The EMBED Study brief summary.

Women at high risk of developing breast cancer often undergo a double mastectomy to eliminate the future development of breast cancer. Detecting the disease at an earlier stage, therefore, could be a major step forward for a reduction in the need to have risky surgery.

There has been considerable interest recently in using blood-markers in the early detection of cancer. ctDNA (circulating tumour DNA) enters the bloodstream through the death of cancer cells and is now an important blood marker in advanced disease. The aims of this study are two-fold: (i) to further develop and use sensitive state-of-the art assays to detect small amounts of ctDNA in the blood with increased positivity and sensitivity and (ii) to measure ctDNA over time in women who develop breast cancer. Using these blood-markers it may be possible to detect breast cancer at a much earlier stage.

We aim to recruit two cohorts of women to answer these research questions:

1. Approximately 2700 women with a strong family history of breast cancer; we estimate that about 40 of these women will develop breast cancer during the life-time of the study. All women with a strong family history of breast cancer are offered annual mammograms at their local hospital Breast Unit. We aim therefore to recruit patients through the Breast Units, in East Anglia initially. These women will be asked to provide annual blood samples and complete some questionnaires for up to five years or until they receive a clinical diagnosis of breast cancer.

For women who develop cancer we will first analyse the ctDNA in the blood samples collected at the time closest to diagnosis. We will also obtain mammogram images and tumour samples to guide a more sensitive analysis. Following this, we will analyse the blood samples collected at other time points to establish the earliest point at which ctDNA can be detected in the blood.

1. 100 women who have been recalled to the clinic because of abnormalities seen on their recent mammogram (in Cambridge only). We will take blood samples from these women to refine and validate the ctDNA assays in readiness for use in the main study.

We would like your comments and suggestions on a number of patient information documents. We have to be upfront about our research and we do have certain legal requirements to meet but we are aware that, in group 2 particularly, women will be already highly anxious and we do not want to add to that.

We aim to provide a layered approach to the information provided. In the first instance, patients will receive a brief flyer in with their appointment letters from the hospital. They will then be able to ask for further information, which will be a more detailed information sheet.

Joanna Proctor (Study Coordinator)

Centre for Cancer Genetic Epidemiology

University of Cambridge

Jm294@medschl.cam.ac.uk

01223 748670