BRCA-DIRECT: A randomised UK study evaluating a digital pathway for germline genetic testing and non-inferiority of digitally-delivered information in women with breast cancer.

Overview

The BRCA-DIRECT study is:

**Evaluating** the feasibility, acceptability and safety of a digital pathway for end-to-end management of germline genetic testing of BRCA1, BRCA2 and PALB2.

**For** unselected breast cancer patients within UK National Health Service (NHS) oncology clinics.

Using saliva-based sampling and supported by a Genetic Counselling hotline, we hypothesise that testing for the majority of breast cancer patients could be managed end-to-end more rapidly and efficiently via a clinically-integrated digital system providing consistent, accurate information (BRCA-DIRECT digital pathway). This would reduce the clinical resource and the overall cost of the testing pathway.

Background

Germline BRCA and PALB2 testing in women diagnosed with breast cancer affords multiple opportunities for:

1. Treatment of a current breast cancer
2. Management of risk of future cancers
3. Identification of at-risk family members through cascade testing

However, whilst the wet-lab costs have plummeted, the NHS clinical infrastructure around delivery of germline genetic testing remains laborious, low-throughput and thus low volume.

Accordingly, access to genetic testing is currently limited (<20% of women diagnosed with breast cancer are eligible) and national assentainment of pathogenic variant carriers remains poor (<5% identified)².

Trial Design

Patients are randomised 1:1 to either the fully-digital or partially-digital arm for evaluation of non-inferiority of digital information versus an appointment with a genetics professional (primary outcome measure).

Additional outcome measures include:

- Genetic test uptake
- Hotline usage
- Patient anxiety, knowledge about genetic testing, and satisfaction
- Healthcare professional satisfaction

Recruitment

**Inclusion:**

- Over 18
- Diagnosis of Invasive Breast Cancer or High-Risk Ductal Carcinoma in situ (DCIS)
- Good comprehension of the English Language
- Access to a smartphone or email + internet access

**Exclusion:**

- Previous genetic testing (BRCA1, BRCA2, PALB2)

Target: 1000 women from two UK regional oncology centres (Royal Marsden NHS Foundation Trust (London) and Manchester University NHS Foundation Trust).

Current Progress

Since July 2021, 724 patients have been recruited, with 611 having consented to genetic testing (84.39%). Of these, results have been returned to 529 women (96.58%) with a positive result (pathogenic variant or hotspot Variant of Uncertain Significance identified) and 9.6% with a negative result (normal or variant of Uncertain Significance).

Data from 13 April 2022

References


Contact

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Ethics Approvals

The study received ethical approval from the London – Chelsea Research Ethics Committee (REC) on 04 January 2021 (REC reference: 20/LO/1200)