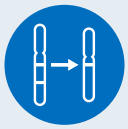
Single, Genome-wide Assay

The current approach to cytogenetics involves a cascade of methods that includes sequential testing via karyotyping, chromosomal microarray (CMA), and fluorescence *in situ* hybridization (FISH). Combined, these methods can be used to create a map of chromosome abnormalities to inform the diagnosis and treatment of genetic conditions. This approach is time-consuming, labor-intensive, expensive, and leaves undefined or ill-defined areas on the chromosomal map.

The CytoTerra™ Platform takes cytogenetics to the next level. It eliminates the gaps and challenges associated with the current approach to deliver a comprehensive picture of the genomic landscape. Powered by Phase Genomics' proprietary technology, the CytoTerra Platform leverages the unique strengths of ultra-long-range sequencing to characterize the breadth of chromosome abnormalities, at higher resolution than standard cytogenetic analysis, CMA, and FISH combined.

Feature/Abnormality	CytoTerra Platform	Cytogenetics (karyotyping)	FISH	CMA
Genome-wide detection	Yes	Yes	No	Yes
Unbalanced **chromosomal alterations (deletion/duplication/amplification)**	Yes	Yes	Yes	Yes
Balanced **rearrangements (reciprocal and Robertsonian translocation/ inversion/insertion)**	Yes	Yes	Yes	No
Complex rearrangements	Yes	Yes	No	No
No dedicated instrumentation	Yes	No	No	No

Identify SVs and CNVs, and Characterize other Complex Rearrangements in both Pre- and Postnatal Specimens



Detect cryptic rearrangements associated with recurrent pregnancy loss (RPL). Uncover these karyotypically undetectable rearrangements with a faster, cheaper alternative to CMA.

Unlock genomic information hidden within non-viable and/or FFPE samples. Ultra-long-range sequencing technology does not require actively dividing cells or high molecular weight DNA extraction and requires a very low starting cell volume.

From a single sample, resolve complex and challenging fetal and adult phenotypes caused by multiple congenital anomalies—with a comprehensive, cost-effective complement for CMA.

Automated Analytics Yield Actionable Results

The CytoTerra™ Platform comprises a new molecular technology with companion analytics. The novel, fully automated analysis and reporting module builds on Phase Genomics' industry-leading capabilities in biological computation, and delivers:

- comprehensive and actionable results
- reports in standard ISCN and sequence-based nomenclature

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CYTOTERRA™ REPORT

PATIENT INFORMATION		SPECIMEN INFORMATION		PROVIDER INFORMATION	
Accession ID	657636	Specimen ID	309110	Ordering Physician	Joanna Johnson, MD
Name	Loren Smith	Type	Blood	Institution	Comprehensive Cancer Center University of Medicine
Date of Birth	07/20/69	Collection Date	03/22/2022		

RESULT SUMMARY

CHROMOSOME NUMBER	SEX CHROMOSOME COMPLIMENT	DETECTED ABNORMALITY
46	XY	del(11)(q24.2),del(11)(q24.3q25)

DETAILED RESULTS

FIGURES	CHANGE	ISCN	GENOMIC COORDINATES	GENES
Figure 1A	deletion	del(11)(q24.2)	chr11:124100000-127250000	ACRV1, CCDC15, CDON, CHEK1, DCPS, DDX25, EI24, ESAM, FAM118B, FEZ1, FOXRED1, HEPACAM, HEPN1, HYLS1, KIRREL3, MSANTD2, NRGN, OR10D3, OR8A1, OR8B12, OR8B2, OR8B3, OR8B4, OR8B8, OR8D1, OR8D2, OR8G1, OR8G5, PANX3, PATE1, PATE2, PATE3, PATE4, PKNOX2, PUS3, ROBO3, ROBO4, RPUSD4, SIAE, SLC37A2, SPA17, SRPRA, ST8GAL4, STT3A, TBRG1, TIRAP, TMEM218, VSIG10L2, VSIG2, VWA5A
Figure 2A	deletion	del(11)(q24.3q25)	chr11:128550000-133600000	ADAMTS15, ADAMTS8, APLP2, ARHGAP32, BARX2, C11orf45, ETS1, FLI1, KCNJ1, KCNJ5, NFRKB, NTM, OPCML, PSDM10, SNX19, ST14, TMEM45B, TP53AIP1, ZBTB44

ANALYSIS

Phase Genomics has played a leading role in delivering ultra-long-range sequencing technology and computational tools to the life science research community, with a proven track record of enabling deeper insights into the architecture, variation, function, and complexity of genomes, epigenomes, and metagenomes. That same know-how and expertise has been used to create the CytoTerra™ Platform. This platform is our first offering that leverages ultra-long-range sequencing to deliver a cost-effective, high-throughput assay for uncovering potential genomic etiological underpinnings of human health and disease. Through the platform's impactful and actionable insights, we are enabling translational discoveries that will ultimately lead to faster precision diagnoses, treatment, and improved health outcomes.

Learn more

The CytoTerra Platform is currently offered as a comprehensive sample-to-report service. Contact us to inquire about RUO kits compatible with existing low- and high-throughput sample processing pipelines.

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