

Lindor, 2015). However, identification of a hereditary BRCA1/BRCA2 mutation in a family is often via initial diagnostic testing in an individual with cancer. The benefits of identifying a germline mutation soon after diagnosis can have implications for early treatment decisions and access to more effective therapies e.g. poly (adenosine diphosphate ribose) polymerase (PARP) inhibitors. (*Konecny and Kristeleit, 2016*). Breast cancer risk management in BRCA1/BRCA2 carriers with advanced ovarian cancer is an under explored area of genetic counseling research.

CASE STUDY

A 46 year old female was diagnosed with stage IV high grade serous ovarian cancer. The patient provided adequate family history information, reporting breast cancer in paternal aunts under the age of 50 and a sister with bilateral breast cancer. There was a suspicion of ovarian cancer in her paternal grandmother (unconfirmed). The patient's treating medical oncologist informed the patient about the BRCA1 and BRCA2 genetic testing but no formal pre-test genetic counseling was conducted.

Genetic testing was carried out at CORE Diagnostics. The genetic testing revealed a frameshift mutation BRCA1 mutation, previously reported in the literature as a pathogenic variant (*Heterozygous variant c.5074+1G>A*) causing a high risk of breast and ovarian cancer. The patient was referred to the genetic counseling clinic at which point she asked about the implications of the BRCA1 mutation on ovarian cancer prognosis, breast cancer risk management and the risk for her first degree relatives. The genetic counselor initiated discussion included the broader implications for relatives and the psychosocial impact of learning about BRCA1 mutation status. The patient had an excellent partial response to neo-adjuvant chemotherapy (Carboplatin and Paclitaxel) with no residual disease after total abdominal hysterectomy and no evidence of disease after six months of adjuvant chemotherapy.

The patient was interested in accessing risk reducing breast surgery and after an initial discussion 2 months post ovarian cancer diagnosis she requested a follow up in the cancer genetics clinic 14 months post diagnosis for a further discussion to weigh up the risks and potential benefits.