A Study of Ovarian Cancer Patients Tested With a 25-Gene Panel of Hereditary Cancer Genes

Lucy R. Langer, MD, Heidi McCoy, MS, Kelsey Moyes, MStat, Jennifer Saam, PhD, Brian Abbott, MD, Larry J. Geier, MD

Compass Oncology, Portland, OR
Myriad Genetic Laboratories, Inc., Salt Lake City, UT
Kansas City Cancer Center, Kansas City, MO

Presented at ASCO- June 2014
## Background

- Patients with epithelial ovarian cancer have a relatively high rate of a hereditary basis for their cancer (11-15%).
- Identifying patients with hereditary cancer provides an opportunity to:
  - Prevent 2nd cancer
  - Notify family members to cancer risk
  - Patients to enroll in clinical trials for new treatments
- All patients with epithelial ovarian cancer meet NCCN guidelines for BRCA1 and BRCA2 testing (Risch et al. AJHG 2001).
- Patients with ovarian cancer and personal or family history of colon and/or endometrial cancer may also meet guidelines for Lynch syndrome testing.
- With next-generation sequencing, patients receiving hereditary cancer testing can be tested for more genes using a multi-gene panel approach.

## Methods

- We queried a commercial laboratory database for patients affected with ovarian cancer (including fallopian tube and peritoneal cancer).
- All patient data regarding clinical history was obtained by health care provider report on test requisition forms.
- Analysis includes 648 patients (September 4, 2013 - April 17, 2014).
- Panel based on next generation sequencing and rearrangement analysis of 25 genes with cancer risk data: BRCA1, BRCA2, MLH1, MSH2, MSH6, PMS2, EPCAM, APC, MUTYH, CDKN2A, CDK4, PALB2, CHEK2, SMAD4, BMPR1A, STK11, TP53, CDH1, PTEN, ATM, NBN, BARD1, BRIP1, RAD51C, and RAD51D.
- Panel limited to genes with strong evidence of cancer association.

## Results

- Ovarian cancer patients represented 7% of the panel testing population.

## Conclusions

- 15.4% of patients with ovarian cancer had a deleterious mutation on a 25 gene hereditary cancer panel.
- 59.6% in BRCA1 and BRCA2
- 5.8% in the Lynch syndrome genes
- 34.6% in other hereditary cancer genes
- ATM and BRIP1 were the most common genes in this category.
- Panel testing led to a 63% increase in the identification of deleterious mutations over BRCA1 and BRCA2 testing alone.
- Panel testing in this series led to the identification of mutations in genes that would not otherwise be suspected by clinical or family history alone.
A Study of Ovarian Cancer Patients Tested With a 25-Gene Panel of Hereditary Cancer Genes