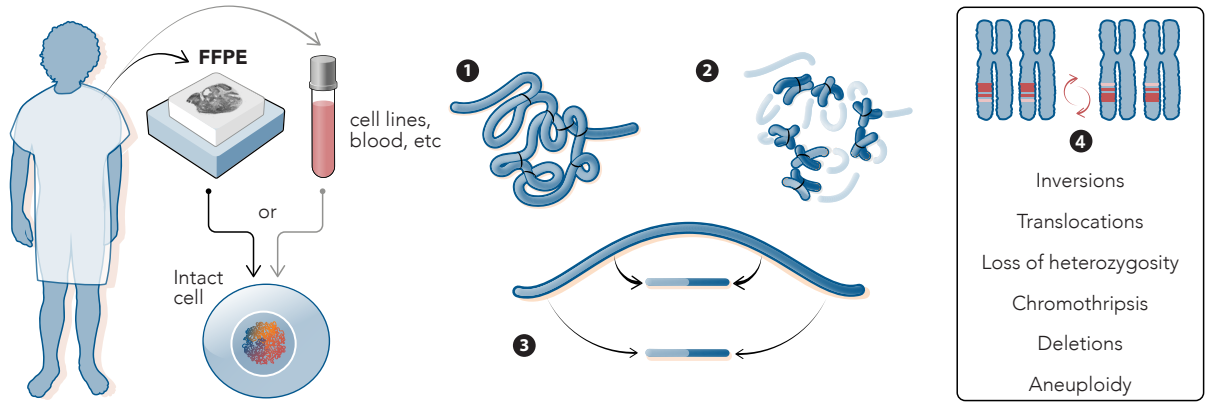
PHASE GENOMICS

CytoTerra® Platform

Access New Insights in Precision Oncology

Improving acute myeloid leukemia (AML) assessment with CytoTerra



How it works: The CytoTerra® Cytogenetics Platform utilizes genomic proximity mapping (GPM)™ technology to determine the structure of chromosomes from fresh-frozen diagnostic or archival FFPE samples. Chromatin within the intact nucleus is crosslinked (1) prior to fragmentation (2). Proximity ligation generates chimeric molecules that are converted to a paired-end DNA sequencing library (3). Using the interaction frequency data from the library, the full range of cytogenetic-scale chromosome rearrangements can be identified (4).

Case study: Identification of novel AML variants undetected by traditional cytogenetics

The CytoTerra Platform was used to analyze a set of 48 samples (including peripheral leukemic blood and bone marrow biopsies) collected at the time of AML diagnosis. Variants previously observed with standard-of-care cytogenetics were confirmed and additional variants were identified. This impacted the risk stratification of 25% of the patients based on the European Leukemia Network (ELN) 2022 guidelines.[1]

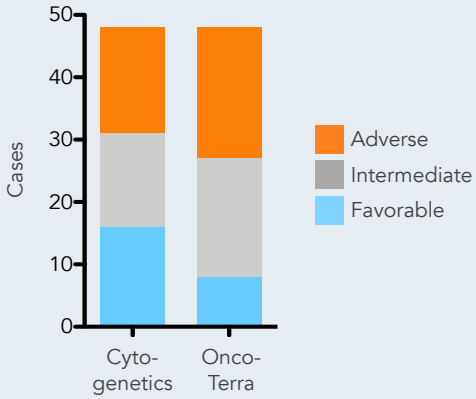
Read the preprint here

Table 1. Summary of variants detected in pilot cohort*

Variant	Translocations	Inversions	CNVs	Total/Overall
True positive (TP)	4	5	11	20
False negative (FN)	1	0	2	3
True Negative (TN)	37	36	17	90
Additional variants	1	3	24	28
Specificity	**0.80**	**1.00**	**0.85**	**0.87**

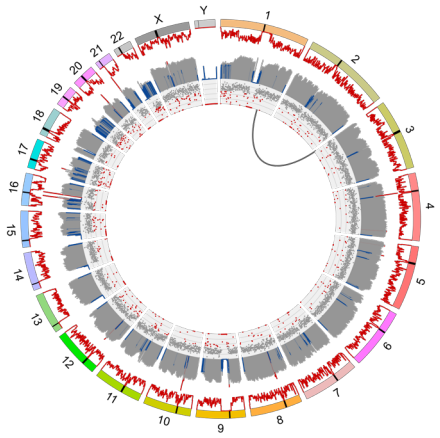
*Reporting all variants at >20% abundance that involve >10 kb of sequence, *n*=48.

Risk stratification

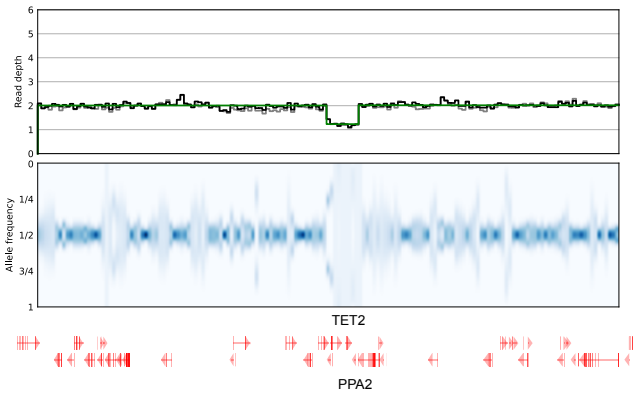


1. https://aml-hub.com/medical-information/2022-eln-recommendations-for-the-diagnosis-of-aml-in-adults

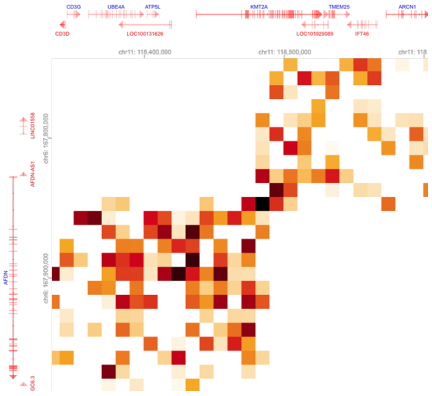
High-resolution view of inversions, translocations, and copy number variants from a single, NGS-based assay



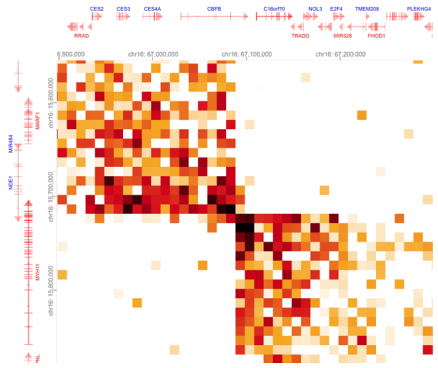
Circos plot provides a genome-wide view



Coverage and minor allele frequency support deletion calls



Detection of t(6;11) creating a KMT2A::AFDN fusion



Identification of inv(16) creating a MYH11::CBFB fusion

Standard-of-care cytogenetics requires a battery of tests which may include karyotyping, FISH, and chromosomal microarray (CMA) to capture clinically relevant variants and guide treatment. With the CytoTerra® Platform, these results are captured in a single assay, providing a high-resolution view of translocation and inversion breakpoints not attainable with other methods. CytoTerra is compatible with FFPE samples, enabling cytogenomic analysis of archival samples for retrospective studies. From one sample to a plate of 96, the CytoTerra Platform meets modern cytogenetic needs without specialized instrumentation.

Learn more

PHASE GENOMICS

info@phasegenomics.com
www.phasegenomics.com

Phone: 1-833-742-7436
Twitter: @PhaseGenomics