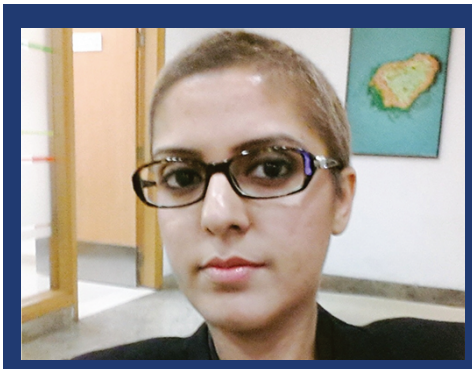
Strand Germline Test: Identification Of A Pathogenic *BRCA1* Mutation In A Patient With Aggressive Hereditary Breast Cancer



Gender: Female

Age: 36 years

Diagnosis: Triple negative breast cancer

Strand Test: Strand Germline Cancer Test

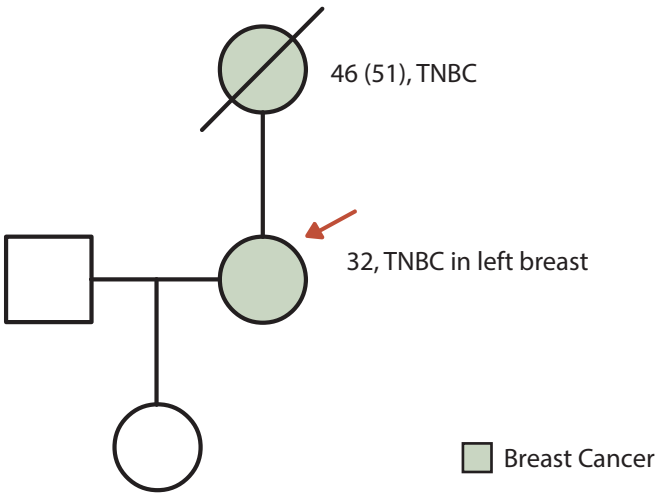
Conclusion: Identification of a *BRCA1* mutation enables intensive screening for patient, and options for prophylatic surgery.

Patient Profile

Breast cancer was definitely not dinner-time conversation in 32-year-old Dimple Bawa's* life, a successful entrepreneur. Her life centered around her family, her work and was sailing smoothly. Pain experienced during an accidental trauma to the left breast prompted a medical check-up. She was diagnosed with triple-negative breast cancer (TNBC), an aggressive form of breast cancer, which had spread through most of her left breast. Suddenly, her life turned around completely and this aggressive disease took centrestage in her life.

Family Tree- Pre-test Genetic Counselling

Dimple had lost her mother to breast cancer, when she was 26 years old. Her mother was diagnosed with breast cancer at the age of 46 years. In Dimple's mother's case, she was undergoing treatment for her breast cancer, but the disease recurred during treatment, and had metastasized to the brain, liver and lungs leading to her death at the age of 51 years.



Initial Treatment

Dimple was treated with 6 rounds of chemotherapy as well as radiotherapy. A mastectomy was also performed on her left breast to remove residual cancerous tissue.

Three years after her treatment, Dimple decided to undergo genetic testing to ascertain the genetic cause of her breast cancer, and to identify if her cancer was hereditary in nature.

*Name and other details shared post patient consent

Genetic Testing

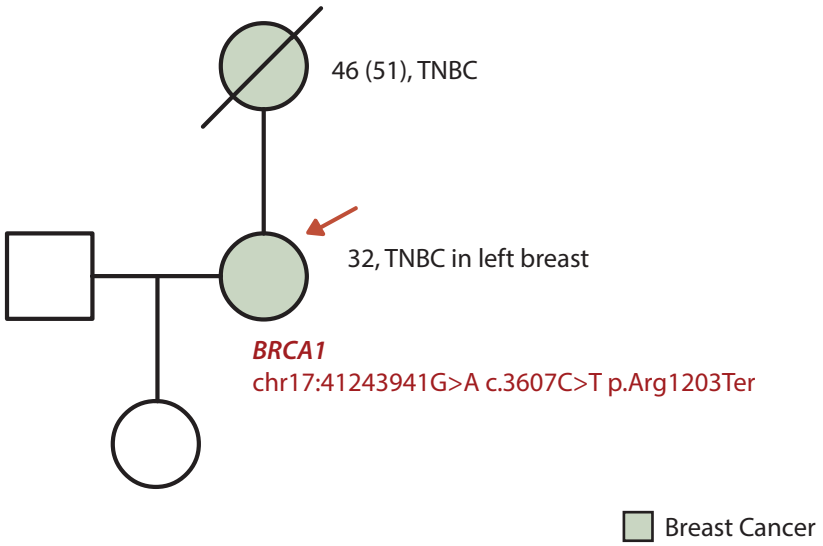
The Strand Germline Cancer Test was performed on the sample provided by Dimple. A hereditary pathogenic mutation in exon 10 of the *BRCA1* gene was identified in her genomic DNA.

RESULT ⊕ Positive for a heterozygous **'pathogenic'** variant, which was detected in exon 10 of the *BRCA1* gene.

Key Findings

Gene	Variation	Zygosity	Clinical significance
BRCA1	chr17:41243941G>A c.3607C>T p.Arg1203Ter	Heterozygous	Pathogenic

Family Tree – Post-test Genetic Counselling



Discussion

Dimple carries one copy (heterozygous) of a pathogenic variant in the *BRCA1* gene, which has been shown to be associated with hereditary breast and ovarian cancer predisposition.

- Women with a *BRCA1* germline pathogenic variation in heterozygous state are at an increased lifetime risk for breast cancer (50%-80%) and ovarian cancer (24%-40%). In addition, there is an increased risk for pancreatic cancer[1].
- Germline variations in the *BRCA1* gene in familial breast and ovarian cancer are inherited in an autosomal dominant manner, which means one copy of the altered gene in an individual is sufficient to increase the risk of developing cancer. Each first-degree relative (children, siblings and parents) of this individual has a 50% chance of having this variation.

*Name and other details shared post patient consent

Conclusion

- Awareness of her own status as a heterozygous carrier of a *BRCA1* mutation has enabled her to undergo intensive surveillance, and given the option to undergo prophylatic surgery for cancer risk reduction.

References

Petrucelli, N., Daly, M. B. & Pal, T. BRCA1- and BRCA2-Associated Hereditary Breast and Ovarian Cancer. GeneReviews® (1993).

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