Docket #: S23-400

Digital telomere measurement by Oxford Nanopore sequencing

There are few means existing methodologies to accurately measure biomarkers of human aging. Telomeres are the nucleoprotein caps at chromosome ends that shorten with aging. However, techniques to measure telomere lengths are imprecise, of low resolution, and typically reflect an indirect measurement of telomere content.

The Artandi Lab at Stanford has developed an experimental and bioinformatic pipeline that uses long read DNA sequencing on the Oxford Nanopore platform to enable highly accurate, single molecule measurement of human telomeres from human clinical samples. By leveraging molecularly barcoded oligonucleotides, the technique allows for the sequencing enrichment of telomeric DNA following library preparation for nanopore sequencing. To complement the experimental component, the inventors also created a bioinformatic pipeline that accurately and reproducibly identifies telomeric reads while measuring telomere length from long-read sequencing data. This approach involving high-resolution telomere measurement not only allows an estimate of organismal age to be accurately measured, but also provides a powerful tool to improve the efficacy of telomere length as a diagnostic and prognostic biomarker. The innovative methodology can be leveraged as a research tool to assess the underlying function of the telomere maintenance machinery in human cells and even, assess the lifetime risk of cancer development.

Stage of Development: Pre-clinical

Applications

- Diagnostics
- Consumer genomics
- Therapeutics

- Research tool/service

Advantages

- Directly measures telomere length of individual, intact telomeres with chromosome specificity.
- Can be applied towards DNA from any source.
- Requires very minimal DNA sample compared to current techniques.

Publications

- Sanchez, S.E., Gu, Y., Wang, Y. et al. Digital telomere measurement by long-read sequencing distinguishes healthy aging from disease. Nat Commun 15, 5148 (2024).

Patents

- Published Application: WO2025117914

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