Supplementary Appendix S1: Introducing new genetic testing with case finding for familial hypercholesterolaemia in primary care: qualitative study of patient and health professional experience

Patients Interview Schedule

Patient Study ID:_____ Date: _____

Location:_____ Time: _____

Interviewer: _____

Interviewer: Switch on recorder and read the above information aloud

Introduction:

Thank you for consenting to take part in this interview. You recently took part in a research study that looked at how to improve identification of people at risk of familial hypercholesterolaemia at your doctors' surgery. Just to remind you, the aim of that study is to help doctors and nurses to identify people who may have inherited familial hypercholesterolaemia.

This interview would like to explore your views and opinions on how you felt when you were informed that you were identified as being at risk of familial hypercholesterolaemia, also, known as "FH".

Is that ok with you? Yes: ☐ No: ☐

People at risk of FH would have been identified by looking at their medical records and their family history. Some people would have been asked to contact their doctor for further assessment and some people who may have been identified as very high risk of FH would have been invited to have a genetic test to see if they had inherited FH.

Everything you say is confidential and it is also anonymous, so feel free to be as honest as you like. However, there is a caveat to this, in that if you disclosed something that could result in harm to you or anyone else, this will be discussed with the Chief Investigator and may be reported to the appropriate authorities. Is there anything you'd like to ask before we start?

Topic 1: The Patient Journey

1. When you were first invited to take part in the study, you would have received a letter informing you that you were at risk of inheriting the cholesterol disorder familial hypercholesterolaemia. What do you understand about the condition?

   Prompts:
   - What do you think can contribute to how the condition will develop?
   - Do you think there's anything that people can do to influence this?

2. What do you know about cholesterol and high cholesterol?

3. Can you remember how you felt when you received the letter?

   Prompts:
   - Can you remember any particular emotions that you felt when you'd read the letter?
   - Do you feel that the letter contained enough information for you to understand about FH?
   - Did you have anyone that you could ask, or anywhere that you could look, for more information on FH?
   - Had you heard of the term FH before?
   - What did you like or dislike about the flowchart that outlined the study steps for patients?

4. Can you remember what happened after you received the letter at your doctors' surgery? Who was it that made contact with you?

Prompts:
- Did you receive any written information from your doctor or nurse about the risk of familial hypercholesterolaemia?
- Did you talk to anyone in the family about the letter informing you that you may be at risk of FH?

5. When you received your letter informing you that you were at risk of FH or to have a genetic test, how did you feel about the information provided?

Prompts:
- Do you feel you received enough information with the letter to make the decision as to whether or not to have the test?

  Prompts:
  - Can you remember what it was that influenced your decision?
  - Is there any other information you would have liked to have received at that time?
  - Did you have any concerns or did any unexpected issues occur during this time?

6. Being detected as potentially at risk of having FH is linked in part with having high cholesterol. This can also put people at risk of developing conditions that are related to high cholesterol, such as heart disease and strokes.

   …How do you feel about this?/What do you think about this?

   Have you thought about how you might be at risk of those things?

   Prompts:
   - Have your thoughts about your own vulnerability to these conditions changed now compared to before you took part in the study?
   - What does that mean to you personally? (more at risk than before/anything you can do to reduce your risk)

7. Has taking part in this study made you think about your own lifestyle?

   Prompts:
   - Have you made any changes? (exercise/alcohol/smoking/diet)

Topic 2: Genetic Testing

ONLY FOR PARTICIPANTS WHO HAD A GENETIC TEST

8. What influenced your decision about whether to take the test?

9. How long did you wait for your genetic test result?

   Prompts:
   - How did you feel waiting for the results of the genetic testing?
   - Did you feel supported and could contact somebody like a doctor, nurse or support group whilst you were waiting for your results?

10. And, how did you feel when you received your results for the genetic testing?

    Prompts:
    - Can you remember feeling any particular emotions when you'd received your results?
    - Did you understand the result of the genetic test?
    - Is there anything you would like to have changed about the way that you received your results?
    - What was the key message(s) that you picked up from the health professional about your genetic test and the result?

11. When people have genetic tests, they are sometimes asked if they would like to ask other members of the family to have a genetic test too. This is called cascade testing. How would you feel about talking to other members of your family about having a genetic test for familial hypercholesterolaemia?

    Prompts:
    - Did you talk to any family members about the possibility that they may have FH too?
    - Did you feel you had enough information to relay the information to family members?
    - Do you know the contact details of support networks for people with familial hypercholesterolaemia and if so, did you look at any of the information that they provide?
    - Did you find the information helpful?

12. Do you have any questions that you would like to ask about the study, or have you anything else that you would like to add about identifying and inviting patients with possible familial hypercholesterolaemia for genetic testing?

Thank you for taking part in the interview. The results will help doctors and nurses to give better advice and treatment to people with a high cholesterol result.

Interviewer: Switch off the recorder

Health Professionals Interview Schedule

Participant Study ID:_____ Date: _____

Location:_____ Time: _____

Interviewer: _____

Interviewer: Switch on recorder and read the above information aloud

Introduction:

Thank you for consenting to take part in this interview. The practice recently took part in a research study that looked at improving identification of familial hypercholesterolaemia in primary care using a new case ascertainment tool (FAMCAT). Therefore, we are inviting practitioners who took part in the study for their views and opinions on the feasibility of using FAMCAT to identify those individuals at risk of familial hypercholesterolaemia in primary care.

Is that ok with you? Yes: ☐ No: ☐

Everything you say is confidential and it is also anonymous, so feel free to be as honest as you like! However, there is a caveat to this, in that if you disclosed something that could result in harm to you or anyone else, I am required to discuss this with the Chief Investigator and may have to be reported to the appropriate authorities.

Is there anything you'd like to ask before we start?

TOPIC 1: FAMCAT PROCESSES

Introduction

As you will know familial hypercholesterolaemia (FH) is an autosomal dominant inherited disorder with family members having 50% chance of inheriting the disorder. However, 85% of affected individuals still remain unidentified. Therefore, we developed the predictive FAMCAT tool to see if this would be an effective way to identify individuals in primary care.

1. What are your thoughts on patients receiving a letter to inform them that they may be at risk of FH?

   Prompt:
   - Do you recall any instances or events that may have occurred with patients at this practice as a result of receiving the study letter?

***Show GP copies of the letters ***

(Show study flowchart)

2. Having been involved in the study using FAMCAT, what are your thoughts on the processes involved?

   Prompt:
   - When using FAMCAT, what do you feel are (perception) or have been (experience) the most helpful steps used in identifying patients at risk?
   - What do you think are the least helpful?
   - Can you think of any barriers or advantages to the process of identifying patients at risk of FH in primary care?
   - Did you get any feedback from colleagues/practice staff regarding the study processes? (Please do feel free to provide negative feedback as well as positive as this will help inform the study outcomes)

*** Prime the GPs on their patient cases – get them to pull up the cases on the system***

3. Some patients would have been asked to contact the practice for a genetic test for FH.

   Can you tell me anything about your experiences of discussing/ communicating with any patients about having a genetic test for FH?
   Prompt:
   - Do you recall any incidents or events that may have arose with any of these patients?
   - Have any concerns been expressed by practice staff relating to the process?

TOPIC 2: PATIENT MANAGEMENT

We're interested in developing ways to support care, and are aware that few practitioners in primary care are likely to have expert knowledge about FH. May we ask…

13. From the study, XXX patients at the practice were identified as being at possible risk of FH using the tool. Eventually, XX patients were diagnosed with FH. What are your thoughts about this detection rate?

14. As a practitioner, following a patient being identified as either at risk, or as having FH, what do you feel are the most important message(s) that you then need to relay to patients about familial hypercholesterolaemia?

    Prompts:
    - Do you feel overall that there have been any barriers to colleagues in the practice relaying information to patients about FH?
    - Have there been any key facilitators to this?
    - Do you think there are any training needs for staff around this?

15. How did/would you feel about relaying the results of genetic tests for familial hypercholesterolaemia to your patients?

    Prompts:
    - Is there any further knowledge that you think would help facilitate those conversations?
    - Can you identify if there are any specific training needs?
    - Do you feel that patients would need any specific written information when relaying information on FH?

TOPIC 3: FURTHER EXPERIENCE OF USE

16. Having been involved in the research study and used the FAMCAT tool in this practice would you recommend FAMCAT to other practices as a tool for identifying patients at risk of FH in primary care? Please do feel free to be as honest as you like as this will help inform the study outcomes?

    Prompts:
    - What have you liked about FAMCAT/what were the positive things about using it?
    - What do you think we could change or could be doing better?

17. Is there anything further you would like to talk about in the interview that we haven't covered?

Thank you very much for taking part in the interview. Your responses will be collated with the results from the other practitioners being interviewed and will help us to improve the design and implementation of the FAMCAT tool. I will forward a summary outline of our interview today so that you can verify that this is a true reflection of our discussion.

Interviewer: Switch off the recorder