Requesting Physician: B. Smith
Collected Date: 21-Apr-2019
Date received: 21_Apr-2019
Specimen type: Blood

FAMILIAL BREAST & OVARIAN CANCER GENE SCREEN REPORT

Test requested: BRCA1 & BRCA2 gene screening

Clinical details: Personal history of TNBC diagnosed at age 39

Test result: **A PATHOGENIC VARIANT WAS DETECTED**

BRCA1 NM_007294.3: c.[45dupT];[=]: p.Asn16* in exon 2

Results interpretation:

The heterozygous mutation c.[45dupT];[=]: p.Asn16* was identified in exon 2 of the BRCA1 gene. This variant is not listed on the Breast Information Core (BIC) database. However, this mutation introduces a premature stop codon, resulting in a truncated BRCA1 protein and based on current knowledge is considered pathogenic.

This mutation is consistent with a diagnosis of hereditary breast or ovarian cancer syndrome. This result has diagnostic and clinical management implications for both the patient and other at-risk family members developing breast and/or ovarian cancer. First degree relatives of this patient have a 50% risk of inheriting the abnormal gene and predictive testing for this mutation is available through the appropriate genetic service.

Please note that common polymorphisms, synonymous changes and intronic variants outside of splice sites have not been reported.

Comment:

For counselling and assessment of the risk to the patient and family members please contact the appropriate genetic service.

Test description:

DNA sequencing analysis: Automated Next-Generation Sequencing of all coding exons and flanking intron junctions of the BRCA1 and BRCA2 genes.

MLPA: Gene dosage was assessed using Multiplex Ligation-Dependent Probe Amplification (MLPA) and kits available from MRC-Holland. The specific kits used were BRCA1 (P002D) and BRCA2 (P045B). This analysis detects large rearrangements.

This analysis does NOT exclude the possibility of other mutations not amenable to our analytical methods being present.

The nomenclature used throughout this report is in accordance with the Human Genome Variation Society (HGVS) guidelines, which can be found at www.hgvs.org.

Reference Sequence GenBank Accession Number: BRCA1 KM_007294.3; BRCA2 NM_000059.3.

In all family DNA studies, the accuracy of the report assumes stated relationships within the kindred, clinical diagnosis and identification of samples to be correct. Data are not available to accurately quantitate the very small but finite errors inherent in the use of molecular biological techniques for diagnosis.

Reported by: A Brown

Authorised by: S Barnes