# Editorials

# Dealing with family history of breast cancer:

## something new, something old

### BACKGROUND

Breast cancer is a major burden to society and individuals with 49 564 new cases and 11 633 deaths in the UK in 2010. A woman in the UK has an up to 1 in 8 lifetime risk of developing breast cancer[1] and 20% of women with breast cancer will have a positive family history.[2] While the management of breast cancer is carried out largely in specialist care, the ability to potentially intervene in people at increased familial risk of breast cancer will enhance the role of primary care.

### ASSESSMENT OF RISK

The National Institute for Health and Care Excellence (NICE) developed guidelines on classification and care of women at risk of familial breast cancer in 2004. These were partially updated in 2006 and again earlier this year.[3] The primary care-relevant recommendations from the original guideline in 2004 have been retained in the current update. These recommendations advise that a family history enquiry would be appropriate when a patient presents with breast symptoms, with concerns about familial risk or in situations where collection of family history is clinically relevant, such as at oral contraceptive checkups. In these circumstances, first and second-degree family history should be collected, not forgetting second–degree relatives include paternal relatives. When a relative with breast cancer is identified the following details should be collected to accurately assess breast cancer risk: age at diagnosis, site of cancer(s), and if the patient is of Jewish ancestry.[4] The guidelines do not suggest specific approaches to stratify the familial risk but recognises the role of family history questionnaires to collect family history, and decision-support computer packages to facilitate risk assessment. Available family history tools are predominately self-administered by the patient, with a few tools automatically collating the family history into risk categories.[5] An integrated primary care-relevant electronic family history collection and decision support tool still needs to be developed and validated.[6] In the absence of such tools, the GP should eyeball collated family history against the guideline recommendations on stratifying familial risk. The stratification categorises women into near-population risk, moderate risk and high risk (<17%, 17–<30%, ≥30% lifetime risk respectively: Table 1). This assessment may be enhanced by suggesting that the patient explores the family history with relatives. Having categorised the patient into specific risk categories, the guidance recognises primary care will need extra support, including standardised patient information and educational materials and, perhaps most important, a designated secondary care contact to discuss 'uncertain cases'. Uncertain cases include relatives with sarcomas diagnosed aged <45 years, gliomas, or childhood adrenal cancers. Specific patient information is available through charitable organisations.[7,8] The patient should also be offered information on breast awareness and lifestyle advice.

### POSSIBLE PREVENTATIVE TREATMENT

Once family history has been assessed, those at near-population risk will remain under the care of primary care but the patient will be reminded to return if the family history has changed or breast symptoms develop. The emergence of a further relative with early onset breast cancer may shift the patient into a higher risk category. Those assessed at moderate or high familial cancer risk should be referred to specialist care for assessment leading to increased surveillance if confirmed. Based on emerging evidence on the effectiveness of magnetic resonance imaging (MRI), the 2006 update included recommendations on MRI surveillance in women aged 30–49 years at increased risk of BRCA1/BRCA2 gene mutations,[9,10] while the 2013 update recommended women, aged 20–69 years, at increased risk of TP53 gene mutation should also be offered MRI surveillance. These women are currently not offered screening through the National Breast Cancer Screening programme. New to the revised 2013 guidelines, and receiving intense media attention, is the incorporation of the recent evidence that those at higher risk of (oestrogen-receptor

**Table 1. Summary of breast cancer risk categories, related care settings and management recommendations in women with a family history but no personal history of breast cancer**

| | Definition of category | | | | Recommendations | | |
|---|---|---|---|---|---|---|---|
| Breast cancer risk | Lifetime breast cancer risk from aged 20 years | Breast cancer risk age 40–50 years | Probability of a breast cancer genetic mutation[a] | Care Setting | Surveillance | Risk-reducing surgery[b] | Chemoprevention for 5 years[b] |
| Near population risk | <17% (= <1 in 6) | <3% | Very low | Primary care | – | – | – |
| Moderate risk | ≥17% but <30% (= >1 in 4) | 3–8% | <10% | Secondary care | Offer annual mammography for women aged 40–49 years and consider for women aged 50–59 years | – | If aged >35 years, consider treatment unless woman has had bilateral mastectomy |
| High risk | ≥30% (= ≥1 in 3) | >8% | >10% chance of a faulty gene in the family | Specialist genetic clinic | Offer annual MRI or mammography. Actual procedure based on age of patient and genetic mutation (identified or predicted) | If appropriate, offer mastectomy and bilateral salpingo-oophrectomy | If aged >35 years, offer treatment unless woman has had bilateral mastectomy |

*[a] BRCA1, BRCA2 or TP53 mutation. [b] Only offer treatment after assessment and counselling by genetic specialists.*

positive) familial breast cancer should be given the option of chemoprevention with tamoxifen or raloxifene for 5 years, unless at increased risk of thromboembolic disease or endometrial cancer.[11–14] In premenopausal women the evidence is limited to tamoxifen.[10] Specifically, it is recommended that those at high breast cancer risk are offered chemoprevention, while treatment could be considered in those at moderate risk. Although initial counselling is likely to occur in specialist care, repeat prescribing will come under the domain of primary care. With tamoxifen there is a slight additional risk of post-menopausal endometrial cancer, highlighting the need to advise women to contact their GP immediately if they develop atypical uterine bleeding or post-menopausal bleeding. Further, women should stop chemoprevention at least 2 months before trying to conceive and 6 weeks before elective surgery.

## WOMEN WITH BREAST CANCER

The updated 2013 guidelines also includes advice on familial risk assessment of patients with breast cancer.[4] Enquiries about family history and testing for predisposing gene mutation should now take place in secondary care for newly-diagnosed patients. However, these cancer patients are likely to also discuss their concerns with their GP. Patients with breast cancer with increased genetic risk of recurrent or new contralateral breast cancer may be offered increased surveillance: annual mammography in women aged >50 years or MRI in younger women. All women should also be offered written information about the risk and benefits of the procedure prior to undergoing surveillance.

GPs may also care for patients who have previously been treated for breast cancer and are no longer in contact with secondary care. Enquiries should be made about family history of breast cancer and if testing for cancer-predisposing gene mutations has been performed in relatives, with referral to specialist genetic clinics if a significant family history is identified.

## FURTHER IMPLICATIONS

With the media publicity about this guideline there is a possibility that patients previously diagnosed with breast cancer or those discharged from specialist services, after being previously assessed as at moderate familial risk, will consult their GPs. The drop in threshold for genetic testing, from 20 to 10% probability of a BRCA1 or BRCA2 mutation will lead to more patients being identified with genetic variants that currently are not linked to cancer-predisposition ('genetic variants of unknown significance'). It is assumed that clinical genetic services will inform the patient, in the future, if these genetic test results become clearer. However, the concerned patient is likely to return to their GP.

On the surface, this guideline may appear within the specialist arena, but primary care is the first port of call for patients with concerns about familial breast cancer risk and in those with relevant family histories embedded in their general practice records. Do we have a systematic way of exploring such family histories? Should we be more energetic about identifying and collecting family history, in particular, in patients with breast cancer? There is insufficient time in a 10-minute general practice consultation to collate and interpret a family history. One option is to develop resources for patients to collate this information outside of the consulting room, with speedy access in the GP consultation to the decision support information. Further, even if a patient has been classified at low (near-population) familial risk, it is important for GPs and patients to keep the family history up-to-date, with a process to interpret this information. As well as decision support tools, this includes designated specialist contacts. We are no longer passive bystanders in the management of this group of patients.


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**Provenance**
Freely submitted; not externally peer reviewed.

**Competing interests**
Gareth Evans and Nadeem Qureshi were members of the guideline development group that produced the NICE Familial Breast Cancer CG164 guideline.

DOI: 10.3399/bjgp14X676267


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