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From National Guideline Recommendations to Familial Cancer Risk Assessment Decision Support in Primary Care: UK Experience

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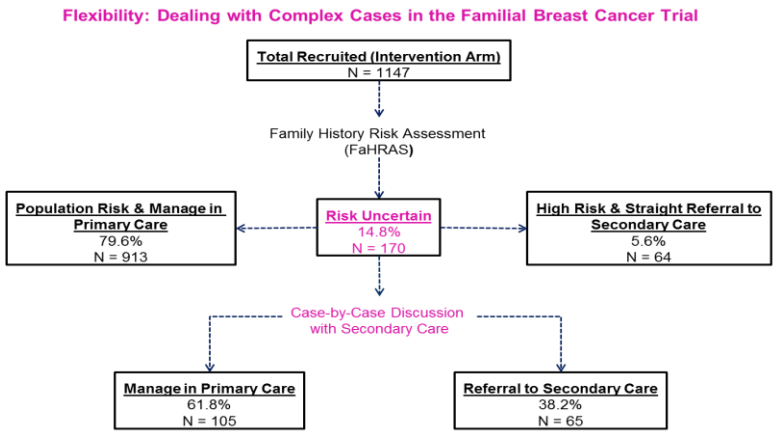
BACKGROUND

The English national (NICE) guidelines on managing women with a family history of breast cancer provides criteria for specialist care referral (in those at moderate/high risk) and for patients that can be followed up in primary care (in those at near-population risk). Decisions for referral are based on details in the family history and previous genetic testing in relatives. The assessment, as well as indicating probability of genetic mutations (in current NICE guidelines set at a 10% threshold), also determines lifetime risk of breast cancer from 20 years age : High risk > 30% risk; Moderate risk 17-29%; Near-population <17%.
Also there is a category, with unclear family histories, where the guidelines recommend that patients are not referred directly but discussed with specialists. We have explored the nature of family histories that fall into this category and how to integrate this information into primary care decision support software.

MATERIALS AND METHOD

In our exploratory trial all women aged 30 to 60 in four General Practices, in Central England, were invited to complete a family history questionnaire. The family histories were assessed against national guidelines and Manchester Scoring System using bespoke primary care familial history risk assessment software (FAHRAS). The Manchester Scoring System estimates the probability of identifying mutations in the BRCA1 and BRCA2 genes.

RESULTS



14.8% (170) of 1147 women completing the questionnaire were recommended by guidelines to discuss with familial cancer specialist. These include family histories with;

- Ovarian cancer in living and deceased relatives
- Other cancers, particularly in the young
- Cancers in more distant relatives
- Jewish ancestry

Manchester Scoring System was applied to all 170 pedigrees.

- 3 (1.76%) participants had a 1st degree relative with a Manchester score over 10%
- 12 (7.06%) participants had a 2nd degree relative with a Manchester score over 20%.

CONCLUSION

Based on the current study over 1 in 7 patients assessed for familial breast cancer risk have unclear family histories and need to be discussed with the familial cancer specialists. This would overload familial breast cancer services in the UK. To operationalise national guidelines and implement decision support software in primary care, uncertain family histories need to be reduced. This could be achieved by combining national guidelines with consensus opinion of specialists and key attributes of evidence-based referral tools, such as Manchester Scoring System.

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