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Ethnic Differences in Germline Genetic Testing For Breast Cancer

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Introduction: Ethnic variations in uptake of genetic testing and differences in findings of germline mutations within ethnic groups, are not well understood. The goal of this research is to assess for any such differences or similarities within a genetic counseling and testing program at an urban Cancer Center.

Methods: This is a non-comparative, descriptive epidemiology study assessing individuals with a diagnosis of breast cancer undergoing genetic counseling at the TJUH Sidney Kimmel Cancer Center in Philadelphia between 2014 and 2019. Data were compiled onto Research Electronic Data Capture (REDCAP) and analyzed statistically.

Results: Patients with Breast Cancer (n=1075) were included in the analysis, 807 of whom had genetic testing conducted. In total, 81 Caucasians had pathogenic/likely pathogenic mutations (13%) and 16% had VUS. African Americans had the highest prevalence of VUS (32%) and 16 pathogenic/likely pathogenic mutations (13%). Asians (n=44) had no pathogenic but 2 likely pathogenic mutations (6%), and 3 VUS (9%). Comparatively, no statistically significant differences were observed. Asians presented for genetic counseling younger (mean age 49) than African Americans (mean age 54) and Caucasians (mean age 58) ($p<0.001$). Caucasians were more likely to undergo genetic testing (89%) than African Americans (78%) and Asians (79%) ($p<0.002$).

Discussion: These results point toward ethnic differences in utilization and followthrough with genetic testing, as well as variations in genetic mutations. The high prevalence of VUS

mutations in African Americans and few mutations in Asians suggests that the mutational spectrum in these populations is not well understood and warrants further study.