CASE STUDY

Preventive Genetic Testing Eliminates Anxiety About Hereditary Breast and Ovarian Cancer

Gender: Female

Age: 40 years

Reasons for genetic testing:

- Family history of breast and throat cancer
- Preventive testing recommended

Conclusions:

- Proband negative for germline mutations in genes for HBOC
- Reassurance and ability to plan health management
- Awareness about risk for sporadic cancer

Patient Profile

Familial cancer is not a strange phenomenon at all. In fact, 5-10% of all cancers are caused by inheritance of genes that are mutated (changed in a way that affects their normal functions) (Romero-Laorden & Castro 2017; Felix et al. 2014). So, how does one protect oneself from cancer? Is there a way to understand whether one is at risk? Is there a simple test available? These were the questions swirling in Ruchira's*, mind, as she went about her daily life. She is a 40- year-old chartered accountant running her own firm and has several prominent clients in Hyderabad. Her paternal grandmother had breast cancer. One of her cousins from her father's side of the family had been diagnosed with throat cancer.

She was worried about her own risks for developing breast and /or ovarian cancer. After some deliberation, she decided to consult Dr. Praveen Dadireddy, a prominent oncosurgeon at Continental Hospitals, Hyderabad.

Genetic Testing

Dr. Dadireddy understood Ruchira's family history and her concern about inheriting genes that may increase her personal risk of cancer. He then suggested getting her DNA tested for the presence of mutations in high risk genes that are known to be strongly associated with breast and ovarian cancer predisposition. Ruchira was asked to get in touch with a representative from Strand Life Sciences, a leading company in genetic diagnostics in India.

The Strand Germline Cancer Test for breast and ovarian cancer was advised to her. A small saliva sample was provided by Ruchira, which was shipped to Bangalore for analysis.

Results of Genetic Testing

The Strand Germline Cancer Test is designed to assay for presence or absence of mutations in 12 genes that are strongly associated with hereditary breast and ovarian cancer. In Ruchira's case, DNA analysis showed that she did not have pathogenic (disease-causing) mutations in any of the 12 genes tested.

*Name changed to protect patient privacy

RESULT **Negative** (Uninformative) for disease-causing or likely disease-causing variants in the genes tested in this sample.

Key Interpretations

- Ruchira does not have germline mutant (abnormal) copies of the 12 genes tested, in her DNA.
- Her risk for developing inherited breast and ovarian cancer is low. However, the risk for developing sporadic (by chance) cancer is still present. These facts were explained to her by a genetic counselor from Strand Life Sciences.
- Her family history of cancer could also be a result of mutations in other genes or other regions of the tested genes, not covered by the test.
- Ruchira's chances of suffering from hereditary cancer are low. However, a healthy lifestyle and periodic health check-ups would still be advisable for her.

Conclusions

- Ruchira*, a 40-year-old chartered accountant was concerned about her risk for developing inherited cancer.
- She was prescribed the Strand Germline Cancer Test in order to determine her personal risk.
- The test returned a negative result, which indicates that germline mutations in the 12 genes tested, were not present in her DNA.
- Ruchira is now relieved at finding out that she does not have a high risk of developing hereditary breast and ovarian cancer.
- *Ad Hoc* testing (prior to the actual incidence of cancer) has made her aware of her risk and has allowed her to plan health management strategies.

Strand Germline Cancer Test

The Strand® Germline Cancer Test is a Laboratory Developed Test (LDT) that was developed and its performance characteristics determined by Strand Center for Genomics and Personalized Medicine at Strand Life Sciences.

Genes evaluated for risk of breast and ovarian cancer: 12 genes.

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

This test has now been expanded (April 2017 onwards) to cover 19 genes:
ATM, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH6, NBN, NF1, PALB2, PMS2, PTEN, RAD51C, RAD51D, STK11, TP53

References

Felix, G.E. et al., 2014. Germline mutations in *BRCA1, BRCA2, CHEK2* and *TP53* in patients at high-risk for HBOC: characterizing a Northeast Brazilian Population. *Human genome variation,* 1, p.14012. Available at: http://www.ncbi.nlm.nih.gov/pubmed/27081505 [Accessed June 6, 2017].

Romero-Laorden, N. & Castro, E., 2017. Inherited mutations in DNA repair genes and cancer risk. *Current Problems in Cancer*. Available at: http://www.ncbi.nlm.nih.gov/pubmed/28454847 [Accessed June 6, 2017].

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