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# BRCA1 & 2 Somatic Gene Sequencing Report

| Specimen Type | |
|---|---|
| FFPE Block - RG9723 A1 | |
| Specimen Collection Date & Time | Date & Time of Accessioning |
| 04/06/2024 00:00 Hrs | 06/06/2024 17:51 Hrs |

| | |
|---|---|
| Case Number: | **102240091589** |
| Patient Name: | **B. A. K. M. Weerakoon** |
| Dob/Sex: | **62 Yrs/Female** |
| Patient Location: | **Colombo** |
| Hospital Name: | **Aegle Omics (Private) Limited** |
| Physician Name: | **Dr. Mahendra Perera** |
| Date & Time of Reporting: | **22/06/2024 14:31 Hrs** |

## TEST INFORMATION

The ***BRCA1*** and ***BRCA2 somatic* gene sequencing test** aims is a Next Generation Sequencing (**NGS)** based test in order to determine pathogenic and likely pathogenic **Single Nucleotide Variants (SNVs)** and **small Insertion/ Deletions (Indels),** that are clinically relevant for PARPi therapy.

## CLINICAL HISTORY

Invasive carcinoma of breast, grade III, Er- positive, pr/her2 - negative, Family history is not available.

## RESULTS

**Negative**

| Genes Tested | Mutation Detected |
|---|---|
| *BRCA1* | No clinically relevant variant identified |
| *BRCA2* | No clinically relevant variant identified |

## INTERPRETATION

- **No clinically relevant variant was identified in the *BRCA1* or *BRCA2* genes by NGS.**

## RECOMMENDATIONS

- Genetic counseling is recommended for the patient.
- The test results should be interpreted in the context of the patient's medical evaluation, family history, and racial/ethnic back-ground in order to arrive at accurate diagnosis, prognosis or for therapeutic decisions.

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Dr. Shivani Sharma
DCP, DNB, DipRCPath. Reg. No. 1906

Dr. Rahul Katara, Ph.D.



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Case Number: **102240091589** Patient Name: **B. A. K. M. Weerakoon** Ordering Physician Name: **Dr. Mahendra Perera**

## ADDITIONAL INFORMATION

**Test details:**

The CORE Diagnostics BRCA Panel includes *BRCA1* and *BRCA2* genes associated with hereditary breast and ovarian cancer (HBOC) syndrome and to a lesser extent other cancers such as prostate cancer, pancreatic cancer, and melanoma. The test utilizes Oncomine BRCA Research Assay, which is based on the proven Ion AmpliSeq technology. The assay has been validated on clinical research samples and provides 100% exonic coverage, including flanking intronic sequences with high average depth of coverage (>500X).

**For the index patient, Percent reads on target was 99.13% with a 87.23% Target base coverage at 500X.**

Genomic DNA is isolated from the blood sample and quantified using Qubit Fluorometer. Approximately 20 ng DNA is used for target amplification. The amplified DNA is subjected to adapter ligation and Ion Xpress™ Barcode generation for specific library preparation. The generated high quality library is subjected to next generation sequencing (NGS) on the ION S5 sequencing platform. The output sequences are aligned to the human reference genome hg19 (GRCh37). The alignments and variant calling is done using the ION S5 torrent server. Variants are identified and interpreted using Ion Reporter Software. The identified variant(s) is(are) annotated according to HGVS sequence variant nomenclature. Multiple in-silico predictors, such as SIFT, PolyPhen, MutationTaster, NNSPLICE, and ASSP etc. are used for variant impact on the protein function. Population and literature databases such as dbSNP, Exome Aggregation Consortium (ExAC), genome Aggregation Database (gnomAD), ClinVar, HGMD and PubMed etc are used for variant summary and classification. Variants are labelled based on the American College of Medical Genetics (ACMG) recommendations for 5-tier variant classification system: Pathogenic, Likely Pathogenic, Variant of Uncertain Significance (VUS), Likely Benign and Benign. Clinically relevant variants identified by NGS are continuously validated in-house by a second independent method (Sanger) for quality aspects; therefore those variants which do not meet our internal QC criteria (based on extensive validation processes) are confirmed by Sanger sequencing.

**Limitations and disclaimer:**

Despite all precautions taken, the error (administrative and technical) associated with these types of molecular diagnostic tests can be as high as 1% to 2%. Rare polymorphisms may be present that could lead to false negative or false positive results. The quality of sequencing and coverage varies between regions; many factors such as homopolymers, GC-rich regions etc. influence the quality of sequencing and coverage. This may result in an occasional error in sequence reads or lack of detection of a particular genetic variant. Variants that have not been confirmed by an independent analysis could represent technical artifacts. Not all variants detected may be listed in the report. Inclusion of variants is dependent upon our assessment of their clinical significance. Additionally, the presence of a pathogenic/likely pathogenic variant may not be predictive of response to therapy in all patients. The selection of any potential treatment/course of action based on this report rests solely within the decision and judgment of the treating physician and patient. Decisions on patient care should be based on the independent medical judgment of the treating physician based upon all available clinical information, according to the applicable standard of care and should not be based solely on the tests and information contained in this report. Accurate interpretation of this report is dependent on provided detailed clinical history of the patient. In the event of unavailability of detailed clinical history, the lab cannot guarantee the accuracy of the interpretation. The classification of variants can change over the time. Please feel free to contact CORE Diagnostics (info@corediagnostics.in) in future to determine if there have been any changes in classification of any reported variants.

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| Aditi Aggarwal, Molecular Scientist | Dr. Shivani Sharma<br>DCP, DNB, DipRCPath. Reg. No. 1906 | Dr. Rahul Katara, Ph.D. |
|---|---|---|



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## Question?

Contact us at **+91 124 4615 615**

Toll Free Helpline **+91 8882899999**

## CONDITIONS OF REPORTING

1. The tests are carried out in the lab with the presumption that the specimen belongs to the patient named or identified in the bill/test request form.
2. The test results relate specifically to the sample received in the lab and are presumed to have been generated and transported per specific instructions given by the physicians/laboratory.
3. The reported results are for information and are subject to confirmation and interpretation by the referring doctor.
4. Some tests are referred to other laboratories to provide a wider test menu to the customer.
5. Core Diagnostics Pvt. Ltd. shall in no event be liable for accidental damage, loss, or destruction of specimen, which is not attributable to any direct and mala fide act or omission of Core Diagnostics Pvt. Ltd. or its employees. Liability of Core Diagnostics Pvt. Ltd. for deficiency of services, or other errors and omissions shall be limited to fee paid by the patient for the relevant laboratory services.



**CORE Diagnostics National Reference Lab - Gurugram (102)**

406, Udyog Vihar, Phase III, Gurgaon 122016

**CORE Diagnostics Lab - New Delhi (103)**

C-13, Green Park Extension, New Delhi – 110016

**CORE Diagnostics Lab - Bangalore (105)**

1st Floor, KMK Tower, 142 KH Road, Bangalore - 560027

**CORE Diagnostics Lab - Lucknow (109)**

J.S. Tower, Plot No. K-702, Sector-K, Ashiyana,

Near Raj Luxmi Sweets, Lucknow-226012

**CORE Diagnostics Lab - Bhubaneswar (108)**

Plot No. 249, Near Police Academy, AIIMS Nagar,

Patrapada, Bhubaneswar-751019

The test was processed in Lab 102.