

## CASE STUDY

# Strand Germline Test Provides Therapeutic Options for Ovarian Cancer Patient

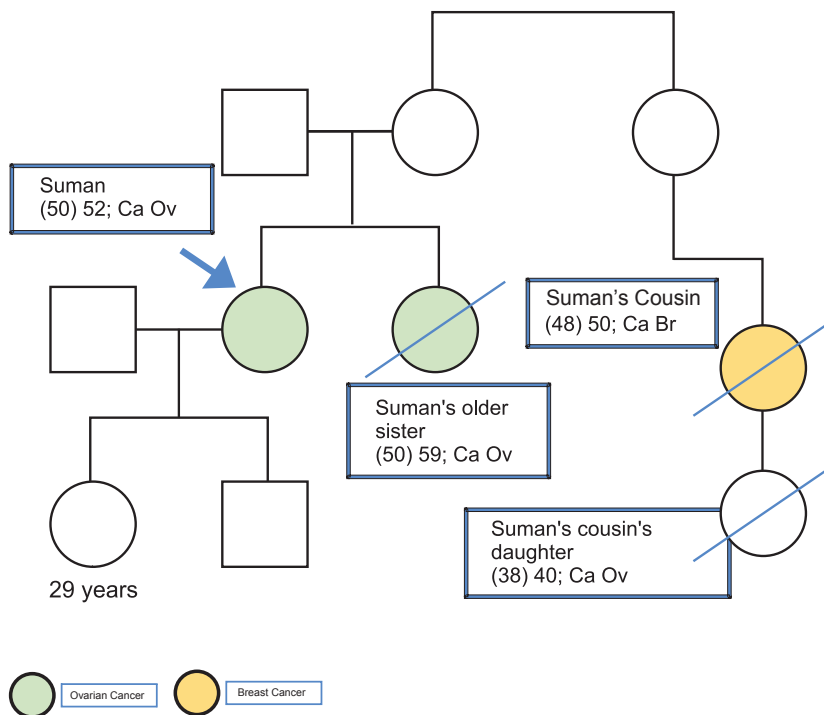
### Patient Profile

Suman\*, a 52-year-old woman from Mumbai was diagnosed with ovarian cancer in 2016. Suman was advised standard chemotherapy for ovarian cancer, as well as genetic testing to understand the cause of her ovarian cancer.

### Family History

Suman has a daughter who is married. Suman's sister was diagnosed with ovarian cancer, at the age of 50 years, nine years earlier. She had succumbed to the disease. Suman also had a cousin sister who lost her life to breast cancer at the age of 48 years. Family history revealed that the same cousin's daughter had died, because of ovarian cancer at the age of 38 years!

### Family Tree - Pre-Test Genetic Counselling



**Gender:** Female

**Age:** 52 years

**Location:** Mumbai, Maharashtra

**Diagnosis:** Ovarian Cancer & Breast Cancer

**Strand Test:** Germline

**Conclusion:**

Precise Treatment Plan, Early diagnosis and preventive action for family members established, QoL improved for patient

\*Name changed to protect patient privacy

## Genetic Analysis

Suman was advised to take the Strand Germline Cancer Test, based on the prevalence of breast and ovarian cancer amongst her relatives. Genetic testing revealed that Suman has a mutation in the *BRCA1* gene that caused her ovarian cancer.

### RESULT



Positive for a heterozygous ‘**pathogenic**’ variant, which was detected in exon 2 of the *BRCA1* gene.

### Key Findings

Gene	Variation	Zygoty	Clinical significance
<i>BRCA1</i>	chr17:41276045_41276046delCT c.68_69delAG p.Glu23ValfsTer17	Heterozygous	Pathogenic

## Treatment Plan

Suman had been advised standard chemotherapy prior to the genetic analysis of her DNA. Additionally, the identification of the *BRCA1* mutation in her DNA makes her eligible to receive PARP inhibitor therapy, as well. Treatment of ovarian cancer patients with PARP inhibitors results in longer periods of progression-free survival in patients<sup>1-5</sup>.

## Conclusion

- Suman, a 52-year-old woman from Mumbai was diagnosed with ovarian cancer and was advised genetic testing.
- Suman’s family history indicated a strong likelihood of hereditary breast and ovarian cancer in the family.
- The Strand Germline Cancer Test facilitated the identification of a *BRCA1* founder mutation (***BRCA1 c.68\_69delAG***) in the patient’s DNA.
- Identification of this mutation indicates her eligibility to receive PARP inhibitor therapy instead of standard chemotherapy.
- Application of recommended genetic testing guidelines at the time of diagnosis of Suman’s sister’s ovarian cancer would have possibly alerted the whole family to the presence of hereditary breast and ovarian cancer in their family tree. Given that her sister was diagnosed with ovarian cancer at the age of 50 years, a few years earlier than the patient, she should have been considered as an index case as per recommended guidelines and advised a germline cancer test.
- Suman’s daughter now has a chance to get her DNA tested, plan her children, and avail of medical surveillance, if she is also positive for the same mutation.

## References

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2. Oza, A. M. et al. Olaparib combined with chemotherapy for recurrent platinum-sensitive ovarian cancer: a randomised phase 2 trial. *Lancet Oncol.* 16, 87–97 (2015).
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4. Jenner, Z. B., Sood, A. K. & Coleman, R. L. Evaluation of rucaparib and companion diagnostics in the PARP inhibitor landscape for recurrent ovarian cancer therapy. *Futur. Oncol.* 12, 1439–1456 (2016).
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## Strand Germline Panel

The Strand Germline Cancer Test is designed to identify genes that are involved in several inherited cancers. The following genes are analyzed in samples from breast and ovarian cancer patients, as per international genetic testing guidelines.

*ATM, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, NBN, PALB2, PTEN, RAD51C, RAD51D, TP53*



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