

CASE 051

AUTHORS: Dr. Avshesh Mishra, Dr. Atreye Majumdar, Rajan Pal, Akash Kumar, Arti Verma, Vipin Kumar.

ACKNOWLEDGEMENTS: Dr. Shivani Sharma, Dr. Aditi Dewan.

EFFICACY OF NGS TECHNOLOGY TO DETECT NOVEL VARIANTS IN THE BRCA1/2 GENES

Abbreviations: BRCA1/2 – BRCA1 and BRCA2, HBOC -Hereditary Breast and Ovarian Cancer, Indels - Insertions or Deletions, NGS - Next generation sequencing, SNVs Single Nucleotide Variations.

PATIENT DETAILS

A 47 years old female diagnosed with recurrent carcinoma ovary was referred to us for BRCA1 and BRCA2 gene sequencing test. There was no known family history of cancer.

CHIEF COMPLAINTS AND HISTORY OF PRESENT ILLNESS

The patient was a follow up case of carcinoma ovary post surgery (8/16) and adjuvant chemotherapy (Paclitaxel+Carboplatin x 6 cycle). She then had a recurrence for which she was given Lipodox+Cisplatin x 6 cycles, had a clinical remission and was kept under observation. However, again the patient had a rise in CA 125 levels, was put on oral Etoposide, and is now under observation.