Briefing: Familial Hypercholesterolaemia in England

About Familial Hypercholesterolaemia (FH)

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. Left untreated, FH may lead to premature death from coronary heart disease (CHD). 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.

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 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 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 **(“**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 will mean 101 cardiovascular deaths are 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 per cent) relatives of FH index cases are identified and treated optimally over a 55 year period, or £6.9m per year.

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 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).

The **British Heart Foundation** has agreed to provide over £1m of funding to support the employment of specialist FH nurses or other key staff, but clinical commissioning groups (CCGs) will be instrumental in delivering sustainable FH services; their partnership will be required if BHF funds are to be unlocked. Improving FH detection and management is a worthwhile, cost and clinically effective objective, which is long overdue being given some priority. If we can do better many lives will be saved, and many families spared the current inevitability of living with or dying from premature cardiovascular disease. CCG help and support would be greatly appreciated.

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Useful resources

Cardiovascular Outcomes Strategy:
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 FH Report – Saving lives, saving families
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/