# Patients' Perceptions and Experiences of Familial Hypercholesterolemia, Cascade Genetic Screening and Treatment

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## Abstract

**Purpose**. Familial Hypercholesterolemia (FH) is a genetic health condition that increases the risk of cardiovascular disease. Although FH can be effectively managed with appropriate pharmacological and dietary interventions, FH detection rate through genetic screening remains low. The present study explored perceptions and experiences of FH patients (N=18) involved in a genetic cascade screening programme.

**Methods**. Face-to-face interviews were conducted to assess patients' knowledge and understanding of FH, explore factors linked to adherence to health-protective behaviours, and examine perceptions of genetic screening.

**Results**. Thematic analysis of interviews revealed four themes: *disease knowledge*, *severity of FH*, *lifestyle behavioural change*; and *barriers to cascade screening and treatment*. Participants recognised FH as a permanent, genetic condition that increased their risk of CHD and premature mortality. Many participants dismissed the seriousness of FH and the importance of lifestyle changes because they perceived it to be effectively managed through medication. Despite positive attitudes toward screening, many participants reported that relatives were reluctant to attend screening due to their relatives' 'fatalistic' outlook or low motivation. Participants believed that they had insufficient authority or control to persuade family members to attend screening and welcomed greater hospital assistance for contact with relatives.

**Conclusions**. Findings support the adoption of direct methods of recruitment to cascade screening led by medical professionals, who were perceived as having greater authority. Other implications included the need for clinicians to provide clear information, particularly to those who are asymptomatic, related to the seriousness of FH and the necessity for adherence to medication and lifestyle changes.

**Key words**. Familial hypercholesterolemia, cascade screening, disease knowledge, attitudes, qualitative interviews.

## Introduction

Familial Hypercholesterolemia (FH) is a serious genetic disorder affecting approximately 1 in every 300 to 500 individuals [1, 2] and is characterized by excessively high low-density lipoprotein (LDL) cholesterol levels, substantially increased risk of early-onset coronary heart disease (CHD), and premature mortality [3, 4]. If FH is untreated it leads to a greater than 50% risk of CHD in men by the age of 50 and at least 30% in women by the age of 60 [5, 2]. FH can be diagnosed through genetic screening and effectively managed through pharmacological treatment and lifestyle changes [6]. Aggressive lipid-lowering drugs have been shown to be effective in inhibiting the progression of arteriosclerosis and reducing CHD risk in FH patients [7, 8]. Lifestyle changes are also recommended for optimal prevention of disease in FH patients [6]. However, research suggests that most patients with FH are undiagnosed [9, 10]. Of the estimated 20 million people worldwide with FH, it is thought that only between 10% and 50% have been identified, and of these; only between 10 and 25% receive adequate treatment [11, 12]. To address this poor detection rate, genetic diagnosis using *cascade* screening procedures has been advocated. Cascade screening procedures have been shown to be cost-effective [13, 14]. and may lead to significant reductions in morbidity and mortality from CHD [15].

Cascade screening involves identifying a specific gene mutation in an FH patient (an 'index' case) and using a family tree to identify relatives likely to have the mutation. Relatives are subsequently invited for screening and the procedure repeated for their relatives [12]. The success of screening programmes depends on the index case providing consent to screen relatives and the uptake of relatives into the programme [4]. However, research indicates that consent given by index cases to have family members contacted for screening is suboptimal with a substantial number of refusals [12]. Further there is evidence that even after consent is given, uptake is comparatively low despite repeated attempts to engage

family members [16]. This has led to increased calls for research on the experiences of FH patients involved in cascade screening programmes to identify the factors that may affect consent for relatives to be screened and reasons contacted relatives fail to respond to invitations to be screened [16].

Only one study to date has explored patient experiences of indirect genetic screening for FH [11]. This study focused solely on the experiences of index patients and did not include relatives. Given that the success of cascade screening depends on uptake by relatives, research is needed to explore the perceptions of relatives in addition to those of index patients. These perspectives may provide insight into understanding decisions to accept or reject screening. In addition, there is also no data on patient views and preferences as to whether cascading should be direct (i.e., contact from the clinic) or indirect (i.e., mediated informally by patients). Contact made directly by practitioners' results in higher response rates and more relatives coming forward for cholesterol testing [18]. However, it is thought that such direct contact may be coercive and invasive [19, 20]. Furthermore, there is evidence that communication about genetic screening frequently takes place within the family without the need for direct intervention [21]. Indirect contact is considered a cost-effective option since it saves time needed to contact relatives [11].

Another under-researched area is how patients interpret their genetic diagnosis of FH and how their understanding of the condition impacts their risk perceptions and motivation to adopt behaviours required to manage their condition including medication adherence and lifestyle changes. It is also important to examine whether there are differences in understanding, risk perceptions and behavioural responses in index cases compared to relative cases and between symptomatic and asymptomatic cases. Some researchers have suggested that genetic diagnosis for conditions like FH could lead to a sense of fatalism, resulting in non-compliance with any kind of treatment because the disease is viewed as

uncontrollable [22]. Other research has suggested that genetic screening for FH is highly acceptable to patients [23, 20] and that a positive result can lead to patients becoming more knowledgeable of the risks of CHD and increase their medication compliance [24, 25]. Research is needed to determine how patients who have been involved in such screening programmes conceptualise FH and how such beliefs affect treatment compliance and lifestyle changes.

### The Present Study

We investigated the perceptions and experiences of patients with a genetic diagnosis of FH involved in a cascade screening programme, including both index and relative cases. The study adopted a qualitative design with semi-structured interviews to explore experiences of cascade screening, living with FH, and behavioural responses to the diagnosis. We expect our results to broaden findings from previous literature pertaining to FH patients' views on their condition, experience of screening and treatment compliance. In particular, we expected results to provide insight into patients' beliefs and attitudes toward decisions to uptake screening, preference for direct and indirect screening, and how their result affects decisions to take medication and engage in lifestyle changes.

## Methods

### Design and Participants

The present study adopted an inductive qualitative research design [26]. Interviews were conducted in a sample of patients who had been genetically tested for FH from the Royal Perth Hospital (RPH) lipid disorders clinic. Ethical approval was granted by the Curtin Human Research Ethics Committee and the RPH Research Ethics Committee. Patients were recruited at the lipid disorders clinic as part of the Familial Hypercholsterolemia Western Australia (FHWA) programme, which provides a publically-funded genetic screening and

treatment service for FH. Invitation letters were sent to 248 individuals selected from a pool of 415 patients according to the following inclusion criteria: aged over 18 years, living in metropolitan Perth, and had received a genetic diagnosis for FH. Sixty-seven (27%) patients responded to the letter. Respondents were excluded because they had received a clinical rather than a genetic diagnosis (n = 9), declined to participate (n = 4), or did not speak English (n = 1) leaving 53 patients eligible to participate the study. From these, the final sample closely represented the characteristics of those in the FHWA programme, based on age and gender, but with an even distribution of index cases and relatives to prevent one perspective being dominant. This resulted in a pool of 21 participants. Three of these were unavailable to be interviewed. The final sample therefore comprised 18 participants (10 males, 8 females) with ages ranging from 25 to 74 years (*M* age = 50.2, *SD* = 14.0). The sample was equally split between index (n = 9) and relative cases (n = 9).

**Procedure**

A semi-structured interview protocol was adopted with participants asked a series of open-ended questions to elicit perceptions and experiences of FH patients. An interview schedule was developed based on questions adopted in previous research on patients' views and experiences with FH [27, 11] and in discussions with a steering group comprised of researchers and clinicians. The interview was designed to be flexible, with an opportunity for follow-up questions to clarify and probe participants' responses. Participants were contacted via email or phone to arrange an interview date. Interviews were conducted in a quiet room by one of two trained interviewers in a building affiliated with RPH away from the lipid clinic. Interviews lasted an average of 40 minutes and were audio-recorded and later transcribed verbatim.

**Data Analysis**

The analysis followed the five-stage process of thematic analysis outlined by Braun and Clark [26]: (1) familiarisation with the data; (2) generating initial codes; (3) searching for themes; (4) reviewing themes; and (5) defining themes. The first step, involved initial 'immersive' readings of the data to gather potential patterns and meanings relevant to the research topic. The second initial code-generation step involved chunking interesting features of the data and labelling them in a meaningful way. The third step involved identifying higher order themes. Initially, a theme was created if several of the coded threads in the transcripts converged on an issue of importance with respect to the research questions. New themes were created if initial codes were inconsistent with existing themes. All interview transcripts were analysed by an experienced qualitative researcher that had not been directly involved in the data collection.

## Results

The analysis identified four main themes in relation to research aims: *knowledge of FH*, *severity of FH*, *lifestyle behavioural change*, and *barriers to screening and treatment*. The latter was a dominant theme and included sub-themes of *avoidance*, *lack of motivation*, and *deferment of responsibility*. Participants demonstrated a good understanding of FH knowledge including that it was a permanent, genetic condition that increases the risk of developing CHD and premature mortality. Most participants did not acknowledge the seriousness of FH because they believed it could be controlled effectively by medication. Participants held positive views towards screening and a desire for relatives to be screened. However, many reported frustration at not being able to persuade adult family members to attend screening or comply with uptake of medication. Details of the themes follow with

quotes that exemplify and illustrate the themes[1]. A summary of the themes, a brief description of the themes and illustrative quotes are provided in Table 1.

**Knowledge of FH**

All participants described explicitly that FH is a permanent genetic condition associated with elevated plasma cholesterol levels. The majority of participants also highlighted the contribution of genetics to their raised cholesterol irrespective of their lifestyle: "It doesn't matter what I eat or how much exercise I'm still going to have high cholesterol without tablets" (M, 46, R). Participants often emphasised the hereditary aspect of FH and their family history of CHD and premature mortality: "It's always been something which is in the family. Mum died from it, I never knew her…she lost all her siblings… from the same disease" (M, 49, I). Participants provided conflicting views of the extent to which their condition is controllable. FH was viewed as something over which patients have little control, but, was also viewed as one that could be effectively managed through medication:

"The illness… is something that's controllable; it can't be changed by diet per se… I need medication and providing I continue to use my medication…I'll probably have cholesterol [levels] like most people" (M, 29, R).

The representation of FH as effectively managed through medication was also evident in expectations of the future progression of their condition: "It's here forever… it's treatable but not curable" (F, 74, R). Overall, participants had a fairly consistent view of the genetic basis of their condition and that it was permanent, but also viewed it being something that was effectively managed through medication.

[1]All quotes are matched to participants by gender (F for female; M for male), followed by age and classification (Index or Relative); for example, a 40 year old male index case would be M, 40, I.

**Severity of FH**

All participants reported knowledge of the increased risk of developing CHD due to FH and the possible consequences, including premature death: “Perhaps your life won’t be as long as you’d like it to be” (F, 69, R). However, the perceived controllability of FH appeared to affect participants’ perceptions regarding both the severity of living with FH and their susceptibility of developing CHD: “I don’t think it’s a really terrible disease… it’s one of the better chronic diseases” (M, 37, I). Another participant alluded to the effective management of the condition which alleviated the perceived severity and consequences of FH: “If the medication is working…then technically I don’t have high cholesterol because it’s being reduced by medication” (F, 50, I).

Participants who were either relatively young compared to the sample average (<30 years of age) (n = 3), or asymptomatic (n = 16), were more likely to refer to CHD as remote and generally viewed FH as less serious: “I never viewed it as a death sentence” (M, 56, I). By asymptomatic, we refer to individuals with FH that are not currently affected by CVD or those presenting with no physical symptoms of disease associated with FH. Quotes from other asymptomatic participants included: “You don’t really experience any problems…because it’s a silent disease, you don’t see it affect you from day to day” (M, 37, I). In contrast, those who described FH in more severe terms generally had a symptomatic form of FH: “You don’t realise how serious it is until something goes wrong…I was told [by the cardiologist] if I don’t control my cholesterol… I’d be lucky to reach 40” (M, 49, I) and “I had sort of puffy eyes and sallow skin and [was not] feeling well… and I’m sure without treatment, I’d probably be dead” (F, 74, R). Others, describing FH in more severe terms had experience of close family members dying prematurely due to undiagnosed or poorly controlled FH: “the diagnosis basically coincided with my brother’s death… he was 35, he had a heart attack and died” (M, 37, I).

Overall, there was convergence of opinion in the acknowledgement of the seriousness of FH and the need for cholesterol-lowering drugs to manage the condition. There were no discernable differences in knowledge of, and attitudes toward, FH between index cases and relative participants. Many expressed the view that living with FH was not a particular burden and that they could live a relatively normal life provided they adhered to their medication regimen.

**Lifestyle and Behavior Change**

The majority of participants spoke positively about lifestyle changes and considered healthy eating and PA as important to their overall health. However, views on the importance of adhering to lifestyle changes for cholesterol control varied widely. Whilst some espoused lifestyle changes to be important in reducing cholesterol: "I watch what I eat constantly…generally it's a low fat diet" (F, 57, R). Alternatively, over half the participants claimed that lifestyle made little or no difference. Several indicated that lifestyle changes had little impact on their cholesterol levels and instead they highlighted the efficacy of the medication:

"Once you're on medication…you can actually eat a lot of fat and the medicine takes care of it… I can have a McDonalds once a week or fried chicken a couple of nights…it never seems to affect my cholesterol levels. They are rock solid at that level irrespective of what I do" (M, 56, I).

Some participants were aware of the importance of lifestyle changes but indicated poor self-control: "I think I shouldn't be eating this, it's not good for my condition. I should have more self-control" (F, 56, I). Several participants expressed barriers to making lifestyle behavior changes: "My wife... loves fatty, salty, oily food. Fried. I tried to steam it she wants

to fry it" (M, 56, I). Participants mentioned that they desired further support from the clinic in relation to the specific details of making behavior changes:

"Don't tell them they can't eat chicken skin, gravy, sugar, butter, fried foods… send them on a low fat cooking course how you can prepare healthy meals without tasting like sawdust… Everyone knows the theory, but putting it to practice is quite hard." (M, 49, I)

It would appear that clinics need to help patients build the practical techniques to facilitate lifestyle behavioral changes such as increasing physical activity and healthy eating. Advice giving only would appear to be insufficient.

**Barriers to Cascade Screening and Treatment**

All participants expressed positive attitudes toward genetic testing. The desire to prevent harm to their family from CHD was reported across all participants and echoed in the common belief regarding FH as a serious but controllable condition: "I think the sooner they know the better, and so they can change their lifestyle" (F, 57, R). However, despite positive attitudes toward screening and encouragement to other family members to attend screening, many participants reported reluctance from relatives to be screened. The main barriers to screening for relatives were clustered around three sub-themes: *avoidance, lack of motivation,* and *deferment of responsibility*.

*Avoidance*

Many participants stated that other family members did not want to find out whether or not they have FH saying they were in denial and avoided screening. This theme of avoidance was reflected by a participant who spoke of her father who refused an invitation to be screened: "He doesn't want to listen to me…he's just like I live my life, life is short, I want to live it…eat what I want" (F, 25, I). Another participant expressed frustration at her adult children

not being willing to be screened: “I have to nag my son and daughter to have their test…my son he’s now 40 and he doesn’t want to know” (F, 69, R) and acknowledged that with adult children there are limits in the extent to which they could be persuaded to be screened without being seen as interfering: “You can’t push her very much…if I say too much I’ll lose them and their kids”. The issue of family dynamics and concern for relationship breakdown on the grounds of ‘nagging’ meant that participants overwhelmingly favoured the hospital to make initial contact with relatives to offer screening.

*Lack of motivation*

The other main barrier to either attending screening or accepting treatment once diagnosed with FH, was a lack of motivation. Advising adult children to attend screening was seen as particularly difficult: “They could make an informed decision about whether to be screened or not…I nagged him when he was 18 or 19 but I won’t do that again”. Another participant living with FH was not receptive of treatment and this lack of motivation appeared to be linked to her young age, and a perception that FH was not very serious: “I put it on the back burner like you don’t need to worry about it just yet…when you’re young you don’t think about it….I didn’t think it was life threatening like being told you’ve got cancer” (F, 27, R). Another participant referred to his brother who had been diagnosed with FH: “He refuses to take medication…he’s more like live for the moment and if it happens it happens” (M, 49, I). Participants also referred to relatives that had been diagnosed with FH but refused treatment. In summary, lack of motivation appeared to be a key factor in seeking to be screened or receiving treatment once diagnosed. Many participants reported relatives who, despite numerous attempts, gave excuses for not being screened or receiving treatment.

*Deferment of responsibility*

Despite positive attitudes towards the involvement of family members in the cascade programme, many participants considered that they were not in a position of authority and expressed frustration in attempting to persuade adult family members to attend screening: "The frustration of them not wanting to find out…I mean you can't make a 37 year old go and do something" (F, 50, I). Participants felt there was little they could do to control whether the relative would respond in a satisfactory way; that is, accepting the information, undergoing screening, and engaging in treatment. Often, participants felt that information on screening should be disseminated through medical professionals because they were perceived to have greater authority: "Okay they'll listen to Mum but 'what does she know'? But when you get a nurse or a doctor talking to you…I think you're much more likely to take notice" (F, 69, R). Another participant endorsed the deferment of responsibility to contact relatives, to the hospital:

"It's not my responsibility… you can lead a horse to water… I've got a cousin who has not consented to screening… We know he's got high cholesterol and doesn't look after himself very well… I wouldn't want that responsibility to be chasing him up and that's why I'd rather leave it" (F, 50, I)

Participants also referred to the issue of responsibility and to weak emotional ties as reasons for not contacting more distant relatives: "The reason I haven't [contacted relatives] is because I'm not closely affiliated with them…they live overseas and I don't really see or talk to them much" (M, 29, R).

In summary, participants expressed multiple barriers to screening and treatment. Participants' perceived their relatives wanted to avoid screening. Participants also expressed a lack of motivation, which was usually because they dismissed the seriousness of the condition and its outcomes. Finally, participants also expressed that they were unable to

persuade their relatives, suggesting that medical staff should contact relatives due to their perceived credibility and authority.

## Discussion

The present study examined beliefs concerning, and responses to, a genetic diagnosis of FH and experiences of cascade screening among index and relative cases. There were five main findings: 1) participants demonstrated good knowledge of FH including that it was a permanent, inherited genetic condition and that FH increased the risk of developing CHD and premature mortality; 2) most participants dismissed the seriousness of FH because they felt the disease could be effectively managed through medication; 3) participants expressed positive attitudes toward screening; 4) many participants reported that their relatives were not motivated or unwilling to attend screening; and 5) many participants felt that they were not in a position of authority to persuade adult family members to attend screening or adhere to medication.

The favourable attitudes found towards genetic testing for FH is consistent with previous research finding that screening is highly acceptable [23, 20]. We also found that as a result of screening, patients were more aware of the risks of CHD as a result of screening, a finding that has been reported previously [25]. In relation to disease severity, all participants were aware that FH is a permanent genetic condition and tended to emphasise the need for medication in controlling FH. Medication was perceived to be a very effective treatment with the majority reporting being compliant with their medication. These findings are consistent with previous research in which FH patients view their condition as manageable and having minimal impact on their lifestyle [17, 28, 29].

Participants viewed cascade screening as important to prevent their family members from long-term chronic illness like, coronary heart disease, consistent with previous research

[30]. All participants, both index or relative cases, consented to passing on information about FH from the clinic and encouraged family members to be screened. There appears to be a high degree of tolerance and advocacy for this screening procedure and this is advantageous for health professionals given the acknowledgment that it is one of the most cost-effective means of identifying previously undiagnosed cases of FH [2]. However, the level of consent for passing on information did not necessarily translate into relatives being contacted by patients or being screened. The main barriers to screening for relatives appeared to be related to avoidance of wanting to know if they have FH or lacking motivation to get tested or starting on medication. It must be acknowledged that it's an individual's right to refuse treatment and that all treatment regimens have deleterious side-effects. It could be that those refusing screening and/or treatment are autonomously choosing to do so rather than them simply exhibiting a lack of knowledge of FH or its severity. Further research is needed with those family members who do not attend genetic screening to better understand the reasons underlying poor uptake and whether these are related to misinformation, health beliefs or motivational factors. It would be useful for further research to determine if intervening with motivation through techniques known to improve motivation to change, such as motivational interviewing, may help to improve uptake of screening if relatives were contacted directly by clinic staff.

In relation to the importance of lifestyle for FH, most patients expressed good knowledge of dietary guidelines. However, many tended to devalue the importance of lifestyle changes in controlling FH and place their hope firmly in medication. In addition, participants reported lapses in adhering to dietary advice and few mentioned physical activity as important. Similar findings have been reported elsewhere where medication has been viewed as more effective, with dietary control and physical activity participation as less effective in controlling their condition [31, 32]. Research by Claassen et al [24] also found

optimal medication adherence in FH patients but less than half reported following recommendations concerning diet and physical activity. Participants also tended to underestimate their risk of cardiovascular disease [24].

The finding that participants did not feel responsible to inform their wider family about their FH diagnosis is consistent with previous research [16]. Participants desired more direct contact from the hospital. The preference for direct contact differs from previous research suggesting that patients prefer indirect, patient-mediated methods of cascading [11]. The findings from the present study support increased direct contact for genetic screening and this preference was reported across both index and relative participants. Such direct methods of contact would ensure that relatives are actually contacted and that any information provided will be correct. As has been found in other studies, participants also believed that recommendations given by health care providers were more likely to be followed [27]. In addition, in a study of public attitudes to screening for FH, almost three-quarters of the participants desired contact by a doctor [33]. Other research findings also indicate that patients appreciate information from the hospital when contacting relatives [16]. The efficacy of direct contact is supported by research showing that contact by a member of the clinical team maximises the number of family members accepting FH screening [18]. Our study is the first to reveal that both cascaded family members and index patients may endorse greater support from the clinic in contacting relatives to attend screening. Previous research has suggested that patients' reticence to pass on information to relatives may be due to weak emotional ties or little contact [34, 29]. This may be true for more distant relatives and is supported by previous research findings [16]. However, our study indicates that strong emotional ties can also be a barrier, with individuals feeling they had to 'tread very carefully' around close relatives, and afraid to push family members for genetic testing due to fears of

damaging the relationship. It appears that both emotional distance and closeness can pose barriers to the discussion of FH screening with relatives.

### Strengths and Limitations

This is the first study to explore the experiences and perception of patients with a genetic diagnosis of FH toward the screening and management of their condition in an Australian context. It is also the first to recruit a sample comprising both relative and index cases within a cascade screening service and, as such, should provide useful information for clinics involved with screening and management of FH. One limitation is that data were collected from a single clinic and as such, the findings may not directly transfer to the wider population of individuals with FH. In addition, the sample was not randomly selected and as such may not be representative of all FH patients. Future research should explore relatives' experiences of cascade screening, in addition to understanding why relatives may decline to participate in genetic screening and how uptake could be enhanced. Such research would entail an intensive approach to participant recruitment as those refusing screening or treatment are likely to be a hard-to-reach sample due to their lack of engagement with FH services. There should also be future endeavours to collect data across sites (urban and rural for example) and across a variety of patients (socio-economic status, age, ethnicity).

## Conclusion

The main finding was the reported reluctance and unwillingness from even close relatives of patients to attend genetic screening and the desire for more hospital assistance in making contact with relatives. This study lends support for the adoption of more direct methods of cascading for FH, by highlighting the value that patients placed on information and advice disseminated through medical professionals, who were perceived to have greater authority. However, given the exploratory nature of this study, these implications need to be

confirmed by future more focused research. For example, researchers should investigate clinicians' beliefs with respect to pro-actively contacting relatives of index cases and investigate the motivational factors associated with lifestyle changes (e.g., increasing physical activity and modifying diet) alongside medication for FH patients managing their condition. Given the low levels of diagnosis of FH through genetic screening, such direct approaches from a clinic may result in more relatives coming forward for screening. Other practice implications include the need for clinicians to provide clear information, particularly to those who are asymptomatic, related to the seriousness of FH and the necessity for adherence to medication and lifestyle changes.

## Informed Consent

All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (Curtin University Health Research Ethics Committee and National Health and Medical Research Council guidelines) and with the Helsinki Declaration of 1975, as revised in 2000. Informed consent was obtained from all patients for being included in the study.

## Conflict of Interest

Sarah J. Hardcastle, Chris Laundy, Ellen Legge, Sarah J. Egan, Rosemary French, Gerald F. Watts, and Martin S. Hagger declare that they have no conflict of interest.

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