# Time to rethink the capture and use of family history in primary care

### Box 1. Case history

Mrs X is 32 years old and worried. She wants to discuss the breast cancer risk for her newborn daughter after learning that her paternal aunt was diagnosed with breast cancer in her 30s and died before she was 40. Her paternal grandmother was also diagnosed with breast cancer in her 70s. Should her GP: a) Advise Mrs X to remember to discuss this with her daughter when she is older? b) Add a code on the daughter's electronic record to indicate Family History of Breast Cancer? Or c) Recommend future referral to a genetics clinic?

## FAMILY HISTORY AND EARLY DIAGNOSIS

Since Hippocrates, doctors have recognised the value of family history.[1] It is less clear how GPs should use this information now that gene sequencing and electronic medical records are redefining what is possible. In rare genetic diseases, family history highlights important risks but shared ethnicity, culture, diet, and environment mean that family history is also a risk factor for many common conditions such as cancer, cardiovascular disease, and dementia. The UK's relatively poor outcomes in cancer, for example, have been partly attributed to reluctance by primary care physicians to investigate possible cancer.[2] Genetic clinics require patients to be identified in primary care before a referral is made, and GPs could make significant use of family history to do this.

In the US, every Thanksgiving Day since 2004 has been declared a 'National Family History Day' with promotion of My Family Health Portrait (https://familyhistory.hhs.gov), an online tool for families to record their relationships and relevant diseases.[3] Such patient-led tools can link to online resources such as Microsoft HealthVault but not to clinical systems, at least not yet. Rubin *et al* presented a model in the *BJGP* to improve early diagnosis from referral to treatment.[2] In this Debate and Analysis we argue for an earlier step — linking patient-led online family pedigree tools with primary care electronic health records to inform early risk assessment, lifestyle choices, and diagnostic screening *instead of* waiting for patients to present with signs of illness.

Patients often present to discuss their genetic risk following a family gathering (Christmas, weddings, or funerals) where information about a relative's illness is shared. This patient-initiated process is different from, but should be linked to, public health screening. Marmot states that everyone should have access to screening to reduce health inequalities[4] and Emery argues that computer-assisted decision support could empower GPs to assess genetic risk.[5] National Institute for Health and Care Excellence familial breast cancer guidelines recommend that direct relatives of female patients diagnosed under the age of 40 years should be referred for genetic advice and all relatives of male patients with breast cancer should be referred regardless of age at diagnosis.[6] We believe that GPs should be more proactive in recording and using family history information to make sure these referrals happen at the appropriate time (in Box 1, options a, b and c are all appropriate).

## CURRENT FAMILY HISTORY CAPTURE AND USE

To investigate our hypothesis that family history is currently under-recorded in electronic patient records, we used patient questionnaires at a single practice and a population-based study across an estimated 700 practices in England, focusing on breast cancer. Studies suggest that 5–10% of breast cancer is hereditary,[7] with a strong case for proactive identification, screening, and possibly genetic counselling.[8]

We sent a family history questionnaire to all 107 patients with a personal history of breast cancer at one urban GP practice with 15 000 patients (response rate 54%). The results indicated that 29% of these patients with breast cancer had relatives who were at increased risk of breast cancer, but many were unaware of this.[9] These relatives require screening at the appropriate age, but there are no national guidelines for recording family history coding within the primary care records, or ensuring appropriate risk assessment of relatives.

To further explore this issue, we conducted a data analytics exercise using the ResearchOne service (www.researchone.org). ResearchOne provides anonymised extracts of records on TPP SystmOne, a clinical system used by more than 2500 practices in England. SystmOne includes features to code cancer diagnosis, family relationships, and family history of cancer. We studied how well these features were used in practice. At the time of the study there were over 4 million patient records in ResearchOne, of which 867 were for adults under the age of 40 years when diagnosed with breast cancer (669 female, 198 male). These adults were linked to their child's records using either a coded relationship link or a probabilistic algorithm. This algorithm identified children using a shared surname and address, age difference >15 years, patient for 5+ years while aged 0–16 and no looked after/adoption codes or flags. Each linked child record was then reviewed to check family relationships and history, and whether need for cancer screening was recorded.

Based on the national average of 1.7 children per adult[10] we expected to find 1474 children, but identified only 94 children using relationship codes and 288 using our probabilistic algorithm. For these 382 children of young adults diagnosed with breast cancer, only 117 (31%) had a family history recorded, coded as Family history of cancer (65), Family history of breast cancer (50), or Family history of neoplasm of breast (2). Only one child of the 198 male patients with breast cancer had a family history recorded.

## OPPORTUNITIES FOR AND BARRIERS TO IMPROVING THE CAPTURE OF FAMILY HISTORY DATA

It can take 15 to 30 minutes for practitioners to collect family history, and completeness depends on the individual's recall during the consultation.[11] Getting patients to record their family history online, outside of a pressured consultation and where they can harness the family's collective memory, may prove significantly more effective. In the US up to 20% of patients have demonstrated a willingness to do this online.[11]

The US national support for online family history tools has not been matched in the UK or elsewhere, but UK general practice can be proud of getting 100% coverage of the population using longitudinal, lifelong electronic health records.[12] Inspired by US online patient tools, UK leadership in lifelong electronic health records and emerging computerised decision support[13] can radically improve the early detection and prevention of multiple diseases where family history is a risk factor. Box 1 summarises the pros and cons of this approach.

## A WAY FORWARD

Patients have an interest in ensuring

## Box 1. The pros and cons of patient-captured family history

| Advantages | Disadvantages |
|---|---|
| Getting patients to record their family history online, outside of a pressured consultation and where they can harness the family's collective memory, may prove significantly more effective | The deterioration of patient memory and accuracy of information with time means that some people are unaware of or forget their family history. Time constraints, apathy, and reluctance to find out negative health information have been reported as barriers[3] |
| Public enthusiasm for genealogy has led to respected online tools, for example, www.ancestry.com for family pedigree trees, Facebook apps, and data exchange standards such as GEDCOM to support interoperability | These systems do not have the functions required to include coded medical history but they are evolving quickly |
| Self-service DNA testing and diagnostic screening services such as Werlabs in Sweden (werlabs.se) are also growing | Such services may not be accurate, nor respect privacy guidelines |
| Family members often collaborate using social media and other digital tools, and, as consumers, can be faster to embrace innovation than their healthcare providers | Linking online family history tools to clinical GP systems is not simple, but common interfacing mechanisms (for example, Endeavour CIM) are emerging |
| Patient-centred approaches reduce pressure on healthcare providers | Privacy may be an issue, especially if the proband[a] requests access to the GP record of another family member |
| Patients can be in control over what personal information they decide to share | Current consent models assume each individual's record is private to them. Debate has been concerned with sharing patient information between health organisations, and few studies have examined patient attitudes to sharing specific information with relatives |

[a]*Proband: a person serving as the starting point for the genetic study of a family.*

their information is kept up to date and in monitoring their own risks. Our proposal is to give patients the opportunity and the necessary information to manage their own family history, and also to support them with online tools that link directly to primary care clinical systems. To be effective such a solution should include:

- tools for drawing family trees;
- standards for coding relevant diseases, interoperability, and data exchange;
- strong security and authentication;
- consent models for sharing with family members;
- support for family members to collaborate;
- links to GP systems; and
- patient-centred and clinical decision support tools.

## FUTURE CAPTURE AND USE OF FAMILY HISTORY IN PRIMARY CARE

For GPs, identifying patients at risk is not a current priority among their many competing pressures. However, many patients are currently unaware that they are at increased risk of developing multifactorial genetic diseases. It is now time for a rethink. In the future, online, patient-driven tools will reduce pressures on consultations and patients will come to the GP with their history ready, for an appropriate detailed discussion. The family history will be used to develop personalised care plans and inform lifestyle choices to reduce family-history-related risk. This approach will also benefit targeted screening.

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**Ethical approval**
Medical ethics approval number for ResearchOne: NRES 11/NE/1084.

**Provenance**
Freely submitted; externally peer reviewed.

**Competing interests**
Susan Clamp and Owen Johnson are directors of X-Lab, an e-health software provider.

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**Acknowledgements**
This work would not have been possible without the assistance and support of Professor Rick Jones at the University of Leeds, who unfortunately passed away in the summer of 2014.

DOI: 10.3399/bjgp16X688273

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