



Risky knowledges: the sociocultural impacts of personal genetics in a knowledge-driven economy

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Abstract

The rapid developments in modern genetics are changing the way disease and wellness may be considered. New concepts are emerging such as predictive genetic testing and forecasting future disease, as well as internet genetic analysis available to the public. In this short communication we consider some of the implications relating to predictive genetic testing in the public domain.

Twenty-five years ago, genetics had a small part to play in general medicine. Although it was known that genes held the 'recipe' for the human body, and could provide clues relating to why health was affected, the understanding of the information and its application was a distant goal. There was little that could be done to predict how an individual's genes may lead to an illness or how a disease may be prevented in which genes played a part.

The inheritance of single gene disorders such as cystic fibrosis was understood, but how the incorrect genetic information led to the disease was poorly understood. Such a poor understanding of the molecular basis of disease led to inadequately formulated intervention strategies based on symptoms alone.

As molecular and biotechnology knowledge progressed, the specific identity of genes associated with both health and disease has increased producing changes in the conventional perceptions of disease. Genes have been identified which make certain individuals susceptible to a disease rather than actually having the disease. Equally in families, various members can be identified as either having a mutated gene, thereby increasing the risk of a disease or not having the mutation and therefore having a 'genetic advantage' over parents, siblings, or other members of the extended family.

The impacts of such knowledge became increasingly apparent as the draft of the Human Genome programme began to unfold in 2001 identifying approximately 30,000 genes from which we have both normal and abnormal function. Information relating to human genetics and health now escalates weekly—demonstrating that not only are genes implicated in disease but also they influence or are influenced by environmental factors on our bodies, including nutrition and how we repair, age, and respond to treatment.

Contemporary health applications of genetic knowledge

Rapid development of genetic testing and information has led to very specific tests for certain single gene disorders and implicated many other genes in the disease process. Genetic diseases not yet manifest can now be identified which has led to the development of predictive genetic testing—i.e. testing for a genetic disease which could (e.g. Huntington's Disease) or may (e.g. inherited breast cancer) develop in the

future. In effect, individuals are being identified as being genetically susceptible to *future* disease, changing the concept of health being defined as the presence or absence of disease to the concept of increased or decreased statistical probability of future disease developing.

These biotechnological advances can also predict whether a pre-implantation embryo may be susceptible to a genetic disease which (if it developed) would appear many years after the infant was born. Such a predictive test cannot, however, specifically identify that the individual will develop the disease.

Future innovations in genetic testing

Based on the use of the new genetic technologies, several major pharmaceutical and biotechnology companies¹ are promoting the concept of ‘predictive medicine’ or ‘predisposition profiling’. This is the use of genetic tests to predict the chances that an individual will develop a serious illness (e.g. cancer, mental illness, heart disease).

A positive aspect of using this information is offering advice on lifestyle changes which may offset the physiological impacts of the abnormal gene—i.e. shifting the emphasis of medicine² from ‘diagnosis and treatment’ to ‘disease prediction and prevention’. However, concerns arise regarding the use of the information.

For example, pharmaceutical companies may not only sell the genetic testing kits but also manufacture the drugs for treating those at high risk, or supply special dietary supplements when testing for ‘nutrition genes’.

Medication may be given to otherwise healthy people who may never develop the disease thereby changing the resourcing structure of the health system, and information may be given to people who will worry unnecessarily or seek treatment for a health problem which may never occur. It places very personalised information into the broader community-raising issues relating to genetic identity, employment, insurance³, paternity⁴, and forensic use.

Generally, the assumption has been that genetic testing would occur in a medical context. However, recent developments in the United Kingdom and the USA (where the availability of ‘over-the-counter’ genetic tests exist in an unregulated environment) have raised major issues relating to:

- Quality and reliability of information,
- Interpretation of information,
- Entitlement of ‘not-to-know’ in other family members,
- Unexpected information,
- Testing for genes with social implications,
- Security of individual DNA and intellectual property rights,
- Role of informed consent, and
- The perceptions of society at large on identifying individuals with potential genetic disease.

Horizon scanning for unregulated practice

Although this is a relatively new area of applied health biotechnology, the transition from the disease gene identification process and the marketing of a diagnostic test will be rapid.

There are at least 10 multinational companies planning to sell genetic testing kits, which include Abbott, Bayer, Johnson and Johnson, and Roche. All of these and their related companies have links or agreements with at least 17 gene discovery companies who are searching for patentable gene products or data from abnormal gene function (www.forbes.com).

Currently companies are selling personalised genetic testing over the Internet with services being offered for cancer susceptibility genes ('The best time to beat cancer is before you ever get it'—www.myriadtests.com); nutrition-related genes (www.scicona.com); paternity, maternity, immigration testing, and genealogy (www.genetrack.com); and home paternity testing (www.dna-worldwide.com).

More recently, a new DNA test has been offered in the UK and USA for fetal sex testing at 6 weeks gestation as well as gene profiling for diseases with Mendelian inheritance patterns in at-risk groups such as Ashkenazi Jews ("Ashkenazi kits"—www.elugicene.com).

Conclusion

Direct-to-consumer genetic testing⁵ is a new and rapidly emerging area both for health-related and non-health related applications, which moves the traditional concept of testing from health providers to the individual and a third party. This raises significant issues relating to accuracy and reliability of the testing services information, the competency of the genetic testing provider, the interpretation and reporting of the results to an individual or family (with little or no support or skills to interpret the information), and the loss of genetic privacy—all of which could have significant impacts on major life decisions.

Whether obtaining such results will provide benefits to the individual is still not known, but we believe that there is potential for significant societal impacts resulting from the use of unregulated personal genetic-testing technologies. The creation of a new social group of the 'at-risk well'; the requirement for health professionals to understand and communicate the implications of the 'new knowledge'; and the demands that might be placed on medical practitioner consultations and use of the diagnostic services will increase as more genetic information becomes available and disease linkages are made with specific genes.

Rabinow (1999)⁶ predicted of this time of rapidly increasing personal genetic knowledges as the age of biosociality in which the most minute of social interchanges will be governed by our biological identities and their associated risk benefits. Rose and Novas (2005) talk of it as 'biological citizenship'.⁷

Many of the genes may ultimately have significant social implications—such as genes associated with mental illness, alcohol susceptibility, and infectious diseases such as HIV and hepatitis—thus creating the potential for the increasing misuse of this genetic information.

These dramatic societal impacts of this style of genetic testing suggest that empirical research in New Zealand should be undertaken before this new area of applied health biotechnology becomes embedded into public practice.

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References:

1. Read J. Schizophrenia drug companies and the internet. *Soc Sci Med.* 2007;Sep 7. Abstract at http://www.unboundmedicine.com/medline/ebm/record/17826878/abstract/Schizophrenia_drug_companies_and_the_internet
2. Gooding H, Organista K, Burack J, Biesecke B. Genetic Susceptibility testing from a stress and coping perspective. *Soc Sci Med.* 2006;62(8):1880–90.
3. Van Hoyweghen I, Horstman K, Schepers R. Making the normal deviant: The introduction of predictive medicine in life insurance. *Soc Sci Med.* 2006;63(5):1225–35.
4. Williams S. Genetic Genealogy. The Woodson Family's Experience. *Culture Medicine Psychiatry*, 2005;29(2):225–52.
5. Williams-Jones B. 'Be ready against cancer, now' direct-to-consumer advertising for genetic testing. *New Genetics and Society.* 2006;25(1):89–107.
6. Rabinow P. Artificiality and Enlightenment: from socio-biology to biosociality. In: Samson C, editor. *Health Studies: A Critical and Cross Cultural Reader.* Oxford, UK: Blackwell Publishers; 1999, p50–60.
7. Rose N, Novas C. Biological Citizenship. In: Ong A and Collier S, editors. *Global Assemblages. Technology politics and ethics as anthropological problems.* Oxford, Blackwell Publishers; 2005, p439–63.