

World Congress on

Human Genetics

November 07- 08, 2016 Barcelona, Spain

Prevalence of the four common *BRCA1* and *BRCA2* founder mutations in Colombian breast patients of African descent

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In Colombia breast cancer is the first most common cancer among women. The contribution of *BRCA1/2* (BRCA) germline mutations to hereditary breast/ovarian cancer has been poorly investigated in minority populations, such as African-Americans. The large size of the BRCA genes and the diversity of the mutations described across the world make genetic testing using DNA sequencing analysis expensive and time-consuming. It has been observed that in some ethnic populations, a limited number of common mutations account for the majority of familial cases of breast and ovarian cancer. In Colombia, Torres and colleagues identified three common founder mutations in BRCA (*BRCA1*: 3450delCAAG and A1708E; *BRCA2*: 3036delACAA) in white breast/ovarian cancer families that accounted for approximately 80% of all BRCA mutations identified in this cohort. Recently, an additional small range founder mutation was identified in *BRCA2*. To assess the contribution of the four BRCA founder mutations previously identified in white Colombian breast cancer patients, 59 families of African descent were screening using PCR-based restriction-fragment-length polymorphism (RFLP) analysis and RT-PCR. One founder mutation (*BRCA1*: A1708E) was identified 59 breast cancer families (1.7%). These findings imply that other mutations may occur in these patients, which need to be identified in a future study. However, given that our results are based on a relatively small number of breast cancer patients, they have to be considered preliminary. This research project was supported by AstraZeneca Colombia and Diana Torres by a postdoctoral fellowship from the Alexander von Humboldt Foundation.

Biography

Elizabeth Vargas has completed her Master's degree in Biological Sciences at the Pontificia Universidad Javeriana. She is currently working at the Faculty of Natural Sciences and Mathematics at the Universidad del Rosario and has been involved as a Researcher in several projects in cell and molecular biology.

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