Familial Hypercholesterolaemia (FH) Pilot Cascade Screening Project

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Familial hypercholesterolaemia (FH) is an inherited disorder of cholesterol metabolism leading to premature coronary heart disease (CHD). The early diagnosis and treatment of FH can delay or prevent the onset of CHD.

The Office of Population Health Genomics in collaboration with the Department of Internal Medicine (RPH), and the UWA School of Medicine and Pharmacology are supported by the Australian Better Health Initiative to run a pilot program of family cascade screening of FH cases in WA.

A model of care has been developed for screening adults and children at risk of FH. Index cases are identified through lipid and cardiology clinics, as well as through selected general practices. Once participants consent to the cascade-screening program, their relatives may be recruited into the program. All cases have an initial consult with the RPH Lipid Disorders Clinic, with ongoing medical care through their GP. So far, 69 index cases have been assessed by the FHWA clinic and 28 relatives are under clinical review.

This is the first report of an Australian FH program that has the potential to prevent 26 heart attacks in every 100 cases identified and treated, with a potential cost-effectiveness of $14,000 per life year gained. This program provides information on appropriate service frameworks, support for FH families and opens issues of targeted genetic screening, sharing family health information and the translation of research into improvements in community health.

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