

How Cost-effective Is Screening for Familial Hypercholesterolemia?

Marks D, Wonderling D, Thorogood M, et al. Cost effectiveness analysis of different approaches of screening for familial hypercholesterolaemia. *BMJ* 2002;324:1303–8.

Study Overview

Objective. To identify cost-effective strategies for detecting and treating familial hypercholesterolemia.

Design. Cost-effectiveness analysis.

Setting and participants. Simulated population aged 16 to 54 years in England and Wales.

Methods. Computer modelling was used to investigate the following screening strategies: universal screening, opportunistic screening of primary care patients, screening of patients hospitalized for premature myocardial infarction, or tracing relatives of patients with diagnosed familial hypercholesterolemia.

Main outcome measures. Cost-effectiveness of interventions calculated as cost per life-year gained, including estimated costs of screening and treatment.

Main results. The most cost-effective screening strategy was tracing the relatives of patients diagnosed with familial hypercholesterolemia (\$4479 per life-year gained). Screening limited to 16-year-old patients also was cost-effective, at \$4087 per life-year gained. Universal screening was the least cost-effective at \$19,176 per life-year gained.

Conclusion. Testing relatives of patients diagnosed with familial hypercholesterolemia was the most cost-effective screening strategy.

Commentary

Familial hypercholesterolemia is an underdetected autosomal dominant disorder associated with a high risk for premature coronary artery disease. Effective treatment from statin medications is available for both primary and secondary prevention of heart disease. The disorder can be detected clinically by measuring plasma cholesterol and obtaining a family history or with genetic testing. Because it is not identified in 75% of patients with the disease [1], population-based screening to increase early detection is desirable.

Determining the cost-effectiveness of different screening strategies is one of the most important steps before making new screening recommendations. The authors' analysis shows that testing all patients for a genetic mutation is insensitive and not cost-effective; therefore, alternative methods are needed. Marks et al report that contacting relatives of patients already diagnosed with the disorder and screening 16 year olds universally are the most cost-effective strategies. However, tracing relatives does not detect families where no member has yet been diagnosed. Additionally, screening 16-year-old patients would not help the many undiagnosed persons over that age. Also, it would have been useful to know the incremental cost-effectiveness of adding the universal screening approach to the tracing strategy. Finally, it is important to note that all of the screening strategies presented by the authors except universal genetic testing are cost-effective by U.S. standards, where a \$60,000 per life-year gain is a commonly accepted threshold.

Applications for Clinical Practice

Various screening strategies for familial hypercholesterolemia are cost-effective by U.S. standards; universal screening of 16-year-old patients and tracing families of patients already diagnosed are the 2 most cost-effective strategies. If tracing family members or universal screening is impractical, then opportunistic screening in the primary care or hospital setting is an acceptable compromise.

—Review by Josh F. Peterson, MD

References

1. Neil HA, Hammond T, Huxley R, et al. Extent of underdiagnosis of familial hypercholesterolaemia in routine practice: prospective registry study. *BMJ* 2000;321:148.

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