



**Dorset
Clinical Commissioning Group**

NHS Dorset Clinical Commissioning Group

FAMILIAL HYPERCHOLESTEROLAEMIA



Supporting people in Dorset to lead healthier lives

SCHEDULE 2 – THE SERVICES

A. Service Specifications

Service Specification No.	03/CVDS/0014
Service	Familial Hypercholesterolaemia Service - Advice, guidance, Diagnostic Assessment, Cascade Testing and Initial Management
Commissioner Lead	Cardiovascular Clinical Commissioning Programme
Provider Lead	<i>Chemical Pathology</i>
Period	<i>In line with contract</i>
Date of Review	<i>September 2014</i>

1. Population Needs

1.1 National/local context and evidence base

The cardiovascular disease outcomes strategy 2013 requires CCGs to put in place processes for identifying families with inherited cardiac conditions. Familial hypercholesterolaemia (FH) is a genetic condition which affects lipid metabolism resulting in raised total and LDL (low-density lipoprotein) cholesterol. The gene mutation of heterozygous FH estimates a prevalence of 1 in 500 UK population, whereas the homozygous FH is 1 per million. In Dorset the estimate is that approximately 1540 people have (FH).

If people at risk of FH are identified and treated with lipid lowering medication before the establishment of CHD they have normal outcomes. If treatment is not instigated until CHD symptoms present, treatment is less effective. NICE predict that with treatment there will be 2 Myocardial Infarctions avoided per year and 1 revascularisation event, as well as better management of risk for heart failure.

2. Outcomes

2.1 NHS Outcomes Framework Domains & Indicators

Domain 1	Preventing people from dying prematurely	√
Domain 2	Enhancing quality of life for people with long-term conditions	√
Domain 3	Helping people to recover from episodes of ill-health or following injury	
Domain 4	Ensuring people have a positive experience of care	
Domain 5	Treating and caring for people in safe environment and protecting them from avoidable harm	

2.2 Local defined outcomes

The expected outcomes of this service will be:

- Improve the detection of families at risk of heart disease and achieve 90% detection rates within 5 years
- Identify through Cascade testing individuals and their families who require on-going management in primary and secondary care
- Identify those adults requiring on-going management within secondary care. Expected level is 20% of newly diagnosed
- Maintain all information uploads to the national database
- Provide advice and guidance to primary care in the management of people at risk
- Provide an educational programme for primary care to support introduction of the service and use of the referral criteria
- Provide advice and guidance to primary care on the appropriateness of referrals
- To network with other providers of FH services in NHS England (Wessex)

3. Scope

3.1 Aims and objectives of service

The aim of the service is to:

- support primary care in identifying patients appropriate for referral to the service, including an electronic advice and guidance service
- ensure lipid testing is undertaken in line with NICE guidance
- Provide advice to primary care on diagnosis and lipid management
- Assess individual patient risks, and draw up a family pedigree and if genetic testing is appropriate. Support individuals who do not wish to undertake genetic testing.
- Enter individual/ family into national database to help inform type of genetic tests to be requested

- Work with a genetic testing service and request tests ensuring best value
- Review results of genetic tests and family pedigree to inform cascade genetic testing within the family
- Provide advice to family members who live outside of Dorset, who will not receive testing within Dorset, to see their local GP and the details of their family mutation
- Provide advice and support to primary care where genetic testing is negative
- Support families post genetic testing and agree if on-going management in secondary care is required and where this support will be provided (model for on-going care for children still to be agreed). One consultation follow up expected for most people.
- Provide a management plan to primary care after diagnosis
- Ensure advice and guidance to primary care is available through electronic and telephone means
- Provide additional support to pregnant women, women considering pregnancy, people with multiple statins intolerance or contraindications to statins, and homozygous FH
- Refer to adult cardiology service and other specialist services when appropriate.
- Provide advice and guidance to people managed by other specialist teams
- Provide an accessible service carried out by appropriately trained and qualified staff
- Provide patients with timely information at all stages of the care pathway
- Provide support and guidance on lifestyle behavioural change required

3.2 Service description/care pathway

The service will receive referrals from primary care in Dorset CCG and secondary care. The service will look to develop clinics in the East and West of the County as the service develops.

The service will:

- provide telephone and email advice to referrers to inform referrals
- on receipt of referral checks will be made of the lipid profile and family history to ensure compliance with the referral criteria
- referrals will be returned to referrers when the request does not meet the criteria in 3.3 below. .
- provide advice and guidance to the individual on FH and implications
- discuss limitations of genetic testing
- Draw up a family pedigree
- Gain consent for genetic testing
- Entry of individual and family on national database
- Initiate genetic testing or testing for known familial mutation
- Following positive genetic test co-ordinate cascade testing for family members registered with Dorset CCG practices
- Following negative genetic test provide advice to primary care and discharge the individual

- Following positive genetic test work with the practice to identify the adults requiring management in secondary care (those where lipid levels are not reduced by 50%). This is assumed to be about 20% of adults and will require on-going monitoring
- Work with specialist teams already supporting adults with FH eg. People with established heart disease and diabetes
- When genetic testing identifies mutation of unknown significance consider if age, gender, specific LDL concentration and patient's phenotype is sufficiently suggestive of FH to support cascade testing to family. Ensure introduction of lipids management regimes if appropriate and provide advice to primary care
- Develop patient information about local services and signpost to nationally developed material
- Follow a health coaching approach to care to support lifestyle behavioural change
- Develop a clinical network with other providers of FH services

3.3 Any acceptance and exclusion criteria and thresholds

The service will not assess and initiate cascade testing for people who are not registered with Dorset CCG.

The service will accept referrals from local practices and other secondary care services in line with the criteria below:

Possible FH

Total LDL >5.5 mmol/l in adult AND a triglyceride concentration less than 4.0 mmol/l (measurements either pre-treatment or highest on treatment)

PLUS ONE OF THE FOLLOWING:

Family history of myocardial infarction before age 50 in 2nd degree relative or before age 60 in 1st degree relative

Family history of raised total cholesterol concentration above 8.0 mmol/l in 1st or 2nd degree relative

Definite FH

The above criteria plus Tendon xanthomata (or evidence of these in 1st or 2nd degree relatives)

And /or DNA-based evidence of an LDL receptor mutation, familial receptor defective apo-B100, or a PCSK 9 mutation in patient or 1st or 2nd degree relative

3.4 Interdependence with other services/providers

The practices will be referring to the service using the referral criteria detailed in 3.3. Close working will be required with all practices within the CCG and with the established lipid clinic in the East of the County. The service will develop a primary care education programme to support introduction of the new service.

The maintenance of close links with the Wessex genetic service will be required to ensure on-

going evidenced based treatment.

The service will require close working relationships with:

- Cardiology teams in: Yeovil, Salisbury, Bournemouth, Poole and Dorchester
- Commissioners
- Genetics testing provider

The service will co-ordinate care with the provider of long term follow up for children and adults (this will is still under consideration).

4. Service

4.1 Applicable national standards (e.g. NICE)

NICE clinical guideline 71, Identification and Management of Familial Hypercholesterolaemia

NICE clinical guideline 67, Lipid Modification

4.2 Applicable standards set out in Guidance and/or issued by a competent body (e.g. Royal Colleges)

4.3 Applicable local standards

The service will develop training plans to ensure that staff undertake a recognised health coaching training programme.

5. Applicable quality requirements and CQUIN goals

5.1 Applicable quality requirements (See Schedule 4 Parts A-D)

Service monitoring requirements on a rolling monthly basis are defined below. This is a new service and the quality standards will be developed further after the service set up period which will be defined as 2 years:

- Follow up rates
- Percentage of total referrals managed through telephone or email advice
- Percentage of referrals returned as not meeting referral criteria
- Percentage of adults referred to be managed in specialist care

Performance criteria to be reflected within contract:

- 1) Total number of referrals by practice
 - a. Given advice and guidance
 - b. Returned pre consultation following triage as they did not meet referral criteria
 - c. Discharged from service with advice and guidance
- 2) Number of individuals and family members undergoing genetic testing, adult and children separately identifiable
- 3) Number of positive tests

- 4) Number of children referred into on-going care
- 5) Number of adults referred into on-going care and care provider
- 6) Follow up rate – Future updates of this specification will agree a rate of follow up

CCG will monitor :

Waiting times from referral to decision to genetic test or discharge with advice and guidance.

The Provider will engage with the CCG on an annual basis to agree patient engagement. At the end of the first year this will include patient/family interviews.

5.2 Applicable CQUIN goals (See Schedule 4 Part E)

None

6. Location of Provider Premises

The Provider's Premises are located at:

To be agreed

7. Individual Service User Placement

Not relevant