

Biomnis



BRCA1/2 by long-read sequencing

A new quick and accessible test for prescribing PARP inhibitors



The presence of a germline mutation of *BRCA1* and *BRCA2* genes determines whether PARP inhibitors are prescribed in the treatment of some types of ovarian, breast, pancreatic and prostate cancer, thus playing a crucial role in cancer management.

The faster the treatment is correctly adapted, the better the therapeutic outcomes for patients.

To meet this demand, Eurofins Biomnis has developed an analysis of the *BRCA1* and *BRCA2* genes within 10 days.

Long-read sequencing (third-generation sequencing)

Long-read sequencing can produce significantly longer reads than second-generation shortread sequencing, with several thousand of base pairs compared to a few hundred.



Comparative alignments of long-read and short-read sequencing on the BRCA2 gene

Driven notably by Oxford Nanopore Technologies, the long-read approach offers numerous advantages:

- Comprehensive study of exonic and intronic regions, detection of sequence variants, deletions, and insertions,
- Reduction of sequencing time,
- Optimisation of sequencing costs.

Eurofins Biomnis uses the long-read approach in routine lab work for the study of the *MEFV*, *TTR* and *PMS2* genes and now also *BRCA1/2*.

Key points of the BRCA1/2 test by Eurofins Biomnis



Results delivered within 10 days



ISO 15189 accredited test



Optimised cost of execution



Sensitivity and specificity at least equivalent to exome sequencing techniques for SNVs, indels, and CNVs



Comprehensiveness

Amplification and sequencing of the entire BRCA1/2 gene, studying both exonic and intronic regions, and detecting point variants, indels, and copy number variations

Benefits of the BRCA1/2 test by long-read sequencing

- A response to urgent or therapeutic situations
- Improved test accessibility due to optimised cost
- Reduction of uncertainty period for patients and their families



* Mean Qc metrics for 1 sample : Mean read length = 4,163.9 Median read length = 1,732.0 Number of reads = 76,520.0 Read length N50 = 10,477.0 STDEV read length = 5,545.3

Why Eurofins Biomnis?



- Specialised clinical pathology laboratory
- ISO 15189 accreditation
- Authorisation to carry out germline genetic tests
- Certified clinical pathologists
- Mastery of sequencing techniques in diagnostic practice
 - Ongoing bio-pathological support



Sequencing of BRCA1/2 Genes	
Test code	BRCAN
Pre-analytical	5 mL Total Blood EDTA, Refrigerated
Turnaround time	13 days
Technique	Long-read Sequencing, Oxford Nanopore Technologies
Required document	Test request form B109-INTGB available on www.eurofins-biomnis.com >
	Test guide > Test code BRCAN
Price	Please contact us
Related test	Somatic BRCA1 and BRCA2 mutations (Test code BRCAS)

For more information

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