Hereditary and hormonal risk factors for breast cancer

Studies on family history, incidence, genetic counseling and testing, and hormonal factors

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Background
More than 8,000 women are diagnosed with invasive breast cancer (BC) annually in Sweden. Most often, women are affected later in life. However, approximately 1.5% are 35 years or younger at diagnosis. A major risk factor for being affected early in life is a previous family history of BC. Other factors associated with an increased risk for BC in women in general are hormonal factors, e.g., use of oral contraceptives and hormone replacement therapy.

Methods and Preliminary Results
Paper I: Cohen’s Kappa analysis was performed to determine concordance between self-reported and registry-reported information regarding cancer in first-degree relatives of 231 patients diagnosed with BC at \(\leq 35\) years, registered at the Oncogenetic Clinic in Lund between 1993 and 2013. Almost perfect agreement was observed between self-reported and registry-reported information regarding BC and ovarian cancer (OvC). However, lesser agreement was observed regarding other types of cancer.

Paper II: Logistic regression analysis was performed to identify factors influencing the probability of registration at the clinic for 279 early-onset BC patients, diagnosed between 2000
and 2013, in the South Swedish Health Care Region. BC patients from Blekinge and the southern part of Halland, and patients from rural settings with a population of less than 10,000 inhabitants, were significantly less likely to be registered.

Paper III: We plan to send a questionnaire to the 100 early-onset BC patients, diagnosed between 2000 and 2013, who have not been registered at the clinic. Analysis of their answers will subsequently provide us with further knowledge regarding reasons to why they have not been in contact with the clinic.

Paper IV: We plan to evaluate the interaction between heredity, hormonal factors, and cancer risk in BRCA tested families by comparing BRCA carriers in all tested families, registered at the clinic between 1993 and 2017, with controls.

**Significance**

Our results demonstrate that physicians and genetic counselors can rely on self-reported information regarding BC and OvC in first-degree relatives. However, self-reported information regarding other types of cancer is not communicated as effectively. Furthermore, our results indicate a need for extended oncogenetic service in regional hospitals to improve care.

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