Abstract for midway evaluation report - May 2018 - Hildegunn Høberg Vetti

# Hereditary Breast and Ovarian Cancer:

# Evaluation of new procedures for genetic testing

The aim of this PhD project is to explore and evaluate alternative procedures for genetic testing and counselling of patients newly diagnosed with breast or ovarian cancer, in order to meet the increasing need of this health service in the era of personalized medicine.

In the first paper we offered *BRCA1/2* testing and familial risk assessment to all new patients with breast (N=893) or ovarian (N=122) cancer diagnosed between September 2012 and February 2015. The results showed that 19 of 83 patients (22.3 %) with ovarian cancer and 7 of 405 patients (1.7 %) with breast cancer carried a germline *BRCA1* or *BRCA2* mutation. Current test criteria were sufficient to identify most *BRCA1/2* mutation carriers.

In the second paper, recently submitted for publication, we have investigated the psychosocial aspects of this new practice for genetic testing.

In the third paper the objective is to determine the pathogenicity of a specific intron variant in *BRCA1* which we published in the first paper.