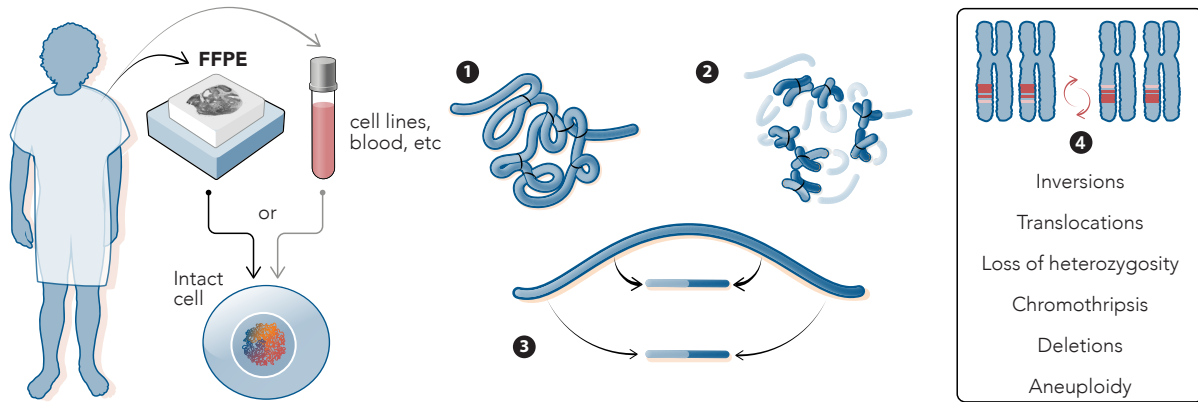


Improving acute myeloid leukemia (AML) assessment with OncoTerra



How it works: The OncoTerra Platform utilizes proximity ligation 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). Sequences that are closer together along the linear length of a chromosome are more likely to interact and form proximity ligation junctions. These junctions generate 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).

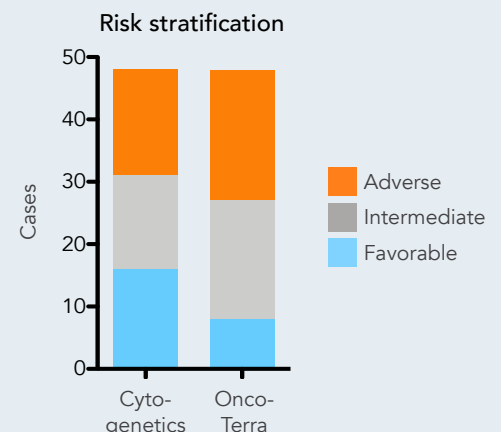
Case study: Identification of novel AML variants undetected by traditional cytogenetics

The OncoTerra 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

Table 1. Summary of variants detected in pilot cohort*

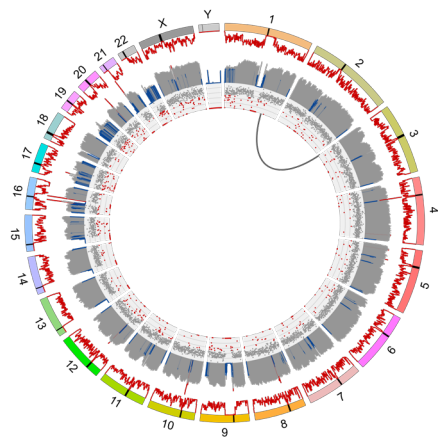
Variant	Translocations	Inversions	CNVs	Total/Overall
True positive (TP)	4	5	11	20
False negative (FN)	1	0	2	3
True Negative (TN)	37	36	17	90
Additional variants	1	3	24	28
Specificity	0.80	1.00	0.85	0.87

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

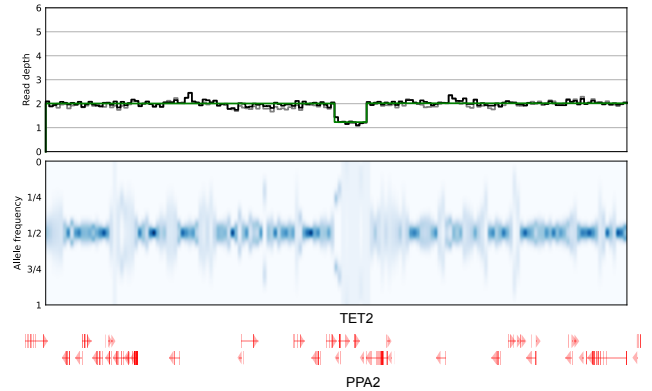


1. Döhner, H. et al. Diagnosis and management of AML in adults: 2022 recommendations from an international expert panel on behalf of the ELN. *Blood* 140, 1345–1377 (2022).

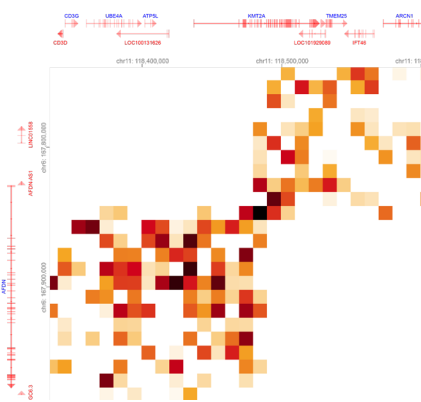
High-resolution view of inversions, translocations, and copy number variants from a single NGS-based assay



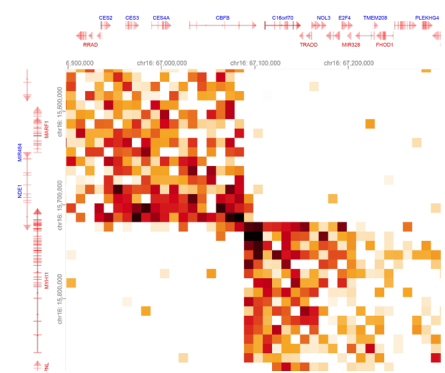
Circos plot provides a genome-wide view



Coverage and minor allele frequency support deletion calls



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



Identification of inv(16) creating a CBFβ::MYH11 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. OncoTerra™ captures these results in a single assay, and provides a high-resolution view of translocation and inversion breakpoints not attainable with other methods. OncoTerra is compatible with FFPE samples, enabling cytogenomic analysis of archival samples for retrospective studies. From one sample to a plate of 96, OncoTerra meets modern cytogenetic needs without specialized instrumentation.

Learn more

The OncoTerra 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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