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# Debate and Analysis

## Is it time for a rethink? Improving the Capture and Use of Family History Information in Primary Care.

Authors: Paul A Nathan, Owen A Johnson, Jeremy C Wyatt, Susan Clamp
Yorkshire Institute for Health Informatics, University of Leeds

Contact details paul.nathan@nhs.net
Dr Paul Nathan RCGP Innovation Fellow and General Practitioner. Hollybrook Medical Centre Littleover, Derby, DE23 3TX
Telephone 01332523300

**Case history**
Mrs X is 32 years old and worried. She wants to discuss the breast cancer risk to 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 grand-mother 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?
c) recommend referral to a genetics clinic?

**Family History and Early Diagnosis**
Current thinking recognises the value of using family history. It is less clear how general practitioners should capture and use this information in a world where advances in electronic medical record systems and genome sequencing are redefining what is possible. GP's have an important role to play in the early diagnosis of illnesses such as cancer, cardiovascular disease and dementia. In cancer for example, the UK's relatively poor outcomes have been attributed, in part, to reluctance by primary care physicians to investigate for possible cancer.[1]. In this journal, Rubin *et al* presented a model of pathways to treatment to improve early diagnosis by shortening the sequence of steps from "detection of bodily changes" through consultation and diagnosis to "start of treatment" (see Figure 1 in BJGP, August 2014, page 429)[1] In this discussion we make the case for an earlier step - the use of family history, properly managed through electronic health records, to inform risk assessment and pre-emptive diagnostic testing before "bodily changes" occur.

When patients present to discuss their genetic risk for a condition such as breast cancer their concern is often triggered by a relative becoming unwell or a family gathering where information is shared. However GPs currently do not use adequate means for collecting and using this data for assessing individual risk. Marmot[2] discussed the need to ensure all people

have access to screening to reduce health inequalities and Emery[3] argued that computer assisted decision support using electronic health records could empower GPs to assess genetic risk in primary care. The benefits of family cancer screening units was confirmed in a Cochrane review in 2012[4] however NICE familial breast cancer guidelines[5] do not suggest active identification of relatives at, even when patients are diagnosed under the age of 40 years. We believe current systems are inadequately used.

**Current Family History Capture and Use**

To investigate our hypothesis that family history is under recorded in electronic patient records we conducted a population-based study using an anonymous data set supplied by ResearchOne (www.researchone.org) of 4 million patient records extracted from the SystmOne electronic health record system widely used in UK primary care. Our focus was on breast cancer where studies suggest 5-10% of breast cancer is hereditary.[6] Our earlier work, determined that 29% of the disease register are at higher risk of breast cancer with many unaware of their risk.[7] There is an urgent need for proactive identification and screening for those at higher risk of breast cancer who require enhanced screening and possibly genetic counselling.[8]

In common with similar systems in the UK, SystmOne includes features to code cancer diagnosis, family relationships and family history of cancer. We analysed 867 adult patients who were diagnosed with breast cancer under the age of 40 (of which 669 female and 198 male). These adults were linked to the records of their children, identified either by a coded relationship link between records or probabilistic linkage. The probabilistic linkage identified children using an age difference > 15 years, shared surname, same address, patient for 5+ years at some point while aged 0-16 and no looked after / adoption related codes or flags. The linked children data was reviewed to see if family history was recorded correctly and whether screening for cancer was recorded in the children's records.

Based on the national average number of children from the 2011 census (1.7 per adult) we would expect to find 1474 children but only 94 children were appropriately recorded in the records using relationship codes with a further 288 inferred through our probabilistic linkage. For these 382 children of adults diagnosed with breast cancer only 117 had a family history recorded on their record (Family history of cancer (65), Family history of breast cancer (50), Family history of neoplasm of breast (2)). Our sample suggests poor collection and coding of parent child relationships and poor coding of family history risks even for breast cancer in adults under 40 where the family history risk is recognised as high.

**Improving the use of Family History**

Our investigation examined root causes for poor coding with a view to identifying opportunities for improvement. Clinical systems do include features for recording relationships, use of national family history codes and free text to explain who had the condition within the patient records with any additional information such as age of diagnosis, age of death, or metastatic disease. However these systems do not have the functions required to construct family pedigrees which include all the family medical history to make an appropriate decision about genetic risk for members of the family. Clinical systems need clinical decision support to bring such genetic information into clinical practice.[9]

There are no established means of sharing patient data between records and current consent models assume each individual's record is private to them. Existing recall systems do not transfer between GP systems and therefore even if patients are identified as being at risk in the future, there is no means of recalling patients who move between GP practices. Instead people rely on family members to remind them at the appropriate age. A further problem being the deterioration of patient memory and accuracy of information with time - people are unaware or forget their family history, particularly important for grandchildren.[10]

There are no nationally agreed approaches to recording this information in clinic letters, coding within the primary care clinical record and for ensuring appropriate risk assessment of affected relatives. One option is to use pedigree drawing tools within records.[11] There are many popular genealogy tools available online and it might be better if patients could construct their own family trees, linking to these their clinical records. Progress is needed developing consent models that will allow specific health data to link to pedigree drawing tools supported by decision support systems. Electronic data sharing of patient records between organizations might also be extended to include sharing data between records of relatives with their consent.

Genetic clinics will tell us they are now able to perform DNA testing for a variety of conditions. However patients still need to be identified in primary care in order that a referral to a specialist clinic can be made. Amongst the competing pressures placed on primary care, seeking to identify patients at risk is not a current priority. Many patients are not currently aware whether they are at increased risk of developing multifactorial genetic diseases. It is now time to rethink.

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