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Grŵp Cydlynu Rhwydwaith y Galon
Cardiac Networks Co-ordinating Group

National Clinical Audit of the Management of Familial Hypercholesterolaemia 2009: Pilot

FULL REPORT
June 2009

Funded by:
Department of Health, England
Cardiac Network Co-ordinating Group, Wales

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To reference this work:

Young KV, Humphries SE, Grant R. National Clinical Audit of the Management of Familial Hypercholesterolaemia 2009: Pilot - Full Report. Clinical Effectiveness and Evaluation Unit, Royal College of Physicians, June 2009

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ISBN 978-1-86016-364-7

Acknowledgements

The time and effort given by all of the people who inputted into the development of the audit tools and for the NHS staff who undertook the pilot audit is acknowledged.

In particular the following are thanked:

- members of the Working Group and Steering Group (see page 4)
- those who inputted at pre-pilot stage:
 - Dr Mark Vella - William Harvey Hospital, East Kent University NHS Trust
 - Bridget Galaskar - Royal Salford NHS Foundation Trust
 - Professor Paul Durrington, Sister Ruth Eatough and Dr Handrean Soran - Manchester Royal Infirmary, Central Manchester and Manchester Children's University Hospital NHS Trust
- Netsolving who undertook the development of the audit webtool
- the sites who completed the pilot audit:
 - Dr Charles van Heyningen & Claire Ashton, University Hospital Aintree, Aintree University Hospitals NHS Foundation Trust
 - Dr Chris Hendriksz & Dr Anupam Chakrapani, Birmingham Children's Hospital, Birmingham Children's Hospital NHS Foundation Trust
 - Dr Handrean Soran & Sister Ruth Eatough, Manchester Royal Infirmary, Central Manchester and Manchester Children's University Hospital NHS Trust
 - Dr David Cassidy & Dr Dev Datta, Prince Charles Hospital, Cwm Taf NHS Trust
 - Dr Andar Gunnberg & Delyth Townsend, Morriston Hospital, Abertawe Bro Morgannwg University NHS Trust
 - Jane Jones, Emma Neves & Dr M Barbir, Royal Brompton Hospital, Royal Brompton & Harefield NHS Trust
 - Dr John Reckless & Dr Andrew Taylor, Royal United Hospital, Royal United Hospitals Bath
 - Dr Alan Taylor, Furness General Hospital, University Hospitals of Morecombe Bay Hospitals NHS Trust
 - Clare Holtby & Michael Addley University Hospital Lewisham, The Lewisham Hospital NHS Trust
 - Dr Nigel Capps, The Shrewsbury and Telford Hospital NHS Trust, The Shrewsbury and Telford NHS Trust
 - Dr Emmanuel Abu & Dr Paul Cook, Southampton General Hospital, Southampton University Hospitals NHS Trust
 - Dr Andrew Iversen & Dr Katherine Harding, Royal Sussex County Hospital, Brighton and Sussex University Hospitals NHS Trust
 - Adie Viljoen & Ann Ainsworth, East & North Hertfordshire NHS Trust, East & North Hertfordshire NHS Trust
 - Dr Alistair Munro, Worcestershire Royal Hospital, Worcestershire Acute Hospitals NHS Trust
- all those who supported us through giving advice, ideas and other support.

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* Resigned after 2nd Steering Group due to change of role

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Pilot Audit Report - June 2009

Report of Pilot audit project: Clinical Audit of Management of Familial Hypercholesterolaemia 2009

CONTENTS

| | Page |
|---|------|
| Acknowledgements | 3 |
| Steering Group | 4 |
| Useful addresses | 6 |
| Glossary | 7 |
| Foreward | 9 |
| EXECUTIVE SUMMARY | 10 |
| Introduction | 12 |
| Methods | 13 |
| Pilot Phase Evaluation | 17 |
| Results | 19 |
| Part 1: Organisational Audit | 20 |
| Section 1: Set Up | 20 |
| Section 2: Clinical Time | 23 |
| Section 3: Management of FH | 24 |
| Section 4: Cascade Testing | 25 |
| Section 5: Trust Services | 26 |
| Section 6: Patient Information | 27 |
| Section 7: Service Improvement | 28 |
| Part 2: Clinical Audit | 29 |
| Section 1: Diagnosis | 32 |
| Section 2: Pedigree | 36 |
| Section 3: Assessment | 39 |
| Section 4: Treatment | 41 |
| Section 5: Advice | 44 |
| Section 6: Cascade Testing | 45 |
| Section 7: (Annual) Review | 48 |
| Section 8: Children | 50 |
| Appendices | |
| 1 Audit proformas: organisational audit; clinical audit | 51 |
| 2 Inter-rater reliability analysis | 72 |
| 3 Participants | 75 |
| 4 Pilot Phase Evaluation - Survey of Pilot Sites | 76 |

Useful addresses / information

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www.bhf.org.uk
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Heart UK 0845 450 5988 Helpline hours: Tues & Thurs 10am to 4pm
- National Institute for Health and Clinical Excellence
www.nice.org.uk
- Royal College of Paediatrics and Child Health
www.rcpch.ac.uk

UK WHO Growth Charts
www.rcpch.ac.uk/Research/UK-WHO-Growth-Charts

Glossary (Adapted from NICE CG71)

| | |
|--|---|
| Adults with FH | For the purposes of this guideline, ‘adults’ includes all persons with familial hypercholesterolaemia (FH; heterozygous or homozygous) who are 16 years and older. |
| Cascade testing | Cascade testing is a mechanism for identifying people at risk of a genetic condition by a process of family tracing. For FH the test employed is measurement of low-density lipoprotein cholesterol (LDL-C) in the blood, and/or a DNA test if a disease-causing mutation has been identified in the index individual/proband (see below). |
| Children/young people | For the purposes of the NICE guideline and the audit tool, ‘children’ refers to persons younger than 10 years; ‘young people’ refers to persons from 10 years of age up to the age of 16 years. |
| Child-focused setting | Child-focused refers to valuing the child’s view and validating their voice in making decisions impacting their lives. A child-focused facility or space is one designed from the viewpoint of the service recipients. |
| Family history | The structure and relationships within the family that relates information about diseases in family members. |
| First-degree relative | A person’s biological parents, full brothers and sisters, and children. |
| Heterozygous FH | High LDL-C concentration in the blood caused by an inherited mutation from one parent only. People with FH are at increased risk of cardiovascular disease. |
| High-intensity statin | Statins are classified as high intensity if they produce greater LDL-C reductions than simvastatin 40 mg (for example, simvastatin 80 mg and appropriate doses of atorvastatin and rosuvastatin). |
| Homozygous FH | <p>Very high LDL-C concentration in the blood caused by an inherited mutation from both parents. When a person inherits exactly the same affected gene from both parents this is called truly ‘homozygous’ FH. When the mutations in the LDL receptor gene (or equivalent) are different, this state is called ‘compound heterozygous’. In general, the overall effect in both states is similar, in that LDL-C concentrations are very high. Both groups of patients have the same clinical pattern and high risk of cardiovascular disease.</p> <p>For clinical purposes, both homozygous FH and compound heterozygous FH can be regarded as behaving in a similar manner. Therefore, for the purposes of this audit the term ‘homozygous FH’ is used to also encompass compound heterozygous FH.</p> |
| Index individual (synonymous with ‘proband’) | The original patient who is the starting point for follow-up of other members of a family when investigating for possible causative genetic factors of the presenting condition. |
| Interquartile range (IQR) | The middle half of data, chosen so that one quarter of patients / sites are below it and one quarter above it. It provides an indication of how diverse clinical practice or case mix is. |
| Lipid measurements/concentrations/levels | These terms refer to the measurement of total cholesterol (TC), triglycerides (TGs), high-density lipoprotein cholesterol (HDL-C), and LDL-C. LDL-C is not usually measured directly but calculated from the TC, TGs and HDL-C, most accurately by using a fasting sample. Such tests are usually done in a clinical biochemistry laboratory. |
| Median | When data are arranged from lowest value to highest, the median is in the middle. It is an average which is not unduly influenced by very high or very low values which may include some element of error. |
| Pedigree | A method of characterising the relatives of an index individual/case and their family relationship as well as problems or illnesses within the family. This information, often represented graphically as a family tree, facilitates analysis of inheritance patterns. Study of a trait or disease begins with the affected person (the index individual). The |

| | |
|--------------------------------------|--|
| | pedigree is drawn as the relatives are described. One begins with the siblings of the index individual and proceeds to the parents; relatives of the parents, including brothers, sisters, nephews, nieces, grandparents, and so on. At least three generations are usually included. Illnesses, hospitalisations, causes of death, miscarriages, abortions, congenital anomalies, and any other unusual features are recorded. |
| Premature coronary heart disease | For the purpose of this guideline, this refers to a coronary event that has occurred (1) before 55 years of age in a male index individual or 65 years of age in a female index individual, (2) before 60 years of age in a first-degree relative, or (3) before 50 years of age in a second-degree relative. |
| Proband | The affected (index) individual through whom a family with a genetic disorder is ascertained. |
| Second-degree relative | A person's biological grandparent, grandchild, uncle, aunt, niece, nephew, half sister or half brother. |
| Simon Broome register | A computerised research register of people with FH, based in Oxford. Research from this voluntary register has led to several publications describing the natural history of FH in the UK. The 'Simon Broome criteria' for diagnosis were based on a study of this group of people with FH. |
| Specialist | One who has expertise in a particular field of medicine by virtue of additional training and experience. For this audit, we use specialist to refer to a healthcare professional with an expertise in FH. |
| Specialist centre | The definition of a specialist centre is not rigid and is based on a combination of patient treatment services, numbers and ages of people attending there, the presence of a multi-disciplinary team (which may include, for example, physicians, lipidologists, specialist nurses and dietitians), the ability to manage the more unusual manifestations of the condition and the additional functions such as research, education and standard setting. Care is supervised by expert healthcare professionals but shared with local hospitals and primary care teams. Although details of the model may vary between patients and areas, the key is that specialist supervision oversees local provision with the patient seen at diagnosis for initial assessment and then at least annually for review. |
| Specialist services for young people | Specialist services for young people provide an area of care that is family focussed. |
| Tendon xanthomata | A clinically detectable nodularity and/or thickening of the tendons caused by infiltration with lipid-laden histiocytes (macrophages in connective tissue). A distinctive feature of FH that most frequently affects the Achilles tendons but can also involve tendons on the back of the hands, elbows and knees. |
| Third-degree relative | A person's biological great grandparent, great grandchild, great aunt, great uncle, first cousin, grand nephew or grand niece. |

Foreward

By the National Director for Heart Disease and Stroke

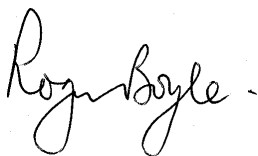
The best way to limit the damage caused by coronary heart disease is to identify those at risk as early as possible. The greatest opportunity for such disease prevention lies in diagnosing and treating people with familial hypercholesterolaemia and screening their families. Currently there are some 100,000 undiagnosed people in the UK at risk of heart disease due to familial hypercholesterolaemia.

NICE has published guidelines setting out how people with familial hypercholesterolaemia should be diagnosed and treated, and very importantly, how their families should be screened.

The pilot project reported here evaluates services against the NICE guidelines. The results do provide extremely helpful information, although care must be taken in interpretation as it is a pilot study. The current treatment of people identified with hypercholesterolaemia is good. However there are clearly apparent inadequacies in the screening programme, both in terms of cascade testing and in the use of DNA testing.

While it is reassuring that those with hypercholesterolaemia are being well managed [by the sites entered into the pilot] the results do indicate a great missed opportunity, if cascade testing is not being effectively implemented.

I am extremely grateful to all who have contributed to the work of the pilot. Not only does it demonstrate how a full national audit can most effectively be carried out but it also gives us initial indications of the ways in which services need to be improved to help reduce the burden of coronary heart disease.

A handwritten signature in black ink that reads "Roger Boyle". The signature is written in a cursive style with a small dash at the end.

Professor Roger Boyle CBE
National Director for Heart Disease and Stroke

June 2009

Executive Summary

Following the publication of the NICE Guideline for familial hypercholesterolaemia (FH)¹, we report here the findings of the pilot clinical audit to investigate the care received by individual patients who have FH.

Why do this audit?

FH is one of the most common monogenic inherited conditions in clinical practice. The prevalence of FH is about 1 in 500 (very similar to type 1 diabetes). FH patients have an increased risk of premature coronary heart disease (CHD). Approximately 50% of men, and 30% of women with FH, if untreated, will have developed clinically evident coronary heart disease by the age of 55 years².

Effective treatment is available to prevent early onset heart disease for individuals with FH. This comprises treatment with a statin to reduce their LDL-cholesterol combined with life style changes, particularly smoking cessation. This clinical approach results in a very significant reduction in their CHD mortality risk, such that well-treated patients with FH can achieve a normal life expectancy³. In the UK over 85% of the estimated 120,000 people who are thought to be affected remain undiagnosed⁴. National audit based on agreed standards and evidence based guidelines is expected to improve clinical practice, and thereby significantly reduce the mortality and morbidity associated with FH.

NICE have estimated the annual cost impact of fully implementing the guideline in England to be £7.9 million in the first year of implementation, which equates to additional costs of £16,000 for a population of 100,000⁵. Within three years considerable savings are estimated because of coronary events avoided and these will increase long term. Ongoing treatment costs would be expected to reduce progressively as higher intensity statins (often indicated for FH) come off patent.

How was the audit carried out?

Audit standards and indicators were developed from the NICE Clinical Guideline for the Identification and Management of Familial Hypercholesterolaemia (2008)⁶.

The web-based tool developed to capture the audit data worked well. Data were supplied for 248 patients, and data were duplicated for 26 of these as part of an assessment of data reliability (see Appendix 2 for details). There were very few missing or contradictory data, and the validation checks and balances in place on the webtool worked well. Some modifications have been suggested by the audit sites and the steering group to streamline and improve the tool, which will allow focus on the most important questions. Feedback suggested that, overall, sites found the audit a positive experience, and that it demonstrated their current progress in implementing the NICE guideline and identifying areas for improvements. Sites stated they would encourage others to take part in future national audits.

¹ ⁶ National Institute for Health and Clinical Excellence (NICE) - CG71: Clinical Guideline for the Management of Familial Hypercholesterolaemia (2008) <http://www.nice.org.uk/nicemedia/pdf/CG071>

² Slack J. Risks of ischaemic heart-disease in familial hyperlipoproteinaemia stats. Lancet 1969;2:1380-2

³ Neil A, Cooper J, Betteridge J, Capps N, McDowell I, Durrington P, Seed M, Humphries, SE on behalf of the Simon Broome Familial Hyperlipidaemia Register Group. Reductions in all-cause, cancer, and coronary mortality in statin-treated patients with heterozygous familial hypercholesterolaemia: a prospective registry study. European Heart Journal (2008) 29, 2625-2633.

⁴ Marks D, Thorogood M, Farrer JM, Humphries SE. Census of clinics providing specialist lipid services in the United Kingdom. J Public Health (Oxf) 2004;26:353-4

⁵ National Institute for Health and Clinical Excellence (NICE) National costing report: Familial hypercholesterolaemia Implementing NICE guidance (August 2008) <http://www.nice.org.uk/nicemedia/pdf/CG071>

What did the audit find?

The Key Findings are:

- For individual patients who have been diagnosed with FH, the clinical management in lipid clinics is of a good standard.
- Organisational issues for the care pathway of FH patients are still being developed, but it appears that centres will need additional resources to cope with the identification of the predicted additional 100,000 FH cases UK wide. This includes access to trained staff (nurses), IT needs and pedigree drawing.
- There is a major lack of systematic family “cascade” testing, whether carried out on the basis of lipid levels, or more effectively by a DNA diagnosis.
- There is limited access to DNA diagnosis and that which is available is being carried out in a research environment. Access to DNA services needs to be more widely available across the UK.
- There is a shortfall in child-focused services throughout the country, so that the ability to diagnose and treat FH in children and young persons in the health service is limited. Where such services were audited they are of a good standard.

As it is only based on 14 sites we recognise that the data are limited, and services for FH patients may be less favourable in a UK-wide audit. However, there are several key areas of clinical practice which appear to be sub-optimal in many sites, and Trusts providing services for FH patients could already consider ways to improve these deficits by developing the necessary structures and funding streams that could improve the identification of people with FH and thereby enable treatment to prevent premature heart disease.

KEY RECOMMENDATIONS

Acute trusts (England) / Integrated Trusts (Wales)

- Care pathways for FH patients need to be implemented. This must include shared care arrangements between hospital and primary care and better links between with several other specialities, including paediatrics.
- Additional resources (clinic sessions) will be needed to cope with the identification of the predicted additional 100,000 FH cases UK wide. At present there is a shortage of both specialists and lipid clinic nurses.
- Systems need to be developed and implemented to carry out systematic family “cascade” testing. This will require trained nursing/genetic services to follow up the families of index patients, improved IT needs, including an FH patient database, and pedigree drawing.
- Resources are needed for DNA diagnosis and Clinical Genetics input.

National Organisations:

- A system for coordination of cascade testing systems on a national basis is recommended, with links to genetic testing services, given that FH families are geographically dispersed.
- The lack of paediatric services may be best coordinated at a national level to ensure that appropriate child focused services are developed.
- On the basis of this pilot study it is recommended that a national audit of services is very feasible and should be commissioned

Introduction

Familial hypercholesterolaemia [FH] is a common inherited disorder of lipid metabolism causing high levels of low-density-lipoprotein cholesterol [LDL-C], which leads to early coronary heart disease [CHD]. Roughly half of men with FH, if untreated, will have developed clinically evident CHD by the age of 55 years, and roughly one third of women by 60 years⁷. A significant reduction in the mortality and morbidity of the disease can be achieved through changes in lifestyle and the use of statins to lower cholesterol, but currently the vast majority of affected individuals in the UK remain undiagnosed [probably 85% of the predicted 120,000 cases]⁸. Testing the families of known cases of FH [cascade testing] can identify those with FH, and data obtained from a large on-going project in Holland and a DH pilot project in the UK indicates this is cost-effective.

In August 2008 the National Institute for Health and Clinical Excellence (NICE) published evidence-based recommendations for the diagnosis and management of FH (CG71, 2008)⁹. Using these recommendations as the standards, we present here a pilot project to develop tools and determine feasibility and acceptability of audit of the management of FH. This pilot project initiated and carried out by the Royal College of Physicians between July 2008 and June 2009, and was funded by the Department of Health [DH], with part funding from Cardiac Network Coordinating Group in Wales. Subject to funding, it is the intention that this audit will be rolled out for use on a national basis in England and Wales, Scotland and Northern Ireland.

The considerable efforts of NHS hospital health professionals and clinical audit staff have enabled a maximum return rate of 100% of the invited acute trusts in England and Integrated Trusts in Wales. Based on audit of these 14 sites, this pilot provides a snapshot of the range of clinical out-patient services provided to and the quality of management of FH patients across England and Wales. It details the current level of implementation of the standards set out in the NICE FH guidelines in the participation sites, and identifies several key areas where NICE recommended standards are not being achieved and from which recommendations for service improvement can be made. We recognise that the data are limited and a full national audit will need to be undertaken to get a complete picture of the level of implementation surrounding the NICE guideline. However, if confirmed by a national audit, the correction of these deficits could be achieved by promoting the development of the necessary structures and funding streams that could dramatically improve the identification of people at risk of premature heart disease because of FH and enable preventative treatment.

You are encouraged to consider the key messages and recommendations in the report.

Finally, thank you to everyone who helped in the design, performance and analysis of this first pilot of this FH clinical audit.

⁷ Slack J. Risks of ischaemic heart-disease in familial hyperlipoproteinaemia stats. *Lancet* 1969;2:1380-2

⁸ Marks D, Thorogood M, Farrer JM, Humphries SE. Census of clinics providing specialist lipid services in the United Kingdom. *J Public Health (Oxf)* 2004;26:353-4

⁹ National Institute for Health and Clinical Excellence (NICE) - CG71: Clinical Guideline for the Management of Familial Hypercholesterolaemia (2008) <http://www.nice.org.uk/nicemedia/pdf/CG071>

METHODS

Standards

The pilot audit was based on the standards in the NICE Guideline 71 “Identification and Management of Familial Hypercholesterolaemia” published in August 2008¹⁰.

Objective

The aim of the pilot audit was to develop and evaluate audit tools to measure current practice in the management of FH against the recommendations identified in the NICE Guideline (NICE CG71, 2008).

The audit had two elements:

Organisational audit to explore the organisation and resources available for people with FH in clinics with expertise in lipid disorders (mostly lipid clinics). The Organisational Survey was divided into seven elements:

- Set Up
- Clinical Time
- Management of FH
- Cascade Testing
- Trust Services
- Patient Information
- Service Improvement

Clinical audit to review the clinical management and process of care of consecutive sample of people (adults and children) attending outpatient clinics in relation to their FH, assessing:

- Section 1: Diagnosis
- Section 2: Pedigree
- Section 3: Assessment
- Section 4: Treatment
- Section 5: Advice
- Section 6: Cascade Testing
- Section 7: (Annual) Review
- Section 8: Children

The audit required data to be collected from policy documents and patient records, suggestions of where the data may be located were provided in accompanying help notes.

Target population for the clinical audit

It was decided to focus the audit on out-patient FH services for adults and children.

Governance of the audit

The project was supervised by a multi-disciplinary steering group including one person with FH as a patient representative. The project management and governance standards were in line with those employed by the Royal College of Physicians Clinical Effectiveness and Evaluation Unit. Quarterly monitoring meetings were undertaken with the main funders (Department of Health).

¹⁰ National Institute for Health and Clinical Excellence (NICE) - CG71: Clinical Guideline for the Management of Familial Hypercholesterolaemia (2008) <http://www.nice.org.uk/nicemedia/pdf/CG071>

Development of Audit Indicators

The Working Group and Steering Group developed indicators derived from the NICE evidence base guideline.

The audit criteria were drawn from the NICE guideline's key priorities for implementation, in particular:

- prescribing a high intensity statin to achieve a greater than 50% reduction in LDL concentration
- offering a DNA test
- using systematic methods of cascade testing for the identification of people with FH
- testing children by the age of 10 years, which should be carried out in a child/young person focused setting,
- giving women and girls on statins information about contraception (because of the slight risk of birth defects in babies conceived whilst taking a statin),
- having an annual structured review.

Pre Pilot exercise

Three acute sites were recruited and undertook a pre-pilot exercise, providing feedback on the hard copy tools.

Inclusion criteria

The audit sampled adults and children with a definite or possible diagnosis of FH (as per the NICE guideline). For the purposes of this audit children are considered under 16 years of age. Participating sites were encouraged to choose an appropriate sample ideally capturing any paediatric cases of FH.

The focus of the audit was people with FH who were accessing outpatient services for the third clinical appointment or greater. Sites were requested to select cases for clinical audit from the first 40 consecutive cases with a diagnosis of FH (ICD 10 Code E78. 0) visiting a clinic with expertise in lipid disorders for the third time or more between 12 January - 3 April 2009.

The numbers were chosen, with statistical advice, to provide the minimum necessary to enable a meaningful analysis comparing individual sites so that local services can be benchmarked against the overall national figure. It was recognised at pre-pilot stage that the majority of sites would not get 40 cases.

Exclusion criteria

1. Patients who have a possible or definite diagnosis of FH attending their first or second clinic appointment
2. Patients with lipid disorders other than FH

Recruitment of pilot sites

Sites were selected to reflect geographical spread, type of hospital and level of service provided. Selected Acute hospital trusts were recruited via letters to their chief executive, medical director, any identified FH lead, and clinical audit or effectiveness managers. They were asked to provide details of leads and contacts.

Participation

Of the 18 sites invited to participate, 14 sites signed up for the pilot project. Of these all sites supplied data on one or both groups of patients (adults and children). Over 248 patients were in the sample, 226 adults (91%); 22 children (9%) in the primary cases. 26 adults were duplicated as part of the inter-auditor reliability analysis (see page 16 for further details).

Data collection tool

The Organisational Survey required entry of hospital management data to the web-based data collection tool. It required the auditor to have access to the information and an understanding of how each element is organised.

The Clinical Audit comprises a clinical case note audit of consecutive admissions of patients with FH, prospectively identified between Monday 12 January and Friday 3 April 2009.

Sites were encouraged to develop a mechanism for identifying all applicable FH cases attending out-patient clinics. This may be done by:

- allocating a doctor or nurse to contact all clinics to identify patients, and log these for subsequent data collection from case notes shortly after the clinic visit OR
- using a log book

It may be that the Patient Administration System (PAS) or local FH database can be used to cross check the capture of all FH cases.

The audit tools were designed to measure the implementation of the NICE Clinical Guideline. The NICE guideline is primarily aimed at adults and the audit tool was designed to reflect this.

Support and Information for participating sites

As soon as sites were recruited and had provided their contact details they were kept up to date with monthly newsletters or by email. The FH Audit website was regularly updated and included a frequently asked question sheet. Support information was provided for auditors, giving definitions and guidance on how to answer each audit question for the patient groups. Sites were provided with guidance on how to undertake the audit based on feedback from the pre-pilot sites.

They were advised to gather data initially on the paper copy of the data collection tool and to enter this onto the web tool once data collection from various sources was complete. As questions arose from sites these were compiled into a frequently asked question sheet and made available on the FH Audit website. All sites were fully prepared for the audit by the second week of January 2009 in readiness to begin data collection. Data collection continued into April 2009.

Information governance

Data were collected via a secure website (<https://audit.rcplondon.ac.uk/fh>). Individual patient consent is not required where unidentifiable patient data is used for audit purposes. No patient identifiable information was collected. Sites were asked not to put any information that may identify the patient in the free text comments boxes.

Data entry to the web tool

Web tool help notes were provided and a help desk was available to sites during data collection by email and phone. Sites were asked to refer to the web tool user notes to aid

data entry and the support information to clarify any of their answers. Each site was provided with their own unique password and site code. Many data entry items required a simple 'YES' or 'NO' response but sometimes other options were possible, i.e. 'not relevant'.

To improve quality of data entry, the web tool had routing and consistency checking built-in to it which meant in particular that if the answer to a stem question was 'No' then it was not possible to answer any of the sub section questions; conversely if the answer to the stem question was 'Yes' then an answer to any sub section question was required in order to progress further with data entry. The web tool was designed for both groups of patients with all sections pertinent to both except for children.

Inter-auditor reliability

Sites were asked to re-audit their first 5 cases, using a different auditor, and returning to the clinical notes again as the source of audit data. 6 sites submitted 26 cases. Reliability (agreement between auditors) is not the same as validity (suitability or accuracy of measure). However establishing good agreement between auditors is an important part of the process of validation, and is therefore critical to piloting a new audit tool. Given the small numbers, the kappa statistic was not used to quantify agreement. The levels of agreement were generally encouraging. There were some questions with poorer results where comments made by participants shed light on possible ambiguity, and here detail can in future be added to the help notes or on the data collection tool. For a more detailed summary of the reliability analyses see Appendix 2.

Post export quality checks

After all data had been entered by a site they were asked to lock their forms and to export their data to a spreadsheet. They were asked to quality check their exported data against a sample of their hard copy data collection forms. If there were errors caused by transcribing these were amended. Sites could do local analyses on their own data.

Analysis of data

Data were held securely and analysed at the Royal college of Physicians' Clinical Standards Department. The software used was SPSS version 15 and Stata version 8/SE. Because of small numbers of children in this pilot audit, the graphs displaying individuals' lipoprotein results show only the adults.

PILOT PHASE EVALUATION

Objective

The audit pilot successfully developed audit tools to measure the current practice in the management of FH against the recommendations identified in the NICE Guideline (CG71, 2008)

Target population

The audit was successful in its aim to audit adults and children with FH. Data were supplied for 248 patients, and data were duplicated for 26 of these as part of an assessment of inter-rater reliability (see Appendix 2 for details).

Development of audit indicators

In general the audit tool enabled the level of implementation of the NICE guideline to be measured. There is need for the audit tool to be more concise in some areas.

Lipoprotein measurements at diagnosis

There was evidently some difficulty for a minority of the patients in finding the correct set of lipoprotein measurements used at diagnosis. This can be seen by comparing the years recorded in the data. In future rounds of audit, this should be used to validate the data as it is typed into the webtool.

Pedigree

Section 2 of the clinical audit proforma relates to the FH patient's pedigree. NICE advises that 3 generations should be captured in the pedigree. This can be interpreted in one of two ways: capturing first, second and third generation or first-degree, second-degree and third-degree relatives. The two sets of definitions are different (see glossary). Despite rigorous scrutiny by the Working Group and Steering Groups and testing at pre-pilot stage this only came to light during the pilot audit. There are always likely to be mistakes in first attempts at audit tools, hence the reason why we pilot. This section would need to be rectified in the full national audit.

Annual review

The audit only included patients with FH attending their third clinic appointment or greater. The audit asked whether the patient had a structured annual review since the initial diagnosis. Patients who are routinely discharged would not be included in the audit. By including all patients in a full national audit the likelihood of bias would be removed.

Paediatrics

It is of relevance that the NICE guidelines only partially address some important differences between adult and paediatric services. For example in paediatrics there is a need for smoking prevention programs rather than smoking cessation programs.

Inclusion criteria

Overall, sites found it straightforward to select eligible patients. Only one site was able to enter 40 cases.

Audited patients were only those attending their third clinic appointment or greater, so as to obtain information about key activities that occur later in the care pathway, such as cascade testing, annual review etc. There is however the possibility that this may have introduced bias into the sample. Feedback from sites suggests that if a future audit

included all FH patients this would require auditing an estimated further 5 patients during a 3 month data collection period.

Recruitment of pilot sites

The aim of the audit was to audit 10 sites; 18 sites were invited to participate and 14 sites registered to undertake the audit. Sites reflected the geographical spread, type of hospital and level of service provided.

Participation

All of the 14 sites who registered to participate completed the organisational and clinical audit.

Data collection tool

The validation checks and balances in place on the webtool worked well. The data collection tool requires minor enhancements to improve data validation.

Inter-rater reliability

Analysis of how different data collectors agreed on the information required for the clinical audit showed that most of the questions produce reliable answers. Some others provided us with feedback which has informed improvements in the helpnotes and the webtool.

Support and information

Feedback suggests that overall sites were happy with the written information (helpnotes and newsletters) and helpdesk support provided.

Evaluation of audit process

Sites were encouraged to provide written/verbal feedback at all stages of the audit process. After the data collection period participating sites were asked to complete a survey detailing their experience of completing the audit process. A summary of the results can be found in appendix 4.

Conclusion

Overall the pilot audit was considered a success meeting the objectives of the project. There was a high level of participation. The pilot project has provided useful data on the level of implementation of the NICE guideline. A full national audit would be required to get the national picture.

RESULTS

Summary of results

This first round of data collection and analysis has shown significant needs for improvement and development of services for people with FH. Acute Trusts, as providers of service, and Primary Care Trusts in England (PCTs) and Local Health Boards in Wales (LHBs), as Commissioners of services, should work together to develop an action plan to address the gaps in service provision.

- Overall patient management of FH is good; there is good documentation of family history and the majority of case notes contained a pedigree and diagnosis included secondary causes of FH being considered.
- The majority of sites had no formal policy for the diagnosis and cascade testing of FH, or an integrated care pathway or clinical management strategy for the management of FH.
- While many sites have shared care arrangements with General practice, Cardiology, Obstetrics, etc, most of these are informal.
- Currently only 20% of sites report either a paper or electronic database for FH patients.
- Half of sites had some arrangements in place for DNA testing although only two sites received funded for this; this is thought to be through research rather than NHS funding.
- Only 15% of sites had a designated cascade testing service. The audit suggests that very little cascade testing is systematic, being rather done on an opportunistic basis.
- Provision of written advice about diet, exercise, smoking and alcohol consumption was good, although for 45% of females of reproductive age there was no documentation of discussion of contraceptive advice.

Presentation of results

The NICE Guideline recommendations audited are presented at the beginning of each section. Following this there is a summary of the results with figures reflecting the national picture. Unless otherwise stated the percentages reported are of all relevant patients, the denominator being dependent on which aspects are applicable.

Throughout this report the results for the two patient groups (adults and children) are shown separately.

PART 1: ORGANISATIONAL AUDIT

Section 1: Set up

NICE GUIDELINE RECOMMENDATION

Healthcare professionals should offer all children and young people diagnosed with, or being investigated for, a diagnosis of FH a referral to a specialist with expertise in FH in children and young people. This should be in an appropriate child/young person-focused setting that meetings the standards within the 'National service framework for children, young people and maternity services' (available from www.dh.gov.uk).

14 sites returned data for this audit.

The fourteen sites have a wide geographical spread across England and Wales, and include lipid clinics in district general hospitals, teaching hospitals and tertiary referral centres, as well as one specialised paediatric centre. We therefore believe they are likely to be representative of lipid clinics throughout the country, although as with any pilot, those “volunteering” to take part are those more likely to be research orientated than average. The results mirror that found in the 2008 Heart UK survey, that FH patients are managed in a spectrum of directorates, with the majority in Chemical Pathology, Diabetes and Cardiology. Although the majority of sites have a lead clinician responsible for FH care, less than 50% have someone with operational managerial responsibility for FH service and most sites do not run a dedicated FH clinic. Currently only 15% of sites have a designated cascade testing clinic, with another 15% in development of this service. Slightly over 50% of sites have a specialist service for the management of young people with FH. While many sites have shared care arrangements with General practice, Cardiology, Obstetrics, etc, most of these are informal. Currently only 20% of sites report either a paper or electronic database for FH patients.

Type of hospital

1.1 What type of hospitals are in your Trust? *Sites were asked to tick all that apply.*

| | |
|---------------------------|-------|
| Teaching Hospital | 7/14 |
| District General Hospital | 10/14 |

Provision of FH services

1.2 Does your Trust provide out-patient services for the clinical management of:

| | |
|---|-------|
| a. Adults with FH? | 13/14 |
| b. Children/young people (under 16s) with FH? | 11/14 |

Of those sites who provide outpatient services for adults with FH (i.e. *responded positively to 1.2 a*)

1.3 *Who provides the out-patient services for the clinical management of adults with heterozygous FH?*

Sites were asked to tick one answer option

| | |
|---|-------|
| Provided by Trust | 13/13 |
| Provided by a visiting service from another Trust | 0/13 |
| By referring to another service or centre | 0/13 |

Sites answered this question if they responded positively to 1.2 a:

1.4 *Who provides the out-patient services for the clinical management of adults with homozygous FH? Tick one option*

| | |
|---|------|
| Provided by Trust | 4/13 |
| Provided by a visiting service from another Trust | 0/13 |
| By referring to another service or centre | 9/13 |

Sites answered this question if they positively to 1.2 b:

1.5 *Who provides the out-patient services for the clinical management of children with FH?*

Sites were asked to tick one answer option

| | |
|---|-------|
| Provided by Trust | 11/11 |
| Provided by a visiting service from another Trust | 0/11 |
| By referring to another service or centre | 0/11 |

Directorate / Care Group that FH is under

1.6 *What Directorate/ Care Group is FH under? Tick all that apply*

| | |
|-------------------------------------|-------|
| Diabetes and Endocrinology | 4/14 |
| Cardiology / Cardiovascular | 2/14 |
| Chemical Pathology | 10/14 |
| General Medicine / General Internal | 1/14 |
| Specialist Medicine | 1/14 |
| Paediatrics | 3/14 |
| Other | 2/14: |

“Inherited metabolic disorders”

“University department of medicine”

Managerial responsibility

1.7 *Does your Trust have someone with operational managerial responsibility for FH services?*

Yes: 6/14

1.8 *Does your Trust have a lead clinician responsible for FH care?*

Yes: 12/14

Out-patient clinics

1.9 How many of the following out-patient clinics are there per month:

| | Adults - morning clinical (am) | Adults - afternoon clinics (pm) | Paediatric clinics (am) | Paediatric clinics (pm) |
|--|--|--|--------------------------------|----------------------------|
| Dedicated FH clinics | 1 site responded: 4 clinics | 1 site responded: 4 clinics | 1 site responded: 2 clinics | No sites |
| General lipid clinics | 11 sites: median 2 IQR 1-9 | 4 sites: 2 had 1 clinic 1 had 2 clinics 1 had 4 clinics | No sites | No sites |
| Other medical clinics (where FH patients are seen in relation to FH) | 3 sites: 1 had 1 clinic 1 had 2 clinics 1 had 8 clinics | 2 sites: both had 4 clinics | 1 site: 1 clinic | 1 site: 1 clinic |

Designated cascade testing clinic

1.10 Does your Trust have a designated clinic to test for FH by cascade testing?

| | |
|-----------------|-------|
| Yes: | 2/14 |
| In development: | 2/14 |
| No: | 10/14 |

Out patient clinics

1.11 Please estimate how many patients with FH were seen in out-patient clinics in the last year (this should include definite, probable/possible):

| | Adults | Paediatrics |
|--------------|---|--|
| New patients | 1 site reported none median 20 IQR 14-49 | 4 sites reported none median 5 IQR 1-7 |
| Follow-up | 1 site reported none* median 102 IQR 50-201 | 5 sites reported none median 10 IQR 0-18 |

* A Pediatric only site

Specialist services for management of young people

1.12 Does your Trust have specialist services for the management of young people with FH?

Yes: 8/14

Sites answered 1.12 a & b if they answered 'yes' to 1.12

1.12 a. do your specialist services have access to paediatric phlebotomy?

Yes: 7/8

b. Do you have family clinics?

Yes: 4/8

Shared care arrangements

“Formal arrangements” are considered to be where there are written/ documented and agreed share care arrangements, with “informal arrangements” being where arrangements are in place however there is no written formal documentation to support this.

1.13 Do you have shared care arrangements in place with:?

- a. General practice
 - Yes, formal: 0/14
 - Yes, informal: 10/14
 - No: 4/14
- b. Cardiology
 - Yes, formal: 0/14
 - Yes, informal: 8/14
 - No: 6/14
- c. Obstetrics
 - Yes, formal: 0/14
 - Yes, informal: 5/14
 - No: 9/14
- d. Paediatrics
 - Yes, formal: 2/14
 - Yes, informal: 8/14
 - No: 4/14
- e. LDL Apheresis services
 - Yes, formal: 1/14
 - Yes, informal: 5/14
 - No: 8/14

Database of patients with FH

1.14 Do you have a paper database of patients with FH?

| | |
|-------------------------------------|------|
| Paper database - complete | 2/14 |
| Paper database - partially complete | 6/14 |
| No | 6/14 |

1.15 Do you have an electronic database of patients with FH?

| | |
|--|------|
| Electronic database - complete | 3/14 |
| Electronic database - partially complete | 7/14 |
| No | 4/14 |

Sites with either paper or electronic databases:

| | |
|---|------|
| At least one form complete | 3/14 |
| Neither complete, but at least one partially complete | 9/14 |
| Neither | 2/14 |

Section 2: Clinical time

Although all sites report some clinician and nurse time for FH patient management, it appears that this is unlikely to be adequate if GP referral numbers increase markedly.

Clinical time

There are 10 Consultant Programme Activities (PAs) per week. Sites were asked to indicate how many Consultant's PAs are devoted to specialist lipid management and how many Lipid Specialist Nurses they had in whole time equivalent (WTE) - for the purposes of the audit, one WTE is 37.5 hours.

2.1 In your Trust, how much clinical time is devoted to specialist lipid management/FH by your employed staff?

- a. How many Consultant's programmed activities (PAs) are devoted to specialist lipid management?
All 14 sites reported some
Median 2
IQR 2-4

- b. On average, what proportion (%) of this is estimated to be spent on the management of FH?

| | |
|---------|------|
| <10% | 3/14 |
| 11-20% | 7/14 |
| 21-40% | 1/14 |
| 41-60% | 1/14 |
| 61-80% | 1/14 |
| 81-100% | 1/14 |

- c. How many Lipid specialist nurses are there (WTE)?
2 sites reported 1 WTE each

- d. On average, what proportion (%) of this is estimated to be spent on the management of FH?

| | |
|---------|------|
| <10% | 0/14 |
| 11-20% | 0/14 |
| 21-40% | 0/14 |
| 41-60% | 1/14 |
| 61-80% | 1/14 |
| 81-100% | 0/14 |

Section 3: Management of FH

Only 65% of trusts formally classify patients according to the Simon Broome criteria or offer an annual review. Most sites do not have a formal policy for the diagnosis and cascade testing or an integrated care pathway for FH patients.

Patient classification

- 3.1 Does your Trust formally classify patients according to the Simon Broome criteria?
Yes: 9/14

Annual review

- 3.2 Does your Trust routinely offer FH patients an annual review?
Yes: 9/14

Routine discharge

- 3.3 Does your Trust routinely discharge FH patients once lipid levels are optimised?
Yes: 5/14

Written policy/protocol for clinical management of FH

3.4 Is there a written policy/protocol for the clinical management of FH?

Yes: 2/14
In development: 2/14
No: 10/14

Sites were asked to answer a-d if they answered yes to 3.4

a. Has this been formally approved by your Trust?

Yes: 2/2
In progress: 0/2
No: 0/2

b. Does the policy include diagnostic criteria for FH?

Yes: 2/2

c. Does the policy include advice on cascade testing?

Yes: 2/2

d. Does the policy include advice on annual review?

Yes: 1/2

Integrated care pathway/clinical management strategy

3.5 Is there an integrated care pathway/ clinical management strategy for the management of FH?

Yes: 2/14
In development: 1/14
No: 11/14

Section 4: Cascade Testing

NICE GUIDELINE RECOMMENDATION:

Healthcare professionals should offer all people with FH a referral to a specialist with expertise in FH for confirmation of diagnosis and initiation of cascade testing.

Cascade testing is as yet considerably under developed in the fourteen sites. Only 21% have access to a formal system, with 29% saying this is in development. Only 15% have any form of dedicated IT software to help manage this service.

4.1 Is there access to a family cascade testing system for FH in your Trust?

Yes: 3/14
In development: 4/14
No: 7/14

4.2 If yes, how is this provided?

| | |
|--|-----|
| Provided by Trust | 2/3 |
| Provide by visiting service from another Trust | 0/3 |
| By referring to another service or centre | 1/3 |

4.3 If yes, who is responsible for the clinical governance of the cascade testing for families?

| | |
|------------------------------------|-------------------------------|
| Trust | 2/3 |
| Regional Clinical Genetics Service | 0/3 |
| Other | 1/3: “Welsh pilot FH project” |

4.4 If provided by Trust, does this include:

a. Dedicated staff?

| | |
|-----------------|-----|
| Yes: | 2/2 |
| In development: | 0/2 |
| No: | 0/2 |

b. Formal input from the local Genetic services?

| | |
|-----------------|-----|
| Yes: | 1/2 |
| In development: | 1/2 |
| No: | 0/2 |

4.5 Do the staff who are looking after individuals and families with FH routinely have access to:

a. dedicated IT software

| | |
|-----------------|-------|
| Yes: | 2/14 |
| In development: | 2/14 |
| No: | 10/14 |

b. pedigree drawing software?

| | |
|-----------------|-------|
| Yes: | 2/14 |
| In development: | 1/14 |
| No: | 11/14 |

Section 5: Trust services

NICE GUIDELINE RECOMMENDATION

Cascade testing using a combination of DNA testing and LDL-C concentration measurement is recommended to identify affected relatives of those index individuals with a clinical diagnosis of FH.

While all sites have access to an accredited laboratory for lipid measurements and the majority for smoking cessation or dietary support, only 15% of sites have access to funded DNA testing.

5.1. Does your Trust have arrangements for DNA mutation testing for FH patients attending your clinical service?

| | |
|--------------|------|
| Yes - funded | 2/14 |
|--------------|------|

| | |
|---------------------------------|------|
| Yes - not funded | 3/14 |
| Available only in special cases | 2/14 |
| No | 7/14 |

5.2 Does your Trust have a laboratory that is currently accredited (CPA) for testing lipid measurements?

Yes: 14/14

5.3 Does your Trust have routine access to HDL measurements?

Yes: 14/14

5.4 Is there a smoking cessation support service that FH patients can be referred to?

Yes: 11/14

Smoking cessation support services included Hospital/PCT/LHB or NHS Stop Smoking Services.

5.5 Is there access to specialist dietary advice?

| | |
|----------------------------|-------|
| Yes - in clinic | 4/14 |
| Yes - separate from clinic | 10/14 |
| No | 0/14 |

Section 6: Patient information

NICE GUIDELINE RECOMMENDATIONS

During the assessment and communication of familial risk, people should receive clear and appropriate educational information about FH, the process of family testing, DNA testing and the measurement of LDL-C concentration.

A healthcare professional with expertise in FH should provide information to people with FH on their specific level of risk of coronary heart disease, its implications for them and their families, lifestyle advice and treatment options.

The sites appear to have a wide range of information leaflets to help FH patients, but 15% report no leaflets available.

Patient information

6.1 Which, if any, of the following information leaflets do you provide to FH patients/carers?

| | |
|--|--------------------------|
| a. British Heart Foundation leaflet(s) | 3/14 |
| b. Heart UK leaflet(s) | 10/14 |
| c. NICE information for the public | 1/14 |
| d. Trust leaflet | 3/14 |
| e. Other | 6/14: |
| | “alcohol advice sheet” |
| | “appt card/results/info” |

“created by lipid nurse”
“FH booklet”
“lipid clinic patient info.”
“To My Healthy Heart (local)”

2/14 sites gave none of these.

4/14 gave one only, and for all these sites it was Heart UK.

6.2 If yes, does the information provided cover the following subject areas?

| | |
|---------------------------|-------|
| a. Diet | 11/12 |
| b. Exercise | 10/12 |
| c. Smoking | 9/12 |
| d. Alcohol | 11/12 |
| e. FH as a condition | 10/12 |
| f. Inheritance | 9/12 |
| g. Role of DNA testing | 5/12 |
| h. Treatment options | 9/12 |
| i. Treatment for children | 8/12 |
| j. Monitoring | 6/12 |
| k. DNA Testing | 5/12 |
| l. Cascade Testing | 5/12 |

Section 7: Service improvement

Currently only 15% of sites report specific links to a patient or service user group, with 21% reporting that this is in development. Only 7% of sites report that FH care is subject to regular audit.

Patient/service-user group

7.1 Does the FH service have specific links to a patient/service-user group?

| | |
|-----------------|------|
| Yes: | 2/14 |
| In development: | 3/14 |
| No: | 9/14 |

Sites answered question 7.2 if they answered yes to 7.1

7.2 Is the patient/service user group involved in planning and delivery of FH services?

| | |
|-----------------|-----|
| Yes: | 1/2 |
| In development: | 0/2 |
| No: | 1/2 |

Regular audit

7.3 Is FH care delivered by the service subject to regular audit?

| | |
|------|-------|
| Yes: | 1/14 |
| No: | 13/14 |

Regular audit is considered to be at least annually. This could be process and/or outcome audit.

PART 2: CLINICAL AUDIT

Sites were asked to select cases for clinical audit from the first 40 consecutive cases with a diagnosis of FH (ICD 10 Code E78. 0) visiting a clinic with expertise in lipid disorders for the third time or more between 12 January - 3 April 2009.

Data were submitted on 248 patients and duplicates were entered for 26 of these. 14 sites collected these data, ranging from 3 patients to 40, with median 16 patients and inter-quartile range 8-28.

Section 0: General information

The audit information reflects the wide spectrum of FH patient care facilities throughout the UK. The age spectrum is very similar to that published by Neil et al in BMJ, and it is possible that the bimodal peak at age 45 represents males and that at 65 represents females. Children are clearly under-represented. The survey has identified slightly more women than men which is similar to that found in the DH Audit project. Possible reasons for this include that a survivor effect and that, since these were people attending their third clinic visit, that women are more likely to re-attend than men. The ethnicity range is as expected, with 70% being White British. The 21% of individuals where ethnicity was not recorded is an important audit issue.

Auditor speciality/grade

0.1 Auditor Speciality:

| | |
|------------------------------|---|
| Chemical Pathology | 102 (41%) |
| Clinical Pharmacology | 0 |
| Diabetology | 10 (4%) |
| Endocrinology | 9 (4%) |
| Paediatrics | 10 (4%) |
| Specialist Lipid | 53 (21%) |
| Clinical Effectiveness/Audit | 15 (6%) |
| Other | 49 (20%) - 27 (from 1 site) were entered by LDL apheresis nurse |

a. Auditor Grade:

| | |
|----------------------|-----------|
| Consultant | 118 (48%) |
| Specialist Registrar | 10 (4%) |
| Specialist Nurse | 64 (26%) |
| Nurse | 15 (6%) |
| Other | 26 (10%) |
| Missing | 15 (6%) |

It should be noted that professionals involved in data entry for a pilot project may not be representative of those who would retrieve and submit data for a national audit.

Out-patient clinic type

0.2 In what clinic was the patient seen?

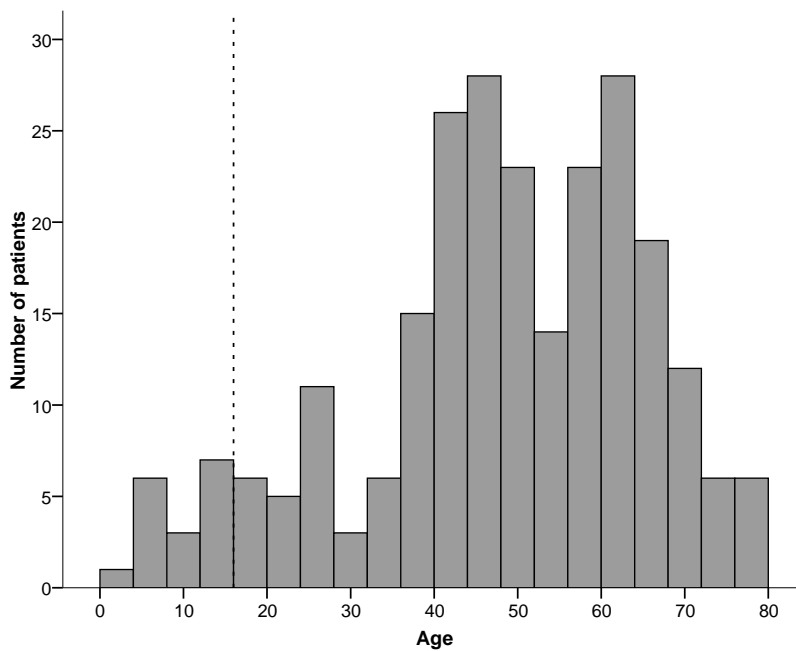
| | |
|---------------------------------------|-----------|
| Familial hypercholesterolaemia clinic | 15 (6%) |
| Lipid Clinic | 217 (88%) |
| General Medicine clinic | 11 (4%) |
| Paediatric clinic | 5 (2%) |

Of 23 patients up to the age of 18, 5 were seen in paediatric clinics.

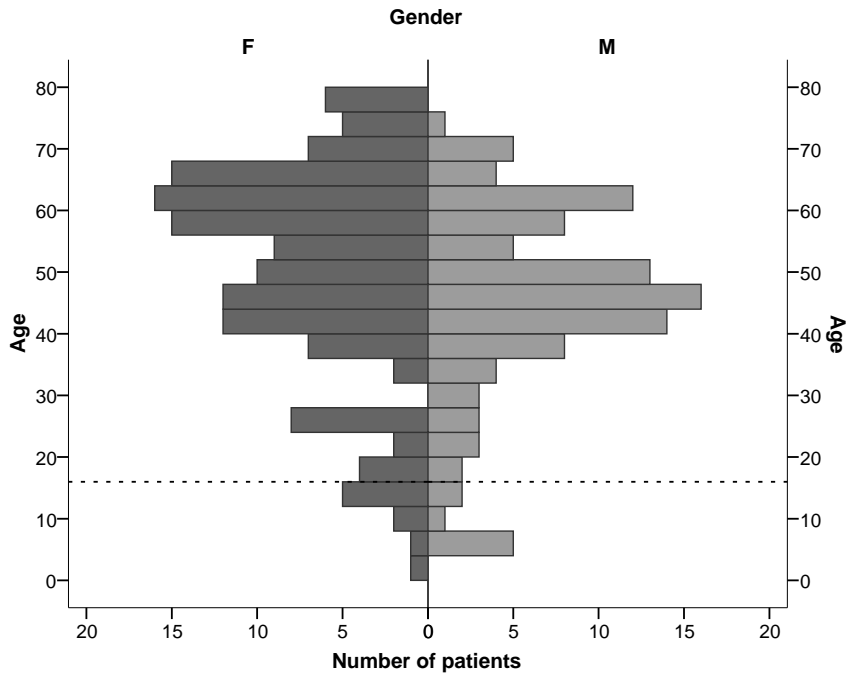
Of 17 patients up to the age of 16, 5 were seen in paediatric clinics.

Patient demographics

0.4 Patient's age at clinic



The vertical dotted line shows the child/adult divide.



0.5 Patient's gender ? Male 109 (44%)
 Female 139 (56%)

0.6 Ethnic group

| | | |
|-------------------------------|----------------------------|-----------|
| White | British | 165 (66%) |
| | Irish | 1 |
| | Any other White background | 9 (4%) |
| Asian or Asian British | Indian | 3 (1%) |
| | Pakistani | 1 |
| | Bangladeshi | 1 |
| | Any other Asian background | 0 |
| Black or Black British | Caribbean | 1 |
| | African | 1 |
| | Any other Black background | 1 |
| Mixed | White and Black Caribbean | 1 |
| | White and Black African | 1 |
| | White and Asian | 0 |
| | Any other mixed background | 0 |
| Chinese / Other ethnic groups | Chinese | 0 |
| | Any other ethnic group | 7 (3%) |
| Not recorded | | 53 (21%) |

Section 1: Diagnosis

NICE GUIDELINE RECOMMENDATIONS

Healthcare professionals should exclude secondary causes of hypercholesterolaemia before a diagnosis of FH is considered.

A diagnosis of FH should be made using the Simon Broome criteria, which include a combination of family history, clinical signs (specifically tendon xanthomata), cholesterol concentration and DNA testing.

A family history of premature coronary heart disease should always be assessed in a person being considered for a diagnosis of FH.

Healthcare professionals should offer people with a clinical diagnosis of FH a DNA test to increase the certainty of their diagnosis and to aid diagnosis among their relatives.

The proportion of possible to definite FH patients of roughly 1.5:1 is similar (but lower) than that reported in the DH audit. It may be that the more difficult to manage DFH patients are being kept in the clinics for management. The 27% of individuals who had a DNA test is higher than expected UK wide, and is likely to reflect the research interests of the 14 sites volunteering for the audit. The 0.4% of patients who had refused a DNA test is encouraging. The fact that the diagnosis was made using a non-fasting sample in only 5% of individuals is encouraging. There is good evidence that secondary causes of hypercholesterolaemia were excluded before a diagnosis of FH was considered (all greater than 88%). The range of lipid traits measured is appropriate and as expected for FH patients. 52% of the individuals had evidence in the notes that a baseline ECG was considered.

Patient diagnosis

1.1 What is the patient's diagnosis? (*Sites were asked to tick one option*)

| | |
|--|-----------|
| Definite Homozygous FH | 2 (1%) |
| Definite Heterozygous FH | 94 (38%) |
| Possible FH | 136 (55%) |
| FH not clearly stated as definite/possible | 16 (6%) |

1.2 Year of diagnosis (FH possible or definite)? Median 2005, interquartile range 2000-2007, range 1978-2009 Not recorded 14 (6%)

The "not known" responses are all from patients diagnosed in the last few years, and there is also a higher proportion of "possible FH" responses for these people.

History of CHD

Coronary heart disease includes MI, CABG, PTCA, coronary angiography and angina.
Premature Coronary Heart Disease (CHD) = <55 male, <65 female in relatives

1.3 Is there evidence in the notes that there is a history of CHD in the patient?

Yes: 80 (32%)

Blood relatives

1.4 Is there evidence in the notes that there are any blood relatives?

Yes: 238 (96%)

Premature coronary heart disease

1.5 Is there a record of premature coronary heart disease in the patient's blood relatives?

Yes: 179 (75%)

DNA testing

1.6 Is there evidence that the patient accepted or refused a DNA test for FH?

| | |
|---|-----------|
| Patient had DNA test - evidence of consent | 56 (23%) |
| Patient had DNA test - no evidence of consent | 10 (4%) |
| Patient refused DNA test | 1 (0.4%) |
| Patient not offered DNA test | 119 (48%) |
| Not recorded | 62 (25%) |

Diagnosis

1.7 Is there evidence that the diagnosis included assessment of the following clinical signs:

| | |
|------------------------|-----------|
| a. corneal arcus? | 194 (78%) |
| b. tendon xanthomata ? | 191 (77%) |

Lipid profile

1.8 Was the lipid profile on which the diagnosis made using a fasting sample or non fasting sample?

| | |
|--------------|-----------|
| Fasting | 217 (88%) |
| Not fasting | 13 (5%) |
| Not recorded | 18 (7%) |

Secondary causes of FH

Diabetes: This could be either a fasting Blood glucose or HBA1C.

Renal Failure: This is U&E hypothyroidism levels TSH.

1.9 Is there evidence that the following secondary causes of hypercholesterolaemia were excluded before a diagnosis of FH was considered?

| | |
|-------------------|-----------|
| a. Diabetes | 218 (88%) |
| b. Renal failure | 223 (90%) |
| c. Hypothyroidism | 226 (91%) |
| d. Cholestasis | 225 (91%) |

Most of these patients had all four excluded (215, 87%), or none (19, 8%).

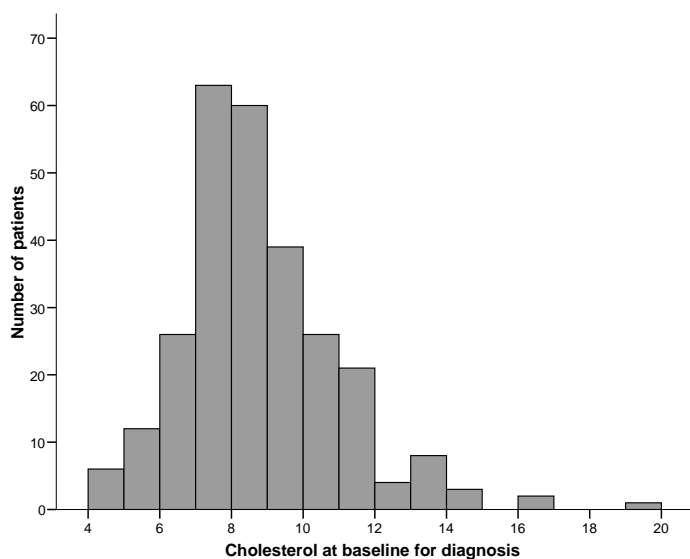
Lipoprotein measurements

Sites were asked to record the lipid results of the blood test that confirmed the diagnosis of FH before treatment.

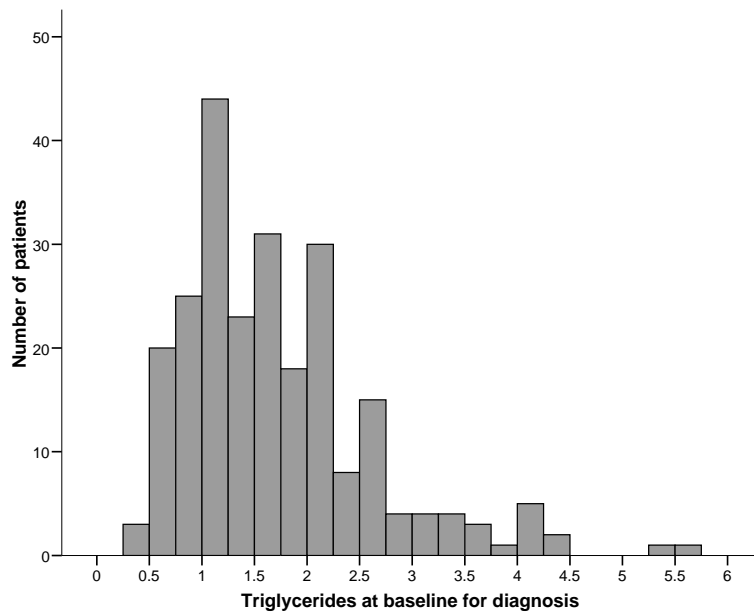
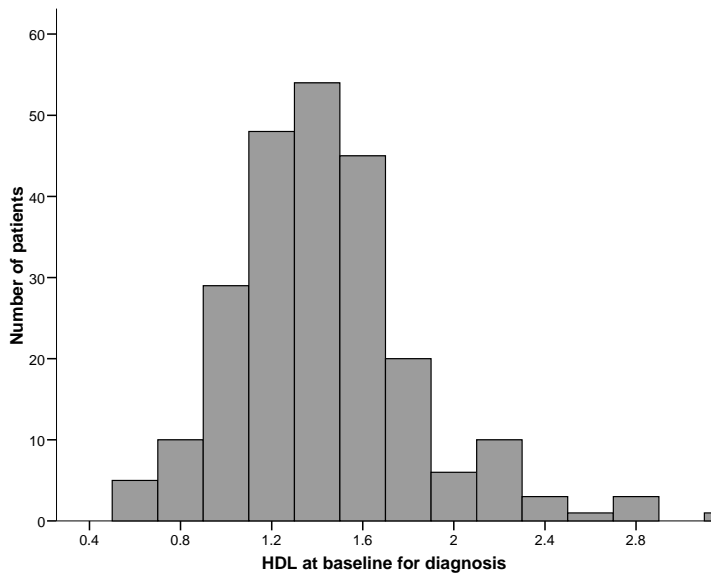
1.10 What were the lipoprotein measurements used to make the diagnosis?

Pre-treatment levels in adults and children

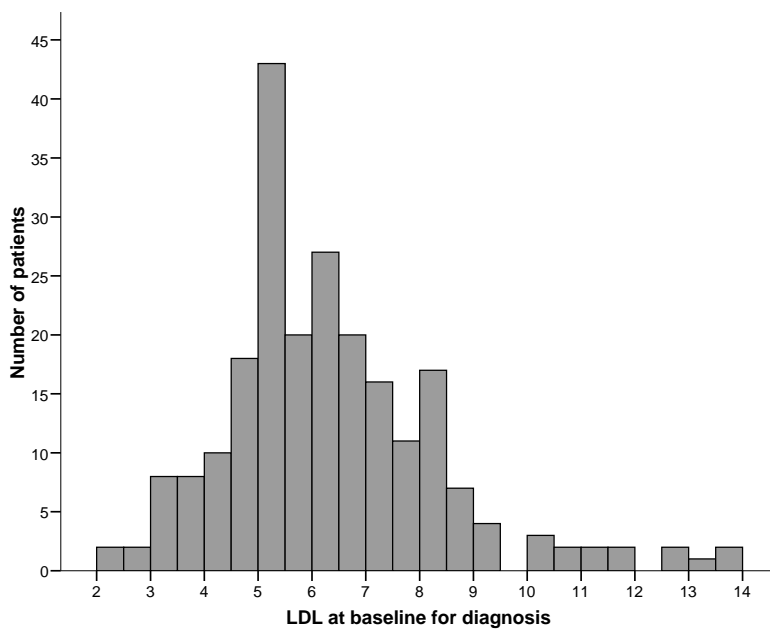
| Adults (n=252) | | | | | |
|---|-----------|---------------------------------------|--------------------------------------|--------------------------------------|---------------------------------------|
| Date | Fasting | Cholesterol | HDL | Trig | LDL |
| median 2004 IQR 1997-2007 range 1978-2009 | 233 (92%) | n=249 median 8.4 IQR 7.5 - 10.0 | n=217 median 1.3 IQR 1.1 - 1.6 | n=221 median 1.6 IQR 1.1 - 2.1 | n=209 median 6.1 IQR 5.1 - 7.15 |
| Children (n=22) | | | | | |
| median 2007 IQR 2005-2008 range 1999-2009 | 9 (41%) | n=22 median 7.0 IQR 6.2 - 7.5 | n=18 median 1.4 IQR 1.2 - 1.5 | n=21 median 1.1 IQR 0.7 - 1.6 | n=18 median 5.2 IQR 4.3 - 5.8 |



The levels below the Simon Broome 7.5mmol/l cut off for adults in a small proportion of subjects suggests that some of these patients were treated in general practice prior to referral to the specialist centre where the baseline levels were recorded. The same applies to the LDL-C levels shown below



The high Tg levels (>4.5mmol/l) in a small proportion of patients suggests that these subjects may possibly have familial combined hyperlipidaemia rather than FH, particularly as triglycerides are also raised on therapy (see data on page 42).



1.11 Is there evidence in the notes that a baseline ECG was considered?

| | Yes | No | Not applicable |
|-----------------|-----------|-----------|----------------|
| Children (n=22) | 6 (27%) | 10 (45%) | 6 (27%) |
| Adults (n=252) | 130 (52%) | 122 (48%) | 0 (0%) |

All the “not applicables” were children, but not all children had “not applicable” entered against this question. If we consider yes/no answers for those aged 18 and over, then a baseline ECG was considered in 124/226 (55%)

Sites answered question 1.12 if they answered “yes” to question 1.11.

1.12 Is there evidence in the notes that an ECG was carried out?

Yes: 121/130 (93%)

Note: a further 11 patients had data suggesting that though no baseline ECG had been considered, one had been conducted (free-text comments suggest that this may have been done for other clinical reasons and then used at baseline).

Section 2: Pedigree

NICE GUIDELINE RECOMMENDATION

When considering a diagnosis of FH, healthcare professionals with expertise in FH should use standardised pedigree terminology to document, when possible, at least a three-generation pedigree. This should include relatives’ age of onset of coronary heart disease, lipid concentrations and smoking history. For deceased relatives, the age and cause of death, and smoking history should be documented. If possible, the index individual should verify this information with other family members.

Almost 90% of the notes contained a pedigree or family tree, with the majority being written or drawn by hand. 97% have information about the first generation, but as expected this number fell off slightly in the second and markedly to the third generation. The frequency of included information about the relatives age, age of onset of CHD, smoking history etc, was quite varied and there is obvious room for improvement in the way these data are collected.

Pedigree

2.1 Is there a pedigree/ family tree in the notes?

Yes: 220 (89%)

Answer 2.2 if answered yes to 2.1

2.2 How is the pedigree / family tree recorded?

- a. Drawn using software 28/220 (13%)
- b. Drawn by hand 110/220 (50%)
- c. Written 137/220 (62%)

2.3 If yes, does the pedigree map the following family members:

- a. First generation 214/220 (97%)
- b. Second generation 162/220 (74%)
- c. Third generation 104/220 (47%)

Sites who answered "yes" to 2.2 (a) (b) or (c) were asked to answer question 2.3

Data items recorded in pedigree

If the patient has advised that they did not know the relatives age and/or cause of death, and the notes captured this, then the answer would be 'Yes'.

Sites were asked to include information about half siblings but not for adopted siblings.

2.4 Are the following data items recorded:

a. Relatives age

- Yes - for all 48/220 (22%)
- Yes - for some 159/220 (72%)
- No 13/220 (6%)

b. Age of onset of coronary heart disease (if present)?

- Yes - for all 59/220 (27%)
- Yes - for some 112/220 (51%)
- No 49/220 (22%)

c. Information about tendon xanthomas?

- Yes - for all 3/220 (1%)
- Yes - for some 13/220 (6%)
- No 204/220 (93%)

d. Information about total cholesterol?

- Yes - for all 18/220 (8%)

Yes - for some 130/220 (59%)
 No 72/220 (33%)

e. Smoking history of blood relatives with CHD?

Yes - for all 6/220 (3%)
 Yes - for some 18/220 (8%)
 No 196/220 (89%)

f. (i) Are there any deceased blood relatives?

Yes: 169/220 (77%)

If answered yes to f(i):

(ii) Please indicate which blood relatives have died?

| | |
|--------|---------------|
| Mother | 77/220 (35%) |
| Father | 104/220 (47%) |

| Number of 1 st generation siblings | Number of patients |
|---|--------------------|
| 1 | 25 |
| 2 | 6 |
| 3 | 2 |
| 4 | 2 |
| 6 | 2 |
| 8 | 2 |

| | |
|------------------------|--------------|
| Grandmother (maternal) | 29/220 (13%) |
| Grandmother (paternal) | 25/220 (11%) |
| Grandfather (maternal) | 33/220 (15%) |
| Grandfather (paternal) | 23/220 (9%) |

| Number of 2 nd generation siblings | Number of patients |
|---|--------------------|
| 1 | 15 |
| 2 | 5 |
| 3 | 2 |
| 4 | 1 |
| 5 | 1 |
| 8 | 1 |

g. If yes to (f), is the age and cause of death recorded?

| | Age of death recorded | Cause of death recorded |
|-----------|-----------------------|-------------------------|
| Mother | 71/77 | 63/77 |
| Father | 98/104 | 95/104 |
| Sibling 1 | 28/39 | 31/39 |
| Sibling 2 | 7/14 | 7/14 |
| Sibling 3 | 2/8 | 2/8 |
| Sibling 4 | 1/6 | 1/6 |

| | | |
|-----------|-----|-----|
| Sibling 5 | 1/4 | 1/4 |
| Sibling 6 | 1/4 | 1/4 |
| Sibling 7 | 0/2 | 0/2 |
| Sibling 8 | 0/2 | 0/2 |

| | Age of death recorded* | Cause of death recorded* |
|------------------------|------------------------|--------------------------|
| Grandmother (maternal) | 10/29 | 11/29 |
| Grandmother (paternal) | 10/25 | 9/25 |
| Grandfather (maternal) | 14/33 | 14/33 |
| Grandfather (paternal) | 8/23 | 8/23 |
| Sibling 1 | 20/25 | 22/25 |
| Sibling 2 | 5/10 | 6/10 |
| Sibling 3 | 2/5 | 2/5 |
| Sibling 4 | 0/3 | 0/3 |
| Sibling 5 | 0/2 | 0/2 |
| Sibling 6 | 0/1 | 0/1 |
| Sibling 7 | 0/1 | 0/1 |
| Sibling 8 | 0/1 | 0/1 |

Section 3: Assessment

75% of the patients were reviewed by the consultant at their most recent clinic appointment. Height and weight was recorded in the vast majority of individuals' notes.

3.1 Who reviewed the patient at their last (most recent) out-patient clinic appointment?

| | |
|----------------------------|-----------|
| Consultant | 183 (74%) |
| Specialist Registrar (SpR) | 38 (15%) |
| Specialist Nurse | 12 (5%) |
| Other (Please list) | 15 (7%) |

Height and weight

For Adults only

3.2 At the first clinic appointment was the patient's weight recorded?

Yes: 214/231 (94%)

3.3 At the first clinic appointment was the patient's height recorded?

Yes: 204/231 (88%)

3.4 At the last (most recent) clinic appointment was the patient's weight recorded?

Yes: 201/231 (87%)

Question 3.5 applied to paediatric cases only

3.5 At the first clinic appointment was the patient's height and weight recorded?

Yes: 16/17 (94%)

3.6 At the last (most recent) clinic appointment was the patient's height and weight recorded?

Yes: 16/17 (94%)

Section 4: Treatment

NICE GUIDELINE RECOMMENDATION

Healthcare professionals should consider prescribing a high-intensity statin to achieve a recommended reduction in LDL-C concentration of greater than 50% from baseline (that is, LDL-C concentration from treatment).

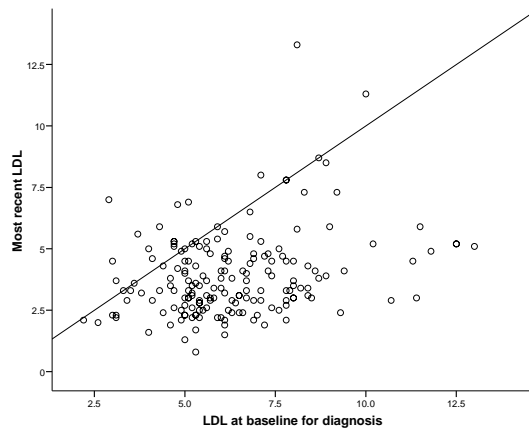
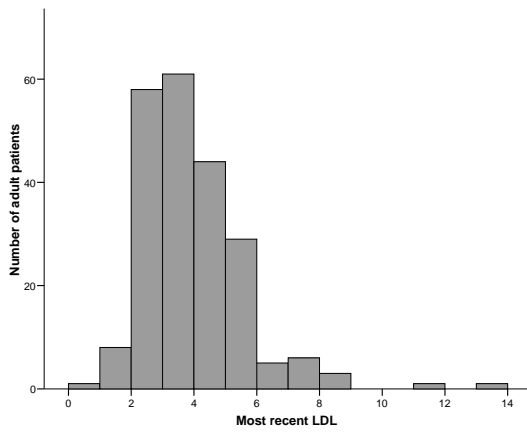
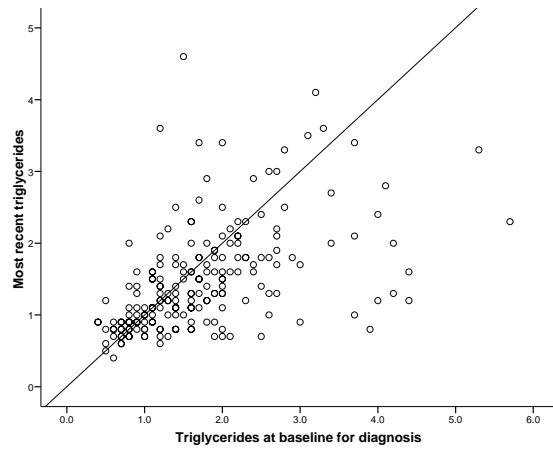
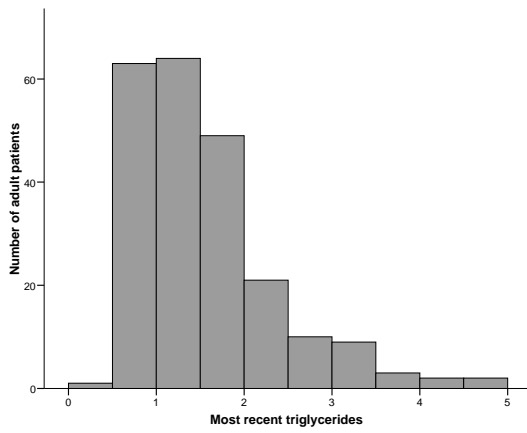
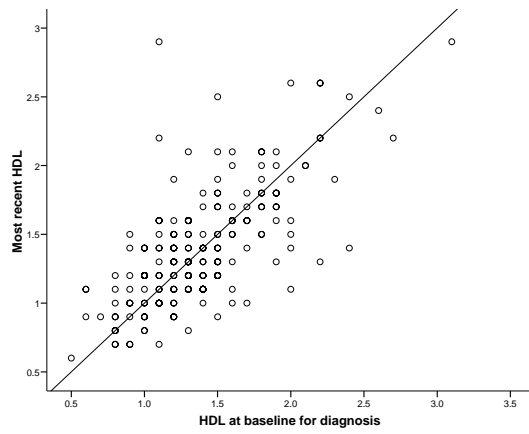
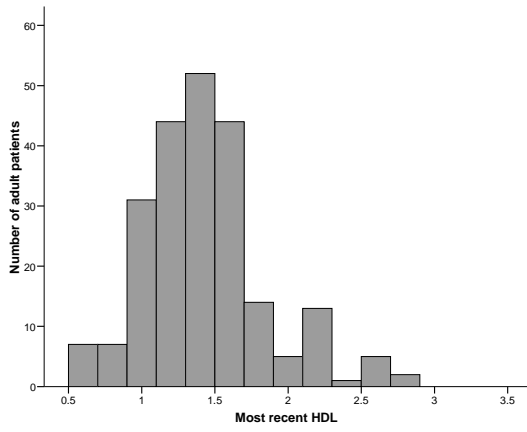
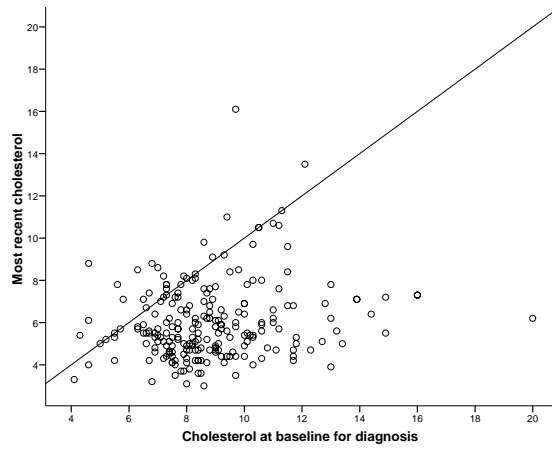
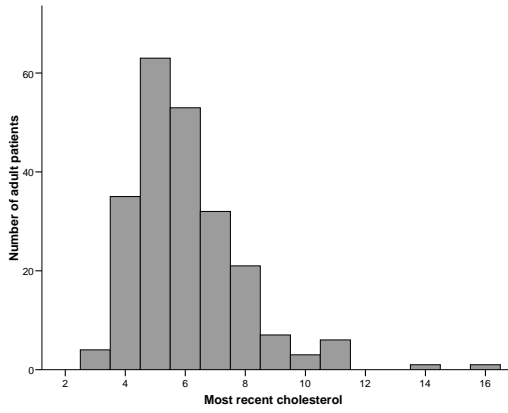
In the adults, the treated lipid levels all show a significant reduction from the untreated levels, with LDL down to 3.5mmol/l from a median of 6.1mmol/l (the mean reduction for individual patients was 33%, median 39%, IQR 16% - 56%). This does fall short of the target 50% reduction recommended by NICE, although since this is only the third clinic visit for some subjects some improvement may still be achieved. In these data, 64/192 (33%) of the patients achieved the 50% target. The most common statin treatments are Atorvastatin and Rosuvastatin (35% each) and the reasons for not taking statins appear to be clearly stated. 48% of patients are also on Ezetimibe, with only 6% also on fibrates.

Lipoprotein results

4.1 What are the most recent lipoprotein results?

| Adults (n=252) | | | | | |
|-------------------|-------------|---------------|---------------|---------------|---------------|
| Date | Fasting | Cholesterol | HDL | Trig | LDL |
| 229 (90%) in 2009 | 251 (99.6%) | n=252 | n=251 | n=250 | n=243 |
| 20 (9%) in 2008 | | median 5.6 | median 1.4 | median 1.3 | median 3.5 |
| Remainder earlier | | IQR 4.8 - 6.9 | IQR 1.1 - 1.7 | IQR 0.9 - 1.8 | IQR 2.8 - 4.7 |
| Children (n=22) | | | | | |
| 15 in 2009 | 8 (36%) | n=22 | n=17 | n=17 | n=17 |
| 6 in 2008 | | median 6.1 | median 1.3 | median 1.0 | median 4.4 |
| 1 in 2006. | | IQR 5.2 - 6.8 | IQR 1.1 - 1.6 | IQR 0.9 - 1.4 | IQR 3.6 - 5.0 |

The graphs below show the most recent lipoprotein results on the left, and then these compared to the patient's "baseline for diagnosis" results on the right. The diagonal line in each case indicates equality, so dots above the line are patients with higher recent results than baseline, and those below the line have lower recent results than baseline.



Lipid lowering drug therapy

4.2 What is the patient's current lipid lowering drug therapy (i.e. as recorded at the most recent out-patient clinic appointment)?

- a. Any Statin: 199 (80%)
 - Atorvastatin: 86 (35%)
Doses: 10 (13 patients), 20 (14), 40 (20), 60 (3), 80 (36)
 - Fluvastatin: 1 (0.4%)
Dose : 80
 - Pravastatin: 7 (3%)
Doses : 10 (2), 20 (1), 40 (4)
 - Rosuvastatin: 86 (35%)
Doses: 5 (8), 10 (22), 20 (24), 30 (2), 40 (30)
 - Simvastatin: 19 (8%)
Doses: 20 (2), 40 (14), 80 (3)

b. (if applicable) Please indicate the reason(s) that this patient is not receiving statin. Sites were able to select more than one answer option.

| | |
|---|-------------|
| Patient intolerant to statin | 23/49 (47%) |
| Patient declined | 4/49 (8%) |
| Patient planning to become or is pregnant/ is breastfeeding | 2/49 (4%) |
| Child under 10 years | 7/49 (14%) |
| Child 10 and over of but risk low | 1/49 (0.4%) |
| Other | 16/49 (33%) |

- c. Resin 3 (1%)
Doses: 4 (1 patient), 8 (2)
- d. Ezetimibe 119 (48%)
Date started: median October 2006, IQR September 2004 - March 2008
- e. Fenofibrate 12 (5%)
Doses: 160 (8), 267 (4)
- f. Bezafibrate 2 (1%)
Doses: 200 (1), 400 (1)
- g. Fish oil / Ω fatty acids 13 (5%)
- h. Nicotinic acid or derivative 3 (1%)
- i. plasma LDL-C apheresis 1 (0.4%)
- j. Other 33 (13%)

Paediatrics only:

k. If the patient is receiving lipid lowering treatment what date was it started?

Median January 2008, IQR May 2007 - March 2009

Adults and Paediatrics:

4.3 Is there evidence that the following were measured before initiation of statin treatment:

- a. Muscle enzyme (CK) 157/199 (79%)
- b. Transaminases (ALT or AST) 180/199 (90%)

Paediatrics only:

c. Was this repeated within 3 months of starting statin treatment?

Yes: 2/7 (29%)

Section 5: Advice

NICE GUIDELINE RECOMMENDATION

People with FH should be given advice on diet, physical activity, weight management, smoking and alcohol consumption (see NICE guideline for full recommendations)

When lipid-modifying drug therapy is first considered for women and girls, the risks for future pregnancy and the fetus while taking lipid-modifying drug therapy should be discussed. This discussion should be revisited at least annually.

Healthcare professionals should give women and girls with FH specific information tailored to their needs and should offer a choice of effective contraceptive methods.

The written advice to FH patients appears in the majority of patients to contain information about diet, exercise, smoking and alcohol consumption, with smoking status recorded in 90% of notes. Since the majority of the female patients were post-menopausal, the notes do not document discussion of contraceptive issues while on statins.

In 45% of female patients of reproductive age the need for contraception whilst taking statins was not documented.

This section covers adults and children/young persons.

Life-style advice

5.1 Is there written evidence that the patient/carer was given the following life-style advice?

- a. Diet 212 (85%)
- b. Exercise 180 (73%)
- c. Smoking (*Question applies whether smoker or not*) 165 (67%)
- d. Alcohol 147 (59%)

Smoking status

5.2 Is the smoking status of the patient recorded? 224 (90%)

a. If yes, please select the status:

Smoker 35/224 (16%)

Ex smoker 51/224 (23%)
Non smoker 138/224 (62%)

Smoking cessation advice/referral

b. If yes, were they given advice about or offered a referral to an intensive support service/smoking cessation?

Offered referral 15/35 (43%)
Given written advice 10/35 (29%)
No referral or advice 10/35 (29%)

Contraception

Question 5.3 applies to females of reproduction age who have received, or are under consideration for receiving, lipid modifying drug therapy.

Lipid-modifying therapy should only be started after a discussion with the patient about the risks for future pregnancy and the foetus.

5.3 Was the need for contraception whilst taking statins discussed and documented?

Yes 34/139 (24%)
No 28/139 (20%)
Patient not of reproductive age 77/139 (55%)

Section 6: Cascade Testing

NICE GUIDELINE RECOMMENDATION

Cascade testing using a combination of DNA testing and LDL-C concentration measurement is recommended to identify affected relatives of those index individuals with a clinical diagnosis of FH. This should include at least the first- and second- and, when possible, third-degree biological relatives.

In individuals where DNA testing is being carried out a mutation was detected in 60%; this is encouraging and suggests that majority of test requested are in DFH patients. While cholesterol measures have been made in approximately 24% of (living) parents, approximately 40% of brothers and sisters and approximately 45% of children of an index case, cascade testing to other more distant relatives and to those outside the catchment are is not systematically being carried out in any of the sites.

This section applies to Adults only

Mutation testing

6.1 Is there evidence that the mutation has been tested? Yes: 63/231 (27%)

a. If yes, was the mutation identified?

Yes 23/63 (37%)
Result not yet reported 25/63 (40%)

No

15/63 (24%)

Cascade testing

6.2 Is there evidence that the need for cascade testing (testing of relatives cholesterol) has been discussed with the patient?

Yes: 193/231 (84%)

6.3 Has the process of cascade testing been initiated?

Yes: 147/193 (76%)

Sites who answered "yes" to question 6.3 answered 6.4

6.4 Please detail cascade testing undertaken in parents:

| | Relative within catchment area? | Relative contacted | Full lipid undertaken | DNA testing undertaken |
|--------|---------------------------------|---|-----------------------|------------------------|
| Mother | 82/231 (35%) | <ul style="list-style-type: none">• Yes: 40/231 (17%)• Of those in catchment area: 38/82 (46%) If not: <ul style="list-style-type: none">• No consent to contact: 12/190• Contact details unknown 31/190• Deceased 74/190• Other 73/190 | 29/40 (73%) | 1/40 (3%) |
| Father | 69/231 (30%) | <ul style="list-style-type: none">• Yes: 22/231 (10%)• Of those in catchment area: 21/69 (30%) If not: <ul style="list-style-type: none">• no consent to contact relative: 11/209• Contact details unknown 35/209• Deceased 102/209• Other 61/209 | 17/22 (77%) | 3/22 (14%) |

a. How many children does the patient have?

141/231 (61%) report some children

32 have 1, 80 have 2, 19 have 3, 10 have more than 3

- b. How many brothers/sisters does the patient have?
 162/222 (73%) report some siblings
 9 adults are missing data for this question
 74 have 1, 35 have 2, 21 have 3, 32 have more than 3

Please provide details of cascade testing in children:

| | Relative within catchment area? | Relative contacted | Full lipid undertaken | DNA testing undertaken |
|---------|---------------------------------|---|-----------------------|------------------------|
| Child 1 | 99/132 (75%) | <ul style="list-style-type: none"> • Yes: 65/141 (46%) • Of those in catchment area: 60/99 (61%) If not: <ul style="list-style-type: none"> • No consent to contact: 10/76 • Contact details unknown 2/76 • Deceased 1/76 • Other 63/76 | 47/64 (73%) | 9/64 (14%) |
| Child 2 | 71/101 (70%) | <ul style="list-style-type: none"> • Yes: 51/109 (47%) • Of those in catchment area: 45/71 (63%) If not: <ul style="list-style-type: none"> • no consent to contact relative: 5/58 • Contact details unknown 1/58 • Deceased 1/58 • Other 51/58 | 30/51 (59%) | 4/50 (8%) |
| Child 3 | 17/23 (74%) | <ul style="list-style-type: none"> • Yes: 13/29 (45%) • Of those in catchment area: 10/17 (59%) If not: <ul style="list-style-type: none"> • No consent to contact: 1/16 • Contact details unknown 0/16 • Deceased 0/16 • Other 15/16 | 8/13 | 1/13 |

Subsequent children are too few to provide reliable estimates of proportions.
 A small number of miss data are excluded.

Please provide details of cascade testing in brother(s)/sister(s)

| | Relative within catchment area? | Relative contacted | Full lipid undertaken | DNA testing undertaken |
|-----------|---------------------------------|--|-----------------------|------------------------|
| Sibling 1 | 66/146 (45%) | <ul style="list-style-type: none"> • Yes: 61/157 (39%) • Of those in catchment area: 44/66 (67%) If not: <ul style="list-style-type: none"> • No consent to contact: 9/96 • Contact details unknown 11/96 • Deceased 19/96 • Other 57/96 | 48/59 (81%) | 5/59 (8%) |
| Sibling 2 | 37/77 (48%) | <ul style="list-style-type: none"> • Yes: 34/86 (40%) • Of those in catchment area: 23/37 (62%) If not: | 24/32 (75%) | 3/32 (9%) |

| | | | | |
|-----------|--|---|------------|-----------|
| | | <ul style="list-style-type: none"> • No consent to contact: 5/52 • Contact details unknown 6/52 • Deceased 5/52 • Other 36/52 | | |
| Sibling 3 | 20/46 (43%) (5 patients have missing data) | <ul style="list-style-type: none"> • Yes: 16/51 (31%) • Of those in catchment area: 12/20 (60%) If not: <ul style="list-style-type: none"> • No consent to contact: 6/35 • Contact details unknown 5/35 • Deceased 3/35 • Other 21/35 | 9/13 (69%) | 0/13 (0%) |

Subsequent siblings are too few to provide reliable estimates of proportions. A small number of miss data are excluded.

Second and third degree biological relatives

6.5 Is there evidence that the patient was asked if second & third degree biological relatives have been tested?

Yes: 101/193 (52%)

Contact with GP

6.6 Has a letter been written to the index case's GP stating the diagnosis of FH has been made? Question 6.6 was only answered if question 6.1a is answered as 'Yes'.

Yes: 23/23 (100%)

a. Does the letter emphasize the need for referral and testing of at-risk relatives?

Yes: 20/23 (87%)

Section 7: (Annual) Review

NICE GUIDELINE RECOMMENDATIONS

All people with FH should be offered a regular structured review that is carried out at least annually.

A baseline electrocardiogram (ECG) should be considered for adults with FH.

Structured review should include assessment of any symptoms of coronary heart disease and smoking status, a fasting lipid profile, and discussion about concordance with medication, possible side effects of treatment the patient may be experiencing, and any changes in lifestyle or lipid-modifying drug therapy that may be required to achieve the recommended LDL-C concentration.

Currently the vast majority of FH patients are having annual review in secondary care, with only 5% in primary care. In general, the notes indicate good achievement of the NICE targets with regard to questions addressed in the annual review.

Structure annual review

7.1 Has the patient had a structured review carried out at least annually since the initial diagnosis?

| | |
|-------------------------------------|-----------|
| Yes - in secondary care | 227 (92%) |
| Yes - in secondary and primary care | 12 (5%) |
| Yes - in primary care | 0 (0%) |
| No - patient offered and declined | 1 (0.4%) |
| No - not offered | 8 (3%) |

b. If undertaken in secondary care, please give the dates of the last 3 reviews (if 3 have been carried out):

Date 1: 83% of patients have had a review in January - April 2009.

Date 2: 87% have had a review since January 2008. Median June 2008.

Date 3: 86% have had a review since January 2007. Median September 2007.

7.2 If review undertaken in secondary care, is there evidence in the notes that the review covered the following issues:

a. Response to drug(s)?

Yes: 193/215 (90%)

b. Side effects?

Yes: 191/215 (89%)

c. CHD status?

Yes: 148/215 (69%)

d. Fertility issues (females only)?

Not applicable: 72/115

Yes: 21/43 (49%)

No: 22/43 (51%)

e. Lifestyle?

Yes: 174/215 (81%)

f. ECG?

Not applicable: 51/215

Yes: 76/164 (46%)

No: 88/164 (54%)

g. Progress with cascade screening?

Yes: 120/215 (56%)

Section 8: Children

NICE GUIDELINE RECOMMENDATION

In children at risk of FH because of one affected parent, the following diagnostic tests should be carried out by the age of 10 years or at the earliest opportunity thereafter.

- *A DNA test if the family mutation is known*
- *LDL-C concentration measurement if the family mutation is not known. When excluding a diagnosis of FH a further LDL-C measurement should be repeated after puberty because LDL-C concentrations change during puberty.*

Although the data from the one Paediatric site suggests good management of children, the number of cases with information on children with FH is too small in this survey for any strong conclusions to be drawn.

This section applies to patients who are under 16 years old.

8.1 Is there evidence that the mutation has been tested ?

Yes: 5/17

a. If yes, was the mutation identified?

Yes 1/5

Result not yet reported 4/5

8.2 If the family mutation is known (*yes to 8.1(b)*) has the child been offered a DNA test?

Yes 1/1

Month and year when offered: November 2006

8.3 If the family mutation is not known has the LDL-C concentration been measured?

0/0 - no data

8.4 If the family mutation is not known (*answer to 8.1(a)*) either result not reported or no) and the initial LDL-C concentration suggested not-FH has the LDL-C level been re measured after puberty?

0/0 - no data

Appendix 1: Audit proformas - organisational and clinical

Royal College of Physicians

National Audit of Familial Hypercholesterolaemia (Pilot Project)

ORGANISATIONAL AUDIT PROFORMA

Please enter the name of each person completing questionnaire including job role/title & contact details.

General information

Name
Job Role/Title
Work Address
Email address
Telephone number

Name
Job Role/Title
Work Address
Email address
Telephone number

Name
Job Role/Title
Work Address
Email address
Telephone number

Section 1: Set up

1.1 What type of hospitals are in your Trust? *Tick all that apply*

Teaching Hospital
District General Hospital

1.2 Does your Trust provide out-patient services for the clinical management of:

| | Yes | No |
|---|--------------------------|--------------------------|
| a. Adults with FH? | <input type="checkbox"/> | <input type="checkbox"/> |
| b. Children/young people (under 16s) with FH? | <input type="checkbox"/> | <input type="checkbox"/> |

If responded positively to 1.2 a:

1.3 *Who* provides the out-patient services for the clinical management of adults with heterozygous FH? *Tick one option*

- Provided by Trust
- Provided by a visiting service from another Trust
- By referring to another service or centre

If responded positively to 1.2 a:

1.4 *Who* provides the out-patient services for the clinical management of adults with homozygous FH? *Tick one option*

- Provided by Trust
- Provided by a visiting service from another Trust
- By referring to another service or centre

If responded positively to 1.2 b:

1.5 *Who* provides the out-patient services for the clinical management of children with FH? *Tick one option*

- Provided by Trust
- Provided by a visiting service from another Trust
- By referring to another service or centre

1.6 What Directorate/ Care Group is FH under? *Tick all that apply*

- Diabetes and Endocrinology
- Cardiology / Cardiovascular
- Chemical Pathology
- General Medicine / General Internal
- Specialist Medicine
- Paediatrics
- Other Please specify

1.7 Does your Trust have someone with operational managerial responsibility for FH services?

- Yes
- No

1.8 Does your Trust have a lead clinician responsible for FH care?

- Yes
- No

1.9 How many of the following out-patient clinics are there per month:

| | Adults - morning clinical (am) | Adults - afternoon clinics (pm) | Paediatric clinics (am) | Paediatric clinics (pm) |
|---|--------------------------------|---------------------------------|-------------------------|-------------------------|
| Dedicated FH clinics | | | | |
| General lipid clinics | | | | |
| Other medical clinics (where FH patients are seen in relation to FH) | | | | |

1.10 Does your Trust have a designated clinic to test for FH by cascade testing?

| | | |
|--------------------------|--------------------------|--------------------------|
| Yes | In development | No |
| <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> |

1.11 Please estimate how many patients with FH were seen in out-patient clinics in the last year (this should include definite, probable/possible):

| | |
|--------|-------------|
| Adults | Paediatrics |
|--------|-------------|

a. New

b. Follow up

1.12 Does your Trust have specialist services for the management of young people with FH?

| | |
|--------------------------|--------------------------|
| Yes | No |
| <input type="checkbox"/> | <input type="checkbox"/> |

Answer 1.12 if yes to answer a & b

1.12 a. do your specialist services have access to paediatric phlebotomy?

| | |
|--------------------------|--------------------------|
| Yes | No |
| <input type="checkbox"/> | <input type="checkbox"/> |

c. Do you have family clinics?

| | |
|--------------------------|--------------------------|
| Yes | No |
| <input type="checkbox"/> | <input type="checkbox"/> |

1.13 Do you have shared care arrangements in place with:?

| | | | |
|---------------------|--------------------------|--------------------------|--------------------------|
| a. General practice | Yes - formal | Yes - informal | No |
| | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> |

| | | | |
|---------------|--------------------------|--------------------------|--------------------------|
| b. Cardiology | Yes - formal | Yes - informal | No |
| | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> |

| | | | |
|---------------|--------------------------|--------------------------|--------------------------|
| c. Obstetrics | Yes - formal | Yes - informal | No |
| | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> |

| | | | |
|----------------|--------------------------|--------------------------|--------------------------|
| d. Paediatrics | Yes - formal | Yes - informal | No |
| | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> |

| | | | |
|---------------------------|--------------------------|--------------------------|--------------------------|
| e. LDL Apheresis services | Yes - formal | Yes - informal | No |
| | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> |

1.14 Do you have a paper database of patients with FH? *Tick one of following*

| | |
|-------------------------------------|--------------------------|
| Paper database - complete | <input type="checkbox"/> |
| Paper database - partially complete | <input type="checkbox"/> |

No

1.15 Do you have an electronic database of patients with FH? *Tick one of following*

Electronic database - complete
Electronic database - partially complete
No

Section 2: Clinical time

2.1 In your Trust, how much clinical time is devoted to specialist lipid management/FH by your employed staff?

- a. How many Consultant's programmed activities (PAs) are devoted to specialist lipid management?
- b. On average, what proportion (%) of this is estimated to be spent on the management of FH?

<10% 11-20% 21-40% 41-60% 61-80% 81-100%

- c. How many Lipid specialist nurses are there (WTE)?

- d. On average, what proportion (%) of this is estimated to be spent on the management of FH?

<10% 11-20% 21-40% 41-60% 61-80% 81-100%

Section 3: Management of FH

3.1 Does your Trust formally classify patients according to the Simon Broome criteria?

Yes No

3.2 Does your Trust routinely offer FH patients an annual review?

Yes No

3.3 Does your Trust routinely discharge FH patients once lipid levels are optimised?

Yes No

3.4 Is there a written policy/protocol for the clinical management of FH?

Yes No In development

If answer to 3.4 is yes, answer a.-d.

- a. Has this been formerly approved by your Trust?

Yes In progress No

b. Does the policy include diagnostic criteria for FH?

Yes No

c. Does the policy include advice on cascade testing?

Yes No

d. Does the policy include advice on annual review?

Yes No

3.5 Is there an integrated care pathway/ clinical management strategy for the management of FH?

Yes No In development

Section 4: Cascade Testing

4.1 Is there access to a family cascade testing system for FH in your Trust?

Yes No In development

4.2 If yes, how is this provided?

Provided by Trust
Provide by visiting service from another Trust
By referring to another service or centre

4.3 If yes, who is responsible for the clinical governance of the cascade testing for families?

Trust
Regional Clinical Genetics Service
Other Please specify

4.4 If provided by Trust, does this include:

a. Dedicated staff?

Yes No In development

b. Formal input from the local Genetic services?

Yes No In development

4.5 Do the staff who are looking after individuals and families with FH routinely have access to:

a. dedicated IT software

Yes No In development

b. pedigree drawing software?

Yes No In development

Section 5: Trust services

5.1. Does your Trust have arrangements for DNA mutation testing for FH patients attending your clinical service?

Yes - funded
Yes - not funded
Available only in special cases
No

5.2 Does your Trust have a laboratory that is currently accredited (CPA) for testing lipid measurements?

Yes No

5.3 Does your Trust have routine access to HDL measurements?

Yes No

5.4 Is there a smoking cessation support service that FH patients can be referred to?

Yes No

5.5 Is there access to specialist dietary advice?

Yes - in clinic Yes - separate from clinic No

Section 6: Patient information

6.1 Which, if any, of the following information leaflets do you provide to FH patients/carers?

a. British Heart Foundation leaflet(s) Yes No

b. Heart UK leaflet(s)

c. NICE information for the public

- d. Trust leaflet
- e. Other Please specify

6.2 If yes, does the information provided cover the following subject areas?

- | | | |
|---------------------------|--------------------------|--------------------------|
| a. Diet | Yes | No |
| | <input type="checkbox"/> | <input type="checkbox"/> |
| b. Exercise | Yes | No |
| | <input type="checkbox"/> | <input type="checkbox"/> |
| c. Smoking | Yes | No |
| | <input type="checkbox"/> | <input type="checkbox"/> |
| d. Alcohol | Yes | No |
| | <input type="checkbox"/> | <input type="checkbox"/> |
| e. FH as a condition | Yes | No |
| | <input type="checkbox"/> | <input type="checkbox"/> |
| f. Inheritance | Yes | No |
| | <input type="checkbox"/> | <input type="checkbox"/> |
| g. Role of DNA testing | Yes | No |
| | <input type="checkbox"/> | <input type="checkbox"/> |
| h. Treatment options | Yes | No |
| | <input type="checkbox"/> | <input type="checkbox"/> |
| i. Treatment for children | Yes | No |
| | <input type="checkbox"/> | <input type="checkbox"/> |
| j. Monitoring | Yes | No |
| | <input type="checkbox"/> | <input type="checkbox"/> |
| k. DNA Testing | Yes | No |
| | <input type="checkbox"/> | <input type="checkbox"/> |
| l. Cascade Testing | Yes | No |
| | <input type="checkbox"/> | <input type="checkbox"/> |

Section 7: Service improvement

7.1 Does the FH service have specific links to a patient/service-user group?

- | | |
|--------------------------|--------------------------|
| Yes | No |
| <input type="checkbox"/> | <input type="checkbox"/> |

If answered yes to 7.1

7.2 Is the patient/service user group involved in planning and delivery of FH services?

- | | | |
|--------------------------|--------------------------|--------------------------|
| Yes | No | In development |
| <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> |

7.3 Is FH care delivered by the service subject to regular audit?

- | | |
|--------------------------|--------------------------|
| Yes | No |
| <input type="checkbox"/> | <input type="checkbox"/> |

National Audit of Familial Hypercholesterolaemia (Pilot project)

CLINICAL AUDIT PROFORMA

The patient should be selected from the first 40 consecutive cases with a diagnosis of Familial Hypercholesterolaemia (ICD 10 Code E78. 0) visiting a clinic with expertise in lipid disorders for the third time or more between 12 January - 3 April 2009. See accompanying guidance notes for full methodology and data definitions.

Section 0: General information

0.0 Patient ID: [[Automatically generated](#)]

0.1 Auditor Speciality:

- | | | |
|------------------------------|--------------------------|---------------------------|
| Chemical Pathology | <input type="checkbox"/> | |
| Clinical Pharmacology | <input type="checkbox"/> | |
| Diabetology | <input type="checkbox"/> | |
| Endocrinology | <input type="checkbox"/> | |
| Paediatrics | <input type="checkbox"/> | |
| Specialist Lipid | <input type="checkbox"/> | |
| Clinical Effectiveness/Audit | <input type="checkbox"/> | |
| Other | <input type="checkbox"/> | Please specify discipline |

a. Auditor Grade:

- | | |
|----------------------|--------------------------|
| Consultant | <input type="checkbox"/> |
| Specialist Registrar | <input type="checkbox"/> |
| Specialist Nurse | <input type="checkbox"/> |
| Nurse | <input type="checkbox"/> |
| Other | <input type="checkbox"/> |

0.2 In what clinic was the patient seen?

- | | |
|---------------------------------------|--------------------------|
| Familial hypercholesterolaemia clinic | <input type="checkbox"/> |
| Lipid Clinic | <input type="checkbox"/> |
| General Medicine clinic | <input type="checkbox"/> |
| Paediatric clinic | <input type="checkbox"/> |

0.3 Date of clinic

0.4 Patient's age at clinic years months

0.5 Patient's gender ? Male / Female

0.6 Ethnic group *tick appropriate box*

| | | |
|-------------------------------|----------------------------|--------------------------|
| White | British | <input type="checkbox"/> |
| | Irish | <input type="checkbox"/> |
| | Any other White background | <input type="checkbox"/> |
| Asian or Asian British | Indian | <input type="checkbox"/> |
| | Pakistani | <input type="checkbox"/> |
| | Bangladeshi | <input type="checkbox"/> |
| | Any other Asian background | <input type="checkbox"/> |
| Black or Black British | Caribbean | <input type="checkbox"/> |
| | African | <input type="checkbox"/> |
| | Any other Black background | <input type="checkbox"/> |
| Mixed | White and Black Caribbean | <input type="checkbox"/> |
| | White and Black African | <input type="checkbox"/> |
| | White and Asian | <input type="checkbox"/> |
| | Any other mixed background | <input type="checkbox"/> |
| Chinese / Other ethnic groups | Chinese | <input type="checkbox"/> |
| | Any other ethnic group | <input type="checkbox"/> |
| Not recorded | <input type="checkbox"/> | |

0.7 Is this a reliability case (validation check)? Yes No

Only answer question 0.8 if answer to 0.7 is "yes".

0.8 Patient Number (reliability case)

Section 1: Diagnosis

1.1 What is the patient's diagnosis? (*Tick one option*)

- Definite Homozygous FH
- Definite Heterozygous FH
- Possible FH
- FH not clearly stated as definite/possible

1.2 Year of diagnosis (FH possible or definite)? Not recorded

1.3 Is there evidence in the notes that there is a history of CHD in the patient?

- Yes
- No

1.4 Is there evidence in the notes that there are any blood relatives?

- Yes
- No

1.5 Is there a record of premature coronary heart disease in the patient's blood relatives?

- Yes
- No

1.6 Is there evidence that the patient accepted or refused a DNA test for FH?

- Patient had DNA test - evidence of consent
- Patient had DNA test - no evidence of consent
- Patient refused DNA test
- Patient not offered DNA test
- Not recorded

1.7 Is there evidence that the diagnosis included assessment of the following clinical signs of :

a. corneal arcus?

- Yes
- No

b. tendon xanthomata ?

- Yes
- No

1.8 Was the lipid profile on which the diagnosis made using a fasting sample or non fasting sample?

- Fasting
- Not fasting
- Not recorded

1.9 Is there evidence that the following secondary causes of hypercholesterolaemia were excluded before a diagnosis of FH was considered?

- Yes
- No

- a. Diabetes
- b. Renal failure
- c. Hypothyroidism
- d. Cholestasis

1.10 What were the lipo-protein measurements used to make the diagnosis?

Pre-treatment levels

| Date | Fasting | Cholesterol | HDL | Trig | LDL |
|--------------------------|----------|----------------------------------|-----|------|------|
| Year (from 1930 onwards) | Yes / no | 0 - 20 for hetero, 0-26 for homo | 0-4 | 0-6 | 0-18 |

1.11 Is there evidence in the notes that a baseline ECG was considered?

- Yes
- No
- Not applicable (*Paediatrics only*)

If answered "yes" to question 1.11 answer 1.12

1.12 Is there evidence in the notes that an ECG was this carried out?

- Yes
- No

Section 2: Pedigree

2.1 Is there a pedigree/ family tree in the notes? Yes No

Answer 2.2 if answered yes to 2.1

2.2 How is the pedigree / family tree recorded?

- a. Drawn using software
- b. Drawn by hand
- c. Written

2.3 If yes, does the pedigree map the following family members:

- a. First generation Yes No
- b. Second generation Yes No
- c. Third generation Yes No

If answered "yes" to 2.2 (a) (b) or (c) answer 2.3

2.4 Are the following data items recorded:

a. Relatives age

Yes - for all Yes - for some No

b. age of onset of coronary heart disease (if present)?

Yes - for all Yes - for some No

c. Information about tendon xanthomas?

Yes - for all Yes - for some No

d. Information about total cholesterol?

Yes - for all Yes - for some No

e. Smoking history of blood relatives with CHD?

Yes - for all Yes - for some No

f. (i) Are there any deceased blood relatives?

Yes No

If answered yes to f(i)

(iii) Please indicate which blood relatives have died?

1st Generation

| | |
|--------|--------------------------|
| Mother | <input type="checkbox"/> |
| Father | <input type="checkbox"/> |

| | |
|---|--------------|
| Number of 1 st generation siblings | enter number |
|---|--------------|

2nd Generation

| | |
|---|--------------------------|
| Grandmother (maternal) | <input type="checkbox"/> |
| Grandmother (paternal) | <input type="checkbox"/> |
| Grandfather (maternal) | <input type="checkbox"/> |
| Grandfather (paternal) | <input type="checkbox"/> |
| Number of 2 nd generation siblings | enter number |

h. If yes to (f), is the age and cause of death recorded?

1st generation

| | Age of death recorded | Cause of death recorded |
|-----------|-----------------------|-------------------------|
| Mother | Yes/No | Yes/No |
| Father | Yes/No | Yes/No |
| Sibling 1 | Yes/No | Yes/No |
| Sibling 2 | Yes/No | Yes/No |
| Sibling 3 | Yes/No | Yes/No |
| Sibling 4 | Yes/No | Yes/No |
| Sibling 5 | Yes/No | Yes/No |
| Sibling 6 | Yes/No | Yes/No |

2nd generation

| | Age of death recorded* | Cause of death recorded* |
|------------------------|------------------------|--------------------------|
| Grandmother (maternal) | Yes/No | Yes/No |
| Grandmother (paternal) | Yes/No | Yes/No |
| Grandfather (maternal) | Yes/No | Yes/No |
| Grandfather (paternal) | Yes/No | Yes/No |
| Sibling 1 | Yes/No | Yes/No |
| Sibling 2 | Yes/No | Yes/No |
| Sibling 3 | Yes/No | Yes/No |
| Sibling 4 | Yes/No | Yes/No |
| Sibling 5 | Yes/No | Yes/No |
| Sibling 6 | Yes/No | Yes/No |

Section 3: Assessment

What was the date of the patient's last (most recent) out-patient clinic appointment?

3.1 Who reviewed the patient at their last (most recent) out-patient clinic appointment?

- | | |
|----------------------------|--------------------------------------|
| Consultant | <input type="checkbox"/> |
| Specialist Registrar (SpR) | <input type="checkbox"/> |
| Specialist Nurse | <input type="checkbox"/> |
| Other | <input type="checkbox"/> Please list |

For Adults only

3.2 At the first clinic appointment was the patient's weight recorded?
 Yes No

3.3 At the first clinic appointment was the patient's height recorded?
 Yes No

3.4 At the last (most recent) clinic appointment was the patient's weight recorded?
 Yes No

For Paediatrics only

3.5 At the first clinic appointment was the patient's height and weight recorded?
 Yes No

3.6 At the last (most recent) clinic appointment was the patient's height and weight recorded?
 Yes No

Section 4: Treatment

4.1 What are the most recent lipoprotein results?

| Date | Fasting | Cholesterol | HDL | Trig | LDL |
|------|---------|-------------|-----|------|-----|
| | | | | | |

4.2 What is the patient's current lipid lowering drug therapy (i.e. as recorded at the most recent out-patient clinic appointment)?

- a. Statin:
- | | Yes | No | |
|------------------------------|--------------------------|--------------------------|--------------|
| Atorvastatin | <input type="checkbox"/> | <input type="checkbox"/> | Dose 10-80mg |
| Fluvastatin | <input type="checkbox"/> | <input type="checkbox"/> | Dose 80mg |
| Pravastatin | <input type="checkbox"/> | <input type="checkbox"/> | Dose 80 |
| Rosuvastatin | <input type="checkbox"/> | <input type="checkbox"/> | Dose 5-40 |
| Simvastatin | <input type="checkbox"/> | <input type="checkbox"/> | Dose 80mg |
| Patient not receiving statin | | <input type="checkbox"/> | |

b. (if applicable) Please indicate the reason(s) that this patient is not receiving statin. *More than one option may be selected.*

- | | |
|---|--------------------------|
| Patient intolerant to statin | <input type="checkbox"/> |
| Patient declined | <input type="checkbox"/> |
| Patient planning to become or is pregnant/ is breastfeeding | <input type="checkbox"/> |
| Child under 10 years | <input type="checkbox"/> |
| Child 10 and over of but risk low | <input type="checkbox"/> |
| Other, please list | <input type="checkbox"/> |

- | | Yes | No | | |
|------------------------------------|--------------------------|--------------------------|--------------|-------|
| c. Resin | <input type="checkbox"/> | <input type="checkbox"/> | Dose | 4-24g |
| d. Ezetimibe | <input type="checkbox"/> | <input type="checkbox"/> | Date started | |
| e. Fenofibrate | <input type="checkbox"/> | <input type="checkbox"/> | Dose | |
| f. Bezafibrate | <input type="checkbox"/> | <input type="checkbox"/> | Dose | |
| g. Fish oil / Ω fatty acids | <input type="checkbox"/> | <input type="checkbox"/> | | |
| h. Nicotinic acid or derivative | <input type="checkbox"/> | <input type="checkbox"/> | | |
| i. plasma LDL-C apheresis | <input type="checkbox"/> | <input type="checkbox"/> | | |
| j. Other, please list | | | | |

Paediatrics only:

- k. If the patient is receiving lipid lowering treatment what date was it started?

4.3 Is there evidence that the following were measured before initiation of statin treatment:

- a. Muscle enzyme (CK - creatine kinase also known as creatine phosphokinase) Yes No
- b. Transaminases (ALT or AST) usually as part of liver function test profile) Yes No

For Paediatrics only

- c. Was this repeated within 3 months of starting statin treatment? Yes No

Section 5: Advice

This section covers adults and child/young persons.

5.1 Is there written evidence that the patient/carer was given the following life-style advice?

- a. Diet Yes No
- b. Exercise Yes No
- c. Smoking (*Question applies whether smoker or not*) Yes No

d. Alcohol Yes No

5.2 Is the smoking status of the patient recorded? Yes No

a. If yes, please select the status:

Smoker

Ex smoker

Non smoker

b. If yes, were they given advice about or offered a referral to an intensive support service/smoking cessation?

Offered referral Given written advice No referral or advice

Question 5.3 applies to females of reproduction age who have received, or are under consideration for receiving, lipid modifying drug therapy

5.3 Was the need for contraception whilst taking statins discussed and documented? Yes No Patient not of reproductive age

Section 6: Cascade Testing

This section applies to Adults only

6.1 Is there evidence that the mutation has been tested? Yes No

a. If yes, was the mutation identified?

Yes Result not yet reported No

6.2 Is there evidence that the need for cascade testing (testing of relatives cholesterol) has been discussed with the patient?

Yes No

6.3 Has the process of cascade testing been initiated? Yes No

If answered "yes" to question 6.3 answer 6.4

6.4 Please detail cascade testing undertaken in parents:

| | Relative within catchment area? | Relative contacted | Full lipid undertaken | DNA testing undertaken |
|--------|---------------------------------|--|-----------------------|------------------------|
| Mother | Yes/No | <ul style="list-style-type: none"> • Yes • No consent to contact relative • Contact details unknown • Deceased • Other, please list | Yes/No | Yes/No |
| Father | Yes/No | <ul style="list-style-type: none"> • Yes • no consent to contact relative • Contact details unknown • Deceased Other, please list | Yes/No | Yes/No |

a. How many children does the patient have?

b. How many brothers/sisters does the patient have?

Please provide details of cascade testing in children:

| | | | | | |
|---------|--------|--------|---|--------|--------|
| Child 1 | Yes/No | Yes/No | <ul style="list-style-type: none"> • no consent to contact relative • Contact details unknown • Deceased Other, please list | Yes/No | Yes/No |
| Child 2 | Yes/No | Yes/No | <ul style="list-style-type: none"> • no consent to contact relative • Contact details unknown • Deceased Other, please list | Yes/No | Yes/No |
| Child 3 | Yes/No | Yes/No | <ul style="list-style-type: none"> • no consent to contact relative • Contact details unknown • Deceased Other, please list | Yes/No | Yes/No |
| Child 4 | Yes/No | Yes/No | <ul style="list-style-type: none"> • no consent to contact relative • Contact details unknown • Deceased Other, please list | Yes/No | Yes/No |

Please provide details of cascade testing in brother(s)/sister(s)

| | | | | | |
|--|--------|--------|---|--------|--------|
| Brother/sister 1 | Yes/No | Yes/No | <ul style="list-style-type: none"> • no consent to contact relative • Contact details unknown • Deceased Other, please list | Yes/No | Yes/No |
| Brother/sister 2 | Yes/No | Yes/No | <ul style="list-style-type: none"> • no consent to contact relative • Contact details unknown • Deceased Other, please list | Yes/No | Yes/No |
| Brother/sister 1 | Yes/No | Yes/No | <ul style="list-style-type: none"> • no consent to contact relative • Contact details unknown • Deceased Other, please list | Yes/No | Yes/No |
| Brother/sister 3 | Yes/No | Yes/No | <ul style="list-style-type: none"> • no consent to contact relative • Contact details unknown • Deceased Other, please list | Yes/No | Yes/No |
| Brother/sister 4 | Yes/No | Yes/No | <ul style="list-style-type: none"> • no consent to contact relative • Contact details unknown • Deceased Other, please list | Yes/No | Yes/No |
| Brother/sister 5 | Yes/No | Yes/No | <ul style="list-style-type: none"> • no consent to contact relative • Contact details unknown • Deceased Other, please list | Yes/No | Yes/No |
| <i>Opportunity to add extra children / brother/sisters</i> | | | • | | |

6.5 Is there evidence that the patient was asked if second & third degree biological relatives have been tested?

Yes No

6.6 Has a letter been written to the index case's GP stating the diagnosis of FH has been made?
Yes No

a. Does the letter emphasize the need for referral and testing of at-risk relatives?

Yes No

Section 7: (Annual) Review

7.1 Has the patient had a structured annual review carried out at least annually since the initial diagnosis?

- Yes - in secondary care
- Yes - in secondary and primary care
- Yes - in primary care
- No - patient offered and declined
- No - not offered

b. If undertaken in secondary care, please give the dates of the last 3 reviews (if 3 have been carried out):

7.2 If review undertaken in secondary care, is there evidence in the notes that the review covered the following issues:

| | Yes | No | Not applicable |
|-------------------------------------|--------------------------|--------------------------|--------------------------|
| a. Response to drug(s)? | <input type="checkbox"/> | <input type="checkbox"/> | |
| b. Side effects? | <input type="checkbox"/> | <input type="checkbox"/> | |
| c. CHD status? | | <input type="checkbox"/> | <input type="checkbox"/> |
| d. Fertility issues (females only)? | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> |
| e. Lifestyle? | <input type="checkbox"/> | <input type="checkbox"/> | |
| f. ECG? | | <input type="checkbox"/> | <input type="checkbox"/> |
| g. Progress with cascade screening? | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> |

Section 8: Children

This section applies to patients who are under 18 years old.

8.1 Is there evidence that the mutation has been tested? Yes No

a. If yes, was the mutation identified?

Yes Result not yet reported No

8.2 If the family mutation is known (yes to 8.1(b)) has the child been offered a DNA test?

Appendix 2: Inter-rater reliability analysis

26 cases were duplicated for this analysis, which aims to test the reliability of the whole system of understanding the questions and supporting help notes, identifying the information needed and entering the data. Errors can happen at any point in this process and although a certain small rate of typographical errors is to be expected, it is important in piloting an audit tool to consider ways in which any deficits in reliability can be addressed through design of the data collection tool or clarification of the questions.

The sample size in a pilot study (here, 26 replicates) does not allow for statistical analyses of any greater precision than to say whether or not there is an obvious problem with a given question. Differences are therefore described quantitatively rather than attempting to use kappa scores or correlation coefficients. To illustrate, the ethnicity question shows some disagreement between auditors as shown in the table below:

| | White British | Not recorded |
|---------------|---------------|--------------|
| White British | 16 | 3 |
| Not recorded | 2 | 5 |

Formally, this has a Cohen's kappa score of 0.53, but because there are few data, the approximate 95% confidence interval is wide (0.17, 0.89) and no firm conclusion could be drawn.

A further consideration is that the data are drawn from 14 sites, so subtle ambiguity in the questions might not be detected quantitatively until a larger group of auditors start using the tool.

All duplicate cases were entered, as requested, by a person of different profession to the auditor who entered the corresponding primary case. 9 of the 26 were entered within a week of the primary case, and 22 were entered within 30 days.

In summary, the levels of agreement show some areas which need to be made more reliable and in most cases it is obvious that better definitions in the helpnotes and on-screen will go a considerable way to achieving this. The disagreements are not far beyond what would be expected in a well-established audit so it seems that this dataset and collection method has potential to be used reliably at a national level in the near future.

Questions with perfect agreement are not listed below.

Date / year variables:

- Q0.3 clinic date: 1/26 pairs disagreed (by 28 days across Feb/March - likely to be caused by a data entry error)
- Age: 1/26 pairs disagreed by 1 year and 1 month
- Q1.6 Year of diagnosis: there were 4/23 disagreements, of 1, 2, 6 and 18 years' difference.
- Q1.10a Year of baseline lipoprotein measurements: 2/26 disagreements, by 1 year and 18 years.

Categorical variables:

- Q0.6 Ethnicity: 5/26 pairs disagreed between White British and Not Recorded. In some cases the latter had been entered in slightly different words under “other ethnicity”.
- Q1.1 Diagnosis: 8/26 disagreements, 4 heterozygous vs possible, 1 homozygous vs possible, and 3 not known vs possible. The “possible” option probably needs to be defined more rigorously.
- Q1.5 Premature CHD in blood relatives: 2/26 disagreements
- Q1.6 Patient accepted/refused DNA test: 9/26 disagreements. There are 5 categories in response to this question and there may be ambiguity between these.
- Q1.7a Corneal arcus: 4/26 disagreements
- Q1.7b Tendon Xanthomata: 4/26 disagreements
- Q1.8: Fasting samples: 2/26 disagreements
- Q1.9: Exclusions in diagnosis: 4/26 disagreements in each of these.
- Q1.10b Fasting baseline samples: 4/26 disagreements. Perhaps it is not clear, if baseline measurements from different appointments were all used in diagnosis, which to use in answer question 1.10.
- Q1.11 ECG considered: 3/26 disagreements.
- Q1.12 ECG performed: 4/26 disagreements.
-

Biochemistry variables:

- Q1.10c-f Baseline lipoprotein measurements: 8 disagreements in cholesterol, 5 in HDL, 6 in triglycerides, and 8 in LDL. Two sites are responsible for most of these disagreements, which suggests a systematic error in selecting the information from the notes. See also the note above against 1.10b.
- Q4.1c-f Most recent lipoprotein measurements: 2 disagreements in cholesterol, 4 in HDL, 2 in triglycerides, and 5 in LDL. This is a bit better than the baseline disagreements, perhaps reflecting the ease of finding the most recent set of results. The errors tended to group together in the same patients, indicating that the wrong set of results had been found.

Appendix 3: FH Audit pilot participants

| Pilot Site | Trust Name | Audit Lead(s) |
|--------------------------------------|--|--|
| Morrison Hospital | Abertawe Bro Morgannwg University NHS Trust | Dr Andar Gunnberg Delyth Townsend |
| University Hospital Aintree | Aintree University Hospitals NHS Foundation Trust | Dr Charles van Heyningen Claire Ashton |
| Birmingham Children's Hospital | Birmingham Children's Hospital NHS Foundation Trust | Dr Chris Hendriksz Dr Anupam Chakrapani |
| Royal Sussex County Hospital | Brighton and Sussex University Hospitals NHS Trust | Dr Andrew Iversen Dr Katherine Harding |
| Manchester Royal Infirmary | Central Manchester and Manchester Children's University Hospital NHS Trust | Dr Handrean Soran Sister Ruth Eatough |
| Prince Charles Hospital | Cwm Taf NHS Trust | Dr David Cassidy Dr Dev Datta |
| East & North Hertfordshire NHS Trust | East & North Hertfordshire NHS Trust | Adie Viljoen Ann Ainsworth |
| Royal Brompton Hospital | Royal Brompton & Harefield NHS Trust | Jane Jones Emma Neves Dr M Barbir |
| Royal United Hospital | Royal United Hospitals Bath | Dr John Reckless Dr Andrew Taylor |
| Southampton General Hospital | Southampton University Hospitals NHS Trust | Dr Emmanuel Abu Dr Paul Cook |
| University Hospital Lewisham | The Lewisham Hospital NHS Trust | Clare Holtby Michael Addley |
| The Shrewsbury and Telford NHS Trust | The Shrewsbury and Telford Hospital NHS Trust | Dr Nigel Capps |
| Furness General Hospital | University Hospitals of Morecombe Bay Hospitals NHS Trust | Dr Alan Taylor |
| Worcestershire Royal Hospital | Worcestershire Acute Hospitals NHS Trust | Dr Alistair Munro |

All sites completed the Organisational Audit.

Appendix 4: Pilot Phase Evaluation -Survey of Pilot Sites



Royal College of Physicians

National Audit of Familial Hypercholesterolaemia (Pilot project)

SURVEY OF PILOT SITES - RESULTS

Version 1 (3 June 2009)

Audit site / form completed by

[Removed for data protection purposes]

1) *Did you find taking part in the audits useful?*

| | |
|-------------------------------|---|
| Very useful | 2 |
| Fairly useful | 1 |
| Neither useful nor not useful | |
| Not very useful | 1 |
| Not at all useful | |

Free text comments

2) *Did you receive any support from the nominated Audit Lead for your Trust (or your Trust's Audit Dept?)*

| | |
|-----------------------|---|
| Yes - lots of support | 1 |
| Some support | 1 |
| No support as yet | 2 |

3) *Would you encourage others to take part in any future national FH audit?*

| | |
|----------|---|
| Yes | 3 |
| No | 1 |
| Not sure | |

Why? Free text comments

4) *Have you changed any of your practice as a result of this audit?*

Yes 1
No 3

If yes, please provide details.

Free text comment

- Would consider the need for more regular weight observations. At follow up.
- Better family screening, patient interaction and advice

5) *Was selecting eligible patients straightforward?*

Very straightforward 2
Fairly straightforward 1
Neither straightforward nor not straightforward
Not very straightforward
Not at all straightforward 1

Free text comment

- This was easy for us because we already have a set up FH clinic of four years.
- Tend to use terms probably or possible FH
- The PCT/LHB will not pay for any follow up visits, so we are under pressure to deal with an FH patient in a single visit. If we do DNA testing, we are forced to a single follow up visit. So the audit for a third visit FH is not a typical patient.

6) *If the audit included all FH patients please estimate the number of additional FH patients (i.e. those attending their first and second clinic appointments) you would have been able to include in the data collection period (Jan 12-April 17)*

Free text comment

- Approximately 30-40 more patients.
- 5
- 3-5 additional cases
- 25

7) *Did you find webtool-based data collection straightforward?*

Very straightforward

Fairly straightforward 3
Neither straightforward nor not straightforward 1
Not very straightforward
Not at all straightforward

Free text comment

- bit awkward entering family history and screening data

8) *Once you were familiar with the audit proforma and tool, on average how long did it take you to audit each patient?* Please include time to collect data from notes and enter on to web tool.

Free text comment

- Twenty minutes.
- 10-15minutes
- 30 minutes
- 25 minutes

9) *Are there any ways in which the webtool be improved?*

Free text comment

- Data base could be improved in area of pedigrees as it was unclear whether it wanted dead or alive relatives also cascade method was unclear to start.
- Have a standardized database of all patients for future data mining
- Just collect the data that you are going to use

10) *Are there any audit questions which could be clearer? How?* (Feedback previously provided need not be repeated.)

Free text comment

- See above.
- We will tell the patient to tell relations to go to their GP for a chol test because we have no lipid clinical follow up nurse. I don't care where they live. How do I enter this?

11) *Are there any questions that you felt were not worth asking?*

Free text comment

- Catchment area was not necessary also did not give option to explain where you were in the cascade process for relatives.
- Age in years and months. This awkward question got me off to a bad start with the form. Is this to the nearest month or the completed month. Suggest take a risk and use the date of birth. This stops inadvertent

duplicate entries. Logging in was annoying - especially the warning screen.

12) *In general was the information required to answer the questions possible to obtain:*

| | |
|---------------------------|---|
| Very easily | 1 |
| Easily | 1 |
| Neither easy or difficult | |
| With difficulty | 2 |
| With great difficulty | |

13) *Were there any questions for which the information could only be obtained with great difficulty?*

Free text comment

- No
- no

14) *Did you find the help desk support useful?*

| | |
|-------------------------------|---|
| Very useful | 2 |
| Fairly useful | |
| Neither useful nor not useful | 1 |
| Not very useful | |
| Not at all useful | 1 |

Free text comment

- Not easy to contact at times.

15) *Did you find the newsletters useful?*

| | |
|-------------------------------|---|
| Very useful | 2 |
| Fairly useful | 1 |
| Neither useful nor not useful | 1 |
| Not very useful | |
| Not at all useful | |

Free text comment

16) *Is there anything the FH Audit could do to make participating easier in the future?*

Free text comment

- Consider the quotes we have added.
- Help with filling in data which is time consuming e.g. by a designated nurse

- Ask a busy lipid clinic what matters. Decide what questions are worth asking. Show us the draft report on this pilot data and demonstrated that all the data fields were useful.

17) Have you developed any quality improvement materials during/following participation in the audits that could be useful for other FH professionals? What do they consist of?

Free text comment

- Our clinic is set up based on the N.I.C.E. guidelines for FH management. Everything is in place, we are willing to share our knowledge.
- Family tree drawing skills
- No
- Not sure what you mean

18) Are there any materials that you would find useful for quality improvement that we could develop for use nationally?

Free text comment

- We use an initial assessment form for our new patients this could be developed and used. This form covers most of what is needed along with our family files for any further audit use.
- Software for electronic pedigree drawing
- Specific paediatric management guidelines (current NICE guidelines are too non-specific)
- Happy to consider suggestions