Genomic Characterization and Genetic Testing Gaps in Early-Onset Breast Cancer in Colombia

M. A. Bravo, D. C. Sotelo-Rodríguez, J. Caicedo, S. Cervera, A. Ruiz-Patiño, W. A. Mantilla, S. X. Franco;
Breast Cancer Unit, Fundación Centro de Tratamiento e Investigación sobre Cáncer(CTIC), Bogota, COLOMBIA.

SAN ANTONIO BREAST CANCER SYMPOSIUM
DECEMBER 10–13, 2024
SABCS.ORG

Abstract

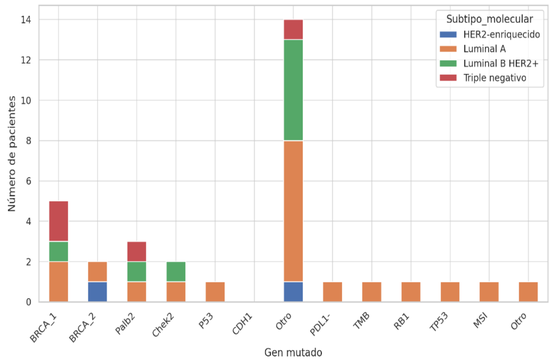
Germline pathogenic variants are frequent in early-onset breast cancer patients in Colombia, with distinct patterns across tumor subtypes. BRCA1 predominates in triple-negative cases, while PALB2 and CHEK2 are more common in hormone receptor-positive tumors. Notably, 24.1% of patients carried alterations in non-canonical genes, reflecting marked genomic heterogeneity. Mutation carriers were diagnosed at a significantly younger age compared to non-carriers, emphasizing the importance of early and comprehensive genetic testing in all young patients, irrespective of tumor subtype. These findings support the implementation of broad germline testing to guide personalized care and familial risk assessment in this high-risk population.

Introduction

Early-onset breast cancer in Colombian women shows aggressive behavior and frequent hereditary alterations. This study used genomic and germline analysis to highlight the need for early genetic testing to guide personalized treatment and family risk assessment.

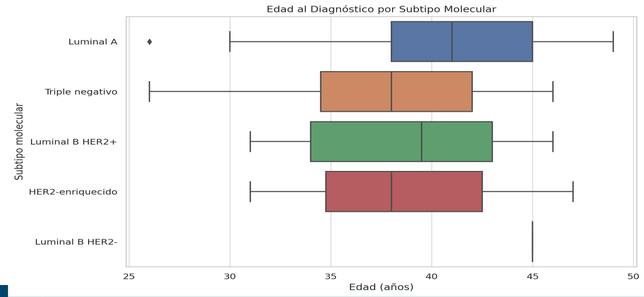
Methods and Materials

A retrospective cohort of 113 women <50 years diagnosed with BC at the Luis Carlos Sarmiento Angulo Cancer Treatment and Research Center (CTIC) between 2019 and 2024 was analyzed. Demographic, clinical, histopathologic, imaging, and molecular data were extracted. Tumor subtypes were defined based on ER, PR, and HER2 status. Germline sequencing was performed in 95% of cases (n=108). Chi-squared and t-tests were used to assess associations between mutation status, age, and clinical variables.



Results

Among 108 women with breast cancer diagnosed at ≤50 years, 43.5% carried at least one germline alteration. The most common subtype was Luminal (47.4%), followed by triple-negative (23.7%) and HER2-enriched (10.5%). BRCA1 mutations predominated in triple-negative tumors (18.2%), while PALB2, CHEK2, and non-canonical genes (MUTYH, POLE, APC) were frequent in hormone receptor–positive and HER2-enriched cases. Overall, 24.1% harbored variants in non-canonical genes, highlighting genomic heterogeneity. Mutation carriers were diagnosed at a significantly younger age (mean 34.9 vs. 41.8 years; p<0.001), underscoring the role of hereditary predisposition in early-onset breast cancer and supporting comprehensive germline testing for all young patients, regardless of subtype.



Discussion

Our findings reveal a high prevalence of germline pathogenic variants in early-onset breast cancer among Colombian women, with subtype-specific patterns such as BRCA1 in triple-negative and PALB2/CHEK2 in hormone receptor–positive tumors. The frequent detection of non-canonical gene alterations highlights the need for comprehensive multigene testing beyond BRCA1/2. Early genetic assessment is crucial for guiding targeted therapies and prevention strategies for patients and relatives. Despite its retrospective design and limited sample size, this study supports expanding genetic testing to improve early detection and personalized care in young breast cancer patients.

Conclusions

Germline pathogenic variants are common in early-onset breast cancer, particularly in triple-negative and luminal subtypes. BRCA1 predominates in triple-negative cases, while PALB2 and CHEK2 occur in hormone receptor–positive tumors. The detection of non-canonical gene alterations (24.1%) underscores the genomic heterogeneity of this population and supports early, comprehensive genetic testing to guide personalized management and familial risk assessment.

Contact

Maria Alejandra Bravo
Centro de Tratamiento e Investigación sobre Cancer Luis Sarmiento Angulo - CTIC
Email: malebravo87@gmail.com
Website: https://www.cticsarmientoangulo.org/
Phone: +57 310 8627483

References

1. Copson et al. Nat Rev Clin Oncol. 2018;15(12):737-750.
2. Metcalfe et al. Lancet Oncol. 2018;19(4):e182.
3. Paterson & Phillips. Expert Rev Anticancer Ther. 2017;17(11):991-1002.
4. Tung et al. Cancer. 2015;121(1):25-33.
5. Zhu et al. Breast Care (Basel). 2025 Jul 2. doi: 10.1159/000547029.
6. Lin et al. Oncologist. 2014;19(6):583-91.

This presentation is the intellectual property of the author/presenter. Contact them at <<malebravo87@gmail.com>> for permission to reprint and/or distribute.

© Poster Template by Genigraphics® 1.800.790.4001 www.genigraphics.com