CASE STUDY

Breast Cancer in a Man: Effects of an Inherited *BRCA2* Mutation

Gender: Male

Age: 63 years

Location: Hyderabad, Andhra Pradesh

Diagnosis: Breast Cancer

Strand Test: Germline Cancer Test

Conclusion:

- Hereditary Breast and Ovarian cancer in the family confirmed; Patient and siblings counselled about their risk for developing cancer

Introduction

Hereditary breast and ovarian cancer (HBOC) is emerging as a serious health issue in the Indian population. A recent study has shown that there are about 50 different mutations that can cause HBOC in the Indian population (1). More often risk is higher for women as compared to men and mostly HBOC patients are women.

As per medical guidelines accepted worldwide, all women who have relatives diagnosed with breast and / or ovarian cancer should get their genome tested for the presence or absence of mutant genes that can cause HBOC. The key word here is 'relatives', so are men also included in this category? Are men also likely to suffer from breast cancer? Looking at the data, it turns out, that although cases of male breast cancer (~1% of all cancer cases in men) are rare, men can also inherit genes that cause HBOC and some can actually develop male breast cancer.

Patient Profile

KV Arumugham*, a 63-year-old man, was referred to a prominent oncologist in Hyderabad. Diagnosis of male breast cancer was confirmed by histological examination of a biopsy of his breast tissue. Development of breast cancer in men indicates a strong likelihood of inheritance of genes associated with HBOC and hence, Arumugham was referred to Strand Life Sciences to get his genome tested.

Results of the Germline Test

A blood sample, provided by Arumugham for genetic testing was analyzed using the Strand Germline Cancer Test. Arumugham was found to be heterozygous for a pathogenic variant in the *BRCA2* gene.

Key Findings

- Arumugham has inherited a 'pathogenic' (disease-causing) variant in the *BRCA2* gene, which might be the causative factor for his breast cancer.
- Pathogenic mutations in the *BRCA2* gene are inherited in an autosomal dominant manner. Therefore, although Arumugham has a normal second copy of this gene, his chances of developing breast cancer (1-10%) as well as prostate cancer (39%), throughout his life, remain higher than those of people who do not have a mutant *BRCA2* gene (2).
- Due to the identified mutation, Arumugham also has an increased risk of developing melanoma, a type of skin cancer.

** Name changed to protect patient privacy*

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RESULT **Positive** for a heterozygous **'pathogenic'** variant in exon 11 of the *BRCA2* gene.

Results

Positive for a heterozygous **'pathogenic'** variant, which was detected in exon 11 of the *BRCA2* gene.

Key Findings

Gene	Variation	Zygosity	Clinical significance
BRCA2	chr13:32911303_32911306delins TATATGGTG c.2811_2814delinsTATATGGTG p.Gln937HisfsTer4	Heterozygous	Pathogenic

Treatment Plan

Arumugham underwent wide excision of the left breast lump with axillary clearance and oncoplastic reconstruction. Subsequently, he also underwent 2 cycles of chemotherapy with inj. Adrim- 60 mg/m2 – 110mg in 10 ml Ns- 15 minutes and inj. Endoxan- 60 mg/m2 – 110mg + mesna 100 mg in 500 ml NS over 2 hours with pre-hydration medicines.

Conclusions

- Arumugham was advised about his risk for development of prostate cancer in the future and was provided guidance about regular medical surveillance.
- Arumugham's children were advised to undergo genetic testing in order to understand their own risks for developing HBOC as well as other cancers.
- Genetic analysis confirmed the hereditary nature of Arumugham's breast cancer. He was given adjuvant chemotherapy

Strand® Germline Test

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, EPCAM, MLH1, MSH2, MSH6, NBN, NF1, PALB2, PMS2, PTEN, RAD51C, RAD51D, STK11, TP53

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References

1 Mannan AU, Singh J, Lakshmikeshava R, Thota N, Singh S, Sowmya TS, et al. Detection of high frequency of mutations in a breast and/or ovarian cancer cohort: implications of embracing a multi-gene panel in molecular diagnosis in India. J Hum Genet [Internet]. 2016;61(6):515–22. Available from: http://www.ncbi.nlm.nih.gov/pubmed/26911350

2 Petrucelli N, Daly MB, Pal T. BRCA1- and BRCA2-Associated Hereditary Breast and Ovarian Cancer [Internet]. GeneReviews®. 1993 [cited 2017 Feb 22]. Available from: http://www.ncbi.nlm.nih.gov/pubmed/20301425

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