Challenges in the care of familial hypercholesterolemia: a community care perspective

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**Title:** Challenges in the care of familial hypercholesterolemia: a community care perspective

**Summary:** Familial hypercholesterolaemia (FH) remains under-diagnosed and under-treated in the community setting. Earlier evidence suggested prevalence of 1:500 worldwide but newer evidence suggests it is more common. Less than 15% of FH patients are ever diagnosed with children and young adults rarely tested despite having most to gain given their lifetime exposure.

Increasing awareness among primary care teams is critical to improve detection profile for FH. Cascade testing in the community setting needs a sustainable approach to be developed to facilitate family tracing of index cases. The use of the Dutch Lipid Clinic Network Criteria score to facilitate a phenotypic diagnosis is the preferred approach adopted in Australia and eliminates the need to undertake genetic testing for all suspected FH cases.

**Keywords:** Familial hypercholesterolaemia, Cardiovascular disease, Prevalence, Statins, Primary - Tertiary care management, Index case, Cascade testing, Dutch Lipid Clinic Network Criteria, Phenotypic testing

**Expert Commentary Body of Article**

**Public health burden**

Awareness that inherited factors can play a significant part in the development of atherosclerotic cardiovascular disease (CVD) is generally not well appreciated amongst the general population in Australia. Whilst it is often well known among family members and other more distant relatives that premature deaths attributed to heart disease have occurred in their families, the significance of such events in relation to their personal risk is often not fully understood and easily forgotten. It is against such a background that attempts to achieve a higher profile for CVD risk assessment have often met with very limited success.

Familial hypercholesterolaemia (FH) is an autosomal dominant inherited condition [1] that causes significantly raised total and low-density lipoprotein cholesterol (LDL-c) levels in affected individuals from birth. Individuals with heterozygous FH (HeFH) have a 50% chance
of passing the condition to their offspring. The lifelong exposure to high circulating lipids significantly increases their chances of developing premature CVD and death.[2] For males with HeFH, the risk of developing coronary heart disease before age 50 years is 50% while for females it is 30% before age 60 years.[3, 4] The CVD risk for individuals with homozygous FH (HoFH) is much greater. Left untreated these patients may manifest CVD events and other vascular complications in childhood or adolescence.[5]

The marked acceleration of coronary artery disease (CAD) seen with FH means that the young have much to gain from early diagnosis and treatment. Effective and early treatment of FH with statin therapy reduces the lifetime exposure to very high cholesterol levels with the concomitant reduction in CVD events largely offsetting the cost of prolonged treatment.[6, 7] The key therefore is to detect and treat FH as early as possible.

The prevalence for FH was generally reported as 1:500 [1, 3, 8] but more recent European [9, 10] and Australian [11] research suggests it is more common in the general population. FH is also especially frequent among communities exhibiting founder gene effects.[3] [1] In countries such as Australia with historically high immigration rates the possibility of increased FH presence amongst patients from such communities should always be considered. The prevalence of FH amongst indigenous Australians is unknown.

The majority of FH cases are caused by mutations in the gene encoding the LDL-receptor (LDLR) while less common mutations in the apolipoprotein-B (apo-B) and Proprotein convertase subtilisin/kexin type 9 (PCSK9) genes cause similar functional consequences.[2] [12] There are over 1200 mutations known to cause FH,[13] and testing for all mutations is not practical. Currently a mutation is only detected in 60-90% of individuals with clinically definite FH.[14] Thus, failure to detect a mutation does not exclude a diagnosis of FH. This makes genetic testing for population screening purposes less feasible, expensive and controversial, both for patients and the health systems involved.

Low detection rates in children and young adults
Despite this potential for early intervention, less than 15% of patients with FH are ever diagnosed \[8\] with children, teenagers and those aged 20 - 40 years amongst the least likely to be offered opportunistic lipid testing during routine primary care visits. This occurs principally because most of these patients are not recognised as potentially at risk, and do not routinely have serum cholesterol estimations undertaken – at least not until much later in middle adult life.

Similarly, because of poor awareness of FH,\[15\] children of FH patients are often not recognised as being at risk, or in need of early diagnosis and management to prevent development of CAD. Their plight is often compounded by reluctance on the part of the primary care physician to institute appropriate treatment at an early age (from age 10 years in boys and one year post menarche in girls is recommended \[16\]). This is sometimes exacerbated by a certain ‘statin stigma’ and attacks on the efficacy of such medication in the popular media.\[17, 18\] This situation is unlikely to change unless the profile of the condition receives much greater prominence and the benefits and cost effectiveness of early intervention are brought to the attention of patients, General Practitioners (GPs), Practice Nurses (PNs) and primary care teams.

**Absolute and relative CVD risk in FH patients**

In assessing CVD risk, it is important for clinicians to be aware of the potential presence of FH in patients whose lipid results show very elevated LDL-c levels. Absolute CVD risk calculators are not advised in FH patients as they underestimate risk for this condition that has been present from birth. The relative risk posed by markedly raised LDL-c levels in patients with FH is so great that the over-riding significance of this single risk factor needs to be acknowledged and such patients should be treated with lifelong, maximum tolerated statins with or without ezetimibe. An LDL-c level above 5.0mmol/L should raise a high degree of suspicion for the presence of FH and clinicians should flag such patients for careful assessment and management. Patients with personal or family history of CVD and untreated LDL-c level above 4mmol/l should also be considered for assessment of FH risk.
It is estimated that the ‘cholesterol-life years burden’ accumulated by a patient with FH at age 45 years is roughly equivalent to that of a similar patient without FH by age 70 years.[19, 20] Effective treatment is available with statin therapy and such treatment is most beneficial and cost effective for younger patients as they have not yet developed severe atherosclerosis due to the lower exposure to elevated LDL-c, and thus severe atherosclerosis may be prevented.

*Unique features of primary care populations*

In Australia, over 81% of the population consult a GP annually [21] with GPs requesting 91.8% of all LDL-c tests undertaken through a community based pathology laboratory.[22] Research has shown that it may be possible to use a serum LDL-c cut-off point alone to facilitate detection of FH [23, 24] with GP consultations potentially offering unique opportunity to help detect unknown index cases of FH in the community.[25]

Current Australian State and Federal Government policy aims to increase primary care management of most chronic conditions. The ensuing increase in time and resource implications means that a primary care based model of care (MoC) for FH needs to be simple, efficient and sustainable.

Data from the [Bettering the Evaluation and Care of Health (BEACH)](https://www.beach.org.au/) studies show that only 5.3% of patients seen by GPs are referred for specialist management.[26] Whilst GPs refer selected populations to their specialist colleagues, they themselves constantly sift diseases that often present in very early stages and are exposed to major serious illnesses infrequently.[27] This information is critical to appreciate the different demographics of the unselected, undifferentiated populations and presentations seen in primary care compared to the highly selective, well differentiated populations referred to specialists for tertiary level care. The primary care physician requires considerable adaptability and flexibility to meet the health needs of individual patients often with multiple morbidities [28-30] and contrasts with
the specialist tertiary clinic that deals with the health needs of a group of patients all with the same range of diseases or illnesses.

In essence, much of the work of primary care physicians is grounded in knowledge that incorporates biomedical, social, psychological, interpersonal and intrapersonal perspectives.[27] For GPs, the consultation is much more patient-centred rather than disease focussed and offers a comprehensive, holistic approach to care that is not just community based but also offers an opportunity to incorporate unique sociological as well as environmental aspects to patient care. Their knowledge of patient and family history can often transcend generations as well as care for extended families.

However, taking a family history and recording it in patient records is usually poorly documented in primary care with the average time taken for family history discussion being less than three minutes.[31, 32] Better systems are needed to assist primary care clinicians to include comprehensive family history records and to ensure such histories are regularly updated.

Against such a background, the involvement of primary care in the early recognition and management of FH offers both challenges and opportunities. High on the list of opportunities must be the ability to help detect the condition in younger patients who have inherited FH but have never been screened, never formally diagnosed or offered appropriate treatment.

**Opportunities in primary care**

Primary care is recognised as being ideally placed to provide a more important role in both opportunistic and systematic case finding especially in recognising the association between premature coronary heart disease (CHD) and markedly elevated serum cholesterol concentrations.[8, 15, 33] An analysis of the cost effectiveness of different approaches to screening for FH found that targeting family members was the most successful option and far superior to simply targeting patients admitted to hospital with premature myocardial infarction.[34] When this approach is combined with institution of treatment with lipid-
lowering agents and subsequent targeted referral of high risk cases for specialist lipid management, the outcome is to substantially reduce morbidity and mortality from CHD among younger FH patients.[35]

The presence of other cardiovascular risk factors is recognised in individuals with FH, although these conditions are also normally managed in the primary care setting, such as smoking, obesity, excess alcohol intake, lack of exercise, hypertension and diabetes.[36] The detection of FH provides added incentive for such patients to optimise treatments for these risk factors as well as their FH. Primary prevention of CVD forms part of the everyday workload of general practice.[36]

Primary care, in combination with dedicated specialist lipid clinics, has the potential to offer a more cost effective approach that may prove more acceptable to patients and practice staff as well as instituting a better disease prevention strategy for affected families. Further research is needed to evaluate innovative programs that seek to undertake detection and management of index cases at the primary care level as well as providing an approach to facilitate cascade testing for newly diagnosed index cases with support from specialist lipid clinics for more difficult and hard to manage cases.

Innovative approaches from primary care

The importance of developing an acceptable approach to increase detection and diagnosis of FH index cases at the primary care level is only partly recognised.[37] An innovative approach to opportunistic screening for FH utilising the community laboratory has been investigated. GPs request over 90% of the LDL-c levels from a community laboratory.[22] Interpretative comments highlighting the possibility of FH were associated with a significant additional reduction in LDL-c and increased specialist referrals if such an approach was included in the interpretative comment.[23] A phone call between the chemical pathologist and requesting primary care physician led to increased specialist assessments, which resulted in over 70% of individuals diagnosed with clinical FH.[24] A combined opportunistic
and retrospective approach using the primary care database needs to be evaluated to ascertain acceptance for patients, GPs, PNs, Nurse practitioners (NPs) and practice reception staff.

Potential secondary causes of raised lipids including liver and renal disease, hypothyroidism and steroid usage need to be considered and excluded before making a phenotypic diagnosis of FH. Central to improving FH care is early identification of the index case or first family individual with the diagnosed condition. Cascade testing (family tracing) of index case subsequently identifies relatives with FH. A subsequent shared care approach involving lipid specialists and GPs can help facilitate cascade testing of index cases to help identify relatives with possible FH.

The current MoC for FH in Australia is designed primarily for lipid clinics in tertiary centres [16] but the need to develop a MoC for general practice is recognised.[37] Currently, FH is diagnosed through a number of different routes [38] and managed mainly through hospital-based lipid clinics that may undertake genetic testing particularly if the clinical features are highly suggestive of FH. Diagnosis of FH has been shown to prompt action by patients [38] and the active involvement of primary care teams in FH identification and optimum management is increasingly recognised as the best and most sustainable approach for the future.[7]

There are several tools used worldwide for the phenotypic diagnosis of FH, clinically with Due to its ease of clinical use, greater sensitivity and comprehensiveness the Dutch Lipid Clinic Network Criteria (DLCNC) score (Figure 1) is the preferred tool of choice in Australia advocated by the Familial Hypercholesterolaemia Australasia Network Consensus Group.[16] The DLCNC score encompasses personal LDL-c levels; personal and family history of premature coronary artery and/or vascular disease and physical examination to detect tendon xanthomata or premature arcus cornealis.
The Consensus Statement of the European Atherosclerosis Society [14] and the International FH Foundation [39] recommend that most individuals with FH should be treated in primary care and preferably in a family context. They advise that more complex cases and children with FH should be referred to specialist lipid or FH clinics. The Netherlands (71%) and Norway (43%) are leaders in Europe for estimated per cent of individuals known to harbour a diagnosis of FH.[14] In the UK, the detection and management of FH has improved after the introduction of the NHS Clinical Services initiative.[8] A network of regionally located specialist lipid clinics can be accessed by GPs to support primary care management.[40] A case ascertainment tool FAMCAT has also been recently produced.[41]

Cascade testing

In the Australian setting, an approach through tertiary level specialist centres undertakes cascade testing of relatives of index cases. This involves measurement of LDL-c levels, DNA testing and statin treatment and has been shown to be cost-effective in preventing CHD.[42] However, not all patients at increased risk of FH, including those highlighted in primary care setting, end up being referred for specialist assessment.[24] This is likely secondary to the low awareness of FH among many health care providers.[15, 43] Costs for the application of universal genetic testing to cascade screen relatives of index cases detected in primary care could be prohibitive. However, the application of DLCNC score to detect possible, probable and definite adult FH cases should be more feasible with use of LDL-c measurements more appropriate for cascade testing children and adults.[16, 44] The DLCNC score can be effectively used in primary care to segregate individuals into high and low risk for FH.[45]

The active involvement of GPs, PNs, NPs and primary care teams could potentially offer a much improved detection rate from FH in the community setting. The use of dedicated field workers to undertake contact tracing of index cases has been very successful in the Netherlands. Other factors including the impact of population migration into Australia are likely to reduce the potential to employ a similar approach in this country. In addition, higher population densities in smaller geographic areas make logistics of contact tracing easier –
Netherlands 17 million compares with Australia’s 23 million in land mass just 0.5% of the latter.

Despite these factors, primary care with its ease of access and frequent patient consultations offers potential to improve on current detection rates. Increasing awareness through health promotion campaigns, highlighting of elevated LDL-c levels on lipid testing and help with developing family pedigrees would be essential elements in any future proactive approach. A commitment on the part of the primary care team will inevitably be the key as any approach that imposes an additional time, manpower or financial burden on the practice is likely to be unsustainable in the long run. Building the infrastructure and knowledge base from within the practice offers the best long term option for success. The UK NICE guidelines [4] have recognised the difficulties of undertaking cascade screening from primary care and have suggested the use of specialist lipid clinics to undertake the work instead. The use in Australia of the DLCNC score rather than genetic testing to establish a diagnosis of FH will make the process easier and offers a better chance of success.

Obtaining consent from index cases to approach relatives about their risk of having FH is critical in the cascade testing process.[46] There is extensive evidence to show that progressing to family cascade testing would be clinically [4, 14, 16] and financially effective.[35, 42, 47] A diagram to show how to proceed with cascade testing, undertake risk notification and suggestions as to how to approach families is shown (Figure 2). If no consent is forthcoming and relatives are also patients of the practice, there may be opportunities to encourage such relatives to undertake LDL-c testing as part of their regular health checks and advise accordingly.[11, 16, 44] Similarly, other relatives who are not patients of the practice should be advised by the index case of their potential high FH risk and encouraged to have their LDL-c levels assessed at their local practice. Any breach of confidentiality should only occur in very specific circumstances such as detailed in the relevant jurisdiction. Where uncertainty exists, federal and state government legislation,
NH&MRC guidelines and local health service protocols about disclosure of medical information without consent should be followed.

It is important, where families and index cases have agreed to co-operate, that the patient’s privacy and autonomy are respected and that information concerning the nature of FH that is sent or given to a particular individual is transmitted in such a manner that it can be easily understood and not cause undue alarm.

Consultations with relatives should involve pre-test counselling to ensure such patients have a good understanding of the nature of the condition and are given an uncomplicated explanation as to why early detection for themselves and first degree relatives including children might be undertaken, as well as the rationale for treatment if they are shown to have FH. It is likely that undertaking a blood test as well as a clinical examination for specific features of FH will prove to be more acceptable to patients rather than offering genetic testing. Good communication skills and knowledge of the patient and their family background should make the process easier for primary care teams. The development of specific skills in constructing a family pedigree (Figure 3) and undertaking cascade screening by a specific doctor and nurse at a particular practice would be especially valuable for the longer term sustainability of the process.

Management

Primary care offers a unique opportunity through ease of access and frequent patient contact to use a strategy of combining effective patient counselling on the chronic nature of FH and the importance of the need for lifelong lipid lowering therapy.

**LDL-c targets:** A significant reduction in LDL-c levels is essential to minimise the risk of future CVD complications. A reduction of 50% in levels present at diagnosis has been suggested with further advice to achieve levels under 3mmol/l and under 2mmol/l respectively for those assessed as being in need of more aggressive or intensive management. Baseline creatine kinase, liver and kidney function tests can be useful when
undertaken prior to commencement of statin treatment in these patients as it allows potential to compare future levels if patient complains of muscle aches or other symptoms subsequent to commencement of statin therapy. In instances where it is difficult to reach recommended LDL-c target levels and for higher risk patients newer therapies (such as PCSK9 monoclonal antibodies) may be indicated.[48, 49]

Compliance: Regular patient contact helps to ensure good compliance with medications as well as with diet, exercise and other lifestyle modifications. Such a holistic approach is essential to ensure lifestyle advice is adhered to and other non-lipid cardiovascular risk factors are also addressed. High on the list of priorities would be to target smoking cessation. FH patients should be left in no doubt that continuation of smoking by themselves or by other family members in their immediate household should not be tolerated under any circumstances.

Rural setting

Rural GPs and their patients can be considerably disadvantaged by geographical distance from specialist lipid clinics. In these circumstances, both patients and doctors have to rely on increased use of electronic communications in order to bridge this gap. The use of the DLCNCC score to achieve a phenotypic diagnosis can be especially useful in these circumstances as it facilitates local assessment and risk stratification without having to resort to genetic testing at a specialist centre. It is advisable that expert advice should always be sought for patients requiring enhanced or intensive management. (Figure 4). A positive aspect is the greater local community knowledge of index cases and their extended families by GPs which can help facilitate cascade testing of relatives. In addition, the construction of family pedigrees (Figure 3) which can be undertaken as part of the contact tracing process for newly diagnosed index cases, can help considerably with achieving greater community awareness of FH risk.

Pregnancy
It is essential that women of childbearing age who have FH are adequately counselled about the risks of statin use in pregnancy.\cite{11, 16} All statins and most other lipid modifying drugs need to be discontinued for a minimum of three months prior to conception as well as during pregnancy and breastfeeding. For FH patients with increased risk of CVD the advice of a specialist should be sought to enable a more comprehensive assessment to be undertaken while access to lipid specialist support is advised for the duration of a pregnancy.\cite{16, 39, 50, 51}

*Children and adolescents*

In children and adolescents, a key focus should be on early dietary and lifestyle modifications with avoidance of smoking a key area to be targeted. The use of risk stratification can help with approaches to treatment but the importance and need for lifetime treatment for this cohort needs to be stressed. This message is likely to need ongoing reinforcement as the exuberance and indestructability of youth can make compliance with strict treatment regimens especially difficult.

*Drug treatment*

Simvastatin, pravastatin, \textit{rosvuastatin} and fluvastatin are licensed by the Therapeutics Goods Authority for use in children in Australia. Primary care physicians may wish to seek the advice and support of lipid specialists and/or paediatric cardiologists if they have concerns about initiating statin treatment or want additional guidance for this age group and their families. Baseline measurements of liver and kidney function as well as creatine kinase are advisable for FH children and adolescents. In addition, other developmental aspects including growth patterns, body weight as well as sexual and physical maturation should also be monitored on a regular basis.

*Family clusters*

The probability exists that other siblings and first degree relatives will also have FH and in such circumstances it may be possible to arrange for management and follow-up of such
family clusters to be undertaken at a specialised FH clinic either in primary care setting or with support from tertiary level lipid clinic. For those patients requiring more intensive management – especially those who are homozygous or compound heterozygous as well as those refractory or intolerant of cholesterol lowering medications – the option of LDL-apheresis needs to be considered and advice sought from lipid specialist with experience in the area.

**Conclusions**

There is increasing awareness worldwide that despite various strategies to increase detection of FH, most people with the disorder remain undetected and untreated. It is equally well known that for affected patients to get the best possible outcome for the condition it is imperative that the diagnosis of FH is made early, preferably in childhood, adolescence [5] or young adulthood. Improved education, especially for younger FH patients, greater community awareness of the disorder including that of specialists, GPs, PNs, NPs and primary care teams, together with an ongoing chronic disease management plan with regular review visits, are seen as essential to achieve adherence to life-long treatment.

Newer evidence suggests that the prevalence in Australia is about 1:200-300 [9-11] yielding a total population of 80,000 nationwide with the overwhelming majority remaining unrecognised. It is hoped that improved community screening programmes, involving targeted, universal and cascade testing and better integration of specialist and primary care, will play a major role in redressing this shortfall.

The diagnosis of FH is cost-effective and presents the greatest opportunity to reduce the overall CVD burden. GPs and primary care teams need to be alert and recognise that raised LDL-c together with personal or family history of premature vascular disease may indicate FH. The use of absolute cardiovascular risk calculations are not appropriate for this population group as they are already at high risk of CVD.
The DLCNC score is an effective tool to achieve a phenotypic diagnosis of FH in primary care setting in Australia, but secondary causes of hypercholesterolaemia need to be excluded. Once an index case is recognised in primary care, cascade testing of relatives should be undertaken and assistance sought from specialist lipid clinics as necessary.

**Expert Commentary**

Familial hypercholesterolaemia has a poor public awareness profile in Australia among both patients and medical practitioners. In comparison with the Netherlands where 71% of affected patients are diagnosed, only 1% of Australian FH patients are currently detected. Based on recent evidence from Denmark, [9] it is likely that up to half of the patients known to have FH are not being treated appropriately.

Currently, most of the diagnosis and management of FH in Australia occurs in tertiary level specialist centres. It is felt that shifting the detection and management into the primary care sector has the potential to radically alter the management of the condition while still maintaining the option of specialist involvement for more complex and hard to manage patients. In addition, the use of genetic testing for DNA mutations is costly and not all mutations can be detected. The use of the DLCNC score offers a more primary care friendly approach to achieve a phenotypic diagnosis and offers a greater likelihood for a sustainable model in primary care.

A key plank in this approach will be to increase both patient and medical practitioner awareness of the condition and thereby facilitate improved diagnoses. The use of labour saving data extraction tools on general practice databases has the potential to alert primary care teams to those patients attending the practice considered to have increased risk of FH. Such patients can then be systematically reviewed by their GPs who can use the opportunity of recall visits to update personal and family history, undertake physical examination and apply the DLCNC score to establish a phenotypic diagnosis of FH.
The subsequent MoC will involve identification of the index cases in primary care, the provision of advice on institution and continuation of appropriate dietary and lifestyle modifications, a discussion on commencement of lifelong drug treatment, the construction of family pedigree trees and the application of cascade testing of their first degree relatives.

The sustainability of the proposed MoC in the Australian primary care setting lies in the recognition of FH as a lifelong, chronic condition that is ideally suited to Care Plan development and follow-up with involvement of allied health support in ongoing patient management. The ease of access and frequent patient contact in primary care will help facilitate this approach. Specialist help should still remain available for higher complexity patients. This MoC could be adapted for use in other countries.

As part of this proposed policy shift from tertiary level care to primary care, we are undertaking research Australia wide that employs data extraction techniques to identify high risk patients, apply the DLCNC score, estimate FH complexity, institute appropriate drug and lifestyle treatment and undertake cascade testing of relatives. The hope is to provide scientific evidence that our proposed model of care works in the busy clinical practice setting and that it becomes embedded and sustainable into the future. Key outcomes for this will include increased awareness about FH among patients and primary care teams, improved confidence in detection and management, increasing numbers of index cases and relatives subsequently diagnosed and appropriate responses to treatment reflected in LDL-c levels.

**Five-year view**

The use of targeted data extraction from existing electronic medical records could help identify patients at high risk of FH in the community setting. A sustainable method tailored to the needs of primary care could help with community based detection and treatment. An integrated care program in primary care involving key GPs, PNs and NPs with specialist help as needed, would be ideal. Further assistance from lipid specialists for more difficult to
control and complex cases, pregnant women and children will still be required. High risk patients, including those intolerant or inadequately managed on standard treatment, may require newer therapies in addition to statins.

**Key issues**

- General community awareness of the potential for FH among patients, primary care physicians, cardiologists and other health professionals is low and needs to be improved if optimum management is to be achieved.

- The prevalence of FH is now recognised as about 1 in 250 compared to the 1 in 500 reported in the past.

- Using genetic testing to confirm a clinical suspicion of FH may not be conclusive as up to 40% of individuals tested will not have a positive result for a mutation.

- The Dutch Lipid Clinic Network Criteria score offers another approach that achieves a phenotypic diagnosis without having to undertake expensive genetic testing for all patients.

- Children, adolescents and young adults have most to gain from the early detection and appropriate management of FH.

- Once an index case for FH has been identified in the community, a process to initiate contact tracing and cascade testing of relatives should be implemented at the primary care level.

- Undertaking cascade testing in the community setting will involve developing a sustainable approach delivered through primary care teams with specialist support and advice for patients requiring more intensive management.
References


Reference Annotations

*Of interest

3. Useful overview of the extent of problem that FH presents.

5. Good paper highlighting the benefits that can be gained from early diagnosis and treatment of FH in children and adolescents.

6. Helpful systematic review that focusses on the cost effectiveness of diagnosing and treating FH.

11. This paper shows the prevalence of FH in Australian population is considerably higher than previously reported.
39. Useful paper highlighting the benefits from using an integrated approach for FH management.
** Of considerable interest

14. Useful consensus statement that highlights the under-diagnosis and under-treatment of FH among the general population.

16. Good overview of current clinical problem in Australasia with helpful information on model of care for FH.

25. This paper provides a helpful introductory primary care perspective with advice on how to approach detection of FH in the community.

Financial Disclosure

None