

ACCELERATING BIOMARKER DISCOVERY, DISEASE DIAGNOSIS AND NOVEL THERAPIES THROUGH SPATIAL GENOMICS

CHARACTERIZE MECHANISMS BEHIND DISEASE

Many modalities of detection, like RNA profiling, assist in diagnosis, but characterization of sequence alone often cannot capture the mechanisms behind the disease. This can result in poor classification and ineffective therapies for patients.



Arima Genomics enables characterization of sequence, structure and regulation by delivering 3-dimensional folding information via spatial reads.

CAPTURE THE SPATIAL GENOME TO IMPROVE DISEASE CLASSIFICATION & PATIENT STRATIFICATION

Molecular techniques like gene expression profiling are used to help diagnose and classify diseases and their subtypes. However, poor classification can result in mischaracterization of disease.

For example, gene expression profiling of lineage ambiguous leukemia resulted in poorly classified clusters of patients. However, capturing the spatial genome illuminated rearrangements and the corresponding enhancer-hijacking events that led to the disease (1).



CAPTURE THE SPATIAL GENOME TO IDENTIFY & VALIDATE NOVEL THERAPEUTIC TARGETS

In a recent Nature Genetics publication, spatial characterization of a mis-folding event allowed researchers to correctly diagnose a TAD-fusion event resulting in a MYC promoter and a distal super-enhancer. Furthermore, these disease cells were effectively targeted with a small-molecule inhibitor, leading to disease correction (2,3).

References

1. Lindsey E Montefiori, et al. Enhancer hijacking drives oncogenic BCL11B expression in lineage ambiguous stem cell leukemia. Cancer Discov June 8 2021 DOI: <u>10.1158/2159-8290.CD-21-0145.</u>

2. Konstantin O., Camgoz A., Park D. E., et al. Oncogenic 3D genome conformations identify novel therapeutic targets in ependymoma, 05 November 2020, PREPRINT (Version 1).

3. Kloetgen, A., Thandapani, P., Ntziachristos, P. et al. Three-dimensional chromatin landscapes in T cell acute lymphoblastic leukemia. Nat Genet 52, 388–400 (2020). <u>https://doi.org/10.1038/s41588-020-0602-9</u>



Linear genome: shows presence of disease, but offers poor characterization.



Spatial genome: shows presence AND mechanism of disease



Correction of disease through intervention

AN END-TO-END PLATFORM FOR BIOMARKER DISCOVERY, VALIDATION AND SCREENING

Biotech and pharmaceutical companies can leverage the Arima Genomics technology platform to discover, validate and apply spatial genomic signatures to to (1) identify novel diagnostic or prognostic markers, (2) identify novel therapeutic targets, or (3) assess therapeutic response to treatment.







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