Journal of Community Genetics (2020) 11:413–420
https://doi.org/10.1007/s12687-020-00476-2

ORIGINAL ARTICLE

# Is family history still underutilised? Exploring the views and experiences of primary care doctors in Malaysia

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Received: 2 March 2020 / Accepted: 8 July 2020 / Published online: 14 July 2020


## Abstract

Family history has long been recognised as a non-invasive and inexpensive tool to identify individuals at risk of genetic conditions. Even in the era of evolving genetic and genomic technology, the role of family history in predicting individual risk for genetic testing and guiding in preventive interventions is still relevant, especially in low-resource countries. The aim of this study was to explore primary care doctors' views and experiences in family history taking and how they utilised family history in day-to-day clinical consultations in Malaysia. Four focus group discussions and six in-depth interviews involving 25 primary care doctors were conducted. Three themes emerged from the analysis: (1) primary care doctors considered family history as an important part of clinical assessment, (2) proactive versus reactive approach in collecting family history and (3) family history collection was variable and challenging. Family history was documented in either free text or pedigree depending on the perception of its appropriateness during the consultation. This study highlighted the need to improve the approach, documentation and the implementation of family history in the Malaysian primary care settings. Integrating family filing concept with built-in clinical decision support into electronic medical records is a potential solution in ensuring effective family history taking in primary care.

**Keywords** Family history · Risk assessment · Primary care · Qualitative

## Introduction

Genetic and genomic advances are expanding and being mainstreamed into clinical practice. Primary care doctors are increasingly exposed to these advances in clinical consultations, from the recognition of genetic carrier status, identification of familial cancer risk to modifying drug management in response to genomic advances. Primary care doctors, often the first point of healthcare contact, offer a unique opportunity to initiate genetic assessment, provide appropriate support and counselling and refer to specialists when appropriate; this allows timely risk mitigation intervention to be instituted (Qureshi and Raeburn 1993). Although genetic and genomic technology has evolved rapidly in the past decade, family history assessment still plays an important role in predicting individual risk for genetic testing and guiding in preventive interventions (Rich et al. 2004) particularly in resource-limited healthcare settings.

Taking family history (FH) is often the first step in identifying an individual with genetic risk. It is described as the 'gateway to recognise inherited disorders in a patient' (Bennett 2012). It is an inexpensive way to identify people who are at risk and a positive family history can inform decisions about genetic carrier testing (Bennett 2012). An ideal FH collects health information of at least three generations. Ancestry history forms an essential part of a family history questionnaire, and genetic conditions such as thalassaemia, sickle cell disease, cystic fibrosis and Tay-Sachs disease predominate in certain ancestry backgrounds (Johnson et al. 2006). Traditionally, FH has been taken in several steps

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including family medical questionnaires and patients are encouraged to contact relatives to obtain or confirm information (Bennett 1999). To ease documentation, FH is presented graphically in the form of a pedigree (Bennett 1999). A pedigree represents family members and their relationships using standardised symbols, which were introduced by the National Society of Genetic Counselors in 1995 (Bennett et al. 2008). In primary care, clinicians have been encouraged to document a 3-generation pedigree; however, there is limited evidence to support its clinical utility in primary care (Wolpert and Speer 2005). Taking thorough FH needs considerable time and effort; thus, it has been recommended that primary care doctors should accumulate a complete family history over several patient visits (Rakel 2007). Promising tools have been developed to serve this purpose, but more rigorous validation is needed before they can be used in clinical practice (Trotter and Martin 2007; de Hoog et al. 2014; Emery et al. 2014). The emphasis on using FH for risk stratification as one of the preventive strategies has been documented in various international guidelines for cardiovascular disease (Greenland et al. 2010), diabetes (American Diabetes Association 2011), breast cancer (Smith et al. 2012) and colorectal cancer (Levin et al. 2008). A systematic review reported that obtaining FH has increased the uptake and adherence to breast screening; however, limited evidence is available on the benefits of FH in other genetic conditions in primary care (Qureshi et al. 2009).

Despite the recognised importance of FH, challenges remain in implementing FH taking as part of routine care in a busy clinical practice. Some studies found that self-reported FH can be inaccurate and misleading (Hunt et al. 2001; King et al. 2002). Lack of time is a significant barrier to asking and documenting FH from patients (Trotter and Martin 2007). Studies have shown that the average time of collecting FH by primary care doctors is 17.9 min, and not all primary care doctors asked FH in their routine consultations (Acheson et al. 2000). Other reported challenges include the following: lack of training and skills to utilise three generation FH, failure to collect FH in a manner that is culturally sensitive to different ethnic groups and ensuring emotional appropriateness when asking about deceased family members (Fry 1999; Watson et al. 1999; Maradiegue and Edwards 2006). Although the use of pedigree has the advantage of having a standard structure, some doctors find it difficult to accurately interpret this information (Fry 1999; Watson et al. 2001).

To date, international guidelines have emphasised the importance of utilising FH in primary care; however, there is very little guidance on how to implement them in low-resource settings. Realising the importance of FH as one of the preventive strategies, we aimed to explore the views and experiences of taking family history among doctors working in the primary care settings from both public and private sectors in Malaysia. The findings from this study could potentially inform future interventions to improve clinical utility of FH in identifying individuals or families at risk of genetic conditions so that preventive strategies, i.e. lifestyle changes, and offering genetic testing can be initiated.

## Methods

### Participant recruitment

This study was conducted in three primary care settings in an urban area in Klang Valley in Malaysia: the public health clinics, public university–based clinics and private clinics. Malaysia has a dual-sector healthcare system: public and private sectors. The public sector, which comprises the public health clinics and public university–based clinics, is government-funded with patients receiving almost free or heavily subsidised healthcare services and treatment. In Malaysia, there were 878 public health clinics and they are mostly run by Family Medicine Specialists, primary care trainees and medical officers (Hwong et al. 2012). On the other hand, the private clinics operate on fee-for-service with patients paying out-of-pocket, through private health insurance or employer payment. It was reported that there were 5468 private clinics which were run by general practitioners (GPs), as either solo or group practice (Hwong et al. 2012). The participants were recruited using purposive sampling. They were invited if they have practised in the primary care setting for at least 1 year and have managed patients with genetic conditions in terms of screening, assessment, diagnosis or treatment. Primary care doctors who have special interest in different areas of preventive care, i.e. cardiovascular risks, women's health, antenatal or hereditary malignancies, were also invited. A few GPs with postgraduate qualifications in Family Medicine were also recruited.

### Data collection

Ethical approval was obtained from the University of Malaya Medical Centre Medical Research Ethics Committee and the Ministry of Health (Malaysia) Medical Research Ethics Committee. Every participant went through the participant information sheet before a written consent was obtained. In-depth interviews (IDIs) and focus group discussions (FGDs) were conducted at a mutually convenient time and clinics where the participants were practicing. The method (either IDI or FGD) was decided based on place of practice and participants' preference. IDIs were conducted among GPs as they worked in solo practices. While FGDs enrich the data through interactions among the participants (Krueger 1994), IDIs captured the views and experiences of primary care doctors who preferred to be interviewed individually. The participants in each FGD had a similar level of seniority in terms of working experience as a medical practitioner. All interviews

were conducted by the researchers who used the same semi-structured topic guide for both IDIs and FGDs. The topic guide was developed based on literature review, brainstorming among the researchers and the Theory of Planned Behaviour. The Theory of Planned Behaviour proposes that subjective norms, attitude and perceived behavioural control indirectly influence behaviour through intentions (Ajzen 1991). It helps to answer 'what is the practice and attitude', explains their practice ('why') and identifies the barriers and facilitators. The interviews were audio-recorded, transcribed verbatim and checked for accuracy. The length of the interviews ranged from 40 to 80 min. Field notes were taken during and immediately following the interviews to capture the context of the group discussions such as setting, group dynamics and ideas from the participants that were of vital interest (Krueger 1994). All the interviews were conducted between July 2017 and February 2018.

### Data analysis

Three researchers independently analysed each transcript, followed by a discussion of the analysis before a consensus was reached on the final analysis. Data were analysed using a thematic approach through open and axial coding. The researchers coded the transcripts line-by-line, followed by revising the coding scheme based on constant comparison across the transcripts and initial coding outputs. Once coding was completed, data were re-organized and condensed into categories and themes. The data were managed by the QDA Miner Lite Version 2.0.6 computer-assisted qualitative data analysis software programme.

## Results

### Demographic

Four FGDs and six IDIs were conducted with 25 primary care doctors. Each FGD involved between four and six doctors. The first focus group (FG-1) was attended by doctors who were undergoing final year of postgraduate training in Family Medicine in a public university–based clinic; the second focus group (FG-2) was attended by doctors who were undergoing the third year of postgraduate training in Family Medicine in a public university–based clinic; the third focus group (FG-3) was attended by medical officers who were working at a public health clinic; and the fourth focus group (FG-4) was attended by Family Medicine Specialists who were working at a public health clinic. IDIs were conducted among GPs who were working in private clinics. The participants were largely female and were from age group of 30 to 40 years. The majority has worked in primary care for at least 5 years. Many of them were undergoing postgraduate training in Family Medicine ($n = 11$) (see Table 1 for demographic and characteristics of participants).

### Themes

Three key themes emerged from the data and they are described below with quotations.

## Theme 1: taking a family history is important

The participants consistently agreed that FH is an important part of clinical assessment, and primary care doctors have a role to play. FH helps to assess patients who are at risk of genetic conditions as part of screening and for early intervention to be carried out. The doctors acknowledged that FH

**Table 1** Sociodemographic profile of participants

| | $N = 25$ | Percent |
|---|---|---|
| Age (years) | | |
| 30–39 | 20 | 80 |
| 40–49 | 3 | 12 |
| 50–59 | 1 | 4 |
| 60–69 | 1 | 4 |
| Gender | | |
| Male | 7 | 28 |
| Female | 18 | 72 |
| Years from medical school graduation | | |
| < 10 | 12 | 48 |
| ≥ 10 | 13 | 52 |
| Years of working in primary care | | |
| ≤ 5 | 10 | 40 |
| 6–10 | 12 | 48 |
| 11–15 | 0 | 0 |
| 16–20 | 2 | 8 |
| ≥ 21 | 1 | 4 |
| Health system sector | | |
| Private | 5 | 20 |
| Government | | |
| Public health clinic | 9 | 36 |
| Public university–based clinic (primary care trainees) | 11 | 44 |
| Practice type | | |
| Solo | 3 | 12 |
| Group | 22 | 88 |
| Postgraduate qualification in Primary Care Medicine | | |
| Yes | | |
| Completed | 9 | 36 |
| Undergoing primary care training | 11 | 44 |
| No | 5 | 20 |

helps to identify conditions that are known to be hereditary, and health promotion should be instituted when familial risk is identified.

> For me it is, it is very important, because from the family, genetic is like a 'subset' of the family history, by taking a family history, several family members who have sudden syncope or sudden death then you can maybe relate to a cardiac condition. If you find a few family members have cancer, then you would like to think of familial cancers (Dr Z, trainee, FG 1)

> It is very important to ask…., if the patient comes to see us for upper respiratory tract infection but has background family history of diabetes, maybe from there we can advise on lifestyle intervention (Dr Z, FMS, IDI 2)

> I encourage them to do regular annual screening and also, checking on their parents. If there is anyone in the family lines with medical problems, this will be an opportunity for us to screen (Dr R, GP, IDI 1)

> If I want to ask family history for all patients, then it will be family history related to common diseases (Dr A, MO, FG 3)

> I usually ask the family history of diabetes and hypertension because that's the most common (Dr M, GP, IDI 3)

FH was taken as part of routine assessment of a newly registered patient at the private clinic.

> We have the operating procedure, we take most of the patients' information at the registration, we take their simple biodata and their medical history including their current medication and also, their family, if possible. If you see our notes that will be caught during the registration. (Dr R, GP, IDI 1)

'Family filing concept' was mentioned as a method to capture the risk of genetic conditions among family members.

> We have the family filing, and put together family names, so when they come in, ok what happen to your grandmother? (Dr R, GP, IDI 1)

## Theme 2: proactive versus reactive approach in collecting family history

Although primary care doctors considered FH as an important part of clinical assessment, FH was not collected consistently and systematically; the doctors collected FH either proactively or reactively. They did not take FH routinely in all patients but only if they felt it was necessary or relevant to the patient.

### Proactive approach

They would proactively take family history when patients come for annual medical screening in which FH is part of the screening questions or if the patients are attending the clinic regularly.

> …..I don't usually ask unless required for example full medical check-up… (Dr M, GP, IDI 3)

> I ask more if they are regular patients or registered to my clinic as a family unit. (Dr R, GP, IDI 1)

The prevalence of the condition was an important factor in influencing whether primary care doctors would ask for FH; family history of multifactorial genetic conditions like diabetes and cardiovascular diseases was the most asked.

### Reactive approach

FH was asked only if the doctors felt that the symptoms are relevant or related to specific genetic conditions, not routinely.

> Maybe I didn't really ask every patient do you have a family history of cancer, unless they have some symptoms (Dr L, trainee, FG 1)

> If the patient comes to the clinic only for an acute problem, to be frank, I don't take the family history.
> It felt very odd if I ask the patient's family history when they come for upper respiratory tract infection or when the patient was about to leave the clinic, suddenly we ask family history! (Dr R GP, IDI 1)

## Theme 3: family history collection was variable and challenging

The content and method of asking and documenting family history varied across participants. Primary care doctors tended to ask FH as a general question and would usually asked up to first-degree relatives.

I just ask in general whether the parents or siblings had any medical conditions or any death and its cause in the family. (Dr B, Medical Officer, FG 3)

I think we just ask about what is your immediate family, your father, your mother then maybe we go up to grandmother, grandfather. (Dr L, Medical Officer, FG 3)

While primary care doctors recognised the importance of documenting FH, they seldom drew a pedigree diagram in their actual practice. The cited reasons such as the electronic medical record system were not user-friendly and did not support the drawing of a pedigree diagram. For some, documenting a pedigree diagram was not considered important because these conditions were uncommon in their primary care practice.

I think the most common would be thalassemia because we have been screening in the health clinic, so usually when we deliver the result, drawing (pedigree) would help them to understand better, I feel. (Dr D, Medical Officer, FG 3)

Just the only thing…in teleprimary care (TPC), we can only type. We cannot draw, so that is a little bit difficult, so that's why we write everything (Dr A, Medical Officer, FGD 3)

Personally, I don't think it is effective at all, it doesn't add any value for me. I think I very seldom treat single gene in primary care, even haemophilia we don't treat, that's why I think drawing the pedigree it's not really necessary. (Dr L, Medical Officer, FG 3)

Some doctors found drawing a pedigree difficult and time consuming. They also found it challenging to elicit a detailed family history from patients who had difficulty recalling their FH.

Some people who do not do it regularly it can become time consuming to draw it and explain it to the patient. (Dr S, Family Medicine Specialist, FG 4)

I find it a challenge, of course it would be very difficult to draw a very nice family tree for a patient and you know, it's not always easy. (Dr Z, Family Medicine Specialist, IDI 2)

Sometimes the patient might not be able to recall of hand at the period of time whether their cousin or their aunty or their great grandmother had something similar. (Dr C, Medical Officer, FG 3)

## Discussion

This study found that primary care doctors considered FH as an important genetic risk assessment tool in the primary care settings. However, the approach to taking FH during consultations was either proactive or reactive. Collection and documentation of FH also varied, and FH was documented either in a free text or by pedigree depending on their perception of its appropriateness during the consultation. Lastly, several challenges were identified when collecting FH such as lack of support and skills, time and patients' difficulty in recalling information.

The primary care doctors tended to ask for FH of conditions that are prevalent such as multifactorial genetic conditions commonly managed in primary care such as diabetes and cardiovascular diseases. This is consistent with other studies which also found that FHs of cardiovascular and diabetes were the conditions routinely asked (Mathers et al. 2010; Daelemans et al. 2013; Endevelt et al. 2015). This study reveals that patients, who require annual medical screening and patients on regular follow-ups, were the main targets for collecting FH; this is unlike the study conducted by Acheson et al. (2000) who reported that FH was discussed with new patients rather than established patients (Acheson et al. 2000). Regarding the reactive approach, primary care doctors enquire FH only if it is relevant to the patient's reasons for clinical consultation even though they are newly registered. While this 'reactive' approach to taking FH might seem appropriate and tailored to the individual patient, it seemed that the primary care doctors perceived taking FH as a practice that is 'good to have' rather than a 'must have'. This is likely to occur in setting with heavy patient load, where an average consultation time was reported to be 12–13 min in a Malaysian outpatient clinic (Raja Lexshimi et al. 2009).

Another important finding from this study is the variation in the enquiry and documentation of FH, from asking general health conditions among family members and writing down in free text to drawing a pedigree. Wood et al. (2008) and Daelemans et al. (2013) also agreed that the approach in taking FH varied considerably where most would ask general questions about health conditions of family members (Wood et al. 2008; Daelemans et al. 2013). Daelemans et al. (2013), in her study in Belgium, noted that the participants captured FH in free text, and none of the participants used pedigree (Daelemans et al. 2013) due to the lack of protocol or tools to assist in FH taking and documentation in the clinics (Wood et al. 2008; Daelemans et al. 2013). Similarly, the practice of documentation also differs. While an ideal family history collects information of at least three generations, primary care doctors in this study asked only up to first or, at most, second degree (up to grandparents). Inadequate FH taking and documentation may lead to inaccurate assessment and, hence, provide incorrect advice to patients. The views and practices of

using a pedigree are also contradictory; while the visual presentation might facilitate patient's understanding, it is often practised when needed to explain rarer genetic conditions which are seldom encountered in the primary care setting. Another approach found by this study is to collect FH using the family filing concept where information from medical records of family members is used to identify individual FH. However, there are serious concerns in the risk of breaching patient confidentiality and autonomy when doing this. A more pragmatic and safer approach to asking and documenting individual FH is needed. For example, using technology, such as artificial intelligence to 'crawl' the electronic medical record for relevant information pertaining to FH and hereditary conditions, is promising and might be more cost-effective (Emery 2015).

Despite the unanimous agreement on the importance of FH, it is often not practised in the real-world clinical setting across all health sectors (public, public university–based and private sectors). The barriers to taking FH routinely among primary care doctors in this study were similar to those in other studies. Primary care doctors do not have the necessary skills to take and document FH competently, the process is time-consuming, there is a lack of guidelines and simple clinical support tool and patients recall difficulty. An earlier study had stated that there is a lack of training among primary care doctors (Watson et al. 1999), and this could be an important factor that contributes to lack of skill and confidence.

Primary care doctors acknowledged the benefits of FH; however, our study did not provide evidence on how using FH has helped them in their clinical practice. To overcome this mismatch between attitude and practice, it is important to emphasise the value of FH and the importance of appropriate documentation and interpretation of FH to the primary care doctors. It is apparent that focused educational effort is required to enhance primary care doctors' confidence and skills not only in collecting but also in interpreting FH. In addition, studies have found that FH takes time and there is a lack of effective tools and practical guidelines to assist primary care doctors in FH taking (Wood et al. 2008; Williams et al. 2010). Therefore, interventions have been developed to tackle these implementation challenges. Firstly, Watson et al. (1999) recommend that greater emphasis should be placed on genetics in medical education (Watson et al. 1999). Secondly, in the context of a busy primary care clinic, a self-administered screening questionnaire on family history can be used to identify patients with genetic risks who warrant further investigations (Qureshi et al. 2005; Emery et al. 2014). Thirdly, the use of technology to record family history via an online system may not only help to reduce time for documentation but also harness the family's collective memory of any inherited conditions (Murray et al. 2013). The idea of compiling FH of all family members in one file may be used to reduce errors due to patient recall bias. Rich et al. (2004) suggest that healthcare providers can clarify with another family member or via retrieval from medical records of family members (Rich et al. 2004). However, this may have potential confidentiality issues.

## Strength and limitation of the study

To our knowledge, this study is one of few studies conducted in the low and middle-income countries to explore primary care doctors' experiences, perceptions and barriers when taking and documenting family history. To ensure maximum variation, participants from different background, practice setting and experience in primary care were recruited. However, all information captured during the interviews was self-reported, and the doctors' actual practice on FH collection was not observed.

## Practical implication

Accurate FH documentation is crucial if it were to be used as a tool for health promotion and predictive genetic testing. Even with the advances of genetic and genomic technology, the role of family history is still relevant especially in the low-resource countries. There is a need to improve the approach, documentation and the implementation of FH in the primary care settings. A three-generation pedigree which is a graphic presentation of a family history has been reported as a comprehensive way to assess risk and patterns of inheritance of genetic conditions (Wattendorf and Hadley 2005). Its use should be considered in primary care to decide the appropriateness of genetic testing. A user-friendly template for taking FH needs to be developed and tested. This could be in the form of a checklist captured during a consultation that will automatically generate a pedigree in the electronic medical record. This will save up time as drawing pedigree manually may take up to 30 min (Frezzo et al. 2003) which is not feasible due to time constraint in primary care consultations. Although the clinical evidence is still lacking on whether pedigree is superior to documenting family history in text, a graphic presentation of a family history would capture the significance of familial risk (Wolpert and Speer 2005). Hence, a pragmatic approach should be the way forward to ensure that pedigree is implemented effectively in primary care.

Another suggestion to improve the use of FH would be a clinical decision support system that mirrors the family filing concept that could be built to manage family members with genetic risk. However, confidentiality, ethical and legal issues must be evaluated before its implementation as it involves other individuals within the family.

The importance of FH as a mean of identifying genetic inheritance must be re-emphasised in clinical practice, and in

particular, the medical curriculum of undergraduates and postgraduates. Regular, continuous medical education (CME) would be an avenue to improve the skills, confidence and practice in FH taking especially documentation and interpretation. This could be carried out as hands-on workshops and educational outreach.

Ultimately, it is vital that current national guidelines on genetic conditions, for example, familial breast or colon malignancies and familial hypercholesterolemia enforce the use of FH as a prime tool for the identification of those who could benefit from more intensive preventive strategies.

## Research recommendation

An effective family history tool should be developed to facilitate doctors to collect FH. An existing validated FH questionnaire in primary care (Emery et al. 2014) could be adapted to local setting as a guide. Potentially, this can be embedded into a clinical decision support system on family history assessment. Once this is developed, it should be validated and tested to improve the quality of care in genetic risk assessment and management.

## Conclusion

This study has highlighted the views and experiences of primary care doctors on FH utilisation in primary care in Malaysia. Improved ways of taking and documenting FH must be carried out to facilitate its entry and use in clinical practice. Electronic medical record integrating family filing concept with built-in clinical decision support is a potential avenue for FH collection and documentation. Reinforcing FH in national clinical guidelines and conducting educational programmes in genetic risk assessment would be beneficial to refine primary care doctors' skills, confidence and practice.

**Acknowledgements** The authors would like to thank all primary care doctors who have participated in this study.

### Compliance with ethical standards

**Conflict of interest** The authors declare that they have no conflict of interest.

**Ethics approval** Ethical approval was obtained from the National Medical Research & Ethics Committee (Malaysia) KKM/NIHSEC/P16-1766. Informed consent was obtained from all patients for being included in the study. This study was performed in accordance with Helsinki Declaration of 1975, as revised in 2000.

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