



British Heart
Foundation

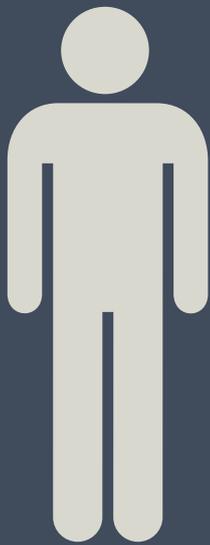
DELIVERY OF FAMILIAL HYPERCHOLESTEROLAEMIA SERVICES:

Identifying Enablers and Barriers



**FIGHT
FOR EVERY
HEARTBEAT**

bhf.org.uk



34
MILLION

people around the world are affected
by FH. The majority of whom will
NOT have been diagnosed.

I. EXECUTIVE SUMMARY	2
2. INTRODUCTION	7
2.1 FH and the burden of the disease	7
2.2 NICE guidelines and cost analysis	7
2.3 The BHF programme	7
2.4 Evaluation and analysis	8
3. THEMATIC ANALYSIS	9
2.1 Services and Pathways	9
2.1.a Variations across sites	9
2.1.b Referrals and contact method	10
2.1.c Key messages	11
2.2 Testing and databases	13
2.2.a Genetic testing	13
2.2.b Utilisation of PASS and other software	14
2.2.c Dealing with VUSs	15
2.2.d Paediatric register	16
2.2.e Key messages	16
2.3 Establishing and improving relationships	18
2.3.a Champions of FH	18
2.3.b Managing and forming relationships	20
2.3.c Engagement with primary care	20
2.3.d Key messages	21
2.4 Training and education	22
2.4.a Conferences and education sessions	22
2.4.b Training for nurses	23
2.4.c Use of resources and patient engagement	23
2.4.d Key messages	24
2.5 Enablers and barriers to cost, data and sustainability	25
2.5.a Risks to sustainability	25
2.5.b Expenditure	25
2.5.c Priority setting	26
2.5.d Evidence and data sharing	28
KEY RECOMMENDATIONS	30
4. APPENDIX	32
References	37

EXECUTIVE SUMMARY

Familial Hypercholesterolaemia (FH) is a genetic condition affecting approximately 1 in 250 people, resulting in high cholesterol levels from childhood and a high risk of early heart disease. Children have a 50 per cent chance of inheriting the conditions if one of their parents has FH. In the UK, FH affects an estimated 260,000 individuals, whilst globally affecting 34 million, the majority of whom will not have been diagnosed.

In 2008, the National Institute for Health and Care Excellence (NICE) in England published guidelines recommending that genetic testing is provided for individuals suspected of FH as well as their family members. Although NICE guidelines and low cost per QALY are seen as powerful incentives, genetic testing is not offered widely at present and services are patchy across the UK. The situation is further fragmented in England where the responsibility for commissioning FH services is divided amongst 211 local Clinical Commissioning Groups.

The BHF funding to support implementation of FH cascade services across the country have brought to light a variety of local and system-level barriers and enablers that are contributing to the current variation in access to services across the country.

SERVICES AND PATHWAYS

The majority of current services are secondary care consultant-led services where FH patients are managed and reviewed by lipid consultants.

This is not the case in areas where lipid clinics are not well established. Paediatric services are also varied in their remit and delivery but primarily delivered jointly by a paediatrician, lipidologist and/or the FH nurse. Referrals into services have been cited as a rate-limiting step in cascade testing in areas where services only accept referrals through lipidologists. Some areas have no access to lipidologists which has meant that key referrals are not reaching cascade testing services. Access to the FH patient care pathway for individuals of different ethnic background has not yet been addressed, however, the issues primarily lie within the lack of engagement by these groups.

Methods for contacting relatives of index patients vary across the sites where a mixed approach of direct and indirect contact is offered depending upon the patients' consent and preferred choice, however the general consensus is that direct contact is more effective and should be considered in the first instance. There is evidence to suggest that the nurse posts have had a positive impact on the delivery of cascade testing and has driven up referrals through a systematic approach. The main causes of concern however remain that a lot of referrals and tests are being accepted based on people's goodwill such as when out-of-area relatives are based in areas with no service provision, or when cost of testing is not covered by CCGs. Paediatric FH services also fall into a grey area where funding of service provision is not defined and people may slip through these gaps in the system.



TESTING AND DATABASES

Genetic tests for FH are offered in eight accredited labs across the UK and some have noted an increase of up to 50% in cascade testing over the last year or so.

There is, however, considerable variation in the methods used for genetic testing and the turnaround time for test results. It is felt that a national initiative should be considered on using fewer labs and utilising high throughput techniques which would drive down costs, allow better communication between labs and work as a sustainable and cohesive model nationally.

Genetic services are largely averse to the direct method of contact and acquiring patient consent for which they have been heavily criticised. Across services, variation has been reported in the criteria used for genetic testing where some services use the using the UK FH “Simon Broome” register criteria while others are using a much higher threshold for genetic testing. Stakeholders report that numbers are too large to consider when using Simon Broome and primary care would be burdened by this.

The PASS database has had a patchy uptake from the nations. It is currently used in about 90% of the BHF-funded FH services and a handful of non-funded areas, and it has gained popularity over the last couple of years, however, it still remains underutilised, and the uptake across the UK is inconsistent. It is fully implemented in Wales and now the Northern Ireland services are in the process of implementing it, but uptake in England remains fragmented and the services in Scotland are not inclined to use it because they have a system in place already. The recurring issues with PASS are centred on its governance, IT, and cost related to its single user per license policy. The BHF-funded PASS coordinator has been able to liaise with IT and information governance teams, bringing clinicians on board, which has worked well in most cases. This has, however, been a tedious exercise and there is a need for standardised protocols for use of PASS and its data.

A few tools exist on the market that can potentially be utilised to undertake case-finding in primary care however further clarity is needed around their effectiveness and at the moment no data exists on comparative efficacy and costs of different tools. These tools may address the issue of low pick-up rates and the time and effort needed for primary care case-finding.

When a Variant of Uncertain Significance (VUS) is found in one of the sequenced genes the result cannot be reported. The issue of how to deal with VUSs featured strongly in conversations on genetic testing, and there is consensus amongst stakeholders that a consolidation function, across the labs, is needed to record and classify VUSs in a systematic fashion, with clear follow-up protocols.

ESTABLISHING AND IMPROVING RELATIONSHIPS

The momentum gained around FH at a national and local level has primarily been the result of a few keen and engaged individuals and organisations: ‘the champions of FH’.

Despite FH services not featuring in the local priorities, it has managed to gain substantive traction over the last 10 years within pockets of the health system. Relationship building and networking appear to be key success factors in establishing and embedding these services. FH services around the country have cited examples of how previously established or long standing relationships within the healthcare community have facilitated promotion of FH services and driven up good quality referrals. In some instances where relationships did not exist, services were met with some resistance. Some areas were uncomfortable with a nurse-led model and had to be reassured on the competency of the service.

In small-knit communities and rural geographies, GPs appear to be more engaged with services however in almost all other areas it has been extremely difficult to communicate and gain access.

Time pressures and lack of awareness and/or interest in FH appears to be the most common reason for the response received from GPs. CCG engagement has been a big issue, with areas in England reporting back that commissioners are, on occasion, not able to engage even when there isn't a request for funding. Relationships with CCGs in many areas have been slow to establish and are further confounded by the ‘forever changing workforce’.

TRAINING AND EDUCATION

All the FH services are running education sessions and the nurses are attending conferences and training in some form or capacity. Primary care is receiving a lot of educational information through FH nurses which would otherwise not have happened.

There has been a significant increase in engagement due to the events and networking opportunities and a subsequent increase in referrals for FH testing. Networking opportunities have been abundant due to events organised by the BHF and HEARTUK and because funding is available to attend other events and conferences. Some stakeholders felt that although this has been extremely valuable, locally tailored regional and multi-disciplinary meetings are needed rather than big academic or insular meetings.

It is felt that the BHF training for FH nurses offers a good foundation to get started into the programme.

Locally, nurses have been part of joint education sessions and working in genetics and lipids has added to their knowledge. There is however a skills gap in their ability to write business cases and a lack of understanding on how to escalate or progress service optimisation issues. Clinicians also felt that FH nurses should be trained to address both paediatrics and adults services and some expressed concerns that front line staff are not confident about genetics. A consistent training programme which is easy to access is needed to address this skills gap.

BHF and Heart UK resources on diet, lifestyle and FH have been widely utilised by all services. The resources have been received positively by patients as well as the nurses, who found the resources educational. The stakeholders unanimously highlighted the gap in paediatric resources.

A pressing need for resources tailored to the younger demographic has been presented by all stakeholders.

ENABLERS AND BARRIERS TO COST, DATA AND SUSTAINABILITY

Risks to the sustainability of services and nurse posts have been highlighted across services which have been largely dependent on priority setting and expenditure.

CCGs are facing financially challenging times in the face of competing priorities and dwindling budgets. Most question the relevance of the service for their population as well as the likelihood of cash savings within their short budgetary cycle. Enablers such as BHF funding for nurse posts, Scottish government and Welsh assembly government funding for genetic testing have significantly progressed the case for FH however issues on further commissioning (in England) of services must be addressed. Presenting a strong case with evidence of clinical impact, effectiveness and robust cost modelling has helped in some areas but not all. Clinicians indicated that health economic modelling is helpful if it is articulated with complementary information such as average age of diagnosis, cost of set up and testing, number of MIs prevented etc. from a national perspective.

The argument on costs has featured significantly in the development of genetic testing in paediatrics with no specific guidelines on commissioning responsibility and tariffs.

At a local level, setting priorities for service provision is inextricably linked to costs and the data available to make the case for change. Although FH has gained priority at a national level through the efforts of the BHF, Heart UK, PHE, academics and representatives from the upper echelons of the NHS, it remains a low priority locally and for CCGs. NICE guidelines do not appear to be gaining traction in the local agenda setting and most commissioners are not willing to look at FH in isolation. Commissioners were not convinced that having a national directive for FH would be helpful given the multitude of priorities they have to tackle. They did however express a desire to have more information on the impact on primary care, local population prevalence, a balanced approach to shorter vs longer term efficiencies and risk sharing with other CCGs.

Access to data and opportune data sharing have resulted in significant benefits as seen in the case of cost savings and linking through PASS or delivering successful business cases. There are however glaring gaps and a lack of consistent culture of data sharing in the FH community as highlighted by key stakeholders. Operational inefficiencies have been highlighted where it has been difficult to access

data due to not knowing where to look or who to approach. Several services have struggled to develop robust business cases due to lack of knowledge on national and local level data, lack of access to data necessary for developing cost-modelling or relevant templates, and lack of expertise. Services from across the country have expressed the

need to be able to share evidence from their respective programmes as well as access learning and data from other programmes. Another key area where data is an issue is the loss of organisational memory and learning due to workforce migration.

KEY RECOMMENDATIONS

The national FH steering group and wider stakeholders should review and consider these recommendations to assess the best course of action for resolving the challenges in the sector around FH. Ultimately, efforts must be concerted to simultaneously address the key issues highlighted to achieve system-level change.

- A national exercise should be considered on assessing and selecting fewer accredited labs offering genetic testing for FH.
- National stakeholders should undertake a mapping exercise to ascertain where paediatrics features in the FH pathway and where the budget responsibilities lie.
- Services should discuss and collectively publish evidence on the efficacy of various referral routes between services such as primary, secondary and out-of-area services and build them into standard service delivery models.
- A standardised programme should be developed for FH nurses detailing training requirements to address the skills gap and ensure services across the country are consistently performing to a high standard.
- All relevant stakeholders within health services must tackle the issues around the ethics of directly contacting family members and accessing patient consent.
- National bodies should undertake an appraisal exercise to assess if PASS or another database will be beneficial to roll out across the nations with a concerted effort. A national toolkit must then be developed to help support its implementation by addressing issues on governance and IT.
- Guidelines should be developed for classifying and logging VUSs and services should negotiate for budgets to allow testing of relatives of those with VUSs on clinical grounds, where deemed appropriate.
- Services should develop a public engagement plan for FH to raise public awareness and outline benefits of treatment to combat the media hype against it.
- Services need to address the issue of equal access to and better engagement with FH testing across all ethnic minority groups
- Efforts should be concentrated nationally to develop approaches on FH cascade testing that can subsequently drive local efforts to championing FH.
- Services should develop and implement a consistent approach to gathering organisational learning so that workforce migration does not affect future negotiations in contracts and sustainability agreements.
- Stakeholders should consider the development of a national FH hub that will give users access to resources crucial for both existing and new services being established.

1. INTRODUCTION

1.1 FH AND THE BURDEN OF DISEASE

FH is a genetic condition affecting approximately 1 in 250 people, resulting in high cholesterol levels in the blood from childhood and a high risk of early heart disease.¹

As the high cholesterol levels occur at a very young age, it is not unusual for a person with FH to have a heart attack as early as in their 20s or 30s. Children have a 50 per cent chance of inheriting the conditions if one of their parents has FH. In the UK, FH affects an estimated 260,000 individuals, whilst globally affecting 34 million, the majority of whom will not have been diagnosed. If left untreated, at least 50 per cent of men with FH will develop coronary heart disease (CHD) by the age of 50, and 30 per cent of women by the age of 60.^{2,3} The risk of death from CHD can be 80 times greater with FH than without the condition at the ages of 20-39.⁴ Once individuals with FH are identified, treatment with statins can substantially reduce the risk of CHD in people with FH, restoring life expectancy to that of the general population.⁵

1.2 NICE GUIDELINES AND COST ANALYSIS

In 2008, the National Institute for Health and Care Excellence (NICE) in England published guidelines recommending that genetic testing is provided for individuals suspected of FH, cascade testing of relatives, and LDL-C treatment targets for those with a confirmed diagnosis.⁶

The NICE guideline was grounded on cost-effectiveness analysis which estimated that the incremental cost per quality adjusted life year (QALY) of testing people with suspected FH was at £2,676. This is significantly below the £20,000/QALY cost-effectiveness threshold used by NICE.

Since NICE guidelines were published, patents have expired on some of the widely prescribed statins used to treat FH, leading to reductions in treatment costs. Advancement in DNA sequencing techniques has reduced the cost of genetic tests and the turnaround time for results. Moreover, the establishment of FH cascade services in parts of the UK have offered the opportunity to collect real-time data. These findings have implications for the cost-effectiveness modelling originally proposed by NICE.

The British Heart Foundation (BHF) has commissioned an economic modelling exercise to update the previously proposed figures to inform services across the UK and to make a strong case for setting up FH cascade testing services more widely across the four nations.

The latest data from cost-modelling analysis indicates that the net cost and the incremental cost per QALY is highly sensitive to the number of relatives tested per index case. Details on the costings will be made available as the paper is published however preliminary findings show that the cost per QALY is well below the NICE threshold.⁷

1.3 THE BHF PROGRAMME

Although NICE guidelines and low cost per QALY are seen as powerful incentives, genetic testing is not offered widely at present and services are patchy across the UK.

The situation is further fragmented in England where the responsibility for commissioning FH services is divided amongst 211 local Clinical Commissioning Groups (CCGs), and the majority of the country has little or no provision for FH testing programmes.

The BHF funded a pilot scheme in Wales shortly after the NICE guidelines were published. With an initial investment of £450,000, BHF worked with NHS Wales and the Welsh Assembly Government with support from HEART UK and the Genetic Alliance, to provide a proactive interface between primary, secondary and tertiary care to support FH services. The service has since been sustained with funding from the Welsh Government. When cascade testing began in autumn 2010, Wales had just 97 known FH patients. At the end of the 3-year programme there had been 589 patients diagnosed and treated.

Following on from the success of the Welsh pilot, and the proven effectiveness of FH testing and diagnosis, the BHF granted two further waves of funding across England and Scotland, investing well over £1million to support the wider roll out and adoption of cascade testing.

The funding covered the 2-year cost of newly created posts of FH nurses on the proviso that the cost of genetic testing is covered by existing systems in place and that there is buy-in from CCGs to sustain the model after the BHF investment comes to an end.

INTRODUCTION

The map shows all known FH cascade testing services in the UK

The services listed below in red (marked with a star on the map) are currently or have previously been BHF-funded.

1. Bristol and Bath FH Service
2. Cornwall FH Service
3. Coventry FH Service
4. Doncaster & Bassetlaw FH Service
5. Dorset FH Service
6. Gloucester FH Service
7. Harefield FH Service
8. Heart of England FH Service
9. Manchester FH Service
10. North East Cardiovascular Network FH Service
11. Royal Free Hospital FH Service
12. Russells Hall Hospital, Dudley FH service
13. Peterborough and Stamford FH Service
14. Sandwell and West Birmingham FH Service
15. South Devon FH Service
16. South Yorkshire and North Derbyshire FH Service
17. Walsall FH Service
18. Wessex FH Service
19. Northern Ireland FH Service
20. NHS Grampian/NHS Tayside FH Nurse
21. Western Isles (FH Nurse)
22. West of Scotland Genetic Service (Glasgow)
23. East of Scotland Genetic Service (Dundee)
24. South East Scotland Genetic Service (Edinburgh)
25. Wales FH Service



1.4 EVALUATION AND ANALYSIS

The BHF funding to support implementation of FH cascade services have brought to light a variety of local and system-level barriers and enablers that are contributing to the current variation in access to services across the country.

To synthesise and highlight the emerging issues and inform senior decision makers within the healthcare sector, the BHF commissioned a qualitative evaluation to gather evidence from key stakeholders involved in delivering, planning and commissioning services.

The evaluation data is comprised of primary and secondary research. Primary data is presented as a thematic analysis of 28 semi-structured qualitative interviews and secondary data has been obtained from national FH meetings and national and international peer-reviewed literature. Further information on the evaluation methodology can be found in the Appendix. Reasonable care has been taken to verify the accuracy of the information provided by respondents however it is not possible to warrant the completeness of the information.

2. THEMATIC ANALYSIS

The analysis of the fieldwork has revealed key themes which have been classified into five broad categories with interdependencies and overlap in several areas which is explored in the sections below. While some themes touch upon issues and developments that are operational in nature, all themes broadly reflect upon the system-level enablers and barriers encountered across the services.

2.1 SERVICES AND PATHWAY

FH services can be largely categorised into three distinct models of delivery⁸:

- A specialist led model with patients' annual reviews based in secondary care
- A primary care led model where full responsibility for all FH patients, including annual reviews, sits with the GP
- A dual care model where primary care is responsible for on-going patient management and referral coordination, whilst specialist advice and genetic testing is carried out in secondary care

Although most services can be classified within the categories listed above, in reality the lines around these models of delivery are significantly blurred with the emergence of nuanced models of delivery.

2.1.a Variations across sites

The evidence emerging from the fieldwork suggests that delivery models are dictated by the existing infrastructures and workforce capacity rather than an appetite for major change or redesign.

A major influencing factor in how the services are delivered is the placement of the FH nurse post. This varies significantly across regions and structures, where FH nurses are based in cardiac nursing, pathology services, genetics, joint lipids and genetics or GP surgeries.

The majority of current services are secondary care consultant-led services, where most FH patients are managed and reviewed by lipid consultants. In some areas where this is not the case this is due either to a lack of established lipid clinics or limited access to consultants -for instance in the Scottish Highlands. The type of work FH nurses are involved in can at times be dependent on their background, such as experience in paediatrics, cardiac nursing and genetics, which can influence the consultants to devolve responsibilities. Culture change has been cited as a factor where some parts of the system, such as genetics, are more familiar and comfortable with nurse-led models of delivery than others.

The paediatric FH clinics are primarily delivered jointly by a paediatrician, lipidologist and/or the FH nurse. A handful of sites bypass the paediatrician owing to the FH nurses' or lipid consultants' ability to handle paediatric cases themselves which usually takes the form of family clinics and provides a more convenient forum for families to get tested. In some cases this is also due to restricted access to paediatricians in areas with high levels of demand or in remote areas.

2.1.b Referrals and contact method

Referral routes vary significantly across services. The source of referrals (primary or secondary care) appear to have some dependence on pre-existing relationships, but also on awareness-raising work carried out in primary and secondary care by the FH nurses.

Some areas receive high volumes of referrals from coronary care units, cardiac rehabilitation services and percutaneous coronary intervention (PCI) services whereas others receive more referrals from GPs. Although all cascade testing services accept referrals from lipidologists, some services only accept referrals through lipidologists. In this case, all referrals from primary or secondary care must go through a lipidologist before reaching the genetic testing service. This variation in referral routes can be a rate-limiting step in cascade testing. Some areas have no or limited access to lipidologists, which has meant that key referrals are not reaching cascade testing services in a timely manner or at all.

Accepting direct referrals from other services is perceived to work better than models where referrals between two cascade services in different areas is not possible unless a GP referral is made. This latter model is sometimes seen as an impediment and results in people not engaging with services. Several sites have local agreements in place with CCGs, allowing them to bypass the GP and take in direct referrals. The GPs are then sent an information letter once test results have come through for the patient.

Methods for contacting relatives of index patients vary across the sites, where a mixed approach of direct and indirect contact is offered depending upon the patients' consent and preferred choice. Whilst most services report that direct contact offers better engagement and follow-up, some were unconvinced as there is little evidence comparing direct effectiveness of the two methods for FH.

Some stakeholders expressed a need to generate evidence to compare the methods of contact to improve service efficiency and increase pick-up rate of relatives.



This is touched upon again in **section 2.2** in the context of genetic testing.

Most services are still dealing with pent up demand in the system, and are picking up cases of FH that are seen as the 'low-hanging fruit'. In a large proportion of these cases a clinical diagnosis has been made but cascade testing has not happened for other family members.

There is a view amongst stakeholders that unless proactive measures are put in place the referrals will eventually dry up.

Some sites are pursuing active case-finding in primary and secondary care. There is learning to be gleaned from these services to assess whether active case-finding makes for a more sustainable and effective service model. Research into this area could be a valuable asset when planning service delivery models. NICE is examining the cost effectiveness of different methods of finding new cases for primary and secondary care databases and will report this in the middle of 2017.

2.1.c Key messages

There is evidence to suggest that the nurse posts have had a positive impact on the delivery of cascade testing and has driven up referrals through a systematic approach.

Consultants and commissioners feel that the nurse posts are essential in delivering and shaping FH cascade testing. There are differences in opinion on the remit and specialist skillset required of the nurse role where some feel that having experience of paediatrics and genetics is advantageous whilst others feel that the role should encompass wider CVD or cholesterol pathway services.

Almost all sites report that administrative burden was greater than anticipated which meant nurses have less time to see patients. Moreover, a nurse working across different hospital trusts has a further impediment, owing to the need for repeated statutory training at each trust and onerous paperwork involved for honorary contracts. Physical barriers to running the service have also been cited, such as not having access to clinic space.

Although the nurse role is seen as key to delivering services, some stakeholders feel that services would have been easier to establish if cost of genetic testing for index cases had been funded instead of the nurse post. There is disagreement with regards to this amongst the stakeholders. The BHF's view is that systems are much harder to change therefore it is necessary for services to ensure some level of buy-in and some allocation for cost of genetic testing at the start, as the BHF's role as a catalyst would not have been enough to bring about a system-level change in multiple services at the set-up phase.

The issue remains that a lot of referrals and tests are being accepted based on people's goodwill, such as obtaining samples for testing from relatives who live in areas with no service provision or when cost of testing is not covered by their local CCGs. Paediatric FH services also fall into a grey area, where cost of service provision is not defined and people may thus be denied access to care and slip through these gaps in the system.

The discrepancies in the referral route to cascade testing, particularly around the need for GP referrals or lipidologist-only referrals, are often cited as barriers to better engagement with services and as decreasing pick-up rate of relatives per index

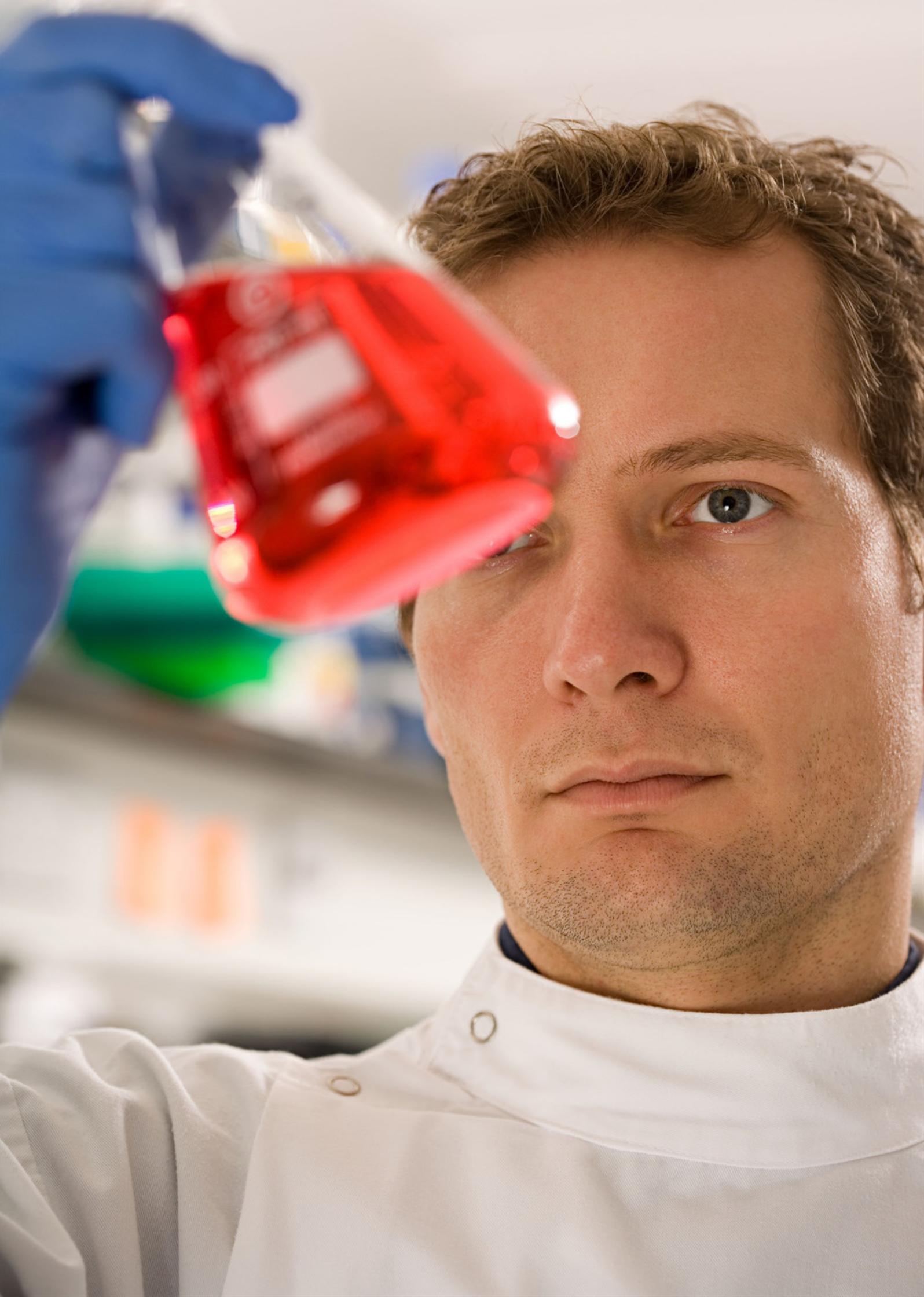
case. Services are keen to access learning from areas where local agreements exist that bypass these requirements, and to understand whether it results in a more efficient and engaged service. Further barriers to not reaching as many relatives as anticipated were lack of engagement due to complacency around FH (in relatives) and its perceived impact, relatives living in areas with no services, or where relatives are living outside of the UK, an important issue when the index case is a recent immigrant or from an ethnic minority group. Geography is also a significant issue in some areas, where one clinic may cover a vast area and patients have to travel several hours to attend clinics.

Some sites are pursuing active case-finding but have queried the value or impact of this approach. There is currently no systematic access to this data from sites implementing this approach, to compare pick-up rates and assess whether this is a worthwhile investment. Barriers to case-finding in primary care have been mainly on engagement with GPs and operational issues such as access to GP databases, clinic space and paperwork involved in honorary contracts. "We are offering to do this work and yet still they (GPs) are saying no. They said we are just so busy that it is difficult to even think about."

Although case-finding is happening routinely in some parts of the country it is felt that learning from such areas is not easily accessible, and that this is akin to re-inventing the wheel. Engagement with primary care is discussed further in **section 2.3**.

Many nurses have come into established posts and services with little knowledge of how services were set up. Without the insight into roles and responsibilities and what the mechanism is for bringing about change, it has been difficult for some to optimise their services. Some felt that this lack of defined governance and structures was unsustainable as it relied solely on few individuals pushing things forward but that this is now starting to change.

"A lot of personal enthusiasm on a shoestring has kept this going; it's not the most sustainable thing."



2.2 TESTING AND DATABASES

2.2.a Genetic testing

According to the UK Genetic Testing Network (UKGTN) database, genetic tests for FH are offered in eight accredited labs across the UK, whereas some institutions such as Professor Humphries' lab at UCL was previously offering genetic testing as part of a research programme.

Data from one of the larger labs shows an increase of up to 50% in cascade testing over the last year or so, however, there is considerable variation in the methods used for genetic testing and the turnaround time for test results. The advent of Next-Generation sequencing and high-throughput screening have cut down the cost of genetic tests and significantly improved processing times, nonetheless, to date only a handful of labs utilise these techniques.

The mechanism for acquiring services from a particular lab is unclear, as there does not appear to be a competitive procurement exercise required. This is in stark contrast to the experience of two sites; Birmingham and Greater Manchester, who are at different stages in the process of establishing services that cover vast and populous areas. The two areas were required to undergo an EU-wide procurement exercise for the genetic testing service based on the size of their service. This is largely uncharted territory for services, and it has slowed down progress and poses a challenge around longer-term sustainability in the post-Brexit climate.

Stakeholders have expressed the need for information on how many labs are used and what the minimum criteria are for using them. It is felt that a national initiative should be considered for using fewer labs and utilising high throughput assays which would drive down costs, allow better communication between labs and work as a sustainable and cohesive model nationally.

Currently, about two-thirds of the BHF programmes are having their genetic tests funded by CCGs or local health boards in the case of Scotland, however issues around sustainability remain and will be discussed in detail in **section 2.5**. Prior to cascade testing services, several parts of the country had been picking up FH patients based only on a clinical diagnosis, however having access to genetic testing has added robustness to the approach and allowed cascade testing to become established, which is

now identifying many children and relatives. A handful of services have had to fund genetic testing by finding money within their system and it will be difficult to sustain this on just on goodwill. A genetics lab cited a few examples where members of the public have approached them for access to testing who were from areas where there are no services. The lab put them in touch with the local lipid centres and reported that "We are now seeing more and more of this." Quite a few services have offered genetic testing to out-of-area relatives but not all services have the flexibility in their budgets to facilitate testing for out-of-area relatives.

Acquiring patient consent and the method of direct contact has been flagged as a concern.

Primarily, genetic services are averse to this method of contact and acquiring consent. There is comparative literature for breast cancer showing the superiority of direct contact, but the labs and services do not seem to be aware of this. A handful of services are either planning or would like to carry out research on the rate of engagement and relative pick-up rates when comparing the indirect and the direct method of contact. Some anecdotal evidence suggests that the direct method is more effective however the ethical implication of contacting someone "out of the blue" about their risk for a disease they may be unaware of is of concern. Some practical issues were highlighted, such as impact on people's ability to get life insurance or a mortgage. However, since people already usually know they have a family history of early heart disease, and are often aware that they have elevated LDL-C, the additional information of having a genetic confirmation of the disorder is very unlikely to influence underwriting practice. There was heavy criticism from outside the sphere of genetics that they (genetics) are basing this on the (untreatable) Huntington's model whereas FH is completely treatable and completely different

"People already know that they have heart disease in the family, you inherit the condition not the disease."

Across services, variation has been reported in the criteria used for genetic testing. Although some services use the Simon Broome criteria for the degree of elevation of total cholesterol > 7.5mmol/L, others are using a much higher threshold before recommending for genetic testing.

Stakeholders report that numbers are too large to consider when using the Simon Broome cut-off and primary care would be burdened by this. The proposed approach is to start at a much high cut-off to identify index cases who are most likely to have FH which will result in a high mutation detection rate and then identify the rest of the population through cascade testing so that services are selective about the type of work that is carried out in GP practices. Service delivery teams claimed that the 2008 NICE guidelines did not fully appreciate the consequence of recommending examination of everyone with total cholesterol of over 7.5mmol/L. On the opposite end of the spectrum, some services have reported seeing FH patients who have lower than expected level of total cholesterol and finding a significant number who carry an FH-causing mutation. Currently no evidence exists on the different approaches of service delivery and their efficiency or the return on investment.

2.2.b Utilisation of PASS and other software

For efficient and cohesive FH cascade testing services across the UK, having a database that connects primary, secondary and genetics services is a major requirement. At present, there is no single database that fulfils all these requirements.

The PASS database, which is currently used in 80% of the BHF-funded FH services and a handful of non-funded areas, has gained popularity over the last couple of years, however, it still remains underutilised and the uptake across the UK is inconsistent. It has been fully implemented in Wales and Northern Ireland are in the process of implementing it, but uptake in England remains fragmented and services in Scotland already have an established system and are not keen to incur additional expense or time needed to adopt PASS.

Services in Scotland are utilising the Aberdeen clinical genetics database. This database allows storage of patient information and has family pedigree functionality. Patient notes from the database are now being made accessible to GPs and consultants. However, reminders and work streams have to be managed manually. PASS was too expensive at the time when Scotland was considering a database and the cost per license model of PASS would not have been sustainable with a distributed service model. Although IT costs have fallen, Scottish services are apprehensive to consider PASS, as cost per license is still a barrier and the

genetics lab will also need the license to ensure a seamless flow of information. Their current database costs are minimal and the one-off fee covers all members of staff and encompasses all genetic conditions.

As with the Aberdeen database, PASS also allows patient data and notes to be stored on the system and has a family pedigree function. The added value of PASS comes from its unique shared workflow functionality that links up all FH services and labs that use PASS. It allows services across the country to link up and identify when a mutation has already been reported in a family. This allows services to save money on genetic testing, and according to staff it only takes a couple these instances of families being linked per annum to make up the cost per license. All stakeholders agreed that there is no substitute for the workflow functionality in PASS.

“Linking up nationally is great and its working and we share family members across other services.”

The recurring issues with PASS are centred on its governance, IT and on the cost due to its single user per license policy. Most services have multiple users on single licenses and the PASS coordinator has developed SOPs for data entry and modification. Audit trail is an issue on one license and work is currently in progress with the development company to enable shared workflows for multiple users. The governance of data can be an issue, as data is shared outside a given hospital trust due to it being a national database, however the BHF-funded PASS coordinator has been able to liaise with IT and information governance teams, bringing clinicians on board, which has worked well in most cases. This has been a tedious exercise and there is a need for standardised SOPs around use of PASS and its data as well as mechanisms for embedding it into hospital systems.

Further criticism of PASS has been on data sharing and lack of integration with primary and secondary care systems. Stakeholders made an emphatic case for hospital and GP systems to be able to ‘talk’ to PASS. In Wessex there are plans (pending ethical approval) to extract data from GP practices and look at follow up, annual reviews, medication optimisation and other routine data to marry up with data from PASS on referrals routes and pick up rates, etc.

A possible impediment to this work is that most services are not utilising PASS for collecting routine data as it is slow and rather cumbersome to extract data from, it therefore services prefer using their own local systems to log and extract data. Moreover, the local systems contain years

of historic patient data which is not on PASS, making it quicker and easier to work with local systems. An urgent need for labs as well as consultants to have access to the PASS software for sharing data and updating VUSs has been highlighted. Areas where this is happening have reported that referrals, test results and follow ups are much more efficient.

As mentioned earlier, a few services are moving into the phase of active case-finding as they get close to completing genetic testing on indexes and relatives historically identified by lipid services. The FAMCAT tool⁹ developed by University of Nottingham is one of the tools being considered however further clarity is needed around its effectiveness in primary care.

The Medway report¹⁰ refers to the Medway audit tool for FH but it is unclear how well-utilised it is at the moment and no data exists on comparing its efficacy against FAMCAT or other equivalent tools. These tools may address the issue of low pick-up rates and the time and effort needed for primary care case-finding. Feedback from commissioners indicates that these tools are expensive to implement, however analysis comparing other methods of case-finding with the tools may allow services and commissioners to see longer-term benefits, both clinical and monetary. NICE is examining the cost effectiveness of different methods of finding new cases for primary and secondary care databases and will report in the middle of 2017. For active identification in secondary care, North Wales implemented an automatic alert system where anyone coming into the hospital with high LDL levels is flagged to the FH service.

“There is a lot of movement of people with high cholesterol within the hospital and that can be a good way of unmasking FH in people if done carefully.”

The BHF-funded role of PASS coordinator has been a catalyst for embedding PASS into services and providing continual training on its use. This role is also a mediator between services and the development company in negotiating system improvements and correcting glitches etc. In collaboration with HEART UK, the coordinator role has raised awareness of PASS and its potential benefits, and there are speculative proposals on handing over PASS to NICOR which can act as a lever for highlighting benefits of PASS nationally. Although the coordinator role is funded to primarily work with BHF-funded services, there is merit in working with non-funded sites in promoting PASS, which may help improve uptake and subsequently improve connectivity within the BHF sites as well. There

are indications that the BHF may develop a business case toolkit for PASS prompted by different areas experiencing the same issues. A toolkit may address some of these issues and help incentivise uptake in other services currently not using PASS.

2.2.c Variance of unknown significance (VUS)

There is consensus amongst stakeholders that a consolidation function, across the labs, is needed to record and classify VUSs in a systematic fashion.

It has been highlighted that staff based in genetics are not always aware of historic pathogenic classification of VUSs. Professor Humphries has been involved in classification of VUSs and through his knowledge of the literature was able to reclassify about 30% of the VUSs that had been reported back from the labs in the recent months. However, there is no consistent mechanism for doing this or feeding this information back to the labs.

Segregation studies are not a priority in several local areas because of funding issues. The feasibility of conducting segregation studies can also be a barrier, for instance relatives are often dispersed across vast geographies and offering genetic testing to family members of someone with a VUS is not covered within existing budgets for genetic testing. Areas where flexibility in budgets exists or there is a research budget allocated, are able to conduct segregation studies if the majority of family members reside within the given region. Some areas are now drafting protocols to deal with VUSs and conduct studies. Services that are not able to conduct segregation studies often flag and discuss VUSs at MDT meetings and notify patients if a VUS is later classified as pathogenic, allowing them to extend genetic testing to relatives.

“We try to see all patients with VUSs to explain results but we are not doing any further testing for segregation as we don’t have funding for that. We advise them to get their relatives to have their total cholesterol checked.”

A lot of services do not have standardised guidelines to deal with VUSs once they are classified which is a serious concern. Services and labs have expressed a need to ensure that some guidelines are established that can be implemented across the country. There is some apprehension and a general lack of knowledge on VUSs in the healthcare professional community.

2.2.d Paediatric register

The paediatric register, initially funded by the BHF, hosted at the Royal College of Physicians and directed by Professor Humphries aspires to register all the children diagnosed with FH in the UK.

Based on the prevalence figures of 1 in 500, it is estimated that 28,000 children under 18 years in the UK could have FH however the register currently has 380 logged cases.

Paediatric cases are dealt with some variability across services as mentioned in previous sections. Since responsibility for testing children for FH is sometimes split across paediatricians, lipidologists and the FH nurse, the subsequent logging of data on the paediatric register is not systematic. In some areas it is the nurses that enter the data whereas in other areas it is the paediatric consultant (if one is available). Consultants find it cumbersome to access the register due to constant password changes (necessitated to ensure security of the database) and time restraints, whilst some questioned if this was the best way to extract patient information. Although the majority of the services are aware of the paediatric register most expressed a lack of clarity on the utility of the register as well as future use of data.

Some stakeholders were more aware of the potential use of the register; "We assume we are doing the right thing by putting children on statins, and by having them on the register we can follow them through longitudinally and be convinced that they have avoided an MI for example, and they have benefitted from these treatments."

The team maintaining the register is aware that consultants are busy and don't always fill the data in. Feasibility to hire staff to move around the country and review doctors' notes and log children was considered, but it was felt that it might be difficult to review hand-written notes and to gauge comprehensiveness of the information. Furthermore, some areas may have as little as five cases whereas some may have well over 50, therefore it would be challenging to judge the caseload. The team have been awarded a grant to roll out the register across Europe and Australia and to develop information sheets and videos for children to highlight the importance of FH services for children.

There is a perception that paediatricians are generally not interested in FH, as it is a very small part of their client base and they mostly see metabolic disorders. Although

evidence does suggest that a significant proportion of children are seen in adult services, there is also evidence to suggest that some areas are now seeing more cases of children with FH than metabolic disorders and the demand for these services has grown steadily over the last five years. There appears to be a clear gap in children's services, with inconsistent access to paediatricians and family clinics. Despite clear guidance on this from NICE in 2008, there is current debate on what should be the appropriate pathway for children and what training and support needs have to be met to deliver this. There appears to be no national consensus or recent guidelines around this. Ultimately there is a sense that paediatric services need to grow and more children need to be tested and logged on the register

2.2.e Key messages

Despite several barriers cited in the sections above on genetic testing and databases, there is positivity amongst most stakeholders that things are moving in the right direction.

There is agreement that conditions have improved for FH in terms of awareness and availability of genetic testing, however more work needs to be done to avoid losing momentum. One of the key issues around genetic testing appears to be the uncertainty on longer term funding for genetic tests. Cost of genetic testing is not covered consistently across the UK, as it is not part of specialised commissioning but rather falls into local CCGs' responsibilities, who are often averse to investing scarce funds for FH services. This will be discussed further in section 2.3 and 2.5.

It is worthwhile assessing whether eight labs, utilising variable techniques, are required across the country which subsequently affects speed of throughput and delay in obtaining a results. Long waiting times, especially when research labs are used, have left patients frustrated and on occasion waiting for up to a year for results. This is partly due to lack of local funding for genetic testing which has stretched existing budgets, and the only economically viable option has been to participate in academic research programmes to facilitate cascade testing. "If you look at the figures it looks like we have diagnosed a lot less now but it is because

of the time it takes to get results back ...whereas before we were diagnosing based on cholesterol results. We use a research lab because of the agreement in place around costs and with us not having PASS it's harder to keep a record and harder to see if people have engaged with the service."

Furthermore, the UKGTN is under financial pressure and being reorganised, therefore it is likely that smaller diagnostic labs may close. Professor Humphries and Professor Gray are planning on putting forward proposals that there should be at least two labs across the UK that offer FH genetic testing. An efficient system needs to be maintained, with a balanced demand and supply model for this to work, so that labs are not waiting months to receive an adequate amount of samples for batch processing.

A conservative approach to genetic labs that offer FH genetic tests should help embed consistent and high quality techniques for optimal sample processing.

Furthermore, it may allow areas to experiment with evolving approaches in genetics (e.g. Polygenic SNPs and VUSs) which could be included within the same price as current tests. Lipid teams have said this would prove useful and could also be a way for patients to get engaged with the Genomics England 100,000 genome project.

There appears to be a divide in the healthcare professional community on the method of contact and patient consent; genetics take a conservative view whereas the rest of the stakeholders are keen not to classify FH under the same umbrella as diseases like Huntington's and Cystic fibrosis. This may be combatted in the general public through raising awareness of FH and its treatment, so as to alleviate fears of getting diagnosed.

Apart from the issues highlighted earlier with PASS on its governance, IT and licenses, there is a wider debate about whether PASS would be better placed as a national database as part of NICOR. Moreover, although implemented in Wales, Northern Ireland and England, the national interfaces are not integrated at present. The workflow functionality has worked really well for some services but equally some areas have not reaped

any benefits, as many relatives of index cases live in places with either no service or no access to PASS. PASS becomes sustainable and cost-efficient only if enough services use it. Not having standardised guidelines on embedding and utilising the database has meant that some areas are struggling to get buy-in. In one example it was reported: "We are still trying to get PASS even though we have been trying for years-it took so long because we had to keep providing assurances around data sharing and governance. Then agreement was reached but that person left and another person refused and so on."

Although procuring a central database for the UK would be very challenging, the current systems need to be able to integrate with each other, providing seamless access to data for services, labs and primary and secondary care systems to work cohesively.

Genetic labs expressed a need for utilising PASS so as better to deal with classifications of VUSs, such as an electronic alert system in place to ensure consistent spread of knowledge, and standardised protocols on dealing with a VUS once it has been re-classified as pathogenic.

Some of the main challenges in paediatrics have been raised earlier but the most significant of them appears to be on the cost of genetic testing for children. There is contention around whose budget this would fall under, and the lack of clarity is causing children to miss out on access to testing. Some areas are unable to cover these costs either through paediatrics or through adult services, so children are left untested and without treatment. Concerns have also been raised on the appropriate pathway for children and what training and support needs have to be met to deliver this. Although this was covered in depth in the NICE 2008 FH guidelines there appears to be no accepted national consensus around this, which could go a long way towards addressing the discrepancies in children's services across the UK.

2.3 ESTABLISHING AND IMPROVING RELATIONSHIPS

2.3.a Champions of FH

The momentum gained around FH at a national and local level has primarily been the result of a few keen and engaged individuals and organisations: ‘the champions of FH’.

Despite FH services not featuring in the local priorities, it has managed to gain substantive traction over the last 10 years within pockets of the health system. The champions of FH have been a driving force behind this movement and the stakeholder analysis has shed some light on their dedication to the cause.

“People passionate about FH are driving this whole movement forward...nothing else.”

The BHF has been championing FH for years, mainly by funding the research that identified FH-causing genes and the subsequent diagnostic tests, and funding the Simon Broome FH Register, the 2010 National Audit of FH and the Children’s FH Register. Furthermore they have been raising the profile of FH through networking and collaborative working with PHE and leaders within the NHS. This has helped prioritisation within some national bodies. The CVD outcomes strategy published in 2013 had listed FH as a priority area but that did not lead to any significant momentum, given that FH services span multiple systems within a complex and constantly evolving environment. This resulted in further impetus for the BHF to fund a second wave of FH cascade testing services across the country in order to support wider buy-in and sustainability plans.

In Scotland, the movement came about through a group of lipidologists and geneticists who came together and developed a case for centrally funded genetic testing for FH. In Wales, it was the BHF-funded initiative and the patient voice that was the catalyst. South Wales had just a few established clinics with genetic testing available so the patients developed and took the case to the senate to establish a centrally funded and all-encompassing service which was then successfully established.

FH has locally gained traction in patches cross England through the work of FH champions driving the agenda forward against resistance within the system. The following

excerpts from stakeholders provide a snapshot of how the foundations have been laid and the case for FH services has been made:

“We have had movement on this from about 6 years ago. The cardiovascular network commissioned a health economics piece of work around costs for setting up services which made an argument to commissioners for services. We updated NICE costings as statins came off patent so it was a lot cheaper and made a powerful case. Luckily, some key people from the locality wanted to set up services and championed it to the commissioners - was able to join these zealots and make a contribution.”

“In 2004 we were fortunate to become part of a Department of Health pilot to improve diagnosis of FH as a research project looking at DNA testing. Once this finished I couldn’t just stop it so I found the money for it with great difficulty. The genetic testing is funded by me through soft money generated by other means and when this funding dries out the services may stop.”

“I linked up with lipid consultants at an educational evening many years ago and we came up with idea of building a regional FH service-I volunteered to take this case forward to the consortium of CCGs translating the clinical case based around NICE guidelines.”

“It all started when our medical director, a cardiologist by trade who had a personal and professional interest in FH, got us to bring together lipidologists in the region and representatives from tertiary centres, adult and paediatric services to work out how we can work together to provide a standardised service.”

“The local SCN have pushed it forward and the GP CVD lead has been extremely engaged and has tabled it as a draft business case”.

The cases outlined above demonstrate that it is key individuals and the networks of like-minded professionals supported by organisations like the BHF and Heart UK that have put FH on the map.



2.3.b Managing and forming relationships

Relationship building and networking appear to be key success factors in establishing and embedding services.

For instance, the BHF have a long standing relationship with PHE and the National Clinical directors within the NHS through representation on their system leadership boards. This relationship has lent BHF influence in developing the CVD outcomes framework as well as shaping conversations around FH. The charity and the national bodies are utilising the systems leadership approach for FH to influence the system and the government, building on their experience of working together.

“We are a catalyst for change and our role is to facilitate the convening of different parts of the system.”

FH services around the country have cited examples of how previously established or long standing relationships within the healthcare community have facilitated promotion of FH services and driven up good quality referrals. The FH nurse background has also played an important role in promotion of services and driving up referrals from primary, secondary and tertiary care; nurses from cardiac rehabilitation, coronary care or PCI units have helped increase awareness of FH within respective services due to the interactions and pre-existing relationships between nurses and their old teams.

“We have very good relationships through the cardiology background of the nurse which has helped raise awareness of FH cascade testing and driven up referrals.”

Even in the case of commissioning, pre-existing relationships and awareness of FH have gone a long way in gathering support for sustainability of services. “We brought up the issue of permanent nurse posts with the CCGs and they were supportive as they already knew of these services from their previous PCT days and that has helped pushed the rhetoric of having FH cascade testing”.

In many cases relationships have had to be established to improve connectivity with other services and to make them aware of the FH service being offered. Nurses have forged relationships with other healthcare professionals not only to improve service uptake but also to help organise educational events and ensure high levels of attendance. Working with new services is seen as a positive

in many cases, especially where gains have been made in learning and development– “working with lipids and genetics is interesting and more holistic in terms of patient management.” Establishing new relationships yielded positive outcomes such as better engagement with GPs, connectivity with wider services based on SCN’s existing links as well as engagement with AHSNs, who are open to have further discussions on collaborations.

In some instances where relationships did not exist, services were met with some resistance. Some areas were uncomfortable with a nurse-led model and had to be reassured on the competency of the service. Similarly, when services had to be run across different hospital trusts, it was easier to do it at nurses’ home-base hospital trust due to existing links. Stakeholders also reported on the laborious process of establishing links with the managerial side of commissioning as well as with financial directors of trusts. It was evident that services ran more smoothly and efficiently once relationships had been established.

In Wales relationships appeared to be easier to establish and maintain given primary and secondary care have the same employer as opposed to a competitive commissioning landscape.

The services in Northern Ireland have developed largely independently to the other nations however key BHF staff have been involved in helping the setup, sharing best practice and delivering training on PASS. The links between Northern Ireland and other national services, however, are largely amorphous at this stage.

2.3.c Engagement with primary care

Contrasting experiences have been reported when it comes to engaging with GPs and CCGs.

In small-knit communities and rural geographies, GPs are well informed and engaged with services however in almost all other areas it has been extremely difficult to communicate with them. Time pressures and lack of awareness and interest in FH appears to be the most common reason for the response received from GPs. Despite being offered help for case-finding by FH nurses, GPs either don’t have the desk facility/room, spare surgery time or they are so inundated with work that they are unable to respond to these requests. GPs have come under further criticism about not grasping the wider implications

of FH, such as testing children, , and were perceived to be treating FH as an individual's condition not a condition that affects the whole family. There were debates in the clinical cabinet around how much work this could possibly generate for primary care and there is some apprehension from GPs on paying for the cost of genetic testing. This has largely stemmed from the view that if a patient is on statins then there is no further need for testing however this is not to say that all GPs hold this view. Massive improvements are being reported in the understanding of wider implications of FH amongst GPs over the last two years.

“A lot of GPs now ring us and ask for advice so engagement and relationships have progressed much further.”

CCG engagement has been a big issue with areas in England reporting back that commissioners are on occasion not able to even discuss promotion of existing services despite where no requests are made for funding. Relationships with CCGs in many areas have been slow to establish and further confounded by the ‘forever changing workforce’.

Cases have been reported of where services have had to communicate with various CCGs to go into GP practices for index identification which has turned into a ‘logistical nightmare’. Governance, IT, SOPS and confidentiality agreements are at the core of the issue. It was felt that none of these issues are new but the CCGs are not aware of the best way to deal with them not having come across them before.

Another big challenge has been identifying who the appropriate contact is within a CCG. “Just getting your foot in the door is an issue in the first instance...not easy to know who to go to.”

On the contrary, commissioners felt that relationships are reasonably established but alluded to the constant turn-over in the workforce. “I think they are frustrated that things haven’t progressed as fast as they would like but it is challenging for us.”

On the positive end of the spectrum, a couple of CCGs acknowledged that there was no systematic testing and case-finding and have been very supportive of a business case proposal. In these cases the commissioners have met and advised regularly on how to compile the business case and provided further support in the form of CCG personnel’s time to assist with economic modelling.

2.3.d Key messages

Relationships and networking is fundamental to gaining traction and buy-in from wider services as well as commissioners.

Previous understanding, exposure or links with FH appear to facilitate better promotion of services and increase referrals for cascade testing. Where relationships have not been established it has been more difficult to assert the utility of the FH service, and to discuss the establishment or sustainability of the service.

Champions of FH have been a primary driving force behind the movement on making FH genetic testing available across the UK but there are significant risks to this model. Lack of a robust infrastructure around services and the source of funding can easily result in dissolution of existing services. As clinicians and other professionals retire there will be a significant loss of momentum and knowledge on these issues if steps are not taken to formalise local arrangements.

Engagement with primary care has continued to be a significant barrier for services in improving visibility and embedding sustainability plans. Involving GPs has been difficult due to time pressures and the lack of clarity around primary care’s role in the FH pathway. Progress has been made where GPs have been interested in FH and feasible arrangements have been made with nurses to facilitate primary case-finding.

Building relationships with CCGs on the other hand has been more difficult due to the financial pressures they face, the competing priorities in the local agenda setting and the frequent movement of the workforce. Only in circumstances where historic relationships have existed, services have managed to have productive dialogues around cost of funding and longer term running of services.

2.4 TRAINING AND EDUCATION



2.4.a Conferences and education sessions

All the FH services are running education sessions and their staff are attending conferences and training in some form or capacity.

The majority of the services are targeting primary care through practice nurse forums, cardiology events and GP study days. Primary care is receiving a lot of written material through FH nurses, which would otherwise not have happened. A lot of time has been invested in increasing awareness locally within the healthcare professional community (e.g. GPs, pharmacists, practice nurses), and services have been promoted at primary care conferences, BHF and HEART UK events and at the British Cardiovascular Society annual meeting.

Response from primary care has been positive, resulting in valuable gains in learning. Stakeholders have felt that as a result of this training and education, GP services are delivering the best and most relevant referrals. There are plans in areas to gradually increase the out-reach to primary care, and to use engaged GPs as champions of spreading the message to gain further traction within primary care.

Education sessions have also been delivered to cardiac services in secondary care, resulting in more referrals. Sessions in coronary care have been centred on making services aware of the referral process for FH, and to allow systematic index identification. A handful of areas are planning a regional event to bring together

GPs, lipidologists, commissioners and hospital trusts to disseminate the FH story and spread the word regionally.

There has been a significant increase in engagement due to the events and networking opportunities and a subsequent increase in referrals for FH testing. Networking opportunities have been abundant due to events organised by the BHF and funding available to attend other events and conferences. Some stakeholders felt that although this has been extremely valuable, locally tailored regional and multi-disciplinary meetings are needed rather than big academic or insular meetings where “we are preaching to the converted”.

In areas where primary care has not been approached for training or education, the quality of referrals has been variable and services reported that there is a lot of to-ing and fro-ing involved. Concerns were also raised that many experienced doctors now retiring and their successors do not have the appropriate awareness or training on treating FH. Time has also been an impediment to delivering more education sessions. To combat this problem, one area has included FH on the mandatory training for hospital trusts to raise awareness in clinical staff.

The genetic labs also attend conferences and networking events where they share data and establish contacts, which acts to promote their services.

Overall, it is felt that there has been an increase in awareness around FH and testing but more work needs to be done on general public awareness as they are a powerful voice that needs to be better utilised.

2.4.b Training for nurses

The FH nurses revealed that networking events have been helpful in gaining and sharing learning with other nurses in the programme.

It is felt that the BHF training also offers a good foundation to get started into the programme. Locally, nurses have been part of joint education sessions and working in genetics and lipids has added to their knowledge. This has helped nurses engage better with families to explain results and treatment options. FH nurses have also had training days on motivational interviewing which they found beneficial for interacting with patients in clinic. BHF have also funded Masters in genetics for the FH nurses.

Some clinicians felt that FH nurses should be trained to address both paediatric and adult services. Some have also expressed concern that front line staff are not confident about genetics, and that a consistent training programme which is easy to access is needed to address this skills gap. There appears to be an emerging need to develop a tailored and standardised training programme for nurses that is applicable across the country to ensure consistent and high quality performance and to address the emerging skills gap.

There appears to be an emerging need to develop a tailored and standardised training programme for nurses that is applicable across the country to ensure consistent and high quality performance and to address the emerging skills gap.

“There is a belief that you need doctors to do this clinic, but you don’t...if you train the nurses well and develop them as lipid nurses it works well”.

2.4.c Use of resources and patient engagement

BHF and HEART UK resources on diet, lifestyle and FH have been widely utilised by all services.

The resources have been received positively by patients as well as the nurses, who found the resources educational. A handful of the services produced their own leaflets and felt that a more tailored approach was needed to promote the local relationships. Patients found the BHF and HEART UK resources interesting as well as easy to understand.

The stakeholders unanimously highlighted the gap in paediatric resources. A pressing need for resources tailored

to the younger demographic was presented by all stakeholders. Some mentioned a video hosted by HEART UK however felt that a printed resource is needed to help in a clinic setting. Based on interactions and feedback from patients, a need was also raised to develop some educational resources on use of statins as families have a lot of questions due to the conflicting media coverage around statins. “There isn’t much on statins to level out the media hype on risks of statins...we need something to reinforce the benefits in this case”. There is a need to separate the general statins debate from the use of statins in the FH cohort due to its unequivocal benefits.

Patients and families feel supported and satisfied with the service provided. Some patients are able to access a full CVD risk assessment depending on the service model, whilst others feel comforted by having a longer nurse appointment and being able to get in touch with the nurse at any point over the phone. Some people have been waiting a long time for their relatives to be tested, and receiving a firm diagnosis has had a positive impact on families. One nurse explained the impact of the service by highlighting one particular family’s experience:

“We had a patient come in whose nephew had not had a proper FH diagnosis but was just based on the fact that his dad had died at age 38. It was very badly managed and they were not given any information which left them feeling anxious. As a result of this, the patient didn’t want his son tested but has now been convinced otherwise. This just highlights how important it is having a service that can support the family through treatment and management.”

There are some barriers to service uptake such as people’s perception on statins and their lack of understanding around risks.

This issue is further exacerbated where the first generation of patients who are on statins are doing well which then gives the false impression that the future CHD risk posed to their children is minimal. In some instances patients expressed that they would rather not be diagnosed and are happy to carry on taking statins. Some educational messaging is needed around this so that the risks to their relatives and children can be clearly articulated. Some services have patient support groups and forums whilst some would like to establish forums to provide support and promote education and awareness on FH. In Wales patient forums are used for patient involvement in service development. This has helped build a stronger case when taking forward decisions to management level.

2.4.d Key messages

It is clear that a nurse led service is crucial for FH cascade testing which would not have happened without BHF funding.

Training and education for both nurses and the wider healthcare community has been instrumental in improving awareness of the FH service and increasing high quality referrals. Networking events have enabled nurses to learn from others' experiences and has helped establish relationships in both primary and secondary care.

Services need continual promotion otherwise momentum will decrease and number of referrals and patient engagement will decrease. There is a perception of increased awareness of FH, as relatives are now phoning in to engage with services however this does not include the general public. Delivering FH education days for the public may be beneficial in addressing myths around FH and statins, and raising general awareness on the benefits of testing and the ease of treatment. Another key concern on the potential loss of momentum is due to clinicians historically engaged and championing FH retiring, and many the junior doctors do not appear to have the same level of understanding or interest in FH. There is a clear need to continue training and education on FH to pass on knowledge and build support from the wider healthcare professional community.

There is a pressing need to establish standardised guidelines or a training programme for FH nurses to allow for consistency in workforce skill development. Primarily, education is needed on genetics, lipid pathways, leadership skills and business case development. There is a need to review the FH nurse role to ensure that they are embedded well within their local systems and their skills are utilised in an optimal manner. Some nurses have stated that an exclusive FH cascade testing role is not very fulfilling and it would work better as a more generalised lipid-based role that also incorporates wider cholesterol pathways and CVD risk assessment.

The family members have been, on occasion, cited as a barrier to uptake of testing due to limited understanding of risks and the genetic aspect of the condition. If people are already on statins then there is often complacency around further testing of family members. Nurses reported that information does not always get relayed to the right people and family dynamics can also be an influencing factor.

Resources on FH were generally positively received and praised by service users as well as providers however a significant gap exists in the market around resources geared towards young children. Furthermore there was a need highlighted to develop resources on why statins are important to alleviate fear amongst parents who are preventing their children from taking statins.

2.5 ENABLERS AND BARRIERS TO COST, DATA AND SUSTAINABILITY

2.5.a Risks to sustainability

After the successful uptake of FH cascade services in Wales, BHF invested in areas across England and Scotland to stimulate the set-up of services with a vision of sustainable service provision. Sustainability of the services still remains a risk in several areas and is a multifaceted issue.

In a couple of areas, longer term funding for nurse posts has not been fully agreed and the areas have submitted business cases for continuation of the posts. The remaining services have indicated that the nurse posts will largely be sustained by the hospital trusts, although it has not been easy to reach agreements. Concerns were raised on the importance of securing funding for the nurse post at the beginning of the programme to avoid losing the work to date, however, given that most areas had to collect evidence of impact to make the case for sustaining the post, it would have been a tall order.

In Wales and Scotland, the health board centrally cover the cost of genetic testing. In England this responsibility falls with CCGs, however all stakeholders highlighted that getting buy-in from them has been the biggest hurdle. Although more than half of the services have agreements in place with CCGs to cover the cost of genetic testing, it is unclear how long this might be for. The remaining handful of areas are developing their business cases, whilst a couple of areas are unable to gain any traction in the funding discussions. It should be noted that with financial restraints and shifting priorities there is no room for complacency even when the cost of testing has been secure

A further challenge highlighted by stakeholders was navigating the CCG i.e. not having access to relevant contacts within the CCG and in several cases not even knowing who to contact. The services are also reliant on their ability to continually pay for the PASS database. As highlighted earlier, the cost of licence, the restriction of one user per licence, governance and IT issues and licenses held by pharma are key risks to its wider utility.

The cascade testing services have principally been at risk due to the entwined issues around expenditure, FH featuring low in priorities and difficulty in accessing relevant data. These themes are explored in greater detail below with some overlap as they cannot be discussed in isolation.

2.5.b Expenditure

CCGs are facing financially challenging times in the face of competing priorities and dwindling budgets.

Most approach FH with a bleak outlook and question the relevance of the service for their population as well as the likelihood of cash savings within their short budgetary cycle. CCGs do not perceive cash savings in the service and there is a fear that costs will be driven up further if uptake is low. Several commissioners are concerned that the benefits of the services do not start accruing until year four in the case of any tangible savings. Some commissioners, upon reviewing cost modelling, felt that the savings will be made within hospitals therefore the cost of testing should be picked up by the hospital's budget. Commissioners are aware that there are upfront costs for the first three years, during which services are predicted to saturate identification however many don't have the money to invest upfront. Although many commissioners concede that NICE guidelines are not being adopted, they feel unable to make changes in the current climate.

“...know that no one in this area is doing DNA testing in accordance with NICE guidelines but I'm sure this is the case in other areas too. The costs are just too high.”

An example of the current picture on the ground is further highlighted by the example of one CCG where the highest numbers of heart attack for people under 50 are reported, however there was little appetite to get involved as there was no money to fund genetic testing. The fear around cutting other services was articulated by a stakeholder: “they don't want stories where they cut mental health services and someone commits suicide. ...FH deaths go unnoticed because someone has a heart attack.”

Although population outcomes and quality of life are a central premise of the services CCGs invest in, they expressed: “We are all in a difficult position financially, pragmatically we would look for financial input and outputs and that would be a big thing that would get traction as with anything else... also knowing the prevalence and where we outlie in relation to it would be very useful.”

Clinicians and nurses cited further obstacles within the current system that are discouraging CCGs from engaging in discussions on sustaining FH services; payment systems

and tariffs are not intuitive and in cases hospital tariffs are not sufficient to cover the cost of testing for the index; due to patchy service provision, out of area relatives are accessing services in other CCGs which can disincentivise CCGs from investing; some felt that although the cost per QALY for FH is very cost effective, it is difficult to decommission other existing services; the cost of primary care case-finding is viewed by many as a non-sustainable model in its current form.

Some services have also approached hospital trusts to cover the cost of genetic testing however most hospitals are unable to prioritise it as a chronic condition, and thus to cover the cost of testing indexes, due to the shortfall through existing tariffs. There are exceptions to this, for instance, there are indications that Central Manchester trust may fund cost of testing with a strict criteria and geographic boundaries if the Greater Manchester business case does not move ahead as planned.

Enablers such as BHF funding for nurse posts, Scottish government and Welsh assembly government funding for genetic testing have significantly progressed the case for FH, however issues on further commissioning (in England) of services must be addressed. As explored in **section 2.3** it is clear that relationships can play a huge role in gaining traction in such matters, however, in addition to this, stakeholders expressed further views on what might serve as incentives for commissioners. Those who have been successful in their business cases claimed that, although laborious, speaking to directors of finance and the managerial side of commissioning is key, especially by making arguments that they can understand. Presenting a strong case with evidence of clinical impact, effectiveness and robust cost modelling has also helped in some areas but not all.

Clinicians indicated that health economic modelling is helpful if it is articulated with complementary information such as average age of diagnosis, cost of set up and testing, number of MIs prevented etc. from a national perspective. Piloting services as a collective has also appealed to many areas and appears to be the way forward as risk to individual CCGs is minimal and costs will be better contained. In fact some models have been developed on the basis of a per capita contribution from CCGs rather than a block payment.

The discussion around costs also extends to the gaps within the current system as in the case of VUS. There is no money set aside for further testing or segregation studies for VUSs. There is discrepancy on how this is handled across the UK, where some areas have the flexibility to table it under research, some areas where local doctors are happy

to cover the cost as long as all relatives reside within a specified geographic area whilst the rest of the areas do not have the means nor the inclination to pursue it any further.

The argument on costs has featured significantly in the development of genetic testing in paediatrics, with no specific guidelines on commissioning responsibility and tariffs. Although several clinical stakeholders feel that there should be a larger focus on paediatrics, most services are developed primarily as adult services, with variability on their capacity to absorb the cost of genetic testing for children. Children's services are perceived to deliver benefits and efficiencies over a much longer period than adults services, which is a significant issue given the barriers on commissioning outlined above.

2.5.c Priority setting

At a local level, setting priorities for service provision is inextricably linked to costs and the data available to make the case for change.

Although FH has gained priority at a national level through the efforts of the BHF, HEART UK, PHE, academics and representatives from the upper echelons of the NHS, it remains a low priority locally and for CCGs.

NICE guidelines do not appear to be gaining traction in the local agenda setting. Stakeholders have variable and sometimes divided points of view on how to approach this issue.

The majority of stakeholders expressed that nothing short of a national directive would do and that cascade testing needs to be mandated at a national level. There was agreement that this should be set up in a model that works well locally however the top-down directive is necessary. Some expressed the need to frame FH within a broader pathway of cholesterol, CVD or prevention in general for it to become part of core services. Some felt that tabling it under prevention would also address it from a health inequalities perspective and there is a perception that the prevention ethos is carried by all CCGs.

Some people's experience is contradictory and in stark contrast to what has been discussed in previous sections: for instance, one stakeholder felt that their case was successful based on the emphasis they had placed on paediatrics because they claimed that long-term health is high on the CCG's agenda. Others found success by



approaching confederations of CCGs and framing FH as a regional issue rather than limited to individual CCG areas. Others felt that avoidance of mortality within a 1-2 year period was not a sufficient hook, but rather knowledge of the wider CVD events that are being avoided and what it might have cost the CCG is more powerful.

In contrast, some felt that there is enough evidence available for commissioners and health bodies to access and more evidence will not make a difference but rather an incentivised approach to FH is needed-similar to that of QOF. Lobbying to MPs, using celebrity ambassadors who may have FH and people's stories along with the patients' voice were also considered important levers to tackle prioritisation.

There was hesitation amongst some to feature FH with other CVD conditions or within the genetics umbrella as they felt that with FH the risk is so high and the benefits so great that this approach will not be helpful either to commissioners or the people affected.

However, most commissioners were not willing to look at FH in isolation. Some felt that CVD is a huge priority for them whereas something niche like FH is hard to prioritise. Moreover commissioners expressed scepticism on tabling it under prevention or using incentives given that QOF is being reviewed and there is an appetite to move away from incentivised tick-box practices. Commissioners were not convinced that having a national directive for FH would be helpful given the multitude of priorities they have to tackle. They did however express a desire to have more information on the impact on primary care, local population prevalence, a balanced approach to shorter vs longer term efficiencies and risk sharing with other CCGs.

The BHF are looking to influence and make the case for change through different means rather than limiting themselves to negotiating with individual CCGs: "Our biggest opportunity to influence is the evolution of STP footprints convening local government and confederations of CCGs as well as through the personalised medicine agenda especially if NHS leaders showcase cascade testing as an exemplar of good practice."

The BHF as well as the majority of other stakeholders agree that a combination of several approaches outlined above may be the best course of action in bringing about the required changes in practice.

2.5.d Evidence and data sharing

Data has been central to the barriers highlighted throughout other themes. Access to data and opportune data sharing have resulted in significant benefits as seen in the case of cost savings and linking through PASS or delivering successful business cases.

There are however glaring gaps and a lack of consistent culture of data sharing in the FH community, as highlighted by key stakeholders. Operational inefficiencies have been highlighted, where it has been difficult to access data due to not knowing where to look or who to approach. Several services have struggled to develop robust business cases due to lack of knowledge about national and local level data, lack of access to data necessary for developing cost-modelling or relevant templates, and lack of expertise.

The issues around data and PASS have previously been highlighted, such as underutilisation in the case of VUSs, governance and IT agreements, preventing data sharing across trusts and lack of interfaces between primary and secondary care. Some areas are utilising PASS for inputting and extracting richer and comprehensive patient data, whereas others are not, and this in itself has created discrepancies in access to consolidated data sources. For details on PASS and data please refer back to **section 2.2**.



Services from across the country have expressed the need to be able to share evidence from their respective programmes, as well as to access learning and data from other programmes. Some areas have developed robust business cases, whereas others are struggling to even understand where to start. Several business cases have been made available on the area's local SCN or CCG's website but are well hidden and not promoted to other services. Some barriers had been encountered when data was requested from accounts managers on tariffs and costing, to work out what lipid services were currently doing and to compare low level and high level activity. Several services have expressed that it would be useful to share business cases and to be able to use them as templates to develop further cases. Most importantly, services wanted a business case which will demonstrate the impact of the FH service on cardiology: MI, stroke, etc. and if a CCG was to invest then what would be the possible return on investment.

“When I was doing this I thought....goodness has no one ever done this before! It would have been fantastic to have the right language and info around this- having the national context and example of local context to have at hand would be so beneficial.”

Some areas said that they are generating the evidence and can use it effectively by marrying it to human stories which are powerful. Some said they have years' worth of data for metrics such as indirect contact and want to compare it to direct contact. It is unclear whether other services are aware of what is happening in other parts of the country and they could capitalise on this by collaborating. Another example of a success story in West Midlands is detailed below where some steps are now being taken to highlight it nationally:

“BHF have given us money to pump-prime this and after a lot of negotiations we have buy in from all 22 CCGs- we developed a robust business case outlining why we need the service, case for change, potential for savings over a period of time regarding MI, stroke etc. and we did very good financial modelling and based it on per capita per CCG rather than splitting it evenly across all 22 CCGs. We based per capita costs on expected prevalence in each CCG area, this was quite detailed financial modelling - the service that we proposed is the biggest in the country and now we have been approached by Huon Gray's team from the NHS to turn our business case into a national document- FH has been on the cards for us for about the

last 10 years and there has always been a grey area around its commissioning but we have finally worked together to make this happen.”

Service such as the one outlined above will make for a valuable resource as a case study to access learning and to utilise their methods and templates to help overcome barriers experienced by other services. It will also serve as an opportunity to learn about unique challenges the service may be facing due to its size, for e.g. EU wide tendering for genetic services.

Many in the sector feel that although there is one objective to be achieved, everyone is going about it in different ways without any knowledge of what may be the best route. They expressed a need for access to this information and to explore which pathways may be appropriate for localities guided by underpinning evidence and toolkits around each model to set up and embed services. “Having SOPs, honorary contract templates etc. in place to guide how cross-area working can be facilitated is really needed here.”

There is agreement among key stakeholders that there is a need to create a community of practice to develop a national approach with local variations rather than local versions where services do things differently which is inefficient. “You can't have everyone doing their own thing otherwise you will have a typical ad-hoc NHS approach.”

Another key area where data is an issue is the loss of organisational memory and learning due to workforce migration. A consistent approach to gathering organisational learning is needed so that workforce migration does not affect future negotiations in contracts and sustainability agreements and so that others systems can learn from this. In several areas service providers were unaware of how funding agreements were reached: “CCG will carry on funding for the genetic test but it was already negotiated by the time we came into post so not sure how we gained traction in it or how long it will last.” Having access to this type of information can help ensure that new staff or new services do not have to start from the drawing board, contrary to the momentum gained in FH.

3. KEY RECOMMENDATIONS

The following recommendations have been developed based on evidence and feedback generated through the qualitative fieldwork with key stakeholders as well as conducting secondary research. The national FH steering group and wider stakeholders should review and consider these recommendations to assess the best course of action for resolving the challenges in the sector around FH and ensuring that the constantly evolving nature of the health services does not impede further progress in embedding cascade testing and making it accessible across the country. Ultimately, efforts must be concerted to simultaneously address the key issues highlighted to achieve system-level change.

- **A national exercise should be considered on assessing and selecting fewer accredited labs offering genetic testing for FH.**
- **A conservative approach can help embed consistent and high quality techniques for optimal sample processing, drive down costs and work as a sustainable and cohesive model nationally, given the financial pressures facing the UKGTN.**
- **National stakeholders should undertake a mapping exercise to ascertain where paediatrics features in the FH pathway and where the budget responsibilities lie.**
- **There is a need to develop a consistent approach to deal with paediatric cases in the context of budgets, access points into a service, treatment and management plans, consolidating information on the paediatric register and alignment with the family clinics.**
- **Services should discuss and collectively publish evidence on the efficacy of various referral routes between services such as primary, secondary and out-of-area services and build them into standard service delivery models.**

This will help eliminate inconsistencies across services, help utilise service level agreements where they exist and may improve pick-up rate of relatives per index.

- **A standardised programme should be developed for FH nurses detailing training requirements to address the skills gap and ensure services across the country are consistently performing to a high standard.**

It may be necessary to review the skills needed for FH nurses to work in a variety of backgrounds that may incorporate a wider lipid/cholesterol or CVD pathway. It will also be worthwhile to consider nurse bands for the roles detailed.

- **All relevant stakeholders within health services must tackle the issues around the ethics of directly contacting family members and accessing patient consent.**

Services must be allowed to consider direct contact as a first line of action once appropriate ethics approval has been agreed. This needs to be bespoke around FH in order to distinguish it from other genetic conditions such as Huntington's and Cystic fibrosis, due to the ease of treatment and positive prognosis.

- **National bodies should undertake an appraisal exercise to assess if PASS or another database will be beneficial to roll out across the nations with a concerted effort.**

A national toolkit must then be developed to help support its implementation by addressing issues on governance and IT. To satisfy requirements, the database must have shared workflow functionality, ability to share interfaces with systems in all four nations, accessibility within genetic labs and ability to share data with different parts of the health system. In the case of PASS, licenses must be managed directly from developers or through a national PASS coordinator role to allow negotiations for improvements.

- **Guidelines should be developed for classifying and logging VUSs and services should negotiate for budgets to allow testing of relatives of those with VUSs on clinical grounds, where deemed appropriate.**

Upon classification of a pathogenic VUS, a clear protocol must be developed outlining communication to and management of families affected.

- **Services should develop a public engagement plan for FH to raise public awareness and outline benefits of treatment to combat the media hype against it.**

This may help in thwarting myths around FH and statins and raise awareness on the potential risks to children where family members are resistant to testing and treatment. This can also be utilised to engage with and lobby the patient voice in the sector. Furthermore, there is a need to prioritise and recognise FH when it comes to high cholesterol and separate it from the wider statins debate.

- **Services need to address the issue of equal access to and better engagement with FH testing across all ethnic minority groups**

- **Efforts should be concentrated nationally to develop approaches on FH cascade testing that can subsequently drive local efforts to championing FH.**

Bringing together a community of practice as well as local champion practitioners will be key in striking a balance between a top-down and a bottom-up approach and ensure that areas are not inefficiently developing approaches in isolation. This will also lend some robustness to approaches and may help alleviate the pressures on keen individuals driving the momentum in their localities.

- **Services should develop and implement a consistent approach to gathering organisational learning so that workforce migration does not affect future negotiations in contracts and sustainability agreements.**

Lack of a robust infrastructure could result in dissolution of existing services. As clinicians and other professionals retire there will be a significant loss of momentum and knowledge on these issues if steps are not taken to formalise arrangements and protocols within services.

- **Stakeholders should consider the development of a national FH hub that will give users access to resources crucial for existing and new services being established.**

The hub should contain information such as patient resources in development, local service agreement templates, honorary contract agreements, successful business case templates, cost-modelling protocols and research data on contact methods and case-finding tools.

4. APPENDIX

LOGIC MODEL

This logic model further explains how the programme intends to achieve its aims and objectives – outlining the activities, subsequent outputs, and interim outcomes that lead to eventual impact.

Objectives	Activities	Outputs (include KPIs)	Anticipated Outcomes	Impact
Develop FH cascade testing services across multiple sites & identification of index cases and relatives.	<ul style="list-style-type: none"> Funding and recruiting FH nurses to posts Case-finding processes established in primary care Case-finding processes established in secondary care Processes established with pathology labs to identify people with abnormal cholesterol levels Work with index cases to identify relatives at risk of having FH Work with FH colleagues across the UK to identify and invite relatives of index cases and offer cascade testing 	<ul style="list-style-type: none"> # of nurses recruited to post Proportion of sites with PASS licenses # of functioning sites No. of courses/modules attended Case studies of integrated work with GPs and other service providers Nurses have appropriate support to run services 	<ul style="list-style-type: none"> Services successfully set up for cascade testing and attain funding for continued and sustainable service provision Growth in reach of cascade testing programmes and increased number of referrals Increased diagnoses and coverage across the UK 	<ul style="list-style-type: none"> All high risk individuals identified in primary care are offered FH genetic testing and subsequent cascade testing for families
Undertake DNA testing to identify people with an FH genetic mutation.	<ul style="list-style-type: none"> Nurses coordinate referrals for genetic testing Consenting patients for genetic testing Nurses send samples to genetic labs & coordinate results Children identified with FH placed on FH paediatric register 	<ul style="list-style-type: none"> # of index cases identified # of positive cases # of cascade tests carried out # of consultation carried out Qualitative account/case studies of FH paediatric register utilisation and functionality 	<ul style="list-style-type: none"> All index cases and family members have had a negative 	<ul style="list-style-type: none"> All high risk individuals identified in primary care are offered FH genetic testing and subsequent cascade testing for families
Provide education and support to families prior to genetic testing and following testing for those diagnosed with FH.	<ul style="list-style-type: none"> Counselling patients and relatives prior to genetic testing Distribute / discuss resources with patients and their families following diagnosis Developing resources and Identifying gaps 	<ul style="list-style-type: none"> Engagement & resource uptake Quality of resource/information provided 	<ul style="list-style-type: none"> All patients and their families have access to appropriate information 	<ul style="list-style-type: none"> FH patients receive appropriate support and treatment
Promote awareness of FH in communities (public and professional).	<ul style="list-style-type: none"> Provide education and training in primary and secondary care – public and professionals Awareness raising in primary and secondary care – public and professionals Developing FH awareness resources 	<ul style="list-style-type: none"> # and type of outreach activities, education sessions etc. with public and professionals Conferences – presentations etc. Research conducted parallel to the programme (Kate) 	<ul style="list-style-type: none"> Population has awareness of testing for FH, particularly where family history of heart disease exists Increase in number of people voluntarily engaging in testing Increase in awareness and benefits of FH testing amongst HCPs 	<ul style="list-style-type: none"> Increases in proportion of individuals who are tested for FH

Objectives	Activities	Outputs (include KPIs)	Anticipated Outcomes	Impact
Utilise and populate the PASS databases that links FH services across the UK	<ul style="list-style-type: none"> PASS software / database / licenses acquired Nurses input data, develop workflows, communicate with other FH specialists Nurses utilise PASS database to both record and identify people with FH Initial training on PASS database and ongoing support for PASS users (Kate) Development of e-learning tool for PASS users, template letters, referrals, etc. (Kate) Development of SOPs and trouble - shooting of problems (NHS IT infrastructures) (Kate) Create workflows for each PASS site and reports for PASS users (Kate) Liaise between PASS (Denmark) and PASS sites (Kate) 	<ul style="list-style-type: none"> Variance in site usage of PASS and usage in region (other service providers) Feedback from nurses regarding PASS usage Database has incorporated data from all services and is usable and accessible to service providers 	<ul style="list-style-type: none"> Service providers are actively using database and regularly inputting new data Improved identification of new cases, reduced burden on service providers, improved efficiency of care provision Appropriately handle VUS cases to facilitate additional understanding of genetic variants 	<ul style="list-style-type: none"> FH services are joined up and increasing overall diagnoses across the UK
Commission work to outline QALYs and cost effectiveness of FH cascade testing	<ul style="list-style-type: none"> Comparison of different service models Cost effectiveness analysis and economic modelling SLA template development for CCG Development of template business case Top tips for commissioners 	<ul style="list-style-type: none"> Evidence based understanding of FH models and associated costs and service provision 	<ul style="list-style-type: none"> Evidence is used and disseminated to influence commissioners and public health bodies 	<ul style="list-style-type: none"> Local commissioners support FH testing nationally
Facilitate implementation and scaling up of FH cascade testing nationally	<ul style="list-style-type: none"> Development of paediatric database Analysis of lipid pathways National partnerships and steering groups Develop national education for FH NICE guidelines input SIGN guidance input 	<ul style="list-style-type: none"> Investigating development of paediatric FH services in parallel with FH service development Path lab usage – barriers to use and access Investigate VUS databases and their utilisation Evidence based understanding of barriers to implementation and scaling up 	<ul style="list-style-type: none"> Evidence is used and disseminated to influence commissioners and public health bodies 	<ul style="list-style-type: none"> Increase in number and efficiency of FH services nationally

TOPIC GUIDE

NURSES

Group members/Interviewees

- FH Nurses

Facilitators

- Group interviews
 - Lead facilitator – introduces session, leads discussion by asking questions, closes session
 - Assistant facilitator – runs recording equipment, takes notes, assists in running session
- 1-on-1 interviews
 - Interviewer – Introduces topic, leads conversation using topic guide, runs recording equipment

Resources required

- Paper, pens
- Recording equipment
- Water, glasses
- Watch, time check reminders

Introduction (Interview)

1. Welcome, name, facilitator job title and explanation, and thank interviewee(s) for their time
2. Explain purpose of session:
 - a. To gather more in-depth understanding of FH cascade testing in practice
 - b. To discuss improvements that could be made to the programme
 - c. To look at specific benefits and/or challenges
3. Why interviewee has been invited:
 - a. Best positioned to help improve the scheme and let us know how it actually went
4. Session guidelines, recording and anonymity:
 - a. We will allow up to 1 hour for the interview
 - b. Session is being recorded for transcription purposes – the recordings will only be used to transcribe comments, not shared with anyone
 - c. Answers are confidential however given nature of the programme, comments may be identifiable
 - d. Informal chat, not an interview. I have a list of set questions, but purely as a guide. Feel free to discuss broadly and I will only turn to the questions to keep us on track if necessary

FH NURSES	
SECTION	QUESTION
Service delivery	<ul style="list-style-type: none"> • Which pathways have been used for development of service and how has experience of this been? • How were relationships developed with partners (other providers) along patient care pathways and were these successful? • How do you interact with pathology services? • Where are the patient referrals coming from? • What happens after FH + (relatives contacted by direct or indirect method?) • What happens to those with high cholesterol but FH-
Databases	<ul style="list-style-type: none"> • How do you log patient data? What databases (e.g. PASS) do you use?/any issues with using database? • How do you handle VUS cases? • How do you handle paediatric cases? How do you log data for these cases?
Patient support	<ul style="list-style-type: none"> • What resources do you use to inform patients about testing and FH? • Are resources appropriate? How could these be improved? • How well supported do patients feel? • What particular successes has the intervention had? (if possible, please use case studies to demonstrate)
Outreach	<ul style="list-style-type: none"> • What outreach activities have you conducted (e.g. education with public / professionals, conferences / presentations)? Please could you talk more about these? • Have you noticed any increases in the number of people voluntarily engaging in testing and / or increased awareness of FH testing?
Interaction with public health bodies	<ul style="list-style-type: none"> • Have you had input in development of national guidelines, education, and practice for FH? • What interaction (if any) do you have with local commissioners and public health bodies? • How has HCP training raised awareness of FH improving identification, diagnosis and patient care? • Would the HCPs have acquired this knowledge over time anyway without the intervention?
Lessons	<ul style="list-style-type: none"> • How has the experience of developing service been for you? • What barriers and challenges were encountered and how could these can be avoided / overcome? • What are the key lessons that have been learnt (including for processes such as setting up the project, project management or for the partnership) and what are your recommendations for the future? • Are there any indirect outcomes emerging from the intervention?
Future of the programme	<ul style="list-style-type: none"> • How sustainable is the intervention after BHF funding has ceased and what exit strategies are in place? • Do you have any further comments that would assist in evaluating the programme?

CONSULTANTS / OTHER HCPS	
SECTION	QUESTION
Service delivery	<ul style="list-style-type: none"> • Describe how the service/pathway works in your area, where do you feature into it? • How were relationships developed with nurses and other partners? (primary/secondary) • What would have happened anyway, without the intervention in place ('counterfactual')? • What are the system levers for such a service? • What are the barriers to this service and its wider roll out?
Databases	<ul style="list-style-type: none"> • How do you handle paediatric cases? How do you log data for these cases? • How do you deal with VUS? • How is the use of PASS for you? Any issues?
Lessons	<ul style="list-style-type: none"> • How has the experience of developing service been for you? • What barriers and challenges were encountered and how could these can be avoided / overcome? • Are there any indirect outcomes emerging from the intervention? • What are the key lessons that have been learnt (including for processes such as setting up the project, project management or for the partnership) and what are your recommendations for the future?
Interaction with public health bodies	<ul style="list-style-type: none"> • What interaction (if any) do you have with local commissioners and public health bodies? • Has your awareness of FH improved since the start of the programme in terms of identification, diagnosis and patient care? • Would you have acquired this knowledge over time anyway without the intervention? • How can evidence from this programme be applied so that cascade testing can be rolled out more broadly across the UK?
Public opinion	<ul style="list-style-type: none"> • Have you noticed any increases in the number of people voluntarily engaging in testing and / or increased awareness of FH testing?
Patient support	<ul style="list-style-type: none"> • What resources do you use to inform patients about testing and FH? • Are resources appropriate? How could these be improved? • How well supported do patients feel? • What particular successes has the intervention had? (if possible, please use case studies to demonstrate)
Future of the programme	<ul style="list-style-type: none"> • What is the future direction for FH testing?
Further comments	<ul style="list-style-type: none"> • Do you have any further comments that would assist in evaluating the FH testing programme?

COMMISSIONERS	
SECTION	QUESTION
Service delivery	<ul style="list-style-type: none"> • Do you commission FH cascade testing in your area? • What are the system levers for such a service? • What are the barriers to this service and its wider roll out?
Public opinion	<ul style="list-style-type: none"> • Have you noticed any increases in the number of people voluntarily engaging in testing and / or increased awareness of FH testing?
Interaction with service	<ul style="list-style-type: none"> • How do you interact with FH service providers (nurses, other HCPs, pathology services)? • Has your awareness of FH cascade testing increased since the start of the BHF programme? • What evidence are you looking for in order to (further) support FH cascade testing? • How can evidence from this programme be applied so that cascade testing can be rolled out more broadly across the UK? • Would FH cascade testing have been established without this intervention being piloted?
Future of the programme	<ul style="list-style-type: none"> • What is the future direction for FH testing?
Further comments	<ul style="list-style-type: none"> • Do you have any further comments that would assist in evaluating the FH testing programme?

Closing

1. Summary of key comments noted through the course of session ["is this an accurate summary?"]
2. Do you have anything else to add? Have we missed anything?
3. Reminder of data confidentiality
 - a. Comments transcribed
 - b. Name / comments not linked
4. Thank participant for their time and ask if they would like follow-up information – e.g. summary of days discussion by email

REFERENCES

- 1 Goldberg AC, and Gidding, SS. Knowing the Prevalence of Familial Hypercholesterolemia Matters. *Circulation*, 2016; 133 (11).
- 2 Slack J. Risks of ischaemic heart-disease in familial hyperlipoproteinaemic states. *Lancet*, 1969; 2(7635).
- 3 Stone NJ, Levy RI, Fredrickson DS, Verter J. Coronary artery disease in 116 kindred with familial type II hyperlipoproteinemia. *Circulation*, 1974; 49(3).
- 4 Scientific Steering Committee on behalf of the Simon Broome Register Group. Mortality in treated heterozygous familial hypercholesterolaemia: implications for clinical management *Atherosclerosis*, 1999; 142.
- 5 Versmissen J, Oosterveer DM, Yazdanpanah M, Defesche JC, Basart DC, Liem AH, et al. Efficacy of statins in familial hypercholesterolaemia: a long term cohort study. *BMJ*, 2008; 337.
- 6 NICE Clinical guideline 181.
- 7 Marrion's economic modelling paper.
- 8 Kerr M, Pears R, Watson M, Cather M, Haralambos K, Miedzybrodzka Z, Gray H, Humphries SE. Cost effectiveness of cascade testing for familial hypercholesterolaemia. (unpublished)
- 9 Weng SF, Kai J, Andrew Neil H, Humphries SE, Qureshi N. Improving identification of familial hypercholesterolaemia in primary care: derivation and validation of the familial hypercholesterolaemia case ascertainment tool (FAMCAT). *Atherosclerosis*. 2015 Feb;238(2):336-43.
- 10 Green P, Neely D, Humphries SE. Improving detection of familial hypercholesterolaemia in primary care using electronic audit and nurse-led clinics. Medway FH Audit Steering Committee. *J Eval Clin Pract*. 2016 Jun;22(3):341-8.



**British Heart
Foundation**

For over 50 years we've pioneered research that has transformed the lives of millions of people living with heart disease. Our work has been central to the discoveries of vital treatments that are changing the fight against heart disease.

But heart and circulatory disease still kills around one in four people in the UK, stealing them away from their families and loved ones.

From babies born with life threatening heart problems, to the many mums, dads and grandparents who survive a heart attack and endure the daily battles of heart failure. Join our fight for every heartbeat in the UK.

Every pound raised, minute of your time and donation to our shops will help make a difference to people's lives.

Text FIGHT to 70080 to donate £3*

**FIGHT
FOR EVERY
HEARTBEAT**

bhf.org.uk

*This is a charity donation service for the BHF. Texts cost £3 + 1 standard rate msg.
The BHF will receive 100% of your donation to fund our life saving research.
To opt out of calls and SMS text NOCOMMS BHF to 70060, or if you have any
questions about your gift call 02032827863.

© British Heart Foundation 2016, a registered charity in England and Wales (225971) and Scotland (SC039426)