POLICY OPTIONS

A proof of concept study to identify Familial Hypercholesterolaemia in primary care

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Policy context

Familial Hypercholesterolaemia (FH) is an inherited condition resulting in excessively high levels of cholesterol in the bloodstream from birth. This increases the risk of heart attacks and angina by age 40 or earlier if not treated. One out of every two relatives (parents, siblings and children) of affected people has the condition. At present only about 15% of affected patients are diagnosed while the other 85% remain at high risk of progressing to heart attacks at a young age. Early diagnosis and treatment are very effective in preventing heart disease developing.

This study examines the feasibility (acceptability, usefulness, and convenience) of using a new method of detecting the condition through GP clinics using a finger prick blood test in patients aged 18-40 years presenting for routine clinical appointments. If the test is positive, a fasting pathology test is then used to confirm the result and further clinical assessment undertaken. If FH is confirmed, cascade testing of close family relatives is strongly encouraged. This family tracing is the best available method to identify new patients with the condition as they stand to benefit most from early treatment. Until recently the condition has been managed mainly at hospital clinics.

The proposed new innovative approach will allow the condition to be managed by the patient's GP with support from the hospital specialist in keeping with the current thrust of State and Federal Government policy to increase primary care management of most chronic conditions. The key targets of this study are younger patients with most to gain from early treatment if they have the condition. This work will potentially have significant impact as patients and affected family members who have their condition treated can expect to live a normal life span without early heart disease complications.

Policy options

We found that 18 to 40 year olds were generally favourable towards being offered the opportunity for point of care testing (POCT) of FH as part of their routine primary care consultation. Most (30 out of 34 interviewees) were positive in their response to the value of early testing especially if there was a family history of heart disease or of lipid lowering treatment. Whilst the number of patients that were eventually opportunistically screened was below what we initially envisaged, those that were targeted did not find the process obtrusive or off-putting with most happy to be re-assured that at that particular point in time, their blood test showed no evidence of FH risk.

A positive benefit from the interaction with patients was that it made them more aware of the nature of FH and how early detection and treatment might be of future benefit. Apart from patients, practice reception staff, practice nurses (PNs) and GPs also benefitted through gaining increased awareness of the condition, increased knowledge about specific diagnostic criteria like the Dutch Lipid Score and clinical parameters as well as personal and family history of premature cardiovascular disease that might increase an individual's risk of having the condition. The fact that the entire POCT process was demonstrated to work in the opportunistic primary care setting was a very positive outcome. Patient acceptance and on-the-spot POCT for FH integrated well with the routine work of primary care consultations apart from busy periods, such as flu injection season, when the demands on practice staff and PNs were stretched and some opportunities for FH testing were inevitably missed.

One limiting factor that had a negative impact in the early phase of the study was the fact that the first batch of supplied lancets was not suitable for purpose. Whilst this problem was eventually identified and acknowledged by the supplier, it did result in some patients being lost, impaired recruitment and probably caused a loss of momentum in the early phase of the study.

The POCT model is well suited to the primary care setting and may present opportunities for further development and refinement in the future. Delivery of the process will need improvement but the general acceptance of the process together with the increased awareness of the condition among patients, reception staff, PNs and GPs was encouraging for future use.

Key findings

- POCT is feasible in implementation and acceptable within a GP setting provided there is good engagement with practice staff (reception staff, practice nurses, GPs)
- Increasing awareness of FH in the community will be a key factor in improving detection and management
- Encourage younger age group blood testing in patients with a family history of premature coronary artery disease
- For the undertaking of future research, it is important to:
    - Have a dedicated research person to work with practice staff
    - Have a research study champion from within the practice
    - Ensure research protocol fits with day to day running of the practice
    - Minimize paperwork not to overly burden participants

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