For immediate Release:

Research from Ambry Genetics & Mayo Clinic Reveals Differences in the Prevalence of Breast Cancer Genes among Racial and Ethnic Populations

Abstract to be presented at the 2019 American Society of Clinical Oncologists (ASCO) Annual Meeting

(Aliso Viejo, CA) May 30, 2019: Researchers from Ambry Genetics (Ambry), a leading clinical genetics testing company, and Mayo Clinic, have documented significant differences in the prevalence of hereditary genetic mutations, also called germline mutations, linked to breast cancer among racial and ethnic populations compared to non-Hispanic White women. Results will be presented at the 2019 American Society of Clinical Oncology (ASCO) annual meeting to be held May 31 to June 4, 2019 in Chicago, IL.

“The prevalence of germline mutations and associated germline genetic drivers of breast cancer risk in racial and ethnic populations is largely unknown, and much of the known risk that is cited in the literature has been studied in non-Hispanic White populations,” stated Dr. Siddhartha Yadav, Hematology/Oncology Fellow at the Mayo Clinic working in the lab of Fergus Couch, PhD, Zbigniew and Anna M. Scheller Professor of Medical Research Chair, Division of Experimental Pathology and Laboratory Medicine Mayo Clinic. “That’s a clinical ‘blind spot’ that needs to be addressed because a better understanding of racial or ethnicity-specific genetic risk for breast cancer is key to improved targeting of genetic testing and more informed management of patients in these populations.”

In a collaborative study between Mayo Clinical and Ambry Genetics conducted from March 2012 through December 2016, a population of 77,900 women with breast cancer, comprised of 57,003 non-Hispanic White, 6,722 Black, 4,183 Asian, 5,194 Hispanic and 4,798 Ashkenazi-Jewish subjects, underwent germline multigene panel testing of cancer genes. The prevalence of gene mutations in racial and ethnic populations relative to non-Hispanic Whites was assessed.

Results revealed numerous differences between populations that may have important clinical implications. Among them: the frequency of pathogenic mutations in known breast cancer genes was 7.5% for Asians and Ashkenazi-Jews and 8.7% for non-Hispanic Whites, compared to 9.7% for Blacks and 9.9% for Hispanics. Distribution of variants of uncertain significance (VUS), a variant classification indicating unknown relevance to disease, also differed, being most frequent among Asians (29%), followed by Blacks (27%), Hispanics (21%), non-Hispanic Whites (16%) and Ashkenazi-Jews (14%). However, across all ethnicities and races, mutations in BRCA1, BRCA2, and PALB2 were significantly associated with a four-times higher risk of breast cancer.

Brigette Tippin Davis, Ph.D., FACMG, senior vice president of research and development of Ambry Genetics made this observation: “As we pursue further research in this area, the now documented differences in the prevalence of mutations in breast cancer genes among major racial and ethnic population in the U.S. underscores two issues: the importance of considering all ethnicities when developing guidelines and best practices for genetic testing, screening and management of patients predisposed to breast cancer; and a need for larger study cohorts in ethnic minority populations.”

Presentation Information: Abstract #1514
Session: Cancer Prevention, Hereditary Genetics, and Epidemiology
Poster Session: Monday June 3, 1:15 PM to 4:15 PM, Location: Hall A
Poster Discussion Session: Monday June 3, 4:30 PM to 6:00 PM, Location: S103
About Ambry Genetics

Ambry Genetics, as part of Konica Minolta Precision Medicine, excels at translating scientific research into clinically actionable test results based upon a deep understanding of the human genome and the biology behind genetic disease. Our unparalleled track record of discoveries over 20 years, and growing database that continues to expand in collaboration with academic, corporate and pharmaceutical partners, means we are first to market with innovative products and comprehensive analysis that enable clinicians to confidently inform patient health decisions. We care about what happens to real people, their families, and the people they love, and remain dedicated to providing them and their clinicians with deeper knowledge and fresh insights, so together they can make informed, potentially life-altering healthcare decisions. For more information, please visit ambrygen.com.

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