

HEVA pro

Hereditary and somatic variants profiling in Breast and Ovary Cancer

HEVA pro

4bases HEVA pro is kit for the identification of mutations in genes related to Breast and Ovary cancer, Familial adenomatous polyposis (FAP) and hereditary nonpolyposis colorectal cancer (HNPCC), two syndromes of colorectal cancer predisposition, inherited in an autosomal dominant fashion.

HEVA pro allows the search for variants linked to a defect in homologous recombination repair (HRD), similarly to pathogenic variants in *BRCA1* or *BRCA2*. The defect in HR causes error-prone DNA repair pathways in the cell, resulting in increased genomic instability, which may be responsible for susceptibility to poly-(ADP)-ribose polymerase (PARP) inhibitors.

HEVA pro is a kit for the analysis of 50 genes through a molecular protocol based on Next Generation Sequencing (NGS) technologies. The kit is validated for germline and somatic analysis (SNPs, indels, CNVs) of DNA extracted from blood or body tissues (fresh, frozen, FFPE, FNA) samples. HEVA pro kit contains all reagents required for the preparation of the capture of specifically designed probes and for the NGS analysis using ThermoFisher sequencers.

TECHNOLOGY

The HEVA pro kit is part of a DNA-to-variant solution that offers streamlined content, easy-to-perform library preparation, push-button sequencing systems, and simplified data analysis.

WORKFLOW

Library preparation follows a straightforward, capture-based protocol that can be completed in as little as 36 hours, with < 3 hours hands-on time.

Resulting libraries can be normalized, pooled, and then loaded on to a chip for sequencing. Prepared libraries are sequenced on any compatible ThermoFisher sequencers.

VALIDATION

To demonstrate assay capabilities, clinical samples were run in a clinical setting. DNA quality and quantity of the libraries prepared were verified using Qubit and Agilent Bioanalyzer.

Profile of the prepared libraries in Figure .

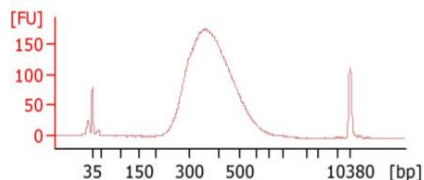


Figure 1. HEVA PRO Bioanalyzer library profile

Table 1: List of genes in HEVA pro

| | | | |
|-------|--------|---------|--------|
| ATM | MSH6 | FAM175A | LZTR1 |
| APC | MUTYH | MRE11 | CTNNB1 |
| BARD1 | NBN | PIK3CA | KRAS |
| BRCA1 | PALB2 | PMS2CL | ERBB2 |
| BRCA2 | PMS2 | XRCC2 | ALK |
| BRIP1 | PTEN | NF1 | EGFR |
| CDH1 | RAD50 | SPRED1 | RB1 |
| CHEK2 | RAD51C | MEN1 | CDKN2A |
| EPCAM | RAD51D | RET | CDK4 |
| MLH1 | STK11 | SMARCB1 | FLCN |
| MSH2 | TP53 | NF2 | RUNX1 |
| XPC | WRN | PTCH1 | CDKN1C |
| SMAD4 | HRAS | | |

SAMPLE PER RUN

| Instrument | Samples per run* | |
|-----------------------------|------------------|---------|
| | Germline | Somatic |
| Ion 316™ Chip/Ion 510™ Chip | 8 | 0 |
| Ion 318™ Chip/Ion 520™ Chip | 16 | 0 |
| Ion 530™ Chip | 48 | 0 |
| Ion PI™ Chip/Ion 540™ Chip | 160 | 8 |
| Ion 550™ Chip | 480 | 16 |

*the estimated maximum number of samples per chip assumes a reading depth of 300x for the germline and 5000x for the somatic. The optimal number of samples can be empirically estimated on the local setup.

The volume present in the kit is calculated to allow the subdivision into multiples of 8 analysis sessions. Dividing the kit in different ways decrease the total number of tests that can be performed.

ORDERING INFORMATION

| Product | REF |
|-----------|----------------------|
| HEVA pro | RC3040Y-16 (16 test) |
| HEVA pro | RC3040Y-96 (96 test) |
| Adapter | |
| Y ADAPTER | R9001-16 (16 test) |
| Y ADAPTER | R9001-96 (96 test) |