Reviewer’s report

Title: CHEK2 1100delC is prevalent in Swedish early onset familial breast cancer, a case control study

Version: 1 Date: 13 April 2007

Reviewer: Petr Pohlreich

Reviewer’s report:

General

This paper by Margolin and coworkers describes analysis of the prevalence of the CHEK2 1100delC mutation in a set of 763 breast cancer patients and 760 controls from the Stockholm region. 2.2% of all familial cases carried the mutation compared to 0.7% of the controls. The prevalence of 1100delC was increased in familial but not in sporadic breast cancer. The variant lowered age at onset in carriers and might constitute a modifier of risk in syndrome.

Major Compulsory Revisions (that the author must respond to before a decision on publication can be reached)

Minor Essential Revisions (such as missing labels on figures, or the wrong use of a term, which the author can be trusted to correct)

1. Page 4: Familial Risk Cohort ... For these cases only age at diagnosis ... Dividing of breast cancer cases into groups could be a bit clearer. Do the following cases “For these cases only age at diagnosis was available (missing in 16 patients)” belong to the familial risk cohort?

2. Page 5: CHEK2del100C-R ....t aat-3´ A primer set that is specific for the mutation contains one mismatch.

Discretionary Revisions (which the author can choose to ignore)

What next?: Accept after minor essential revisions

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'