

Cell free DNA detection of breast cancer using single molecule methylation sequencing

Malignant tumor cells shed their DNA into the bloodstream of breast cancer patients in the form of cell-free DNA (cfDNA) and can be assayed to identify cancer-associated biomarkers. This minimally invasive approach, also known as a liquid biopsy, allows for early detection of breast cancer in asymptomatic individuals with ready accessibility. cfDNA can be monitored for early detection of breast cancer and provide accurate information of evolving tumors and breast cancer state. Thus, developing more sensitive techniques for the monitoring of cfDNA would allow for better and faster understanding of treatment response and disease progression, enabling more dynamic care.

The Ji lab at Stanford developed a novel method for the detection of breast cancer from a blood sample by utilizing a nanopore system to directly identify methylated DNA biomarkers from cfDNA without chemical or enzymatic conversion. This test process avoids the use of PCR to generate sequencing libraries. It also uses nanogram amounts or less of cfDNA per sample. The passage of methylated DNA through the nanopore generates a unique electrical signal compared to unmodified DNA. As a result, methylation can be detected with various machine learning algorithms at high accuracy. Current DNA methods for detecting breast cancer are not being used clinically because of poor performance. The invention has an advantage over existing technologies as the single molecule sequencing can detect modifications from the native DNA molecules. This analysis method maintains the single DNA molecules in their native state.

Stage of Development

Prototype

Applications

- Blood-based early detection of breast cancer
- Breast cancer monitoring
- Cancer diagnostics
- Cell free DNA detection tests

Advantages

- Cost effective compared to imaging tests such as mammograms and MRI
- Does not require chemical or biochemical conversion of methylated DNA
- Specific and efficient method in detecting breast cancer

Patents

- Published Application: [WO2024031097](#)

Innovators

- Hanlee Ji
- Billy Lau
- Lincoln Nadauld

Licensing Contact

Inyoung Lee

Licensing Manager, Life Sciences

[Email](#)