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Familial Hypercholesterolaemia

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Contents

1 Introduction

1.1 About Familial Hypercholesterolaemia (FH)

Familial hypercholesterolaemia (FH) is a genetic condition that causes high cholesterol and coronary heart disease, often resulting in premature coronary heart disease (CHD) myocardial infarction (MI) and reduced life expectancy. Patients with FH will have abnormally high cholesterol from birth.

FH is a relatively common genetic disorder. The estimated prevalence is 1 in 500, suggesting 120,000 affected individuals in Britain. The condition is massively under diagnosed with only 15-17% of cases identified in the UK.

Children of an individual with FH have a 50 per cent chance of inheriting the condition. 50% of males and 30% of females with untreated FH will have developed CHD by the age of 55. This premature disease, often resulting in early death, is avoidable. Unlike many genetic conditions, FH can be diagnosed relatively easily and, with inexpensive treatment, people with FH can lead normal, healthy lives.

1.2 The NICE Guideline and the Benefits of Cascade Testing

In 2008, NICE published a clinical guideline for the Identification and Management of FH (CG71). The guideline recommends identifying cases of FH, using cholesterol measurements and cascade genetic testing of their families. Referral to specialist lipid clinics is recommended for confirmation of the diagnosis, patient counselling and in order to initiate the cascade testing.

The NICE Guideline indicates that the cascade testing model for diagnosing FH is cost-effective, with an estimated ICER (incremental cost effectiveness ratio) of £2,700 per QALY (quality adjusted life year); well below the NICE cost effectiveness threshold of £20-30,000/QALY.

Benefits from initiatives to find cases of FH include a reduction in premature deaths from heart disease; a reduction in long-term morbidity and its associated costs; and the benefits to families no longer trapped in a cycle of premature heart disease. Since the cost of effective therapy is so low, a significant saving could be made by the NHS in England, due to a reduction in CHD events and the reduced number of associated hospital admissions.

1.3 Strategic Direction: Recent Policy and Publications

In March 2013 the Department of Health published its Cardiovascular Outcomes Strategy, endorsed by NHS England, and Public Health England. Among its priorities, the Strategy set the initial ambition of identifying at least 50% of cases of FH in England diagnosed and treated – a substantial jump from the current low levels.

In August 2013, NICE published its Quality Standard on FH (QS41). The Quality Standard includes an FH care pathway and commissioning guidance. Importantly, the Quality Standard supports the delivery of the NHS Outcomes Framework, by helping to fulfil Domains 1 (preventing people from dying prematurely), 2 (enhancing quality of life for people with long-term conditions) and 4 (ensuring people have a positive experience of care).

2 British Heart Foundation Bid

The British Heart Foundation (BHF) has agreed to provide over £1m of funding nationally to support the employment of specialist FH nurses or other key staff.

In June 2015, the West Midlands Clinical Network (WMCN) put forward a bid to the British Heart Foundation as part of their second round of funding applications. The WMCN, in collaboration with clinical colleagues was successful in securing £375,000.00 from the BHF to support the introduction of a West Midlands regional FH service. The funding will cover the cost of 5 specialist FH nurses for a period of 18 months. In addition, patients who meet the referral criteria as being at risk of FH will require genetic screening and family cascade testing. This service provision falls outside of the BHF funding. CCG's will be required to fund genetic testing as part of the delivery of a robust service model across the West Midlands

In making the bid, the WMCN had to ensure there was an agreement from CCGs that they would continue to fund the specialist nurses after the 18 month period and that CCGs would fund the additional genetic testing costs from service.

This bid was presented to the Birmingham Accountable Officers (AO) in July, but the full impact of the bid and the costs that the CCG would be asked to support were not fully presented. This was not known to AOs until Birmingham Cross City (BCC) CCG were asked to be the lead commissioner for the service and work with the BHF in contracting for the service. On review by BCC CCG it was clear that not all CCGs were compliant in agreeing to support the bid and associated costs, nor had all the cost impacts been shared with AOs. The CCG requested, via the FH operational group, that a full proposal was developed for CCGs to consider formally before any of the BHF funding was drawn down and before any recruitment for the specialist nurses commenced. Following the successful bid, the FH operational group considered who could host the service on behalf of the region, and it was felt that University Hospitals Birmingham NHS Foundation Trust would be the most suitable host due to them being the lead NHS organisation for the WM Genomic Medicine Centre (a regional collaboration across 17 NHS trusts). The decision was also supported by all 12 CEO's of the provider Trusts involved. A regional cascade screening service offers the opportunity to evaluate the impacts of embedding genomic medicine as standard practice for a well-defined tracker condition.

3 Case for Change

There is an urgent need for change within the West Midlands. The region is currently not meeting NICE Guidelines for this condition despite the case and strategic direction that sets it out as high priority.

The current service for patients lacks regional co-ordination; care is fragmented, with no agreed pathway, standards or protocols or systematic cascade testing either by genetic typing or by cholesterol measurement. There is a real chance that poorly planned testing will begin based on ability to pay, or individual commissioner decisions that will increase health inequalities and not deliver the potential public health benefits; unless accompanied by systematic cascade testing.

Early identification and treatment of FH is effective at prolonging life. This is most pronounced for those identified at an early age and for women the [1]reduction in risk of

progression to early coronary heart disease with the use of statins is significant. Untreated, the risk of new CHD occurring is 119 per 10,000 person years, whilst for FH treated with statins the risk is reduced to 11 per 10,000 person years. [2]This means that if we were able to treat all FH cases we would prevent 126 cases of CHD per year across the West Midlands. (See table 1)

Table 1: Number of cases of CHD avoided per year if FH programme fully implemented, based on Versmissen (2008)

Total no of CHD cases preventable	
Total Number of cases	11,692
No of new CHD cases per year if all untreated	139.1347
No of new CHD cases per year if all treated	12.8612
Reduction in new CHD cases	126.2735

Using the PbR 2014/15 National Tariff for Myocardial Infarction at a cost of £3,129 non elective (EB10Z), potential cost savings could amount to **£413,966.70 (includes MFF at 5%)** per year based on this number of avoidable heart attacks as a result of CHD. This figure does not take into account additional savings for CCG's for avoided elective primary PCI and prescribing costs.

This figure does not include additional savings for a reduction in the following:

Clinical outcomes

- Strokes
- Heart failure
- Cardiovascular and total mortality
- Transient ischemia attacks
- Unstable angina
- Revascularisations

Organisational

- Hospital bed days
- GP consultations as a result of cardiovascular events
- Stroke rehabilitation

3.1 Care settings and Patient Numbers

In developing the case for change, the FH operational group have undertaken a needs assessment to indicate the likely patient numbers who could benefit from a regional FH service, improving their clinical outcomes and reducing future morbidity.

Whilst, patients with FH may be seen within specialist lipid clinics, by cardiologists or in primary care, those current under the care of local lipid clinics offer the best way to estimate the potential client group this proposal would benefit.

WMCN undertook a recent survey in collaboration with all lipid clinic providers in the West Midlands. This revealed that approximately 1,579 patients attend local clinics as new FH patients per year. Using prevalence data, the number of expected cases in the West Midlands, based on population, is estimated at 11692. Table 2 below shows the number of new patients attending local FH clinics per year in secondary care providers. The current total number of new cases per twelve months equates to approximately only 13% of expected cases, highlighting the current gap to identify and treat those at risk in the region. The service will aim to capture 100% of all FH cases over 8 years. (*See Appendix 1 for Expected number of cases per CCG based on mid 2014 population estimates*)

1 Marks 2002
2 Versmissen

The table below displays current estimated FH *only* activity from current acute based lipid clinics over the past 12 months.

Table 2 - 12 month activity of FH patients per Hospital

Hospital	New activity per wk.	New per year (44 wks)	Per year
UHCW		200	200
QHBT	2	88	88
DGH	2	88	88
HEFT	8.5	374	374
SaTH	1.3	57.2	57.2
UHNM	0.4	17.6	17.6
SWBH	6	264	264
WAHT	1.2	52.8	52.8
RWHT		165	165
UHB	1.2	52.8	52.8
Walsall Hospital	3	132	132
WVT	2	88	88
Total 1579.4			

3.2 The evidence for Genetic Testing

In the West Midlands the majority of lipid clinics are currently performing cascade testing using cholesterol only, in the absence of commissioned genetic testing. The service will see genetic testing that can link with other groups using PASS software. This will help in the identification of individuals within the West Midlands, and ensure that data for relatives in other regions is used to cascade on a national basis. The data collected by the nurses, as patients are screened will be used to evaluate other methods of approaches to identification of affected individuals. These methods will include The HEART UK Medway Project, The Dutch Lipid Clinic Criteria, The Simon Broome Criteria and assessment using the FAMCAT methods. We will use these to audit the approach and determine if these are relevant especially in relation to the ethnic diversity of the population.

There are two elements of Genetic services required to support an FH service:

1. Molecular Genetics – this is the actual genetic DNA analysis to support diagnosis;
2. Index Case/Cascade Testing – this provides specialist genetic counselling to all patients before undergoing any DNA analysis; it will also produce family pedigrees and run all aspects of cascade testing.

3.2.1 Cascade testing

Cascade testing is a cost effective method of case finding compared to general population screening. Relatives of a known case are tested for the condition. In genetic conditions that are dominantly inherited e.g. FH there is a 50% chance that 1st degree relatives will share the same mutation, and is therefore a highly focused method of case finding.

It is unlikely that the majority of patients will be found through general population screening because of the large numbers requiring screening and the exclusion of other causes of high cholesterol.

A NICE[1] commissioned economic evaluation of strategies for cascade testing found that the most cost effective strategy is to use a combination of genetic and cholesterol based cascade testing, using genetic testing where the family mutation has been identified. There are two main reasons for this:

- Cascade testing using cholesterol only was cheaper but less cost effective than using genetic testing where possible. Cholesterol levels are not a sensitive or specific screening test for FH, and will result in false negative and false positive diagnoses, although there is benefit in using cholesterol base cascade testing in clinically diagnosed cases where no mutation has been found.

- Genetic cascade testing alone is more expensive and less cost-effective than cholesterol testing alone. This is because at the moment it is not possible to identify all FH mutations. Restricting cascade testing to DNA-confirmed cases reduces the amount of high risk families tested, resulting in missed chances to identify and treat further cases of FH.

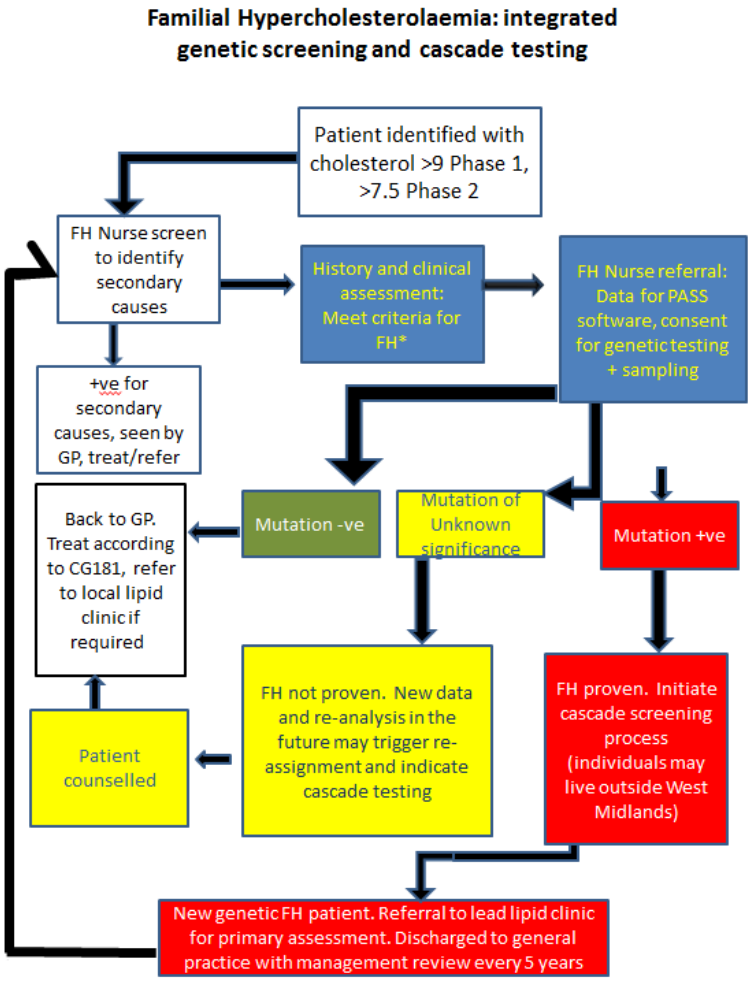
4 The Proposed Model

The regional approach will maximise quality across the region and minimise costs. The service will be hosted by UHBFT with linkage to the Rare Disease Centre at the QEH. The host organisation will provide governance, administration, nursing, management and IT support. The cohort of specialist BHF FH specialist nurses will run peripatetic clinics in the West Midlands region, each nurse covering the populations served by a number of lipid clinics, to optimise equity is geographical access and service efficiency. They will primarily undertake provision of the regional cascade screening service in primary care settings rather than acute hospital settings to deliver care closer to patient needs and ensure maintenance of close links to the patient's primary care provider.

The flowchart below (fig 1) details the proposed integrated genetic screening and cascade testing pathway to be adopted across the West Midlands. The service aims are to treat at least 70% of the FH population in primary care following diagnosis where only the most complex cases are seen in acute settings following diagnosis.

[1] NICE FH guidelines (CG71); the NICE Quality Standard on FH (QS41); https://www.**nice**.org.uk/guidance/**cg71**

Fig 1 West Midlands FH Screening Pathway



5 Implementation

5.1.1 Patient Identification for entry to the FH Cascade Screening Pathway

The FH specialist nurses will receive patients who have been referred to the lipid clinics with potential FH. This can be by either actual (i.e. after having been seen at the lipid clinic) or virtual referral from GP's or hospital colleagues as appropriate (e.g. Advice and Guidance via Choose and Book). Where possible, the FH specialist nurse will also receive direct GP referrals to avoid any unnecessary out patient's referrals. As per NICE guideline and quality standards, patients meeting the Simon Broome criteria will be accepted but assessments of the other methods of identification will be documented to help with audit of the process. They will be seen by the FH specialist nurse who will assess their suitability for genetic screening as index cases by the use of the scoring tools. The nurses will also liaise with the relevant laboratories to identify potential index patients with historically raised cholesterol who can be targeted via primary care to consider genetic referral if appropriate.

The use of a specialist database will be used for rolling out the cascade testing programme. PASS Clinical® Genetic is a fully integrated management information system for clinical genetic centers used for family contact tracing. HEART UK has agreed to partially fund the licence fees for 3 years for the use of the PASS Clinical® Genetic software programme for the regional service.

5.1.2 Cascade testing

In order to target resources effectively and reduce unnecessary tests, cascade testing should be performed on first degree relatives, with the process repeated for the first degree relatives of new cases found and subsequent second and third degree relatives as found. This cascading process will reduce cholesterol testing and definitively identify those at greatest risk.

Cascade testing will be undertaken by BHF FH specialist nurses who have been trained to provide genetic and cholesterol counselling, testing and interpretation of results.

Efficient use of cascade testing resources will be maintained by ensuring that the 5 primary care based BHF FH specialist nurses manage all cascade testing according to protocol. This will reduce unnecessary tests and also ensure that as far as possible medical appointments in secondary care are reserved for complex cases.

The BHF FH specialist nurses will manage a database to map the epidemiology of FH in the West Midlands, and enable audit.

Children will be seen in BHF FH specialist nurse led family clinics supported by the Birmingham Children's Hospital where required.

5.1.3 Recruitment to the 100,000 Whole Genome Project in the West Midlands

It is anticipated that a regional FH cascade screening service will also identify index cases and families who will meet inclusion criteria for entry to the 100,000 whole genome project (WGP). In the event that index cases and family members are thus identified, the nursing posts associated with the FH service in collaboration with the patient's GP and secondary care clinical consultant, will be able to undertake patient recruitment to 100,000 whole genome project. Costs for 100,000 WGP whole genome sequencing, including consumables, will be met by the WM Genomics Medicine Centre, hosted at UHBT.

5.2 A phased implementation approach

It is proposed that the NICE CG71 Guidelines are followed in order to achieve the modelled health benefits. It is anticipated that implementing FH cascade testing in the West Midlands will fall into three distinct phases. During **Phase One**, a high risk group of patients of cholesterol 9 mmol/L and above (approximately 1,500 people) are tested and cascaded. (See table 3).

Table 3 Predicted activity for undiagnosed patients with cholesterol of 9 and above

Hospital	**12 mth activity of cholesterol 9 and above**
UHCW	175
QHBT	126
DGH	*135
HEFT	128
SaTH	119
UHNM	144
SWBH	154
WAHT	150
RWHT	*63
UHB	143
Walsall Hospital	129
WVT	*56
Total	1522

* Average activity calculated where data not provided by lipid centres

Phase Two will assess the populations with a lower cholesterol concentration to a level decided by the operational group to ensure an efficient use of resource and maintained identification of index genetic cases with appropriate cascade screening. This process will require regular review to ensure that the widest possible appropriate population is targeted. Specifically patients presenting with early cardiovascular disease in general practice or referred through cardiology centres after intervention will be targeted for inclusion following acute vascular events. Phase one and two are projected to last approximately 7 years.

Phase Three should begin once the majority of cases have been found and cascaded. At this point regional demand for DNA testing for FH should reduce to 'incident cases' only, largely made up of births within affected families, new FH patients moving into the area, and referrals for testing of relatives of index cases living outside of the West Midlands. As the birth incidence of FH is also 1/500, a maximum of 140 babies per year would be tested where parents are known to be affected.

The regional approach to cascade testing has been designed to manage Phase one and two. It is expected that this will be reviewed once the region moves to Phase three, this would be subject to evaluation of phase 1 and 2. A new business case will be required to commissioners for prioritisation to proceed to phase 2 and 3.

6 Finance

6.1 Cost breakdown for Regional Service, years 1-4.

The tables below outline the cost breakdown for years 1-4 split per capita per CCG. NICE CG71 advises tha potenial savings on reduction of CHD in the FH population will be realised post year 2, therefore cost savings have been calculated based on population and projected savings per CCG from year 3.

6.1.1 Table 4 Year 1 cost breakdown per CCG

CCG	£250.00 per index case	Cascade cost/relative @£75	Year 1 consumble cost	Year 1 non-FH clinical consumable costs	year 1 total
Coventry and Rugby	£12,596	£11,336	£2,054	£1,447	**£27,434**
Herefordshire	£5,093	£4,584	£831	£585	**£11,093**
Redditch and Bromsgrove	£4,655	£4,190	£759	£535	**£10,139**
South Warwickshire	£7,393	£6,654	£1,206	£849	**£16,102**
South Worcestershire	£7,886	£7,098	£1,286	£906	**£17,176**
Warwickshire North	£5,038	£4,535	£822	£579	**£10,974**
Wyre Forest	£3,067	£2,760	£500	£352	**£6,680**
Birmingham Cross City	£19,989	£17,991	£3,260	£2,297	**£43,536**
Birmingham South and Central	£6,846	£6,161	£1,116	£786	**£14,910**
Dudley	£8,352	£7,517	£1,362	£960	**£18,190**
Sandwell and West Birmingham	£14,650	£13,185	£2,389	£1,683	**£31,907**
Solihull	£6,435	£5,791	£1,049	£739	**£14,015**
Walsall	£7,503	£6,753	£1,224	£862	**£16,341**
Wolverhampton City	£7,174	£6,457	£1,170	£824	**£15,625**
Cannock Chase	£3,615	£3,253	£589	£415	**£7,872**
East Staffordshire	£3,697	£3,327	£603	£425	**£8,051**
North Staffordshire	£5,833	£5,249	£951	£670	**£12,703**
Shropshire	£8,215	£7,393	£1,340	£944	**£17,892**
South East Staffordshire and Seisdon Peninsular	£5,750	£5,175	£938	£661	**£12,524**
Stafford and Surrounds	£3,943	£3,549	£643	£453	**£8,588**
Stoke-on-Trent	£7,640	£6,876	£1,246	£878	**£16,639**
Telford and Wrekin	£4,710	£4,239	£768	£541	**£10,258**
Totals	£160,080	£144,072	£26,105	£18,391	**£348,648**

6.1.2 Table 5 Year 2 cost breakdown per CCG

Year 2 costs include 6 months nursing salaries

CCG	£250.00 per index case	Cascade cost/relative @£75	Year 2 including: Salaries, consumables & Non-FH Clinical consumables	Year 2 totals including genetic costs
Coventry and Rugby	£12,596	£11,336	£12,933	**£36,866**
Herefordshire	£5,093	£4,584	£5,229	**£14,907**
Redditch and Bromsgrove	£4,655	£4,190	£4,780	**£13,624**
South Warwickshire	£7,393	£6,654	£7,591	**£21,638**
South Worcestershire	£7,886	£7,098	£8,097	**£23,081**
Warwickshire North	£5,038	£4,535	£5,173	**£14,746**
Wyre Forest	£3,067	£2,760	£3,149	**£8,976**
Birmingham Cross City	£19,989	£17,991	£20,524	**£58,504**
Birmingham South and Central	£6,846	£6,161	£7,029	**£20,036**
Dudley	£8,352	£7,517	£8,575	**£24,443**
Sandwell and West Birmingham	£14,650	£13,185	£15,042	**£42,876**
Solihull	£6,435	£5,791	£6,607	**£18,833**
Walsall	£7,503	£6,753	£7,704	**£21,959**
Wolverhampton City	£7,174	£6,457	£7,366	**£20,997**
Cannock Chase	£3,615	£3,253	£3,711	**£10,579**
East Staffordshire	£3,697	£3,327	£3,796	**£10,819**
North Staffordshire	£5,833	£5,249	£5,989	**£17,070**
Shropshire	£8,215	£7,393	£8,435	**£24,043**
South East Staffordshire and Seisdon Peninsular	£5,750	£5,175	£5,904	**£16,830**
Stafford and Surrounds	£3,943	£3,549	£4,049	**£11,541**
Stoke-on-Trent	£7,640	£6,876	£7,844	**£22,360**
Telford and Wrekin	£4,710	£4,239	£4,836	**£13,785**
Totals	£160,080	£144,072	£164,361	**£468,513.00**

6.1.3 Table 6 Year 3 cost breakdown per CCG

Year 3 includes 12 months nursing salaries. Potential projected savings based on the avoidance of 126 MI's throughout the regional have been included. As per NICE guidance these savings are realised after year 2, from a reduction in CHD within the FH population which can then be offset against the service cost.

CCG	£250.00 per index case	Cascade cost/relative @£75	Year 3 including: Salaries, consumables & Non-FH Clinical consumables	Year 3 totals including genetic costs	projected Cost Savings (reduction in MI's)
Coventry and Rugby	£12,596	£11,336	£22,769	**£46,689**	£ 32,579.00
Herefordshire	£5,093	£4,584	£9,206	**£18,879**	£ 13,164.00
Redditch and Bromsgrove	£4,655	£4,190	£8,415	**£17,255**	£ 12,046.00
South Warwickshire	£7,393	£6,654	£13,364	**£27,405**	£ 19,125.00
South Worcestershire	£7,892	£7,103	£14,265	**£29,242**	£ 20,408.00
Warwickshire North	£5,038	£4,535	£9,107	**£18,676**	£ 13,039.00
Wyre Forest	£3,067	£2,760	£5,544	**£11,368**	£ 7,948.00
Birmingham Cross City	£19,989	£17,991	£36,133	**£74,094**	£ 51,704.00
Birmingham South and Central	£6,846	£6,161	£12,374	**£25,375**	£ 17,717.00
Dudley	£8,352	£7,517	£15,097	**£30,957**	£ 21,609.00
Sandwell and West Birmingham	£14,650	£13,185	£26,481	**£54,302**	£ 37,877.00
Solihull	£6,435	£5,791	£11,632	**£23,852**	£ 16,641.00
Walsall	£7,503	£6,753	£13,562	**£27,811**	£ 19,415.00
Wolverhampton City	£7,174	£6,457	£12,968	**£26,593**	£ 18,545.00
Cannock Chase	£3,615	£3,253	£6,534	**£13,398**	£ 10,845.00
East Staffordshire	£3,697	£3,327	£6,682	**£13,702**	£ 9,562.00
North Staffordshire	£5,833	£5,249	£10,543	**£21,619**	£ 15,068.00
Shropshire	£8,215	£7,393	£14,849	**£30,450**	£ 21,236.00
South East Staffordshire and Seisdon Peninsula	£5,750	£5,175	£10,394	**£21,315**	£ 14,861.00
Stafford and Surrounds	£3,943	£3,549	£7,128	**£14,616**	£ 10,183.00
Stoke-on-Trent	£7,640	£6,876	£13,810	**£28,318**	£ 19,746.00
Telford and Wrekin	£4,710	£4,239	£8,514	**£17,458**	£ 12,170.00
Totals	£160,080	£144,072	£289,361	**£593,361**	£413,966.70

6.1.4 Table 7 Year 4 cost breakdown per CCG

Year 4 includes 12 months nursing salaries. Potential projected savings have again been included as per NICE guidance.

CCG	£250.00 per index case	Cascade cost/relative @£75	Year 4 including: Salaries, consumables & Non-FH Clinical consumables	Year 4 totals including genetic costs	projected Cost Savings (reduction in MI's)
Coventry and Rugby	£12,596	£11,336	£22,965	**£46,886**	£ 32,579.00
Herefordshire	£5,093	£4,584	£9,286	**£18,958**	£ 13,164.00
Redditch and Bromsgrove	£4,655	£4,190	£8,487	**£17,327**	£ 12,046.00
South Warwickshire	£7,393	£6,654	£13,480	**£27,520**	£ 19,125.00
South Worcestershire	£7,892	£7,103	£14,389	**£29,365**	£ 20,408.00
Warwickshire North	£5,038	£4,535	£9,186	**£18,754**	£ 13,039.00
Wyre Forest	£3,067	£2,760	£5,592	**£11,416**	£ 7,948.00
Birmingham Cross City	£19,989	£17,991	£36,445	**£74,406**	£ 51,704.00
Birmingham South and Central	£6,846	£6,161	£12,481	**£25,482**	£ 17,717.00
Dudley	£8,352	£7,517	£15,227	**£31,088**	£ 21,609.00
Sandwell and West Birmingham	£14,650	£13,185	£26,710	**£54,531**	£ 37,877.00
Solihull	£6,435	£5,791	£11,732	**£23,953**	£ 16,641.00
Walsall	£7,503	£6,753	£13,679	**£27,928**	£ 19,415.00
Wolverhampton City	£7,174	£6,457	£13,080	**£26,705**	£ 18,545.00
Cannock Chase	£3,615	£3,253	£6,590	**£13,454**	£ 10,845.00
East Staffordshire	£3,697	£3,327	£6,740	**£13,760**	£ 9,562.00
North Staffordshire	£5,833	£5,249	£10,634	**£21,710**	£ 15,068.00
Shropshire	£8,215	£7,393	£14,977	**£30,578**	£ 21,236.00
South East Staffordshire and Seisdon Peninsula	£5,750	£5,175	£10,484	**£21,405**	£ 14,861.00
Stafford and Surrounds	£3,943	£3,549	£7,189	**£14,677**	£ 10,183.00
Stoke-on-Trent	£7,640	£6,876	£13,929	**£28,437**	£ 19,746.00
Telford and Wrekin	£4,710	£4,239	£8,587	**£17,531**	£ 12,170.00
Totals	£160,080	£144,072	£291,871	**£595,871**	£413,966.70

Appendix 1 gives further detail of population split. See appendix 2 for detailed workings for cost breakdown over years 1-4.

Genetic and cascade test costing assumptions have been made based on the Bristol Genetics Laboratory current guidance. These assumptions may need to be revisited if the genetic service provision associated with this FH service is procured via competitive tender.

Staff costs will be provided 100% in year one by the BHF and then 50% in year two. As patients will be seen in a primary care setting, a tariff will not be allocated for outpatient appointments. Commissioners may seek to move this to a local primary care/community local currency and tariff post year 2 with an all-inclusive index tariff and cascade tariff. Phase two costing assumptions (from 2017-18) have been based on the criteria of testing bought down from cholesterols levels of 9 and above down to 7.5 and above, in line with NICE guidance. This would be subject to evaluation of stage 1 and prioritisation by commissioners in 17/18, to consider whether to lower the threshold or remain with phase 1. This will depend upon the success of stage 1 and other competing calls on their finite resources.

The cost of DNA testing will reduce in phase three, as well as other cascade testing costs although this is negligible.

Staff costs (specialist nurses and admin) have been projected to remain stable, although pending evaluation at the end of phase one and two it may be appropriate to stop these posts or reduce capacity. However as there will be a large patient population, these projections assume at the moment that these staff members will be retained, possibly to reduce the cost and improve the cost effectiveness of the annual review process through the delivery of nurse-led clinics in primary care.

6.1.5 Rate of change

There are factors that may impact upon the rate of uptake and future growth, these include:

- the speed at which known clinical cases are identified and tested
- the number of relatives per index case living in the West Midlands and willing to be tested
- the number of additional cases diagnosed clinically, usually through primary care, for example through NHS Health checks or family history that identifies more patients with cholesterol greater than 9.

7 Benefits of the Model

From the point of view of commissioning organisations, implementing the proposal will enable compliance with NICE Guidance that is already implemented across Wales, Scotland, Northern Ireland and parts of England.

It will also help to meet individual CCG priorities:

- Effective commissioning of a regional and consistent genetic services model will ensure consistent equity of access, enabling preventative actions to be taken for those at genetic risk of developing serious and life threatening illness and disease.
- Identifying and treating FH increases life expectancy in some cases by more than 9 years.
- Identifying and treating FH will have an impact of premature cardiovascular morality.
- Identifying and treating FH will increase control of cholesterol

From the point of both primary and secondary providers it will allow a consistent approach to be adopted across the region, based on agreed standards. It will enable primary care providers to reduce the burden of cardiovascular disease in their populations whilst focusing secondary care resources on those patients who most need them. We also envisage that this service will enable patients to increasingly become accountable care officers for their own care through the provision of a definitive molecular diagnosis, clear identification of associated lifestyle risk factors (e.g. smoking, adiposity) – for themselves and their affected family members, and co-production with patients of interventions (e.g. drug treatments, lifestyle changes) which are known to minimise disease progression in this setting.

7.1 Impact on the health system

7.1.1 Patients' Lives

The best case scenario arising from implementation will be that, for a proportion of patients will have their CHD risk will have been prevented entirely and for others, their serious sequellae of atherosclerosis will be are reduced.

Through the implementation of a regional service the worst case scenario will be that the onset of coronary heart disease will be delayed by many years for many individuals. However the intervention will have added many years to individual lives.

Because of the mode of genetic inheritance, FH is concentrated in specific families, where 50% of close family members will be affected. Untreated FH will therefore have an impact on many specific families within the region where half of all family members may have an MI by the age of 60, and/or symptoms of coronary heart disease before this age.

Research from Wales has found that patients who received a positive DNA test found the explanation for their high cholesterol reassuring, that it provided clarity, and encouraged family cascading. The negative impact in some patients was the worry about the use of their personal data, particularly for their future life however this is in common with many health care scenarios, particularly genetic testing, and highlights the need for robust information governance and consent to testing. We will mitigate for these risks through the implementation stage and ensuring governance arrangements are in place.

7.1.2 Clinical management

Genetic testing for FH is currently not available for patients in the West Midlands. Implementing the NICE Guidelines will enable high quality evidence based personalised medical care to be provided consistently across the region for this high priority condition. Implementing this programme will have an impact on avoidable cardiac mortality.

If successful the total numbers of FH cases may quadruple through the roll out of phases 1-3. Implementing an agreed care pathway and standards will facilitate high quality clinical care across the entire region.

Adopting this proposal will ensure that what is an almost inevitable increase in activity is evidence based and cost-effective, and contributes to improved healthcare outcomes.

7.1.3 Primary care

It is expected that the 5 BHF FH specialist nurses will work peripatetically across the region holding specialist clinics in primary care settings. GPs will be involved at all stages of the care pathway particularly in the long term management of the majority of patients.

The pathway has been designed to ensure that investment is used in the most cost-effective way. This will be achieved through tight control over who can order a genetic test, namely BHF FH nurse specialists.

There is an explicit need for GPs to refer clinical cases to FH specialists in order to access confirmation and testing, which may represent a change in practice for some GPs. It is expected that secondary care lipid clinics will see an increase in new appointment activity of approximately 10% per year which equates to less than 40 patients per lipid clinic. This increase will be offset by the significant reduction in outpatient follow up activity where there is a current average ratio of 1:4 new to follow up appointments. As part of the

implementation stage we will also look to reduce this further with protocols and EReferral processes that enable GPs to refer direct to specialist nurses. There will be an expectation that specialist clinics will refer 70-80% of patients back to primary care for long term management where it is appropriate and the primary care FH workload will therefore rise. An essential part of the BHF FH specialist nurse role will be to deliver training and education in primary care to support this model.

Patients with a cholesterol concentration of 9.0 mmol/L and above are selected because they are likely to have the greatest number of positive genetic tests (Humphries S et al Atherosclerosis April 2015) all of these patients will also be audited using a number of other tools that are available. This will include the Simon Broome criteria (NICE CG71), the use of the Medway study criteria (NICE), the use of the Dutch lipid clinic criteria (RACGP), the Welsh criteria for FH (HeartUK) and the FAMCAT data tool (Quereshi N et al Atherosclerosis 2015;238:336). As we progress through populations, progressively reducing the concentrations of cholesterol that patients have who are screened, the service can audit the outcomes of the positive genetic patients can be reviewed using these screening data criteria to improve the screening criteria. The ethnic diversity of the West Midlands population will be a good environment to test whether these screening tools can detect cases of FH in the ethnic minority groups as FH is a universally expressed cardiovascular risk.

7.1.4 Secondary care

Secondary care services are currently provided in 12 locations throughout the region. Following the commissioning of this regional service it is expected that patient numbers will rise. The NICE Guidelines recommend that the long term management of the majority of adult patients (approximately 70%) can be in Primary Care, which will require FH Specialists to discharge appropriate patients to their GPs. If all 11,000 patients were found the specialist workload should increase to 3,300 long term patients with majority of patients managed in primary care.

7.1.5 Delivering a genomics strategy across populations

The current focus on integrating genomics into main stream medicine suggests that moving forward with a programme like FH cascade screening would provide West Midlands CCGs with an opportunity to develop an experience and evidence base with which to inform future commissioning intentions in this space. The model described for delivery in this paper could for example allow for prospective evaluations of impact through clinical outcome, cost analyses and patient reported outcome measures.

7.1.6 Laboratory services

There is currently limited genetic testing being undertaken within the West Midlands for FH and this will therefore represent additional work.

8 Governance, Audit & Procurement

8.1 Management of the Service

For the first 2 years, the service will be contracted by Birmingham Cross City CCG with the host regional centre UHBFT, separate contracts maybe held with the contracted genetic testing laboratories; this will be scoped as part of implementation planning. At the end of year 2 we may revise contracting arrangements as CCG areas may wish to align other primary care contracting mechanisms.

The service will be implemented and led by a multidisciplinary team composed of West Midlands CCGs, WMSCN, UHB Rare Disease Centre, Lead FH Consultant (UHB), and Lead Paediatrician (BCH). This currently is the FH operational group. This team will form the strategic body that will oversee the implementation and evaluate through the first 2 years the proposed service model and outcomes. *See appendix 2, Fig 1 Strategic Overview organigram.*

The service team will be led by the regional host centre UHBFT. The service delivery team comprise of the Lead Paediatrician (BCH), Lead FH Consultant (UHB), and Lead Geneticist (UHB and genetics provider). The lead FH Consultant will have direct line management of the senior Band 8a BHF FH Specialist Nurse, who will then have line management responsibility for the four Band 7 BHF FH Specialist Nurses. The nursing team will feed into and support local lipid services in their respective geographical catchment areas. *See appendix 2, Fig 2 Operational Overview*

8.1.1 Professional competency, education and training

The BHF FH Specialist Nursing team will attend a regular programme of education, training and support, provided by BHF. The host organisation will ensure safe staffing capacity at all times and will ensure that staff are able to demonstrate that they have participated in organisational mandatory and update training, for example infection control, manual handling, information governance, risk assessment as required. BHF FH Specialist Nurses will attend BHF genetic counselling training provided by BHF.

8.1.2 Clinical audit and review

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

8.1.3 Information management

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

8.1.4 Equipment

Providers will be expected to adhere to Medicines and Healthcare Regulatory products Agency (MHRA) advice and guidance on selection of appropriate equipment, training in its use and ongoing management, troubleshooting, and quality assurance processes that ensure the accuracy and reproducibility of test results. UHBT will be providing IT equipment to the BHF FH Specialist Nursing team whilst acting as hot organisation and will provide IT support. This should be reviewed when BHF funding ceases.

9 Implementation

It is expected that the FH regional service should commence April 2016. To ensure this timeline can be adhered to the following tasks will be completed prior to start date;

- Development and oversight of implementation plan via FH operational group.
- Finalise regional service model via FH operational group.
- Recruitment of BHF FH specialist Nurses via host centre.
- Finalise contract products including specifications, KPIs, PAMs.
- CCG identification and agreement of local primary care clinic space.
- Procurement for genetic testing.
- Procurement of PASS licences from Heart Development of referral forms and service protocols.
- Nurses build case load and attend relevant training via BHF.
- Internal launch.
- Public launch.

10 Recommendations

National strategies to reduce the burden of cardiovascular disease in the UK would be made more effective and more cost effective by incorporating the screening strategy that was recommended in national guidance from NICE for the identification and treatment of people with Familial Hypercholesterolemia

The proposal recommends the following;

1. CCGs approve the proposed West Midlands model of care for the identification and management of FH.
2. CCGs approve the host arrangements for the service should be via the regional centre.
3. CCGs agree to continue to fund FH specialist Nurses post BHF funding.
4. CCGs agree funding for genetic and cascade testing.
5. CCG approve funding for phase 1 at a total cost of £348,648

11 Bibliography

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NICE FH guidelines (CG71); the NICE Quality Standard on FH (QS41); https://www.**nice**.org.uk/guidance/**cg71**

The Wales FH cascade testing Initiative – Heart UK; heartuk.org.uk/images/uploads/healthylivingpdfs/imcdowell.pdf

12 Appendices

Appendix 1 Expected number of cases per CCG based on mid 2014 population estimates extracted from CCG websites (accessed 2014).

Appendix 2 Costings spreadsheet

Appendix 3, Fig 1 Strategic Overview organigram Fig 2 Operational Overview organigram

Appendix 1: Expected number of cases per CCG. Population counts referenced from individual CCG websites (2014).

CCG	General information	
	Population	FH population 1/500
Coventry and Rugby	460,000	920
Herefordshire	186,000	372
Redditch and Bromsgrove	170,000	340
South Warwickshire	270,000	540
South Worcestershire	288,000	576
Warwickshire North	184,000	368
Wyre Forest	112,000	224
Birmingham Cross City	730,000	1460
Birmingham South and Central	250,000	500
Dudley	305,000	610
Sandwell and West Birmingham	535,000	1070
Solihull	235,000	470
Walsall	274,000	548
Wolverhampton City	262,000	524
Cannock Chase	132,000	264
East Staffordshire	135,000	270
North Staffordshire	213,000	426
Shropshire	300,000	600
South East Staffordshire and Seisdon Peninsular	210,000	420
Stafford and Surrounds	144,000	288
Stoke-on-Trent	279,000	558
Telford and Wrekin	172,000	344
Totals	5,846,000	11,692

Appendix 2 – costings spreadsheet hyperlink

FH activity and finance 2015.xlsx

Appendix 3: Fig 1 Strategic Overview organigram

Strategic overview

The following diagram identifies the strategic body that will oversee the running of the proposed service model and encompasses the multidisciplinary team who will deliver the service.

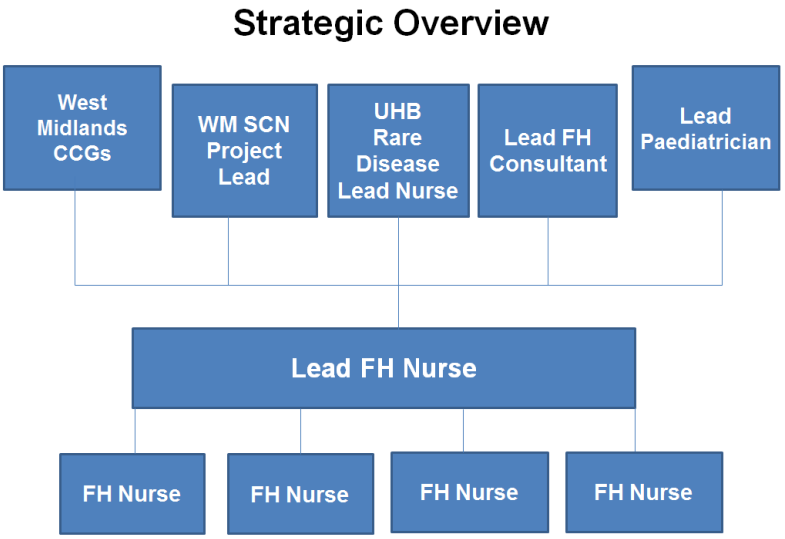

Fig 2 Operational Overview organigram

Operation Overview

The diagram shown below identifies the operational team that will deliver the FH service.