Health Economics and Genomics

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Background

Many diseases are thought to have an underlying genetic basis, caused by mutations in an individual's DNA sequence. However, many patients remain undiagnosed even when all possible tests have been performed. There is now the scientific ability to test much more of an individual's genome and explore many genes simultaneously. So called high-throughput sequencing could have major implications for patient care if adopted into the NHS. However, little is known about whether genomic technologies are likely to be cost-effective and provide value for money for the NHS.

HERC has had a programme of research into the health economic implications of adopting genomic technologies since 2003. We focus on generating evidence to support the translation of genomic techniques into clinical practice through a variety of applied and methodological work. We currently have seven ongoing studies in a mix of clinical areas, including cardiology, leukaemia, breast, ovarian, colorectal, lung and skin cancer, infectious diseases, mitochondria disease and IVF. The presented projects here are a selection of completed and ongoing projects.

Patient Preferences for Genomic Technologies

- A key challenge is how to evaluate the benefits of genomic technologies, as a commonly used measure, quality adjusted life years (QALYs) may not be appropriate.
- A discrete choice experiment (DCE) is an alternative approach and is a survey technique which elicits patient preferences for different attributes of an intervention (e.g. for a genetic test: effectiveness, cost, time to results) and allows us to quantify the strength of these preferences and place a monetary valuation on health outcomes. We are conducting a DCE in chronic lymphocytic leukaemia (CLL) patients to evaluate their preferences for microarray testing and targeted next generation sequencing. Patients are asked to choose between alternative testing scenarios. To date, 220 patients from across the UK have completed the survey.

Figure 1: Example scenario from DCE in chronic lymphocytic leukaemia

EXAMPLE QUESTION

Consider the following features describing two tests (**Test A** or **Test B**). Please indicate with a cross (X) which test you would choose:

Feature	Test A	Test B
Time to receive the test result	8 days	5 days
Cost of the test **to you**	£260	£600
Ability of the test to predict who will **not** respond to the usual chemoimmunotherapy treatment	Test identifies 50 out of every 100 patients who will not respond to usual treatment	Test identifies 90 out of every 100 patients who will not respond to usual treatment
Test reliability	2 out of every 100 tests provide an incorrect result	4 out of every 100 tests provide an incorrect result
Length of time health care professionals spend describing the test to you	10 minutes	5 minutes
Type of health care professional who explains the test result to you	Consultant hospital doctor	Junior hospital doctor

Which test would you prefer? ☐ TEST A ☒ TEST B

Assessing the Mainstreaming of Cancer Genetics

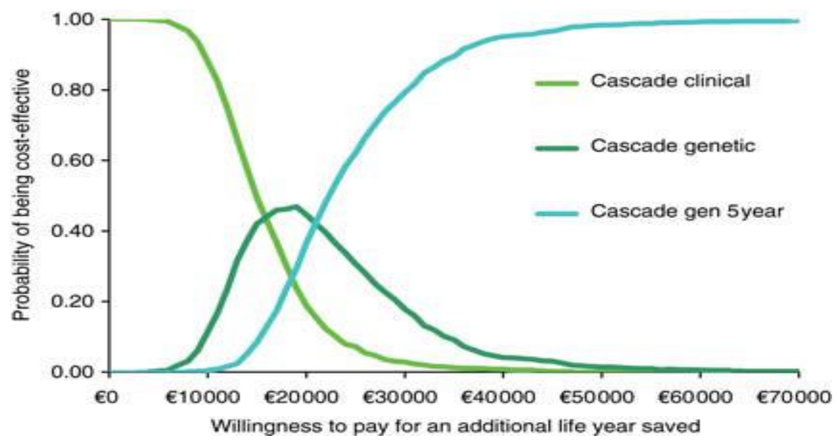
- There is currently variation and inequity in Cancer Genetic Service delivery but potential, through technological advances, to greatly increase availability and affordability of cancer gene testing.
- The Mainstreaming Cancer Genetics (MCG) Programme is a translational research programme aiming to make genetic testing part of routine cancer patient care.
- Currently, all cancer gene testing is undertaken by geneticists. The MCG is implementing a new model in which testing in cancer patients can also be done by oncologists.
- The Institute of Cancer Research, London, in partnership with Ilumina, has developed the TruSightTM Cancer Panel which can analyse 97 cancer predisposition genes at once.
- The programme aims to make the entire process of genetic testing in cancer patients as practical, robust, efficient and cost-effective as possible.
- In collaboration with the MCG Programme and the Royal Marsden Hospital, Sutton, HERC is estimating the cost-effectiveness of both the new model of service delivery and the new sequencing technology.
- Average costs per patient pathway are being determined through a micro-costing analysis and combined with data on effectiveness.

DNA testing for hypertrophic cardiomyopathy: a cost-effectiveness model

Hypertrophic cardiomyopathy (HCM) is the most common monogenic cardiac disorder (1/500) (1) and most frequent cause of sudden cardiac death (SCD) in young people and trained competitive athletes. (2) HCM is defined by unexplained hypertrophy of the ventricular myocardium in the absence of a detectable cause. However, until recently, clinically testing alone was undertaken rather genetic testing. As such, Oxford University scientists and clinicians undertook a large programme of research to assess the viability of DNA (genetic) testing in the management of HCM. HERC undertook the cost-effectiveness analysis (Wordsworth et al, 2010) (2) as described below.

- We aimed to explore the cost-effectiveness of alternative methods of diagnosing family members for HCM.
- We built an economic decision model to compare cascade screening using genetic testing, as opposed to clinical methods.
- The incremental cost per life year saved was €14.397 for the cascade genetic compared with the cascade clinical approach. The costs for cascade molecular genetic testing were slightly higher than clinical testing in the short run, but this was largely because the genetic approach is more effective and identifies more individuals at risk.
- Our conclusion was that the use of DNA testing in the diagnosis and management of HCM was a cost-effective approach to the primary prevention of SCD.

Figure 2: Cost-effectiveness acceptability curve for DNA testing in HCM



The economics of sequencing human pathogens in infectious diseases

Identifying pathogens such as mycobacterium tuberculosis (TB) infection through routine laboratory service is time consuming (six weeks for TB), requires multiple techniques and it is challenging to find out about pathogen drug resistance. Illumina MiSeq, a whole-genome sequencing (WGS) platform, is expected to reduce turnaround time to just 24 hours, resulting in improved patient care at lower cost (through faster receipt of appropriate antimicrobials) and/or reductions in transmission. We are evaluating the cost-effectiveness of WGS compared to current NHS laboratory testing methods. Average costs and effects on a per patient basis are being estimated through micro-costing, with new infections averted and inappropriate antimicrobials avoided as the main outcome measures. This is a multicentre study including laboratories in Oxford, Birmingham, Brighton, Leeds, Newcastle, Lille, Hamburg, Dublin, Vancouver and the National Mycobacterium Reference Laboratory.

Developing Economic Methods

Developing economic evidence in genomic interventions is challenging resulting in gaps in evidence, which is slowing translation into clinical practice. (3) Examples of methodological challenges that we are addressing include:

1.The appropriate analytical perspective, timeframe and timing of analysis are unclear.

2. A broad range of cost data must be collected, frequently, in a data-limited environment.

3.Measuring outcomes is problematic as standard measures (e.g. QALYs) are less useful in this context, and alternative metrics (e.g., personal utility) and approaches (e.g., cost–benefit analysis) are underdeveloped and underused.

4.Effectiveness data quality is weak and challenging to incorporate into standard analyses.

5.Little is known about patient and clinician preferences for genomic technologies.

References

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Our Collaborators and Funders

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