Suboptimal Documentation and Management of Familial Hypercholesterolaemia in Patients Admitted with Suspected Ischaemic Heart Disease

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Introduction Familial hypercholesterolemia (FH) is an inherited disorder often presenting with premature coronary artery disease (P-CAD). Up to 75% of FH cases remain undetected; therefore an easily utilised in-hospital screening tool needs to be implemented in order to identify potential FH index cases, maximise secondary prevention in index and institute proven cost-effective family screening.

Approach and Findings We studied 210 patients (men <55 years and women <60 years) admitted to the Gold Coast Hospital with chest pain of suspected ischaemic origin. We applied FH international diagnostic criteria for FH and compared possible to documented FH diagnosis. The management of possible FH cases was also analysed. FH was considered in 1% of cases; fasting lipid levels were measured in 46% of cases; lipid measurements in first- or second-degree relatives were mentioned in 0 cases; a diagnosis of definite, possible or non-FH could be made was 54% of cases; international criteria for FH diagnosis was used in 0 cases; and 25% of possible FH cases were discharged on appropriate statin therapy.

Conclusions Minimal consideration was made to the diagnosis of FH in patients with suspected P-CAD. Inadequate history, physical examination and ordering of fasting lipid levels made retrospective FH diagnosis impossible in over half the patients. Treatment of possible index FH cases was suboptimal.

Future Directions A flow chart has been developed for use in patients admitted to hospital with suspected P-CAD. Implementation of this flow chart may improve identification and treatment of index FH cases and family screening of proven cases.

Opportunities for Collaboration in Health The identification and treatment of patients with FH ideally should involve GPs, cardiologists, nurses, genetic field workers, clinical geneticists, molecular biologists, dieticians, ophthalmologists and vascular surgeons. A central DNA laboratory is required for the unequivocal diagnosis of FH. Genetic field workers should visit homes for blood sampling and completing questionnaires. Specialised lipid clinics with dedicated dieticians and clinical geneticists or genetic counsellors are vital. Medical and nursing staffs are cognisant of the nature of FH and the effectiveness of CHD prevention with adequate treatment. Finally, families’ participation in cascade screening will diagnose and treat FH at the earliest possible stage.