

Case Study

Unilateral Breast Cancer



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Case presentation

A 30 year old physician was diagnosed with unilateral breast cancer. Now, at the age of 32 years, she is concerned about the risk to her children and her siblings in view of her strong family history. (Image of pedigree to be included)

Genetic testing

The Strand Germline Cancer Test, a multi-gene panel (13 genes - APC, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, NBN, PALB2, PTEN, RAD51C, RAD51D, STK11, and TP53) test was recommended.

Results

A disease causing variant was detected in the BRCA1 gene.

Follow-up after one year

This patient informed that she has three other siblings who were concerned about their own risks of developing cancer. Pre-test genetic counselling was conducted for the family and testing was initiated for other siblings based on this result. One of the sisters was found to carry the exact same mutation.

Literature review

Approximately 10% of breast cancer cases are thought to be hereditary and about 25% of these are caused by gene variations in the BRCA1 or BRCA2 genes. Individuals carrying variations in either one of these genes have a 40-80% probability of developing breast cancer, 11-40% risk for ovarian cancer and 1-7% for pancreatic cancer in their lifetime

Each first degree relative of this individual has 50% chance of carrying this BRCA1 variation. Individuals who are identified to carry a genetic variation in BRCA1 gene are recommended to undergo stringent surveillance, prophylactic measures such as bilateral mastectomy and bilateral salpingo-oophorectomy are offered, if appropriate and options of chemoprevention are suggested.

Ref: NCCN, 2016 guideline

Conclusion

This test helped the family in understanding the cause of cancer in every generation of the family. The sibling who also carried the BRCA1 gene mutation was educated about her lifetime chances of developing any form of cancer, surveillance and prophylactic measure options. A management protocol was put in place for both the siblings to ensure early diagnosis of cancer. Both the siblings are now compliant with the management protocol in place.

Physician comments about case
