

BACKGROUND

Breast cancer incidence has increased in recent years, with notable disparities across populations. Overall breast cancer incidence in the United States rose by approximately 1% per year from 2012-2021, while **incidence among Asian American and Pacific Islander (AAPI) women increased 2.7% annually.**¹ Among women under age 50, breast cancer **cases increased by nearly 50% in AAPI women**, comparable to trends in White women.² Despite rising rates, limited data exist on germline genetic testing outcomes in AAPI women with breast cancer.

AIM: To examine the rates and results of germline genetic testing in AAPI women diagnosed with breast cancer

METHODS

Cohort

A retrospective analysis was conducted of self-identified Asian American and Pacific Islander (AAPI) women with breast cancer identified from a non-consecutive dataset of patients undergoing multigene panel testing (MGPT) at a national clinical diagnostic laboratory (Ambry Genetics) between January 2016 and April 2025.

Analysis

Descriptive statistics were used to characterize the number of AAPI women undergoing testing, most frequent pathogenic/likely pathogenic (P/LP) variants, geographic residence (metropolitan vs rural, based on ZIP code), and insurance type. P/LP variant rates were compared between AAPI women, White women, and all non-AAPI women tested from the same dataset, stratified by age group (<40 vs <50 years and all ages). Differences in variant frequencies between groups were assessed using two-proportion z-tests.

TAKE HOME POINTS

- **AAPI women are underrepresented in genetic testing relative to their breast cancer incidence.**
- **Genetic testing among AAPI women with breast cancer is concentrated in metropolitan populations with private insurance.**
- **Testing disparities may disproportionately impact younger AAPI women (<40), who exhibited higher P/LP variant rates than older AAPI women.**

RESULTS

AAPI Women are Underrepresented in Genetic Testing Cohorts

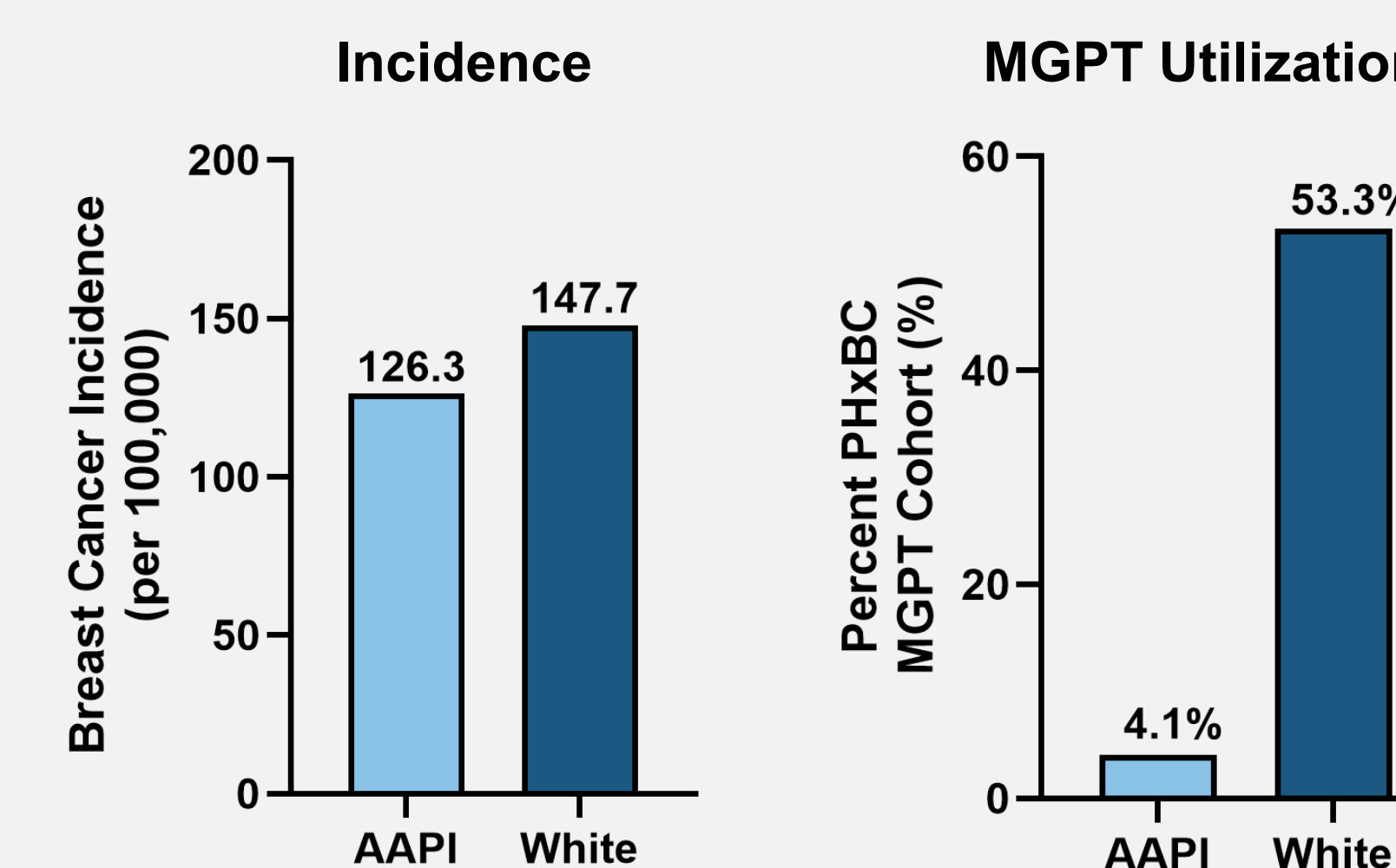
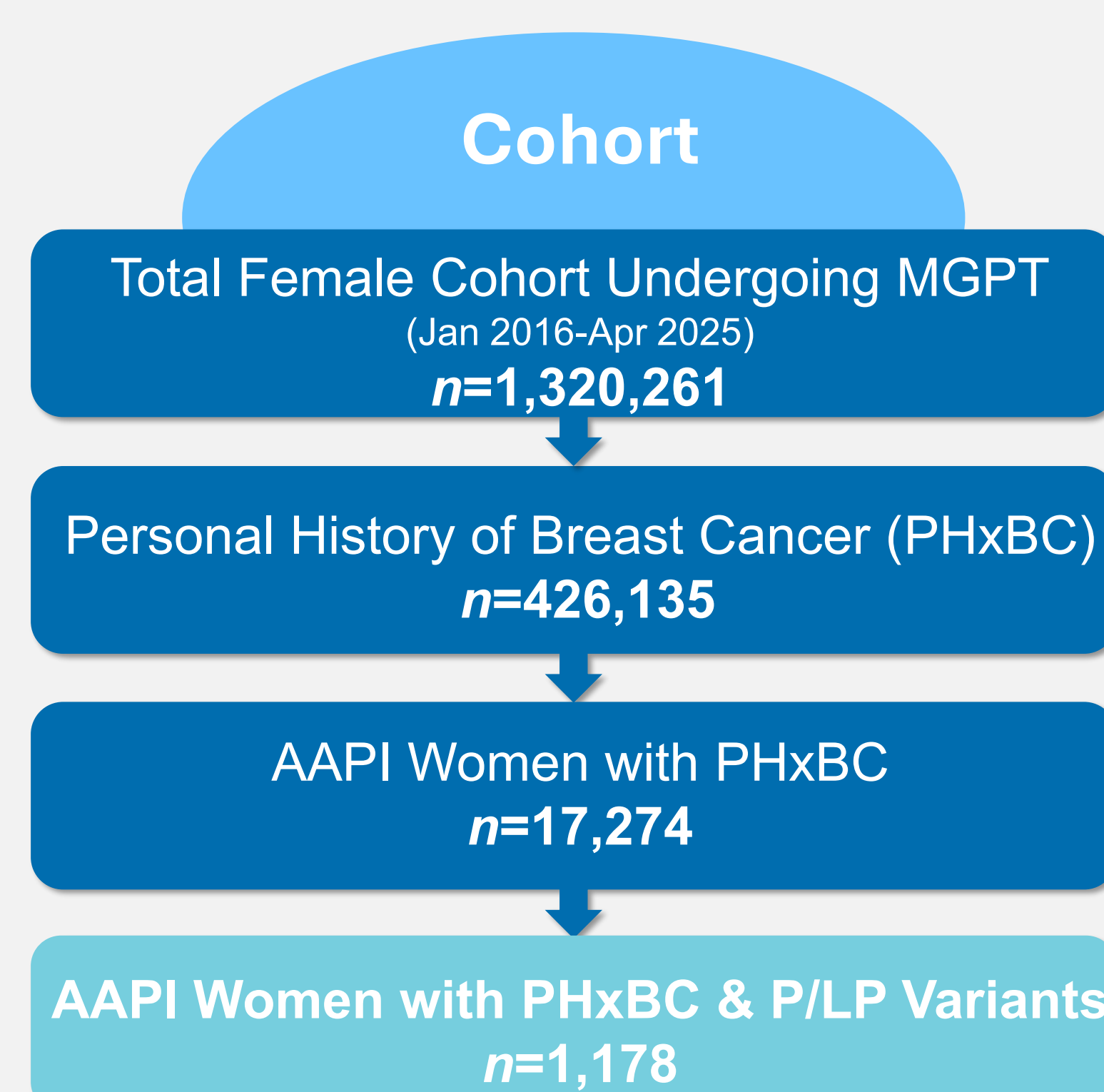


Figure 1. Lower Representation of AAPI Women in MGPT Cohorts. Breast cancer incidence rates among AAPI women approached those observed in White women (126.3 vs 147.7 per 100,000 (left panel));³ however, AAPI women comprised only 4.1% ($n=17,274/426,135$) of women with breast cancer undergoing MGPT compared with 53.3% ($n=227,224/426,135$) for White women (right panel).

Breast cancer incidence in AAPI women was **85%** that of White women BUT AAPI women had **~8-fold lower** MGPT-tested cancer cases

Germline P/LP Variant Rates by Ethnicity and Age

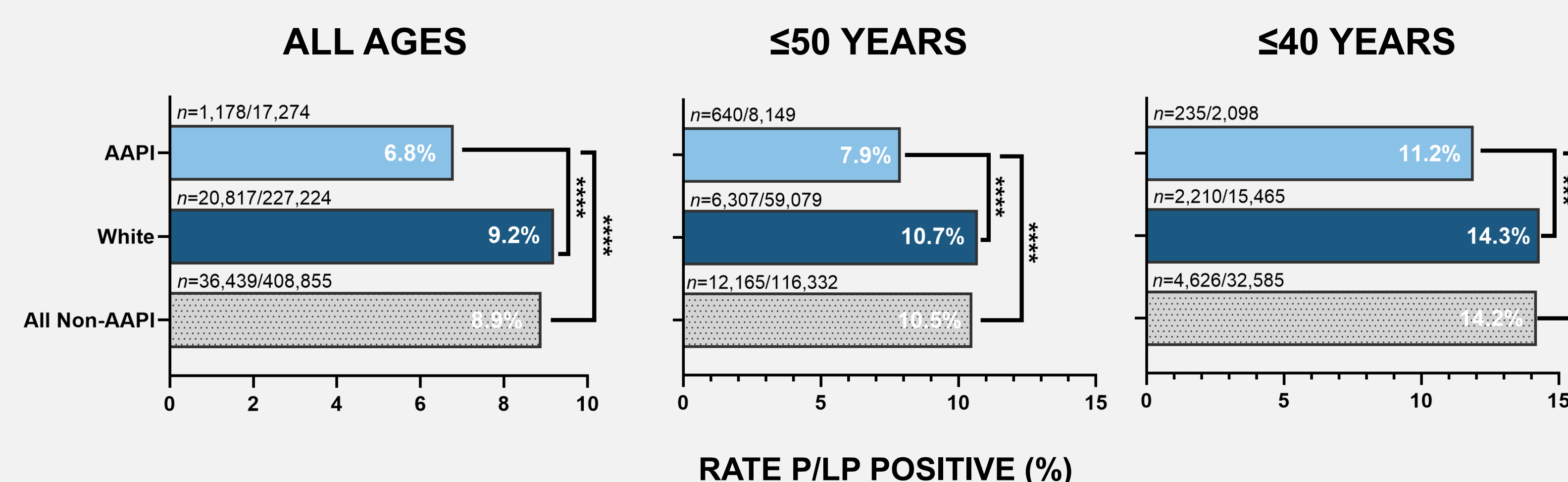


Figure 2. Proportion of Women with Germline P/LP variants by ethnicity and age. P/LP rates are shown for AAPI women, White women, and all non-AAPI women. Rates were calculated as the proportion of individuals with breast cancer and P/LP variants who underwent genetic testing within each subgroup. AAPI women <40 years had significantly higher P/LP variant rates compared with AAPI women ≥40 years ($p<0.0001$).

SES Characteristics of AAPI Cohort

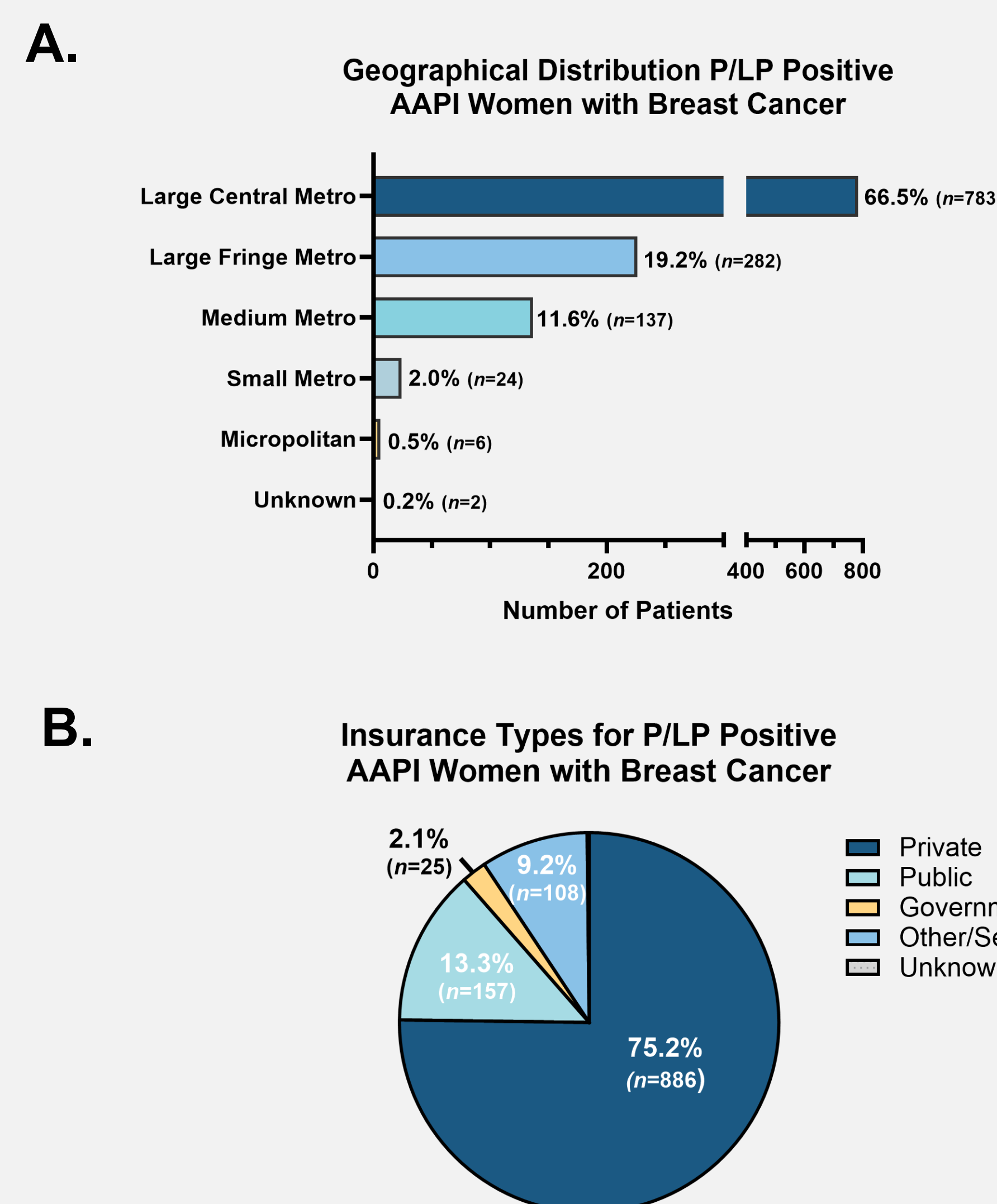


Figure 3. Geographical distribution and insurance type among P/LP positive AAPI women with breast cancer undergoing genetic testing. (A) The majority of AAPI women undergoing genetic testing resided in large central metropolitan areas. Classification based on NCI Urban-Rural Classification Scheme for Counties.⁴ (B) Private insurance was the predominant payor type for the AAPI cohort.

Gene & Variant Findings in AAPI Cohort

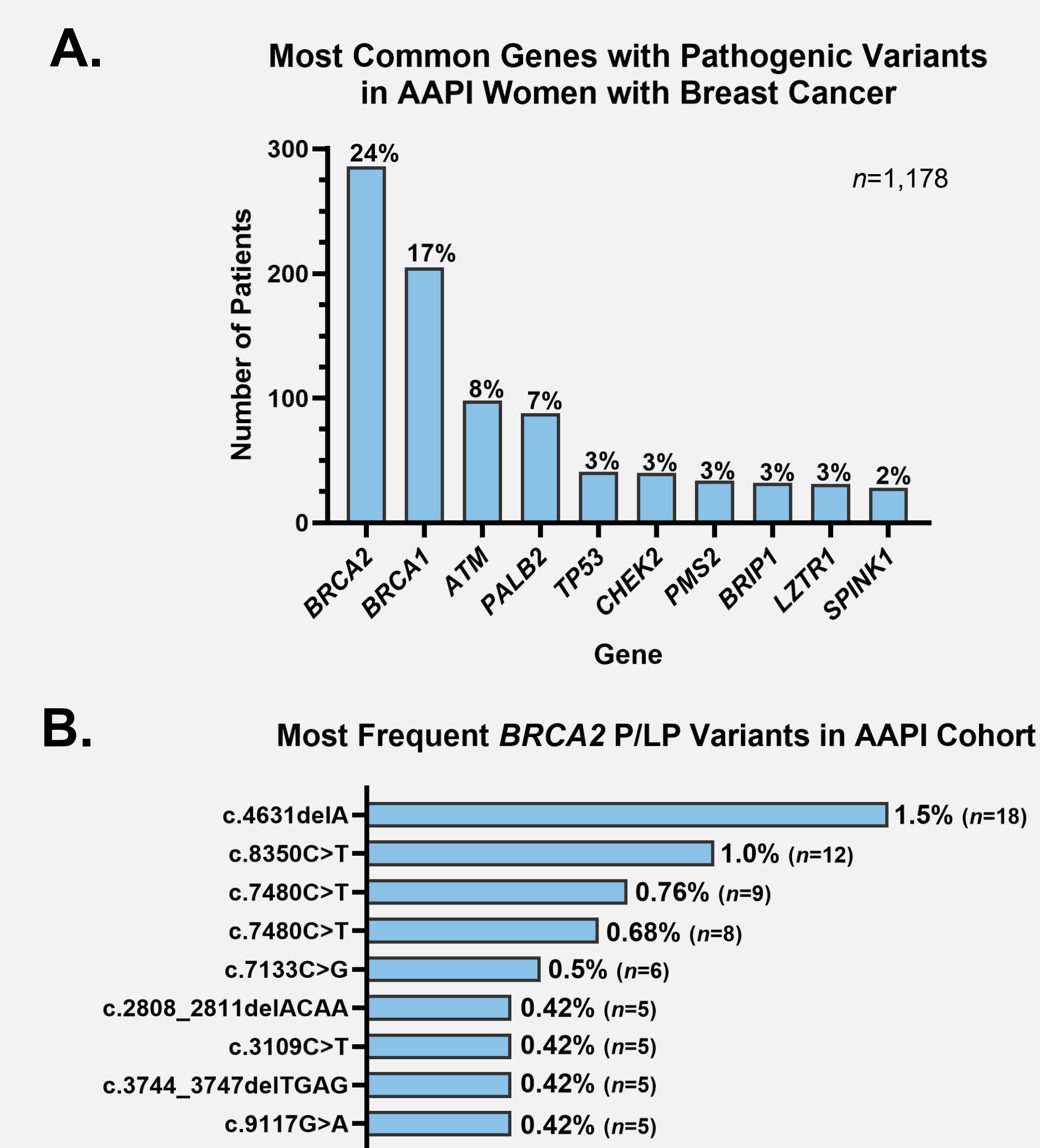


Figure 4. Most frequent germline genes with P/LP and recurrent variants in the AAPI cohort. (A) The most frequently observed genes with P/LP variants in AAPI women with breast cancer were *BRCA2*, *BRCA1*, and *ATM* (top 10 genes shown). (B) Several *BRCA2* P/LP variants were recurrently observed within the AAPI cohort. Lower rates of germline testing among AAPI women may underestimate the true population prevalence of hereditary cancer susceptibility variants.

3. <https://seer.cancer.gov/statfacts/html/breast.html>

4. <https://www.cdc.gov/nchs/data-analysis-tools/urban-rural.html>