IMPACT OF HEREDITARY MULTI-GENE PANEL TESTING FOR CANCER SURVIVORS

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BACKGROUND

- Multi-gene panel testing evaluates up to 43 genes for pathogenic mutations linked to increased risk for breast, ovarian and other cancers.
- Around one-half of pathogenic mutations occur in cancer-related genes that are not BRCA1/2.
- Advances in the capability and efficiency of genetic testing have important implications for cancer survivors and their families.

OBJECTIVES

- The purpose of this study is to evaluate the impact of reflex genetic testing with newer multi-gene panels on patients with prior negative BRCA1/2 tests.

METHODS

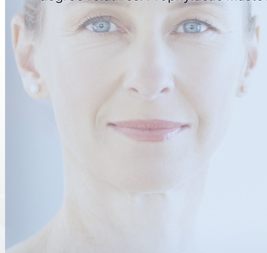
- Data was retrospectively collected from 914 patients who underwent a multi-gene panel test through Ambry Genetics at three Breastlink sites in Orange County, CA from August 2013 to June 2015.
- Patients were included in the study if they previously tested negative for BRCA1/2 and had a personal history of breast or ovarian cancer.
- 71.2% of patients received no prior genetic testing; 28.8% previously tested negative for mutations in BRCA1/2.

CASE STUDY

PREVENTING A SECOND INVASIVE CANCER

ONE PATIENT INCLUDED IN THE STUDY TESTED POSITIVE FOR PTEN AT 63, 19 YEARS AFTER HER INITIAL DIAGNOSIS AND PREVIOUS NEGATIVE BRCA1/2 TESTING.

PTEN is associated with increased risk for multiple cancers. Breast cancer risk estimates range from 67% to 85% in women with pathogenic PTEN mutations. Increased screening is recommended for women with a pathogenic PTEN mutation and predictive testing for first-degree relatives. Prophylactic mastectomy may also be recommended.



INITIAL DIAGNOSIS:	AGE 44
PREVIOUS TREATMENT:	LUMPECTOMY, RADIATION, CHEMOTHERAPY
PREVIOUS BRCA1/2 RESULT:	NEGATIVE
REFLEX PANEL TESTING:	PTEN POSITIVE, AGE 63

After identifying her PTEN mutation, she underwent prophylactic mastectomy and was found to have occult DCIS.

REFLEX GENE TESTING IN CANCER SURVIVORS CAN PREVENT SECOND CANCERS.

RESULTS

- Of 187 patients who met study-inclusion criteria, 10 patients (5.3%) were found to carry 11 pathogenic mutations.
- Pathogenic mutations detected included CHEK2, PTEN, BARD1, NF1, and RAD51C.
- One patient tested positive for two pathogenic mutations: CHEK2 and BARD1.
- Positive multi-gene panel testing results affected cancer-screening decisions, including the addition of breast MRI, colonoscopy and thyroid ultrasound.

CHANGE IN MANAGEMENT FOR CANCER SURVIVORS WHO HAD POSITIVE REFLEX GENETIC TESTING

PATIENT	MUTATION DETECTED	CANCER HISTORY	AGE AT FIRST CANCER Dx	CHANGE IN MANAGEMENT			
				FAMILY COUNSELING & TESTING	BREAST MRI	CONSIDER RISK REDUCING MASTECTOMY	EARLY COLONOSCOPY
A	CHEK2	BREAST	43	✓	✓	✗	✗
B	CHEK2	BREAST	43	✓	✓	✗	✗
C	CHEK2	BREAST	43	✓	✓	✗	✗
D	CHEK2	BREAST	65	✓	✓/✗	✗	✗
E	CHEK2	BREAST/THYROID	35	✓	✓	✗	✗
F	CHEK2/BARD1	BREAST	40	✓	✓	✗	✗
G	PTEN	BREAST	36	✓	✓	✓	✓
H	PTEN	BREAST	43	✓	✓	✓	✓
I	NF1	BREAST	35	✓	✓	✗	✗
J	RAD51C	BREAST/CARCINOID	64	✓	✓/✗	✗	✗

*For patients with PTEN mutations, annual thyroid ultrasound, renal ultrasound, and endometrial surveillance should also be considered.

CONCLUSIONS

- Cancer survivors can benefit from advancements in technology and understanding of cancer even years after treatment has ended.
- Multi-gene panel testing can guide screening recommendations in cancer survivors as well as their families.