

See What You Have Been Missing Discover and Detect Gene Fusions with 3D Genomics

With 3D genomics you gain unparalleled access to the sequence, structure, and regulatory landscape of cancer genomes to reveal novel biomarkers and therapeutic targets.



"Using Arima Hi-C technology and the new FFPE sample preparation method and bioinformatics tools, we've been widely successful in detecting structural variants in a variety of tumor samples. We are hopeful that additional insights gained with this approach will lead to improved understanding of disease mechanisms and, ultimately, the development of new therapeutic options for people with cancer."

- Matija Snuderl, MD, Director of Molecular Pathology and Diagnostics, NYU Langone Medical Center

Identify Novel Gene Fusions

Case Study

A pediatric patient with Stage 2 glioma was initially treated with a subtotal resection of the tumor, and six months post-surgery experienced rapid progression. Comprehensive DNA and RNA sequencing of the primary tumor was inconclusive, with no driver mutations identified.

A subsequent analysis by Arima revealed a novel PD-L1 translocation, and immunohistochemical staining showed strong and diffuse PD-L1 expression. The patient was given pembrolizumab and her disease status is stable¹.



Find Gene Fusions Missed by DNA, RNA, or FISH

Case Study

In a pediatric patient presenting with IDH1/2 wild-type diffuse glioma, RNA sequencing did not reveal a prognostically favorable *MYBL1* or *MYB* gene fusion event, making optimal post-surgical management unclear. All other aspects of the patient's clinical presentation were consistent with a favorable prognosis.

An FFPE sample of the tumor was sent to Arima Genomics, where analysis identified a *MYBL1-MAML2* gene fusion, confirming the favorable clinical presentation and sparing the patient adjuvant chemotherapy¹.

Assay	Result	Treatment
DNA Next Generation Sequencing	Negative for IDH1/2 mutations	Unclear if adjuvant therapy required
RNA Fusion SEQer	Negative for gene fusions	
Arima Technology	Positive for MYBL1- MAML2 gene fusion	No adjuvant therapy needed



Study Fusions in Degraded and Archived FFPE Specimens

Detect gene fusions in rare cancers or precious samples and get results even when standard molecular profiling fails^{1,2,*}.



Leverage Arima's Robust DNA Workflow for Gene Fusion Detection

Our DNA workflow enables you to go from sample to answer quickly with validated protocols and customer support^{2,3,*}.



References

- 1. Snuderl, M. (2022) <u>Webinar: Unraveling the Complex Nature of the Genetic Origins of Brain Tumors with 3D Genomics</u>.
- 2. Application Note: Structural Variant Detection with Hi-C Technology. Arima Genomics.
- 3. Product Flyer: Arima-HiC+ FFPE Kit. Arima Genomics.

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