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Grŵp Cydlynu Rhwydwaith y Galon
Cardiac Networks Co-ordinating Group

National Clinical Audit of the Management of Familial Hypercholesterolaemia 2009: Pilot

EXECUTIVE SUMMARY June 2009

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The Clinical Effectiveness and Evaluation Unit,
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Advised and approved by:

The Familial Hypercholesterolaemia Audit Steering Group

This executive summary forms part of report:

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The Familial Hypercholesterolaemia Audit Steering Group

The following individuals are members of the Steering Group. Those marked with '✦' also form the Working Group.

Professor Steve Humphries (Project Director) ✦	Professor of Cardiovascular Genetics at University College London and FH Clinical Lead.
Dr Philip Adams	Consultant Cardiologist at Royal Victoria Infirmary Newcastle upon Tyne Hospitals.
Rhona Buckingham ✦	Clinical Effectiveness and Evaluation Unit Manager, RCP
Professor Roger Boyle	National Director for Heart Disease & Stroke / Vascular Programme at Department of Health
Dr Cyril Chapman	Consultant in Clinical Genetics at Birmingham Women's Hospital
Dawn Davies	Patient Representative on Nice Guideline and representative for Heart UK
Bridget Galaska*	Lipid Specialist Nurse at Hope Hospital, Salford Royal NHS Foundation Trust*
Dr Gaye Hadfield	Project Co-ordinator for the Department of Health FH Cascade Testing Audit Project, based at University College London
Dr Chris Hendricks ✦	Representative from the Royal College of Paediatrics and Child Health. Consultant in Clinical Inherited Metabolic Disorders, Birmingham Children's Hospital
Leonard Levy**	Vascular Programme, Department of Health
Professor Andrew Neil	Professor of Clinical Epidemiology, University of Oxford and Honorary Consultant Physician at the Churchill Hospital, Oxford
Tessa Ing***	Vascular Programme at Department of Health
Dr Ian McDowell	Cardiac Networks Coordinating Group, Wales
Claire Neuwirth ✦	Lipid Specialist Nurse at MRC Clinical Sciences Centre Imperial College
Judy O'Sullivan	Operations Manager Heart Helpline at British Heart Foundation
Diana Paine / Andrew Earnshaw	Team Leader, NHS Genetics Team at Department of Health
Dr Jonathan Potter ✦	Director of the Clinical Effectiveness and Evaluation Unit, RCP
Dr Mary Seed ✦	Honorary Consultant Physician at Charing Cross Hospital, Imperial College Healthcare and recently member of the GDG for NICE FH guidelines
Dr Tim Wang	Consultant in Clinical Biochemistry at Royal Surrey County Hospital, Guildford and Frimley Park Hospital NHS Foundation Trust
Katharine Young ✦	FH Project Manager, RCP

* Resigned after 2nd Steering Group due to change of role

** Attended 3rd Steering Group meeting representing Professor Roger Boyle

*** Attended 1st Steering Group meeting representing Professor Roger Boyle

Foreward

By the National Director for Heart Disease and Stroke

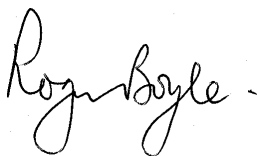
The best way to limit the damage caused by coronary heart disease is to identify those at risk as early as possible. The greatest opportunity for such disease prevention lies in diagnosing and treating people with familial hypercholesterolaemia and screening their families. Currently there are some 100,000 undiagnosed people in the UK at risk of heart disease due to familial hypercholesterolaemia.

NICE has published guidelines setting out how people with familial hypercholesterolaemia should be diagnosed and treated, and very importantly, how their families should be screened.

The pilot project reported here evaluates services against the NICE guidelines. The results do provide extremely helpful information, although care must be taken in interpretation as it is a pilot study. The current treatment of people identified with hypercholesterolaemia is good. However there are clearly apparent inadequacies in the screening programme, both in terms of cascade testing and in the use of DNA testing.

While it is reassuring that those with hypercholesterolaemia are being well managed [by the sites entered into the pilot] the results do indicate a great missed opportunity, if cascade testing is not being effectively implemented.

I am extremely grateful to all who have contributed to the work of the pilot. Not only does it demonstrate how a full national audit can most effectively be carried out but it also gives us initial indications of the ways in which services need to be improved to help reduce the burden of coronary heart disease.

A handwritten signature in black ink that reads "Roger Boyle". The signature is written in a cursive style with a small dash at the end.

Professor Roger Boyle CBE
National Director for Heart Disease and Stroke

June 2009

Executive Summary

Following the publication of the NICE Guideline for familial hypercholesterolaemia (FH)¹, we report here the findings of the pilot clinical audit to investigate the care received by individual patients who have FH.

Why do this audit?

FH is one of the most common monogenic inherited conditions in clinical practice. The prevalence of FH is about 1 in 500 (very similar to type 1 diabetes). FH patients have an increased risk of premature coronary heart disease (CHD). Approximately 50% of men, and 30% of women with FH, if untreated, will have developed clinically evident coronary heart disease by the age of 55 years².

Effective treatment is available to prevent early onset heart disease for individuals with FH. This comprises treatment with a statin to reduce their LDL-cholesterol combined with life style changes, particularly smoking cessation. This clinical approach results in a very significant reduction in their CHD mortality risk, such that well-treated patients with FH can achieve a normal life expectancy³. In the UK over 85% of the estimated 120,000 people who are thought to be affected remain undiagnosed⁴. National audit based on agreed standards and evidence based guidelines is expected to improve clinical practice, and thereby significantly reduce the mortality and morbidity associated with FH.

NICE have estimated the annual cost impact of fully implementing the guideline in England to be £7.9 million in the first year of implementation, which equates to additional costs of £16,000 for a population of 100,000⁵. Within three years considerable savings are estimated because of coronary events avoided and these will increase long term. Ongoing treatment costs would be expected to reduce progressively as higher intensity statins (often indicated for FH) come off patent.

How was the audit carried out?

Audit standards and indicators were developed from the NICE Clinical Guideline for the Identification and Management of Familial Hypercholesterolaemia (2008)⁶.

The web-based tool developed to capture the audit data worked well. Data were supplied for 248 patients, and data were duplicated for 26 of these as part of an assessment of data reliability (see Appendix 2 for details). There were very few missing or contradictory data, and the validation checks and balances in place on the webtool worked well. Some modifications have been suggested by the audit sites and the steering group to streamline and improve the tool, which will allow focus on the most important questions. Feedback suggested that, overall, sites found the audit a positive experience, and that it demonstrated their current progress in implementing the NICE guideline and identifying areas for improvements. Sites stated they would encourage others to take part in future national audits.

¹ ⁶ National Institute for Health and Clinical Excellence (NICE) - CG71: Clinical Guideline for the Management of Familial Hypercholesterolaemia (2008) <http://www.nice.org.uk/nicemedia/pdf/CG071>

² Slack J. Risks of ischaemic heart-disease in familial hyperlipoproteinaemia stats. Lancet 1969;2:1380-2

³ Neil A, Cooper J, Betteridge J, Capps N, McDowell I, Durrington P, Seed M, Humphries, SE on behalf of the Simon Broome Familial Hyperlipidaemia Register Group. Reductions in all-cause, cancer, and coronary mortality in statin-treated patients with heterozygous familial hypercholesterolaemia: a prospective registry study. European Heart Journal (2008) 29, 2625-2633.

⁴ Marks D, Thorogood M, Farrer JM, Humphries SE. Census of clinics providing specialist lipid services in the United Kingdom. J Public Health (Oxf) 2004;26:353-4

⁵ National Institute for Health and Clinical Excellence (NICE) National costing report: Familial hypercholesterolaemia Implementing NICE guidance (August 2008) <http://www.nice.org.uk/nicemedia/pdf/CG071>

What did the audit find?

The Key Findings are:

- For individual patients who have been diagnosed with FH, the clinical management in lipid clinics is of a good standard.
- Organisational issues for the care pathway of FH patients are still being developed, but it appears that centres will need additional resources to cope with the identification of the predicted additional 100,000 FH cases UK wide. This includes access to trained staff (nurses), IT needs and pedigree drawing.
- There is a major lack of systematic family “cascade” testing, whether carried out on the basis of lipid levels, or more effectively by a DNA diagnosis.
- There is limited access to DNA diagnosis and that which is available is being carried out in a research environment. Access to DNA services needs to be more widely available across the UK.
- There is a shortfall in child-focused services throughout the country, so that the ability to diagnose and treat FH in children and young persons in the health service is limited. Where such services were audited they are of a good standard.

As it is only based on 14 sites we recognise that the data are limited, and services for FH patients may be less favourable in a UK-wide audit. However, there are several key areas of clinical practice which appear to be sub-optimal in many sites, and Trusts providing services for FH patients could already consider ways to improve these deficits by developing the necessary structures and funding streams that could improve the identification of people with FH and thereby enable treatment to prevent premature heart disease.

KEY RECOMMENDATIONS

Acute trusts (England) / Integrated Trusts (Wales)

- Care pathways for FH patients need to be implemented. This must include shared care arrangements between hospital and primary care and better links between with several other specialities, including paediatrics.
- Additional resources (clinic sessions) will be needed to cope with the identification of the predicted additional 100,000 FH cases UK wide. At present there is a shortage of both specialists and lipid clinic nurses.
- Systems need to be developed and implemented to carry out systematic family “cascade” testing. This will require trained nursing/genetic services to follow up the families of index patients, improved IT needs, including an FH patient database, and pedigree drawing.
- Resources are needed for DNA diagnosis and Clinical Genetics input.

National Organisations:

- A system for coordination of cascade testing systems on a national basis is recommended, with links to genetic testing services, given that FH families are geographically dispersed.
- The lack of paediatric services may be best coordinated at a national level to ensure that appropriate child focused services are developed.
- On the basis of this pilot study it is recommended that a national audit of services is very feasible and should be commissioned