

Conference Abstracts

Characterizing BRCA1 and BRCA2 mutations in Cameroonian breast cancer patients: Efforts towards bridging the genomic gap in Africa

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Background

There are profound global inequities in cancer genomics research, with publications focusing on cancer genetics from Africa representing only 0.016% of the global total. This genomic underrepresentation creates significant barriers to equitable cancer care and precision medicine for African populations. BRCA1 and BRCA2 mutations remain critically understudied in these communities, limiting our understanding of hereditary breast cancer patterns. This pioneering study characterizes BRCA1/BRCA2 mutations among Cameroonian patients, laying the foundation for future genomic research and addressing critical knowledge gaps.

Methods

This hospital-based study recruited 82 breast cancer patients from 2 major treatment centers in Cameroon, representing an underserved population with limited access to genetic services. Following culturally sensitive pre-test genetic counseling, saliva samples were collected and analyzed using next-generation sequencing for 29 cancer-associated genes. This approach addresses the critical gap in genetic testing accessibility for vulnerable African populations.

Results

Pathogenic mutations were identified in 23 patients (28%), with BRCA1 (15) and BRCA2 (2) accounting for 73.9% of all mutations (18.3% and 2.4% of the total population, respectively). The most frequent BRCA1 mutation was c.4484G>T(p.Arg1495Met), present in 7 patients (46.7% of BRCA1 mutations, 8.5% of total population), followed by c.5155dup(p.Val1719Glyfs6) in 3 (including 1 with bilateral breast cancer) patients (20% of BRCA1 mutations). BRCA2 mutations included c.1813dup(p.Ile605Asnfs11) and c.5572del(p.Thr1858Glnfs*5). Family history of cancer was reported in 73.9% of mutation carriers compared to 62.2% overall. Variants of uncertain significance were detected in 25.6% of patients. Two of those with the c.4484G>T BRCA1 mutation had an associated VUS in the PALB2 gene (c.365A>G(p.Asp122Gly)). They were both under 30 and had at least 4 first- and second-degree relatives with breast and ovarian cancer. Two other patients had the same VUS in APC (c.3760A>G(p.Ile1254Val)), all with at least 2 relatives with cancer, and 1 had the BRCA1 c.4484G>T mutation, and the other had no pathologic mutation identified.

Conclusions

This pioneering study reveals striking health disparities in cancer, with BRCA1/BRCA2 mutation frequencies exceeding those in well-studied populations. The predominance of specific mutations and VUSs suggests population-specific genetic architecture requiring tailored approaches. These findings demonstrate the urgent need for inclusive research and equitable implementation of genetic services in

underrepresented communities. Our work provides a foundation for developing culturally-appropriate cancer prevention strategies, reducing genetic testing disparities, and advancing precision medicine accessibility in vulnerable populations, ultimately contributing to the global effort to eliminate cancer health disparities. Future efforts would focus on implementation studies to test the sustainability of incorporating cancer genetics into routine clinical practice in this low-resource setting.