CASE 074

CŌRE DIAGNOSTICS™

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CASE HISTORY:

We present a case of Breast Carcinoma in a 57yr old female who came to us for BRCA1 and BRCA2 gene testing. She gave history of cancer in right breast followed by second primary in left breast within one-year of chemotherapy. No metastasis was reported in any other part of body as per the PET-Scan reports. Family history of this patient comprised of breast carcinoma in sister who succumbed to it. BRCA1 & 2 full gene testing was ordered; which came out to be positive for heterozygous variant c.4508C>A (p.Ser1503Ter) in BRCA1 gene. Genetic testing became evident in diagnosis of root cause of cancer, also to check predisposition of variant in other unaffected family members. We proposed mutation specific testing for her children for segregation analysis and risk reduction options based on their individual reports. Entire process was looked after by Genetic counsellors; from end to end patient counselling and discussion of the same with treating medical oncologist.

TITLE:

Importance of segregation analysis and role of genetic counselling in multiple family members (affected and unaffected) of family with BRCA1 and BRCA2 gene mutation.

INTRODUCTION:

Breast Carcinoma is one of major cancers in Indian population. Kashmir valley shows a sharp increase in cases with approximately 66% of breast cancer patients having local-stage breast cancer, 28% having regional stage, and 6% having distant (metastatic) disease. The minimum and maximum age of the cases were 19 years and 78 years respectively, with increase in the age there is an incident increase in breast cancer. Kashmir valley experiences a higher death rate of breast cancer patients than rest of India. The high mortality rate of