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BRCA1 and BRCA2 mutations in Senegalese women with hereditary breast cancer

Rokhaya Ndiaye Diallo and Jean Pascal Demba
University Cheikh Anta Diop, Senegal

Statement of the Problem: The majority of breast cancers are sporadic; however 5 to 10% are hereditary. The mechanism of carcinogenesis in hereditary forms involves several tumor suppressor genes such as the BRCA1 and BRCA2 genes. Mutations of these genes conferring genetic predisposition to breast cancer have been described in Caucasian, Asian and American populations but rarely in African populations. The objective of this study was to identify mutations of the BRCA1 and BRCA2 genes in hereditary breast cancer in Senegal.

Methodology & Theoretical Orientation: Fifteen index cases, each with a family history of breast cancer were recruited after free and informed consent at Joliot Curie Institute of Aristide Le Dantec Hospital in Dakar. A family survey was conducted to establish pedigrees. Venous blood sample on EDTA tube was collected for DNA extraction. Mutation screening of BRCA genes was carried out by PCR of all exons followed by Sanger sequencing.

Findings: For BRCA1 gene, a duplication of 10 nucleotides (934_943dupAGCCATGTGG, p.Thr276AlafsX14) in exon 11 was identified in 6 index cases. Several healthy relatives of these index cases were also mutated. This mutation has been reported as having an African origin with a founder effect. A genetic test allowing the identification of this mutation has been performed in the laboratory of cytology of Aristide Le Dantec Hospital for breast/ovarian cancer prevention in women with family history. For BRCA2 gene, a new nonsense mutation c.5447T>G, p.Leu1740X was identified in one index case as well as in 4 healthy relatives.

Conclusion & Significance: This pilot study identified a recurrent mutation of the BRCA1 gene and a new mutation of the BRCA2 gene predisposing to hereditary breast and ovarian cancer in Senegalese women. These results will be used to advocate for setting up cancer genetic testing infrastructures in Senegal.

Biography

Rokhaya Ndiaye Diallo is an Associate Professor of Human Genetics at the Department of Pharmacy of the Faculty of Medicine, Pharmacy and Odontology of University Cheikh Anta Diop of Dakar, Senegal. After PharmD training at University Cheikh Anta Diop of Dakar, she has completed a Human Genetics PhD in 2014 at University Paris 7, France. She was awarded in 2010 a Fullbright Senior Scholar Fellowship in the Department of Pathology of University of Washington, Seattle, USA. Her research interests focused on the role of genetic variation in cancer susceptibility and genetic basis of single gene disorders in African populations. Her current research projects focused on breast and ovarian cancers, head and neck cancer and rare genetic diseases.

rokhaya9.ndiaye@ucad.edu.sn

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