breastlink
A network of comprehensive breast health centers
Orange Laguna Hills Newport Beach Murietta Encino

MULTI-GENE PANEL TESTING
AND THE CANCERS IDENTIFIED IN PATIENTS AT RISK FOR HEREDITARY BREAST CANCER

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BACKGROUND

- Multi-gene panel testing evaluates up to 43 genes for pathogenic mutations that increase risk for breast, ovarian, and other hereditary cancers.
- There is limited information available on the type of breast and other cancers that develop with non-BRCA1/2 mutations.
- Understanding cancer biology will help to identify optimal screening, prevention, and treatment algorithms for patients with a genetic cancer predisposition.

OBJECTIVES

- To evaluate the utility of multi-gene panel testing in a comprehensive breast health practice.
- To explore the relationship between pathogenic gene mutations and the biology of breast cancers.

METHODS

- Data was retrospectively collected from 500 patients who received multi-gene panel testing at 2 of 3 Breastlink sites in Orange and Laguna Hills, CA between July 2013 and September 2014.
- All patients met genetic testing criteria per NCCN guidelines; all patients had personal or family history of breast and/or ovarian cancer.
- Patients underwent pre- and post-test counseling and tests were ordered by a supervising Breast Surgical Oncologist.
- Patients underwent a panel test for at least 5 and up to 43 cancer-related genes.

PATHOGENIC MUTATIONS *n*=33*

MUTATIONS *n*=33

ATM	**2**	6.1%
BRCA1	**2**	6.1%
BRCA2	**10**	30.3%
CHEK 2	**7**	21.2%
MUTYH	**3**	9.1%
PALB2	**3**	9.1%
OTHER (BARD1, MRE11A, MSH2, NBN, RAD51C)	**6**	18.2%

TYPE OF BREAST CANCER *n*=25

INVASIVE DUCTAL CARCINOMA	**15**	60.0%
ER+PR+HER2-	11/15	73.3%
HER2+	2/15	13.3%
ER-PR-HER2-	2/15	13.3%
INVASIVE LOBULAR CARCINOMA	**3**	12.0%
ER+PR+HER2-	3/3	100.0%
DUCTAL CARCINOMA IN SITU	**6**	24.0%
INFORMATION UNAVAILABLE	**1**	4.0%

PERSONAL HISTORY OF CANCER *n*=29

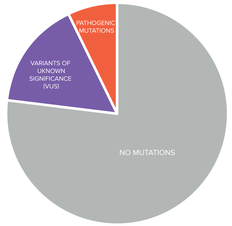
BREAST CANCER	**25**	86.2%
OVARIAN CANCER	**3**	10.3%
THYROID CANCER	**1**	3.4%

* One patient tested positive for more than 1 pathogenic mutation. Multiple patients tested positive for VUS or pathogenic mutation and VUS in separate genes."

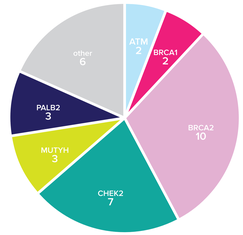
RESULTS

- Of 500 patients, 6.4 % (32) tested positive for at least 1 pathogenic mutation.
- 16.2% carried at least 1 variant of uncertain significance (VUS).
- A majority of patients (79%) did not carry a mutation or VUS.
- Most patients with pathogenic mutations were diagnosed with ER/PR-positive invasive ductal carcinoma.
- Compared to patients with BRCA1/2 mutations, patients with non-BRCA mutations were more likely to have a family history of non-breast or ovarian cancer (58.3% vs 90%, respectively, p=0.0735)

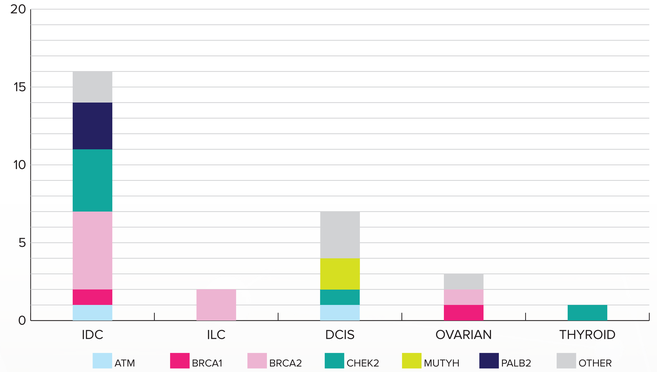
MULTIGENE PANEL TEST RESULTS



PATHOGENIC MUTATIONS



CANCER TYPE & PATHOGENIC MUTATION



CONCLUSIONS

- Multi-gene panel testing identifies more patients at risk for hereditary breast cancer than BRCA1/2 testing alone.
- Most patients with breast cancer and a pathogenic mutation will have Luminal-type Invasive Ductal Carcinoma.
- Breast surgeons and oncologists at the forefront of treatment are ideally situated to initiate discussions about multi-gene panel testing and potential outcomes