

Cascade testing in familial hypercholesterolaemia: how should family members be contacted?

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Introduction

Familial hypercholesterolaemia (FH) is an autosomal dominant condition characterised by elevated cholesterol levels and increased risk of coronary heart disease. At least 75% of individuals with FH in the UK are thought to remain undiagnosed. **Effective primary prevention (with statins and lifestyle advice) is dependent upon early diagnosis.** This may be via lipid or genetic testing or both.

Issue addressed in paper

Cascade testing in relatives of known FH probands is an important mechanism for identifying those at risk of the condition. A 2 year program has been established in the UK to identify new cases of FH from relatives of patients already attending lipid clinics. Relatives are systematically identified, contacted and provided with information and then make an autonomous choice whether to undergo testing.

There are two methods of contact:

- (1) Family contact –The proband is provided with information and clinic contact details and it is their responsibility to pass these to relatives. This method is associated with less risk of psychological harm and protects confidentiality.
- (2) Direct contact – The proband provides family names and contact details and then the clinic contacts them directly. It has been proven that more people present for screening and there is better communication / transfer of information using this method.

Summary of findings

The ethical and legal issues associated with these two methods of contact were outlined. **The paper argues that direct contact is ethically justifiable in the context of FH cascade screening.**

The ethical issues associated with direct contact were divided into 6 main classes:

1. *Psychological harm to relatives*: to minimise this risk it was suggested probands should informally discuss the condition / testing with relatives first.
2. *Right not to know*: because FH is a preventable genetic condition there may be an ethical imperative to provide choice which overrides the right of an individual “not to know” about their risk of a condition.
3. *Actual harm to relatives*: Direct contact versus family contact is not thought to provide any increased risks of discrimination.
4. *Detrimental impact on family dynamics* – this can be combated by discussing family dynamics with the proband and then using a different system of contact if problems are likely to be encountered.
5. *Breach of confidentiality*: It was proposed relatives’ names and addresses should not be added to the cascade screening register until there is a positive response to contact. If the proband refuses to provide details for family members then the paper argues there may be ethical justification to breach the confidentiality of the proband but only once very strong criteria have been met.
6. *Potential for undue influence*: the voluntary nature of testing must be emphasised to ensure the clinical context does not pressurise people into making inappropriate decisions.

The main advantages of direct contact were outlined:

1. *Efficiency of cascade testing programme*: More people receive potentially life saving health information and advice. The cost effectiveness of screening is also higher.
2. *Increased accuracy of information*: Everyone receives the same high quality information about FH.
3. *Preventing harm to the proband*: The burden on the proband of discussing testing with relatives is removed.

It was concluded that initial family contact should be established by the proband (if they wish). The cascade screening programme is then the “second line” of contact, but the main source of counselling, information and advice.