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Identification of a miR-1245a germline variant and its relation with early-onset breast cancer susceptibility

Lilian Jara
University of Chile, Chile

Breast Cancer (BC) is one of the most frequent cancers affecting women worldwide. In Chile, BC has the highest mortality rate among cancers (15.69/100,000 women). Recent evidence supports a role for microRNAs (miRNAs) in BC development and progression. Single-Nucleotide Polymorphisms (SNPs) are the most common type of variation in the human genome. SNPs in miRNA genes can alter expression, maturation or target binding affinity, thus contributing to the development of cancer. Studies have documented that SNPs in miRNA(s) targeting BRCA1/2 genes alter BRCA gene expression levels. miR-1245a targets the BRCA2 gene. Sequencing pre-miR-1245a in 107 BRCA1/2-negative BC probands from high-risk families resulted in the identification of rs60611793, which corresponds to a 1-bp deletion. This variant was detected in an early-onset (diagnosis at 30 years of age), Triple-Negative (TN) BC case without a family history of BC. Given the clinic-pathological features of the patient carrying the deletion, we used Sanger sequencing to screen for rs60611793 in another 181 early-onset and TN BC cases and 192 healthy individuals. The deletion was detected in another four BC cases and 3 of 192 controls. The five cases with the deletion had early-onset BC. However, there was no correlation between rs60611793 and histological BC type. Two of the five cases were poorly-differentiated ductal carcinomas, one was a medullar carcinoma, one was an in-situ carcinoma and the histopathological report for the fifth case was unavailable. Therefore, this deletion could be related to the development of early-onset BC and high-grade malignant neoplasms, with no predominant ER/PR/Her2/Neu status.

Biography

Lilian Jara is a Geneticist with a PhD in Biomedical Sciences and the Head of the Human Genetic Program at University of Chile. She is an expert in cancer genetics, with a research focus on familial breast cancer. She has focused her research in search for new genetic factors involved in the development of familial breast cancer in patients without point mutation in BRCA1/2 genes. She has published several articles with novel findings on Chilean familial breast cancer.

seba.morales.p@gmail.com

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