Awareness of familial hypercholesterolaemia in Australian primary care

A qualitative descriptive study

Caroline Bulsara, Tom Brett, Jan Radford, Clare Heal, Gerard Gill, Charlotte Mary Hespe, Cristian Vargas-Garcia, Ian W Li, David R Sullivan, Alistair W Vickery, Jing Pang, Diane Arnold-Reed, Dick C Chan, Gerald F Watts

Background and objective
A lack of public and health professional awareness about familial hypercholesterolaemia (FH) leads to an estimated 90,000 Australians remaining undiagnosed. The aim of this study was to establish the level of knowledge and awareness of FH in Australian general practices.

Methods
A qualitative descriptive methodology was used to explore baseline knowledge and perceptions of practice staff about diagnosing and managing FH. Overall, 63 interviews were conducted with general practice staff at 15 practices taking part in a National Health and Medical Research Council partnership grant study (GNT1142883).

Results
Data were analysed thematically and coded into themes – knowledge/awareness/recall, management, use of guidelines/referrals, and contacting family members. Most general practitioners treated the high cholesterol component as their primary focus. Guidelines and referrals were rarely used.

Discussion
This research reflected a lack of knowledge, awareness and use of guidelines similar to that shown in other published studies. Improved primary care infrastructure, knowledge and awareness of FH need to be addressed.

Since atherosclerosis caused by familial hypercholesterolaemia (FH) begins in childhood and continues into adulthood, there is an urgent need for early identification and preventive treatment.1 Younger people have most to gain from this, as it enables the prospect of a normal lifespan and avoidance of premature atherosclerotic coronary artery disease (CAD).2–4 The latent period from birth to the onset of CAD in midlife is sufficient for appropriate treatment to be commenced to prevent development and progression of atherosclerosis.

Despite recent exponential growth in research on the disorder, there remains a general lack of public and health professional awareness about FH.5–9 Fewer than 10% of affected individuals in Australia are diagnosed, with most remaining undertreated.8 This lack of diagnosis and treatment exposes these individuals and their close relatives to a high risk of CAD that could be effectively prevented.2,10

Over 88% of Australians consult their general practitioners (GPs) at least once every 12 months,11 offering unique opportunities to detect the disorder early while patients are still young and asymptomatic. FH screening can be undertaken in the community primary prevention setting.12 Such opportunistic screening using low-density lipoprotein cholesterol is commonly employed in general practice but may not always be used effectively to distinguish patients with FH from those without, especially in adults.

A lack of awareness of FH among health professionals (eg GPs, cardiologists and other non-GP specialists) as well as in community and family settings is a major reason for the poor uptake in diagnosing and managing FH in Australia and worldwide.11,12 Little attention has been focused to date on screening, diagnosis and management of FH in general practice, where most affected patients can be identified.12 The lack of suitable infrastructure in primary care to undertake systematic testing for FH remains a major limitation.12 The need for a radical shift and evolution in general community and health professional perceptions of FH and the effect on children and young adults is recognised.1
While evidence strongly suggests FH remains underdiagnosed in primary care, the underlying reasons as to why this continues are unknown. The aim of the present study was to explore the level of knowledge and awareness about FH in several Australian general practices.

Methods

Study sample and setting

The participating practices were involved in an Australia-wide National Health and Medical Research Council (NHMRC) partnership grant study (GN1142883) into ‘Improving the detection and management of familial hypercholesterolaemia in Australian primary care’.14,15

A total of 63 interviews with practice staff were undertaken.16 Fifteen general practices across five Australian states were involved in the study, and all were participating in the planned education intervention after the baseline interviews were conducted. Five practices were from Western Australia, four from New South Wales, three from Queensland, two from Tasmania and one from Victoria. Nine of the practices were in larger metropolitan areas (both inner city and outer metropolitan), while six were in smaller towns and rural areas.

Recruitment and data collection

The selection of practices involved in the study was based on several factors including geographic location, rural–urban spread, availability of Best Practice software and the willingness and ability of the practice staff to become active participants.14

Data collection was accomplished using a semi-structured interviewing technique with GPs, practice nurses (PNs) and practice managers (PMs) to explore any barriers and/or enablers to diagnosing the condition, as well as their current role (if any) in managing the condition. Interviews were audio recorded and transcribed verbatim. Overall, 55 GPs and eight PNs and PMs participated in the study. As a result of the small numbers of PNs and PMs, the results were combined as one for PNs/PMs for this qualitative phase of the study. The data were collected prior to commencement of the NHMRC study14 and represent the initial phase of the study.

Interviewers sought participant feedback on their level of knowledge and awareness about FH prior to delivery of the educational component on the disorder at the study commencement.14 The interviewers also explored whether participants were aware of any patients attending their practice with an established diagnosis of FH and how they might manage such patients.

Participants were also asked about how they would typically manage a patient with known high cholesterol, if they were aware of any specific guidelines for managing FH, and whether they had ever referred such patients to a lipid specialist. Finally, participants were asked for their perceptions and experiences related to cascade testing and screening of patients for FH.

A qualitative descriptive study methodology16 was used to explore the baseline knowledge and perceptions among GPs, PNs and PMs about diagnosing and managing FH.

The qualitative descriptive methodology used by Sandelowski16 enables the researcher ‘to stay close to their data and to the surface of words and events’. Theoretically driven methodologies, such as phenomenology, use interpretative analysis of data to determine key themes. Conversely, qualitative descriptive methodology does not interpret the data, but instead provides a rich descriptive portrayal of an event as told directly by the participants.

By employing this approach, the data are not unduly influenced by the interpretation of the researcher.16,17 Qualitative descriptive methodology is suitable for healthcare research as it helps to focus research questions directly on the experiences of the participants rather than through a more theoretical lens.18

Data analysis

Data were analysed thematically using NVivo software, version 12 (QSR International). A template for the thematic analysis19 of the data was

<table>
<thead>
<tr>
<th>Table 1. Level of confidence and knowledge in familial hypercholesterolaemia diagnosis and management</th>
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<table>
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<tr>
<th>Practice role</th>
<th>Poor confidence/knowledge (%)</th>
<th>Little confidence/knowledge (%)</th>
<th>Moderately confident/knowledge (%)</th>
<th>Somewhat confident/knowledge (%)</th>
<th>Very confident/knowledge (%)</th>
<th>Not applicable/no response (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Own knowledge about FH</td>
<td>GP (n = 55)</td>
<td>5.5</td>
<td>49.1</td>
<td>29.1</td>
<td>14.5</td>
<td>1.8</td>
</tr>
<tr>
<td></td>
<td>PN/PM (n = 8)</td>
<td>62.5</td>
<td>12.5</td>
<td>25.0</td>
<td>–</td>
<td>–</td>
</tr>
<tr>
<td>Confidence in diagnosing FH</td>
<td>GP (n = 55)</td>
<td>12.5</td>
<td>12.5</td>
<td>12.5</td>
<td>12.5</td>
<td>–</td>
</tr>
<tr>
<td></td>
<td>PN/PM (n = 8)</td>
<td>10.9</td>
<td>30.9</td>
<td>38.2</td>
<td>9.1</td>
<td>3.6</td>
</tr>
<tr>
<td>Confidence in managing FH</td>
<td>GP (n = 55)</td>
<td>5.5</td>
<td>16.4</td>
<td>30.9</td>
<td>36.4</td>
<td>5.5</td>
</tr>
<tr>
<td></td>
<td>PN/PM (n = 8)</td>
<td>25.0</td>
<td>25.0</td>
<td>12.5</td>
<td>–</td>
<td>12.5</td>
</tr>
</tbody>
</table>

FH, familial hypercholesterolaemia; GP, general practitioner; PM, practice manager; PN, practice nurse.
Table 2. Main themes, interview questions, brief explanation and exemplar quotes

<table>
<thead>
<tr>
<th>Theme</th>
<th>Explanation</th>
<th>Exemplar quotes</th>
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<tbody>
<tr>
<td>Knowledge, awareness and recall</td>
<td>Unsure of diagnosing FH</td>
<td>‘... I would have thought about it, ... in terms of the confidence, diagnosing it is probably where I lack, yeah.’ [GP, NSW, metro]</td>
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<tr>
<td></td>
<td>Patient youth a major factor in suspecting FH</td>
<td>‘Often if a young person with elevated cholesterol, they knew that there was some inheritance ... it was a matter of those factors.’ [GP, NSW, metro]</td>
</tr>
<tr>
<td>Management</td>
<td>Graded approach – lifestyle modifications before medication</td>
<td>‘... maybe not jump at medications straight off ... talk to them about what we can do, ... what can contribute in terms of diet modifications and exercise, ... activities that can help change it, and set some goals in terms of time.’ [GP, Qld, rural]</td>
</tr>
<tr>
<td></td>
<td>Use of statins to manage high cholesterol</td>
<td>‘... if they reached lipid-lowering guidelines and hadn’t responded to lifestyle modification ... I would be putting them on a statin and referring for secondary opinion if I suspected FH. But I would be initiating statins if it was at that level, yeah.’ [GP, Qld, rural]</td>
</tr>
<tr>
<td></td>
<td>Manage as for cholesterol</td>
<td>‘Pretty similar to standard cholesterol patients in that it’s about compliance issues a lot, watching their risk factors, because there is nothing they can see or feel.’ [GP, Qld, rural]</td>
</tr>
<tr>
<td>Use of guidelines and referrals</td>
<td>Lack of familiarity with guidelines</td>
<td>‘I know there are guidelines ... I searched them for [a] patient who has FH but I can’t remember where they came from, but I know they exist.’ [GP, NSW, metro]</td>
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<tr>
<td>Contacting family members</td>
<td>No follow-up by GP of family but rather to make the patient aware</td>
<td>‘It is hard to follow up family members because it is not necessarily the whole family who come and see the same GP. I ask the family straight through the patient.’ [GP, Qld, rural]</td>
</tr>
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</table>

FH, familial hypercholesterolaemia; GP, general practitioner

created using a priori codes taken from interview questions to create the skeleton code frame on which to base the coding structure. The question guides are listed in the study protocol.¹⁴

Ethics
This study was approved by The University of Notre Dame Australia Human Research Ethics Committee Protocol ID: 016067F.

Clinical Trial Registration Number
The Clinical Trial Registration Number is 12616000630415.

Results
Participants were initially asked for their self-perceived confidence and knowledge of FH with the following questions:
- How confident are you in diagnosing FH?
- How confident are you in managing a patient with FH?

Responses were rated from 1 (poor) to 5 (very confident) and can be found in Table 1. Overall, GPs were more confident even at baseline regarding competencies with FH. Although some PNs/PM noted that they had some knowledge, diagnosis and management were usually outside of the scope of their roles within the practice.

Subsequently, the semi-structured interview data were coded into key themes using the questions asked by the interviewer. The key thematic areas are briefly summarised in Table 2.

Knowledge, awareness and recall
Overall, relatively few GPs and PNs across the 15 practices could recall caring for a specific patient with FH. The diagnosis of FH was perceived to be ‘opportunistic’ for some:

The other [patient] was having cholesterol done as part of a workup to be prescribed

Some participants considered FH to be one of the top ‘easily missed’ diagnoses. At times, the younger age of the patient was the only factor that would raise concern for a GP, especially when cholesterol levels were high.

Testing would not normally be performed unless the patient talked about a family history or the patient was younger. The younger age range of patients with Roaccutane, which is an acne medication that you have to check their cholesterol, so it was just incidentally found. [GP, Qld, rural]

I said, ‘Look, I definitely would just (check) because that’s quite young’. But I said to him to speak to the doctors because that’s not really my decision – [it’s] the doctor’s decision to say that really. But definitely, I would definitely get them checked and he was like, ‘Oh ok, I’ll consider that’. [PN, WA, metro]
FH proved problematic for some GPs in terms of ‘getting patients to take it seriously’ as something ‘more than high cholesterol’, and they identified that this could prove challenging to manage on an ongoing basis. One PN endorsed this by commenting on a patient that she had telephoned regarding their result:

Oh they knew they had FH and they called up for the result, we advised the result and advised Dr [redacted] to see them and they said, ‘Oh well I’m not coming in because I’m not going on medication’, so they knew exactly what the problem was but they were just adamant that, they were one of those patients that just went ‘No, I’m not taking this’. [PN, WA, metro]

Many GPs expressed confusion about making the diagnosis of FH. A few who had prior experience in managing a patient with the condition felt more comfortable with it. In the absence of such prior experience, most GPs treated for high cholesterol instead, with one remarking:

I wouldn’t have even thought of it, I would’ve just carried on treating people like I do for high cholesterol and blaming them rather than their family. [GP, WA, metro]

So I look out for what I would believe the signs of it are but I haven’t picked up someone who I’ve thought, ‘Yep, they have familial hypercholesterolaemia’. [GP, Vic, metro]

One GP regarded the diagnosis as a balance between ‘opportunistic’ case finding for some patients and targeted case finding ‘through genetic screening processes’ for others. Beyond this, there was little mention made by GPs and PNs of any patients who had undergone genetic screening. Only a few GPs were aware of patients who had been genetically tested. Of those who were aware, some reported that often the patient was reluctant to continue with testing over the longer term.

Management

The management of FH was perceived by GPs as a lengthy process commencing with the lowering of cholesterol levels:

I’d initiate treatment including lifestyle modification depending on their risk factors and their clinical setting. I would possibly initiate statin treatment as a first line, though it differs if it’s for primary or secondary prevention. It is a lot more aggressive for secondary prevention. [GP, Vic, metro]

However, at baseline for this study, the screening and treating of patients for high cholesterol was still the primary focus among the GP respondents. Lack of awareness of FH guidelines or FH patients who ‘can’t take statins at all or react’. In such circumstances, they would refer patients to a cardiologist or endocrinologist for advice.

Another GP reported only referring patients who ‘can’t take statins at all or react’. In such circumstances, they would likely refer them to a specialist cardiologist or endocrinologist. The study also found that GPs were largely proficient in using the Dutch Lipid Clinic Network Criteria but were generally unaware of any FH specific guidelines for its use in primary care settings:

Yeah, I wasn’t aware of any guidelines. [GP, WA, metro]

... but I haven’t used any tool to make a diagnosis ... so certainly if they have secondary issues ... like diabetes or cardiac problems ... I use the general guidelines but nothing specific to familial hypercholesterolaemia. [GP, WA, metro]

Yet, most participants stated they were unaware of any specific FH guidelines, with many GPs using their ‘own methods’ for diagnosing and managing the condition. Referral to a lipid specialist was not undertaken by most GPs, who instead preferred to refer patients to a cardiologist or endocrinologist for advice.

That is always fun and that changes throughout their age groups, especially if you have got them when they are really young and you are trying to get them through those teenage years, it’s probably not quite as difficult as some of our other chronic medical conditions but you know when all your peers are eating Pizza Hut and MacDonald’s ... it is working with them and then trying to adjust things as they go along. [GP, Qld, rural]

Many GPs tended to have a graded approach to managing high cholesterol, often commencing with lifestyle modifications unless there were other risk factors. Diagnosis and management were perceived as long-term management strategies. Statins were only prescribed if lifestyle interventions had not been successful in lowering lipid levels or in modifying patient risk over a period of 3–6 months. Most respondents felt the best course of action was to proceed with a graded approach to managing the patient’s high cholesterol:

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To be honest I am not really familiar. I read about FH when I was in the hospital setting, you know the basic physician training I went through, but at the moment there is no trace detail of the FH. [GP, Qld, rural]

Contacting family members
GPs and PNs were also asked about the importance of notifying family in terms of FH risks and their subsequent management. One GP (WA, metro) said that there was an increasing awareness of FH and that ‘we are generally becoming more aware of this category of patients and certainly we would spend more time and effort’ on it. In addition, asking patients about notifying other family members about FH was somewhat ad hoc during a consultation:

No, to be honest, not, because some of them can go to a different practice but usually I encourage them to talk to the family and, as you are aware as well, some of them [may] not be talking to family members and not likely to talk to them. [GP, WA, metro]

There was also an assumption that the cascade testing component would be ‘handled’ entirely by the lipid clinic. One GP felt that family reluctance was ‘just human nature’, while another believed that the length of time taken to offer cascade testing and await results was a barrier for some patients and commented:

So most people when you offer them [advice about] anything that occurs more than five years ahead ... they completely ignore it and discount it, so if you offer them a high reward in five years’ time compared with now, they still don’t bother so this is human nature. [GP, Qld, rural]

GPs perceived that the costs of genetic testing were the reason for reluctance to be tested by some lower income families. One GP spoke of practising in an area with a higher socioeconomic status where most patients were willing to go through the process of notifying family members and being tested.

Some GP respondents said that they were aware of the importance of family screening and testing and would check children as well. Some, however, reported little experience of managing children with high cholesterol, especially given the small population in this category, although they would still test and follow up on any higher cholesterol readings. It was acknowledged that treatment for FH is ‘different from others’ and that it would have to be treated ‘more aggressively’.

Discussion
The qualitative descriptive methodological approach in the pre-education phase of this study revealed that participating GPs had limited awareness and knowledge of the detection, diagnosis and management of FH. Instead, GPs tended to focus more on lowering high cholesterol levels by using statins and encouraging improvement in lifestyle measures for most of these patients. The hereditary component of FH with its ongoing, raised lifetime cholesterol burden from birth was less well appreciated in comparison to the elevated cholesterol levels detected as part of routine medical care during midlife.

Kwok et al. examined the knowledge and awareness of FH among GPs in the north west of England and found they almost universally considered themselves to have a key role in the early recognition of undiagnosed patients with FH in the community. However, gaps existed in their knowledge of FH inheritance and its increased cardiovascular risk.

Pang et al. also examined knowledge and awareness among primary care physicians in the Asia Pacific region. They found a lack of awareness of FH management guidelines, while the physicians’ knowledge of prevalence, inheritability and cardiovascular risk were also suboptimal. The findings from both Kwok et al. and Pang et al. broadly reflect similar findings to the present research.

A UK study by Weng et al. showed that an intervention to identify and manage patients with FH in a primary health setting could be successfully adopted. Their research revealed improvements in best practice for identification and management of FH following an educational intervention among participating general practices.

Findings from the present study showed that the process of managing high cholesterol in patients was graded and protracted, with the GP commencing with lifestyle interventions first (diet, smoking avoidance, exercise) and then following up after a few months to see if those lifestyle measures were successfully implemented. GPs noted an element of ongoing resistance among some patients to statin use.

Previous research has noted that GPs are largely proficient in using the Dutch Lipid Clinic Network Criteria, indicating that the lower rates of diagnosis were more likely attributed to a need for greater education in general practice about diagnosing FH rather than a fault in the diagnostic processes per se.

A greater logistical barrier to managing FH was found in the process of family screening and cascade testing. The current system of contacting close family members is reliant on the patient informing relatives who may or may not be attending the same practice. Studies in the UK and Australia provide a cost–benefit analysis of using specific FH services to not only reduce the lifetime costs associated with the condition, but also to raise the quality of life and survival gains for those affected.

The lack of awareness at the health professional and patient/family levels about the essential hereditary nature of the FH condition (ie that it had a familial/hereditary component as well as the markedly raised cholesterol component) was one of the key concerning findings from the present study. While lifelong treatment with lipid-lowering medications – such as statins in addition to diet and lifestyle modifications – is the key to successful management of FH, such knowledge was not always well appreciated in the participating practices at the time the study commenced.

The present findings are generally consistent with and complementary to Hardcastle et al., who examined Australian patients’ perceptions and experiences of FH and found that many tended to dismiss the serious nature of FH and the importance of lifestyle changes, preferring instead to rely on medications to maintain adequate control.
Awareness of familial hypercholesterolaemia in Australian primary care: A qualitative descriptive study

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Limitations
The present study is limited in that it only reflects the responses of the 63 practice staff interviewees at the 15 practices involved in the study and these may not be representative of other practice staff across Australia. Busy work schedules meant not all potential staff members could be interviewed, and their responses and approaches may differ.

Conclusion
Lack of awareness of the essential hereditary nature of FH combined with a lack of physical and human resources infrastructure to support better screening, diagnosis and management are key elements confronting general practice approaches to improved management of the condition. In addition, the logical progression to cascade testing of first- and second-degree relatives once new index cases are identified represents another major barrier that will need to be addressed if primary care is to optimise its potential in this important underrecognised and undertreated condition.

These qualitative findings have implications for improving health service delivery for FH in the primary care setting. This study has highlighted the need for greater education to improve knowledge and awareness about FH for primary healthcare teams (GPs, PNs and PMs) as well as patients and their families. Community conversations involving patients and families with FH as well electronic/digital supports and face-to-face meetings about FH are planned.

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