Double heterozygous pathogenic variants in the BRCA1 and BRCA2 genes in a patient with bilateral metachronous breast cancer: a case report

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Abstract

Background

Double heterozygosity pathogenic variants in BRCA1 and BRCA2 genes are a very rare finding, particularly in non-Ashkenazi individuals. We described the first case of double heterozygosity variants in a non-Ashkenazi Argentinean woman with metachronous bilateral breast cancer.

Case presentation

The proband is a 65-year-old female diagnosed with invasive ductal carcinoma in the left breast at 45 years old and invasive carcinoma in the right breast at 65 years old. She underwent a multi-gene panel testing indicating the presence of two concurrent heterozygous germline deleterious variants in BRCA1 (c.4201C>T), and BRCA2 (c.5146_5149del) genes. The patient's son (40 years-old) was found to have the inherited pathogenic variant in BRCA2 gene.

Conclusion

There are few reports of double heterozygosity variants in BRCA1 and BRCA2 genes in Latin America. The two pathogenic variants identified in our patient have not been described together so far.

Full Text

Due to technical limitations, full-text HTML conversion of this manuscript could not be completed. However, the manuscript can be downloaded and accessed as a PDF.

Figures

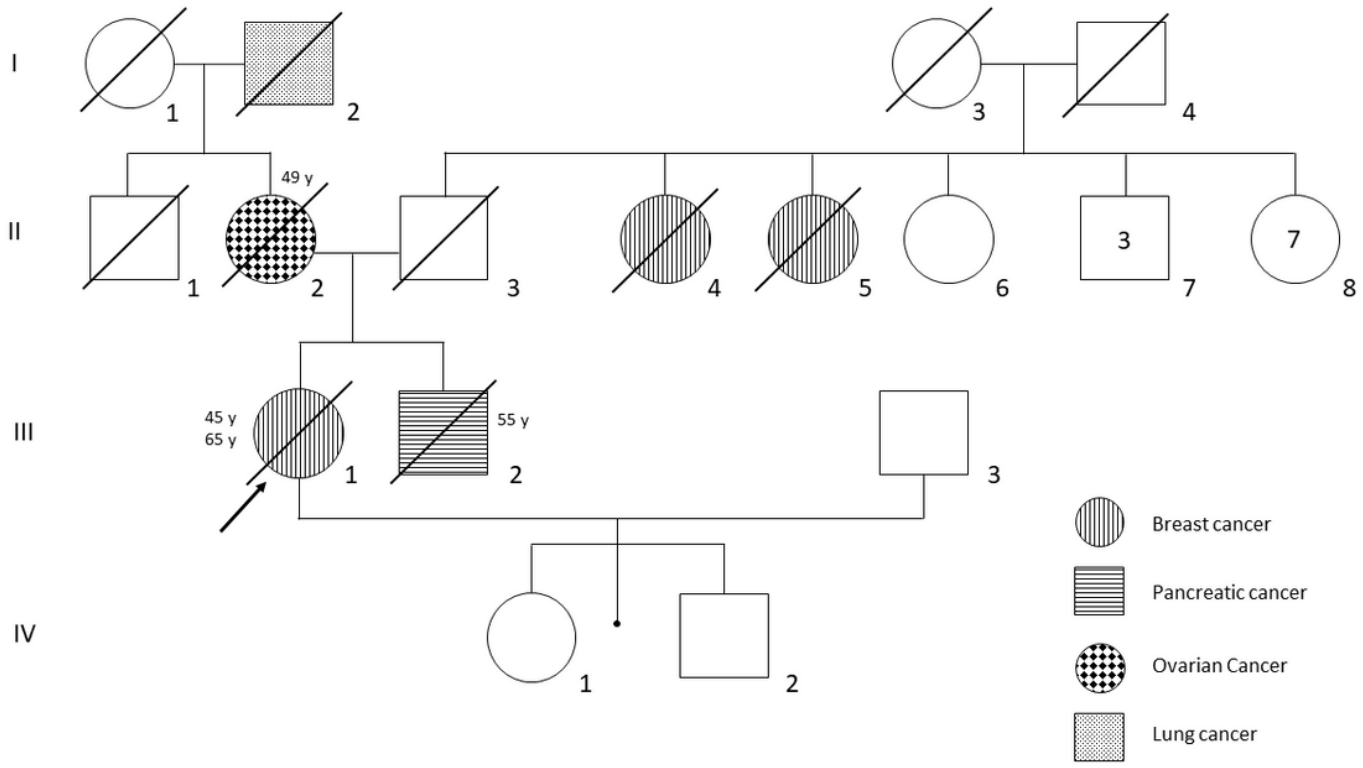

Figure 1

Family pedigree. Age at diagnosis is indicated. Diagonal slash indicates deceased, while the proband is indicated with an arrow. Two pathogenic monoallelic mutations were detected in the proband, one in the BRCA1 gene and one in the BRCA2 gene. Subsequent predictive testing was offered to the children. The patient's son presents a pathogenic monoallelic mutation in the BRCA2 gene.

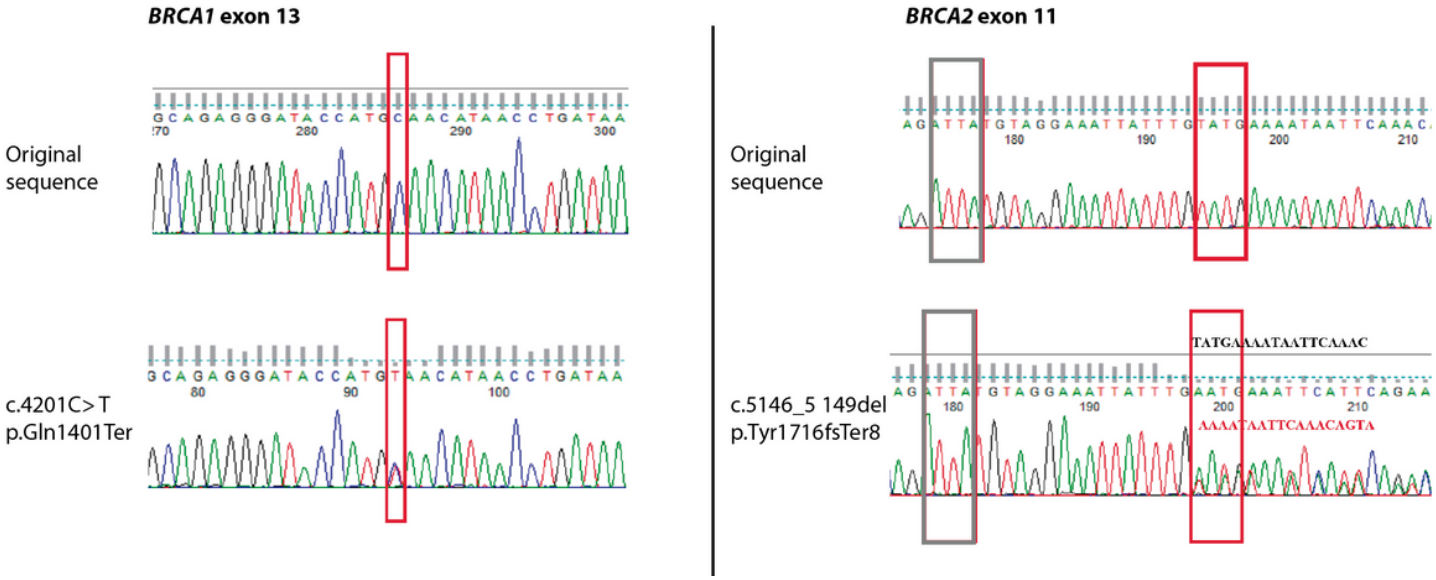

Figure 2

Sanger-type sequencing genomic DNA of the proband. The nucleotide changes in BRCA1/2 genes are indicated on the electropherograms in red. The gray box indicates the region before the frameshift.