Article

Privacy of Genetic Information

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On 9 May 1998, the Bioethics Research Centre saw the first Master of Bioethics and Health Law (MBHL) degree conferred. Hamish Broadbent (25) gained his science and law degrees prior to commencing the Centre's interdisciplinary MBHL programme under the supervision of Barbara Nicholas and Prof. Peter Skegg. Hamish completed his thesis in March 1997, three months after leaving Dunedin to become a policy analyst at the Ministry of Health in Wellington. Hamish now works as a legal officer for the Health and Disability Commissioner.

Hamish's MBHL thesis, 'Privacy of Genetic Information', examined the legal and ethical implications of knowing one's genetic makeup. Reproduced below is Chapter 3 of 10 from his thesis. This chapter introduces the reader to issues of genetic testing and seeks to define 'genetic information'.

The recent advancements in genetic biotechnology have greatly expanded our understanding of the human condition, especially in regard to illness and disease. Identifying individual health problems is no longer reliant on detecting symptoms, but instead can be predicted with some degree of certainty by genetic testing.

Genetic Testing

My thesis, does not address in depth the reasons why an individual may seek genetic testing. However, it is acknowledged that an understanding of genetic testing practices, particularly in New Zealand, will foster an appreciation of the uses for genetic information, help define areas of concern, and identify practices which may, or may not, require some form of control. For instance, to make an initial differentiation, genetic testing encompasses two activities – genetic monitoring and genetic screening.

1.1 Genetic Monitoring

Genetic monitoring involves regularly examining an individual to evaluate modifications to their genetic material due to chromosomal damage or mutations. Such monitoring may be desirable in an employment setting if there is continued and prolonged exposure to hazardous substances, and where genetic changes may increase risk of illness.[1] Although monitoring in the workplace is rarely practised in New Zealand, it is used with increased regularity in overseas countries, particularly the United States.[2] However, as ambient exposure and lifestyle decisions involving environmental factors (e.g. smoking, ultra-violet rays) may also induce genetic change, genetic monitoring is useful outside of the workplace.[3]

1.2 Genetic Screening

Genetic Screening, unlike genetic monitoring, involves a single assay designed to examine the genetic makeup of an individual for certain inherited characteristics. Traits include those which may render an individual susceptible to a pathological effect if exposed to specific agents or environment. For example in the United Kingdom, those who apply to defence forces which operate in atypical atmospheric conditions are required to undergo screening for the sickle cell trait.[4] Such conditions are labelled occupational or multifactorial conditions. Alternatively, individuals can be screened for severe inherited conditions, usually single gene defects which cause late-onset of condition regardless of environmental factors, (e.g. Huntington's Disease (HD) or neurofibromatosis).[5]

As more genes for genetic diseases are identified, predictive presymptomatic tests are destined to become a medical boom industry. Tests are already available for certain disorders in most parts of the Western world.[6] For example, identification of genes that predispose people to hereditary nonpolyposis colon cancer has resulted in ten companies in the United States purchasing rights to develop tests.[7] It is important, therefore, that in developing these tests, regulations are implemented which guarantee quality of screening programmes, the taking and storage of blood samples, and accuracy of results. This may reduce the likelihood of false-positive and false-negative outcomes. In New Zealand the introduction of appropriate genetic services forms part of the overall management scheme.

Genetic Services in NZ

2.1 Why the Need?

Providing genetic testing services, like all medical services, needs an infrastructure to enable genetic testing to operate successfully, and to ensure that testing does not create more problems than it can solve. In July of 1995, the National Advisory Committee on Core Health and Disability Support Services (NHC) commissioned a detailed report entitled 'Priorities for Genetic Services in New Zealand'.[8] In the foreword to the report, the committee advised:

> . . . [the] study of genetic diseases is a field where medical, scientific and social knowledge and technical capacity is expanding rapidly, and as a consequence there has been an increase in the development of screening tests and rapidly arising expectations from health professionals and members of the public that diagnostic and counselling services should be provided.[9]

The NHC report outlines to the committee the make-up, organisation and content of genetic services offered in New Zealand, primarily in the areas of clinical, laboratory and screening services. Further, the report recommends that these services be publicly funded, and more available to the general public.[10]

2.2 Clinical Services

Genetic disorders, together with congenital abnormalities, account for approximately forty per cent of all infant deaths in developed countries, and constitute a significant per centage of adult diseases.[11] However, through genetic testing, diagnosis of these conditions can be made prenatally, and even at the pre-implantation stage.

Despite the inability to cure in many cases, postnatal diagnosis of disorders in infants is equally beneficial, allowing better healthcare management and planning. This may include medical and dietary intervention, special education needs, and attempts to mitigate psychological implications and stigmatisation. In addition, presymptomatic testing of youths or adults for severe late onset conditions also provides opportunities for individual management.[12]

The NHC report also identified that genetic services in New Zealand were limited, and comprised just two clinical geneticists and two genetic counsellors.[13] International recommendations for full time clinical geneticists is one per 500,000 people. Based on these guidelines New Zealand requires seven full-time clinical geneticists to meet projected demands. Ideally two genetic counsellors would be expected to work with each clinician, in addition to other specialised personnel, for example, dieticians, field workers, physiotherapists, occupational therapists and secretarial staff.[14]

Education of the public is also a priority, not only so individuals at risk understand the incidence and likelihood of genetic disease, but also the benefits of self-referral. Healthcare service providers also need to be aware of a patient's increased risk, and understand their obligations to refer patients to clinical genetic centres when necessary.[15]

2.3 Laboratory Services

Laboratory diagnosis of genetic disease supplements the medical evaluation performed by the clinical geneticists who make the initial diagnosis through recording family histories and patient examination. The current services in New Zealand can be split into three specific areas: Cytogenetic Laboratories, Molecular Genetic Laboratories, and Biochemical (Metabolic) Genetic Laboratories. These all operate under the standard and quality parameters of TELARC[16] and the Human Genetics Society of Australasia (HGSA).

2.4 Screening Services

Screening is a separate service which involves testing of the general population for particular conditions. Screening is voluntary and includes, for example, tests which detect newborn metabolic disorders,[17] and maternal serum screening for chromosomal disorders. The NHC report outlines the importance of genetic healthcare services, stating that there is a:

> . . . special obligation on the screening programme . . . to care for subjects who are affected by the screening programme, since these individuals were approached for screening and did not seek intervention by the programme.[18]

Newborn screening[19] is one area which recognises the importance of this ideal, and only screens for conditions which are treatable by way of special diet or medicine. Approximately ninety-five per cent of newborns in New Zealand each year (60,000) have a blood smear taken about two to three days after birth.[20] Of these, approximately thirty to thirty-five newborns are found to have one of the metabolic conditions screened for.[21] Recently the test for cystic fibrosis has changed,[22] and as a consequence identifies the parental carriers of the disease, as well as the affected child. This information is provided to parents with appropriate counselling and support.[23]

The NHC Report indicates that laboratories performing screening services are required to conform to the rigorous standards and guidelines.[24]

Genetic Information

3.1 Genetic Information Defined

The starting point of further discussion in my thesis presumes genetic testing has been performed on an individual. Consequently my thesis does not address that health information derived from analysing an individual's physical traits, or that based on family histories.[25]

Instead, I address only genetic information that is derived from DNA tests, reports interpreting these tests, and diagnoses determined by geneticists. While the special nature of genetic information is highlighted throughout this thesis, it is important at this stage to be aware of the following points.

Genetic information consists of more than just DNA sequences – it also includes the intimate and personal facts about an individual. This includes:

(1) information that determines identity or biological relationship, and,
(2) information that has diagnostic or predictive value about one's health or the health of one's actual or potential children.[26]

In this sense, genes contain much that is relevant to our past and our future, including an individual's faults, susceptibilities and weaknesses. Prior to testing, such information may have been unknown to the individual, unknown to their relatives or employers, and may be information the individual wishes to remain secret. Genetic data can be further classed under one or more of five general categories:

(1) an individual will develop a specific disease, such as Huntington's disease,
(2) an individual has a conditions that will cause a disease (unless treated), e.g. Phenylketonuria,
(3) an individual has a trait which will increase his or her probability of disease,
(4) an individual has an increased probability of disease if exposed to specific environmental factors,
(5) an individual carries a heritable trait, such as Tay Sachs, which can be passed to offspring.[27]

One commentator, Sonya Suter, argues that these various levels of genetic variation share common features which distinguish them from other diseases. For instance, significant differences between genetic and contagious disease are that genetic diseases are only inherited, and 'transmitted' vertically from generation to generation, while contagious diseases are spread horizontally, relying upon contact, not genetic relationships, for their transmission.[28] Further important characteristics of a genetic condition are:

(1) it is caused by no fault of the individual (although it may be accelerated by individual behaviour),
(2) the handicap (if any) does not always appear immediately,
(3) the condition may be alleviated by medical or other assistance,
(4) the effect can be avoided in some cases by environmental change.[29]

Such a list suggests all genetic disease involve a combination of choice and fate. Yet some believe that choice and fate play fundamental roles in all medical conditions and, therefore, genetic conditions are no different. Suter also suggests genetic information may affect one's self identity, with the knowledge or assumption that one carries a certain disease gene, altering

that individual's self-perception. People who believe they have a chance of carrying a particular gene (based on knowledge of family history), develop notions about themselves and their role in the world based upon these assumptions. For instance, a person who believes they are likely to carry the HD gene may choose to live their life based on this presumption. Many may become troubled by test results that contradict such assumptions and consequently challenge established perceptions of their life.[30]

3.2 Other Medical Information

While some disorders are purely genetically derived, the discovery of many multifactorial disorders has elevated the importance of environmental factors in the general field of genetic diseases. Concurrently the discovery of conditions previously thought of as non-genetic disorders (such as hypercholesterolemia, hypertension and osteoporosis), but which in fact have demonstrable genetic components, is escalating. Hence the distinction between genetic and non-genetic disorders appears increasingly blurred, and indeed it has become widely recognised in the scientific world that it is inadvisable to perpetuate the distinction.[31]

Given this thinking, is there any reason why genetic information should be subject to greater protection than other kinds of medical information? Rothstein suggests there may be three reasons for the distinction, all of them more social them medical. First, there is an air of inevitability to genetic disorders, and individuals are often regarded as helpless to prevent its occurrence.[32] In this sense genetic disorders are 'sticky' – they are permanent and while signs or symptoms may be controlled the underlying defect is always there.[33] However it may be argued such permanence is not unique to genetic conditions, and occurs in many other conditions, particularly viral conditions such as herpes and HIV.

Second, the stigma attached to genetic disorders relates to heritability, and consequently there exists a risk of passing it to offspring.[34] This transgenerational aspect makes it especially onerous because it may be viewed as a flaw in one's ancestors and a cloud hanging over progeny in generations to come.[35] Such ramifications force healthcare providers to consider the implications of diagnosis and treatment on siblings, parents and children. Again, however, such problems arise in other conditions: for example, HIV where sexual partners and progeny may also be affected or infected.

Thirdly, many genetic traits fall along racial and ethnic lines. The potential for misuse of genetic information for eugenic purposes is itself enough to compel restrictions on unrestrained discovery of genetic information or compulsory genetic testing.[36] Rothstein concludes that such reasons reinforce the notion that it is not genetic etiology of disorders, but the reactions of other people, including family members and third parties, that causes the deleterious social consequences.

There are perhaps two further qualities of genetic information which are difficult to draw comparisons to other health-related information. One is the ability to predict conditions many years before they become symptomatic. This, it seems, is difficult to compare with non-genetically tested disease. One might suggest, however, that a weak analogy to presymptomatic genetic testing is the detection of carcinoma in-situ as a pre-cancerous growth in the cervical screening programmes.

The second quality unique to genetic information is its implications for 'untested' relatives of tested individuals. A condition 'found' in the tested individuals can sometimes be presumed by medical professionals, employers or insurers, to be present in an 'untested' relative.[37] Such medical and social stigmatisation has created numerous moral, ethical, legal and psychological dilemmas for which there are no easy solutions.[38]

3.3 Probabilities in the Lottery

Everyone possesses a number of genes that may or may not be expressed phenotypically. Where more than one gene is involved, or is environmentally dependent for triggers (i.e. multifactorial), genetic information may do no more than identify one factor of many. In this context, while genetic information may often convey only a 'probability an individual will display a particular characteristic or develop a particular condition; it cannot predict with certainty that this will happen'.[39]

This introduces difficult uncertainties not only into the lives of those tested,[40] but also on the ability for others to use such information with confidence. Some genetic flaws – predominantly single gene defects – indicate a condition will definitely develop (e.g. HD), however many other genetic disorders can only suggest an increased susceptibility to certain medical conditions (e.g. the BRCA genes in determining breast cancer). This difference is important, and highlights the need for definite distinctions between genetic *determination* and genetic *predisposition*.

In cases of complex causation which lack clear contributing genetic and environmental factors, one cannot be sure how particular genes increase the risk of developing the condition. Therefore, genetic information is dependent in large measures on interpretation.[41] In order to ensure that correct interpretations are conveyed, many believe a better understanding of genetic conditions is helped by the use of appropriate language.

3.4 Genetic Nomenclature

How one makes reference to a genetic condition can create confusion. As previously mentioned, the fact that an individual has a genetic condition different from the (supposed) norm, does not necessarily correlate with predispositions or disease. However, to group all genetic variations together using negative terms (e.g. 'disorders', 'mutations', 'flaws', and 'defects'), presumes an individual is unacceptably different. Such terms also implicate the variation as being associated with those who are less than healthy.

While in many cases this may be true (e.g. most Downs syndrome individuals develop heart conditions), many other genetic variations do not correlate with medical problems. For instance, the HD gene will not make an individual any less healthy during their first forty or so years of life. It is only after onset that one's health will deteriorate. Until then, an HD gene carrier would appear to have no greater medical susceptibility than any other individual. As Boyle points out, appropriate language is important, and it is imperative that care be taken. Furthermore to continue '[u]sing words such as *normal* – and its corollary, *abnormal* – is likely to foster stigmatisation and discrimination.'[42]

The risk of medicalising a previously non-medical genetic variation because

of how society perceives a particular trait also poses concern. For example the gene(s) for 'shortness' or 'fatness' (if they exist) may, based upon society's demands, be considered undesirable traits. Once labelled as 'undesirable' it may become as much a medical condition as more severe genetic conditions. Such misnomers may even cause individuals to select against these traits during prenatal screening. Alternatively individuals may seek to 'remedy' these perceived medical conditions through gene therapy.

The Australian Privacy Commissioner notes a distinction must be made 'between genetic makeup of the individual and the way that makeup manifests itself',[43] How one describes such genetic traits should neither unfairly label nor stigmatise. The Privacy Commissioner, therefore, uses 'characteristics' and 'condition' which 'should not be taken as implying anything about whether a particular characteristic of condition is commonly regarded as desirable or otherwise'.[44] While such semantics can only aid in the understanding of genetic information, it is likely to be extremely difficult to maintain.

3.5 Using Genetic Information

A broad overview suggests that there are two main uses of genetic information: these being first for medical, therapeuticor healthcare purposes, and second, for non-medical or non-therapeutic purposes. In later chapters I deal with both of these uses.

The medical uses include diagnosis, reproductive planning, disease prevention, treatment and research. These applications raise numerous legal and ethical issues, including informed consent, privacy, confidentiality, duty to warn, public health screening, and medical malpractice.[45] Of particular importance is the dissemination of the information to the individual tested. The desire, or conversely lack of desire, to know one's genetic makeup is not limited to the patient, but may extend to all those who may have inherited similar genetic material.

However, many argue that the risks involved with foresight into one's future health prospects are yet to be realised. As yet, there are few cures available for genetic conditions, and consequently genetic knowledge will often be unlikely to save lives. Instead genetic testing, and subsequent revelation of genetic disorders, may cause people to refrain from having normal relationships, bearing children, or suffering ongoing depression, advancing arguments that its use should be restricted. Others, however, argue that genetic information can free individuals of the burden of thinking they have inherited a particular genetic trait, and provide some form of relief.

Genetic information may also be of interest to public health authorities (for statistical purposes), insurers, employers, schools, child welfare agencies, adoption agencies, law enforcement officials, and the armed services. Such application has caused worldwide concern about the ability of existing law and policy to regulate non-clinical uses of genetic data, in particular the underwriting of insurers, and the hiring practices of employers.[46]

In both contexts, the broad range of use makes it extremely difficult to formulate any universally applicable principles. However, certain themes do reoccur, notably conflicts between individuals and third parties over control of genetic information, balancing the potential benefits against the cost and consequences of obtaining it, and placing into perspective the evolutionary, psychological, and social power of genetic information.[47]

Such issues are tackled on three fronts in this thesis: first the innately private relationship that exists between doctor and patient and the pressures upon it when identifiable others may be at risk; second the privacy of information in the public realm when society, institutions, or third parties may benefit; and finally, the potential for genetic discrimination of individuals, groups, or races.

The references for this article and copies of Hamish's thesis are held in the Bioethics Research Centre and the Sir Robert Stout Law Library at the University of Otago. Hamish can be contacted at the office of the Health and Disability Commissioner, PO Box 12 299, Wellington.

Open Letter

15 May 1998

The use of stored body parts or bodily substances for research purposes without informed consent

It has recently come to my attention that routinely stored pathology samples may be being used for research without the consent of the consumers involved on the belief that the sample is 'abandoned'. I would, therefore, like to take this opportunity to clarify provider obligations under the Code of Health and Disability Services Consumers' Rights when conducting research on body parts or bodily substances.

While I am able to make general comments and indicate those provisions of the Code which appear to be particularly relevant, I am not able to give advance rulings on interpretation and application of the Code. The complaints jurisdiction under the Health and Disability Commissioner Act requires investigations to be undertaken impartially and with an open mind. This would be open to challenge if I had already said that the practice under investigation was or was not a breach of the Code. However, the following general comments may be useful.

Right 9 of the Code makes it clear that the rights in the Code extend to those occasions when a consumer is participating in, or it is proposed that a consumer participate in, research. In particular, for present purposes, Rights 7(9) and 7(10) set out consumers' rights in respect of decisions about body parts or substances:

(9) Every consumer has the right to make a decision about the return or disposal of any body parts or bodily substances removed or obtained in the course of a health care procedure.

(10) Any body parts or bodily substances removed or obtained in the course of a health care procedure may be stored, preserved, or utilised only with the informed consent of the consumer.

Right 7(6) requires informed consent