Objective

Breast cancer affects more than 360,000 women per year in the EU and causes more than 90,000 deaths. Identification of women at high risk of the disease can lead to disease prevention through intensive screening, chemoprevention or prophylactic surgery. Breast cancer risk is determined by a combination of genetic and lifestyle risk factors. The advent of next generation sequencing has opened up the opportunity for testing in many disease genes, and diagnostic gene panel testing is being introduced in many EU countries. However, the cancer risks associated with most variants in most genes are unknown. This leads to a major problem in appropriate counselling and management of women undergoing panel testing. In this project, we aim to build a knowledge base that will allow identification of women at high-risk of breast cancer, in particular through comprehensive evaluation of DNA variants in known and suspected breast cancer genes. We will exploit the huge resources established through the Breast Cancer Association Consortium (BCAC) and ENIGMA (Evidence-based Network for the Interpretation of Germline Mutant Alleles). We will expand the existing datasets by sequencing all known breast cancer susceptibility genes in 20,000 breast cancer cases and 20,000 controls from population-based studies, and 10,000 cases from multiple case families. Sequence data will be integrated with in-silico and functional data, with data on other known risk factors, to generate a comprehensive risk model that can provide personalised risk estimates. We will develop online tools to aid the interpretation of gene variants and provide risk estimates in a user-friendly format, to help genetic counsellors and patients worldwide to make informed clinical decisions. We will evaluate the acceptability and utility of comprehensive gene panel testing in the clinical genetics context.

Related information

Report Summaries

Periodic Reporting for period 1 - BRIDGES (Breast Cancer Risk after Diagnostic Gene Sequencing (BRIDGES))
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**EU contribution:** EUR 75 000

Last updated on 2017-06-16
Retrieved on 2019-06-26


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