



cfDNA Fragmentomic Detection Of Cancer

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The Invention

UW-Madison researchers have developed a method of identifying and characterizing DNA fragmentation patterns in circulating tumor cells using targeted circulating tumor cell DNA (ctDNA) panels. These researchers trained an algorithm to identify DNA fragmentation patterns using commercially available target ctDNA gene panels to identify the presence of cancer and determine the type of cancer. Presently, whole genome sequencing needs to be performed to obtain and use this fragmentomics information from circulating tumor cells. That process adds an additional step in the genetic analysis of the DNA in these cells adding additional cost. This method can be run simultaneously with genetic mutation and gene expression assays using commercially available targeted ctDNA gene panels streamlining and improving accuracy of cancer diagnostics. The inventors recognize that the data they have is preliminary data and will need further validation, but they wanted to disclose this because this space is very competitive. When using overall and transcription factor fragmentomic patterns in a small public targeted panel dataset, this trained model can predict normal cell vs. lung cancer vs. breast cancer with ~80% overall accuracy. They plan to continue to explore whether this method of extracting fragmentation information from targeted circulating tumor gene panels can be expanded to other cancer types. They also plan to test the method looking for more complex fragmentation patterns.

Tech Fields

- [Diagnostics & Biomarkers : General diagnostics & biomarkers](#)
- [Drug Discovery & Development : Precision medicine](#)

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