Response to NHS Chief Executive’s Open Call for Evidence and Ideas

Respondent ID: 91

Organisation name: Heart UK

Type of response: Email and document
Dear Sir/Madam,

Spreading innovation in the NHS – Submission from HEART UK

HEART UK, the Cholesterol Charity, is passionate about preventing premature deaths caused by high cholesterol and cardiovascular disease, including forms of inherited high cholesterol such as Familial Hypercholesterolemia (FH). FH is a genetic condition which causes high concentrations of cholesterol in the blood. People with FH are at very high risk of developing coronary heart disease (CHD) from a young age – sometimes suffering major or fatal heart attacks or strokes in their 20s and 30s. This is a tragedy for families and a significant burden on the NHS and the economy.

The genetic nature of FH means that siblings and offspring of people with the condition have a 50 per cent chance of inheriting it. It is critical that family members of patients diagnosed with FH are tested so that they can benefit from early management and treatment. Approximately 120,000 people in the UK have FH, but only 15,000 patients have been identified and treated. This means thousands of people are still unaware that they have FH and that their family members are at risk.

HEART UK has been working hard to address this diagnosis gap. In 2008, NICE published a clinical guideline on the identification and management of patients with FH which called for cascade screening of family members. Three years on, implementation is proving very slow. HEART UK has run a series of regional meetings to engage directly with clinicians and commissioners to mobilise action for services to be rolled-out locally. We also developed a comprehensive toolkit to assist this process, which was endorsed by Professor Sir Roger Boyle.

Whilst we have encountered high levels of support at a local level, there is yet to be any real progress in implementing a comprehensive service for FH patients in England. Yet the rollout of FH cascade screening is an innovation that we should better utilise. FH is an obvious priority among genetic diseases because of its high prevalence. Unlike many genetic conditions, FH is easily treatable, with excellent prognosis if diagnosed early and treated properly.

In the long term, we believe that making all NICE guidance mandatory would ensure that clinical guidelines are rolled out as intended.

HEART UK’s recommendations for improving the delivery of FH services in England are outlined in the attached briefing paper. We are currently producing a new Report on FH, which will detail these recommendations. The Report will be launched in the New Year and will include newly-commissioned health economic modelling. The draft research indicates that considerable savings can be made if FH services are properly rolled out. Indeed, these findings echo the recent remarks of Professor Sir John Bell concerning the untapped medical and economic value of genetic technology.

We would be pleased to share these findings with you when available, and would welcome an opportunity to meet to discuss FH and the options for improving rollout of cascade testing.

Thank you for the opportunity to submit to this review.

Your sincerely,
We're passionate about preventing premature deaths caused by high cholesterol.

Thread ‘em red and join the Teamredlaces Challenge today! Click here to find out more
Developing a national programme for Familial Hypercholesterolaemia in England

About HEART UK
HEART UK - The Cholesterol Charity - is passionate about preventing premature deaths caused by cardiovascular disease (CVD). The charity aims to help families with a high risk of premature CVD, especially those with inherited high cholesterol (familial hypercholesterolaemia, FH). More than three-quarters of people with FH are undiagnosed, probably untreated and thus remain at serious risk of premature death from CVD. HEART UK has formed a Guideline Implementation Team, which aims to support the implementation of the NICE Guideline on FH (2008).

About Familial Hypercholesterolaemia
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 15,000 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 CVD. 50% of males with untreated FH will develop coronary heart disease (CHD) before the age of 50 (for females, 50% have CHD before age 60). 50% of untreated males will die before they are 60. It is important to recognise that these deaths are 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 genetic testing of families, by a method known as cascade screening. Referral to specialist lipid clinics is recommended for confirmation of the diagnosis, patient counselling and in order to initiate cascade screening.

The NICE Guideline indicates that the cascade testing model for diagnosing FH is the most 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 £20,000/QALY.

Benefits from initiatives to find cases of FH would 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.

FH in other countries of the UK
The devolved countries each have a national directive or initiative specifically targeting FH, which has helped them achieve higher standards of care for their FH patients than for those in England.

Scotland’s Better Heart Disease and Stroke Care Action Plan (2009) includes the development of a national forum for FH. When a proband (or first case) of FH is found, genetic testing is performed in a central laboratory and the results used for ‘cascade testing’ within the family to find other cases.
The Welsh Assembly has provided support for a cascade testing initiative in Wales. The project combines an FH pilot and the development of an IT system for following up families.

Northern Ireland’s *Service Framework for Cardiovascular Health and Wellbeing* (2009) includes a standard for the identification and treatment for people with FH. Active steps are being undertaken to implement these guidelines, with the recruitment of FH screening nurses to support the five lipid clinics, using a central database.

**The current situation in England**

In 2010, to better understand the extent to which the NICE FH Guideline has been implemented in England, HEART UK conducted a study in which Freedom of Information requests were sent to Primary Care Trusts, asking about their progress to date. The study showed that little has been done to implement the recommendations of the Guideline in England. The findings demonstrate a lack of formal planning for FH and incomplete provision of clinical services and education about FH. There is a paucity of specialist services, including provisions for paediatric, obstetric, as well as adult patients. The NHS Health Checks programme recommends that people identified with total cholesterol > 7.5 mmol/L and with a family history of premature coronary heart disease should be considered as a possible cases of FH, with referral advised according to local clinical guidelines. However, identification continues to take place on an ad hoc basis, and the commissioning of FH services remains very limited.

**Lipid clinics**

Specialist lipid clinics are critical for the assessment and treatment of people with FH and other complicated lipid disorders, as they can provide the expertise required to ensure that the correct diagnosis is made and that genetic tests are used cost-effectively. Additional capacity will be required to accommodate adults and children with FH identified through cascade testing. Access and availability of lipid clinics are important aspects of a comprehensive, equitable health service. A recent paper on cascade testing for FH recommends that a lipid clinic is needed for every 200,000 people. Lipid clinics are currently far too scarce across the UK, with clinics offering specialist services to children and young people being particularly rare.

**HEART UK recommended actions**

- **A national programme is needed to tackle FH in England.** Localised commissioning has failed to deliver results for people with FH. Without a national programme, serious health inequalities will emerge for people in different countries of the UK
- **Rollout of the NICE Guidelines for FH in England.** This should be overseen by the National Commissioning Board, using dedicated, ring fenced funding
- **The development of a family-based follow-up IT system and UK patient register.** The IT system currently being developed in Wales is aimed at helping to coordinate cascade testing and register FH patients. Software based on the Dutch FH screening programme is being adapted and piloted for use in the UK. The learnings from the Welsh experience could inform a UK-wide system. This will enable more efficient tracking and diagnosis of people with possible FH in families throughout Britain
- **Additional lipid clinics should be established** to help manage patients with FH and other forms of inherited high cholesterol causing early heart disease.

**Contact**

HEART UK would be pleased to be involved in developing these actions into a comprehensive plan. For more information, please contact:
Slade Carter, Campaigning and Public Affairs Manager, HEART UK.
Email sc@heartuk.org.uk Tel. 07879 684 111

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