

1 **Awareness of familial hypercholesterolaemia among general practitioners in Australian primary** 2 **care: a qualitative descriptive study**

3 **Introduction**

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

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

14 Over 88% of Australians consult their general practitioner (GP) at least once every 12 months¹¹, offering
15 unique opportunities to detect the disorder early while patients are still young and asymptomatic. FH
16 screening can be undertaken in the community primary prevention setting.¹² Such opportunistic
17 screening using LDL-cholesterol is commonly employed in general practice, but may not always be used
18 effectively to distinguish FH from non-FH patients, especially in adults.

19 Lack of health professional (GPs, cardiologists and other non-GP specialists) and poor community and
20 family awareness of FH are major reasons for the poor uptake in diagnosing and managing FH in
21 Australia and worldwide.^{12,13} Little attention has been focussed to date on screening, diagnosis and
22 management of FH in general practice, where most affected patients can be found.¹² The lack of suitable
23 infrastructure in primary care to undertake systematic testing for FH remains a major limitation.¹² The
24 need for a radical shift and evolution in general community and health professional perceptions of FH
25 and the effect on children and young adults is recognised.¹

26 While evidence strongly suggests FH remains underdiagnosed in primary care, the underlying reasons as
27 to why this continues are unknown. The aim of the present study was to explore the level of knowledge
28 and awareness about FH in a number of Australian general practices.

29

30 **Methodology**

31 **Study sample and setting**

32 The participating practices were involved in an Australia-wide National Health and Medical Research
33 Council (NHMRC) partnership grant study (GNT 1142883) into 'Improving the detection and
34 management of familial hypercholesterolaemia in Australian primary care'.^{14,15}

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

41 **Recruitment and data collection**

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

45 Data collection was accomplished using a semi-structured interviewing technique with GPs, Practice
46 Nurses (PNs) and Practice Managers (PMs) to explore any barriers and/or enablers to diagnosing the
47 condition, as well as their current role (if any) in managing the condition. Interviews were audio
48 recorded and transcribed verbatim. The data were collected prior to commencement of the NHMRC
49 study¹⁴ and represents the initial phase of the study.

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

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

58 We employed a qualitative, descriptive (QD) study methodology¹⁶ to explore the baseline knowledge
59 and perceptions among GPs, PNs and PMs about diagnosing and managing FH.

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

64 By employing this approach, the data are not unduly influenced by the interpretation of the
65 researcher.^{16,17} QD is a suitable method for healthcare research as it helps to focus research questions
66 directly on the experiences of the participants rather than through a more theoretical lens.¹⁸

67 **Data analysis**

68 Data were analysed thematically using NVivo software, version 12 (QSR International, Melbourne, VIC,
69 Australia). A template for the thematic analysis¹⁹ of the data was created using a priori codes taken from
70 interview questions to create the skeleton code frame upon which to base the coding structure.
71 The question guides are listed in the study protocol.¹⁴

72

73 **Ethics:** Study approved by The University of Notre Dame Australia Human Research Ethics Committee
74 Protocol ID: 016067F

75 **Clinical Trial Registration Number:** ID: 12616000630415

76 <https://www.anzctr.org.au/Trial/Registration/TrialReview.aspx?ACTRN=12616000630415>

77 **Findings**

78 The data were coded into key themes using the questions asked by the interviewer. The key
79 thematic areas are briefly summarised in Table 1.

80 **Knowledge, awareness and recall**

81 Overall, relatively few GPs and PNs across the 15 practices could recall caring for a specific FH patient
82 with the diagnosis of FH perceived to be 'opportunistic' for some.

83 "The other one [patient] was having cholesterol done as part of a workup to be
84 prescribed Roaccutane, which is an acne medication that you have to check their
85 cholesterol, so it was just incidentally found." [GP Qld Rural]

86 "I said, "Look, I definitely would just (check) because that's quite young". But I said
87 to him to speak to the doctors because that's not really my decision – (its) the
88 doctor's decision to say that really. But definitely, I would definitely get them
89 checked and he was like, "Oh ok, I'll consider that." [PN WA metro]

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

93 Testing would not normally be done unless the patient talked about a family history or the patient was
94 younger. The younger age range of FH patients proved problematic for some GPs in 'getting patients to
95 take it seriously', as something 'more than high cholesterol' and that this could prove challenging to
96 manage on an ongoing basis.

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

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

102 "So I look out for what I would believe the signs of it are but I haven't picked up
103 someone who I've thought "yep, they have familial hypercholesterolemia." [Vic GP
104 metro]

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

110

111 **Management**

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

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

118 However, at baseline for this study, the screening and treating of patients for high cholesterol was still
119 the primary focus among the GP respondents. Lack of patient compliance with statins was noted with
120 patient resistance seen as an issue by some doctors. Some GPs found it challenging to get patients to

121 appreciate the severity of FH in younger age groups especially alongside the perceived rarity of the
122 condition. Another GP spoke of the challenges of advising and treating a younger patient,

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

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

135 "I assess their general cardiovascular risk, and talk to them about diet and lifestyle,
136 and either would repeat their cholesterol in 3 months with diet and lifestyle changes
137 or would start them on a statin and repeat their cholesterol in (a further) 3 months."
138 [GP Qld Rural]

139 "So it is hypothetical but what I would do would be more of a tendency towards
140 treating them to prevent early onset heart disease." [GP NSW Metro]

141 Some GPs mentioned examination of modifiable and non-modifiable risk factors in their plan to manage
142 a patient. 'Close follow up' was noted as important in managing such patients on an ongoing basis.

143 Regular follow-up and monitoring were seen as key to managing the condition with GPs noting the need
144 to review the patient every few months to check for progress in FH management.

145 **Use of guidelines and referrals**

146 Most participants stated they were unaware of any specific FH guidelines with many GPs using their
147 'own methods' for diagnosing and managing the condition. Referral to a lipid specialist was not availed
148 of by most GPs, preferring instead to refer patients to a cardiologist or endocrinologist for advice.

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

153 "Yea, I wasn't aware of any guidelines." [GP WA Metro]

154 "... but I haven't used any tool to make a diagnosis yeah so certainly if they have secondary
155 issues you know like diabetes or cardiac problems so certainly I use the general guidelines but
156 nothing specific to familial hypercholesterolemia." [GP WA Metro]

157 "I have looked at the guidelines to manage FH, I just can't recall now, I think they're on the
158 Heart Foundation, but I could be wrong, but I have looked at them in the past. " [GP NSW
159 Metro]

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

163 **Contacting family members**

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

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

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

176 “So most people when you offer them (advice about) anything that occurs more than
177 5 years ahead that they completely ignore it and discount it so if you offer them a
178 high reward in 5 years’ time compared with now, they still don't bother so this is
179 human nature” [GP Qld Rural]

180 Costs of genetic testing were seen by lower income families as a possible reason for reluctance to be
181 tested in some instances. One GP spoke of practising in a higher SES area where most patients were
182 willing to go through the process of notifying family members and being tested.

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

188 **Discussion**

189 The QD methodological approach¹⁶ in the pre-education phase of this study revealed that participating
190 GPs had limited awareness and knowledge of the detection, diagnosis and management of FH. Instead,
191 GPs tended to focus more on lowering high cholesterol levels by using statins and encouraging
192 improvement in lifestyle measures for most of these patients. The hereditary component of the FH
193 condition with its ongoing, raised lifetime cholesterol burden from birth was less well appreciated in
194 comparison with the elevated cholesterol levels detected as part of routine medical care in their middle
195 years.

196 Kwok et al.²⁰ have examined knowledge and awareness of FH among GPs in the North West of England
197 and found they almost universally considered themselves to have a key role in the early recognition of
198 undiagnosed FH patients in the community. However, gaps existed in their knowledge of FH inheritance
199 and its increased cardiovascular risk.

200 Pang et al.²¹ also examined knowledge and awareness among primary care physicians in the Asia Pacific
201 region. They found a lack of awareness of FH management guidelines while their knowledge of

202 prevalence, inheritability and cardiovascular risk were also sub-optimal. The findings from both Kwok
203 and Pang broadly reflect similar findings to our research.

204 A UK study by Weng et al.²² has shown that an intervention to identify and manage FH patients in a
205 primary health setting could be successfully adopted. Their research revealed improvements in best
206 practice for identification and management of FH following an educational intervention amongst
207 participating GP practices.

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

212 Our research has noted that GPs were largely proficient in using the Dutch Lipid Clinic Network Criteria²³
213 indicating that the lower rates of diagnosis were more likely attributed to a need for greater education
214 in general practice about diagnosing FH rather than a fault in the diagnostic processes per se.

215 A greater logistical barrier to managing FH was found in the process of family screening and cascade
216 testing. The current systems of contacting close family members is reliant upon the patient informing
217 relatives who may or may not be attending the same practice. Studies in the UK²⁴ and Australia²⁵⁻²⁷
218 provide a cost benefit analysis of using specific FH services to not only reduce the lifetime costs
219 associated with the condition but also to raise the quality of life and survival gains for those affected.

220 The lack of awareness at the health professional and at the patient / family level about the essential
221 hereditary nature of the FH condition (that it had a familial/hereditary component as well as the
222 markedly raised cholesterol component) was one of the key concerning findings from our qualitative
223 research. The fact remains that while life-long treatment with lipid lowering medications - such as
224 statins in addition to diet and lifestyle modifications - was the key to successful management of FH, such
225 knowledge was not always well appreciated in the participating practices at the time the study
226 commenced.

227 Our findings are generally consistent with and complementary to Hardcastle et al.²⁸ who examined
228 Australian patients' perceptions and experiences of familial hypercholesterolaemia and found that
229 many tended to dismiss the serious nature of FH and the importance of lifestyle changes preferring
230 instead to rely on medications to maintain adequate control.

231 **Limitations**

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

236

237 **Implications and conclusion**

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

244 Our qualitative findings have implications for improving health service delivery for FH in the primary
245 care setting. We have highlighted the need for greater education to improve knowledge and awareness
246 about FH for both primary health care teams (GPs, PNs and PMs) and patients and their families.
247 Community conversations involving patients and families with FH as well electronic/digital supports and
248 face-to-face meetings about FH are planned.

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250

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255 GG, CHespe, CV-G, BS, IL, AV, DA-R and JP conducted the study. CB undertook thematic
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Table 1. Main themes, brief explanation and exemplar quotes		
Theme	Explanation	Exemplar quotes
Knowledge, awareness and recall	Unsure of diagnosing FH	"... I would have thought about it, ... in terms of the confidence diagnosing it is probably where I lack, yeah." [NSW Metro GP]
	Patient youth a major factor in suspecting FH	"Often if a young person with elevated cholesterol, they knew that there was some inheritance ... it was a matter of those factors." [NSW Metro GP]
Management	Graded approach – lifestyle modifications before medication	"... 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." [Qld rural GP]
	Use of statins to manage high cholesterol	"... 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." [Qld rural GP]
	Manage as for cholesterol	"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." [Qld rural GP]
Use of guidelines and referrals	Lack of familiarity with guidelines	"I know there are guidelines ... I searched them for patient who has FH but I can't remember where they came from, but I know they exist." [NSW GP metro]
Contacting family members	No follow up by GP of family but rather to make the patient aware	"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." [Qld rural GP]