

## Factsheet: Early diagnosis of people with Familial Hypercholesterolaemia (FH) in England

The benefits of early diagnosis of FH would be reduction in premature deaths from heart disease; a reduction in long-term morbidity and its associated costs and 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 cost of hospital admissions.

All figures per year	England	100,000	Comments
Potential lives saved from intervention	42	0.08	Evidence suggests very few lives saved at over 75. 42 is achievable after a few years of cascade testing. Costs are lower in the early years when there are fewer people to test and treat.
Potential lives saved (U75)	42	0.08	Cascade testing and improved treatment would also reduce AMI. Cost savings from these are included. Costs are from NICE 2008 costing guidance. Staff costs have increased since this time but drugs are off patent, reducing their price significantly.
Reduction in Potential Years of Life Lost (U75) from early diagnosis of FH	1606	3	
Cost saving (£)		£21,570	
Cost (£)		£3,408	
Net cost (£)		£18,162	
<b>Strength of evidence</b>	<b>1</b>		

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 percent of cases identified in the UK<sup>Error! Bookmark not defined.</sup>. Children of an individual with FH have a 50 percent chance of inheriting the condition. Left untreated, FH may lead to premature death from coronary heart disease (CHD). 50 percent of males and 30 percent of females with untreated FH will have developed CHD

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by the age of 55<sup>1</sup>. 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.

### **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 very cost-effective, with an estimated ICER (incremental cost effectiveness ratio) of £2,700 per quality adjusted life year (QALY); well below the NICE cost effectiveness threshold of £20-30,000/QALY<sup>12</sup>.

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 of course 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 cost of hospital admissions.

The cholesterol charity HEART-UK has published a report<sup>2</sup> (“Saving Lives, Saving Families: The health, social and economic advantages of detecting and treating Familial Hypercholesterolaemia”) and have shown how England is falling behind the devolved nations of the UK in diagnosing and treating FH. The devolved countries each have a national directive or initiative specifically targeting FH, which has helped achieve higher standards of care for their FH patients.

- High intensity treatment would mean 101 cardiovascular deaths could be avoided per 1,000 FH patients (aged 30 to 85 years) treated, when compared with no treatment
- The UK could save £378.7 million from cardiovascular events avoided if all (100 percent) relatives of FH index cases are identified and treated optimally over a 55 year period, or £6.9 million per year.

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<sup>1</sup> NHS England (2013) *Briefing: Familial Hypercholesterolemia in England* [Online] available from: [http://www.england.nhs.uk/wp-content/uploads/2013/11/fh\\_eEngland-briefing11\\_2013.pdf](http://www.england.nhs.uk/wp-content/uploads/2013/11/fh_eEngland-briefing11_2013.pdf) [Accessed November 2013]

<sup>2</sup> Heart UK (2012) *Saving lives, saving families* [pdf] available from: [http://heartuk.org.uk/files/uploads/HUK\\_SavingLivesSavingFamilies\\_FHreport\\_Feb2012.pdf](http://heartuk.org.uk/files/uploads/HUK_SavingLivesSavingFamilies_FHreport_Feb2012.pdf) [accessed November 2013]

## Recent policy and publications

In March 2013 the Department of Health published its Cardiovascular Outcomes Strategy, endorsed by NHS England, and PHE. Among its priorities, the Strategy set the initial ambition of identifying at least 50 percent 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. The Quality Standard

supports the delivery of the NHS Outcomes Framework, helping to fulfill 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).

## Useful resources

Cardiovascular Outcomes Strategy:

[https://www.gov.uk/government/uploads/system/uploads/attachment\\_data/file/214895/9387-2900853-CVD-Outcomes\\_web1.pdf](https://www.gov.uk/government/uploads/system/uploads/attachment_data/file/214895/9387-2900853-CVD-Outcomes_web1.pdf)

NICE FH Quality Standard (QS41): <http://guidance.nice.org.uk/QS41>

HEART UK Report 'Saving lives, saving families'

[http://heartuk.org.uk/files/uploads/HUK\\_SavingLivesSavingFamilies\\_FHreport\\_Feb2012.pdf](http://heartuk.org.uk/files/uploads/HUK_SavingLivesSavingFamilies_FHreport_Feb2012.pdf)

HEART UK FH Toolkit (aimed at helping commissioners and clinicians deliver on the NICE FH Guideline): <http://heartuk.org.uk/FHToolkit/>