SYSTEMATICALLY IDENTIFYING FAMILIAL HYPERCHOLESTEROLAEMIA IN PRIMARY CARE

AN AUDIT WITHIN THE MEDWAY CLINICAL COMMISSIONING GROUP

HEART UK
THE CHOLESTEROL CHARITY

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ABOUT OUR CHARITY

HEART UK is the nation's cholesterol charity and aims to prevent premature deaths caused by high cholesterol and cardiovascular disease. The charity works to raise awareness of the risks of high cholesterol, lobbies for better detection of those at risk, provides advice and information to patients and clinicians, and supports healthcare professional training.

TARGET AUDIENCE FOR THIS REPORT

Key opinion leaders and policymakers in health, the Department of Health, NHS England, Public Health England, NHS Trust chief executives, Clinical Commissioning Group (CCG) chief executives, CCG chairs, medical directors, Strategic Clinical Networks, Directors of Public Health, geneticists, a range of clinicians, including GPs, lipidologists, chemical pathologists, and nurses.

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FOREWORD

Professor Huon Gray, National Clinical Director for Heart Disease, NHS England

"Prevention is better than cure" no more so than for those people with the inherited lipid disorder familial hypercholesterolaemia (FH). There is an enormous opportunity to prevent the occurrence of coronary heart disease (CHD) in patients with this disorder through early diagnosis and effective management.

In the UK there are believed to be more than 120,000 people with FH, but fewer than 12 per-cent of them are diagnosed. This suggests that there are more than 100,000 people in the UK living with FH at risk of premature mortality due to CHD.

In 2008, the National Institute for Health and Care Excellence (NICE) issued guidance on the diagnosis and management of FH. In Scotland, Northern Ireland and Wales, significant steps were taken to implement the NICE recommendations by establishing national screening and cascade testing programmes, but a report from HEART UK in 2010 sadly demonstrated that England is lagging behind.

This new Medway report discusses an approach by one Clinical Commissioning Group (CCG) to address the recommendations made in the NICE guidance and improve diagnosis of FH within primary care. It is a huge step forward and offers a model that could be implemented within other CCGs in England; to systematically identify people at risk of FH and offer them appropriate treatment.

I am very grateful to all who have contributed to the work of this audit. It demonstrates how successful collaboration between commissioners (CCG) and a charity (HEART UK), together with industry support (Sanofi), can help improve patient care. There are, of course, lessons to be learnt and any model can be improved. To that end, this report provides a number of useful recommendations. I am delighted that HEART UK are planning to make the FH audit tool widely available to GPs and are seeking support to extend the FH Nurse Advisor Programme beyond the Medway region.

Initiatives such as this demonstrate the way in which services can be improved to help reduce the future burden of coronary heart disease in this group of patients. FH need no longer be a 'silent killer'.

EXECUTIVE SUMMARY

Familial hypercholesterolaemia (FH) is a relatively common autosomal dominant lipid disorder that confers a lifelong risk of premature coronary heart disease (CHD) because of highly raised low-density lipoprotein-cholesterol (LDL-C). FH is one of the most common inherited disorders. Worldwide, between 14 million and 34 million people are likely to have FH, with some 1.8–4.5 million in Europe and at least 120,000 in the UK. The UK 2010 National Audit of the Management of FH estimated that around 15,000 patients are diagnosed with FH in the UK. Therefore, at least 100,000 cases of FH are undiagnosed, and this points to severe under-diagnosis and under-treatment of FH in the UK.

Cascade testing is a mechanism for identifying people at risk for a genetic condition, such as FH, by a process of systematic family tracing. For FH, cascade testing of close relatives who carry a 50% risk of the disorder is a recommended and cost-effective approach to diagnosing new patients. It is estimated that cascade testing may identify approximately 50% of people with FH (at least as estimated for the UK population); therefore, to further improve FH diagnosis, other strategies are required to identify new index cases.

Despite the 2008 National Institute for Health and Care Excellence (NICE) guideline recommendation for genetic testing of index cases and cascade testing, and the publication of the NICE Quality Standard for the management of FH in August 2013, no systematic diagnostic testing programme has yet been introduced in England, although there are active programmes in Scotland, Wales and Northern Ireland.

Primary care provides an opportunity to systematically identify new index FH cases for diagnosis, testing and treatment via data already held within GP computer systems. The Medway FH audit was prompted in response to the failure of the 2008 NICE guidance to produce significant improvements in diagnosis of FH in England.

The Medway FH Audit Tool enabled the identification of patients at risk of FH from data already available within the patients' electronic primary care clinical record. It was piloted in a single practice in September 2011 to test and optimise performance, and in October 2011 it was rolled out across the Clinical Commissioning Group (CCG) (then a Primary Care Trust). The audit first identified patients already diagnosed with FH or possible FH, thus providing a baseline prevalence. Next, all undiagnosed patients with elevated total cholesterol and/or LDL-C were identified and those previously assessed using the Simon Broome criteria were excluded. On-screen prompts highlighted these 'at risk and unscreened' patients to the GP for assessment and diagnosis using the Simon Broome criteria when the patient was next consulted. In addition, a list of patients in need of assessment could be generated at practice level. At a CCG level, patient numbers could be seen to monitor progress and target support.

In October 2011, the baseline prevalence of FH within Medway CCG (population 260,000) was 0.13% (one in 750), and 0.59% of patients (around 1600) were 'at risk and unscreened'. In 2 years, the prevalence of patients diagnosed with FH within the Medway CCG increased by 0.09% to 0.22%, increasing from one in 750 patients to one in 450. However, the proportion of patients at risk and unscreened remained the same.

In October 2013, an FH Nurse Advisor Programme was introduced into the CCG with the aim of further improving the rate of diagnosis of FH in Medway. From initiation of the FH Nurse Advisor Programme in October 2013 until programme end in July 2014 (9 months), the prevalence of patients diagnosed with FH within Medway CCG increased to 0.28% (one in 357). Following the programme, the proportion of patients at risk and unscreened reduced by three-quarters to 0.14%.

HEART UK acknowledge all who have contributed to and supported the work of the audit, in particular the members of the Medway FH Audit Steering Committee, the GP practices within Medway CCG and Sanofi for their support of the project. HEART UK is encouraged by the success of this model and will advocate its rollout to CCGs across England. In the long term, such programmes will improve diagnosis with the inevitable benefit of better management and treatment and ultimately prevention of CHD in these patients.

KEY RECOMMENDATIONS

1. The Medway FH Audit Tool is incorporated into other CCGs and GP IT systems throughout England.
2. An FH Nurse Advisor Programme is supported and established within each participating CCG to further improve diagnosis and treatment.
3. The FH Nurse Advisor Programme is expanded and all patients 'at risk and unscreened' are invited to a clinic for assessment.
4. A Practice Nurse is assigned to the FH Nurse Advisor Programme and attends clinics to gain education and training, to enable them to continue testing and managing FH patients after completion of the programme, thus leaving a lasting legacy and providing continuity of care.
5. Patient participation in future programmes is improved by addressing the methods used to engage and encourage patients to attend FH Nurse Advisor clinics.

To support these recommendations HEART UK will:

6. Continue to support GP practices within Medway and those in other CCGs adopting the model with provision of primary care guidance materials.
7. Support the integration of primary care FH diagnosis with secondary care referral for specialist management, including genetic testing, through its advocacy and projects of the FH Implementation Team.
8. Engage with secondary care facilities to support any potential increase in FH patient diagnosis within the CCG to ensure they are prepared for and are aware of the potential numbers of new diagnoses (based on the Medway practice, this could be a doubling of cases).
9. Continue to promote public awareness of the health risk of elevated cholesterol, the importance of knowing your family history and the importance of an FH diagnosis to yourself and family members.
10. Encourage patient participation and facilitate patient support groups through the HEART UK Ambassador programme.

INTRODUCTION

Familial hypercholesterolaemia (FH) is a relatively common autosomal dominant lipid disorder that confers a lifelong risk of premature coronary heart disease (CHD) because of highly raised low-density lipoprotein-cholesterol (LDL-C).[1]

FH is one of the most common inherited disorders, with an estimated prevalence of one in 500 (0.2%), but more recent estimates suggest that the prevalence may be much higher, approaching one in 200 (0.5%).[2,3] These prevalence estimates indicate that worldwide between 14 million and 34 million people are likely to have FH, with some 1.8–4.5 million in Europe and at least 120,000 in the UK. The European Atherosclerosis Society (EAS) estimates that less than 1% of patients are diagnosed in most countries, although there are exceptions, for example Norway.[2]

Untreated, the elevated LDL-C that characterises FH leads to a greater than 50% risk of CHD in men by the age of 50 years and at least 30% in women by the age of 60 years.[4] Effective treatments for FH include lifestyle modification, including dietary fat restriction, exercise and avoidance of smoking, and lipid-lowering treatments such as HMG CoA (hydroxymethylglutaryl co-enzyme A) reductase inhibitors (i.e. statins). Statins are an effective therapy and clinical trials have shown CHD risk reductions of up to 80% compared with that of the general population, especially if treatment is initiated prior to the onset of CHD.[5,6] The early onset of atherosclerosis caused by FH emphasises the importance of early identification and effective therapeutic intervention. In patients with established CHD, the benefits of preventive measures are significantly attenuated.[7]

FH is commonly caused by single gene mutations in the LDL receptor (*LDLR*), apolipoprotein B (*APOB*) and more rarely in the proprotein convertase subtilisin/kexin type 9 (*PCSK9*) gene, which encode for proteins critical for the normal removal of excess LDL-C from the bloodstream.[8] *LDLR* gene mutations are the most frequent cause of FH, with more than 1200 different mutations identified to date.[9] The majority of people with FH are heterozygotes and have inherited one disease-causing mutation. Heterozygous FH occurs in approximately one in 500 people, and up to one in 70 in certain ethnic groups with founder mutations.[1] Rarely, a person will inherit a genetic mutation from both parents to give them homozygous FH, which affects approximately one in 1 million people.[10] Clinically, the inheritance of two copies of a mutated FH gene results in extremely high LDL-C that requires aggressive lipid-lowering drug therapy and, if available, lipoprotein apheresis from a young age.[2]

In populations in which no founder effect has occurred, such as the UK, approximately 40% of people with clinically suspected FH carry an identifiable mutation.[11] Recently, it has been suggested that FH in mutation-negative patients may be caused by an accumulation of common small-effect LDL-C-raising alleles—so-called polygenic FH;[12] however, even when polygenic cases are combined with proven mutations, there remains a substantial proportion of phenotypic FH cases that do not have a genetic diagnosis.[12] The 2013 National Institute for Health and Care Excellence (NICE) Quality Standard for FH (QS41) now recommends that cascade testing resources should be focused on patients with a confirmed diagnosis of monogenic FH, which is the strategy adopted with great success in Holland and in Wales.[13] Although funding for genetic testing for FH is not yet widely available, it is likely that, overall, the use of such testing will improve the cost-effectiveness of the cascade process, since 50% of first-degree relatives will be affected in monogenic families.

Under-diagnosis of FH – a major gap in coronary disease prevention

It was previously accepted that the prevalence of heterozygous FH was about one in 500, based on calculations using the Hardy-Weinberg equation and the frequency of FH homozygotes; however, recent data suggest that this is an under-estimate.[2] The Copenhagen General Population Study used the Dutch Lipid Clinic Network (DLCN) score to establish the clinical diagnosis of FH and determined that the prevalence in individuals classified as definite or probable FH approached one in 200.[2,3]

Extrapolations from this range of one in 500 to one in 200 suggest that there are between 120,000 and 300,000 people with FH in the UK. The UK 2010 National Audit of the Management of FH estimated that around 15,000 patients are diagnosed with FH in the UK.[14] This estimate matches closely with a survey in 2008, which showed that approximately 15,000 adults and approximately 500 children with FH were being managed in UK lipid clinics.[14] Estimates vary, but fewer than 12% of cases of FH are diagnosed in the UK.[2] Therefore, at least 100,000 cases of FH are undiagnosed, and this points to severe under-diagnosis and under-treatment of FH in the UK.[14]

Despite the 2008 NICE guidelines' recommendation for genetic testing of index cases and cascade testing[4] and the publication of the NICE Quality Standard for the management of FH in August 2013 (QS41),[13] no systematic diagnostic testing programme has yet been introduced in England, although there are active programmes in Scotland, Wales and Northern Ireland.[11]

Diagnosing FH in index cases

The diagnosis of FH relies on five criteria: very high LDL-C on repeat measurements, family history, clinical history of premature CHD, physical examination for xanthomas and corneal arcus, and/or a causative mutation detected by molecular genetics. Secondary causes of hyperlipidaemia should be excluded by establishing that there is no hyperglycaemia or albuminuria and by determining normal levels of liver enzymes, renal function and thyroid hormones.

Clinical diagnostic algorithms for FH are well defined, but there is no one internationally agreed algorithm. In the UK, the Simon Broome criteria[1,15] is recommended to evaluate patients with raised LDL-C, especially if there is a personal or family history of premature CHD. A diagnosis of *'definite FH'* is made based on total cholesterol >6.7 mmol/L or LDL-C >4.0 mmol/L in a child (<16 years) or total cholesterol >7.5 mmol/L or LDL-C >4.9 mmol/L in an adult plus the presence of tendon xanthomas in the patient or a first-degree or second-degree relative, or the identification of an FH-causing mutation. A diagnosis of 'possible FH' is made if there are no tendon xanthomas but a family history of myocardial infarction (aged <50 years in a second-degree relative or <60 years in a first-degree relative) or a family history of raised total cholesterol (>7.5 mmol/L in an adult first-degree or second-degree relative or >6.7 mmol/L in a child or sibling aged <16 years) (**Table 1**).

In Europe, the DLCN criteria is widely used and calculates a numerical score to predict the probability of diagnosing FH. This scoring system is increasingly accepted as simple and comprehensive[16], categorising patients as having definite, probable or possible FH[1] (**Table 2**).

Table 1: The Simon Broome Register criteria (total cholesterol and LDL-C levels either pre-treatment or highest on treatment)[1, 15]

Definite FH	Possible FH
Total cholesterol >6.7 mmol/L or LDL-C >4.0 mmol/L in a child aged <16 years OR Total cholesterol >7.5 mmol/L or LDL-C >4.9 mmol/L in an adult	Total cholesterol >6.7 mmol/L or LDL-C >4.0 mmol/L in a child aged <16 years OR Total cholesterol >7.5 mmol/L or LDL-C >4.9 mmol/L in an adult
PLUS	**AND AT LEAST ONE OF THE FOLLOWING**
Tendon xanthomas in the patient or a first-degree (parent, sibling or child) or second-degree relative (grandparent, uncle or aunt) OR DNA-based evidence of an *LDLR* mutation, familial defective *APOB-100*, or a *PCSK9* mutation	A family history of myocardial infarction: <50 years of age in second-degree relative or <60 years of age in first-degree relative OR A family history of raised total cholesterol: >7.5 mmol/L in an adult first-degree or second-degree relative or >6.7 mmol/L in a child or sibling aged <16 years

Table 2: DLCN score for FH[1]

GROUP	Score
GROUP 1: FAMILY HISTORY	
First-degree relative with known premature coronary and/or vascular disease (men <55 years, women <60 years) OR First-degree relative with known LDL-C above the 95th percentile for age and sex	1
First-degree relative with tendinous xanthomata and/or arcus cornealis OR Children aged <18 years with LDL-C above the 95th percentile for age and sex	2
GROUP 2: CLINICAL HISTORY	
Patient with premature coronary artery disease (ages as above)	2
Patient with premature cerebral or peripheral vascular disease (as above)	1
GROUP 3: PHYSICAL EXAMINATION	
Tendinous xanthomata	6
Arcus cornealis prior to age 45 years	4
GROUP 4: LDL-C (mmol/L)	
≥8.5	8
6.5–8.4	5
5.0–6.4	3
4.0–4.9	1
GROUP 5: DNA ANALYSIS	
Functional mutation in the *LDLR, APOB* or *PCSK9 gene*	8
SCORE	
>8	Definite FH
6-8	Probable FH
3-5	Possible FH
0-2	Unlikely FH

In the US, the MEDPED system was developed to help improve FH diagnosis and treatment. This algorithm relies on plasma total cholesterol and LDL-C and strictly requires that cholesterol measurements are known in first-degree family members.[16] The Japanese criteria is comparable with the Simon Broome criteria but uses population-specific LDL-C measurements and includes radiographic diagnosis of Achilles tendon xanthomata.[16]

The presence of an FH-causing mutation provides a definitive diagnosis of the disorder, and in the UK approximately 40% of people with clinically suspected FH carry an identifiable mutation.[11] Based on the Simon Broome criteria, a UK study identified mutations in 73% of patients with 'definite FH' and 30% of patients with 'possible FH'.[17] Using the DLCN score, mutations were detected in 54% of patients with definite FH (>8), 39% with probable FH (6-8) and 28% with possible FH (3-5).[17]

Cascade testing

Cascade testing is a mechanism for identifying people at risk for a genetic condition, such as FH, by a process of systematic family tracing. For FH, cascade testing of close relatives who carry a 50% risk of the disorder is a recommended and cost-effective approach to diagnosing new patients.[4] Cascade testing for FH can be performed using phenotypic (clinical features) or genotypic approaches, but cascade testing based on an identified pathogenic mutation is more accurate and cost effective.[18]

In the Netherlands, systematic sassessment and family cascade testing was established in 1994[19] and has led to the diagnosis of more than 33,000 patients with FH to date.[2] Based on a population prevalence of one in 500, the programme has identified more than 70% of the expected number of cases of FH in the Netherlands. In the UK, cascade testing initiatives have been established in Scotland, Wales and Northern Ireland, but there is no systematic programme established in England. Identifying relatives allows for significant health-affecting interventions to be administered, which can extend life expectancy significantly, especially if administered before the onset of CHD. It is estimated that cascade testing may identify approximately 50% of people with FH (at least as estimated for the UK population)[4]; therefore, to further improve FH diagnosis, other strategies are required to identify new index cases.

Cascade testing from an index case - making a difference to a family's risk of CHD

Hazel Gallagher was a fit, healthy, young mum of two who ate well, did not smoke and played competitive squash. No-one would have suspected that she had dangerously elevated cholesterol levels. Hazel was diagnosed with FH after visiting a Consultant to discuss an unrelated health issue. The Consultant noticed that Hazel had the visible signs of xanthoma on her knuckles and Achilles heel. A blood test revealed a total cholesterol level of 9.8 mmol/L, she was diagnosed with FH and immediately prescribed lipid-lowering therapy.

Following Hazel's diagnosis, family cascade testing was initiated. Both of her children—James aged 2 and Darren aged 8—were diagnosed and treatment initiated, and her sister, niece and mother were all diagnosed. Hazel had a significant family history of CHD; her uncle had died of 'hardening of the arteries' in his 40s, leaving behind four sons who all developed heart disease in their 30s. Two years ago, Hazel's diagnosis was confirmed by mutation testing and two of her grandchildren were found to also carry the FH mutation. Her eldest grandchild (aged 5 years) is already receiving treatment. Hazel says *"DNA testing is so much less invasive for a small child, as it only requires a small sample of saliva rather than a blood sample."*

Cascade testing has now traced five generations of FH in Hazel's family, from her grandmother down to her 3-year-old granddaughter.

Originally prescribed a bile acid sequestrant (Questran), Hazel has subsequently been treated with all the available cholesterol-lowering drugs. She took part in a clinical trial for a statin in 1985. Thirty years on, Hazel continues to be treated with statins and has tolerated them well. Hazel did not conform to the typical 'high cholesterol' stereotype and her opportunistic diagnosis may not have happened at such a young age but for the interest of her Consultant. Hazel became a pioneering patient advocate for raising awareness of FH and was one of the two founders of the 'Family Heart Association', now flourishing as 'HEART UK'. She is a passionate supporter of the Medway FH audit and the systematic approach to diagnosing new cases of FH within primary care. She is the HEART UK patient representative on the Medway Audit Steering Committee and said *"I really welcome this approach to help increase the identification of index FH cases. The audit supports GPs to make a diagnosis of FH and raises awareness within primary care of the hereditary aspect to high cholesterol that is distinct from acquired high cholesterol. Patients are not always old with a poor diet and an unhealthy lifestyle"*.

Having FH has not prevented Hazel from doing anything she wants to do. After being diagnosed, she went on to become a world-class master sprinter and continues to keep extremely fit through cycling and walking. *"People need to understand that diagnosis of FH can be simple and is easily treated and managed. It is the not knowing that could kill you."*

NICE guidance

In 2008, NICE published its evidence-based clinical guideline for the identification and management of FH [4]. The guidance recommends that:

- Healthcare professionals (HCPs) should consider the possibility of FH in adults with raised cholesterol (total cholesterol >7.5 mmol/L), especially when there is a personal or family history of premature CHD;
- HCPs should exclude secondary causes of hypercholesterolaemia before a diagnosis;
- The Simon Broome criteria is used to establish a diagnosis and;
- All patients with a clinical diagnosis of FH be offered a DNA test to confirm their diagnosis and to aid diagnosis among relatives.

The guidelines advocate cascade testing using the mutation identified in the index case for all first-degree, second-degree and, when possible, third-degree biological relatives. In the absence of a DNA-based diagnosis, cascade testing using LDL-C concentration measurements should be undertaken to identify people with FH. In this case, age-specific and gender-specific cut-offs for determining affected, non-affected or undecided status are recommended.[20] The NICE FH Quality Standard lists eight key priorities (**Table 3**).[13] In light of the finding that a significant proportion of patients with a clinical diagnosis of FH but with no identified mutation are likely to have a polygenic and not monogenic cause of their elevated LDL-C levels,[12] the Quality Standard now recommends that 'Relatives of people with a confirmed diagnosis of *monogenic* FH are offered DNA testing through a nationwide, systematic cascade process',[13] emphasising the importance of DNA testing to focus cascade testing on the families at greatest risk.

Table 3: The NICE Quality Standard* for FH[13]

QUALITY STATEMENT		
1	**Diagnosis**	Adults with a baseline total cholesterol >7.5 mmol/L are assessed for a clinical diagnosis of FH
2	**Specialist referral**	People with a clinical diagnosis of FH are referred for specialist assessment
3	**DNA testing**	People with a clinical diagnosis of FH are offered DNA testing as part of a specialist assessment
4	**Diagnosis in children under 10 years**	Children at risk of FH are offered diagnostic tests by the age of 10 years
5	**Cascade testing**	Relatives of people with a confirmed diagnosis of monogenic FH are offered DNA testing through a nationwide, systematic cascade process
6	**Drug treatment in adults**	Adults with FH receive lipid-modifying drug treatment to reduce LDL-C concentration by more than 50% from baseline
7	**Drug treatment in children**	Children with FH are assessed for lipid-modifying drug treatment by a specialist with expertise in FH in a child-focused setting by the age of 10 years
8	**Annual review**	People with FH are offered a structured review at least annually

*NICE Quality Standards are concise sets of prioritised statements designed to drive measurable quality improvements within a particular area of health or care. They are derived from the best available evidence such as NICE guidance and other evidence sources accredited by NICE. They are developed independently by NICE, in collaboration with health and social care professionals, their partners and service users.

Saving lives: the benefits of identifying FH

In 2012, HEART UK published *Saving lives, saving families*, which described the health, social and economic advantages of diagnosing and managing FH.[21] The health economic modelling, commissioned by the HEART UK Familial Hypercholesterolaemia Implementation Team, demonstrated the financial and health benefits of cascade testing and use of optimal statin treatment through quality-adjusted life-years gained and cardiovascular events avoided. The report estimates that appropriate management and treatment of every 1000 FH patients (between the ages of 35 and 85 years) would lead to 101 fewer cardiovascular deaths when compared with no treatment. Overall, the potential savings to the UK are almost £380 million from CHD events avoided if all relatives of FH index cases are identified and appropriately treated. More realistically, if 50% of patients with FH are diagnosed and treated, the NHS could save £1.7 million per year on health treatment otherwise required for CHD, but not implementing cascade testing is costing the NHS £1.4 million per year.[21]

"The greater the number of FH patients identified and treated, the greater the comparative and accrued health benefits and cost savings to the NHS."

MEDWAY FH AUDIT

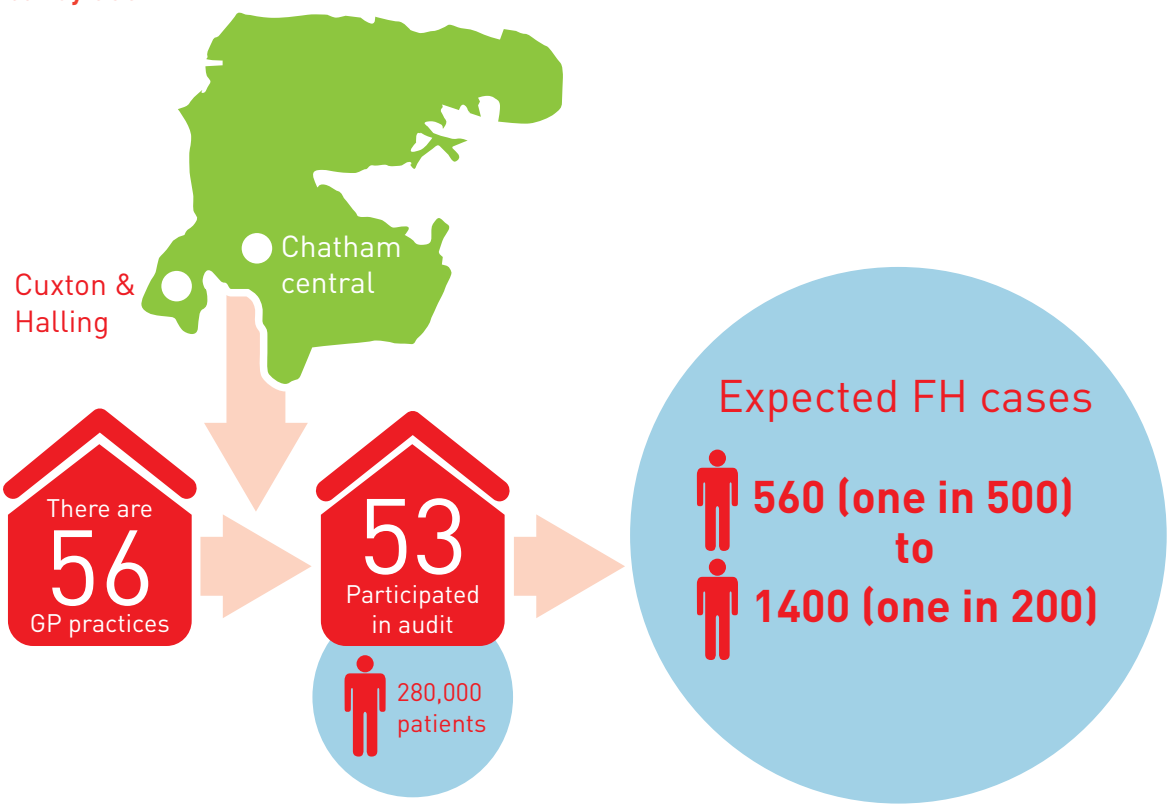
Primary care provides an opportunity to systematically identify index FH cases for diagnosis and treatment via data held within GP IT systems. GP practices are essential in managing patients with elevated cholesterol and as points of referral to secondary care. [21] The Medway FH audit was prompted in response to the 2008 NICE guidance[4], which was failing to produce significant improvements in diagnosis of FH in England.

The Medway Clinical Commissioning Group (CCG) in Kent comprises 56 GP practices. Fifty-three practices took part in the audit (Appendix A), serving approximately 280,000 patients (data accurate at July 2014) (**Box 1**). Across the Medway CCG, the recorded prevalence of FH is significantly below the level predicted for the UK. Whilst the NHS Health Check programme will identify raised cholesterol in those who attend, those with undiagnosed FH would benefit from earlier diagnosis and treatment.

Virtually all GP practices are now computerised, and surgeries have been recording patient data within their IT systems for more than 20 years.[22] These clinical IT systems contain information about cholesterol measurements, a personal history of early ischaemic heart disease (IHD) or a history of a relative with early IHD or raised cholesterol. Major diseases are coded, via the NHS-wide, alpha-numeric coding system of Read Codes. Read Codes are designed to record the everyday care of a patient and enable computerised patient records to be electronically searched. In order to support the Medway audit, additional Read Codes were requested to allow a code for possible FH, probable FH, and DLCN scores, as well as additional modifications made to existing codes.

Technology in the form of 'Audit +' (BMJ Informatica)[23] is in use in GP surgeries, helping practices deliver best practice care through prompts during consultations for a variety of diseases. Prompts built into GP systems can help them achieve improvements in patient care and can be applied to any area requiring improvement that can be audited/measured using Read Codes or numeric or demographic information. The use of prompts, as well as audit reports, acts as a performance-enhancing tool. In collaboration with BMJ Informatica, Dr Peter Green, Chief Clinical Officer of Medway CCG (see **Box 2**), has established a suite of audits utilising the Audit + software to support GP practices to deliver the CCG's first three strategic objectives: prevention, early diagnosis and better care. The Audit + software is compatible with multiple GP clinical platforms and is loaded remotely onto GP clinical systems, requiring no additional work for the practices or clinicians. At Medway, it was felt that the diagnosis of FH was amenable to the Audit + software and an audit was instigated to increase the diagnosis of FH within the CCG.

BOX 1: Medway CCG



BOX 2: Dr Peter Green, Chief Clinical Officer, Medway CCG

Dr Peter Green is the Chief Clinical Officer of Medway CCG and a GP with more than 20 years of experience. Pete has a special interest in quality of care and the systems that can be developed to support this. His interest in medical audit systems led to him becoming Chair of the Medway Medical Audit Advisory Group and Co-Chair of the West Kent Medical Audit Advisory Group. He also works with the *British Medical Journal's* Health Analytics division to help identify areas where care for patients can be improved by the use of technology within a consultation and information collated at practice and CCG levels. In 2007, Pete was appointed Medical Director of NHS Medway Primary Care Trust (PCT) and remained a Co-Medical Director of the NHS Kent and Medway PCT Cluster until the formation of CCGs in April 2013. During his time as Medical Director, he has held roles as Director of Quality for all aspects of commissioned and provided care, Director of Commissioning for all acute, community and primary care services, and Director of Performance for all registered GPs, pharmacists, dentists and optometrists. He oversaw improvements in the quality of General Practice measured by the National Quality and Outcomes Framework to being above those of neighbouring PCTs. He works as a GP 1 day a week, which he sees as essential in helping him keep in touch with what's happening from a patient's perspective. Pete has always seen his involvement in the PCT, and now the CCG, as an extension of what he and many other GPs do on a daily basis when seeing patients: to lessen the risk of them becoming unwell and helping them to get better if they do.

Medway FH Audit Tool

The Medway FH Audit Tool, developed in accordance with the Royal College of GPs' standard criteria for audits, was prompted by the NICE clinical guidance for FH[4] and the National Quality and Outcomes Framework for measuring improvements in GP practices.[24] The aim of the FH Audit Tool was to enable the identification of patients who are at risk of FH from electronic databases in primary care. Patients were identified if they had elevated cholesterol levels, but had not yet been diagnosed or screened via the Simon Broome criteria ('at risk and unscreened'). On-screen prompts highlighted these patients to the GP for assessment, diagnosis and appropriate management. In addition, a list of patients in need of evaluation could be generated at practice level. At a CCG level, the numbers of patients (but not the names or any patient identifiable data) could be seen to monitor progress and target support.

PHASE I – ESTABLISHING THE BASELINE AND IMPROVING FH DIAGNOSIS

NICE guidance recommends that all patients with a diagnosis of 'definite FH' or 'possible FH' based on the Simon Broome criteria are managed in the same way. The NICE guidelines were initially difficult to implement as there was no NHS Read Code for 'possible FH'. The Read Code for possible FH was provided by the NHS in June 2010 and the audit was planned.

The FH Audit Tool was piloted in a single practice in September 2011 to test and optimise performance. In October 2011, the Audit Tool was rolled out across the CCG (then a PCT). Practices across the Medway CCG were familiar with the Audit + software and the audit process and no additional training was required at initiation of the audit but practices were supplied with information on FH and the Simon Broome criteria.

The audit first identified patients already diagnosed with FH or possible FH, thus providing a baseline prevalence. Next, all undiagnosed patients with elevated total cholesterol and/or LDL-C (**Table 4**) were identified and those previously assessed using the Simon Broome criteria were excluded. From this, the Audit Tool produced a list of patients 'at risk and unscreened' for each practice, and added prompts to these patients' notes, which appeared when the clinician saw the patient, recommending them to be assessed using the Simon Broome criteria. Those who met the criteria were diagnosed as having FH or possible FH. In addition, the audit contained a series of triggers that encouraged further management steps at the point of consultation, allowing systematic patient evaluation (see **Table 5**).

Audits could be conducted within practices on a daily basis. At the CCG level, audits could be conducted weekly, but progress was monitored monthly. Formal re-audit and comparison with the baseline was performed at 2 years.

Table 4: Cholesterol levels used as selection criteria for identifying 'at risk' individuals from the Audit Tool – all levels either pre-treatment or the highest on treatment

	TOTAL CHOLESTEROL	LDL-C
Child/young person (<16 years)	>6.7 mmol/L	>4.0 mmol/L
Adults	>7.5 mmol/L	>4.9 mmol/L

Table 5. Triggers and prompts within the Medway FH Audit Tool

TRIGGER	PROMPT
Patients with FH or possible FH whose family has not been informed	Have relatives been informed regarding FH?
Patients with FH, possible FH or probable FH whose latest total cholesterol is >5 mmol/L	Up-titrate statins or consider referral
Patients whose latest cholesterol is >7.5 mmol/L or LDL-C >4.9 mmol/L and who have had a positive genotype test	Diagnose FH
Patients whose latest cholesterol is >7.5mmol/L or LDL-C >4.9 mmol/L and have a family history of premature CHD and/or hypercholesterolaemia and have not had a Simon Broome assessment	Consider possible FH
Patients whose latest cholesterol is >7.5 mmol/L or LDL-C >4.9 mmol/L, have not had a Simon Broome assessment and have a family history of CHD but no details of the age of the relatives	Ask patient if myocardial infarction has occurred before 50 years of age in a second-degree relative or before 60 years of age in a first-degree relative Yes: Consider FH No: Assess using Simon Broome criteria

Note: Prompts contain further information along with relevant Read Codes, which can be added directly into the patient record from the prompt screen.

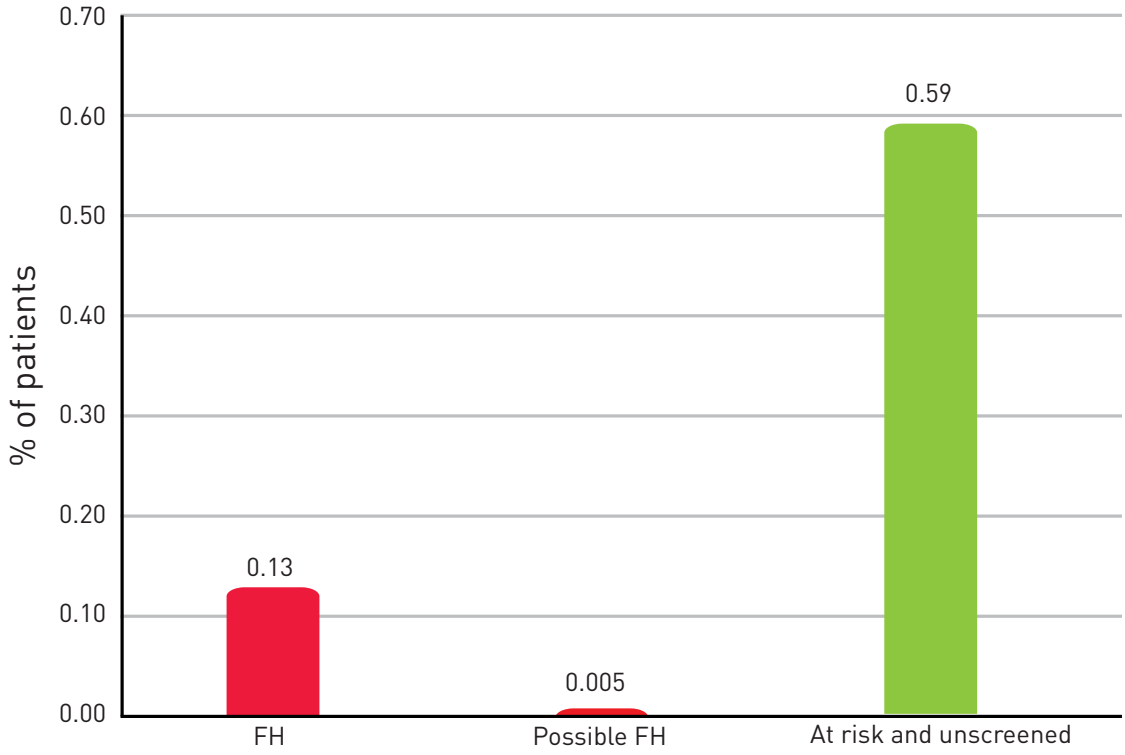
Results: Establishing the baseline

Baseline FH prevalence: The audit identified patients already diagnosed with FH or possible FH and established the baseline prevalence of FH within Medway CCG of 0.13% (one in 750) (**Figure 1; Table 6**).

Baseline 'at risk and unscreened': The audit identified the baseline 'at risk and unscreened' prevalence of FH within Medway CCG of 0.59% (**Figure 1; Table 6**).

Estimated diagnostic workload: In the context of a GP practice with a population of 10,000 patients, there would be approximately 60 'at risk and unscreened' patients.

Figure 1: Medway FH Audit results at baseline (October 2011)



FH diagnoses made by Simon Broome criteria; Patients were considered to be 'at risk and unscreened' if they had a total cholesterol >7.5 mmol/L and/or LDL-C >4.9 mmol/L and had not been assessed using the Simon Broome criteria.

Table 6: Medway FH Audit Tool results at baseline (October 2011)

	NUMBER	POPULATION	PREVALENCE (%)
FH*	331	262, 030	0.13
Possible FH*	12	262,030	0.005
Total FH	343	262,030	0.13
At risk and unscreened	1553	262,030	0.59

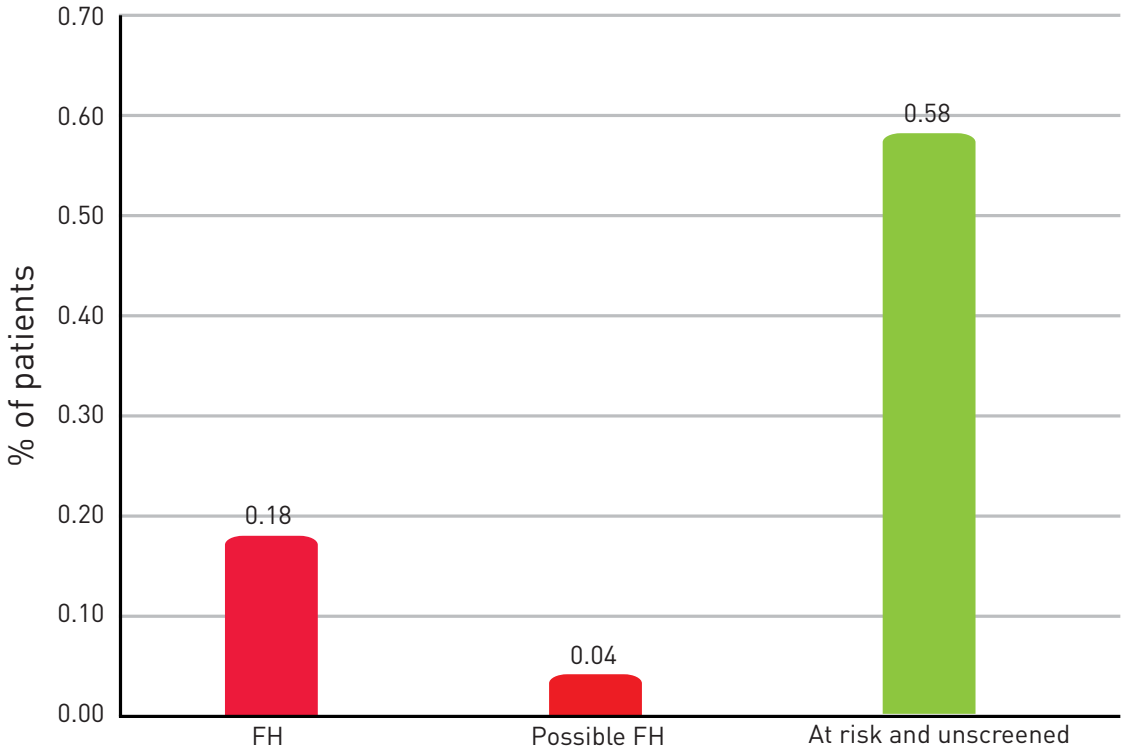
* FH diagnoses made by Simon Broome criteria

Re-audit at 2 years

FH prevalence after 2 years: Re-audit showed a substantial increase in the prevalence of diagnosed FH, increasing to 0.22% (one in 450) (**Figure 2; Table 7**).

'At risk and unscreened': Despite the increase in FH diagnosis, the proportion of patients 'at risk and unscreened' remained almost unchanged at 0.58% (**Figure 2; Table 7**).

Figure 2: Medway FH Audit results at re-audit (October 2013)



FH diagnoses made by Simon Broome criteria; Patients were considered to be 'at risk and unscreened' if they had a total cholesterol >7.5 mmol/L and/or LDL-C >4.9 mmol/L and had not been assessed using the Simon Broome criteria.

Table 7: Medway FH Audit Tool results at re-audit (October 2013)

	NUMBER	POPULATION†	PREVALENCE (%)
FH*	354	199,346	0.18
Possible FH*	88	199,346	0.04
Total FH	442	199,346	0.22
At risk and unscreened	1164	199,346	0.58

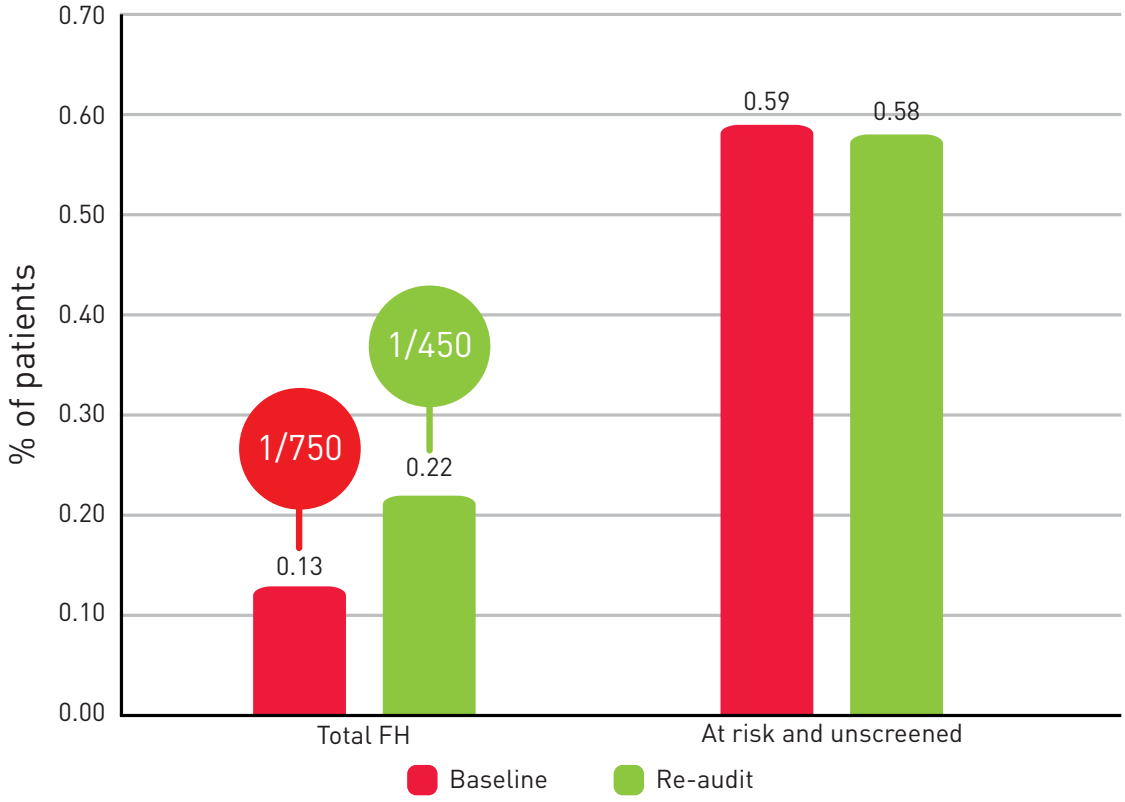
*FH diagnoses made by Simon Broome criteria

† Population (and number of FH) is lower than previous time-point; data could not be extracted from all electronic medical information systems at this time.

Improving FH prevalence

In 2 years the prevalence of patients diagnosed with FH within the Medway CCG improved by 0.09%, increasing from one in 750 patients to one in 450. However, the proportion of patients at risk and unscreened remained the same (**Figure 3**).

Figure 3: Medway FH Audit results at baseline and re-audit at 2 years



PHASE II – THE FH NURSE ADVISOR PROGRAMME

Introduction

In October 2013, an FH Nurse Advisor Programme was introduced into the CCG with the aim of further improving the rate of diagnosis of FH in Medway. The decision to implement the programme was in response to feedback from participating GPs towards the end of Phase I of the audit that highlighted a need for additional resources to assess patients at risk and to diagnose FH.

It was intended that the FH Nurse Advisor Programme would assist practitioners with the implementation of the 2008 NICE Clinical guidelines[4] and that patients would receive optimal management following a clinical assessment and possible secondary care referral. Overall, it was intended that this programme would support a reduction in cardiovascular events in patients through raised awareness, education and early identification of FH.

At this point, a decision was made to revise the audit and incorporate the DLCN score to define the severity of FH and support clinical management. New Read Codes for DLCN score and probable FH were requested and issued. The six sections of the current Medway FH Audit Tool are shown in **Box 3**.

BOX 3: The six sections of the Medway FH Audit Tool

1. FH (definite, possible, probable)
2. High cholesterol (excluding all FH*)
3. DLCN score (excluding all FH*)
4. Simon Broome assessment
5. Personal history of CHD
6. Family history of FH (*all FH = all definite, possible and probable cases)

The FH Nurse Advisor Programme was a collaboration between NHS Medway CCG and HEART UK, supported by Sanofi. Ashfield Healthcare provided the service and employed the Nurse Advisor. A single Nurse Advisor was appointed and visited all practices (**Box 4**).

BOX 4: Medway FH Nurse Advisor

Tanya Sanders was a Community Matron and is currently employed by Ashfield Healthcare Ltd as an FH Nurse Advisor. Tanya's nursing career spans over 20 years beginning at Basingstoke & Winchester School of Nursing, where she qualified as a Registered General Nurse in 1994. She spent several years broadening her nursing experience both in the UK and abroad and covered various fields including vascular surgery, oncology and nursing recruitment. In 2000 she began a career in accident and emergency (A&E) nursing at Chelsea and Westminster Hospital, eventually becoming an Emergency Nurse Practitioner. In 2010, she left A&E to become a Community Matron, a particularly challenging role that involved the management of patients with multiple complex long-term conditions. Tanya has a BSc in Health and Social care and completed her FH training with HEART UK in 2013.

Governance

The FH Nurse Advisor was governed by the Nursing and Midwifery Council Code of Professional Conduct and was subject to pre-employment checks, which include references, qualification verification, competency assessment, Disclosure and Barring Service checks (enhanced disclosure) and pre-employment vaccination assessment.

Ashfield Healthcare is registered with the Care Quality Commission (England and Wales and the equivalent bodies for Scotland and Northern Ireland) for the purposes of the delivery of healthcare services. The FH Nurse Advisor Programme adhered to The Ashfield Healthcare Clinical Governance and Risk Management Framework, The Health and Social Care Act 2008 updated 2012, Caldicott Guidelines, The Data Protection Act 1998, and the NICE guideline for identification and management of FH.

Initial programme set-up

The FH Nurse Advisor contacted each Practice Manager to arrange an initial meeting with key practice personnel, including an identified lead GP, Practice Nurse and Receptionist. The initial meeting was structured to cover:

- Programme objectives
- Service operating procedure
- Audit criteria
- The cascade letter

During the initial meeting a contract was agreed and signed between the practice and the Nurse Advisor provider, which set out the conditions of the service the FH Nurse Advisor would provide to the practice and granted their legitimate access to patient data.

The service provided by the FH Nurse Advisor consisted of three main components:

- Audit list validation
- FH Nurse patient clinics
- Administration visit

Audit list validation

Before any patients were invited to the clinic, the FH Nurse Advisor reviewed the audit list to identify if any clinical or non-clinical parameters were missing in individual patient records that would prevent the DLCN score being calculated. Any missing clinical parameters were sought from the relevant HCP and the DLCN score was calculated; for those patients for whom non-clinical parameters were missing, an invitation to the FH clinic was issued and subsequently the DLCN score calculated. The management pathway of each patient was based on the DLCN score as calculated by the FH Nurse Advisor (**Table 8; Figure 4**).

Table 8: DLCN scores used to determine patient management pathway for the FH Nurse Advisor Programme

DLCN SCORE	RISK CATEGORY	ACTION
>8	Definite FH	Referred to GP for further assessment and management followed by a review by the FH Nurse Advisor for education and discussion
6-8	Probable FH	Referred to GP for further assessment and management followed by a review by the FH Nurse for education and discussion
3-5	Possible FH	Patient referred to other HCPs if necessary and as appropriate (blood pressure check, lifestyle advice etc.)

FH Nurse patient clinics

All patients identified in the audit list validation process who met the inclusion criteria for an FH Nurse Advisor review and had been scored as having a high or probable risk of FH were invited to clinic in the form of a patient invitation letter (**Appendix B1**) issued by the GP practice staff. Appointments were made at approximately 30-minute intervals to allow adequate time to provide individualised patient education and lifestyle advice.

At the clinic, each patient was provided with a copy of the Patient Service User Guide (**Appendix B2**) and a leaflet explaining FH[25] and the purpose of the clinic was explained. The FH Nurse answered any questions and obtained written consent (**Appendix B3**). During the appointment, clinical examination for xanthoma or corneal arcus was conducted, family history and cascade testing were discussed, and patients were provided with cascade letters to pass onto their first-degree relatives (**Appendix B4**). The role of lifestyle factors and family history were discussed with the patient and information and advice were provided, which were aimed at improving patient concordance to prescribed hypercholesterolaemia medication and increasing the patient's understanding of their disease using a HEART UK fact sheet.[26] The FH Nurse Advisors role was limited to discussing the disease severity and any identified management issues for each patient based on the clinical assessment and NICE guidelines.

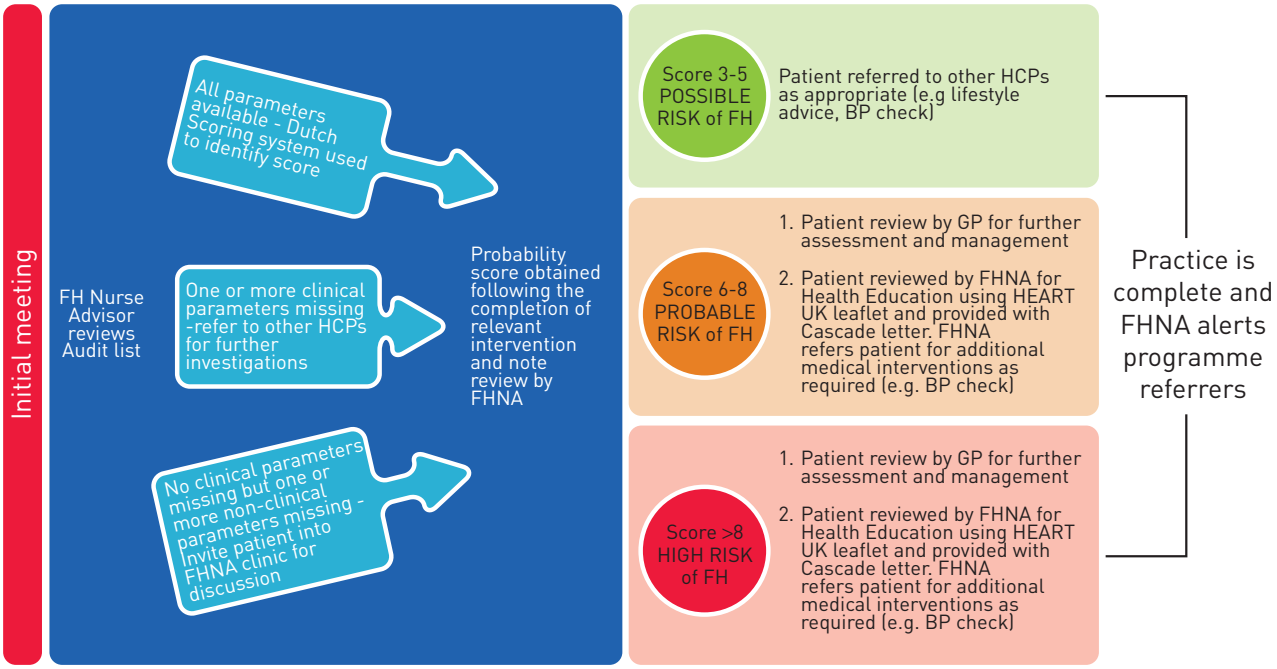
Patients attending the clinic were offered the opportunity to provide feedback on the quality of the services that they had received via a questionnaire (**Appendix B5**).

In the event of a patient not attending their clinic appointment, the FH Nurse Advisor discussed the patient with the GP practice lead clinician for appropriate follow-up by the practice.

Administration visit

Following each patient clinic, the FH Nurse Advisor arranged a suitable date for an 'Administrative Clinic' with the practice lead to ensure all clinical findings were documented in the patient's electronic clinical record. If not already carried out, an individual DLCN score was calculated by the FH Nurse Advisor for each patient. Medical interventions were decided by the GP based on the findings and the individual score for each patient. Patients identified as having definite FH were recommended for referral to secondary care for specialist management. Patients with probable or possible FH were recommended for management in primary care, but were to be referred to secondary care if their cholesterol levels did not stabilise, if their relatives had required more intensive specialist management, or if they had a particularly prominent family history of vascular events.

Figure 4: Overview of FH Nurse Advisor Programme



FHNA: Familial Hypercholesterolaemia Nurse Advisor
HCPs: Healthcare professionals
BP: Blood pressure

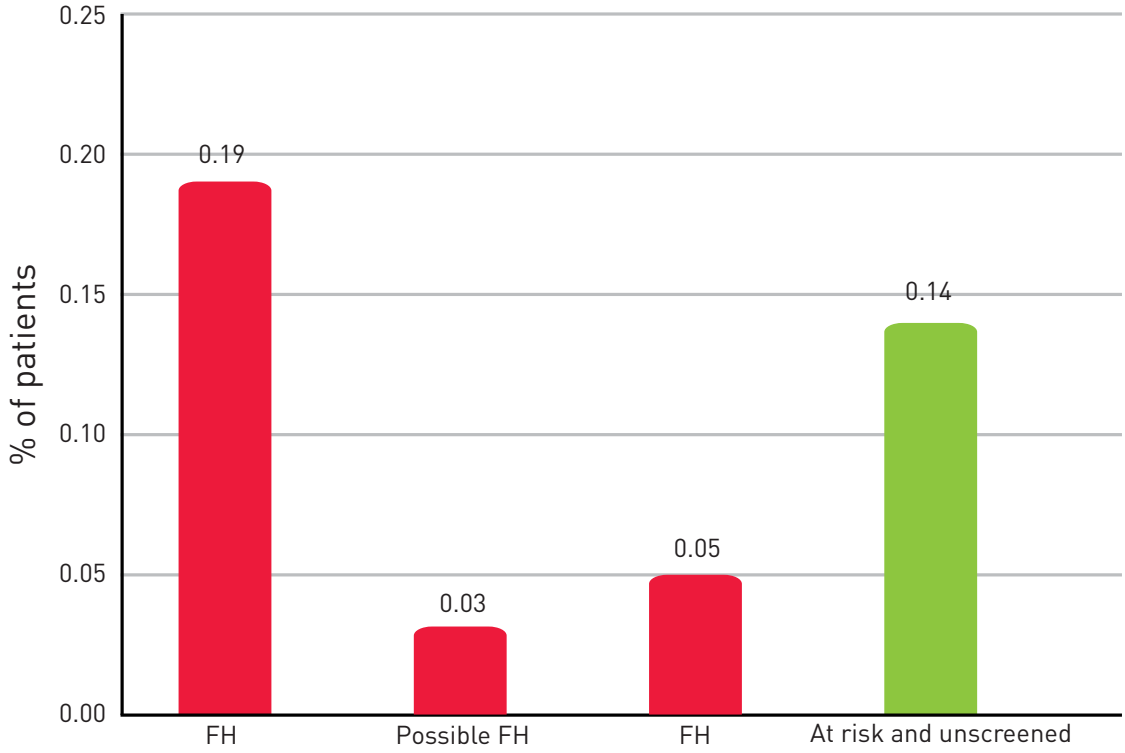
FH Nurse Advisor Programme results

A total of 53 Medway practices were able to take part; 47 practices participated in the FH Nurse Advisor Programme representing 89% of Medway GP practices able to take part. The Nurse Advisor Programme conducted 116 audit reviews and reviewed 1505 patients, of which 210 patients were invited for clinic visits, and 109 (52%) attended.

FH prevalence: From initiation of the FH Nurse Advisor Programme in October 2013 until programme end in July 2014 (9 months), the prevalence of patients diagnosed with FH within Medway CCG increased to 0.28% (one in 357) (**Figure 5; Table 9**).

'At risk and unscreened': Following the programme, this had reduced to 0.14% (**Figure 5; Table 9**).

Figure 5: Medway FH audit results at close of Nurse Advisor Programme (figures at 30 July 2014)*



* FH diagnoses made by Simon Broome criteria and/or DLCN score; Patients were considered to be 'at risk and unscreened' if they had a total cholesterol >7.5 mmol/L and/or LDL-C >4.9 mmol/L and had not been assessed using the Simon Broome criteria.

Table 9: Medway FH Audit Tool results after end of FH Nurse Advisor Programme (figures at 30 July 2014)*

	NUMBER	POPULATION	PREVALENCE (%)
FH*	546	281,655	0.19
Possible FH*	147	281,655	0.05
Probable FH*	83	281,655	0.03
Total FH	776	281,655	0.28
At risk and unscreened	398	281,655	0.14

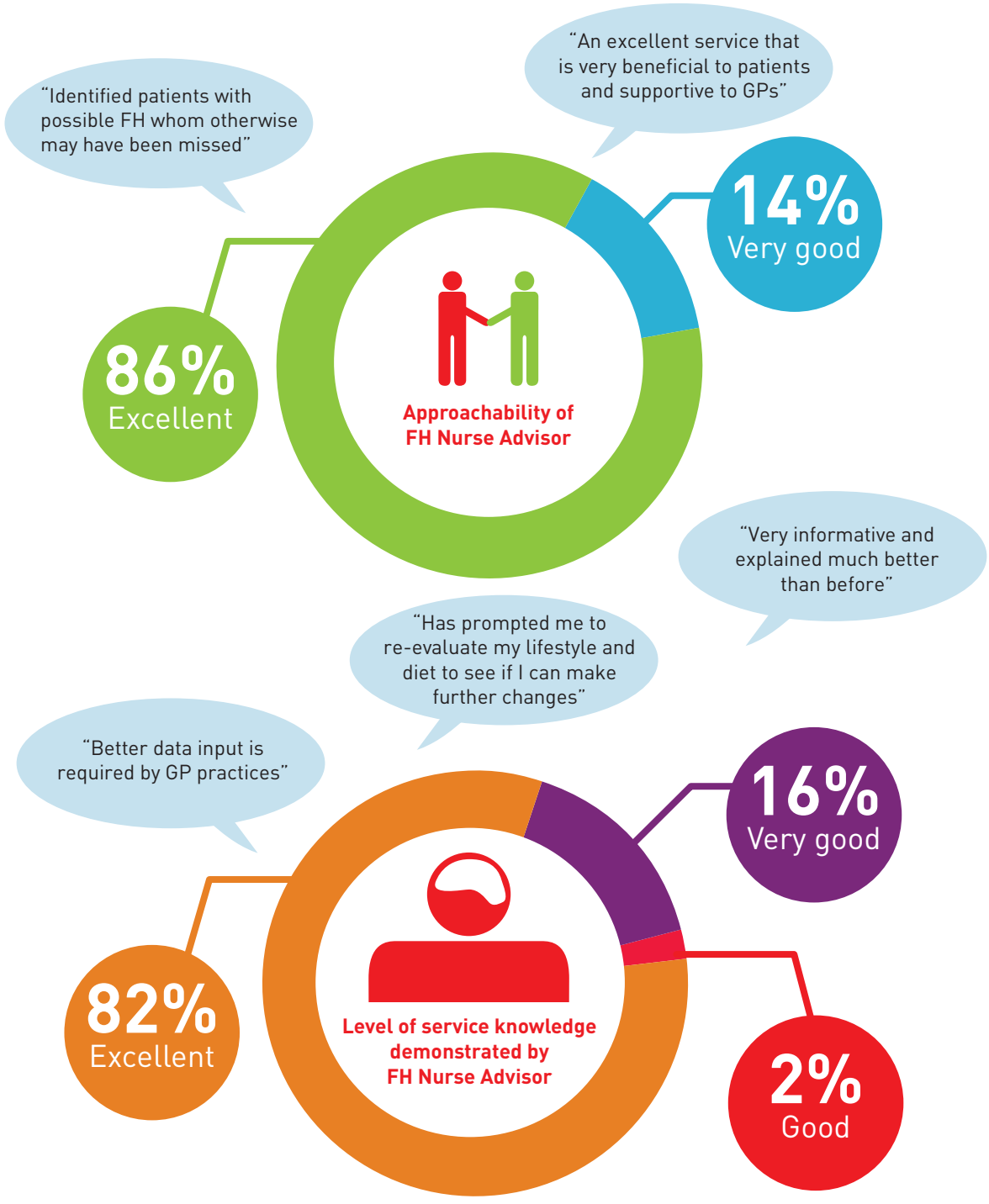
*FH diagnoses made by Simon Broome criteria and/or DLCN score

Patient feedback

Of the 109 patients seen in the FH Nurse Advisor clinic, 64 (59%) responded to the patient feedback questionnaire (**Appendix B5**). All of the respondents felt that the approachability of the FH Nurse Advisor was either excellent (86%) or very good (14%) (**Figure 6**). This was mirrored in the patients' views of the level of service knowledge demonstrated by the FH Nurse Advisor, with 98% responding as either excellent or very good and the remaining 2% as good (**Figure 6**).

97% (62/64) of patients felt that the FH patient review service met their expectations and only 5% (3/64) felt that it could be improved in some way.

Figure 6: Patient feedback from FH Nurse Advisor Programme



Recommended improvements to FH Nurse Advisor Programme

Increasing the number of patients invited to attend the clinic

To aid accurate diagnosis, future programmes could consider clinic assessment for all patients identified as 'at risk and unscreened'. At the clinic, missing information could be obtained and secondary comorbidities identified to allow an accurate assessment and appropriate referral onto specialist treatment (e.g. lipid specialist or diabetes care). This would increase the workload of the FH Nurse Advisor; however, the proposal is not unmanageable, with an estimated 60 patients flagged for a clinic review in a practice of 10,000 patients (based on the proportion of patients unscreened at the beginning of the FH Nurse Advisor Programme), which would equate to 10 FH clinics with six patients seen per clinic.

Improving communication, education and leaving a sustainable legacy

Communication between the FH Nurse Advisor and the GP was often via the Practice Manager, with the potential for information to be misinterpreted. Direct contact between the GP and FH Nurse Advisor would be ideal, but this would add pressure to a GP's already busy schedule. To improve future programmes, a Practice Nurse could be assigned as the practice lead and primary contact for the FH Nurse Advisor. The Practice Nurse would attend the patient clinics, providing an opportunity for training and education, and would allow the Practice Nurse to continue assessing, diagnosing and appropriately managing FH within the practice after the FH Nurse Advisor Programme ended. Based on the audit data after the FH Nurse Advisor Programme, around 0.14% of patients would require evaluation and 0.28% would require management. In real terms these numbers are small: in a practice of 10,000 patients, 15 patients would be 'at risk and unscreened', with around 30 patients diagnosed with FH requiring management.

Improving clinic attendance

The number of patients attending the clinic was low (52%). Future programmes need to encourage greater patient participation, and this could be achieved by:

- Revising the current patient invitation letter to include a more detailed explanation of the programme and the familial aspect of high cholesterol;
- Providing contact details for either the FH Nurse Advisor or the practice lead to allow patients to ask questions prior to or after the clinic;
- Involving a local FH Patient Ambassador as a peer-to-peer voice to endorse the importance of the programme;
- Running evening clinics to make it easier for those patients who work to attend;
- Adding a prompt to the Audit Tool to flag to GPs patients who did not attend a clinic and allow an 'opportunistic' follow-up when the patient next visited the practice;
- Providing the option of completing a self-assessment template at home in paper format returnable via a stamped addressed envelope or accessible online via a webpage.

Improving practice participation

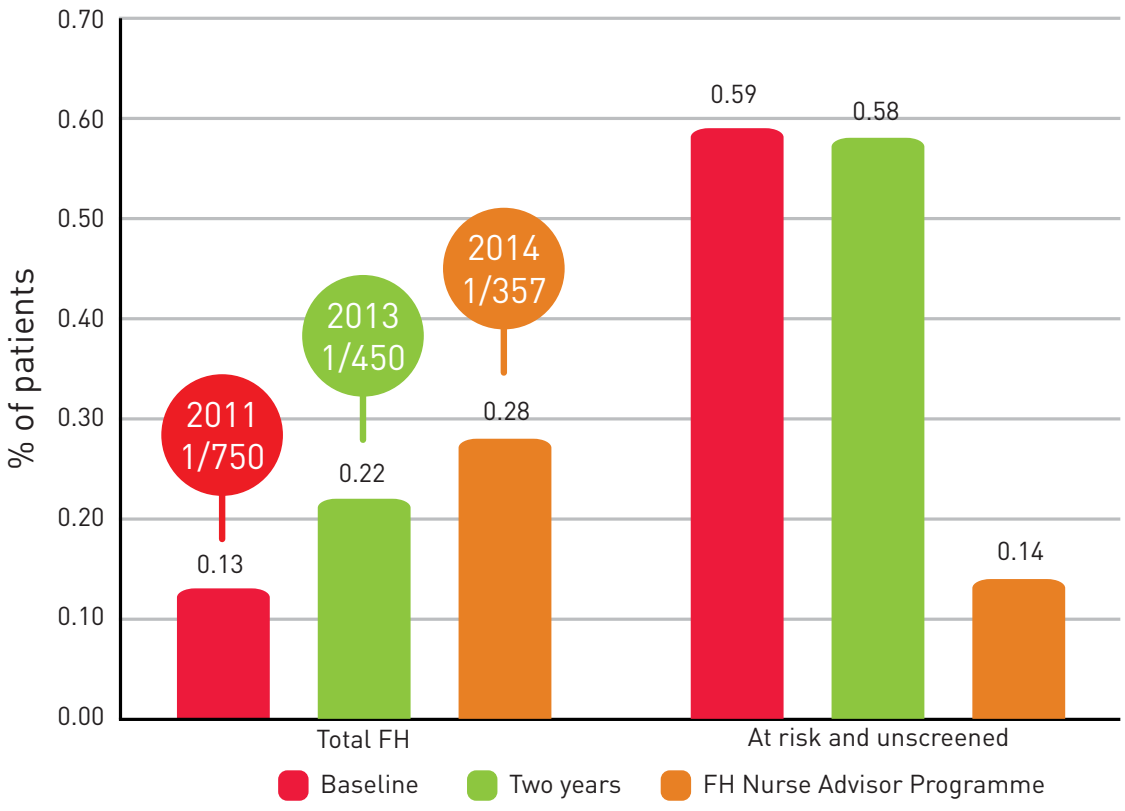
Identification of a GP and nurse lead within each practice was essential to ensure practice buy-in and assist with coordination of the programme. Practice participation was relatively high at 89%, and of the non-participating practices, most were small, with time and resource constraints. Improving practice participation in future programmes could be achieved by additional support from the CCG during the set-up of the programme; a key opinion leader within the CCG and/or a patient champion could advocate the importance of FH diagnosis and the CCG could also incentivise involvement for all practices.

IMPROVING FH PREVALENCE, DECREASING THE NUMBERS AT RISK AND UNSCREENED

FH is a common disorder that remains under-diagnosed and untreated.[2] Recent NICE Quality Standard guidance[13], EAS guidelines[2] and guidance from the International FH Foundation[16] has recognised this as a significant issue to be addressed. In addition, the recent Department of Health Cardiovascular Outcomes Strategy recognised improving identification of inherited cardiac conditions, and FH in particular, as a strategic priority and action.[27]

The Medway Audit model provides a solution to the challenge of improving diagnosis of FH within primary care. The Audit + software and Medway FH Audit Tool and prompts running in the background on GP IT systems improved diagnosis of FH, but the number of patients 'at risk and unscreened' remained the same. The FH Nurse Advisor Programme not only increased the number of FH diagnoses, it also reduced the number of patients 'at risk and unscreened' by almost three-quarters (**Figure 7**). The Medway CCG model could be adopted by other CCGs within England to improve diagnosis, awareness and management of FH in primary care.

Figure 7: Summary of the Medway FH audit results at baseline, after 2 years and after the introduction of the FH Nurse Advisor Programme



EATING THE FH ELEPHANT

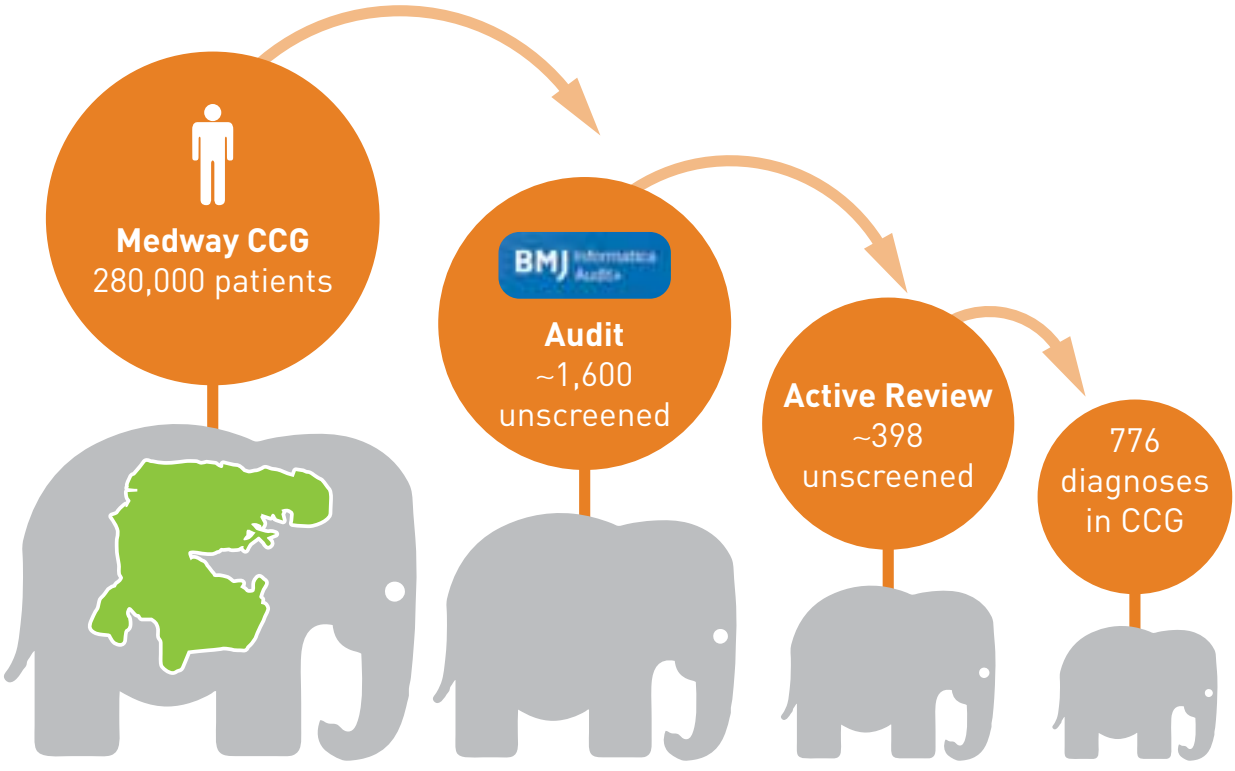
Many GPs and CCGs consider reviewing their patient population to identify FH a mammoth and daunting task. The Medway FH Audit Tool utilises information contained within a GPs IT system and produces a list of patients 'at risk and unscreened' for each GP practice. The Medway FH audit data suggest that in a large practice of more than 10,000 patients, initially around 60 patients would be flagged for review by the GP practice, either at the patient's next visit or by systematic assessment such as that conducted in the FH Nurse Advisor Programme. Once diagnosed, based on the audit numbers, a large practice of 10,000 patients would be managing approximately 30 patients and smaller practices with fewer than 2000 patients would have around five patients to manage.

Within the Medway CCG, the Medway FH Audit Tool reduced the numbers to screen from 280,000 (the number of patients within the Medway CCG) to approximately 1600 patients at risk and unscreened. The Audit Tool and prompts in conjunction with the FH Nurse Advisor Programme led to the diagnosis of 776 patients with FH (**Figure 8**).

Importantly, improving diagnosis and treatment of FH helps CCGs and other agencies fulfil their role in the delivery of quality healthcare in accordance with Government policy and clinical guidance. Relevant framework and guidelines include:

- The NHS Outcomes Framework (with impact on Domain 1 – preventing people dying prematurely; Domain 2 – enhancing quality of life for people with long-term conditions; and Domain 4 – ensuring people have a positive experience of care); [24]
- The NICE FH guidelines (CG71);[4]
- The NICE Quality Standard on FH (QS41);[13]
- The Cardiovascular Disease Outcomes Strategy (2003), with its aspiration to find and treat at least 50% of cases of FH in England.[27]

Figure 8: Reducing the FH "elephant" to a manageable task



ECONOMIC ADVANTAGES OF DIAGNOSING FH

In 2012, HEART UK published *Saving lives, saving families*.[21] The report includes economic modelling that demonstrates the health and cost savings that can be made through improved identification and treatment of FH using methodology recommended in NICE guidelines (CG71).[4]

Key findings of the research:

- High-intensity treatment, compared with low-intensity or no treatment, results in greater reductions in LDL-C and major cardiovascular events, which translates into more quality-adjusted life-years and life-years gained.
- High-intensity treatment will mean 101 cardiovascular deaths are avoided per 1000 FH patients (aged 30–85 years) when compared with no treatment.
- If 50% of patients with FH are diagnosed and treated optimally over a 55-year period, £94.7 million (£1.97 million per 1000 cases) can be saved by the NHS (through reduced cardiovascular events), or £1.7 million per year.
- By not implementing cascade testing as recommended by NICE (identifying 50% of potential relatives cases), the NHS is losing £1.4 million per year.

A recent paper by Pears and colleagues[28] examined three alternative models of care for FH: specialist led, primary care led, and a dual care model in which primary care manages the majority of patients in the cascade testing pathway. The authors concluded that costs for all three models are now less than 50% of the cost of the original estimates undertaken by NICE. By using the latest statin costs, reducing the proportion of patients prescribed more expensive proprietary owned rosuvastatin and managing more patients with FH in primary care, providing an FH service is now much more affordable than predicted by NICE in 2008.

Pears and colleagues assessed their models in a population of 1.95 million, estimating the dual care model to cost £1.89 million over 10 years. If we extrapolate the figure for the Medway population (280,000), the dual care model will cost approximately £271,551 over 10 years. Such a programme would include the cost of medicines, management in primary care, referral for specialist attention and genetic testing, and assessment of family members.

Ultimately, this would generate savings for the NHS, by reducing the number of cardiovascular events.

NEXT STEPS AND SUSTAINABILITY

Rolling out to other GP practices and CCGs

The Medway FH audit programme provides a transferrable model that can be used to improve the detection of FH in primary care. The Audit + software and Medway tool and prompts can be readily integrated into other GP practices and implemented within other CCGs. The FH Nurse Advisor Programme provides a useful model to increase diagnosis and appropriate referral of patients with FH within primary care.

HEART UK is currently in discussions with BMJ Informatica and relevant agencies (eg, CCGs) to make the Audit Tool and prompts widely available to GPs. In addition, HEART UK is seeking support to extend the FH Nurse Advisor Programme beyond the Medway region.

GP information packs

To continue to support GP practices within the Medway CCG, GP practices have been provided with a primary care guidance pack from HEART UK (**Appendix C)**. The packs contain information about FH, including links to the HEART UK FH toolkit,[29] HEART UK patient information leaflets[26] and a series of publications sponsored by HEART UK and published in the Primary Care Cardiovascular Journal.[30]

Referral to secondary care

NICE guidance recommends that HCPs should offer all people with a diagnosis of FH referral to a specialist with expertise in FH for confirmation of clinical diagnosis with DNA testing and initiation of cascade testing of relatives in those patients with a confirmed molecular diagnosis.[4, 13] There was concern at the outset that increasing the diagnosis of FH could stretch secondary care resources. Within the Medway FH Nurse Advisor Programme, referral to local lipid clinics was advised for all newly diagnosed cases of FH. However, in many cases, the patient had already been referred and managed in secondary care. Re-referral was advised if the patients' cholesterol levels were not optimised or if their cholesterol levels had risen after being transferred back to primary care.

The Medway Audit Steering Committee are currently developing FH referral criteria based on the DLCN score, with consideration of the following: number of living relatives a patient has, whether any relatives with FH require specialist interventions, failure of response to primary care treatment, and willingness to accept more intensive treatment.

Genetic testing

NICE guidance recommends genetic testing of all index cases and cascade testing of family members as a cost-effective method for identifying new cases of FH.[4] Genetic testing for FH is not yet routine in England and was not included as part of this audit. However, with the new NHS commissioning structure and its commitment to increased investment into genetic sequencing resources, genetic testing of all FH cases is certainly feasible.[11, 31] When available, genetic testing will allow mutation carriers to be distinguished from those with polygenic FH and focus resources on cascade testing in the 40% of clinical FH patients with an identified single gene alteration.[11]

DIAGNOSING FH IN PRIMARY CARE

Whilst implementation of cascade testing of current index cases may increase FH diagnosis by 50%[21], a systematic strategy for detecting new index cases is essential to improve diagnosis of FH and prevent CHD. Importantly, each new index case is a trigger for cascade testing, whereby further cases can be efficiently discovered. Both methods need to be well integrated if all cases of FH are to be diagnosed.[16]

The Medway FH Audit Tool and the FH Nurse Advisor Programme provide a systematic approach for identifying index cases from data already available within GP systems. This model has not only improved diagnosis of FH, it has raised awareness of FH to both GPs and patients within the CCG. The Audit + software is an inexpensive tool that is amenable to most GP IT systems and can be used to increase the diagnosis of FH. The provision of an FH Nurse Advisor can improve diagnosis even further.

The FH audit and Nurse Advisor Programme have successfully doubled FH diagnosis in Medway CCG, but the increase in patient numbers is manageable. An important next step is the rollout of this system into other commissioning groups and to engage with secondary care practitioners to establish and support the implications of these programmes to their services.

HEART UK is encouraged by the success of this model and will advocate its rollout to out CCGs. In the long term, such programmes will improve diagnosis with the inevitable benefit of better management and treatment and ultimately preventing CHD and alleviating unnecessary anxiety in these patients.

An unexpected diagnosis of high cholesterol in a young mother

Katharine Kear was unexpectedly diagnosed with high cholesterol aged 25. Although she had a known family history of CHD, it took several years before she was diagnosed with FH. Even after diagnosis, her four children remained untested and at risk. After reading about the implications of high cholesterol via a newspaper article about FH, Katherine embarked on a mission to have her children tested.

"The Medway FH audit is a fantastic initiative; such programmes are essential to diagnose FH. Catching that initial patient does not just benefit them, their whole families can be tested and treated. My diagnosis and that of my children's was a long time coming; such a programme would have made a real difference to my family."

Katharine's cholesterol was first measured after she visited her GP with white lumps on her eyelids, which were diagnosed as xanthoma. Despite being a slim, fit and healthy non-smoker, her total cholesterol level was 9.8 mmol/L. She was immediately prescribed a statin and advised to see a nutritionist. Katherine had a significant family history of CHD, her mother had CHD and heart by-pass surgery in her 50s and her grandfather died in his early 50s following a heart attack. Katherine's GP suggested that her high cholesterol could be hereditary, but did not suggest FH. Her cholesterol was not monitored any further.

Although aware of a potential hereditary aspect to her high cholesterol, Katharine didn't consider the implications this could have for her children, until she read a newspaper article on FH. It was then that Katharine realised the impact that high cholesterol could have on the health of her and her family. Now registered with a different GP practice, she sought re-assessment of her cholesterol and was diagnosed with FH.

Recognising the implications for the health of her immediate family, Katharine wanted to establish if she had passed FH on to her four young children; however, she was told that her children were too young to be tested. She was determined to pursue testing, and with the support of her local MP, Katharine and her children were eventually referred for specialist assessment by a lipidologist. Her eldest son did not have raised cholesterol, but for her younger son and daughter (aged 11 and 8 years), a diagnosis of FH was made and treatment was commenced with statins. Her youngest son was too young to be tested. NICE guidance suggests that children are not tested for FH before the age of 2 years. Katherine's diagnosis prompted cascade testing of other family members and several were diagnosed with elevated cholesterol.

Initially, Katharine and her two children were reviewed annually by a specialist, but are now managed by their GP. Katharine has not yet had genetic testing for FH, but she is hoping that recent changes in funding will allow her FH mutation status to be confirmed, which will aid diagnosis of further family members. She is concerned about the long-term implications of FH on her children, particularly the risk to a grandchild, and the risk of homozygous FH if her children were to marry someone with FH.

Katherine believes that educating GPs about FH is vital and says *"It is a relatively easy disease to treat and manage and diagnosing FH saves lives. The work conducted by the Medway FH is a real step forward."*

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APPENDIX A: LIST OF MEDWAY GP PRACTICES PARTICIPATING IN AUDIT

Woodlands Family Practice
Dr Aslam T
Dr JS Birdi & Partners
The Kings Family Practice
Dr Tanday J S & Partners
Dr Silhi R B
Dr Markwick C P & Partner
Dr Jana P P
Dr Sastry M R & Partner
Dr Hubbard D C & Dr Redman J H
Dr Maheswaran S & Partner
Dr Dharan M & Partners
Dr Ramesh N
Dr Mir A R
St Mary's Medical Practice
Dr Green Ph & Partners
Dr Qureshi K N
Dr Patel P & Partners
Dr Raval J K K & Partners
Dr J N Ray & Partners
Dr Ferrin L V & Partners
Dr Elapatha N
Maidstone Road Surgery
Apex Medical Practice
Dr Lakshman J C & Partner
Sunlight Centre Surgery
Dr J Spinks & Partners
City Way Surgery
Dr Patel S K C & Partner
Dr KS Mahapatra & Partners
Dr Singh 0 S & Partner
Dr Y Karim & Partners
St Werburgh Practice
Dr WSB Fernando & Partner
Eastcourt Lane Surgery
Dr S Bhasme
Dr Chaudhry M A
Dr Ma El-Faramawi
Dr Bhatia S
Dr R Vibhuti & Partners
Dr Tandon S L
Dr An Stacey
Dr Jha A B
Malling Health Group
Church View Practice
Parkwood Health Centre
Dr Singh B N
Dr K Padma
Marlowe Park Medical Centre
Dr IM Ali
Dr Balachander C S
Dr SM Lawrence
Dr Selvan S T & Partner

APPENDIX B: FH NURSE ADVISOR PROGRAMME SUPPORT MATERIALS

B1: Patient invitation letter

[Practice headed paper]

Patient Name

Patient Address

Dear

This practice is committed to ensuring all of its patients are receiving the best possible health care. In order to achieve this we have decided to review patients who have a history or family history of raised cholesterol levels and/or premature coronary or vascular disease.

The review will be carried out by a Nurse employed by Ashfield Healthcare Limited on behalf of NHS Medway CCG.

The review will be held in a patient clinic and will include a review of family history and general lifestyle advice.

Since you are one of our patients selected for a review in clinic we have made an appointment for you to attend this clinic which will be held at the surgery on:

Date: ..... Time: .....

Please bring your current medication with you to the appointment.

You will be contacted by telephone 24 hours before your clinic appointment to confirm your attendance.

Should you be unable to attend, then please contact the surgery on: ***[insert telephone number]*** and one of the surgery staff will be happy to rearrange this appointment for you.

Yours sincerely,

Dr/Nurse [Insert name and address of GP/practice Nurse]

B2: Patient Service User Guide

Ashfield Clinical

The Familial Hypercholesterolaemia Patient Review Service

Patient User Guide

Q: What is the Service?

This service is designed to help general practices ensure that patients with Familial Hypercholesterolaemia (FH) receive treatment that reflects best practice according to the National Institute for Health and Clinical Excellence (NICE) guidelines. Familial Hypercholesterolaemia affects 1 in 500 of the population it is a specific genetic defect that causes high cholesterol levels in the blood.

Q: Who is providing the service?

This service is provided by an FH Nurse employed by Ashfield Healthcare Limited on behalf NHS Medway CCG and is sponsored by Sanofi Ltd.

Q: Who is this service for?

This service is for all patients who may be at risk from Familial Hypercholesterolaemia and their GP has requested this service to help screen for potentially susceptible patients.

Q: Can you tell me about the Nurse who will be taking the FH patient clinic?

The FH Nurse is a registered nurse dedicated to providing educational review and lifestyle advice. Each nurse receives specific training, competency assessment and validation which is supplemented by continuing education.

Q: What do I need to know?

1. How has this service developed?

Ashfield Healthcare Limited has developed this service in partnership with NHS Medway CCG, HEART UK and Sanofi Ltd. As our patients are at the centre of service delivery we have included valuable feedback from other patients in the planning and preparation of the FH Patient Review Service.

B2: Patient Service User Guide (continued)

2. What will happen with my confidential information?

Your confidential information will remain within the practice. The nurse will have no need to remove any confidential information from the practice.

If you have any questions regarding your confidential information please contact the Quality Department Manager at Ashfield Healthcare Limited at the address at the end of this leaflet.

3. Do I have to pay for this service?

There are no charges to you for the Familial Hypercholesterolaemia Patient Review Service.

4. If I am not happy about the FH Nurse or the service delivered by the nurse, what shall I do?

If you, a family member or carer are not happy with the FH Nurse or the service provided by Ashfield Healthcare Limited then please contact the Quality Department Manager in writing at this address:

Quality Department Manager

Nursing Services

Ashfield Healthcare Limited
Ashfield House
Resolution Road

Ashby-de-la-Zouch

Leicestershire

LE65 1HW

We will ensure that your comment or complaint is thoroughly investigated and will be acknowledged within 5 working days. We will keep a written record of your comment or complaint with all the details and aim to have the outcome of our investigation completed within 28 days of receipt of your letter.

If you remain dissatisfied with the service you may contact any one of the following as appropriate:

Care Quality Commission
National Correspondence
Citygate
Gallowgate
Newcastle upon Tyne
NE1 4PA

Nursing Midwifery Council (NMC)
23 Portland Place,
London W1B 1PZ
Tel: 0207333 9333

B2: Patient Service User Guide (continued)

The Scottish Commission for the Regulation of Care
Compass House,
11 Riverside Drive
Dundee
DD1 4NY

The Regulation and Improvement Authority Regulate Healthcare provision in Northern Ireland
9th Floor Riverside Tower
5, Layton Place,
Belfast
BT1 2BT

5. What happens if something goes wrong?

Insurance is in place in the unlikely event that anything goes wrong. Ashfield Healthcare Limited has professional indemnity insurance and medical malpractice insurance.

6. What hours does the service operate?

The Patient Review Service will usually operate between 8am and 5pm Monday to Friday, when the surgery is open.

If you would like any additional information please contact the Quality Department Manager at the address above.

Ashfield Healthcare Limited provide high quality healthcare services. The requirements of patients are at the forefront of everything we do.

Our commitment to quality includes the continual review and refinement of our systems. We regularly assess our processes and services and ensure we develop our staff to improve standards and outcomes for patients. The opinions of patients we work with are paramount in measuring the quality of the services we deliver.

The Directors of Ashfield Healthcare Limited have responsibility for achieving quality standards; the Quality Department has day to day responsibility for devising, implementing and monitoring quality standards in all of Ashfield Healthcare Limited' healthcare services. All of the Company's staff have a role to play in achieving the Company's quality objectives.

B3: Patient Consent Form

Ashfield Clinical

Familial Hypercholesterolaemia (FH) Patient Review Service Programme

Patient Consent Form

Patient Name: _____

Date of Birth: _____

Patient Care and Use of Information

The FH Patient Review Service that I will receive will be delivered to me by a Nurse Advisor working on behalf of this practice/healthcare setting. This Nurse Advisor is employed by Ashfield Healthcare Limited and sponsored by Sanofi Ltd.

This programme is being offered to a number of patients within GP Surgeries. As part of the programme and in order to improve patient care, the information collected from this programme may be used to help assess its effectiveness. The data collected will be totally anonymous and it will not be possible for individuals to be recognised from this information. No personal identifiable data of mine will be removed from the practice by the Ashfield Healthcare Limited Nurse Advisor.

The nature and the purpose of the programme have been explained by the Ashfield Healthcare Limited Nurse and I give my consent to participate in this programme and for my data to be anonymised and used for the purpose stated above. I understand that I am able to change my mind about participating in this service at any time.

Patient Name _____ **(Please print)**

Signed _____

Date _____

Nurse Advisor _____ **(Please print)**

Signed _____

Date _____

B4: Cascade letter to relatives from HCP and reply form

[Address]

Dear [Name]

My name is [insert name] and I work at [insert practice] I am writing to you because a member of your family has high cholesterol. The doctor thinks this is a special form of high cholesterol that can be passed down through families so we would like to test you. High cholesterol leads to a greater risk of heart disease but this risk can be lowered by following a healthy diet and exercising. There are also several medicines available to help lower cholesterol levels.

We are asking all the relatives of patients with this special form of high cholesterol to take part in family testing to find out whether other members of the family also have the same form of high cholesterol. You will be asked to have blood tests to check your cholesterol levels and to see if you have the special form of high cholesterol that your relative has.

Please could you telephone me on [insert telephone number] so that we can organise an appointment at a date and time to suit you. Please phone me if you have any questions or would like any further information, or you can return the reply slip and I will phone you.

We hope that you will take part in the testing of family members but if you decide not to you should ask your GP to arrange a cholesterol test. To do this, please take the enclosed letter to your GP.

Thank you very much for your help.

Yours sincerely

[insert signature]

Encl. Letter to GP & response form for GP to fill-in

B4: Cascade letter to relatives from HCP and reply form (continued)

FH Cascade Testing - Reply Form

Name: .....

Address:.....

Home phone: _____ Work phone: _____ Mobile: _____

I understand that someone in my family has high cholesterol and you would like to talk to me about having a cholesterol test.

I **would/would not** (delete as appropriate) like to know more about this

(Please tick one box)

☐ I would like to receive an appointment in the post.

☐ I would like to be telephoned by the FH Nurse/GP (insert name) to arrange a convenient appointment.

I would like to be contacted on my **home/work/mobile** (delete as appropriate) telephone number (as above).

I prefer to be telephoned in the **morning/afternoon** (delete as appropriate).

☐ I do not want to be contacted at this time and I would like to be contacted in 6 months.

☐ I do not want to be contacted now or in the future.

If you do not wish to be contacted it would help us very much if you could tell us why:

_____

_____

Please add any other information _____

_____

Signature **Date**

Please return to: [Insert name & Address of FH Nurse]

B5: FH Patient Review Service - Patient Evaluation

B5: FH Patient Review Service - Patient Evaluation

Ashfield Clinical

FH Patient Review Service - Patient Evaluation

Name of FH Nurse Advisor.....

1. Did this FH Patient Review Service meet with your expectations? Yes ☐ No ☐

Any comments? .....

.....

2. Is there anything that you feel could be improved about the service? Yes ☐ No ☐

If so, please state here what could be improved

.....

.....

3. Please rate the following:

	Excellent	Very Good	Good	Fair	Poor
The approachability of the FH Nurse Advisor who reviewed you in clinic.					
The level of education provided by the FH Nurse Advisor					

4. Please state below any lifestyle changes that you will adopt as a result of receiving this service:

.....

.....

.....

Thank you for completing this evaluation

APPENDIX C: HEART UK FH RESOURCES FOR PRIMARY CARE

NHS Medway Clinical Commissioning Group

HEART UK

Medway familial hypercholesterolaemia audit project:

Resources for primary care

September 2014

Background

Increasing detection of patients with familial hypercholesterolaemia (FH) can improve the prevention of cardiovascular events and identification of additional 'at risk' relatives for screening and evaluation. Medway Clinical Commissioning Group partnered with HEART UK and Sanofi to deliver its innovative FH primary care audit project. Following the development and widespread use of an audit prompt in general practice, a nurse was employed to help identify cases of FH, assess patients and provide support and advice for their care.

GPs are encouraged to continue to use the FH audit prompt, review identified patients, and further assess first and second degree relatives registered at their practice.

These resources aim to provide primary care clinicians with tools that can useful for healthcare professionals and patients alike.

The interim project report, *Systematically identifying familial hypercholesterolaemia in primary care*, shows promising results. The full report will be published in October 2014, but the interim report can be viewed at:

http://heartuk.org.uk/files/uploads/HEART_UK_FH_Audit_project_interim_report_-_July_2014.pdf

Audit information

For detailed information on the audit, please see the separate PDF – Medway CCG FH audit.

Guidelines and service information

HEART UK FH toolkit

Comprehensive information for clinicians, commissioners and patients to help improve diagnosis and treatment of FH.

http://heartuk.org.uk/FHToolkit/

NICE Guideline – Identification and management of familial hypercholesterolaemia (CG71)

http://www.nice.org.uk/guidance/CG071

NICE FH Quality Standard (QS41)

http://www.nice.org.uk/guidance/qs41/chapter/about-this-quality-standard

FH diagnostic criteria

The diagnosis of FH relies of five criteria: family history, clinical history of premature CHD, physical examination for xanthomas and corneal arcus, very high LDL-C on repeat measurements, and/or a causative mutation detected by molecular genetics. Decisions about genetic testing for FH are largely made by secondary care specialists.

Clinical diagnostic tools for FH are well defined, but there is no one internationally-agreed algorithm. In the UK, the Simon Broome criteria is recommended to evaluate patients with raised LDL-C, especially if there is a personal or family history of premature CHD. See Simon Broome criteria at **Appendix 1.**

In Europe, the Dutch Lipid Clinic Network Diagnostic Criteria (DLCNC) is widely used and calculates a numerical score predicting the probability of diagnosing FH (**Table 1**). This criteria is increasingly accepted as simple and comprehensive.[1] The DLCNDC categorises patients as definite, probable or possible FH. In Medway, the Dutch criteria is used to establish the patient's 'score' to indicate the likelihood of FH.

Table 1: Dutch Lipid Clinic Network Diagnostic Criteria ('score') for familial hypercholesterolemia[2]

GROUP	SCORE
GROUP 1: FAMILY HISTORY	
First-degree relative with known premature coronary and/or vascular disease (men <55 years, women <60 years) OR First-degree relative with known LDL-cholesterol above the 95th percentile for age and sex	1
First-degree relative with tendinous xanthomata and/or arcus cornealis OR Children aged less than 18 years with LDL-cholesterol above the 95th percentile for age and sex	2
GROUP 2: CLINICAL HISTORY	
Patient with premature coronary artery disease (ages as above)	2
Patient with premature cerebral or peripheral vascular disease (as above)	1
GROUP 3: PHYSICAL EXAMINATION	
Tendinous xanthomata	6
Arcus cornealis prior to age 45 years	4
GROUP 4: LDL-C (mmol/L)	
LDL-C ≥8.5 8	8
LDL-C 6.5–8.4	5
5 LDL-C 5.0–6.4 3	3
LDL-C 4.0–4.9	1
GROUP 5: DNA ANALYSIS	
Functional mutation in the *LDLR*, *APOB* or *PCSK9* gene	8
SCORE	
>8	Definite FH
6-8	Probable FH
3-5	Possible FH
0-2	Unlikely FH

[1] Watts GF, Gidding S, Wierzbicki AS. Integrated guidance on the care of familial hypercholesterolaemia from the International FH Foundation. *Int. J Card.* 2014; **171**:309–325.

[2] Marks D, Thorogood M, Neil HA, *et al*. A review on the diagnosis, natural history, and treatment of familial hypercholesterolaemia. *Atherosclerosis* 2003; **168**:1–14.

Also see HEART UK advice sheet - Diagnostic criteria for FH using Simon Broome register

http://heartuk.org.uk/files/uploads/documents/HUK_AS04_Diagnostic.pdf

Educational materials for GPs and nurses

HEART UK has written a series of articles for the Primary Care Cardiovascular Journal. They are free to download and carry CPD points for GPs.

Dr David Milne, *It's not just a high cholesterol level, it can be an indicator of genetic disorder*

http://www.pccj.eu/index.php?option=com_content&view=article&id=964:sponsored-fh-series-its-not-just-a-high-cholesterol-level-it-can-be-an-indicator-of-genetic-disorder&catid=938:expedited-publication&Itemid=285

Dr R Dermot G Neely, *The importance of early diagnosis: how to identify patients with FH for diagnosis and referral*

http://www.pccj.eu/index.php?option=com_content&view=article&id=1027:sponsored-fh-series-the-importance-of-early-diagnosis-how-to-identify-patients-with-fh-for-diagnosis-and-referral&catid=938:expedited-publication&Itemid=285

Prof Gilbert R Thompson and Dr Mary Seed, *The management of familial hypercholesterolaemia*

http://www.pccj.eu/index.php?option=com_content&view=article&id=1064:sponsored-fh-series-the-management-of-familial-hypercholesterolaemia&catid=938:expedited-publication&Itemid=285

Drs Atul Kalhan, Vinay Eligar and Alan Rees, *Why do we need new options for managing FH?*

http://www.pccj.eu/index.php?option=com_content&view=article&id=1072:sponsored-fh-series-why-do-we-need-new-options-for-managing-fh&catid=938:expedited-publication&Itemid=285

Information for patients

HEART UK factsheet – FH and FCH

http://heartuk.org.uk/files/uploads/documents/huk_fs_mfsC_inheritedhighcholest.pdf

Family and children's resources. This webpage features a short 4-minute film, A story of Hope, suitable for all family members. The page also links to an e-book, *Buddy's FH adventure*, **specially designed for children with FH aged 7 and above.**

http://heartuk.org.uk/FHchildrensresources

Inherited high cholesterol – familial hypercholesterolaemia. **Patient booklet produced by HEART UK and the British Heart Foundation**.

http://heartuk.org.uk/files/uploads/documents/HUK_InheritedHeartConditions_FH.pdf

Other clinical information and reports

FH paediatric register

https://audit.rcplondon.ac.uk/PaedFH/page.aspx?pc=homepage

Saving lives, saving families: The health, social and economic advantages of diagnosing and treating familial hypercholesterolaemia. T**his HEART UK report makes the case for improved diagnosis and treatment of FH.**

http://heartuk.org.uk/files/uploads/documents/HUK_SavingLivesSavingFamilies_FHreport_Feb2012.pdf

Appendix 1

The Simon Broome Register Criteria (Total Cholesterol and LDL-C levels either pre-treatment or highest on treatment)[3 4]

A diagnosis of *'definite FH'* is made based on total cholesterol >6.7 mmol/L or LDL-C >4.0 mmol/L in a child (<16 years) or total cholesterol >7.5 mmol/L or LDL-C >4.9 mmol/L in an adult plus the presence of tendon xanthomas in the patient, or a first-degree or second-degree relative. A diagnosis of *'possible FH'* is made if there are no tendon xanthomas but a family history of myocardial infarction (aged <50 years in a second-degree relative or <60 years in a first-degree relative) or a family history of raised total cholesterol (>7.5 mmol/L in an adult first- or second-degree relative or >6.7 mmol/L in child or sibling aged <16.

DEFINITE FAMILIAL HYPERCHOLESTEROLAEMIA	POSSIBLE FAMILIAL HYPERCHOLESTEROLAEMIA
Total cholesterol >6.7 mmol/l OR LDL-C >4.0 mmol/l in a child aged younger than 16 years OR total cholesterol >7.5 mmol/l or LDL-C >4.9 mmol/l in an adult	total cholesterol >6.7 mmol/l OR LDL-C >4.0 mmol/l in a child aged younger than 16 years OR total cholesterol >7.5 mmol/l or LDL-C >4.9 mmol/l in an adult
PLUS	**AND AT LEAST ONE OF THE FOLLOWING**
tendon xanthomas in patient, or in first-degree relative (parent, sibling or child), OR in second-degree relative (grandparent, uncle or aunt) OR DNA-based evidence of an *LDLR* mutation, familial defective *APOB-100*, or a *PCSK9* mutation.	A family history of myocardial infarction: <50 years of age in second-degree relative or <60 years of age in first-degree relative OR A family history of raised total cholesterol: >7.5 mmol/l in adult first- or second-degree relative or >6.7 mmol/l in child or sibling aged younger than 16 years.

NHS
Medway
Clinical Commissioning Group

Familial Hypercholesterolaemia - Medway – v 6.1

This audit is based on NICE CG 71 Familial Hypercholesterolaemia.

The reason for identifying patients with Familial Hypercholesterolaemia (FH) is to decrease their risk of premature cardiovascular disease. Familial hypercholesterolaemia covers a range of autosomal dominant genetic conditions which affect lipid metabolism causing high cholesterol. Diagnosis is either made by genetic testing or as 'possible familial hypercholesterolaemia' based on the Simon Broome criteria. The management of patients with 'possible Familial Hypercholesterolaemia' is the same as for those with proven Familial Hypercholesterolaemia. The presence of heterozygous Familial Hypercholesterolaemia is estimated to be 1 in 500. Direct relatives will have a 50 : 50 chance of inheriting the FH abnormalities so contact tracing and getting relatives of index cases checked is important.

This audit takes a pragmatic approach in identifying those patients without a diagnosis who have had a cholesterol of > 7.4mmol/l or a LDL cholesterol of > 4.9mmol/l and have not already been assessed against the Simon Broome Criteria. This is the target population for assessment and a list of these patients can be identified by practices. Assessment may require further blood tests, information about a patient's family history or examination for arcus or xanthoma. In those patients diagnosed, treatment should be initiated with a statin and they should be asked to inform relatives to be tested. In those patients who fail to respond to statins consideration should be made for referral to a lipidologist.

We are working with HEART UK, the cholesterol charity, to improve this audit as more evidence becomes available to further risk stratify patients in a primary care setting.

Sections within the FH Audit

The audit has 6 sections:

- Familial Hypercholesterolaemia
- High Cholesterol (excluding FH/Possible/Probable FH)
- Dutch Lipid Score (excluding FH/Possible/Probable FH)
- Simon Broom Assessment
- History of
- Family History of FH

Each section contains measures some measures have prompts attached to them. Not all measures have prompts.

The prompts are included as a reminder or suggestions of interventions the patient may need. Once the action has been completed that prompt will no longer show for the patient.

Prompts within the FH Audit

Trigger	Prompt
Patients with Familial Hypercholesterolaemia or possible FH whose family have not been informed (by letter)	**Have relatives been informed regarding Familial Hypercholesterolaemia**
Patients with FH, Possible FH or Probable FH whose latest cholesterol reading is more than 5	**Up Titrate statins or consider referral**

Patients whose latest cholesterol is 7.4 or above OR latest serum cholesterol is 4.9 who have had a Genotype test	**Diagnose Familial Hypercholesterolaemia**
Patients whose latest cholesterol is 7.4 or above OR latest serum cholesterol is 4.9 or more and they have not had a Simon Broome assessment. They have a family history of male/female relatives under 55 and 65 with CHD and/or Hypercholesterolaemia	**Consider possible Familial Hypercholesterolaemia**
Patients whose latest cholesterol is 7.4 or above OR latest serum cholesterol is 4.9 or more and they have not had a Simon Broome assessment. They have a family history of IHD or MI (No Age or whether 1st or 2nd degree)	**Ask patient if MI < 50y 2nd degree relative or MI < 60y 1st degree relative** • **If YES Consider Possible Familial Hypercholesterolaemia** • **If NO assess using Simon Broom criteria**

Note: Prompts contain further information along with relevant read codes, which can be added directly into the patient record from the prompt screen.

Measures within the FH Audit

Familial Hypercholesterolaemia

Measure	*Description*
FH01	**Familial Hypercholesterolaemia** Patients with Familial Hypercholesterolaemia diagnosis
FH02	**Possible Familial Hypercholesterolaemia** Patients with Possible Familial Hypercholesterolaemia diagnosis
FH13	**Probable Familial Hypercholesterolaemia** Patients with Probable Familial Hypercholesterolaemia diagnosis
FH03	**Patients with FH, Possible FH or Probable FH latest Cholesterol over 5** Patients with a diagnosis of FH, Possible FH or Probable FH, whose latest Cholesterol level is over 5
FH03a	**Patients with FH, Possible FH or Probable FH latest Cholesterol less than 4** Patients with a diagnosis of FH, Possible FH or Probable FH, whose latest Cholesterol level is less than 4
FH03b	**Patients with FH, Possible FH or Probable FH latest Cholesterol >=4 and <5** Patients with a diagnosis of FH, Possible FH or Probable FH, whose latest Cholesterol level is 4 or more and less than 5
FH03c	**Patients with FH, Possible FH or Probable FH latest Cholesterol >=5 and <6** Patients with a diagnosis of FH, Possible FH or Probable FH, whose latest Cholesterol level is 5 or more and less than 6
FH03d	**Patients with FH, Possible FH or Probable FH latest Cholesterol >=6 and <7** Patients with a diagnosis of FH, Possible FH or Probable FH, whose latest Cholesterol level is 6 or more and less than 7
FH03e	**Patients with FH, Possible FH or Probable FH latest Cholesterol >=7 and <8** Patients with a diagnosis of FH, Possible FH or Probable FH, whose latest Cholesterol level is 7 or more and less than 8

FH03f	**Patients with FH, Possible FH or Probable FH latest Cholesterol >=8 and <9** Patients with a diagnosis of FH, Possible FH or Probable FH, whose latest Cholesterol level is 8 or more and less than 9
FH03g	**Patients with FH, Possible FH or Probable FH latest Cholesterol >=9** Patients with a diagnosis of FH, Possible FH or Probable FH, whose latest Cholesterol level is more than 9
FH14	**Patients with Tendon Xanthoma** Patients with a diagnosis of FH, Possible FH or Probable FH, with a record of Tendon Xanthoma
FH15	**Patients with Arcus Juvenails** Patients with a diagnosis of FH, Possible FH or Probable FH, with a record of Arcus Juvenails

High Cholesterol (excluding FH/Possible/Probable FH)

This section EXCLUDES patients with a diagnosis of FH, Possible FH or Probable FH

Measure	*Description*
FH04	**Patients Cholesterol above 7.4** Patients whose latest cholesterol level is 7.4 or above
FH05	**LDL cholesterol above 4.9** Patient whose latest LDL cholesterol level is 4.9 or above
FH06	**Patients with Genotype** Patients whose latest cholesterol level is 7.4 or above OR latest LDL cholesterol level 4.9 or above and have had a Genotype test

Dutch Lipid Score (excluding FH/Possible/Probable FH)

This section EXCLUDES patients with a diagnosis of FH, Possible FH or Probable FH

Measure	*Description*
DLS00	**Number of patients with a Dutch Lipid Score** Total number of patients with a Dutch Lipid Score recorded
DLS01	**Dutch Lipid Score 8 or above** Patients with a Dutch Lipid score of 8 or above
DLS02	**Dutch Lipid Score between 6 and 8** Patients with a Dutch Lipid score between 6 and 7 (inclusive)
DLS03	**Dutch Lipid Score between 3 and 5** Patients with a Dutch Lipid score between 3 and 5 (inclusive)
DLS04	**Dutch Lipid Score between 0 and 2** Patients with a Dutch Lipid score of 2 or less

Simon Broom Assessment

Measure	*Description*
FH07	**Patients who have had a Simon Broom Assessment** All patients who have had a Simon Broom Assessment

FH08	**Cholesterol > 7.4 or LDL >4.9 without Simon Broom Assessment (excluding FH, Possible FH or probable FH)** Patients without a diagnosis of FH, Possible FH or Probable FH, whose latest Cholesterol level is more than 7.4 OR their latest LDL cholesterol level is more than 4.9 who have not been assessed with the Simon Broom Assessment

History of

Measure	*Description*
HO01	**Male patients under 55 years with IHD or PVD** Male patients under 55 years with a recording of Ischaemic Heart Disease or Peripheral Vascular Disease
HO01a	**Male patients under 55 years with IHD or PVD, without FH diagnosis** Male patients under 55 years with a recording of Ischaemic Heart Disease or Peripheral Vascular Disease, but do not have a diagnosis of FH
HO01b	**Male patients under 55 years with IHD or PVD, without FH who have not been assessed using the Simon Broom criteria** Male patients under 55 years with a recording of Ischaemic Heart Disease or Peripheral Vascular Disease and no diagnosis of FH, who have not been assessed using the Simon Broom criteria
HO02	**Female patients under 60 with IHD or PVD** Female patients under 60 years with a recording of Ischaemic Heart Disease or Peripheral Vascular Disease
HO02a	**Female patients under 60 with IHD or PVD, without FH diagnosis** Female patients under 60 years with a recording of Ischaemic Heart Disease or Peripheral Vascular Disease, but do not have a diagnosis of FH
HO02b	**Female patients under 60 with IHD or PVD, without FH, who have not been assessed using the Simon Broom criteria** Female patients under 60 years with a recording of Ischaemic Heart Disease or Peripheral Vascular Disease an no diagnosis of FH, who have not been assessed using the Simon Broom criteria

Family History of FH

This section is based on patients, who do not have a diagnosis of FH, Possible FH or Probable FH, whose latest cholesterol is >7.4 or latest LDL cholesterol is >4.9 and have not been assessed with the Simon Broom assessment.

Measure	*Description*
FH09	**Patients with a family history of 1st degree relatives (male under 55 or female under 65) of CHD and Hypercholesterolaemia** Patients who have a family history of male relative under 55 or female relative under 65 with CHD and/or Hypercholesteroleamia
FH10	**Patients with a family history of IHD or MI age unknown or whether 1st or 2nd degree relative** Patients who have a family history of IHD or MI (no age or whether 1st or 2nd degree relative