Self-Reported Race vs. Genetic Ancestry and the Risk of Breast Cancer

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African American (AA) women experience lower overall incidence of breast cancer, but higher mortality rates from this disease, relative to European American women. Although it is unclear to what extent these differences are attributable to genetic or environmental factors, AA women are more likely to be diagnosed with aggressive tumors, including a larger percentage of estrogen receptor negative (ER-)/progesterone receptor negative (PR-) tumors, tumors with more advanced stage, and higher grade tumors that are more likely to spread.

Previous studies have reported a relatively high proportion of particularly aggressive triple negative tumors (so called because they do not produce significant amounts estrogen receptor, progesterone receptor or epidermal growth factor receptor proteins) among West African women, the ancestral population for many AA women. This suggests a possible genetic component to breast cancer mortality, yet few studies have evaluated whether genetic admixture predisposes women to certain types of breast cancer.

To address this question, Drs. Kerryn Reding, Chris Carlson and Kathleen Malone of the Public Health Sciences Division, all contributed to a recent analysis to determine whether percent African ancestry was associated with breast cancer tumor characteristics in American-born women. They studied women from five distinct U.S. geographic regions who self-reported as AA in a population-based case-control study of invasive breast cancer. Percent African ancestry was determined using 128 ancestry informative markers, a subset of genetic markers that differ in allele frequencies across different populations of the world.

Based on self-reported race, AA women had a 30% lower risk of ER+/PR+ breast cancer (Odds Ratio (OR): 0.7, 95% Confidence Interval (CI): 0.6-0.99), a non-significant higher risk of ER-/PR- breast cancer (OR: 1.3, 95% CI: 1.0-1.8), a 1.3-fold higher risk of regional/distant rather than localized disease (95% CI: 1.0-1.8), and 1.6-fold increased risk of poor/undifferentiated tumors (95% CI: 1.2-2.1) compared to self-reported White women. In their study, the authors adjusted these statistical differences for a range of potential confounders, including: age at diagnosis, geographic study site, family history of breast cancer, age at first live birth, parity, menopausal status, use of menopausal hormones and body mass index.

In contrast, percent African ancestry based on ancestry informative markers was not significantly associated with breast cancer ER/PR subtypes, stage or grade among AA women (see figure). This lack of associations suggests that percent African ancestry does not further delineate within-race variation in breast cancer risk. However, the mean percent African ancestry in the sample of 863 AA women was 85.7%. Thus, it is possible that there was insufficient variability among the women to detect an effect.

Several studies have demonstrated racial differences in mammographic screening rates and other factors related to health care access and utilization. Ongoing research will be required to determine whether additional non-genetic characteristics associated with race including potential social, lifestyle, and economic factors, can help account for the observed racial patterns in aggressive breast cancer.

Reding KW, Carlson CS, Kahsai O, Chen CC, McDavid A, Doody DR, Chen C, Ornelas I, Lowe K, Bernstein L, Weiss L, McDonald JA, Simon MS, Strom B, Marchbanks PA, Burkman R, Spirtas R, Liff JM, Malone KE. (2012). Examination of ancestral informative markers and self-reported race with tumor characteristics of breast cancer among black and white women.*Breast Cancer Research Treatment*. Epub ahead of print, doi:10.1007/s10549-012-2009-0.

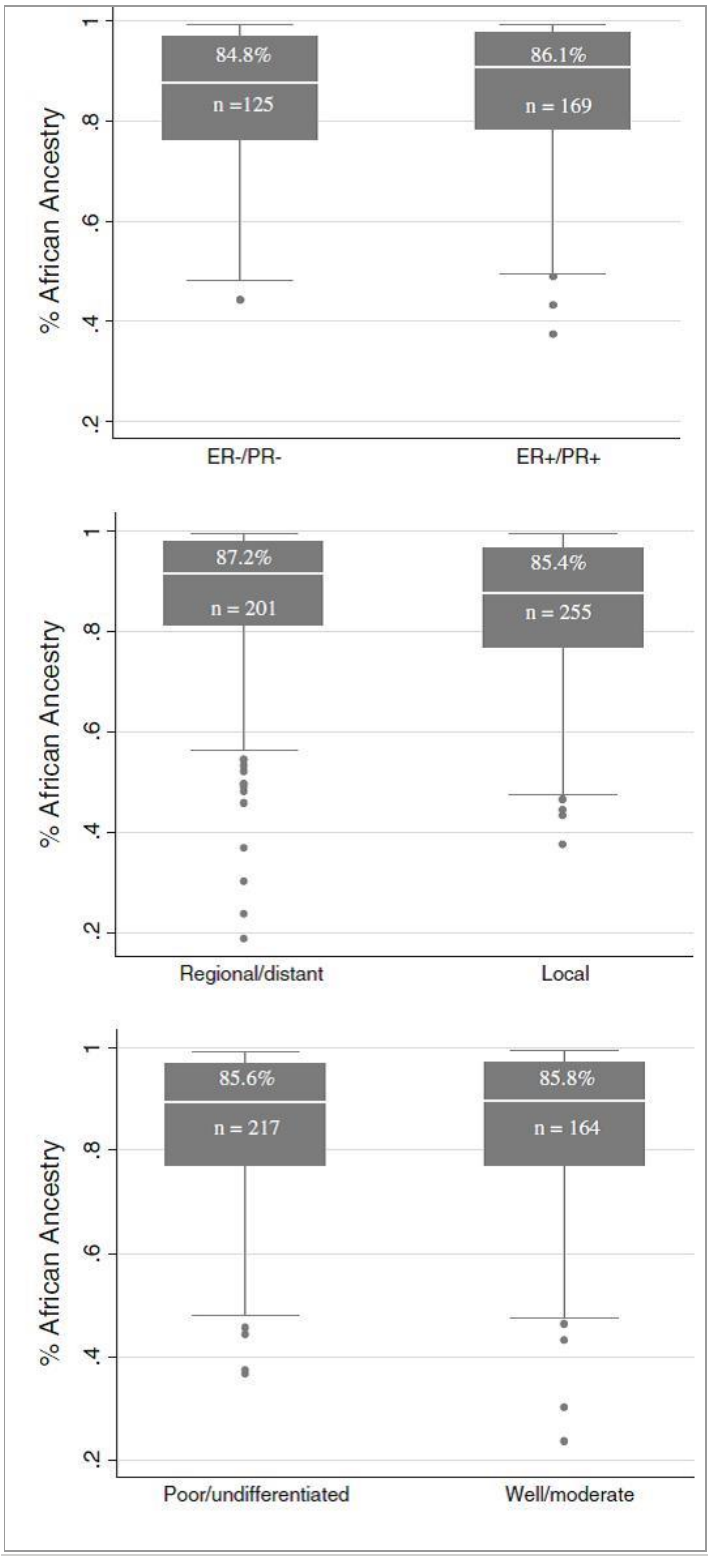

Image courtesy of author

Figure. Percent African ancestry among African American women by i) hormone receptor status, ii) stage, iii) grade. Boxplot showing the 50th, 25th, and 75th percentiles (middle, bottom, top lines of boxes, respectively) along with values outside the 95th percentile (dots).