Ambry Genetics®
A Konica Minolta Company

# Identifying Patients with Hereditary Breast Cancer

## CASE EXAMPLE

## Who is the Patient?

- 48 year old female
- Breast cancer diagnosed at 48
- Previous *BRCA1/2* germline genetic testing was negative

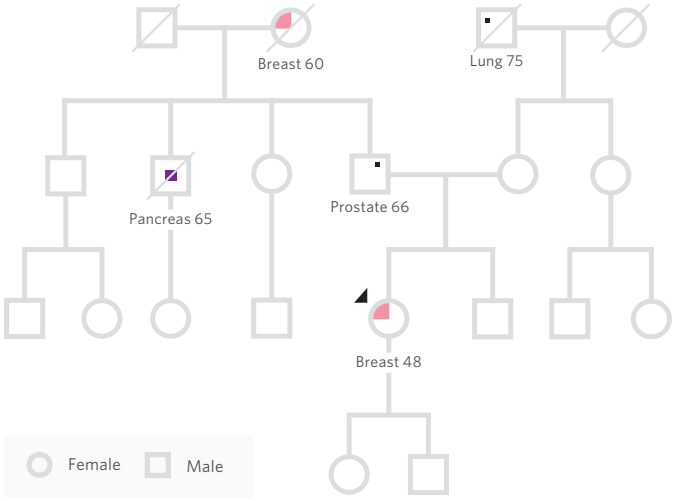
## What is the Family History?

### PATERNAL FAMILY HISTORY

- Father with prostate cancer at 66
- Uncle with pancreatic cancer at 65
- Grandmother with breast cancer at 60

### MATERNAL FAMILY HISTORY

- Grandfather with lung cancer at 75




## What Happened with Genetic Testing?

1. Initial provider ordered *BRCA1/2* testing only which was negative.
2. A second healthcare provider ordered **Ambry's BreastNext, a 17 gene hereditary breast cancer panel.**

### Genetic Testing Criteria:

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

### Genetic Test Results:

- Positive finding on BreastNext
- *ATM* pathogenic (disease-causing) variant
- Consistent with a diagnosis of hereditary breast cancer

## HOW DID GENETIC TESTING IMPACT THE PATIENT AND FAMILY?

### INCREASED LIFETIME CANCER RISKS
(see reverse for details)

- Breast (patient and female relatives)
- Pancreas
- Prostate (male relatives)

### PERSONALIZED SCREENING AND PREVENTION OPTIONS

- Annual mammogram
- Consider annual breast MRI
- Consider risk-reducing mastectomy based on family history

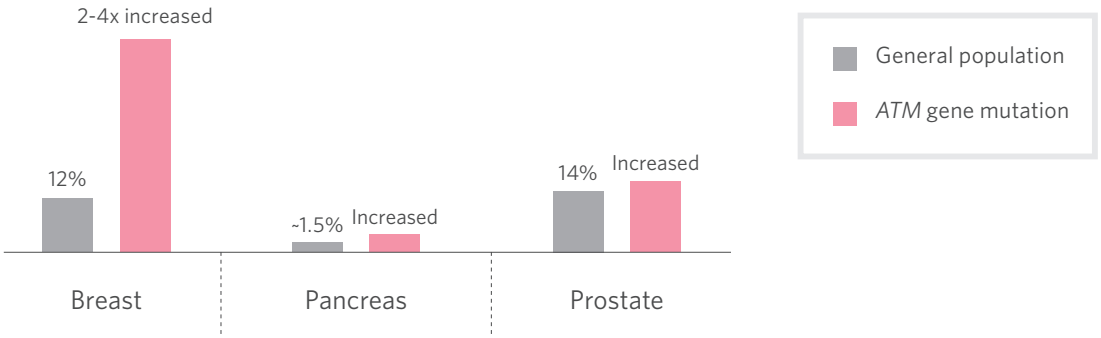
### IMPACT FOR THE PATIENT'S FAMILY

- Siblings and children have a 50% chance of having the same *ATM* 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 *ATM*?

*ATM* 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 breast cancer, as well as pancreatic and prostate cancer. Ongoing research is continuing to further our understanding of the cancer risks for individuals with an *ATM* 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 breast cancer
- Comprehensive multigene panel testing that includes *BRCA1/2*, as well as other breast cancer genes, should be considered for a patient with a personal and/or family history of breast cancer
- Identifying if a patient has hereditary breast cancer allows informed, personalized healthcare decisions for that patient and his or 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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