Product Profile

GeneRead® QIAact BRCA 1/2 Panel

An assay that goes beyond inherited breast cancer

Introduction

The tumor suppressor-encoding BRCA 1 and 2 genes (BRCA 1/2) represent an example of well understood and broadly recognized cancer molecular genetics. Inherited deleterious mutations in these genes greatly increase the risk for familial forms of breast and ovarian cancer. While less widely known and much harder to detect, somatic mutations in these genes also play a critical role in cancer development and progression in non-inherited forms.

The GeneRead QIAact BRCA 1/2 Panel (Cat no. 181920) for use with the QIAGEN GeneReader[™] NGS System, is an amplicon-based NGS panel designed to capture all mutations in both genes. The high level of analytical sensitivity of this integrated NGS assay enables detection of both germline and somatic mutations, making this the first full solution for BRCA 1/2 analysis in FFPE samples.

- Complete coverage of BRCA 1 and 2: all exons, plus flanking intronic regions
- All mutations: germline or somatic, SNVs or InDels
- Proven performance with somatic mutations in FFPE (formalin-fixed paraffin embedded) samples
- Interpretation at your fingertips: part of a complete and automated NGS workflow including full bioinformatics analysis and interpretation with QCI[™] Analyze and Interpret



Figure 1. The complete GeneReader NGS System workflow. The first three steps (nucleic acid extraction to library preparation) may be performed either manually or using one of QIAGEN's automated solutions, using the GeneRead Q family of products.



Sensitive detection of somatic and germline mutations

Currently available BRCA 1/2 NGS assays focus on germline mutation detection in somatic tissues (from blood or buccal swabs). These assays cannot be easily adapted to identify somatic mutations in the cancer tissue, owing to challenges inherent to FFPE sample processing, the need for a much higher analytical sensitivity threshold and required bioinformatics optimization. Furthermore, an NGS solution is only sound when paired with a robust database that provides meaningful and actionable interpretation of all variants identified.

The GeneRead QIAact BRCA 1/2 Panel uses multiplex PCR-based target enrichment technology with a sophisticated and proven primer-design algorithm. This technique enables specific enrichment and unbiased amplification of the BRCA 1/2 genes for NGS, and provides complete coverage of the entire coding and essential non-coding regions of both genes. This enables the study of mutations, including InDels, in FFPE samples at a 5% variant allele frequency, with 100% precision and analytical sensitivity.

A complete sample to insight assay

The GeneRead QIAact BRCA 1/2 Panel is fully incorporated as part of the complete Sample to Insight GeneReader[™] NGS System (Figure 1). All steps from DNA extraction to sequencing and result interpretation have been developed in synchronization with the assay, making the workflow 'plug and play' and ready for implementation in any laboratory.

Integrated bioinformatics for interpretation at the push of a button

The GeneRead QIAact BRCA 1/2 Panel workflow incorporates a custom designed and fully integrated bioinformatics pipeline, comprising QCI Analyze for variant identification, and QCI Interpret for reporting of likely mutation impact based clinical research evidence (Figure 2). Guided by a robust and continuously updated bioinformatics database, our interpretation at the push of a button solution removes the bottleneck of data analysis.

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Figure 2. Example of the Variant Detail tab from a QCI Interpret report.

Ordering Information

Product	Contents	Cat. no.
GeneRead QIAact BRCA 1/2 Panel Kit	For target enrichment of all coding regions of the BRCA1 and BRCA2 genes	181920
GeneReader Platform	Next-generation sequencing instrument	9002312
QCI Analyze	The bioinformatic complement to the GeneReader NGS System	188001
QCI Interpret	For NGS data interpretation and reporting	830371

The GeneReader NGS System is for Research Use Only. Not intended for diagnostic procedures.

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