

Title: A systematic review of economic evaluations of genetic testing technologies

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Background: Genetic test technologies offer hope for early diagnosis and identification of persons at risk for serious diseases. Stemming in large part from the sequencing of the human genome, numerous genetic tests have been introduced into clinical practice. Because many of these tests are costly and applicable to large populations (e.g., screening all newborns for cystic fibrosis), evaluations of the cost-effectiveness of these technologies is important.

Objectives: To conduct a systematic search for and review of economic evaluations of genetic testing technologies.

Methodology: Literature searches were performed using PubMed, Proquest, LexisNexis, Expanded Academic Index, The Harvard Review of Economic Analyses, PsycINFO, National Institute for Clinical Excellence, and The Canadian Council on Technology Assessment in Health Care. For resources amenable to mesh searching, the mesh terms included were: economic(s) and/or cost(s), combined with genetic, gene, and/or genotype. Searches were limited to English language with publication dates from 1990 to present. Selection criteria included original articles in cost-effectiveness (as defined by Drummond et al, Oxford Press 1997) in genetic services. To be considered as a genetic service, either the disease or condition had to be primarily genetic or involve a genetic test, defined as the analysis of human DNA, RNA, chromosomes, proteins, and certain metabolites in order to detect heritable disease-related genotypes, mutations, phenotypes or karyotypes for clinical purposes. Articles were categorized by clinical category and type of economic study (e.g., cost-utility, cost-benefit), and graded independently by the authors using CEA study quality system developed by Chiou et al (Med Care, 2003;41:32).

Results: A total of 149 abstracts were retrieved using the search terms; 63 met selection criteria. Types of economic studies were as follows, cost-utility studies (25%); cost-benefit (19%); cost-minimization (6%); cost-effectiveness analyses (59%). Clinical categories were as follows: preconception carrier testing (8%); prenatal diagnosis (40%); adult testing (57%). (Totals >100% due to some studies having more than one type of analysis). The studies considered interventions for 26 different medical conditions. Study quality using the Chiou et al grading method ranged from 43-100 (average 82). Cost-utility studies were of highest quality (mean 91); cost-minimization studies were of lowest quality (mean 63). Adult studies had the highest rating (mean 86); preconception testing studies were lowest in quality (mean 74). Intraclass correlation among raters was 0.82 (CI 0.70-0.89).

Conclusions: A substantial number of economic analyses have been published in human genetics across a wide range of conditions. A large proportion of studies do not include a measure of benefit that facilitates comparison with other medical interventions. Study quality varied widely. Priority areas for the field include increasing quality and uniformity of measures of outcome.

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