



Managing genetic tests, surveillance, and preventive medicine under a public health insurance system



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ABSTRACT

There is a prospect in the medium to long term future of substantial advancements in the understanding of the relationship between disease and genetics. We consider the implications of increased information from genetic tests about predisposition to diseases from the perspective of managing health care provision under a public health insurance scheme. In particular, we consider how such information may potentially improve the targeting of medical surveillance (or prevention) activities to improve the chances of early detection of disease onset. We show that the moral hazard implications inherent in surveillance and prevention decisions that are chosen to be privately rather than socially optimal may be exacerbated by increased information about person-specific predisposition to disease.

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1. Introduction

It is fair to say that genomic science is now well into its second phase since current research involves not only the identification of so-called “disease genes” or, more appropriately, “disease alleles”, but also the understanding of how specific sequences of genes interact with each other and with environmental factors to affect the onset and influence the treatment of diseases. Claims in the scientific literature and the media suggest that advancements made in genetic information will lead to significant improvements in the effectiveness of prevention and treatment of disease. A rough road map of the human genome has been available since 2003 and currently, according to the NIH-sponsored web site genetests.org, there are over 1600 genetic tests used clinically. With the prospect of the so-called \$1000 genome close to reality (see [Davies, 2010](#)), whole genome sequencing may soon become the norm for developed countries. The information that can be gleaned from an individual’s whole genome has the potential to revolutionize the

practice of medicine with population wide genome sequencing forming the basis of so-called P4 medicine (i.e., medicine that is Predictive, Preventive, Personalized and Participatory). Although the future of P4 medicine has many proponents, not least of whom is Leroy Hood through his P4 Medicine Institute (p4mi.org), there is some controversy over the pace of its progress.¹

Once the relationships between specific genes, environment, and diseases are better understood, harnessing this information to create improved health outcomes in a cost effective manner requires a good understanding of how individuals will behave in the context of such individualized informational change. We provide insight into this debate by focusing on how individuals’ incentives for use of surveillance (monitoring) technologies, such as colonoscopies or mammograms, change in the presence of risk-type specific information about the likelihood of onset of disease. It has been debated in the literature whether population wide screening for diseases such as colon cancer or breast cancer is cost effective and whether monitoring should be restricted to those at

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¹ As noted by [Roukos \(2008\)](#), “although personalized medicine and oncology in clinical practice is still a dream, some isolated first steps have been taken.”

higher risk as identified, for example, by family history. As genetic tests become more wide ranging and less costly, there is the potential of substantial improvements to the targeting of surveillance techniques such as colonoscopies with the potential of improved overall health outcomes in a more cost effective manner. However, we show that the usual moral hazard problems associated with insurance coverage may interact with improved knowledge of individual risks in a way that could blunt the potential for such improvements. Through the use of simple models, we develop a series of results which characterize the possible outcomes that could develop as more genetic information becomes available.

Many genetic tests continue to be expensive and so choosing which tests to make available through health insurance plans, be they private or public, represents a challenge. Insurance or health care providers are concerned about the possibility of escalating costs due to the adoption of more genetic tests (e.g., see report by Miller et al. (2002) funded by the Ontario Ministry of Health and Long Term Care) while others believe improved targeting of surveillance and preventive measures will ultimately reduce health care costs.² It is this aspect or phase of growth in genetic testing and related knowledge that we address here. In particular, we study the implications of improved genetic information about risk of disease in terms of the socially optimal management of surveillance and related health care strategies for public health insurance systems. The results of this exercise can be helpful in developing guidelines to use in determining which genetic tests to offer within the coverage of the public health system. Some aspects of what we find could also be applied to a population covered (or partly covered) by private health insurance, although there are some important differences to consider.

Many of the papers that model the effects of improved information about risk classification involve the private insurance market and exogenously specified (fixed) probabilities of disease and/or financial loss (e.g., Rothschild and Stiglitz, 1976; Wilson, 1977; Hoy, 1982, 1984; Crocker and Snow, 1985, 1986; Tabarrok, 1994; Hoel and Iversen, 2002; Rees and Apps, 2006).³ Our model also involves exogenously determined differences in the probabilities of onset of disease. However, we allow for the possibility of early or late detection of disease through individuals' choices of level of surveillance. For many diseases, early detection leads to improved treatment and outcomes. Information from genetic tests creates (or increases) differential assessment of risk of disease onset across individuals. Thus, although probability of onset may be fixed by genotype, choice of level of surveillance creates endogenous determination of detection being late or early (i.e., at least probabilistically). The possible benefit of a genetic test in this context arises from potential improvements in targeting of surveillance strategies for early detection of onset of disease. The important management issue is in determining the extent to which higher (lower) risks should increase (decrease) surveillance and then trying to encourage the appropriate responses from individuals. We show that a model of differential use of preventative medicine based on genotype is very similar and so determination of the value of genetic tests follows a similar pattern relating to improved targeting of such strategies.⁴

It is well known that in the presence of health insurance, be it public or private, individuals face incentives that lead to actions that are not necessarily socially optimal. In our context,

we presume that individuals do not pay for the financial costs of surveillance or treatment, should onset of disease occur. The result is that individuals may either over-use or under-use medical surveillance or prevention. The moral hazard problems due to insurance are complicated by the introduction of information about differential risk of disease onset. We characterize how genetic testing can lead to changes in the pattern of over- and under-use of surveillance. We find, under a broad range of scenarios, that at least one group (i.e., the average, high or low risk types) will tend to want to over-use surveillance relative to the socially optimal decision. The relative extent to which over-use (or under-use when it occurs) of surveillance reduces social welfare can vary across the groups in counter-intuitive ways. Overall efficiency may fall as improved knowledge about risk type interacts with the standard moral hazard implications of insurance leading to a reduction in social welfare.

In the following section, we introduce a simple model of surveillance, which is also referred to as screening or monitoring. The basic model describes the decision for intensity of monitoring taken by the individual and compares that to the socially optimal decision. In Section 3, we present our results regarding the implications of introducing genetic tests and in Section 4, we briefly consider the case of private insurance and implications of explicitly accounting for costly genetic tests. We provide a discussion, conclusion, and suggestions for further research in the final Section 5.

2. Model of medical surveillance

The role of surveillance is to increase the likelihood of early detection of disease. One key aspect of the model is the relationship between the intensity of surveillance and its effectiveness at early detection, including its financial cost. In the context of screening for colon cancer, one can think of the use of FOBT – fecal occult blood test – as a low level and low cost approach to screening; FSIG – flexible sigmoidoscopy – as an intermediate level and intermediate cost approach; and CSCPYP – standard colonoscopy – as a higher intensity and higher cost method of screening. The relative unit costs of these approaches, quoted in U.S. Congress report OTA-BP-H-146 (1995) are \$10, \$80, and \$285 respectively while the {sensitivity, specificity} in regards to detection of cancer are {40%, 90%}, {90%, 98%}, and {90%, 100%}, respectively. Moreover, FOBT is not very effective at detecting pre-cancerous polyps (sensitivity of only 10%) compared to colonoscopy (sensitivity of 90%). One can then think of an intensity of surveillance as a mixture of the various techniques that one can apply with varying frequency starting at a particular age (e.g., FOBT once yearly with CSCPYP once every five years starting at age 50). We describe the relationship between the intensity of surveillance and the probability of early detection of disease by the function $p^{ED}(s)$, with $p^{ED}(s) > 0$ and $p^{ED'}(s) < 0$; that is, the probability of early detection of disease increases (at a decreasing rate) with the intensity (and/or frequency) of surveillance as measured by s . The financial cost to the health care system of providing an individual with level of surveillance s is $C(s)$, which we assume is increasing and convex in the level of surveillance; i.e., $C'(s) > 0$, $C''(s) > 0$.⁵

The financial cost of treatment, for those who eventually have onset of disease, depends on whether the disease is detected early or late. In our model, we have in mind all future lifetime medical costs conditional on stage of detection of disease. We refer to these as C^{DE} and C^{DL} for cases of early and late detection, respectively. The

² See, for example, Caulfield et al. (2008) for a critical evaluation of such claims.

³ See Hoy (1989), Doherty and Posey (1998), and Hoel and Iversen (2002) for examples of models where self-protection (or prevention) can affect the probability of loss/disease differentially according to risk (geno-)type.

⁴ See Filipova-Neumann and Hoy (2009) for a model describing the implications of genetic testing for differential impacts of prevention strategies based on risk type.

⁵ Generally we may allow $C'(s) = 0$ and still satisfy conditions for an interior optimum. Linearity of $C(s)$ may reflect more frequent (repeated) applications of a given monitoring technology.

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