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ORIGINAL ARTICLE

Optimising the Detection and Management of Familial Hypercholesterolaemia: Central Role of Primary Care and its Integration with Specialist Services

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Familial hypercholesterolaemia (FH) is the most common monogenic lipid disorder associated with premature coronary heart disease (CHD). However, the majority of people with FH are undiagnosed or undertreated. Early cholesterol lowering therapy reduces cardiovascular disease mortality in FH. Low awareness and knowledge of FH in specialty and general practice highlights the need for strategies to improve the detection and management of FH.

We present an algorithm describing a multidisciplinary approach to FH detection and management. We highlight the role of primary care, and where GPs can work with preventive cardiologists to improve care of FH. Novel strategies to detect index cases with FH are presented including the community laboratory, highlighting patients at high risk of FH, and targeted FH detection through searching the general practice database.

General practitioners request over 90% of LDL cholesterol measurements in the community. Once an individual with FH is detected only a small proportion of patients require specialty management with the majority of patients suitably managed in primary care. However, it is crucial to screen family members, as 50% of first-degree family members are expected to have FH due to the autosomal dominant inheritance.

Keywords Familial hypercholesterolaemia • Cardiovascular diseases • Genetic predisposition to disease • Mass screening • Primary health care

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Introduction

Familial hypercholesterolaemia (FH) is the most common monogenic lipid disorder associated with premature coronary heart disease (CHD) [1]. FH is an autosomal co-dominant disorder associated with elevated low density lipoprotein (LDL) cholesterol [2–4]. Currently, the majority of people with FH are undiagnosed, and those who are diagnosed are often undertreated [5,6]. There are 50,000 people estimated to have FH in Australasia [7], and general practitioners (GPs) are well placed to detect people with FH [8,9].

FH occurs in approximately 1 in 500 people, thus the average GP is likely to encounter up to 25 patients with FH every year [9]. If left untreated, 50% of men with FH will have CHD by age 50 years and 30% of women by age 60 years [3]. FH is an autosomal dominant disorder, thus cascade screening of first-degree relatives (i.e. parents, siblings and children) is essential as 50% will also have the condition. Cascade screening of relatives either clinically or genetically, is the most cost effective screening method [10]. However, this requires effective and accurate mechanisms for identifying FH index cases.

Most patients with FH can be simply and effectively treated with life-long cholesterol-lowering treatment, especially with HMG co A reductase inhibitors (statins) which have been demonstrated to reduce mortality [11]. In particular, people with FH under 30 years of age are rarely identified or treated, although they are predicted to benefit most in terms of life years gained [5]. (Figure 1) Early treatment before the development of atherosclerosis is crucial. Individuals with genetically low LDL cholesterol levels have lower risk of CHD [12–14]. Mendelian randomisation studies have shown that exposure to lower LDL cholesterol in early life is associated with a substantially greater reduction in CHD [15].

General practitioners request over 90% of LDL cholesterol measurements in the community [8], and are thus critical to detecting FH index cases [1]. Current models of care have not focussed on the role of primary care [1,5,16]. FH detection in the community may be optimised with a multidisciplinary approach including: a broader primary care awareness and education programs, auditing of electronic patient information on general practice databases, and via pathology providers highlighting people with very elevated LDL cholesterol at risk of FH. We have previously reviewed the role of the GP in screening for FH [9] and a key function for cardiologists in detecting FH amongst patients and their families with established CHD [17]. We now provide further recommendations to improve the detection and management of these patients using an uncomplicated algorithm that allows for the integration of care of between GPs and specialist services, particularly cardiology.

Role of Cardiologist and Awareness of FH

Cardiologists can play a significant role in the care of FH across the continuum of health care from prevention to management of acute coronary syndrome [17]. As many index cases present for the first time with acute coronary syndrome, cardiologists may be one of the first clinicians to encounter patients with FH and their families. 10-15% of patients with premature CHD will have FH although nearly one third did not have cholesterol measured during their coronary care admission [18,19]. Coronary care and rehabilitation units provide an ideal setting to identify FH and to lead the secondary prevention of CHD in FH. However, cardiologists need to lead by example to educate their primary care colleagues that FH is a preventable cause of CHD, that it is an inherited disorder and refer index cases for cascade screening of family members. In spite of this potentially important role, awareness and knowledge of FH amongst cardiologists, as with GPs, remains sub-optimal [18,20].

The role of cardiologists in leading the change of culture towards improved multidisciplinary awareness of the importance of primary and secondary prevention of CHD,

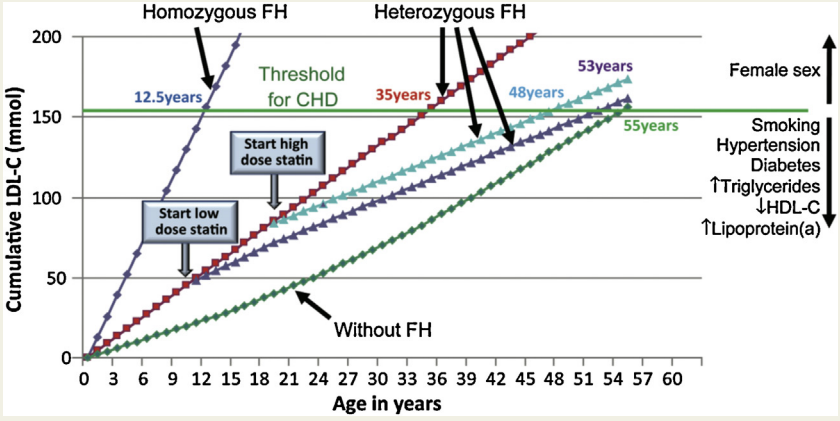

Figure 1 LDL cholesterol burden in individuals with and without FH as a function of the age of initiation of statin therapy.