



Empire Genomics Helps Demonstrate Value of New Technology for Use in FFPE Samples

Background

Clinical management and therapy of many solid tumor malignancies is dependent on detection of medically actionable or diagnostically relevant genetic variation. The fragmented and damaged state of DNA in formalin-fixed paraffin-embedded (FFPE) samples poses a challenge for genetic assays from tumors. There is currently no technology for the generation of long-range DNA sequence data from these highly fragmented DNA and RNA.

Objectives

Because of the lack of technology for generating long-range DNA sequence data from fragmented DNA and RNA, the present study sought to develop a high-throughput chromosome capture approach for FFPE samples, as long-range sequence data is necessary for detection of genomic structural variation and long-range genotype phasing.

Approach

The mechanism the present study developed for high-throughput chromosome capture was named “Fix-C,” as it is similar to Hi-C but with fixed and damaged samples. Fix-C was applied to 15 clinical adenocarcinoma and sarcoma specimens. Results of Fix-C were compared to the results of fluorescence in situ hybridization (FISH) using corresponding break-apart FISH probes from Empire Genomics.

Results

Fix-C was shown to achieve a 90% concordance rate with FISH assays. Furthermore, Fix-C was able to identify novel structural variation undetected by other methods, and to recover long-range chromatin configuration information from FFPE samples, as was previously impossible with this highly degraded DNA. Fix-C appears to be a powerful new approach enabling detailed genome resolution. This new approach may inform targeted molecular diagnostic assays for patient care, and technology from Empire Genomics was integral in helping the researchers reach these results and conclusions.

Structural Variation Detection by Proximity Ligation from FFPE Tumor Tissue

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Lead Organization

University of California Santa Cruz

Diseases

- Many solid tumor malignancies

Biomarkers Mentioned

- MYB
- ROS1