BRCA 1 and 2 profile Complete Gene Sequencing , *PERIPHERAL BLOOD*

BRCA1 & BRCA2 Gene panel by NGS- Germline Study

Clinical Indication:

As mentioned on test requisition form there is Family history of cancer- Mother has breast cancer and Sister has ovarian cancer. Mrs. Anna has requested BRCA 1 and BRCA 2 test for genetic screening purpose.

Result

NEGATIVE

(No clinically relevant variant identified)

Key Findings:

Gene & Transcript	Variant	Exon	Coverage / VAF	Zygosity	Clinical Significance
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*Genetic test results are reported based on the recommendations of American College of Medical Genetics

Gene Summary:

BRCA1: This gene encodes a 190 kD nuclear phosphoprotein that plays a role in maintaining genomic stability, and it also acts as a tumor suppressor. The BRCA1 gene contains 22 exons spanning about 110 kb of DNA. The encoded protein combines with other tumor suppressors, DNA damage sensors, and signal transducers to form a large multi-subunit protein complex known as the BRCA1-associated genome surveillance complex (BASC). This gene product associates with RNA polymerase II, and through the C-terminal domain, also interacts with histone deacetylase complexes. This protein thus plays a role in transcription, DNA repair of double-stranded breaks, and recombination. Mutations in this gene are responsible for approximately 40% of inherited breast cancers and more than 80% of inherited breast and ovarian cancers. Alternative splicing plays a role in modulating the subcellular localization and physiological function of this gene. Many alternatively spliced transcript variants, some of which are disease-associated mutations, have been described for this gene, but the full-length natures of only some of these variants has been described. A related pseudogene, which is also located on chromosome 17, has been identified.

BRCA2: Inherited mutations in BRCA1 and this gene, BRCA2, confer increased lifetime risk of developing breast or ovarian cancer. Both BRCA1 and BRCA2 are involved in maintenance of genome stability, specifically the homologous recombination pathway for double-strand DNA repair. The largest exon in both genes is exon 11, which harbors the most important and frequent mutations in breast cancer patients. The BRCA2 gene was found on chromosome 13q12.3 in human. The BRCA2 protein contains several copies of a 70 aa motif called the BRC motif, and these motifs mediate binding to the RAD51 recombinase which functions in DNA repair. BRCA2 is considered a tumor suppressor gene, as tumors with BRCA2 mutations generally exhibit loss of heterozygosity (LOH) of the wild-type allele.

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Disclaimer:

The performance charactersistics of the test has been verified at Lupin Diagnostics Ltd. This test is limited to BRCA1 & BRCA2 genes analysis only, It should be noted that this test does not sequence all bases in a human genome, not all variants have been identified or interpreted, and this report is limited only to variants with evidence for causing or contributing to disease/clinical details.

This assay is not meant to interrogate most promoter regions, deep intronic regions, or other regulatory elements, and does not detect single or multi-exon deletions or duplications.

References:

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SIN No: MO00074324

If Values are marked with * , they are critical values.

***** End Of Report *****

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