Fred Hutch Cancer Center

Long Range Sequencing for the Detection of Fusion Genes

Business Opportunity

Exclusive license
Sponsored research

Technology Type

Assay/method
Bioinformatics
Diagnostic

State of Development

Assay validated using human samples

Patent Information

PCT Application Filed

Investigator

Cecilia Yeung, MD
Clinical Research Division

Tech ID

22-120

Contact

partnering@fredhutch.org

Brief Description

A CRISPR-Cas9 enrichment Nanopore sequencing assay for long-read sequencing

Technology Overview

Fusion genes occur in 30% of AML patients, resulting in distinct types of AML. Identification of these structural variants help in diagnostic classification and drive targeted therapies. Recognition of fusion variants is also important for detecting clonal heterogeneity and possibly clonal evolution. Available methods for clinical diagnosis have either low sensitivity, require long turnaround time, or the need to know the genes involved in the fusion. Dr. Cecilia Yeung's team has developed a method that enables faster and comprehensive genomic profiling of AML mutations and fusion genes. Specifically, this method uses CRISPR guides to select and enrich genes involved in leukemia-specific fusion gene breakpoints, perform sequencing using nanopores, and a custom bioinformatic tools for fusion detection. This amplification-free, long-read DNA sequencing assay is able detect BCR-ABL1, PML-RARA, CBFB-MYH11, and KMT2A-AF4 fusion genes in patient specimens.

Applications

- Genomic profiling of AML mutations and fusion genes

Advantages

- Does not require previous knowledge of the target
- Open to discovery of novel/different fusion partners
- Rapid turnaround time (within 24 hours) when multiplexing different assays

Market Overview

In 2022, an estimated 20,050 people of all ages in the United States will be diagnosed with AML. The 5-year survival rate for people 20 and older with AML is 27%. For people younger than 20, the survival rate is 69%.