Reviewer’s report

Title: Patients with Pathogenic Variants for Breast Cancer Other Than BRCA1 and BRCA2: Qualitative Interviews about Health Care Experiences

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Reviewer: Mev Domínguez Valentin

Reviewer's report:

The manuscript entitled «Patients with Pathogenic Variants for Breast Cancer Other Than BRCA1 and BRCA2: Qualitative Interviews about Health Care Experiences" provided a qualitative research that describe the gap between the pace of medical advances in genetics and the ability of providers to incorporate this information into clinical practice, mainly for less the well-known breast cancer (BC) associated genes. It is a very interesting focus and congratulations for it!

Some comments:

Abstract

1. Include the total number of individuals you mailed the survey.
2. Use the term "pathogenic variants" in the results and conclusion.
3. If the aim of the manuscript is to explore the personal and health care experiences form patients who had pathogenic variants "other" non BRA1/2" genes, these genes should be stated (e.g. ATM, CHKE2, PALB2 PTEN or TP53).

Page 6 Line 53: It would have been more appropriate to use the term "variant" throughout the manuscript, which is in line with the standards and guidelines for the interpretation of sequence variants [e.g. Richards et al, 2015]. The term "variant" should replace the term "mutation" or "polymorphism" with the following modifiers: (i) pathogenic, (ii) likely pathogenic, (iii) uncertain significance, (iv) likely benign, or (v) benign.
Results

It had stated that the selected individuals had pathogenic variant in CHEK2 (8), ATM (7), PALB2 (5), TP53 (2), NBN (1), MSH6 (1), PTEN (1), and RAD51C (1), however in the methods part, it had been described only ATM, CHEK2, PALB2 PTEN or TP53 genes.

Specify which BC associated genes were included? And, why did you select them?

Level of interest
Please indicate how interesting you found the manuscript:

An exceptional article

Quality of written English
Please indicate the quality of language in the manuscript:

Acceptable

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