Academic Health Science Network
North East and North Cumbria

# Improving the identification of patients with familial hypercholesterolaemia in primary care

AMGEN®

SANOFI

This pilot is delivered as part of a Joint Working Arrangement between the AHSN NENC, Amgen Ltd. and Sanofi Ltd. who have all contributed to the review and approval of this case study.

MAT-GB-2001510(v1.0)
Date of preparation: October 2020

## Introduction

Cardiovascular disease (CVD) is the leading cause of death worldwide, accounting for 31% of all global deaths.[1] An important under-diagnosed cause of premature CVD, specifically coronary heart disease (CHD), is familial hypercholesterolaemia (FH).[2] If left untreated, >50% of men and >30% of women with FH may develop CHD by the age of 50.[2] FH is an inherited disease, characterised by lifelong high cholesterol levels – specifically low-density lipoprotein-cholesterol (LDL-C).[2] As many as 300,000 people in the UK may be living with the condition, but it is estimated that only around 15,000 patients have a formal diagnosis of FH.[2]

Improving the detection and treatment of high cholesterol and conditions such as FH that are associated with CV risk factors is a strategic public health priority. Both Public Health England (PHE) and the NHS Long Term Plan have set targets to prevent 150,000 CVD events over the next 10 years by improving the detection and treatment of high-risk conditions including high cholesterol (hypercholesterolaemia).[1,3] There is also a drive to expand access to genetic testing for FH to support the diagnosis and treatment of patients at genetic risk of sudden cardiac death.[3] According to prevalence estimates, only 7% of patients in the UK with FH are currently identified, but the NHS Long Term Plan aims to improve these diagnosis rates to at least 25% of FH patients in the next five years through the NHS genomics programme.[3]

Despite NICE clinical guidelines (CG71) and quality standards (QS41) recommending health care professionals to consider the possibility of FH in adults with raised cholesterol, especially in those with a personal or family history of CHD,[4,5] substantial improvements in FH diagnosis are not being made, which represents a major gap in CVD prevention in the UK.

**This case study describes an initiative from the Academic Health Science Network for the North East and North Cumbria (AHSN NENC), in collaboration with Amgen Ltd. and Sanofi Ltd., Clinical Commissioning Groups (CCGs), and GP practices to implement a targeted FH risk assessment and screening programme at a primary care level to improve the identification of patients with FH.**

## Rationale for identifying high-risk FH patients in primary care – previous pilot

Between 2011 and 2014, a pilot project, ran by NHS Medway CCG, showed that implementing a FH risk assessment service in primary care can lead to improved FH diagnostic rates.[2]

**Two interventions were introduced to increase the detection of FH:[2]**

1. An FH audit tool systematically searched electronic medical records within GP practices
2. A nurse-led programme reviewed the audit list of at-risk patients and identified any missing clinical or non-clinical parameters and referred suitable patients for genetic testing

## Aims and objectives of the FH case-finding service

The scope of the current project covers the footprint of the AHSN NENC, with a target patient population of approximately 3.5 million. The Joint Working project with AHSN NENC, Amgen and Sanofi is one of the first multi-company instances of a non-promotional industry collaboration. The project involved undertaking searches of primary care electronic medical records and establishment of a nurse-led clinic to identify patients at high-risk of FH.

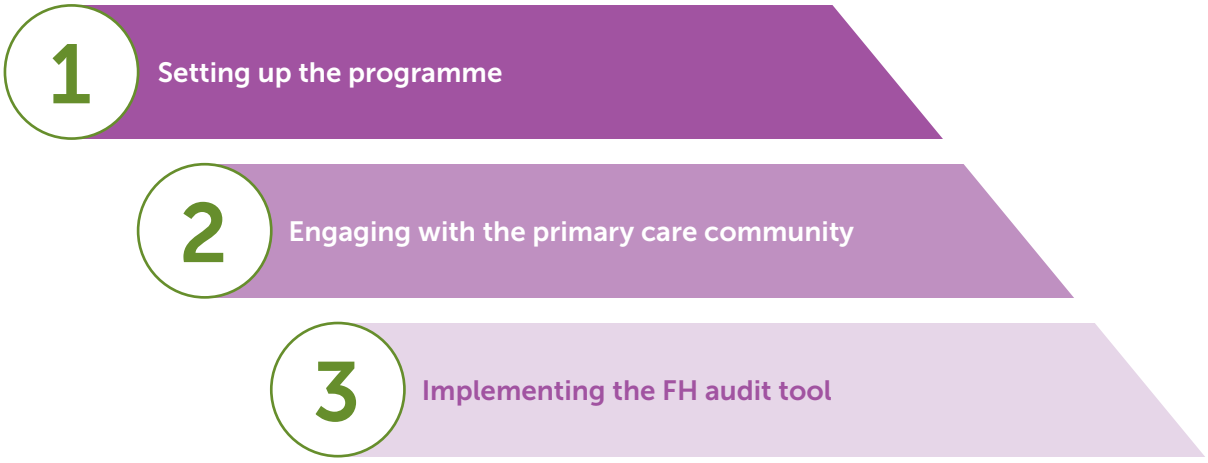
This case study specifically details the initiation and implementation of the FH case-finding service and provides an interim analysis of the learns so far and the audit results to date.

## Key stakeholders

Critical to the success of initiating, implementing and evaluating the FH case-finding service was the collaborative engagement and effective communication between multiple stakeholder groups and individuals.

| Stakeholder groups | |
|---|---|
| AHSN NENC | Primary care data quality leads |
| Sanofi | GP practice managers |
| Amgen | GP leads |
| Northern Molecular Genetics Service | FH nurses |
| Newcastle Upon Tyne Hospitals NHS Foundation Trust | North East commissioning support unit |
| Newcastle University | Regional lipid leads |
| British Heart Foundation | Medicines optimisation leads |
| IT infrastructure support teams | CCG leads |
| NIHR data collection support | CVD prevention networks |

## Process for establishing the FH case-finding service

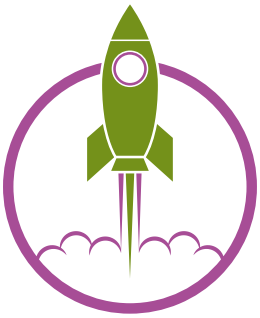



### 1. Setting up the programme

Joint Working agreements were initially established between the AHSN NENC and Amgen and the AHSN NENC and Sanofi. Key stakeholders were identified, and an expert steering committee established to define the scope, requirements and geographical location of the service. The southern region of the AHSN NENC was chosen as the pilot area to improve FH diagnostic rates. A dedicated FH nurse was appointed to coordinate the implementation of the pilot to triage, identify and screen patients for referral for genetic testing.

## 2. Engaging with the primary care community

Raising awareness of FH diagnosis among the primary care community was critical to ensure GPs were engaged in the case-finding service.



**A launch meeting was held within the region to promote the service and educate GPs around the importance of identifying patients at high-risk of FH**

**Speakers included the NHSE National Clinical Director for CVD who endorsed this project from a national perspective**

## 3. Implementing the FH case-finding service

An FH audit tool was developed to identify patients at high-risk of FH in primary care by searching electronic medical records in GP IT systems and providing an FH risk score for each patient.

Initially, a commercial tool identified clinical signs associated with FH including highest total cholesterol, LDL-C, triglyceride levels, previous history of CHD, family history of myocardial infarction (MI), previous FH diagnosis and elevated cholesterol levels.[6]

However, when the tool was used across GP practices, several implementation and operational challenges were identified that proved a barrier to more widespread implementation.

- The tool used the hard drive in GP IT systems and was not integrated with the practice medical records – this presented a security challenge when accessing all GP systems and was impractical for healthcare professionals to use.
- An associated cost per GP practice to run the tool is incurred.
- The tool was not live and therefore only provided a snapshot in time. Each time the tool was run, patients were allocated a new identification, so it was not possible to track the patients.
- The criteria for identifying patients at high-risk of FH were not adequate as a large proportion of patients were identified who did not fit the genetic testing criteria – this resulted in a lengthy triaging process for the FH nurse specialist to ensure all eligible patients were captured, which may not be feasible when considering full roll-out of the service.

A subsequent partnership with the Clinical Digital Resource Collaborative (CDRC) resulted in the development of a free resource that allowed direct integration of FH-specific search templates into GP practices using SystmOne and EMIS. The CDRC FH resource is accessible within the GP IT system and allows health records to be searched in real-time, with notes/flags applied directly to the patient records. The searches were based on NICE guidelines, with a Dutch Lipid Clinic Network Score (DLCNS) applied to relevant patient records.

The CDRC FH resource identified patients according to the specific FH diagnostic parameters and generated the DLCNS. The FH nurse specialist then worked closely with the GP practice leads to triage the records and then invite suitable patients into a dedicated clinic to inform them of the service and indication for genetic testing.

The regional FH genetic testing pathway incorporated a two-stage genetic testing process, utilising an innovative shorter genetic test which looked for pathogenic variants known to be common within the North East population. Patients with positive genetic test results are referred to a local lipid clinic and followed-up with the FH team for cascade testing of family members. Patients with a negative initial genetic test were referred to a multi-disciplinary team (MDT) and proceeded to have full gene sequencing if indicated. Patients who had an ethnic background from outside the North East (e.g. European, black, Asian or minority ethnic) would proceed directly to full gene sequencing as a low number of positive results from the shorter genetic test was expected in this population.

**CDRC FH diagnostic parameters**

**SEARCH 1:**

**Total cholesterol**
>9 mmol/L (>7.5 mmol/L if <30 years)

**Non-HDL cholesterol**
>7.5 mmol/L (>6.0 mmol/L if <30 years)

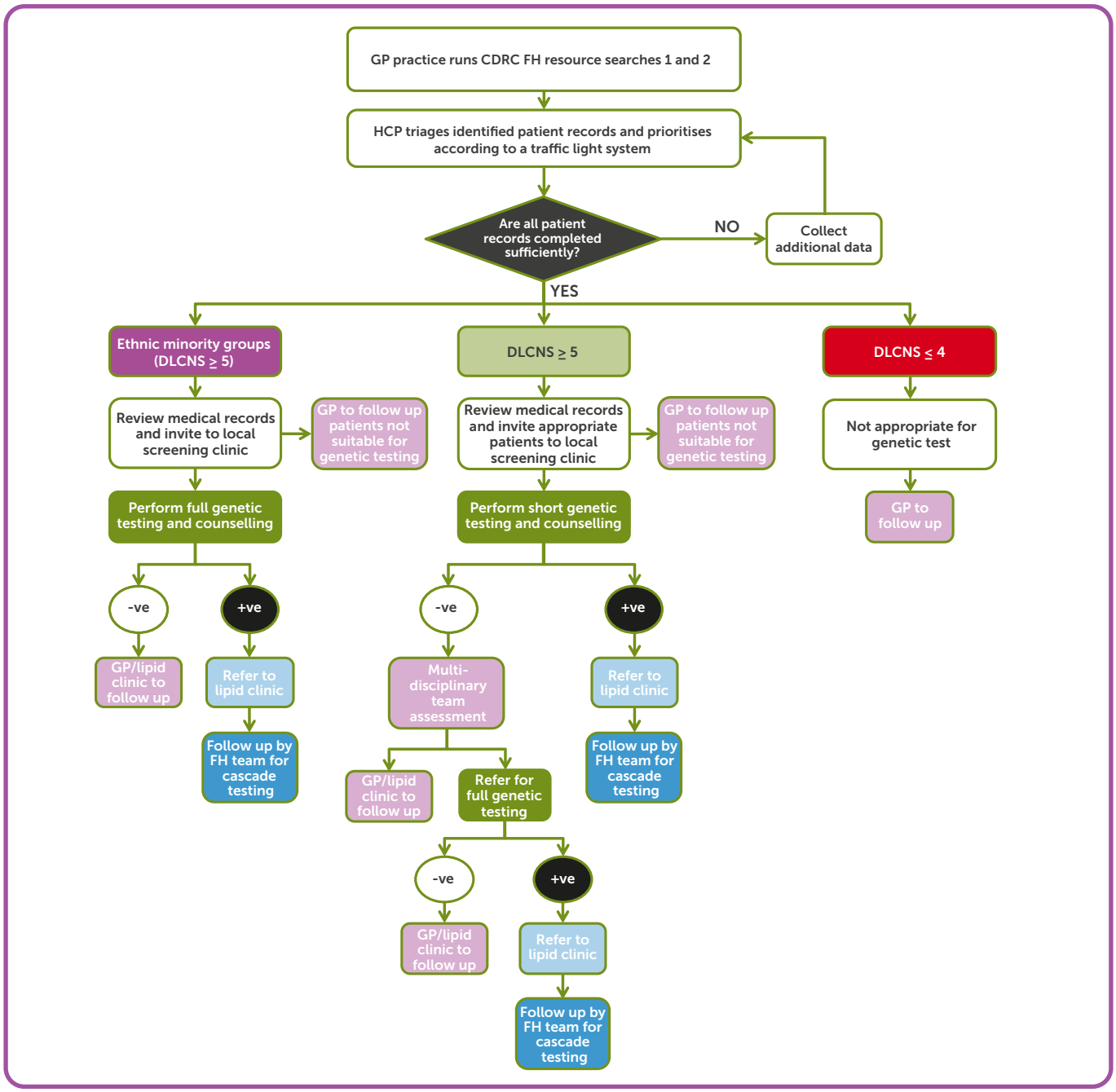
**LDL cholesterol**
>6.4 mmol/L (>5.0 mmol/L if <30 years)

**Triglyceride**
≤2.3 mmol/L

**SEARCH 2:**

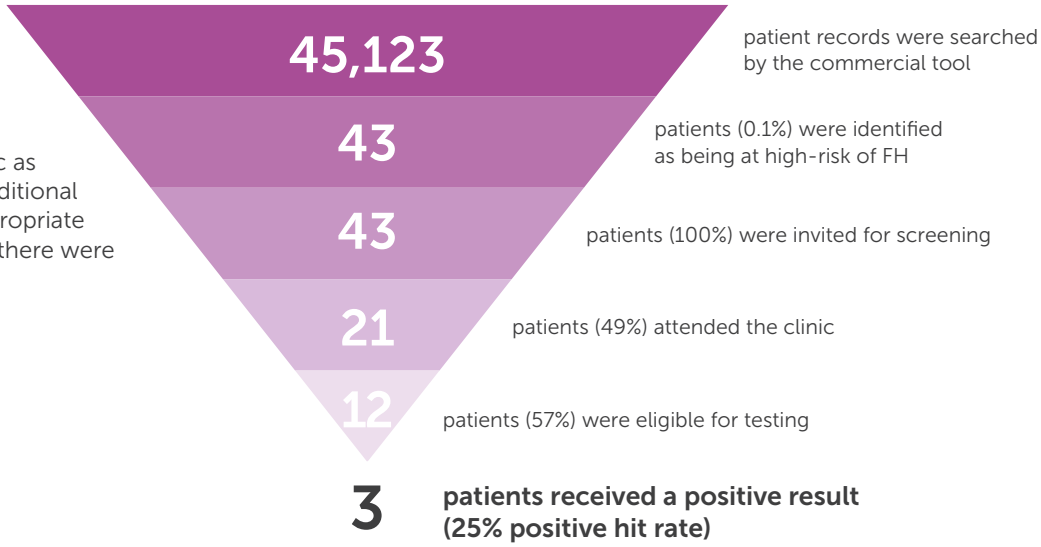
**DLCNS**
≥5

## FH case-finding service patient pathway



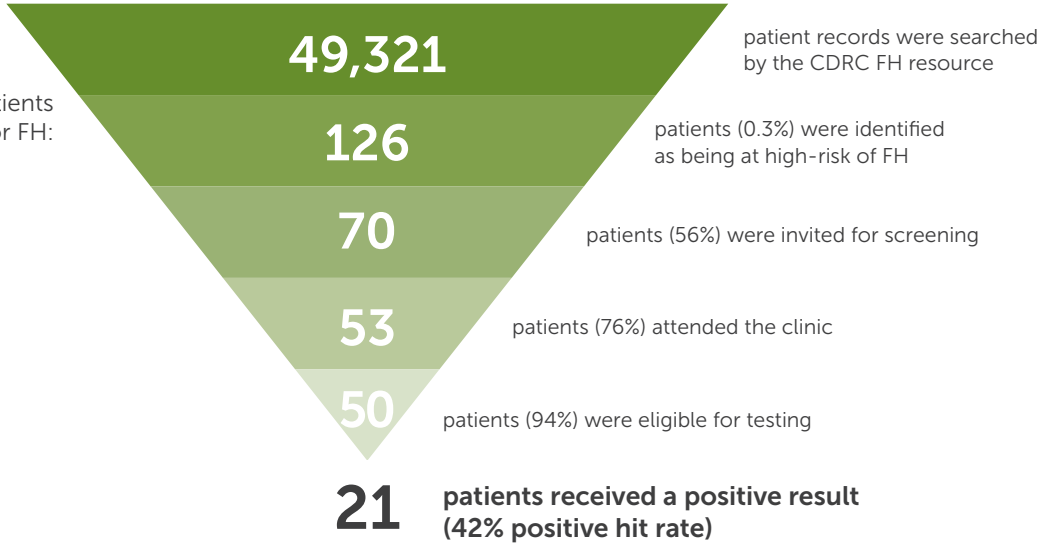
## Key results to date

The commercial tool was used across four GP practices. Although it helped identify some patients at high-risk of FH, it was not as specific as anticipated and took additional work to identify the appropriate patients for testing and there were areas for improvement:

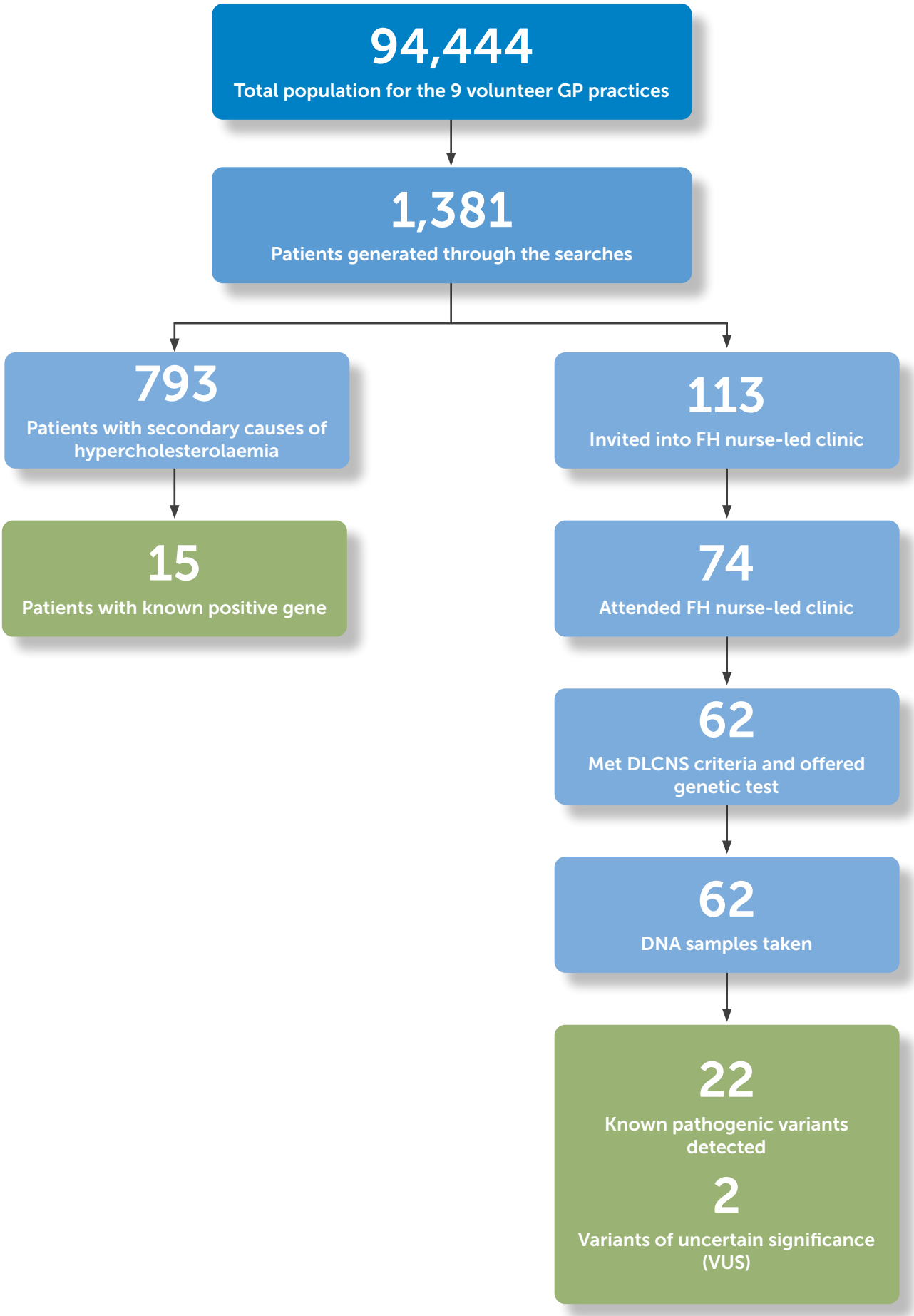



### The CDRC FH resource was then developed and compared to the commercial tool and it resulted in an improved detection rate of patients at high-risk of FH

The CDRC FH resource was refined further to improve the detection rate and numbers of patients identified as high-risk for FH:

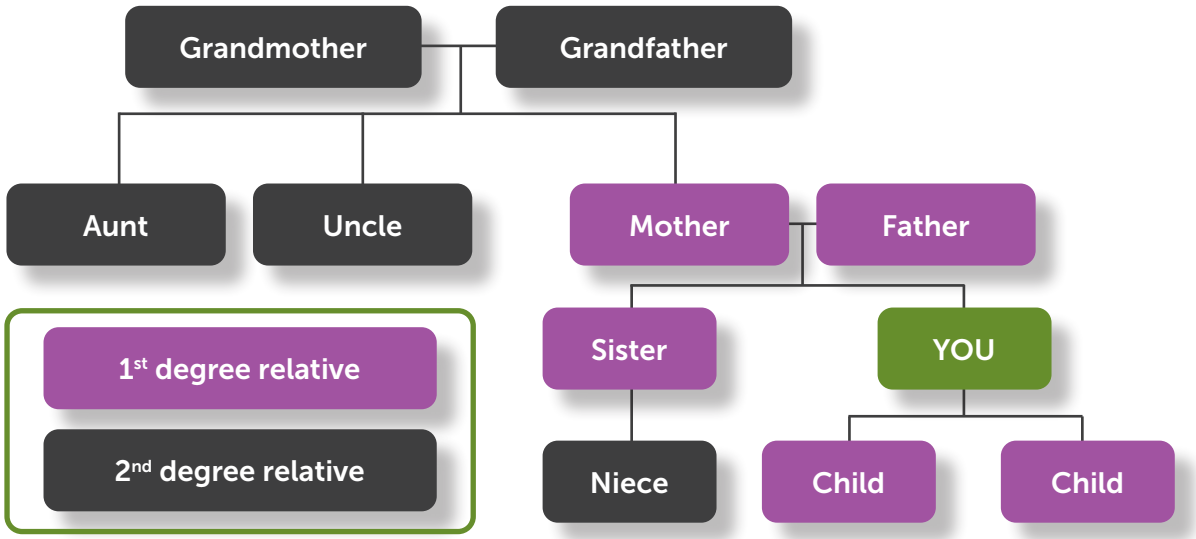


## The project identified 24 index patients with confirmed FH across a footprint of 9 GP practices

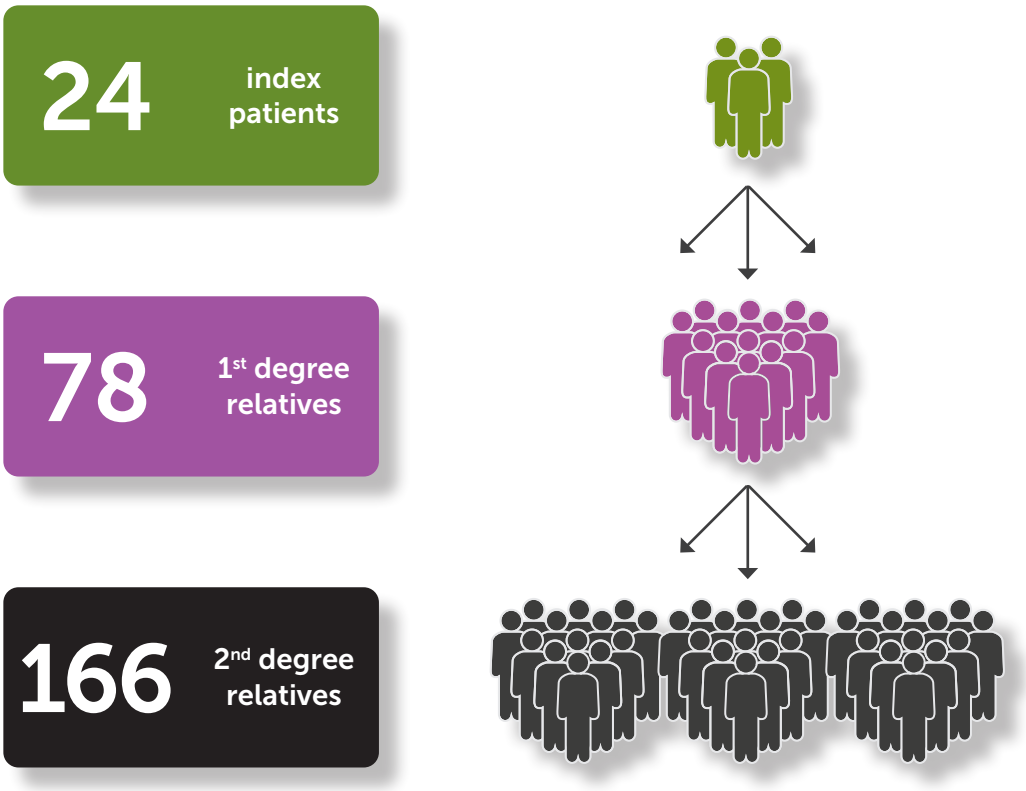


## FH testing was offered to the family members of patients who tested positive

Once a patient had received a positive genetic result, the FH nurse specialist contacted the individual's 1st and 2nd degree relatives to begin cascade testing




## The initial 24 patients identified as FH positive in this study had 244 family members eligible for cascade testing



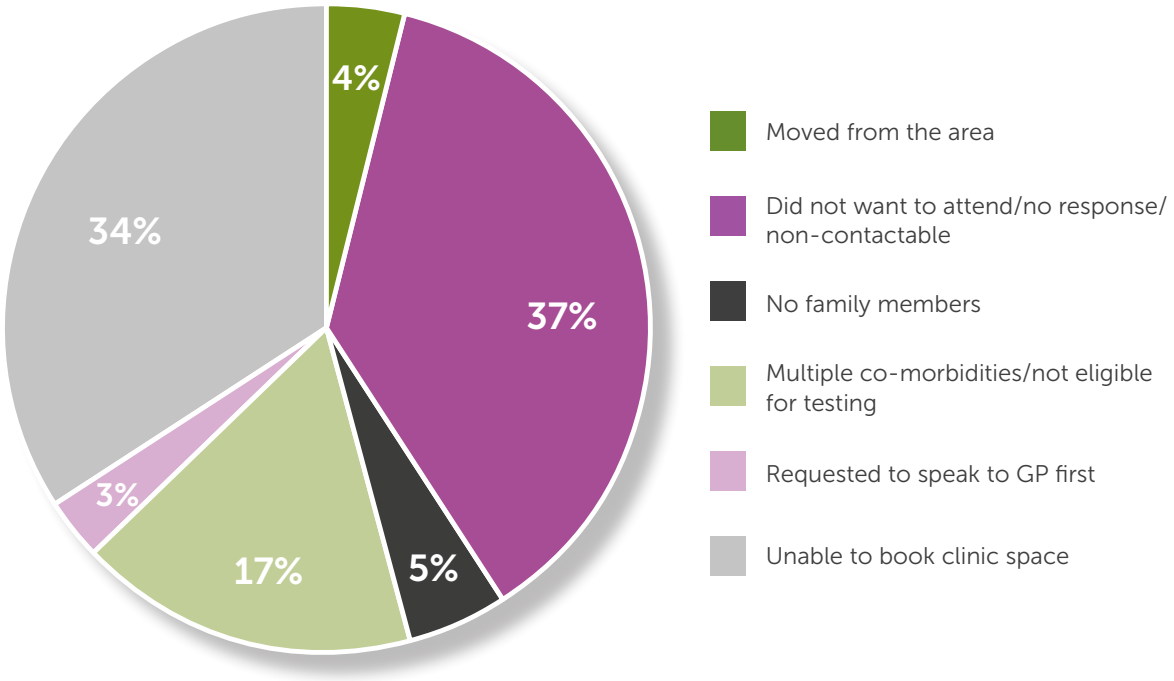

## An additional 80 patients are predicted to be FH positive following cascade testing of these 1st and 2nd degree family members

# Optimising and evaluating the FH case-finding service

## Patient feedback was used to help optimise the service

The invitation letter and patient information sheet were redesigned and a pre-clinic phone call was introduced to reduce the number of patients who did not attend the clinic

**Reasons why patients did not attend the clinic include:**



The FH case-finding service is currently ongoing, and a full evaluation will be performed following completion to build a business case for future funding and commissioning. GPs will be surveyed to evaluate how the FH case-finding service has improved their knowledge and understanding of the diagnosis and appropriate management of FH.

## Patient feedback rated the service as excellent or very good

- **100% of patients felt the service met their expectations**
- **100% of patients felt the level of education provided was excellent or very good**
- **100% of patients felt that the FH nurse specialist was approachable**

Very informative and expertly delivered - has made me aware of how important my diet is in lowering my cholesterol

I received a lot of information about FH and cholesterol, treatments and ways to self-help. Very interesting and well worth attending the appointment

I just want to say thank you

The nurse was very good at explaining what the project was for and I was happy to participate

Everything was explained clearly in an approachable manner

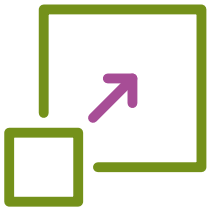
It was very informative and interesting

I found the nurse to be very helpful, understanding and knowledgeable

## Key challenges

The key challenge for successfully implementing the FH case-finding service was ensuring that only the most appropriate patients were identified

Each patient identified was triaged and an accurate family history for CVD and FH risk factors obtained. This could be time-consuming if a large number of inappropriate patients are identified and has capacity and logistical implications for both the patient and practitioner



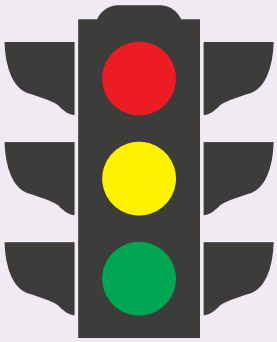
It is important to consider these capacity and logistics implications when scaling-up the service across the region and planning workforce requirements

Initially, 50% of patients did not attend their clinic appointment. After acting on feedback from patient groups, this was reduced to 25%

It was challenging to fit secondary care diagnoses into limited pre-existing codes in primary care and non-lipid specialists in primary care are currently making an FH diagnosis without genetic testing. Out of 176 patients who had an FH diagnosis, only 15 had a confirmed diagnosis with genetic testing

# Key considerations

| Raising awareness of FH testing and clinical risk factors | |
|---|---|
| **Primary care** | GP engagement and provision of educational support on the unmet need for FH, including building advocacy around active screening for FH and appropriate referrals for genetic testing. |
| **Cardiovascular healthcare professionals** | Disease awareness campaigns and medical education. Secondary care healthcare professionals should consider whether a patient who presents with or has a previous history of a premature coronary event, could have FH, and to review not only their most recent lipid results, but also their historical results and family history. |
| **Patients** | Improved awareness of FH risk factors and the importance of familial testing will help to relieve fears around genetic testing and its implications, ensure adherence and that the appropriate level of care is received. |

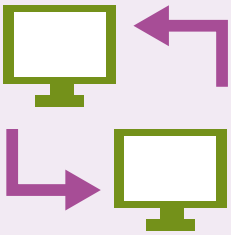
| Consider developing a care pathway for patients who have been identified as high risk of CVD, but are unlikely to have FH | |
|---|---|
|  | The service identified a proportion of patients who are at high-risk of CVD, but either tested negative for FH or were not eligible for genetic testing.<br><br>Patients with a negative genetic test would have a diagnosis based on the clinical assessment and blood results, which could include: clinical familial hypercholesterolaemia; polygenic hypercholesterolaemia; severe polygenic hypercholesterolaemia; primary hypercholesterolaemia; mixed hyperlipidaemia; raised lipoprotein (a) levels (this is important when re-running FH identification searches as well as implications for lipid management for these patients).<br><br>These patients would benefit from a dedicated care pathway to minimise long-term health implications and ensure they are being optimally managed. |

| Use existing working relationships to define an appropriate geography for implementation of the service | |
|---|---|
|  | The successful roll-out of an FH case-finding service requires the collective advocacy and commitment of multiple CCG and GP leads across the region. Utilising existing working relationships and developing steering committees to champion best practice in the region can facilitate this process.<br><br>Targeting a population of around 3 million is suitable for scaling this service and may require collaboration across a whole integrated care system. |

**Ensure dedicated practitioners with a good knowledge of FH risk factors are utilised to support implementation of the service**

Dedicated FH practitioners are crucial to releasing capacity and improving efficiencies, via screening patients and identifying potential FH risk factors in the patient records. They can then refer appropriate patients for genetic testing without significantly impacting the daily duties of the GP.

**Facilitate data sharing between primary and secondary care**



Sharing of patient records between primary and secondary care would allow the most relevant and accurate clinical data to be imported into the FH audit tool and the most appropriate FH risk score to be calculated.

Appropriate data protection and security measures are also necessary when implementing this type of service and when transferring patient records between primary and secondary care.

## Conclusion and next steps

**Advocacy**, **collaboration**, and **commitment** from GPs, CCGs and patients, with a **dedicated workforce** to triage patient records and manage the genetic testing clinics is key to the success of the service. It is important to remember that for every FH patient diagnosed, there will be other members of their family with FH, so it is vital to begin actively identifying FH patients to stem the potential increase in CV events via untreated FH.

This project will be scaled-up across the AHSN NENC footprint in GP practices using SystmONE in the first instance. The CDRC FH resource would also be able to be used on EMIS systems following modification and will be implemented during phase 2 of the roll-out.

This case study highlights the feasibility and process involved in implementing a service in primary care that identifies patients at high-risk of FH. It represents an opportunity to improve FH diagnostic rates and is a strategy to meet the targets set by Public Health England and the NHS Long-Term Plan to substantially reduce the incidence of CVD and premature mortality associated with FH by 2024.

## References


1. Public Health England. Health matters: preventing cardiovascular disease. www.gov.uk/government/publications/health-matters-preventing-cardiovascular-disease/health-matters-preventing-cardiovascular-disease. Published 2019. Accessed September 2020.

2. Green P, Neely D, Humphries SE, Medway FHASC. Improving detection of familial hypercholesterolaemia in primary care using electronic audit and nurse-led clinics. *J Eval Clin Pract.* 2016;22(3):341-348.

3. NHS. The NHS Long Term Plan. www.longtermplan.nhs.uk/wp-content/uploads/2019/01/nhs-long-term-plan-june-2019.pdf. Published 2019. Accessed September 2020.

4. National Institute for Health and Care Excellence. Clinical guidelines and evidence review for familial hypercholesterolaemia: the identification and management of adults and children with familial hypercholesterolaemia. www.nice.org.uk/CG71. Published 2008. Accessed September 2020.

5. National Institute of Health and Care Excellence. Familial hypercholesterolaemia QS41. www.nice.org.uk/guidance/qs41. Published 2013. Accessed September 2020.

6. Weng SF, Kai J, Andrew Neil H, Humphries SE, Qureshi N. Improving identification of familial hypercholesterolaemia in primary care: derivation and validation of the familial hypercholesterolaemia case ascertainment tool (FAMCAT). *Atherosclerosis.* 2015;238(2):336-343.

This pilot is delivered as part of a Joint Working Arrangement between the AHSN NENC,
Amgen Ltd. and Sanofi Ltd. who have all contributed to the review and approval of this case study.

MAT-GB-2001510(v1.0)
Date of preparation: October 2020

Veeva Vault

# CERTIFICATE FOR NON-PROMOTIONAL ITEMS (PMCPA)

| | |
|---|---|
| **Version:** | 1 . 0 |
| **Document Number:** | MAT-GB-2001510 |
| **Document Name:** | case Study from Sanofi & AHSN-NENC Joint Working Project |
| **Country:** | United Kingdom |
| **Product:** | Praluent |
| **Material Type:** | Institutional/Partnership Activities |
| **Material Intent:** | Non-Promotional |
| **Certification Type:** | Certification |
| **Audience:** | Healthcare Professionals, Payers |
| **Additional Audience:** | Output of Joint working project - is being approved in parallel by Amgen as JW stakeholder |
| **Intended Use:** | External Use |
| **Method of Dissemination:** | Digital |
| **Description / Objective:** | Project Title: Improving the Identification and treatment of patients with FH in Primary Care |
| **Material Owner:** | Adam Jakeway |

I have examined the final form of the material and in my belief it is in accordance with the requirements of the relevant regulations relating to advertising and this Code, and is a fair and truthful representation of the facts.

| Role | Signature |
|---|---|
| Arunesh Sil - Medical | Date: 28-Oct-2020 12:10:02 GMT+0000 |