

Name
MRN
Gender
Age / DOB
Client Name
Nationality
Doctor Name :

Lab ID
Pat.File.No
Emirates / Passport ID :
Reg Date
Collection Date
Reporting Date

Recommendations

1. Please correlate clinically.
2. Genetic counseling for accurate interpretation of test results is recommended.
3. MLPA analysis of *BRCA1* and *BRCA2* genes is recommended to rule out the presence of large deletion/duplication or copy-number variations (CNVs).
4. There may be mutations in genes other than those tested that may be responsible for carcinogenesis, such as **TP53** (Li Fraumeni Syndrome), **PTEN** (Cowden syndrome), **CHEK2**, Lynch Syndrome, et c. Thus we would advise testing the patient via the NGS Hereditary Cancer Panel, to assess for mutations in other inherited Cancer genes.

Technical Notes

Methodology: Massively Parallel Sequencing (Next Generation Sequencing) Genomic DNA from the peripheral blood was enriched for the complete coding regions and splice site junctions of *BRCA1* and *BRCA2* genes using AmpliSeq for Illumina *BRCA* Panel system. The panel consists of three primer pools (167 amplicons) that target the entire coding region, including 10 to 20 bp of intronic [flanking sequences](#) around all coding exons, of both genes. Paired-End Sequencing was performed with 2x150 chemistry, on an Illumina platform. Reads were assembled and aligned to reference sequences based on NCBI RefSeq transcripts and human genome build GRCh37/UCSC hg19. The following quality control metrics are generally achieved: >95% of target bases are covered at >20x and mean coverage of target bases $\geq 100\times$. Sequence variants are reported according to the Human Genome Variation Society (HGVS).

Tools and Databases employed for analysis: Clinvar, OMIM, BIC, HGMD, UCSC genome browser, Uniprot, Ensembl, dbSNP, gnomAD, ExAC, Pubmed, Dgap, icgc, Kaviar, various bioinformatics analysis, predictive tools and disease-specific databases used as available and appropriate.

Gene Coverage

Indication Based Analysis:

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
BRCA1	100%	BRCA2	99.8%				

QC Metrics

Total aligned reads	99.12 %
Total reads	3.43 (M)
Total data generated	0.49 (Gb)
Total reads which passed mapping quality cutoff	0.45 (Gb)

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Important

- These test results should be interpreted by the referring clinician only in conjunction with the patient's clinical history, other test results, and any previous analysis of appropriate family members.
- Each first-degree relative of this individual has a one-in-two chance of having this mutation. Family members can be tested for these specific mutations with a single site analysis for each deleterious mutation.
- Variants of unknown significance are mutations detected on sequence analysis that have either not been reported before, or whose effect cannot be determined based on the current knowledge standards and reporting guidelines. We recommend periodic review of this report on an annual basis, to determine change in pathogenicity status of reported mutations in the future as new research comes to light. If variants of unknown significance (VUS) with or without other abnormal clinical/lab findings are detected, follow-up testing of parents and other available family members is generally recommended to aid in determining the segregation pattern of the identified variant(s).
- Unless reported or predicted to cause disease, alterations found that do not result in an amino acid substitution are not reported. These common polymorphisms identified for this patient are available upon request.
- The classification and interpretation of all the variants in this assay reflect the current state of scientific understanding at the time this report was issued. In some instances, the classification and interpretation of such variants may change as new scientific information comes to light. We recommend a re-analysis of this report yearly, in order to take advantage of any new scientific data that may become available. Please contact the laboratory in case a re-analysis of the report is required.
- This test has been validated by the MDC LABS and CAP.

Test Limitations

- Testing has been performed assuming that the sample received belongs to the above-named individual(s) and that any stated relationships between individuals are accepted as true. It is also assumed that consent for the same was provided after pre-test counseling at the point of collection/referral.
- A negative result does not exclude a heritable form of cancer. This test only detects variants within the coding regions and intron-exon boundaries of the *BRCA1* and *BRCA2* genes.
- The test methodology currently does not detect large deletions/duplications, deep intronic variants, and regulatory region variants.
- The reported variants have not been Sanger confirmed. Sanger confirmation is recommended for the same.
- Phenotype variability may be due to modifying genetic/non-genetic factors and is not a part of the current analysis.
- Variants of hitherto unknown significance may be detected and may not be reported subject to analysis by various methodologies.
- This report is for research purposes only, not for use in clinical diagnostic or therapeutic applications.
- For tests performed on specimens received or collected from non-STMPL locations, it is presumed that the specimen belongs to the patient named or identified as labeled on the container/test request and such verification has been carried out at the point of generation of the said specimen by the sender.