KEY MESSAGES

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

March 2016

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Policy context

The significance of Familial Hypercholesterolaemia (FH) is poorly appreciated among both general practitioners (GPs) and patients in the Australian community setting with the result that the condition remains largely unrecognised and undertreated. A newer approach focused on increasing awareness, early detection, lifelong treatment and cascade testing of close relatives is seen as essential in order to improve outcomes of patients with the disorder. Younger patients have most to gain as the condition, being hereditary, is present from birth and thus early diagnosis and treatment can prevent the cumulative cardiac complications that ensue from lifelong exposure to high circulating low-density lipoprotein cholesterol (LDL-c) levels and allow these patients to enjoy a normal lifespan. Our point of care testing (POCT) study looked at the feasibility of identifying incident index cases of FH in patients aged 18 to 40 years presenting for routine clinical appointments in primary care.

Key messages

- 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
- Research in general practice requires a pragmatic approach

The research reported in this paper is a project of the Australian Primary Health Care Research Institute, which is supported by a grant from the Australian Government Department of Health under the Primary Health Care Research, Evaluation and Development Strategy. The information and opinions contained in it do not necessarily reflect the views or policies of the Australian Government Department of Health.