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Order no.: 62511101
Order received: 19 Sept. 2018
Sample type: blood, filter card
Sample collection date: not available
Report date: 25 Sept. 2018
Report type: Final Report

Patient no.: **1324141**, First Name: **Zinnie**, Last Name: **Puri**
DOB: **05 Nov. 1979**, Sex: **female**, Your ref.: **-**

Additional report recipient(s): Dr. Sunil K. Tadepalli, Centogene India PVT. LTD., Management, 107, Wegmans Business Park, Know ledge Park - III, 201308 Surajpar-Kasna Road, Greater Noida, India

Test(s) requested: BRCA1, BRCA2 panel

CLINICAL INFORMATION

The patient with bilateral breast cancer (ductal carcinoma in situ and invasive ductal carcinoma).

NEGATIVE RESULT

INTERPRETATION

No clinically relevant variant has been detected in the BRCA1 or BRCA2 genes by sequencing.

RECOMMENDATIONS

- As large deletions/duplications not detectable by sequencing have been described in the BRCA1 and BRCA2 genes, we recommend proceeding to the MLPA analysis.
- We recommend analysis of further gene panels relevant to clinical diagnosis (eg. CentoBreast panel).
- Genetic counselling is also recommended.

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METHODS

Genomic DNA is enzymatically fragmented and regions of interest are selectively enriched using capture probes targeted against coding regions of panel genes. Libraries are generated with Illumina compatible adaptors and sequenced on an Illumina platform.
For the BRCA1, BRCA2 panel, the entire coding region of the BRCA1, BRCA2 genes including 10 bp of flanking intronic sequences are targeted. Our Plus Panel includes analysis of all reported disease causing deep intronic and regulatory mutations described outside the coding +/-10 boundary. Due to limitations of the method, the targeted sequences within the requested panel may not be covered 100%. Missing regions or regions of poor quality are completed with classical Sanger sequencing to achieve 100% coverage of all genes within this panel. Raw sequence data analysis, including base calling, demultiplexing, alignment to the hg19 human reference genome (Genome Reference Consortium GRCh37) and variant calling is performed using validated in-house software. All identified variants are evaluated with respect to their pathogenicity and causality, and these are categorized into classes 1 - 5. All variants related to the phenotype of the patient, except benign or likely benign variants, are reported. Structural changes (i.e. CNVs, inversions, repeat expansions etc.) are not assessed with the NGS data.
Variants of relevance identified by NGS are continuously and individually in-house validated for quality aspects; those variants which meet our internal QC criteria (based on extensive validation processes) are not validated by Sanger.

ADDITIONAL INFORMATION

This test was developed and its performance validated by CENTOGENE AG. The US Food and Drug Administration (FDA) has determined that clearance or approval of this method is not necessary and thus neither have been obtained. This test has been developed for clinical purposes. All test results are reviewed, interpreted and reported by our scientific and medical experts.

To also exclude mistaken identity in your clinic, several guidelines recommend testing a second sample that is independently obtained from the proband. Please note that any further analysis will result in additional costs.

The classification of variants can change over the time. Please feel free to contact CENTOGENE (customer.support@centogene.com) in the future to determine if there have been any changes in classification of any reported variants.

DISCLAIMER

Any preparation and processing of a sample from patient material provided to CENTOGENE by a physician, clinical institute or a laboratory (by a "Partner") and the requested genetic and/or biochemical testing itself is based on the highest and most current scientific and analytical standards. However, in very few cases genetic or biochemical tests may not show the correct result, e.g. because of the quality of the material provided by a Partner to CENTOGENE or in cases where any test provided by CENTOGENE fails for unforeseeable or unknown reasons that cannot be influenced by CENTOGENE in advance. In such cases, CENTOGENE shall not be responsible and/or liable for the incomplete, potentially misleading or even wrong result of any testing if such issue could not be recognized by CENTOGENE in advance.

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