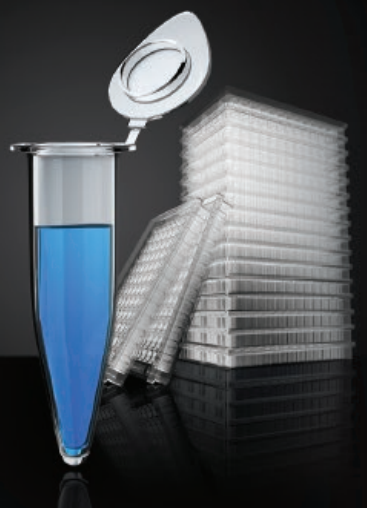
ion torrent
by life technologies™



Ion AmpliSeq™ *BRCA1* and *BRCA2* Panel

A community panel designed with 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 is now being further verified by an additional 5 laboratories. 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 semiconductor sequencing to develop a variant screening solution that meets these goals:

- **High coverage of the target region—**expanded targeted regions to include the entire coding region, including 10–20 bases of padding around all targeted coding exons. Amplicons are designed to overlap for sequence coverage redundancy
- **High-performance amplicons and uniformity—**stringent primer design ensures 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
- **Adoptable by other translational research labs—**accurate, economical, and easy-to-implement end-to-end solution that can be widely adopted

Ion AmpliSeq™ *BRCA1* and *BRCA2* Community Panel	
Targets	Analyzes coding regions of *BRCA1* and *BRCA2* genes
Amplicon length	Average 200 bp
Primer pools	167 pairs of primers in 3 primer pair pools
Input DNA required	30 ng
Amplicon coverage	• Designed for 100% amplicon coverage of all targeted coding exons and exon–intron boundaries • Expanded target regions—amplicons were designed to cover 10–20 bases beyond the targeted coding exon and exon–intron boundaries • Created for sequence coverage redundancy with overlapping amplicons across exons • High-fidelity primers • No SNPs with frequency >0.5% are included in the last 5 nucleotides of the primers • Only SNPs with a population frequency of <0.5% are present at other positions
Verification	Verified by 2 laboratories on 65 samples with known mutations, including homopolymer variants with 7 bases 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

"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
Post-doctoral fellow

Designed with leading researchers:

Dr. Marjolijn Ligtenberg[1]
Dr. Arjen Mensenkamp[1]
Dr. José Carlos Machado[2]
Dr. José Luis Costa[2]

[1]Radboud University Nijmegen Medical Centre, The Netherlands

[2]The Institute of Molecular Pathology and Immunology of the University of Porto (IPATIMUP), 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 (**ioncommunity.lifetechnologies.com/community/datasets**).

"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

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. Create an Ion Community account by going to the Ion Community (**lifetechnologies.com/ioncommunity**)
2. Log in to **ampliseq.com** using your Ion Community account
3. Select "Panels tab" and then the "community panels" subtab to review panel design and order

Ion AmpliSeq™ technology—see our expanded offering of panels for your targeted DNA and RNA sequencing

Ion AmpliSeq™ technology delivers simple and fast library construction for affordable targeted sequencing of specific human or mouse genes or genomic regions. Based on ultrahigh-multiplex PCR, Ion AmpliSeq™ DNA and RNA panels comprise target-specific primers for regions of interest, enabling routine sequencing of precious samples (such as FFPE, FNB or FNA) on Ion PGM™ Systems. With a wide variety of preselected and community-designed panels and custom options, Ion AmpliSeq™ technology provides a flexible solution to meet your specific research needs.

	Human DNA	Human RNA	Mouse DNA
Custom panels	✓	✓	✓
Ready-to-use panels	✓	✓	
Community panels	✓		

Ordering information

Product	Cat. No.
Ion AmpliSeq™ *BRCA1* and *BRCA2* Panel ordered via **ampliseq.com**	
Ion AmpliSeq™ Library Kit 2.0	4475345, 4478378, 4478379
Ion Xpress™ Barcode Adapters Kits	4474517, 4471250, 4474009, 4474518
Ion PGM™ 200 Sequencing Kit and Template Kit	4474004, 4480285
Ion 316™ Chip (4 pack)	4466616

Learn more at **lifetechnologies.com/ampliseqcommunity**

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