Ambry Genetics
A Konica Minolta Company

Identifying Patients with Hereditary Ovarian Cancer

CASE EXAMPLE

Who is the Patient?

- 62 year old female
- Epithelial ovarian cancer diagnosed at 60y
- Previous *BRCA1/2* germline genetic testing was negative

What is the Family History?

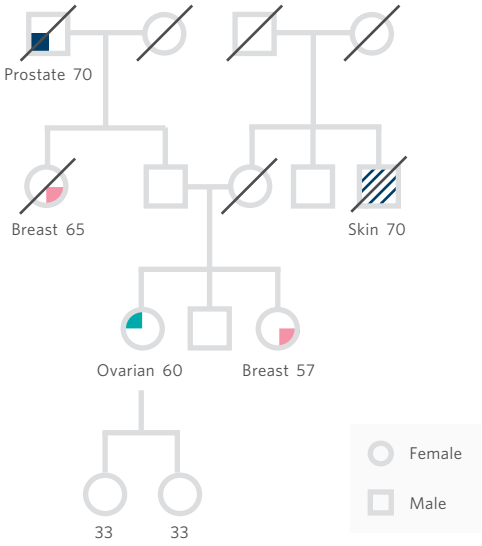
- Sister with breast cancer at 57

PATERNAL FAMILY HISTORY

- Aunt with breast cancer at 65
- Grandfather with prostate cancer at 50

MATERNAL FAMILY HISTORY

- Uncle with skin cancer at 70



What Happened with Genetic Testing?

1. Initial provider ordered *BRCA1/2* testing only which was negative.
2. A second healthcare provider ordered **Ambry's OvaNext, a 25 gene hereditary gynecologic cancer panel**

Genetic Testing Criteria:

- Patient meets NCCN® genetic testing criteria for *BRCA1/2*[1]

Genetic Test Results:

- Positive finding on OvaNext
- *RAD51D* pathogenic (disease-causing) variant
- Consistent with a diagnosis of hereditary ovarian cancer

HOW DID GENETIC TESTING IMPACT THE PATIENT AND FAMILY?

INCREASED LIFETIME CANCER RISKS
(see reverse for details)

- Ovarian
- Breast
- Prostate (male relatives)

PERSONALIZED SCREENING AND PREVENTION OPTIONS

- Consider TAH/BSO (post-childbearing)

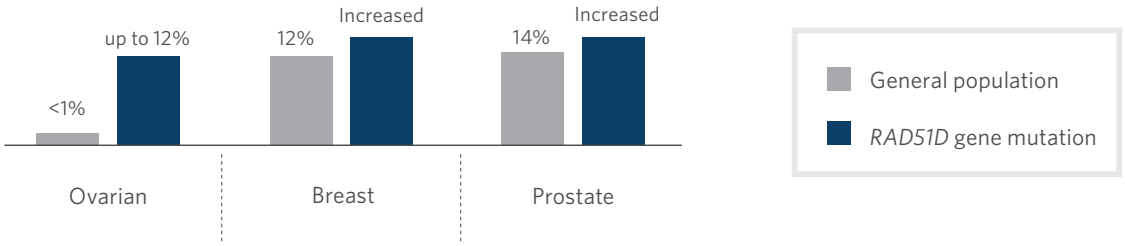
IMPACT FOR THE PATIENT'S FAMILY

- Siblings and children have a 50% chance of having the same *RAD51D* mutation and should be tested for the familial mutation
- Need to determine which side of the family is at-risk, so other relatives, such as aunts/uncles and cousins, can be tested for the familial mutation

What is *RAD51D*?

RAD51D is a gene which is critical for DNA damage repair because of its role in a process called homologous recombination. Germline (hereditary) mutations in this gene have been associated with an increased lifetime risk for ovarian cancer, as well as breast and prostate cancer. Ongoing research is continuing to further our understanding of the cancer risks for individuals with a *RAD51D* mutation.

Cancer Risks



POINTS FOR YOUR PRACTICE

- Negative results from germline *BRCA1/2* testing do not completely rule out hereditary cancer for a patient with ovarian cancer
- Comprehensive multigene panel testing that includes *BRCA1/2,* as well as other ovarian cancer genes such as *BRIP1*, *RAD51C*, and *RAD51D* should be considered for a patient with a personal and/or family history of ovarian cancer
- Identifying if a patient has hereditary ovarian cancer allows informed, personalized healthcare decisions for that patient and her family members.

References

1. National Comprehensive Cancer Network®. NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®). Genetic/Familial High-Risk Assessment: Breast and Ovarian. Version 2.2017

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