Specific BRCA1 gene variations amongst young Moroccan breast cancer patients


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ABSTRACT. Germline mutations in the BRCA1 gene are known predictive markers for the development of hereditary breast cancer. Nevertheless, no comprehensive study has been performed targeting the presence and relevance of BRCA1 mutations in Moroccan breast cancer patients. We here present an analysis of BRCA1 gene regions (exon 2 and exon 11a/b) of 50 female Moroccan breast cancer patients with early disease onset (≤40 years) or familial disease backgrounds. Results showed that no mutation was present in either exon 2 or exon 11a of the BRCA1 gene in any of the 50 patients analysed. However, in exon 11b, a mutation generated by a nucleotide exchange was detected in 8% of patients, most of whom were young women (≤40). This mutation leads to substitution of the amino acid glutamine by an arginine at position 356 of the polypeptide sequence (Q356R). Although this mutation was previously characterised at a lower frequency in western populations, our study is the first to describe it in a young Moroccan population.
Furthermore, another mutation was detected with a high frequency (4%) on exon 11b of the \textit{BRCA1} gene in exclusively young patients (≤40). This mutation was silent, encoding the same threonine residue at position 327 (T327T) as the wild type. The present study is the first to describe this mutation as well, particularly in a young Moroccan population. Analysis of a larger population is required in order to highlight the relevance of the Q356R and T327T mutations in young Moroccan breast cancer patients.

\textbf{Key words:} Breast cancer; Clinical samples; \textit{BRCA1} gene; Exon sequencing; Mutation