

MutREAD – Mutational Signature Detection by Restriction Enzyme-Associated DNA Sequencing

Available Technologies

A cheap, fast, reproducible and scalable method

To detect cancer mutational signatures

Studies of mutational signatures in tumour DNA increased the understanding of the defective cellular processes implicated in cancer development. This approach can be used for patient stratification and can help tailor therapies targeting specific defects and support cancer prevention programmes.

Despite the importance of signatures in cancer biology, the ability to study them in a clinical setting is limited by the requirement of working with high quality and quantity of DNA extracted from fresh or frozen samples.

As the majority of historical samples are stored in formalin-fixed blocks (FFPE), studies of signatures are limited to samples collected specifically for DNA sequencing projects. Despite the recent significant decrease in the cost, WGS procedures are still prohibitively expensive for routine application in the clinical setting.

Technology overview

For providers of DNA sequencing technologies, MutREAD is a method for DNA library preparation and sequence analysis able to identify cancer mutational signatures from extremely low amount of DNA (ng/pg) from both fresh and FFPE fixed samples. MutREAD detects mutational signatures as well as WGS (cosine similarities = 0.95-0.96 compared with WGS-derived mutational

signature profiles) with less time (library ready in few hours) and reduced costs, making mutational signature analysis more accessible to the cancer research and clinical field.

Benefits

- Allows coverage for somatic mutation calling without bias in the type of detected mutations
- A new chemistry of adapters for rapid library preparation (ready in few hours)
- Cheaper than current methods (80% cheaper than 10x sWGS and 96% lower than WES libraries)
- Works with extremely low amount of DNA (ng/pg)
- Works with fresh and FFPE fixed samples (current methods do not work on FFPE)
- Compatible with multiplexing for studying larger cohorts

Applications

Identification of:

- Mutational signatures
- Single nucleotide variants
- Copy number variations
- Tumour mutation burden

for patient stratification, personalised therapies, and early cancer detection.

Opportunity

PCT application filed (PCT/GB2021/051299)
Licensing opportunities available

Inventors

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Publications

[Nature article](#)