PHASE GENOMICS

CytoTerra® Cytogenetics Platform

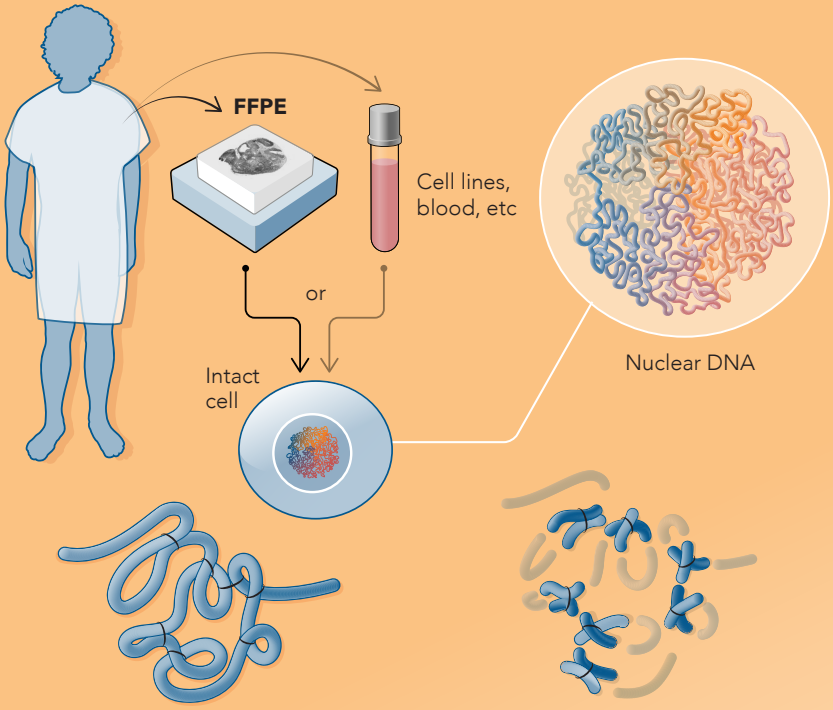
Advance Precision Health and Cancer Research

Survey the entire chromosomal landscape with a single, NGS-based assay

The CytoTerra® Cytogenetics Platform combines the genome-wide structural variation detection power of traditional cytogenetics with the molecular-level precision of chromosomal microarrays (CMA), and FISH in a single, powerful, NGS-based assay. Based on Genomic Proximity Mapping™ (GPM), it enables a comprehensive survey of the genomic landscape and new insights in oncology and genetic disease research.

Key features and benefits:

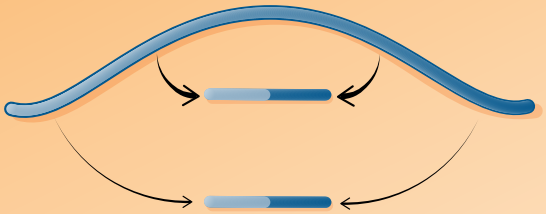
- Genome-wide detection of chromosomal abnormalities in a single, high-resolution, NGS-based assay.
- Comprehensive cytogenetic information from a wide variety of sample types, including blood and FFPE tissue.
- Scalable, fast, and cost-effective—no cell culture or dedicated instrumentation required.
- Integrated sample-to-report product; no advanced NGS experience needed.



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.

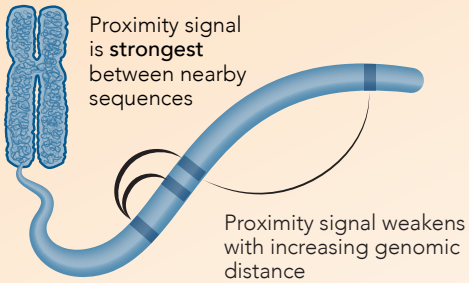


1 DNA is crosslinked *in vivo* to fix DNA sequences inside the nucleus. Crosslinking traps sequence interactions across the entire genome and between different chromosomes.

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.

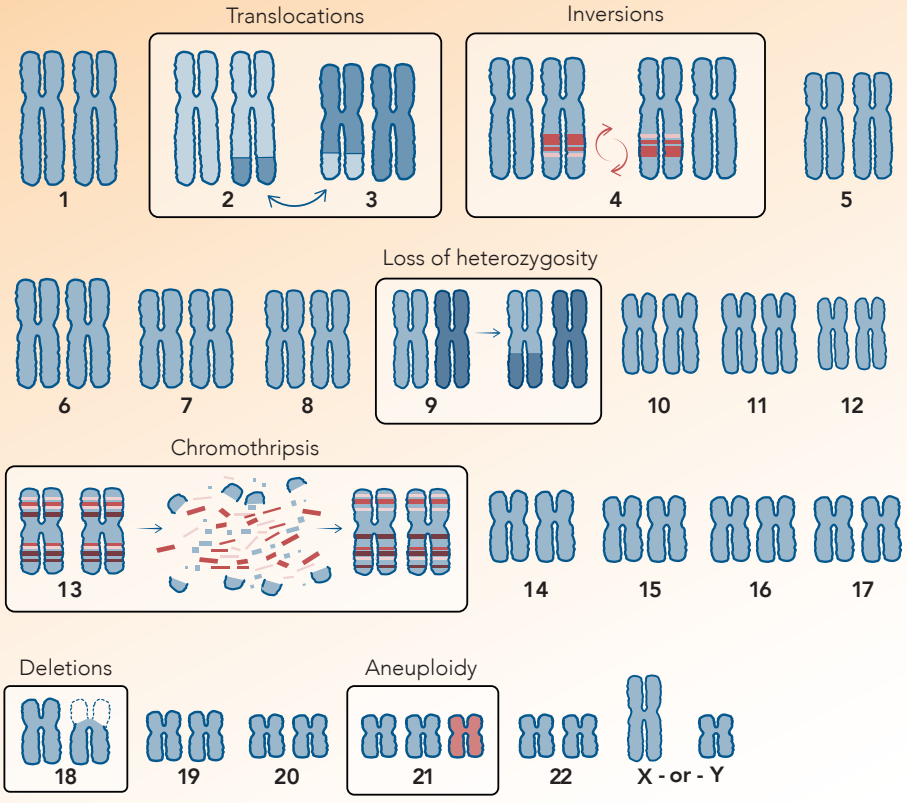


Illustration by Tolpa Studios

Less Time. Less Resources. One Assay.

The CytoTerra® Platform takes cytogenetics to the next level. It eliminates the gaps and challenges associated with the cascade of cumbersome traditional cytogenetics methods that still define the standard of care, but often fall short in identifying the aberrations that underly somatic and inherited diseases. Powered by Phase Genomics' proprietary Genomic Proximity Mapping™ (GPM) technology and AI-based computational tools, 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, FISH, and CMA combined.

Feature/Abnormality	CytoTerra® Platform	WGS	Chromosomal Microarray	FISH	Karyotyping
Genome-wide detection	Yes	Yes	Yes	No	Yes
***Unbalanced* chromosomal alterations (deletion/duplication/amplification)**	Yes	Yes	Yes	Yes	Yes
***Balanced* rearrangements (translocation/inversion/insertion)**	Yes	Yes	No	Yes	Yes
Chromothripsis (cth)	Yes	Yes	Yes	No	No
Complex rearrangements	Yes	No	No	No	No
Resolution	High	High	Medium	Medium	Low

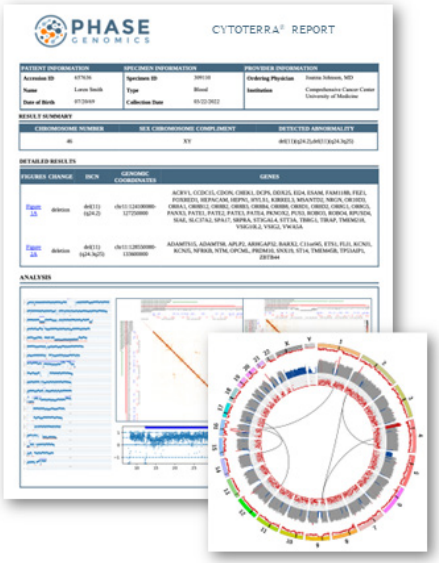
Identify SVs and CNVs, and Characterize other Complex Rearrangements for a Broad Range of Applications

- **Precision Oncology.** Structural variants play an important role in the risk stratification of cancers and inform treatment decisions, especially in the case of hematological cancers. Our technology enables rapid detection and accurate characterization of large structural variants that are difficult to assess with short-read sequencing methods.
- **Genetic disease.** The genetic basis of many diseases is complex, as disease states are often not the result of simple nucleotide changes. Our approach offers a genome-wide view of the structural variants, including large-scale chromosomal abnormalities, that potentially contribute to a disease phenotype.
- **Cell therapies.** Cell therapy products require meticulous quality control. The CytoTerra Platform offers a simple, genome-wide assay for confirming the absence of structural variants that may impact genomic stability.

Download Applications Summary

Automated Analytics Yield Actionable Results

The CytoTerra® Platform provides a sample-to-report NGS-based cytogenetics assay that starts with a blood, FFPE, or other biological sample, and ends with a comprehensive and actionable report in standard and sequence-based nomenclature. Fully automated, cloud-based analysis of paired-end, short-read sequencing data is performed with proprietary computational tools developed at Phase Genomics. The entire process can be completed in less than a week, and is scalable to large numbers of samples.



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® Cytogenetics 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

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