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Ambry Genetics presents groundbreaking TP53 gene data at ASHG; TP53 linked to breast cancer

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Ambry Genetics (Ambry), a leader in clinical genetic diagnostics and genetics software solutions, has announced new data affirming the effectiveness of multi-gene panel testing (MGPT) in identifying *TP53* gene mutations in people who might otherwise not have been tested. The findings, presented as a poster at the American Society of Human Genetics (ASHG) 2015 Annual Meeting in Baltimore, includes data collected from more than 25,000 individuals having *TP53* testing, of which 187 were positive. The tested cohort is the largest to date from a single testing laboratory.

Germline mutations in the *TP53* gene are associated with Li-Fraumeni syndrome (LFS), a rare inherited cancer predisposition syndrome that significantly increases a person's risk of developing breast cancer and many other types of cancer. LFS affects between one in 5,000 and one in 20,000 people. People with LFS have up to a 50% risk of developing cancer by age 30, and up to a 93% chance of developing cancer in their lifetime. Breast cancer is the most common cancer diagnosed in women with a *TP53* gene mutation. Most individuals with LFS inherited the *TP53* mutation from a parent, though an estimated 7-20% are the first in their family to have a *TP53* gene mutation.

"These findings suggest that we have been identifying only the most clinically affected LFS families, and raises the concern that we have overestimated cancer risks for them," said Judy Garber, MD, MPH, Director, Center for Cancer Genetics and Prevention, Dana Farber Cancer Institute in Boston, and one of the study's authors. "The findings make the collection and analysis of unselected data more important than ever, and the kind of data that panels can provide essential to that work."

For the study, researchers reviewed data from 25,182 patients that underwent *TP53* testing conducted at Ambry. Among those positive for a *TP53* mutation, personal and family cancer histories were examined to identify specific patterns and to determine whether any National Comprehensive Cancer Network (NCCN) testing criteria were met, including Classic criteria, Chompret criteria, and breast cancer diagnosis before age 36 years.

In total, 187 patients (0.74%) tested positive for *TP53* mutations. These results came from single gene testing (118/2956, 3.99%) and from MGPT (69/22,226, 0.31%). Of all those tested, 95% who underwent single gene testing (SGT) had a cancer diagnosis, versus 82% of patients who had MGPT.

Among individuals with *TP53* mutations for whom family history data were available, 73% (74/102; 95% CI 63%-81%) in the SGT group and 30% (20/66; 95% CI 19%-47%) in the MGPT group met Classic or Chompret criteria for LFS (p=0.0000001). Adding in the cases meeting the criterion of breast cancer before age 36, the researchers estimated that 85% of patients in the SGT group but only 53% in the MGPT group met NCCN testing criteria.

"Ambry has amassed a large quantity of data from multi-gene panels and we are using and sharing that information to uncover vital insights for physicians and genetic counselors to better guide their patients," said Jill S. Dolinsky, MS, CGC, Senior Manager of Clinical Research at Ambry. "Our research has shown that *TP53* mutations are more common than we thought, and affected individuals don't always meet established criteria for testing. Along with single gene analysis, *TP53* testing is available on all appropriate hereditary cancer panels at Ambry, and frequently benefits people with a suspected cancer predisposition syndrome, conflicting pathology, or missing family history."

ABOUT AMBRY GENETICS®

Ambry Genetics is both College of American Pathologists (CAP)-accredited and Clinical Laboratory Improvement Amendments (CLIA)-certified. Ambry leads in clinical genetic diagnostics and genetics software solutions, combining both to offer the most comprehensive testing menu in the industry. Ambry has established a reputation for sharing data while safeguarding patient privacy, unparalleled service, and responsibly applying new technologies to the clinical molecular diagnostics market. For more information about Ambry Genetics, visit www.ambrygen.com