EDITORIAL

Familial hypercholesterolaemia – a developing English scandal

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As Chairman of HEART UK's Familial Hypercholesterolaemia (FH) Guideline Implementation Team, I am well aware that little has been done in England to implement the recommendations of the National Institute for Health and Clinical Excellence (NICE) guideline for the identification and management of FH (CG71), published in August 2008. This is, of course, in stark contrast to developments in Wales, Scotland and Northern Ireland, where my colleagues have made significant progress in identifying and treating patients with FH. However, even I was surprised by the findings of a study, commissioned by HEART UK – The Cholesterol Charity, in which freedom of information (FOI) requests were sent to primary care trusts (PCTs) in England, requesting information about their progress to date.

Findings

Nearly 70% of eligible PCTs responded to this survey, with many PCTs admitting to a lack of formal planning for FH, and incomplete knowledge about relevant FH services. Looking at the results as a whole, I think it is safe to say that the failure to implement the guidelines at PCT level suggests that national guidance is not always given local priority. More than 40% of the respondents admitted to a shortage of specialist care for people with FH, including adults, children and pregnant women. A number of PCTs indicated that they face barriers to treating FH patients and these concerns need to be addressed immediately if the guidelines are going to be successfully rolled out in England.

The key findings from this study make sober reading for both patients and clinicians working in this field:

- 60% of PCTs do *not* have written plans to support implementation of the NICE guidance on FH.
- Only 5% of PCTs provided us with evidence of written plans to support implementation.
- 28% of PCTs indicated that there are no specialist lipid clinics in their area and 25% "didn't know" where lipid clinics were that FH patients may be referred to. This suggests an imperfect knowledge of local services as there *are* likely to be lipid clinics in the local area – the HEART UK database currently lists 131 specialist lipid clinics in England.[1]

- 27% of PCTs did not know whether lipid clinics in the area could carry out DNA cascade testing in adult FH patients.
- The study showed a definite lack of lipid clinics or specialist centres available to children or young people with FH. Altogether, 41.5% of PCTs answered that there are no appropriate services in their area, and a further 26% did not know whether there were any appropriate services.
- Shared care of discharged FH patients has not been well developed: 63% of PCTs indicated that there are no written plans for shared care of discharged FH patients, with a further 20% of PCTs saying that they did not know if there are shared care arrangements.

Consequences

The estimated prevalence of FH is one in 500,[2] suggesting 120,000 individuals are affected in the UK. The condition is massively underdiagnosed, with only 15,000 cases identified in the UK.[3] The disorder has an autosomal dominant mode of inheritance, with children and siblings of a person with FH having a 50% chance of inheriting the condition. Left untreated, FH often leads to premature heart disease. The guideline recommends measures to find and diagnose cases of FH, using cholesterol and DNA testing of families known as cascade screening. Referral to specialist lipid clinics is recommended for confirmation of the diagnosis, patient counselling and initiating cascade screening.

With appropriate treatment as recommended by NICE, including the involvement of specialist lipid clinics in managing the condition, people with FH can live full and healthy lives.

The responses of English PCTs are very disappointing and suggest a lack of knowledge concerning FH. As we move to new forms of commissioning, informed input in specialised areas will be increasingly important and the new Primary Care Service Framework document on FH is available for advice.[3]

Of course, NICE guidelines are not compulsory, but we do recognise them as best clinical practice. As a result of this study, my colleagues at HEART UK and I have legitimate concerns that, if such guidance is not being implemented, patients cannot enjoy access to the best clinical diagnosis and care. Over the long term, increased diagnosis and management of people with FH will reduce long-term morbidity and early mortality from heart disease. While the health service faces cost pressures, the reduction in morbidity (and long-term disablement) has important implications for cost-savings to the health service over time. Indeed, a recent study by NICE found that PCT implementation of their guidelines can save enormous costs, with proactive health management programmes reducing tertiary care and other expenditure in the long run.[4] In addition, people with FH whose condition is being well managed have the capacity to work, which provides the state with tax revenue.

Without a proper programme of cascade screening we cannot capture the generations of people *within* each family affected by FH. If the attitudes of English healthcare commissioners do not change, we risk the scandal whereby the genetic misfortune of about 100,000 people with undiagnosed FH will be further compounded by where they happen to live.

Resources for PCTs

HEART UK's FH Toolkit is intended to assist with the development of local FH services. Available from: http://www.heartuk.org.uk/FHToolkit/

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Conflict of interest

None declared.

Patient booklet

For patients that you suspect may have FH or have been diagnosed with it, a new booklet entitled *Familial hypercholesterolaemia* is an essential read. Written by HEART UK and published with the support of the British Heart Foundation (BHF), it provides a step by step explanation of what FH is, how it is diagnosed, treatment and tips for everyday living. This fantastic resource is part of a range of publications about inherited cardiac conditions which have been produced by the BHF in collaboration with other heart charities.

The FH booklet can be downloaded from www.heartuk.org.uk/index.php?/been_diagnosed. Single copies can be ordered from HEART UK by emailing ask@heartuk.org.uk or calling 01628 777046. Multiple copies – GPs can order up to 30 – must be ordered from the British Heart Foundation via email orderline@bhf.org.uk or by calling 0870 600 6566 (order code M111F).

References

1. Noted from HEART UK specialist lipid clinic database, June 2010.

2. DeMott K, Nherera L, Shaw EJ *et al*. *Clinical guidelines and evidence review for familial hypercholesterolaemia: the identification and management of adults and children with familial hypercholesterolaemia.* London: National Collaborating Centre for Primary Care and Royal College of General Practitioners, 2008.

3. NHS Primary Care Commissioning. *Primary care service framework: familial hypercholesterolaemia*. Available from: http://www.pcc.nhs.uk/uploads/commissioning/2010/02/primary_care_service_framework_familial_hypercholesterolaemia.pdf

4. National Institute for Health and Clinical Excellence. How NICE could help the NHS save millions. Available from: http://www.nice.org.uk/newsroom/features/HowNICECouldSaveTheNHSOver600million.jsp

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