

CASE 029

AUTHORS: Aparna Dhar MSc (Medical Sciences), Fellowship (Medical Genetics)

ACKNOWLEDGEMENT: Poonam Sharma BSc. & Saurabh Singh

A case study illustrating Genetic Counseling Challenges for when to consider risk-reducing Mastectomy for advanced stage ovarian cancer patient with a BRCA1/BRCA2 mutation.

INTRODUCTION

Germline mutations in BRCA1/BRCA2 significantly increases the risk of breast and ovarian cancer in women. This case study describes a BRCA1 germline mutation identified in a women with stage IV epithelial ovarian cancer and the provision of genetic counseling about BRCA1 associated breast cancer risk in the one and half years following diagnosis. This case study centers on the patient's enquiry about risk-reducing breast surgery.

BRCA1/BRCA2 associated cancer risk management includes increased cancer surveillance and risk reduction strategies. For breast cancer risk, additional screening using mammography and MRI is recommended with the aim of detecting cancer at an early stage (*Kriege et al, 2004*) The most significant breast cancer risk reduction strategy involves double mastectomy, usually combined with breast reconstruction. Risk reducing surgery is usually most relevant between approximately ages 30-60, during the years where relative breast cancer risk is high and the risk benefit balance is more likely to be favorable. A woman's age and general health have a critical impact on these risks. (*Easton et al, 2015*).

Risk reduction strategies are most relevant to women identified as carrying a BRCA1 and BRCA2 mutation who have not yet developed cancer and the role of risk reducing surgery in the management of hereditary breast and ovarian cancer is already well established (*Hartmann and*