Ion AmpliSeq *BRCA1* and *BRCA2* Panel

A community panel designed with input from leading researchers

The Ion AmpliSeq™ *BRCA1* and *BRCA2* Panel contains primer pairs that target the coding regions of the tumor suppressor genes *BRCA1* and *BRCA2*, which have been implicated in hereditary breast and ovarian cancers.

This panel was designed and verified with input from leading cancer researchers from two institutes and has been utilized successfully by numerous cancer research laboratories, globally. These two groups verified the performance of the panel on 65 unique samples that were previously screened using orthogonal technologies. They combined the potential of Ion AmpliSeq™ technology and the affordability of Ion Torrent™ semiconductor sequencing to develop a variant screening solution that helps deliver:

- **High coverage of the target regions**—expanded targeted regions to include the entire coding region, including 10–20 bases of padding around all targeted coding exons. Amplicons overlap for sequence coverage redundancy.
- **High-performance amplicons and uniformity**—stringent primer design helps ensure that primers do not overlap and are not located in regions with high-frequency SNPs.
- **Fast turnaround time coupled with throughput flexibility**—enabling rapid time-to-results in processing either a small or large number of samples.
- **Rapid adoption for translational research labs**—accurate, economical, and easy-to-implement, end-to-end solution.

"Ion AmpliSeq technology has allowed us to substantially improve the turnaround time and cost of sequencing these important genes. The methodology proved robust enough to call even difficult mutations, including homopolymers, in these two genes."

Dr. José Luis Costa
Postdoctoral fellow

"The Ion AmpliSeq *BRCA* panel enabled us to develop a quick and easy high-throughput workflow, with minimal hands-on time. It is an accurate solution—of the 65 samples tested to date, we detected all expected mutations."

Dr. Arjen Mensenkamp
Clinical molecular geneticist

Designed with leading researchers:

Dr. Marjolijn Ligtenberg—Radboud University Nijmegen Medical Centre, the Netherlands
Dr. Arjen Mensenkamp—Radboud University Nijmegen Medical Centre, the Netherlands
Dr. José Carlos Machado—The Institute of Molecular Pathology and Immunology of the University of Porto, Portugal
Dr. José Luis Costa—The Institute of Molecular Pathology and Immunology of the University of Porto, Portugal

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Tested and verified using 65 unique samples

During development and optimization of this panel, collaborators carried out thorough testing and verification using 65 DNA samples previously characterized by capillary electrophoresis. Different types of variants including deletions, duplications, and insertions near homopolymer regions were analyzed, and all of the expected variants were detected with this panel.

Data from two of the 65 tested samples are available for download from the datasets section of Ion Community at **ioncommunity.thermofisher.com/community/datasets**

Ion AmpliSeq *BRCA1* and *BRCA2* Panel	
Targets	Coding regions of *BRCA1* and *BRCA2* genes
Average amplicon length	200 bp
Primer pools	167 pairs of primers in three primer pair pools
Input DNA	30 ng*
Amplicon coverage	• 100% of all targeted coding exons and exon–intron boundaries • Expanded target regions—additional coverage 10–20 bases beyond the targeted coding exon and exon–intron boundaries • Sequence coverage redundancy with overlapping amplicons across exons • High-fidelity primers
Verification	Verified by two laboratories on 65 samples with known mutations, including homopolymer variants with 7 and 9 bases; these samples were previously detected using capillary electrophoresis, and verification on the Ion PGM™ System yielded 100% sensitivity**
Multiplexing recommended	8 samples on an Ion 316™ Chip

* 30 ng input DNA was used for verification studies. Robust results have been observed with input DNA amounts ranging from 1 to 100 ng. Results may vary, pending the quality of extracted DNA material utilized.

** Participating laboratories verifying the Ion AmpliSeq *BRCA1* and *BRCA2* Panel used the following reagents and consumables: Ion AmpliSeq™ Library Kit v2.0, Ion Xpress™ Barcode Adapters Kits, Ion PGM™ 200 Sequencing Kit and Template Kit, andIon 316™ Chip.

Developed with the community, available to the community

The Ion AmpliSeq *BRCA1* and *BRCA2* Panel and other Ion AmpliSeq™ community panels may be reviewed and ordered via the Ion AmpliSeq™ Designer at ampliseq.com:

1. Log in to ampliseq.com using your Thermo Fisher Scientific account
2. Select “Panels tab” and then the “community panels” subtab to review panel design and order

Ordering information

Product
Ion AmpliSeq *BRCA1* and *BRCA2* Panel ordered at **ampliseq.com**

Find out more at **thermofisher.com/ampliseqcommunity**

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