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Simple, quantitative PCR-based detection and characterization of cancer gene fusions

Stanford Medicine's Ji Research Group has developed a simple, quantitative method for detecting and characterizing gene fusions that uses DNA rather than RNA as analyte. In acute myeloid leukemia (AML), certain chromosomal translocations correlate with a good prognosis, while some deletions or duplications are linked to an aggressive cancer with poor prognosis. The ability to quickly determine specific cancer mutations provides a clinical advantage and opportunity for precise delivery of targeted therapies. However, detecting cancer gene fusion is very difficult with current methods.

The Ji Research Group overcomes the associated issues by physically isolating target gene fusion from non-targets and from the wild-type, non-rearranged genes. Their method targets a specific locus prone to rearrangement via an initial PCR based prescreen, and multiplexes for different targets specific to a given cancer, which greatly increases the efficiency of the process. The target sites are excised with CRISPR edits, and the high molecular weight DNA targets are isolated. A simple quantitative PCR assay provides an initial, high sensitivity detection that is then followed by long read nanopore sequencing for more extensive sequence characterization of the gene mutation. This diagnostic method identifies and characterizes specific genomic aberrations in cancer types that can be used to more accurately provide diagnoses, prognoses, and targeted therapies.

Stage of Development – Proof of Concept
Proof of concept studies performed with acute myeloid leukemia (AML). Ongoing research will focus on applying the method to clinical uses and a variety of cancers.

Applications

- **Cancer diagnostics, prognostics, and targeted therapies.**

Advantages

- Stable, high-resolution data pinpointing of fused genes, breakpoints and gene variant information.
- Simple workflow, easy to implement for research and diagnostic labs.
- Broad use in cancer diagnostics

Patents

- Published Application: WO2024081596

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