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Knowledge gaps in the management of familial hypercholesterolaemia. A UK based survey

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ABSTRACT

Background and aims: Untreated individuals with familial hypercholesterolaemia (FH) are at increased risk of developing premature cardiovascular disease (CVD). Early diagnosis and treatment can result in a normal life expectancy. A recent survey commissioned by the European Atherosclerosis Society (EAS) reported a lack of awareness of FH in the general population. We conducted a survey to assess knowledge among healthcare professionals involved in the assessment and management of cardiovascular risk and disease in the United Kingdom.

Methods: A survey designed to assess knowledge of diagnostic criteria, risk assessment, the role of cascade screening, and management options for patients with FH was distributed to 1000 healthcare professionals (response rate 44.3%). The same survey was redistributed following attendance at an educational session on FH.

Results: 151 respondents (40.5%) reported having patients under their care who would meet the diagnostic criteria for FH, but just 61.4% recognized that cardiovascular risk estimation tools cannot be applied in FH, and only 22.3% understood the relative risk of premature CVD compared to the general population. Similarly, just 65.9% were aware of recommendations regarding cascade screening.

Conclusions: The prevalence and associated risk of FH continue to be underestimated, and knowledge of diagnostic criteria and treatment options is suboptimal. These results support the recent Consensus Statement of the EAS and production of quality standards by the National Institute for Health and Care Excellence. Further work is required to formulate interventions to improve FH awareness and knowledge, and to determine the effect these interventions have on patient outcomes.

1. Introduction

The life-threatening effects of familial hypercholesterolaemia (FH) are the result of abnormally high circulating concentrations of atherogenic low-density lipoprotein cholesterol (LDL-C), present from birth due to an inherited defect in low-density lipoprotein clearance [1]. FH is under-diagnosed and often diagnosed late [2]. Lipid-modifying drug treatment reduces LDL-C levels and attenuates the development of cardiovascular disease (CVD) but fewer than one in six patients may be treated, with many of these being undertreated [3,4]. Importantly, the overall mortality in treated Heterozygous FH is similar to that in the general population [5].

Patients with a history of premature CVD are not consistently screened for FH [6,7], and hence even Coronary Care Unit admissions thought to relate to FH may be overlooked [1]. Individuals with CVD secondary to FH may also be missed in primary care where other risk factors for CVD are more common [1,8]. Whilst the U.S. National Lipid Association recently highlighted the limited training available to practitioners to facilitate the screening and appropriate management of patients with FH [9], there has been little discussion of this in Europe beyond a recent survey commissioned by the European Atherosclerosis Society (EAS) that reported

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a lack of awareness among the general population [10]. We therefore carried out a survey of healthcare professionals working in primary and secondary care in the United Kingdom (UK) to assess current knowledge of FH, and explore whether targeted education could improve this.

2. Materials and methods

A structured survey with questions based on expert recommendations and published guidelines (Fig. 1) was distributed to healthcare professionals involved in the assessment and management of cardiovascular risk and disease at local and national educational meetings, and electronically by e-mail with an embedded single-use URL. The survey was introduced to potential participants with an explanation of the aims of the project including planned dissemination of the results to meet the requirements of informed consent. Due care was taken to protect confidentiality.

Demographic data were sought for job title and region within the UK. Participants were asked about their familiarity with FH, knowledge of its prevalence, inheritance, associated risks, awareness of diagnostic criteria and guidelines for management. Questions were also included on current and future treatment options. Participants were asked to choose the most correct statement, or to select one or more answers from a list; the only open questions related to previous experience of FH and suggestions to improve the care of patients with FH.

A second survey was distributed to healthcare professionals following attendance at an educational session on FH. Participants were asked to answer the same questions four weeks after this session.

Data were collected and analyses performed using SurveyMonkey and Microsoft Excel. Statistical tests (student's t-test) to determine occupational and geographical differences in FH knowledge, practices and opinions used position and previous experience as categorical variables. Paired t-tests were used to determine the effect of education. Knowledge scores were calculated from answers submitted for questions on the pathogenesis of FH, and its management.

3. Results

The paper questionnaire distributed at educational meetings was completed by 81 of 100 healthcare professionals (81%). The response rate to electronic distribution of the questionnaire was 40.2% (362 of 900) giving a cumulative response rate of 44.3% (443 of 1000). 35 of 50 (70%) completed a second survey assessing the effect of education (31 of 40 paper questionnaires, 4 of 10 electronic invitations).

Of 443 participants the majority (94.5%) were based in England, with 13 respondents (3%) from Scotland, 9 from Wales (2%), and 2 working in Northern Ireland (0.5%). The professional roles of respondents are shown in Table 1.

80 respondents (20.8%) were currently seeing patients in a specialty lipid clinic while a further 36 (9.4%) had done previously but not at the time of survey completion. 151 participants (40.5%) reported having patients under their care who would meet the diagnostic criteria for FH. Participants currently seeing patients in a specialty lipid clinic scored significantly better on the knowledge-based elements of the survey compared with those who had seen patients previously and those who had never seen patients in a specialty lipid clinic (82.5% *vs.* 62.3% *vs.* 46.1% ($p < 0.001$ *and*

1	The following statements apply to Familial Hypercholesterolaemia (FH): ***True** / False / Don't Know*	• Largely due to acquired nutritional & lifestyle factors; genetic susceptibility involves more than one gene • **LDL-cholesterol is raised from birth** • Use of lipid lowering therapy should be guided by cardiovascular risk estimation using a Framingham or QRISK based calculator • Statin therapy is considered only if estimated 10-year cardiovascular risk >15% • **If untreated 1 in 2 men and 1 in 3 women will suffer a myocardial infarction before age 60** • A diagnosis of definite FH can only be made via genetic testing	
2	Which of the following best describes Heterozygous FH?	• The presence of family members with known hypercholesterolaemia • **A monogenic disorder characterized by hypercholesterolaemia and a family history of premature coronary heart disease** • The presence of multiple lipid abnormalities that may be genetic in nature • An extremely rare, potentially fatal condition caused by cholesterol levels up to 6 times normal • Don't know	
3	Total cholesterol above what level should make the clinician think about FH?	• **7.5 mmol/l** • 10 mmol/l • 12.5 mmol/l	• 15 mmol/l • Don't know
4	Are you aware of any of the following criteria for the diagnosis of FH?	• Simon Broome diagnostic criteria • Dutch diagnostic criteria • NICE guidelines on FH	• MEDPED Criteria (USA) • Local Guidelines
5	After diagnosing an index case, family cascade screening is:	• Indicated only if the index case suffers from premature cardiovascular disease (CVD) • **Recommended by NICE** • Recommended only if there is a family history of premature CVD from both parents • Recommended only if there is tendon xanthoma and / or genetic mutation in the index case • Don't know	
6	The prevalence of FH in the UK is thought to be:	• 1 in 100 • **1 in 500** • 1 in 5,000	• 1 in 100,000 • Don't know
7	First degree relatives of someone with FH have what chance of having FH themselves?	• 10% • 25% • **50%**	• 100% • Don't know
8	In FH, the risk of premature CVD is how many times greater than the risk in the general population?	• 5-fold • 10-fold • 15-fold	• **20-fold** • Don't know
9	Which of the following would improve the care of FH patients? *Tick all that apply*	• Better access to the Lipid Clinic • More Education for General Practitioners • Patient forums	• My region manages FH well • Don't know • Other (please specify)
10	Are you aware of any of the following treatment options beyond statins? *Tick all that apply*	• Ezetimibe • Bile acid sequestrants • Fibrates • Omacor	• Extracorporeal LDL cholesterol removal therapy • PCSK9 inhibitors • None of the above
11	Extracorporeal LDL cholesterol removal therapy (LDL apheresis) ***True** / False / Don't know*	• I am aware of guidelines for its use • **There is evidence to support its use to reduce CVD mortality & morbidity** • Is only indicated for Homozygous FH • I am aware of a local centre providing this service	

Fig. 1. Questions included in questionnaire.