STRUCTURAL VARIANTS

Changes to the structure or number of chromosomes in a genome can contribute to genetic diseases and cancers.

Current sequencing techniques lack the resolution to reliably identify all structural variants.

Neither short-read sequencing nor long-read sequencing can reveal the complete and accurate complement of structural variants in a genome.

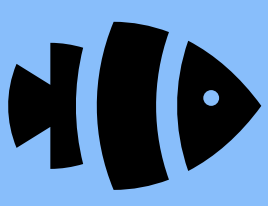
OPTICAL GENOME MAPPING

An emerging genome scanning technology that creates a detailed visual map of the structural variants in the human genome

SOME BENEFITS

5-7 day turnaround (more than one cytogenetic test but less than standard cascade)

Streamlined data analysis

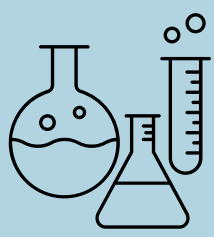


Not restricted to specific probes, fluorescent in-situ hybridisation (FISH)

Provides novel data

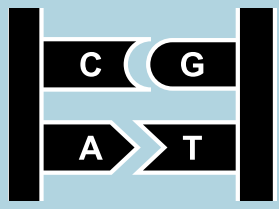
SOME LIMITATIONS

Secondary confirmation still advisable



Sample preparation laborious and fiddly

'On the ground' runtime too long for clinical benefit



No nucleotide data - SNP diseases not covered

RESEARCH

Optical genome mapping is being used in research alongside standard of care tests to investigate its efficacy and reliability.

BLOOD CANCERS

Optical genome mapping has provided increased resolution that has resulted in changes to

- treatment strategies
- risk classifications and
- qualified patients for entry to clinical trials

SOLID TUMOURS

Research is more limited due to tumor DNA quality. An average of 92% sensitivity and 98% specificity has been reported when using optical mapping alone.

RARE DISEASE

A user-friendly pipeline specifically to handle complex structural variants found in rare diseases has been developed.

READY FOR THE CLINIC?

- Optical genome mapping is not a replacement for next-generation sequencing but can work alongside to overcome next-generation sequencing shortcomings.
- More attention to 'on the ground' logistics, especially throughput, is needed