Reviewer's report

Title: First evidence of a CHEK2 duplication involved in cancer predisposition in an Italian family with hereditary breast cancer

Version: 1 Date: 4 April 2014

Reviewer: Pål Møller

Reviewer's report:

This is a report on a large duplication in CHEK2 associated with disease. Also, the introduction gives a valuable overview of the various associations previously found between cancer and inherited CHEK2 variations. As such, this is information which should be available to build the common knowledge on CHEK2 and its associations to cancer.

A clear description of the duplication should be given in both the title and abstract.

A clear statement of why the duplication is demonstrated to cause disease in the family (=result of segregation analysis) should be given in the text.

A clear reason why no other genetic variation found in the family is causing cancer in the family, should be given.

A summary of how many single-nucleotide variants (nonsense, silent, missense), truncating mutations and larger in/dels should be given as numbers in each group.

All above to support the conclusion that the report of a large duplication adds to the knowledge on CHEK2.

Level of interest: An article of importance in its field

Quality of written English: Acceptable

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

Declaration of competing interests:

No conflict of interest