Reviewer’s report

Title: A novel deleterious PTEN mutation in a patient with early-onset bilateral breast cancer

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Reviewer: Robert Pilarski

Reviewer’s report:

This is a well written and well documented case report of a women with Cowden syndrome (CS) who developed bilateral breast cancer. While bilateral breast cancer has been reported in several series of CS patients in the past, the majority were prior to the identification of the PTEN gene as the cause of CS, and the risk of bilateral cancer has never been quantified. The authors have done an admirable job performing laboratory studies supporting the pathogenicity of the patient’s missense mutation, which strengthens their findings. This type of work is typically absent in most case reports. While not presenting entirely new findings, this report does make important teaching points regarding rarer syndromes which may predispose to breast cancer.

Major Compulsory Revisions:
- None

Discretionary Revisions:
- “Clinical History” section: since the initial breast cancer was ER/PR positive, HER2/neu negative, while the relapse was ER/PR negative, HER2/neu positive, the authors should discuss the possibility that this represents a third primary breast cancer, rather than a relapse.
- “Conclusions” section: the first paragraph should include a short discussion of the available data on breast cancer risk in women with PTEN mutations, particularly the limited data on bilateral risk.

Minor Essential Revisions:
- “Background” section, line 16: “PTEN Hamartoma Tumor Syndrome” should be abbreviated “PHTS”, not “PTHS”.
- “Functional assessment of the mutation” section: first line, “length” is spelled incorrectly.

Level of interest: An article whose findings are important to those with closely related research interests

Quality of written English: Acceptable

Statistical review: No, the manuscript does not need to be seen by a statistician.
Declaration of competing interests:

I declare that I have no competing interests.