

Frequency of the 185delAG mutation in the BRCA1 gene in Chilean healthy women with family history of breast cancer Frecuencia de la mutación 185delAG en el gen BRCA1 en mujeres chilenas sanas con antecedentes familiares de cáncer de mama.

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BACKGROUND: Breast cancer is the most common malignancy among women, and is the second cause of cancer mortality among Chilean women. Female mortality due to breast cancer in Chile has shown a steady increase from 9.5 deaths per 100.000 women in 1985 to 12.8 deaths per 100.000 in 1995. A family history of breast cancer is one of the main risk factors for the development of the disease. BRCA1 and BRCA2 are two major hereditary breast cancer susceptibility genes. Mutations in these genes are associated to inherited breast cancer; 664 predisposing mutations have been described, but in specific populations only some of them, such as 185delAG have been found to be associated with susceptibility to breast cancer. **AIM:** To establish the frequency of the 185delAG mutation in the BRCA1 gene in Chilean healthy women with a family history of breast cancer. **PATIENTS AND METHODS:** The 185delAG mutation was studied by mismatch polymerase chain (PCR) reaction in 382 Chilean healthy women with at least