13405

CONFIDENTIAL

BRACAnalysis®

Single Site Analysis Result

PHYSICIAN	SPECIMEN		PATIENT	
Dr. Palanki Satya Dattatreya **Positive Bioscience** **533 Kanta Terrace, Kalbadevi Rd** **Opposite Edward Cinema** **400002 Mumbai** **India**	Specimen Type: Collection Date: Accession Date: Report Date:	**Blood** **22 Jan 2017** **25 Jan 2017** **02 Feb 2017**	Name: Date of Birth: Patient ID: Gender: Accession #: Requisition #:	 **10 Jul 1962** **17806** **Male** **G0017806-BLD** **05219609**

BRCA1 Gene: Deleterious Mutation c.81-2A>C Detected

It is our understanding that this patient was identified for testing due to a personal or family history of a gene mutation.

Functional Significance: Abnormal Protein Production and/or Function
The heterozygous germline BRCA1 mutation c.81-2A>C is located 2 nucleotide(s) upstream of exon 3. This mutation occurs within a consensus splice junction, and it is predicted to result in abnormal mRNA splicing.

A similar mutation at this splice acceptor site has been shown to result in a truncated BRCA1 protein by disrupting the normal acceptor site and utilizing an alternative acceptor site 7 nucleotides downstream (Martin ES et al. Ca Genet & Cytogenet 2004, 150:173-175). c.81-2A>C is expected to have a similar effect.

Clinical Significance: High Cancer Risk
This mutation is associated with increased cancer risk and should be regarded as clinically significant.

Although the exact risk of breast and ovarian cancer conferred by this specific mutation has not been determined, studies indicate that women with deleterious mutations in BRCA1 have a risk of breast cancer of 28%-51% by age 50 and 46%-87% by age 70. The risk of ovarian cancer is 13%-23% by age 50 and 39%-63% by age 70. There is a 20% risk of a second primary breast cancer within five years and a 12.7% risk of ovarian cancer within 10 years of a breast cancer diagnosis. This mutation may also confer an increased (albeit low) risk of pancreatic, prostate and male breast cancer, as well as some other cancers. Each first degree relative of this individual has a one-in-two chance of having this mutation. Family members can be tested for this specific mutation with a single site analysis. For references and additional management information, please see the attached BRCA1 Management tool or visit www.myriadpro.com.

Analysis Description: The Technical Specifications summary (www.myriadgenetics.eu) describes the indications, analysis, method, performance, nomenclature and interpretive criteria of this test. Analysis consists of sequencing of the specific mutation(s) indicated above of the BRCA1 Gene (OMIM 113705/GenBank entry U14680) and/or BRCA2 Gene (OMIM 600185/GenBank entry U43746). The interpretation of this test may be impacted if the patient has a hematologic malignancy or an allogeneic bone marrow transplant. Some or all of the analysis might have been performed at Myriad Genetics Laboratories, Inc. In cases where Myriad identifies benign variants for which there is strong evidence that they are not associated with disease, these variants will not be reported. The classification and interpretation of all variants identified in this assay reflects 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 becomes available. In the future, should Myriad significantly modify the interpretation of one or more variants, an amended report will be issued.

To discuss any questions regarding this result, please contact helpmed@myriadgenetics.eu.

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