

Primary Care Service Framework: Familial Hypercholesterolaemia

1. Preface

Familial hypercholesterolaemia is a genetic disorder characterized by high cholesterol levels, specifically very high low-density lipoprotein cholesterol (LDL-C) levels, in the blood and premature cardiovascular disease.

Familial hypercholesterolaemia (FH) occurs in about one person in every 500 and is one of the most frequently occurring inherited conditions. It is caused by an abnormal gene resulting in exceptionally high LDL-C levels. These high cholesterol levels start from birth and are present throughout life. This leads to early hardening of the arteries and the onset of vascular disease. People with FH are at high risk of early coronary heart disease (CHD).

FH is hugely under diagnosed. Although more than 120,000 people in the UK have the disease, a 2008 survey of lipid clinics indicated that 15,000 cases are known at most. Thus, approaching 90% of people with FH go undetected and may be disabled or die from CHD early in life, often in their 40s or 50s, sometimes earlier.

FH is a genetic problem with a dominant mode of inheritance and can therefore be passed from parent to child. Each family member has an even chance of inheriting the problem. The disease does not skip generations. This means that children and grandchildren of family members who do not have the defect are not at risk from FH.

Effective treatments are available. The key to improving outcomes for people with early FH is early identification which will enable early initiation of preventative treatment. Therefore, whenever FH is diagnosed, it is important that all close relatives are followed up so they can start preventative treatment.

The Institute for Health and Clinical Excellence (NICE) published guidance on the identification and management of FH in August 2008. This guidance clearly states that risk algorithms such as Framingham or QRISK are not appropriate for estimating CHD risk in FH patients, and the high CHD risk in FH patients is often not recognised by GPs who therefore do not offer adequate lipid lowering therapy, or identify the familial nature of the disease (and therefore relatives are not tested).

To date, few commissioners have taken action and FH remains something of a "Cinderella" condition. Once diagnosed, FH can be effectively treated (usually with drug therapy such as statins).

The preventive aspect is therefore important: it is estimated that if the detection rate for FH was improved to 80-90%, the potential saving to the NHS in terms of reduced premature morbidity/mortality would be of the order of £20m. In developing a business case to implement this Primary Care Service Framework (PCSF), commissioners may also find it useful to note:

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| | <ul style="list-style-type: none"> • In terms of quality and productivity, for FH it is necessary to invest in order to save • The NHS Constitution¹ gives people a right to a health check – between 1-2% of these will be found to have very high cholesterol levels. The NHS Health Checks² document, Putting Prevention First, advises that it is important to consider FH if cholesterol levels exceed 7.5mmol/L. • Some FH patients may already be on treatment as a result of their hypercholesterolaemia despite their diagnosis being unrecognised. Therefore, not all new FH patients will represent additional treatment costs • Genetic testing is a one off cost; the savings arising from the treatment of FH are on going • Evidence also suggests that the health burden of people treated for FH is lower than for other people of the same age, particularly if they have yet to sustain a first cardiovascular event. • There is a potential gain to the wider economy given that FH affects people of working age. |
| <p>2. Purpose of this Primary Care Service Framework</p> | <p>The main purpose of this framework is to support commissioners to put in place improved services to identify people with FH and improve their management. This should result in a reduction in unnecessary early deaths and disability from the condition.</p> <p>Commissioners need to ensure that an integrated, patient centred service for people with familial hypercholesterolaemia is in place for their population. The service will comprise several elements: diagnosis, genetic testing, counselling and management.</p> <p>The purpose of this framework, therefore, is to enable commissioners, providers and practitioners with the necessary background knowledge, service and implementation details to deliver improved identification of people with FH through:</p> <ul style="list-style-type: none"> • improved pathways and communications • genetic testing • improved diagnosis of people with FH, especially children and young people. (Some of those aged 40 and over will already be known as they will have already had an event requiring medical intervention. Some others may be picked up through the health checks/vascular checks programmes being commissioned from primary care.) • improvement of early diagnosis of people under 40 with FH (especially children and young people) through family cascade testing • cascade testing to identify other affected relatives • counselling • onward management of patients in normal primary or secondary care services <p>It is important to note that although this will enable better management of people with this condition, this framework does not deal with the last element – the management of FH once it has been identified. For this, PCTs should refer to NICE guidance.</p> |

¹The NHS Constitution is currently out for consultation. It proposes that the NHS Health Check will become a right for all those eligible with effect from 2012.

²NHS Health Check- the NHS health Check programme (formerly Vascular Checks) is a universal and systematic programme for everyone between the ages of 40-74 that will assess people's risk of heart disease, stroke, kidney disease and diabetes on a 5-year rolling basis and will support people to reduce or manage that risk through individually tailored advice. PCTs began implementing the programme in April 2009 and are expected to achieve full roll out by 2012/13. People with existing diagnosed vascular disease, including FH, are excluded from the programme.

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| <p>3. Period of Service</p> | <p>It is recommended that this service is commissioned for a minimum period of 5 years initially, subject to satisfactory annual review. This is to allow time for the service to become sufficiently established in order to reach a detection level of 80-90% of people with FH.</p> |
| <p>4. Scope and Definition of Service</p> | <p>Target population: the service is aimed at all age groups (both index cases and associated family members) and will comprise the following stages:</p> <ol style="list-style-type: none"> 1. Identification 2. Confirmation of diagnosis 3. Undertaking family cascade testing 4. Follow-up <p>Provider: this service may be commissioned from both primary and secondary care providers and joint proposals may be beneficial. The various components of the service may be commissioned from different providers. However, providers will be required to work together to ensure that the patient pathway through the service is managed and seamless.</p> <p>Level: consideration should be given to commissioning this service over a large area – possibly SHA wide – for two reasons:</p> <ol style="list-style-type: none"> 1. the relatives of an index patient may live outside the PCT’s area – possibly in several different PCTs. SHAs may, therefore, wish to consider co-ordinating the commissioning and development of the services across their area and beyond (consideration should also be given to the pathways for relatives living outside the SHA area). 2. there is approximately a 3-fold difference in cost between genetic tests for index cases and their relatives, and working jointly with other PCTs will yield better value as the relative proportion of the (more expensive) index tests will be lower <p>PCTs will also need to review the levels of genetic testing services they commission to ensure existing services are not overloaded.</p> <p>This primary care service framework should not be confused with (and sits outside of) essential and additional GMS or PMS services already provided, current Quality and Outcomes (QOF) indicators and any National Enhanced Services.</p> |
| <p>5. Parties to the agreement</p> | <p>For example: Names of any accountable individuals and organisation details.</p> |
| <p>6. Background</p> | <p>Familial hypercholesterolaemia (FH) is a genetic condition that causes a high cholesterol level in the blood. It is caused by mutations in genes of the pathway that clears low-density lipoprotein (LDL) from the bloodstream (in the majority of patients in the gene for the LDL receptor). Elevated LDL-cholesterol is present from birth and may lead to early development of atherosclerosis and coronary heart disease. The disease is transmitted from generation to generation in a dominant pattern, such that siblings of affected individuals have a 50% risk of inheriting FH. Once diagnosis has been confirmed in an individual, it is important to “cascade” screening to other family members.</p> |

There are two types of FH:

- Heterozygous – where an individual inherits one copy of the defective gene. This is the most common form.
- Homozygous – where an individual inherits two copies of the defective gene.

Levels of understanding about the condition are low. However, the condition is relatively common. The prevalence of heterozygous FH in the UK population is estimated to be 1 in 500. This means that over 120,000 people are affected in the country. Both adults and children are under-diagnosed.

If untreated, having this condition leads to:

- a 50% risk of coronary heart disease by the age of 50 years in men and
- a 50% risk of coronary heart disease by the age of 60 years in women

Homozygous FH is much rarer, with a prevalence of approximately 1 in 1,000,000. Symptoms appear in childhood, and are associated with very early death from CHD.

Besides a very high cholesterol level and family history of premature CHD, the following signs may occasionally be present:

- cholesterol deposits (tendon xanthomata) in the tendons on the back of the hand and in the Achilles tendon on the heel
- cholesterol deposits (xanthelasmas) in the skin around the eye or eyelid
- a pale or white ring (corneal arcus) around the outer rim of the iris

Only tendon xanthomata are specific to FH.

Advice on lifestyle (eg dietary advice and smoking cessation) are essential in the management of FH, although medication is also necessary in almost all cases. Cholesterol lowering drugs are effective in treating the condition (statins being the first choice). Generic statins are a relatively inexpensive treatment, so an increase in drug costs associated with increased identification is not always a major issue. However, the NICE guidance recommends using high-potency statins (Atorvastatin and Rosuvastatin) in order to achieve the LDL target reduction, and these are still under patent (although this will expire soon for atorvastatin).

Each individual identified with a confirmed diagnosis of FH (termed the “index case”) has on average 4-5 first degree relatives, of whom 50% on average will have FH. Each affected first degree relative then becomes a new index case and their first degree relatives (for example their children) can be tested. Thus each index case can be expected to lead to the identification of 4-5 new FH subjects if systematic cascade testing is carried out. (The word “systematic” here implies not just testing first degree relatives but extending this to second and possibly even third degree relatives as recommended by the NICE guidelines.)

NICE clinical guideline no 71 gives the key priorities for implementation as:

- diagnosis
- identifying people with FH using cascade testing
- management (adults and children/young people)

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| | <p>Two case studies will serve to illustrate the issues posed by FH and the importance of diagnosing index cases and identifying family members at risk:</p> <ol style="list-style-type: none"> 1. A man died of an acute MI aged 32, and his son was subsequently referred with a total cholesterol of 5.2 and LDL-cholesterol of 3.7 mmol/L, aged 11. The son had genetic testing done when this became available, and this has confirmed he has FH. 2. A woman with FH (LDL-C of 5.3 mmol/l) marries a man with FH. There is a possibility that any offspring they have will suffer from homozygous FH <p>See also the References & Resources section at the end of this document</p> |
| <p>7. Summary of Local Need</p> | <p>The prevalence of FH (1 in 500) is uniform across the UK, so the likely numbers of people with the condition can easily be extrapolated for a given population. There is thus no need for a specific local needs assessment to be undertaken. However, in implementing this framework, commissioners will need to discount those people who have already been identified with a diagnosis of FH - this will require a rolling gap analysis (to take account of births/deaths). Specialist Dutch software (currently being field tested in Wales) is available to do this. Could reference the relevant section of the HEART UK toolkit here.</p> |
| <p>8. Service Objectives and Intended Health Outcomes</p> | <p>The key service objectives are:</p> <ul style="list-style-type: none"> • to improve the detection level of FH in the population from the present 10-15% to about 80-90% • to offer screening and treatment to those identified with a diagnosis of FH. <p>In clinical terms, the objective is to achieve a reduction >50% in the LDL-C³ of these individuals.</p> <p>This will help achieve:</p> <ul style="list-style-type: none"> • A reduction in premature mortality (and through this, a reduction in health inequalities for this group of people)⁴ • A reduction in morbidity eg avoidance of adverse vascular events, prevention of strokes etc • A reduction in the risk of disability for an economically active group • A saving to the NHS arising from the fact that the health burden of people treated for FH is lower than that for other people of the same age. • Improved survival rates for people with FH both in primary and secondary care. (People with FH who have not yet experienced a CHD event, have a normal life expectancy, once treated, in comparison with the rest of the population. Conversely untreated FH sufferers have a shorter life expectancy.) • Implementation of the NICE guidance relating to FH • Improvement in patient experience: it is possible that the certainty of diagnosis relieves anxiety/guilt in patients who otherwise worry that their condition is a result of lifestyle choices |

³Low density lipo protein cholesterol

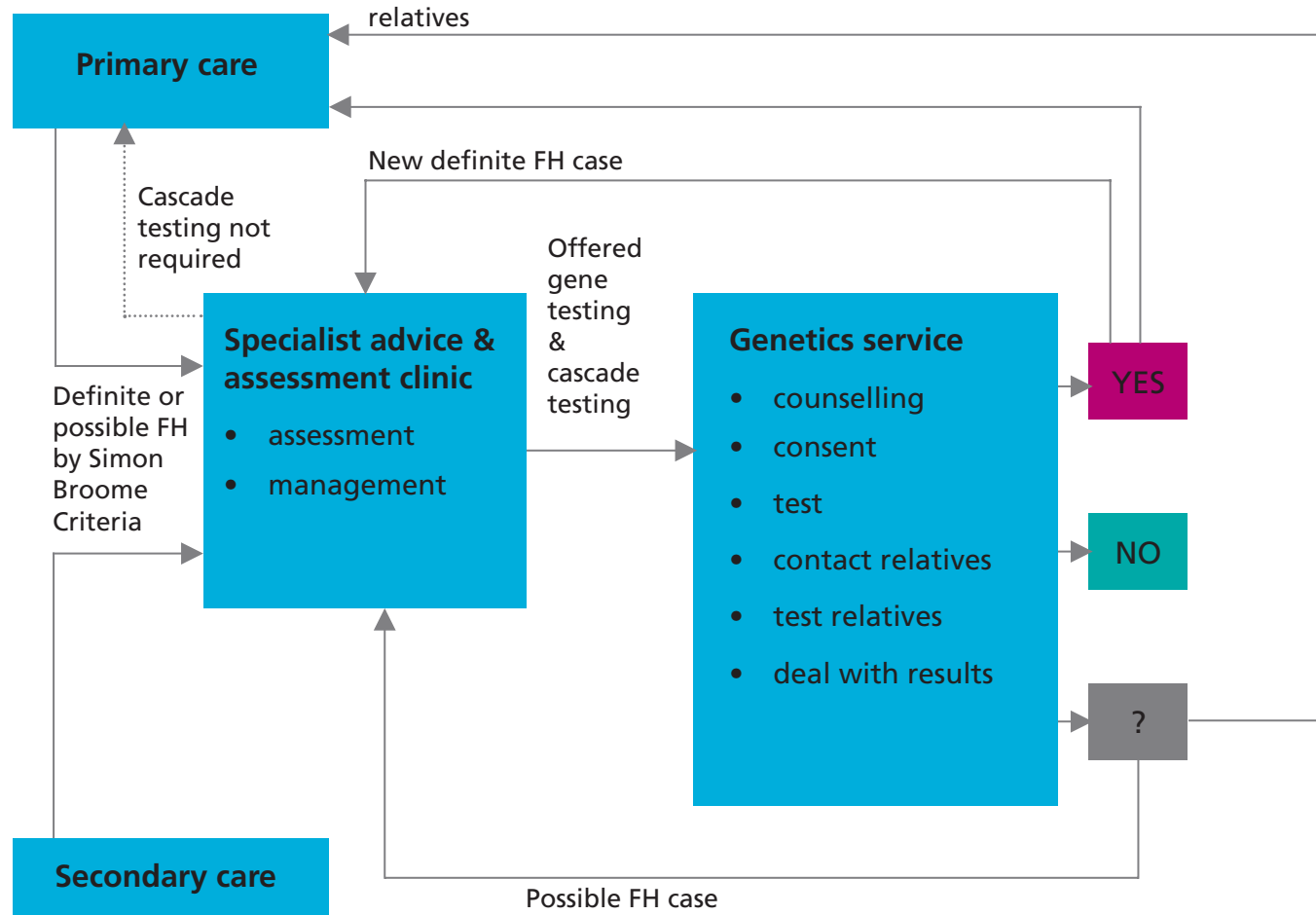
⁴Reductions in all-cause, cancer, and coronary mortality in statin-treated patients with heterozygous familial hypercholesterolaemia: a prospective registry study. Neil A, Cooper J, Betteridge J, Capps N, McDowell I, Durrington P, Seed M, Humphries SE. Eur Heart J. 2008 Nov;29(21):2625-33.

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| | <ul style="list-style-type: none"> • Having a genetic diagnosis also improves likelihood of patient compliance with treatment regimes⁵ • This service should deliver both prevention and improved quality in line with the Quality, Innovation, Productivity & Prevention (QIPP) agenda. |
| <p>9. Service Outline</p> | <p>There is no one service model for FH, and the approach adopted in a particular area will depend to a large degree on local circumstances.</p> <p>The Department of Health funded FH Cascade Audit Project has demonstrated the feasibility and acceptability of cascade testing for FH in the UK⁶.</p> <p>Two possible pathways –a genetics centre-led model and an outreach service –are illustrated below. The following general principles are applicable to any model:</p> <ul style="list-style-type: none"> • Once someone has been identified as a possible FH sufferer, eg in primary care, they should be assessed by someone with specialist knowledge for confirmation of the diagnosis: it is important to be able to differentiate FH from other causes of high cholesterol levels. The Simon Broome criteria are recommended by NICE when deciding when to refer to a specialist. Referral for genetic testing should only be made by someone with specialist training. These activities are frequently carried out in lipid clinics but could be carried out in other settings by someone with the appropriate level of training, eg a GP with a Special Interest or a nurse specialist. • The genetics element has two components: laboratory analysis (to confirm the diagnosis of FH) and counselling to support family cascade testing (which could be linked to lifestyle counselling). Whilst these two components need to work closely together, they do not need to be provided by the same organisation. It is however vital that each component is provided by an organisation with the requisite skills. • Close links are needed between those undertaking the genetic diagnosis and those providing treatment. Whilst services need not be provided by the same provider those delivering the service will need to work as part of a multidisciplinary team • Counselling and family cascade testing should be undertaken by specialist nurses who have been appropriately trained in the interpretation and communication of genetic test results and information and with proper supervision. It should be noted that most of the questions asked by patients are not to do with genetics but are to do with the consequences of hyperlipidaemia, dietary advice, side effects from statins etc. PCTs may need to commission jointly to provide sufficient patient “critical mass”. • Children and young people will need access to specialist paediatric lipid services delivered in an appropriate child/young person focused environment, in line with NICE clinical guideline 71 • NICE guidelines should be used for the ongoing management of people in either primary or secondary care <p>Close collaboration between the various providers and commissioners is required to agree the most appropriate pathway. In developing this pathway, consideration should be given as to how best to ensure that young people do not go undetected – who will derive the greatest benefit.</p> |

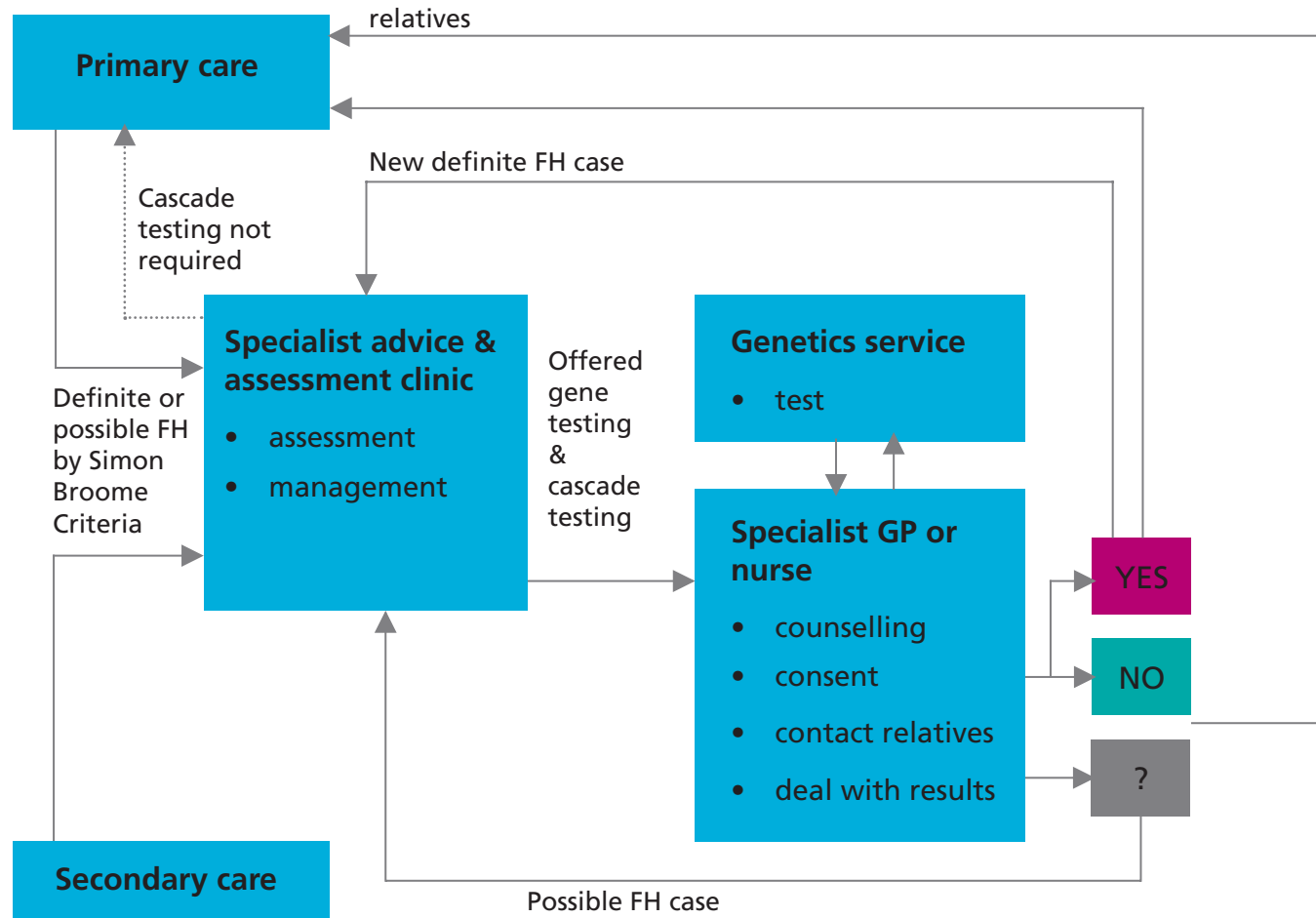
⁵Self reported adherence to cholesterol lowering medication in patients with FH: the role of illness perceptions. Senior V, Marteau M and Weinman J Cardiovascular Drugs Therapy 2004 18: 475-81. Quality of life, anxiety and concerns among statin-treated children with familial hypercholesterolaemia and their parents. de Jongh S, Kerckhoffs MC, Grootenhuys MA, Bakker HD, Heymans HS, Last BF. Acta Paediatr. 2003 Sep;92(9):1096-101

⁶The full manual of Operations and Standard Operating Procedures including templates for letters are available from Professor Steven Humphries at Cardiovascular Genetics BHF Laboratories, Rayne Building, UCL, 5 University Street London WC1E 6JJ, Tel 0207 679 6962

Illustrative Pathway 1 – Genetics-led service



Illustrative pathway 2 – outreach



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| <p>10. Location of Service</p> | <p>Elements of the service are likely to be provided from various locations – the key requirement is that all premises/ locations should be fit for purpose.</p> |
| <p>11. Integrated Governance</p> | <p>Clinical Governance arrangements must be proportionate to the service provided and comply with any local expectations or requirements of the commissioner.</p> <p>Any commissioned service must meet all national standards of service quality and clinical governance including those set out in Standards for Better Health: www.dh.gov.uk/en/publicationsandstatistics/publications/publicationspolicyandguidance/dh_4086665. These core and developmental standards of provision are designed to cover the full spectrum of health care as defined in the Health and Social Care (Community Health and Standards) Act 2003. The seven domains are safety, clinical and cost effectiveness, governance, patient focus, accessible and responsive care, the care environment and public health. Compliance with relevant NICE guidance is required.</p> <p>Professional competency, education and training - Healthcare staff delivering the service will be required to demonstrate their professional eligibility, competence, and continuing professional development in order to remain up-to-date and deliver an effective service which is culturally appropriate. Staff appraisal on an annual basis and at an appropriate level will also be required. Commissioners will need to be reassured that practitioners have the required competencies at an appropriate level.</p> <p>Commissioners should be satisfied that providers who deliver the services described have a planned, regular programme of education, training and support for their staff.</p> <p>Providers should ensure safe staffing capacity at all times and staff should be able to demonstrate that they have participated in organisational mandatory and update training, for example infection control, manual handling, risk assessment as required.</p> <p>Staff undertaking genetic counselling must be appropriately trained. This is a relatively new and expanding field. At present, as a result, this will normally be carried out by a clinical geneticist (although there are currently two recognised courses at Cardiff University and Manchester University). Appendix 1 gives a sample job description for the role of the specialist nurse in FH. Additionally, appropriate supervision for staff undertaking counselling must be in place.</p> <p>Patient, public and staff safety – Providers will be required to demonstrate that evidence based clinical guidelines are being used. Providers should have in place appropriate health and safety and risk management systems and ensure that the PCTs required premises standards are met. They should also ensure that any risk assessments and significant events are both documented and audited regularly and outcomes of these implemented. Services should comply with national requirements for recording using an agreed risk reporting, investigation and implementation of learning from incidents. Further details can be found on the National Patient Safety Agency website: www.npsa.nhs.uk</p> <p>The provider will ensure that staff undertaking patient assessments will have full CRB checks/clearance</p> |

Information management - The protection, use and disclosure of patient information must comply with the information governance policies and guidance set out in the NHS Information Governance Toolkit which can be found at www.igt.connectingforhealth.nhs.uk. This encompasses the NHS Codes of Practice on Confidentiality, Records Management and Information Security and supports delivery against core standard C9 of Standards for Better Health. All staff should undertake the information governance training provided on-line at www.igte-learning.connectingforhealth.nhs.uk/igte/index.cfm.

Equipment – Providers will be expected to adhere to Medicines and Healthcare Regulatory products Agency (MHRA) advice and guidance on selection of appropriate equipment, training in its use and ongoing management, troubleshooting, and quality assurance processes that ensure the accuracy and reproducibility of test results.

Tests - All genetic tests should be carried out in a CPA⁷ accredited laboratory and should meet relevant DH guidelines (e.g. reporting times).

The NHS Centre for Evidence-based Purchasing (CEP) is preparing an Evidence Review on rapid genetic testing for Familial Hypercholesterolaemia. The report will review current clinical, technical and economic evidence on point-of-care and rapid FH microarrays. It will discuss sensitivity and specificity of the tests and help to inform NHS purchasing decisions. The Evidence Review is due for publication in March 2010 and will be available through NHS Evidence: www.pasa.nhs.uk/cep (By way of background, CEP was part of the NHS Purchasing and Supply Agency (NHS PASA) and existed to underpin purchasing decisions by providing technical, clinical, economic and service delivery information on medical equipment and technologies. The CEP work programme is closing along with NHS PASA. The role of evaluating medical technologies now passes to NICE as part of the Evaluation Pathway for medical technologies.)

Clinical audit and review – Providers will be required to demonstrate their coordination of and involvement in regular inter-professional and inter-agency meetings and regular clinical audit of the service interventions and outcomes such as drug therapies or well-being and behaviour changes. This audit can be carried out by extracting data using the Read codes.

Patient and Public Involvement - Providers will be required to demonstrate active engagement with people and local communities in developing services, self care plans or in supporting people to utilise self care opportunities. Providers should demonstrate how they respond to patient feedback and this is to be used to shape and improve services. Involving family carers and supporters will help deliver the components within this service specification. Cardiovascular networks, Local Involvement Networks (LINKs), the voluntary sector and patient advocacy organisations are all further mechanisms to seek active involvement in service planning, delivery and monitoring.

Equality and Human Rights - Delivering good quality care will require organisations to demonstrate competence in identifying and taking action on inequality and also needing to engage with communities that have not found accessing public services easy. Undertaking Equality Impact Assessments (EQIAs) is a specific legal obligation, and conducting EQIAs and using the evidence to create a meaningful dialogue with communities (especially seldom heard from groups) is central to effective commissioning and service provision. This will create an evidence-based approach.

⁷Clinical Pathology Accreditation (UK) Ltd

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| | <p>As a minimum, core standard C7e of Standards for Better Health stipulates “healthcare organisations should enable all members of the population to access services equally and offer choice in access to services and treatment equitably”. To assist this process, organisations may wish to refer to ‘Creating a Disability Equality Scheme: a Practical Guide for the NHS’ www.dh.gov.uk/en/Publicationsandstatistics/Publications/PublicationsPolicyAndGuidance/DH_4139666.</p> <p>Managing complaints – providers should have in place a complaints system that reflects the arrangements introduced on 1 April 2009. This provides an opportunity for all organisations to review their local systems so they can both respond flexibly to complaints, concerns and complements and feed the resulting lessons into their work on learning from patients’ feedback to improve services.</p> <p>Continuous quality improvement – a set of indicators should be selected or developed and then agreed which defines the key quality requirements of the service. The service should also identify how it uses these measures and others to ensure that the quality of the service is continuously improved.</p> |
| <p>12.Information management/ requirements</p> | <p>The prevalence of FH in the UK population is uniform, so commissioners can expect similar levels of activity per head of population⁸. Service levels will, however, be differential.</p> <p>Each index case can be expected to lead to the identification of 4-5 FH subjects if systematic cascade testing is carried out.</p> <p>Commissioners will want to know how many people have been seen at each stage of the pathway and what happened to them. The aim should be to build a database that can inform future commissioning and identify the impact of the FH service on other services.</p> <p>Core data is likely to include:</p> <ul style="list-style-type: none"> • Numbers referred into the service • Number of referrals onwards for genetic diagnosis • Number of patients offered/accepting genetic counselling • Number of patients tested and the outcomes (NB it is important to know the number of people diagnosed with definite FH. Read codes do not distinguish between possible and definite FH, and overcoding tends to occur as a result of the way GP computer systems work – for example, typing in the word “hypercholesterolaemia” automatically brings up FH. The Read codes do not support the diagnostic categories in the NICE guidelines.) |
| <p>13.Service Monitoring and evaluation</p> | <p>Monitoring and evaluation of the services should sit within the PCTs contract monitoring cycle. Service providers will need to demonstrate the effectiveness of the service to commissioners possibly at regular times during the year and, at the least, on an annual basis. This will need to be provided to the commissioners in an annual report, which will inform any annual review process or meeting. The process by which this evaluation is achieved can also be used to show the outcomes of the service to other key stakeholders such as users and family carers. Service evaluation should be built in from the commencement of any service and should cover, as a minimum, the following areas:</p> |

⁸Note the prevalence of FH in people from the Indian subcontinent is also believed to be 1/500 but the prevalence in people of Afro-Caribbean origin is unknown and thought to be lower.

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| | <ul style="list-style-type: none"> • Service Activity – Volume of work against any agreed activity levels and distance from profile, capacity, needs and demand analyses, workforce arrangements, real time referral data to other care pathways or appropriate agencies recorded appropriate Read codes. • Clinical Outcomes – Regular analysis and interpretation of clinical outcomes data as well as regular analysis and interpretation of PPA data for prescribing. • Quality and Governance – Quality criteria will need to be established (in agreement with commissioners) and measured with standards needing to be met on a continual basis. Results of clinical audits will be used to inform service provision during the year. EQIA data should be used to underpin local integrated service provision. • Patient Experience – It is important to note that certainty of genetic diagnosis improves patient experience, eg through a reduction in feelings of guilt. Patients views for both index and family cases on their experiences and satisfaction levels will need to be measured through an on-going, systematic process to test whether the service is engaging with patients, family carers and supporters in a way that supports them. Different and innovative approaches to obtaining these views and experiences will be necessary, eg through capturing stories of community members experiences of the NHS and applying the learning from these. These processes should also be stratified where possible to show any differential impact on disadvantaged groups (e.g. Black and Minority Ethnic groups, deprived groups, males, females etc) and any resultant service changes (planned or achieved) should be highlighted. • Value for Money – Cost effectiveness or ‘best value’ analyses of the primary service outcomes in relation to comparative costs of hospital activity or those services providing equivalent quality of care. Such measures could include attendance rates, waiting times. Other possible analyses include: - Prescribing costs; benefits of increase in social capital and active citizenship; Staff and non-staff costs of running the service; Capital costs etc. |
| <p>14.Funding</p> | <p>There will be no fixed or nationally agreed price for this service. Commissioners and providers may wish to access alternative funding mechanisms, and should agree funding which is reflective of the level of service to be delivered locally and could include:</p> <ul style="list-style-type: none"> • Basic funding for achieving minimum requirements within the service specification • Additional funding or financial incentive for delivering specific local patient outcomes • Indication of national benchmark prices if available <p>Commissioners should note that testing for the index case is likely to cost between £300-£500. Subsequent familial tests will be substantially cheaper, there being in the region of a three-fold difference in the cost of an individual test.</p> |
| <p>15.Contract Management</p> | <p>For example:</p> <p>Name and contact point of the contract manager of both the commissioner and provider. Any specific local arrangements for contract management should also be stated.</p> |

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| 16. Review, variation and re-commissioning process | For example: <ul style="list-style-type: none">• Formal review of the service• Contractual variation procedures• De-commissioning and re-commissioning arrangements• Notice periods• Dispute Resolution and Arbitration procedures• Legal advice and support |
| 17. Signatories | Signatures from both parties as those accountable for the agreement |

Glossary/Definitions

Cascade testing: a method for identifying relatives at risk of a genetic condition following positive diagnosis in an index case

FH Heterozygote: individuals that have inherited one copy of the defective FH gene

FH Homozygote: individuals that have inherited two copies of the defective FH gene

Index cases: the original patient in whom a diagnosis is made and who is the starting point for follow-up of other members of a family when investigating possible genetic factors that are responsible for the presenting condition.

LDL-C: Low density lipoprotein cholesterol

Tendon xanthomata: cholesterol deposits on the tendons in the back of the hand or Achilles tendon in the heel

Xanthelasma: cholesterol deposits in the skin around the eye or eyelid

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Appendix 1

Role of Specialist Nurse in management of Familial Hypercholesterolaemia (draft)⁹

Familial Hypercholesterolemia is an autosomal dominant disorder of chromosome 19 with homozygous and heterozygous forms. It is defined in the WHO classification as a type IIa hyperlipidaemia. Homozygous FH is rare, with symptoms appearing in childhood, and is associated with early death from coronary heart disease. Homozygous FH has an incidence of approximately one case per million. The elevated serum cholesterol concentration that characterises heterozygous FH leads to a greater than 50% risk of coronary heart disease in men by the age of 50 years and at least 30% in women by the age of 60 years. The heterozygous form occurs in one in 500 people (meaning that approximately 110,000 people are affected in the UK). With only 15,000 people known to be affected, a great deal of work is needed to identify the remainder. The FH specialist Service requires a dedicated health professional to ensure that correct diagnosis is made, cascade screening of relatives is carried out and appropriate treatment is given. All of which can be performed by a trained FH specialist nurse.

Recently NICE published guidelines on the identification and management of adults and children with familial hypercholesterolaemia. The purpose of the guideline is to ensure that correct diagnosis is made, cascade screening of relatives is carried out and appropriate treatment is given. All of which can be performed by the FH specialist nurse. The FH specialist nurse should have clinical experience in managing inherited forms of lipid disorders, management of cardiovascular risk factors along with a moderate knowledge of genetics and counselling skills.

The Specialist nurse in FH will be responsible in assisting the lead consultant in the management of the patients with a possible probable or definite diagnosis of familial hypercholesterolaemia attending the clinic. The Specialist nurse in FH would provide:

1. Information on the condition
2. Information on the genetics of the condition where known

3. The importance of cascade testing
4. Genetic counselling
5. General support for not only the patient but their family also

The above would empower the patient with the patient having increased knowledge about their condition enabling them to make informed choices about their care, and increase the uptake of preventative medicine. It would also increase the control for the patient to participate in the planning of their care. Increased patient participation will lead to increased compliance with the regime agreed between the clinician and patient, showing the benefit of shared care. Appropriate training will be provided as specified in the training document and regular assessment will be carried out on all health professionals involved in the management of FH (See Training and assessment for health professionals in managing lipid disorders)

Diagnosis:

Approximately 800 patients are seen in the lipid clinic diagnosed with lipid problem attributable to a genetic cause with up to 10-15 patients seen in the clinic with possible or confirmed diagnosis of FH. Of the new referrals to the clinic at least 4-5 per week are diagnosed to have possible or definite FH. This number is steadily growing due to increased awareness of inherited types of hyperlipidaemia.

Diagnosis of FH is made using the Simon Broome criteria (see Appendix A). The FH specialist nurse re-checks cholesterol levels and other biochemical markers, observes for lipid stigmata (namely tendon xanthomata) and takes a detailed family history. If these findings correspond to the Simon Broome criteria, then cascade screening can be initiated, and DNA testing will be offered as appropriate. A family number will be given to the index case and a family file will be created for documentation of details of consultation arising from cascade testing of the family members.

Also a personal file containing information on FH along with a cumulative results sheet will be created and given to the Index case. Patient held records will contain comprehensive personal records of their results and treatments. This has further advantage to the patient and their family, as due to their condition being inherited they will require continuity of care. The patient held record will facilitate information sharing within the family, as it will also contain relevant information leaflets and genetic test results where available.

Involving the proband in the record keeping helps the health professional to get more accurate family information for diagnosis and treatment for the future generations.

Cascade screening:

FH is an autosomal dominant disorder which means that it is passed on through generations with a 50% chance of inheritance; therefore it is important for cascade screening of families to be carried out. This will primarily take place in the Family Support Centre (see section on FSC). The specialist nurse will have taken contact details of family members during the lipid clinic appointment of the index patient. He / She will then write to relatives asking whether they wish to be involved in family screening. If they agree, a full cardiovascular risk assessment will be carried out by the nurse. In the presence of Simon Broome criteria, a letter will be sent to the GP asking for a referral to a local lipid clinic. If the family member is outside of the sector, the nurse will write to them stating that they should visit their GP to have their cholesterol checked. DNA mutations can also be performed as part of the cascade screening process as long as a specific mutation has previously been identified in the proband.

Documentation will all be placed in the relevant family file (or a new file)

Management:

The FH specialist nurse should have an in-depth knowledge of cardiovascular risk factor management and should be aware that and risk calculators, although these are not to be used in patients with FH, as it is assumed that they are already at high risk and should be offered a high-potency statin with the aim of reducing the LDL-C by 50% of the

original levels. The specialist nurse will follow the medicine management guideline in case of statin intolerance or for use of additional lipid lowering therapy. The decision made by the specialist nurse in the lipid clinic and recommendations are communicated to the GP. General lifestyle advice is also offered at this point. Once appropriate cholesterol levels have been achieved and compliance to treatment is good, the patient is reviewed annually.

Family Support Centre (FSC):

The FSC links in with the lipid clinic but has other functions all of which are co-ordinated and performed by the FH specialist nurse.

- Cascade screening of relatives from the lipid clinic.
- DNA mutation screening of either index patients or their relatives.
- General advice given to people who have concerns regarding cardiovascular risk, lifestyle factors, drug treatments and / or side effects.
- Organisation of both educational and fundraising events.
- Co-ordination of a patient focus group who aims to raise awareness of inherited cholesterol conditions. This is achieved by assisting with the educational / fundraising events, assisting in media coverage, working pro-actively with healthcare professionals and both local and national lobbying.

The Specialist nurse in FH would act as a contact for family members both nationally and internationally regarding their hyperlipidaemia.

The FH specialist nurse forges links with many different organisations such as PCTs, Cardiac Networks, Lipid Networks, Genetic services and pharmaceutical companies.

Liaison with local GP surgeries is important as this helps to not only forge productive relationships but to help raise awareness of FH and the FSC.

Data collection:

Demographic and clinical data are held locally on an encrypted system, and used as an aid for cascade screening. The data is treated with strict confidence in line with the data protection policy.

Skills and knowledge set for health professional managing people with FH

- Interpretation of family and medical histories to assess the chance of disease occurrence
- To become sufficiently familiar with the many components of the medical record in order to be able to find specific data and to recognise what is most likely to be pertinent to the genetic issues
- To develop knowledge of medical terminology format and content of medical and genetic histories
- Knowledge about the natural history of FH Description of phenotype inheritance pattern, genetic testing , management options, prevention , support resources and research
- To construct a pedigree appropriate to the to calculate the risk, recognise gaps in information and/or misinformation
- Counselling to promote informed choices in view of risk assessment, and also ethical values.
- Presenting information in a non-judgemental way relevant to the needs of the client.
- Facilitate use of genetic information in a personally meaningful way minimising psychological distress and increasing personal control
- Understanding that the diagnosis and information discussed may have implications for other family members and its impact on relationships and family dynamics
- Have the understanding that information on Genetic diagnosis if given to the other family members or outside agencies such as insurers or employers has the potential to result in discrimination and stigmatisation so this issue should be discussed during consultation.
- Understand family function -denial, blame, communication difficulties marital problems, family members with different attitudes to a particular issue
- Understand ethical legal confidentiality and management issues
- To develop awareness and understanding of the moral and ethical issues in particular cases
- To recognise when referrals are appropriate both in relation to referral source and nature of the problem and to ensure that the health professional understands the reason for referral
- To construct a pedigree appropriate to the to calculate the risk, recognise gaps in information and/or misinformation
- To anticipate potential counselling problems during the telephone inquiry or the initial meeting that might interfere with the exchange of information language problems , doubtful paternity etc
- Accurate record keeping, literature and database searches
- To be aware of current and developing technologies in all areas of genetics.
- To become empathetic, sensitive, non judgemental and non directive recognise personal limitation seeking additional help where appropriate
- To develop the ability to present all options fairly, accurately and non directly
- To understand the importance of research and to participate where appropriate Understand the implications of enrolment into research projects the role of registers, implications of inclusion into a register, confidentiality issues, insurance implications of testing and testing of children
- To develop the ability to cope emotionally with responses of clients especially unexpected ones anger towards the health professional guilt at having been the one to pass on a harmful trait.
- To learn the technique of conveying bad news
- To recognise people's defence mechanism and decide when to leave them intact
- To develop self awareness about attitudes towards race moral issues social class religion
- To become a lifelong self directed learner who is aware of the available educational resources how to access them effectively and how to critically appraise their usefulness
- To be a willing and enthusiastic participant in education activities for other health care professionals students at all levels and the public
- To work in a multidisciplinary team and within the organisation
- Contact with patients may involve pre-clinic contact, clinic consultation, follow up/review

Pre consultation contact:

- Outline or review the purpose of the consultation as well as explain the consultation process and aims including possibility of physical examination so that concerns that the client may have regarding the nature and conduct of the consultation may be addressed.
- Ascertain needs and expectations of the client/family their agenda identifying any special requirements including social/ cultural issues that may impinge on the consultation identified later.

Preparation and consultation:

- Check all information needed for the consultation has been obtained
- Review relevant health record
- Discuss case with colleagues as necessary
- Review relevant medical literature
- Prepare information that will be given to the client including support group information (FSC) and fact sheets as applicable
- Consultation room and examination facility should ensure total privacy and child friendly. Children should be examined in presence of a parent or guardian by professionals authorised to see children. Female subjects should have another female present as appropriate if examined by a male health professional
- There should be a limit on the number of professionals present during consultation. Rapport with the family should not be compromised by the educational needs of the health professional and trainees. Consent should be obtained from the client for other professionals, such as students and trainees, to be present at the consultation.
- Gathering specific information – several ways - referring doctor, hospital medical records laboratory. Should be aware of confidential nature of medical information and obtain consent
- Informing the client how the information will be stored and who will have access to it
- Making and verifying the diagnosis by history taking physical examination and use of information obtained before or during the consultation.

- Providing information in a non biased way using language and concepts that clients understand about the condition pattern of inheritance, natural history, complications, and treatment options. Discussion of the limitations of current information and/or tests and the offer to review probands/relatives as appropriate.
- Once a diagnosis is made in a family individual risk is estimated using empirical data, inheritance patterns, and pedigree information clinical expertise and test results. They should also be informed of the risk interpretation and the limitations of risk calculations.
- A genetic diagnosis often has a profound impact on the individual and their family, this should be acknowledged and clients must be given a safe environment in which to express their emotional / psychological responses. Emotional support should be offered to clients during consultation and follow up
- While offering genetic tests the proband/family should be informed about the nature of the sample required , appropriateness of the test , the information the test is seeking, the limitations of the test and possible implications of the test results. It is important for the proband or relatives to know how long results will take and a contract should be agreed for arrangements to give the results.
- The appropriate request forms should be completed and a system of follow up or tracking of outstanding results must set in place for efficient reporting and action.
- Any involvement of proband or relatives in research should be supported by local ethic committee approval
- Proband and relatives should be provided with educational material or means of obtaining educational material fact sheets leaflets brochures and internet addresses which are linguistically and culturally appropriate.
- Discussing the medical emotional and social implications of the individual and family. A genetic diagnosis/ or test may have direct risk implications for other family members and counselling should be available as appropriate.
- Considering and discussing with the proband implications for genetic relatives.

- Presenting options including genetic testing and assisting with informed decision making in non judgemental/non-coercive manner
- Arranging genetic tests after obtaining consent, conveying and explaining results, referring to other health professionals as needed addressing interest of third parties
- Providing educational material and/ appropriate references
- Offering contact with community based support groups or persons
- Record keeping

Family files should be separate from the proband's medical notes, for the following reasons- confidential nature of genetic diagnosis, need to maintain the record for future generations, need to have complete and comprehensive file for each family . With the proband and relative's consent the relevant correspondence and reports should be sent to their current health care providers. As per DOH good clinical practice policy, information that additional family file availability should be documented in the notes. With access to this restricted to authorised personnel.

Pedigree drawing:

Taking and documentation of the family history is essential and fundamental skill required by the FH Nurse. In addition to recording relationships and health of at least three generations a thorough history includes information that may contribute to the later management of family/individual. Internationally recognised pedigree genetic symbols should be used in order to be able to interpret findings over time and also over distance.

Phone consultation:

Records must also be kept of all telephone consultation and letters If there are no existing documentation on a client should follow policy for such contact records.

Confidentiality of records:

- Proband/ relative records are confidential and subject to relevant professional standards or legislative requirements related to data protection act and release of information should follow appropriate

legislation.

- In general information about a person must not be released to a third part without written or documented consent of the person or guardian
- If information about a deceased individual needs to be obtained consent may be sought from the next of kin
- If information about an individual who is intellectually disabled needs to be obtained, consent may be obtained from a legal guardian
- If information is to be conveyed to other health professionals consent must be obtained.

Improving clinical practice:

- The continual improvement of clinical practice is important. All aspects of the service must be reviewed: waiting times (urgent and routine), phone consultations and enquiries, documentation and outcomes, face to face consultation including pre- and post- clinic discussions, follow up phone calls, letters to probands and relatives and health professionals as well as referrals to other professionals
- Satisfaction questionnaire
- Service liaison with genetic laboratories
- Maintenance of professional standards
- Regular staff appraisal
- Maintenance and review of database(s)
- Review of protocols and guidelines
- Review of associations with outside agencies
- Review of adverse incidents /event.

References

Recommendations for standardised human pedigree nomenclature
Journal of genetic counselling 4(4) 1995

Practical guide to the family history by Robin Bennett Wiley Liss New York 1999

Appendix A

Simon Broome Diagnostic Criteria for FH

A diagnosis of definite FH requires:

Cholesterol above 7.5mmol/l or LDL cholesterol above 4.9 mmol/l in an adult. Cholesterol above 6.7mmol/l or LDL cholesterol above 4 mmol/l in a child under 16.

PLUS

Tendon xanthomas in patient or a 1st degree relative (parent, sibling, child), or in a 2nd degree relative (grandparent, uncle, aunt).

OR

DNA-based evidence of an LDL receptor mutation, familial defective apo B-100, or a PCSK9 mutation.

A diagnosis of possible FH requires:

Cholesterol above 7.5mmol/l or LDL cholesterol above 4.9 mmol/l in an adult.

Cholesterol above 6.7mmol/l or LDL cholesterol above 4 mmol/l in a child under 16.

PLUS:

Family history of myocardial infarction (MI): Before 50 years in a 2nd degree relative

or below age 60 in a 1st degree relative.

OR

Family history of raised total cholesterol: Above 7.5mmol/l in adult 1st or 2nd degree relative or above 6.7mmol/l in child or sibling aged under 16 years.

Please note:

Cholesterol/LDL cholesterol levels should be those that were recorded before any cholesterol lowering medication was started. If this is not possible, then use the highest that was recorded on treatment.