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Regulating genetic privacy in the online health information era

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Abstract

As the clinical implications of the genetic components of disease come to be better understood, there is likely to be a significant increase in the volume of genetic information held within clinical records. As patient health care records, in turn, come on-line as part of broader health information networks, there is likely to be considerable pressure in favour of special laws protecting genetic privacy. This paper reviews some of the privacy challenges posed by electronic health records, some government initiatives in this area, and notes the impact that developments in genetic testing will have upon the 'genetic content' of e-health records. Despite the sensitivity of genetic information, the paper argues against a policy of 'genetic exceptionalism', and its implications for genetic privacy laws.

Key words: genetic information; genetic testing; health law; health information; electronic health records; e-health

Introduction

The likely future increase in the volume of genetic information within clinical medicine and clinical information systems raises important concerns about privacy. These concerns will be amplified as clinical records increasingly come 'on-line' as part of broader health information networks. Here, as in other contexts where genetic information will be generated and stored, regulators and legislatures face the question of whether we should have special laws to respond to the particular sensitivities of genetic testing information ([Australian Law Reform Commission and Australian Health Ethics Committee](#); hereafter 'ALRC and AHEC', 2001).

This paper will review some current government initiatives in the area of electronic health records, and some of the privacy challenges that arise, while also noting the impact that developments in genetic testing will have upon the volume of genetic information within clinical medicine and patient health records. While genetics itself is not new ([Chalmers 1999](#), pp.204-205), the scale, speed and sophistication of gene mapping and research into the genetic determinants of health certainly are. As the clinical implications of the genetic components of disease come to be better understood, the volume of genetic testing information within clinical databases will increase significantly. Despite these trends, or perhaps *because of them*, I argue in this paper against a policy of 'genetic exceptionalism', and its implications for genetics-specific privacy laws. The electronic storage of genetic health information, like other categories of sensitive health

information, poses important risks to privacy. However, the privacy issues that are unique to genetic information are not specific to on-line information.

What is genetic health information?

Genetic health information can be understood as information relating to an individual's inherited, genetic make-up, which has implications for the health of the individual, and potentially his or her genetic relatives, either now or (with differing degrees of certainty) in the future. Genetic health information can be generated or revealed in several ways, including:

- through DNA testing that identifies the individual as a carrier of particular genes or gene alleles (an allele is a specific form of a gene ([Korf 1996, p.343](#)));
- through the clinical diagnosis of genetic disorders or abnormalities, either apparent or latent; and
- by inference from a person's family medical history (in those circumstances where inferences can properly be drawn).

It is clear that while tissue samples provide 'vectors' for accessing an individual's DNA and genetic information, DNA testing is only one way of generating genetic health information.

Is genetic health information special?

Does genetic information warrant special privacy laws that are stricter than those that apply to health information generally? The case for 'genetic exceptionalism', in privacy terms, usually revolves around several inter-related claims. The first is that genetic information is especially sensitive because of its own subject matter and intrinsic qualities. Genetic information is, for example, unchanging and (to differing degrees) predictive in character, qualities which mean that it impacts heavily upon one's sense of personal identity. Rightly or wrongly, such information 'is widely viewed...as saying something about who the person is, not simply what he or she has done, encountered, or eaten' ([Karjala 1992, p.164](#)). Annas, Glantz and Roche refer to a person's DNA as a 'coded probabilistic future diary'. 'Most of the code cannot now be broken, but parts are being deciphered almost daily' ([Annas, Glantz and Roche 1994, p. 360](#)).

The second claim is that genetic information is particularly sensitive because it has implications for the health of genetic relatives and genetically related communities. While the shared character of genetic information may enhance its sensitivity from the collective perspective of those who share certain genes, the limits of a person's claim to privacy, in circumstances where a third party is at risk of harm through the transmission of 'harmful genes', is more controversial. A good deal of debate in the genetic privacy literature has revolved around the conflict between a duty of confidence owed to an index case and perceived ethical duties to disclose genetic data to third parties ([Skene 1998](#); [Bell and Bennett 2001](#)). In other contexts, the contest is between a person who wishes *not* to know genetic information about themselves, and

third parties who do. The latter situation has arisen, for example, where a person at risk of carrying the Huntington's disease gene is locked in dispute with a pregnant sexual partner who desires foetal testing.

The third claim supporting genetic exceptionalism is that community responses to genetic information create a heightened risk of information-based harm. The 'social construction' of genetic information is such that disclosure may lead to a 'spoiled identity'. In addition, the predictive quality of genetic information means that test data is of special interest to insurers and employers, and this has increased concern about discrimination and loss of privacy.

Opponents of 'genetic exceptionalism' have disputed each of these claims. Murray argues that genetic information is 'neither unique nor distinctive in its ability to offer probabilistic peeks into our future health'. Asymptomatic hepatitis B, HIV status and cholesterol level, he argues, are as cogent and sensitive in their implications for future health as information about genetic predispositions and susceptibilities ([Murray 1997, p.64](#)). Similarly, Gostin argues:

Arguments that genomic data deserve special protection must reckon with the fact that other health conditions raise similar sensitivity issues (for examples, HIV infection, tuberculosis, STDs, and mental illnesses)... Some patients may be just as sensitive about prevalent nongenetic or multifactorial diseases like cancer and heart disease as they are about diseases with a unique genetic component ([Gostin 1995, p.326](#)).

Genetic exceptionalism has a prominent precedent in HIV/AIDS, a life-threatening infectious condition that emerged in an environment of widespread ignorance and prejudice. Gradually, over a period of about ten years, Australian legislatures in most jurisdictions enacted a body of AIDS-specific laws ([Magnusson 1996](#); [Godwin et al. 1993](#)). On the other hand, there is nothing discrete or self-contained about genetic information as a form of health information. As clinical genetics continues to develop, any attempt to quarantine genetic health data from other forms of health information will quickly become nonsensical. A 'genetics-specific' approach to health privacy is likely to lead to additional layers of legal complexity (and health privacy law is already complex enough) while constituting a major irritant to health providers themselves.

Daniel Callahan has argued that modern medicine has come to be influenced by two competing approaches to the reduction and elimination of disease. The first approach seeks to influence health status from the 'outside' by targeting 'the way we live our lives', and 'changing those hazardous health behaviours known to be major causes of illness and mortality'. The second approach seeks to improve health from the 'inside', by looking to 'the genetic bases of disease and mortality and to change and manipulate them to improve our health' ([Callahan 1996, p.17](#)). Rather pessimistically, Callahan fears that the

first approach may lead to a coercive, 'health fascism', while the second approach may translate into a 'fierce, unforgiving, and uncompromising use of genetic knowledge to perfect the human condition' ([Callahan 1996, p.18](#)).

Whatever interpretation we may place on them, Callahan's 'inside' and 'outside' approaches provide useful labels for two important trends in clinical care. Callahan's 'inside' approach usefully directs attention to the central future role that genetic health data will have in shaping and 'individualising' clinical care. The 'outside', or external operating environment, of clinical care, on the other hand, is in the early stages of a fundamental transition from episode-based care towards a more holistic, co-ordinated model that encompasses health surveillance systems and the strategic use of health data within on-line networks. An understanding of trends in both the 'internal' and 'external' aspects of clinical care is important to an appreciation of the nature and scale of the privacy risks posed by on-line health information systems. Nevertheless, while the on-line storage of genetic data poses some important risks to privacy, genetics-specific laws are not the answer.

Surveillance and continuity: The future health care record

Traditionally, medical care focused on patients' immediate illnesses and not much else. Medical interventions (and their underlying assumptions) were reactive rather than pro-active, fragmented rather than coordinated, and reductionist rather than holistic ([Harper, Holman and Dawes 1994, pp.227-230](#)). Increasingly, however, government initiatives are moving towards an integrated electronic health record, which aims to improve the quality and continuity of care by linking all health providers involved in the patient's care, thereby facilitating the sharing of up-to-date information. In addition to cost reductions and efficiency gains ([Commonwealth of Australia 2000](#)), electronic health records will facilitate 'real time' public health surveillance, and a closer alignment between personal health care and public health functions ([Commonwealth of Australia 2000, pp.69, 97-98](#); [Douglas 2001](#)). Supported by data from population health studies, and concerted efforts to integrate the clinical implications of health-outcomes data, the aim is to make clinical care more holistic, more interactive, more evidence-based, more attuned to the demographic and social determinants of health, and more closely aligned with individual needs.

This paper can provide only the briefest review of current progress towards this emerging clinical care environment. Electronic health records have existed for a number of years and are now commonly seen in hospitals and, increasingly, general practice. What *is* new is the push towards integrated on-line health information networks that will facilitate the capturing of longitudinal health data on patients, including episodic summary data from all the patient's interactions with the health care system (the so-called 'cradle-to-grave' e-health record).

National initiatives

In Australia, progress towards health information networks is occurring at both federal and State level. At the national level there are three initiatives for the development of on-line

health records:

- HealthConnect;
- The Better Medication Management System (BMMS);
- The Health eSignature Authority.

HealthConnect is an evolving framework for a coordinated national network of electronic patient health care records. While still in the conceptual stages, trial sites are expected to be operational in late 2002.^[1] HealthConnect would consist of episodic event summaries, containing health and demographic data, collected at point of care and stored in a distributed manner ([Commonwealth of Australia 2000, p.120](#)). Hospital and pathology event summaries, for example, might be held on-line at the relevant hospital, while general practitioner records might be stored at regional level. Such data would be available to authorised health care providers and could be assembled in different ways.

The BMMS, an electronic patient *medication record*, would operate within the framework of HealthConnect, linking prescription information written by different doctors with dispensing pharmacists. It is anticipated that such a system could reduce adverse drug events,^[2] and, no doubt, fraud.^[3] The Health eSignature Authority is a registration authority established and owned by the Health Insurance Commission ([Health Insurance Commission, undated](#)). In its April 2000 report, the National Electronic Health Records Taskforce noted that the transfer of data between sites, in order to generate an integrated electronic patient record, will be secured by encryption, within a national Public Key Infrastructure ([Commonwealth of Australia 2000, pp.121-122, 138-139](#)). The anticipated role of the Health eSignature Authority would include authenticating the identities of participants in HealthConnect and issuing digital certificates. By binding a person's location or identity to their digital keys, digital certificates will ensure the secure transmission of data over electronic health networks.^[4] Secure health data transmission will facilitate a variety of administrative and clinical functions,^[5] including the collection of data stored in various locations (within a distributed data storage network) in order to generate an integrated patient record.

Victorian initiatives

Initiatives for on-line health records are also progressing at State level. In Victoria, it is anticipated that electronic records will develop as an integral part of the 'primary care partnerships' strategy of the Department of Health and Human Services. The strategy aims to improve health outcomes in Victoria, and to reduce the preventable use of hospital services through the creation of partnerships of primary care agencies encompassing GPs, community nurses, physiotherapists, psychiatric disability support, aged care assessment, dental health, drug treatment services and women's health ([Department of Human Services \(Victoria\) 2000](#)). Locally based primary care partnerships ('PCPs') will identify the population health needs and priorities of their communities (through the development of Community Health Plans), and coordinate the services they provide through 'integrated,

multi-sectoral health promotion and disease management programs and services' ([Department of Human Services \(Victoria\) 2001a, p.4](#)). An important feature of these partnerships is the convergence of primary clinical care and public health functions (illness prevention and health promotion). State-wide infrastructures linking locally based PCPs are envisaged, as well as links to national structures ([Department of Human Services \(Victoria\) 2001b, pp. v, 21-22](#)).

The aspiration for 'seamless pathways' of clinical care requires a sophisticated information management capacity encompassing the on-line collection and sharing of patients' health information between service providers ([Department of Human Services \(Victoria\) 2001b, pp.5-6](#)). The information management strategy for PCPs envisages that over the next five years, electronic health records will be implemented within each partnership, with the primary role of assisting the 'coordination of the interactions between consumers and providers' ([Department of Human Services \(Victoria\) 2001b, p.31](#)).^[6] Such records would act as a 'centrally held master file' available to authorised staff, who could view consumer information and download it into their own systems. In the absence of a national approach to patient identification, PCPs would use demographic data to identify clients, rather than a unique patient identifier ([Department of Human Services \(Victoria\) 2001b, p.33](#)). As with other proposals, data security and authentication would depend upon a public key infrastructure ([Department of Human Services \(Victoria\) 2001b, pp.33-35](#)).

NSW initiatives

In contrast to the Victorian primary care-based initiative, plans for an electronic health record (EHR) in New South Wales appear to be somewhat more ambitious. In its March 2000 report, the NSW Health Council recommended the introduction of an EHR by 2010, supported by a unique patient identifier to facilitate linkage of patient records across health facilities ([Department of Health \(NSW\) 2000a, pp.21-29](#)). 'EHR*NET' would link hospitals, general practice, and other primary care systems, providing 'an event summary comprising client demographics, clinical information, outcomes of care and an ongoing management plan' ([Ayres, Horvath and Kidd 2001, p.13](#)). By June 2003, the unique patient identifier (UPI) would be implemented at the Area Health Authority level, with a transition towards a State-wide identifier thereafter.

EHR*NET is intended to comprise several inter-related systems. The first component of the system, scheduled for introduction by June 2004, will be the Patient Administration System (PAS), which would facilitate administrative information exchange within hospitals. This will be complemented at the community level by CHIME, a case-management information system for community-based health workers (scheduled for implementation progressively between 2001-2004). Other elements of the system include the electronic transmission of hospital discharge summaries to GPs, Electronic Prescribing Decision Support (E-PDS), and a Point-of-Care Clinical System (PoCCS), a mobile information resource that captures the latest patient information and can

be linked to evidence-based databases and clinical decision-support tools.^[7]

“Multi-function” health records

The forces fuelling the development of these and other on-line health information systems are complex. They include budget imperatives and the need to use resources in the health care sector more efficiently.^[8] Better health information systems will also facilitate health resource allocation through the monitoring of trends in health status, and the evaluation of health services and programs in terms of health outcomes. Although on-line health information systems, coupled with programs aimed at improving continuity of care (‘seamless care’), are expected to result in improved clinical outcomes for patients, there is also growing interest in ‘multi-function’ electronic record systems whose purpose extends beyond clinical care to include epidemiological and clinical outcomes research, quality assurance, audit and cost-monitoring functions ([Mount, Kelman, Smith et al. 2000](#)).^[9]

Population health researchers have long argued that public health surveillance should enjoy equal status with clinical care, thereby permitting the electronic linkage of clinical data with other government data sources for public health purposes. Douglas argues that:

The personal electronic health record, however it is stored and accessed, should also be the building block for ‘real time’ public health surveillance. Improved efficiency of personal clinical care and improved management of public health both require the same data and should become two sides of the one coin. When a patient presents for care by a general practitioner for a respiratory infection or a manifestation of HIV, that information should automatically become part of [a] national public health monitoring activity. And when the laboratory reports to the GP that the respiratory infection is, or is not, a new strain of influenza that fact should, as well as informing the clinician instantly, feed into a national database that informs public health action ([Douglas 2001, p.242](#); see also [Hammond, Pollard and Straube 1998, pp.28-29](#); [Dever 1997, p.97](#)).

This tension between public health and clinical health is heightened by the influence of ‘the new public health’ in health information policy, and, inevitably, the design of health information networks. Goraya and Scambler point out that what is ‘new’ in the ‘new public health’ is ‘the rediscovery that influences on health are multifaceted and include economic, environmental, ecological, political and behavioural components, as well as the provision of, and equitable access to, medical services’ ([Goraya and Scambler 1998, p.144](#); see also [Baum 1998](#)). In contrast to the role specialisation that characterises clinical care, there is growing realisation that health problems are ‘characterised by interdependence with one another, and by interdependence with life style and environment’ ([Harper, Holman and Dawes 1994, p.227](#)).

Richardson notes that, under the influence of the WHO definition of health,^[10] 'there has been a transition in the conceptualisation of health from a clinical orientation to a greater focus upon the patient in a social context' (Richardson 1998, p.251). It seems likely, therefore, that over time the 'new public health' will have a significant impact on the design and functioning of any integrated health information network. In particular, there will be strong pressure for an architecture that permits linkages between clinical data, disease registers and other government health databases, demographic data, environmental surveillance data, and indices of socio-economic advantage. Anything less would fail to capture the significance of the social, lifestyle and environmental factors that – with the decline of infectious agents as major causes of mortality and morbidity – are key contributors to today's non-communicable, 'degenerative diseases' (Hetzel 2001). Kirby points out that in population health informatics, 'the focus is on surveillance of the health status of entire populations, rather than simply on those who access services' (Kirby 1999, p.S18). In addition, 'population-based health information systems are integrated with a variety of external data sources to provide a comprehensive perspective on the determinants of population health' (Kirby 1999, p.S18).

There is an inevitable tension, therefore, between the development of electronic health records for clinical purposes and the application of these systems for public health purposes. This tension carries with it the potential for surveillance to a degree that many privacy advocates themselves may not appreciate. The appetite for population health and 'outcomes' measurements represents an important privacy issue within on-line health information systems, especially as those systems begin to store genetic health information. At present, the 'release mechanisms' that permit clinical data to be used for the purposes of health research, in the absence of patient consent, are generally restrictive, although far from uniform across jurisdictions and sectors (Magnusson 2002, p.21-38). On the other hand, the use of clinical data for billing, service monitoring, complaints handling, evaluation and accreditation, quality assurance, audit and medico-legal purposes is generally recognised as being within the reasonable expectations of patients themselves. In privacy terms, these purposes are regarded as 'directly related to the primary purpose of collection', thus dispensing with any requirement for specific patient consent.^[11] In general terms, therefore, evolving e-health data networks are likely to serve a variety of purposes, subject to requirements in privacy legislation that restrict the use of data for what are considered to be 'secondary purposes'.^[12] It is certainly appropriate to scrutinise current privacy laws to see if they are capable of adequately protecting genetic health information within an on-line environment. However, the privacy risks that arise will rarely be unique to genetic health information.

Genetic testing and the 'individualisation' of clinical care

I have argued above that the 'external' operating environment of clinical care is undergoing a process of fundamental change. But Callahan is also right to recognise the impact of 'internal' changes, or the influence of genetics, upon clinical care.

Genetic tests may fulfil a number of health-related functions. They can confirm the presence of a specific disorder in a person who presents with symptoms (diagnostic testing); identify genetic mutations that will (in the case of adult-onset disorders) or may (in the context of a multi-factorial disease) result in future illness (predictive or pre-symptomatic testing); and determine whether the individual carries genetic mutations that may increase the risk that genetic offspring will inherit the disorder (carrier testing) ([NHMRC 2000, pp.15-19](#)). As genetic tests become available for a wider variety of diseases, and as the health implications of various genetic factors become clearer, health care services are likely to respond pro-actively, with a greater focus on health monitoring, and service provision that is responsive to individual genetic circumstances.

At present, most genetic testing in Australia is carried out, following referral by doctors, in public hospital laboratories funded by the States and Territories ([ALRC and AHEC 2001, p.256](#)). Historically, different laboratories have specialised in particular tests in order to minimise the duplication of services, and to maintain proficiency of diagnosis and interpretation (given the low prevalence of individual genetic disorders).^[13] However, tests are becoming available for an increasing number of specific disorders.^[14] The identification of genes associated with breast and colon cancer has played a part in shifting the emphasis of clinical genetics services from its traditional paediatric-obstetric focus to include adult medicine and psychiatry ([Department of Human Services \(Victoria\) 1996, para 1.2](#)). Genetic testing is gradually becoming part of the clinical practice of neurologists, oncologists, gastroenterologists and general practitioners ([Department of Health \(NSW\) 2000b, para 2.3](#); [Magnusson 1995, p.215](#)).

In Australia, genetic testing is not regulated by legislation, although in some cases guidelines have been developed by bodies such as the Human Genetics Society of Australia.^[15] The New South Wales Department of Health acknowledges that the range of available tests is increasing, but cautions that testing should be medically indicated, whether by positive family history, high risk or clinical indication.^[16]

Professor David Danks, an Australian pioneer in genetic research, has suggested that a number of trends in genetic testing are likely in future.^[17] First, there is likely to be a gradual increase in pre-natal testing of maternal serum, followed up by amniocentesis, for genetic diseases such as spina bifida. Such testing permits couples to make more informed choices relating to abortion, foetal treatment, the disabilities of the foetus and options for post-natal treatment. Testing for genetic diseases is particularly efficient within families, since genetic problems run in families. Similarly, there may also be an increase in pre-marital carrier testing for diseases such as cystic fibrosis: overall, one in 25 individuals in NSW is a carrier of a mutant CF gene ([Department of Health \(NSW\) 2000b, para 2.1](#)).

Secondly, as genes associated with cancer and other diseases are identified, recently diagnosed persons are likely to undergo

genetic testing to determine if their cancer is one of the inherited forms. Given the cost of population screening, this is an efficient way of identifying carriers and those at risk of disease. For example, of the 2,000-odd cases of colon cancer diagnosed each year in Victoria, genetic testing would identify perhaps 200 with the colon cancer gene mutation. If siblings of the index case were tested, each would have a 50% chance of having that gene. This means that only two persons would need to be tested to identify one with the gene. Unlike population screening, this ensures the efficient use of counselling resources, and minimises false positives.

If the trends identified by Danks continue, it seems inevitable that clinical genetics services will gradually release their de-facto monopoly over genetic testing, counselling, diagnosis and clinical follow-up. Genetic medicine will become an integral part of primary care ([Emery and Hayflick 2001](#)). Hand in hand with this, genetic testing information will become commonplace: a familiar component of clinical records, encompassing an individual's entire lifespan (prenatal and newborn screening,^[18] pre-marital screening, with diagnostic, carrier and predictive testing where indicated). Anticipated developments in nanotechnology leading to the miniaturisation of genetic testing may permit rapid, point-of-care testing, with implications for the determination of drug dosages ([Trent 2002](#)). As clinical records themselves go on-line, therefore, an increasing volume of genetic information will be stored in digital form within integrated electronic health records systems. As customised therapies become possible, such information will have a vital clinical role.

This is not to deny, of course, the ambiguity of, and contingencies associated with, genetic test information. For example, the sensitivity of tests for heterogeneous diseases, such as cystic fibrosis, which can be caused by over 700 different mutations ([NHMRC 2000, p 21](#)), will be affected by whether or not the test covers all the disease alleles, or by whether geneticists know which mutation to test for.^[19] Testing for diseases that are polygenic (involving complex interactions between many genes) or multi-factorial (involving complex interactions between genes and the environment) may only reveal weak, probabilistic information ([ALRC and AHEC 2001, pp.77-78](#)). Up to 98% of human disorders with a genetic component may involve genes that increase an individual's susceptibility to disease in accordance with pathways that are complex and currently unknown ([NHMRC 2000, p.20](#)). Carrier screening performed in a population in which the incidence of the gene is low will yield a high proportion of false positives ([Gostin 1995, p.323](#)). The clinical phenotype for a genetic illness, including onset date, severity of symptoms and susceptibility to treatment, will also depend upon the degree of penetrance ([ALRC and AHEC 2001, pp.85-86; Gostin 1995, p.323](#)).

The fact that genetic test results can only suggest probabilities, susceptibilities or ill-defined risks, together with the psychological impact of test results upon individuals who do not wish to know, may act as a brake on the integration of some genetic test data into individualised health care plans ([Department of Human Services \(Victoria\) 1996, para 3.1](#)). On

the other hand, there will be significant, ongoing research into the genetic determinants of those chronic and degenerative diseases that increasingly account for morbidity and mortality, including cancer, heart disease, hypertension, dementia, diabetes and schizophrenia. As the role of genes within disease pathways becomes better understood, public perceptions about the value of genetic tests for these illnesses may also alter accordingly. Testing for genetic susceptibilities for multi-factorial diseases will remain sensitive, but may well be seen as less threatening than testing for monogenic diseases with high penetrance, such as Huntington's disease ([Wexler 1992, pp.211-243](#)).

Genetic testing is expensive and may well *not* be implemented on a population basis. On the other hand, just as economic factors are partly fuelling the trend towards integrated electronic records, so they will encourage the uptake of genetic testing within clinical care, in circumstances where interventions are available that will (or may) lead to better (and cheaper) health outcomes. Subject to the caveats above, therefore, genetic information is likely to become, in future, an integral part of each person's individualised health care program. Genetic information will be 'smeared across', so to speak, the individual's clinical record as a relevant component of health status in many areas. Whether or not this information will be regarded as especially sensitive will depend, just like non-genetic information, on what it is. The future role of genetic information within clinical care provides, therefore, an important, pragmatic argument against having special privacy laws for genetic information. Ultimately, there will be little point in seeking to compartmentalise and quarantine genetic health information behind additional privacy or security barriers. A generic solution to the privacy challenges of all on-line health information is preferable.

Protecting health privacy in an on-line environment

This paper cannot provide a detailed review of the privacy issues raised by electronic health records. In this section, I will merely seek to identify what I see as the most important issues, with implications for genetic and non-genetic data alike.

Health privacy law is complex and is best conceptualised in several 'layers' ([Magnusson 2002, pp.6-7](#)). Doctors and other health professionals have long been subject to a legal duty of confidentiality owed with respect to patient information, based on equitable and contractual principles and recognised in case law.^[20] Secondly, confidentiality has a statutory dimension, since most jurisdictions have non-disclosure provisions in health services statutes, and public health statutes.^[21] In addition, there are a variety of HIV-specific provisions.^[22] Thirdly, legislatures are progressively enacting privacy legislation. At the Commonwealth level, information privacy principles having statutory force apply to Commonwealth agencies^[23] and, since 21 December 2001, to private sector 'organisations'.^[24] At the State and Territory level, generic privacy legislation applies to public sector agencies in NSW,^[25] and to both public and private sector organisations in

Victoria.^[26] The trend towards health-specific privacy legislation is reflected in legislation applying to both public and private sectors in Victoria and the ACT (NSW is expected to follow).^[27]

The development of on-line health systems has precipitated a fourth layer of regulation. At the Commonwealth level, the protection of health data within the HealthConnect network will exist at three levels.^[28] The recently enacted 'National Privacy Principles' will apply to private sector organisations participating in HealthConnect. Secondly, a 'National Health Privacy Code' is under development that would provide a nationally consistent set of rules covering the private and public sectors. There is also an anticipated third level of legislation applicable, specifically, to e-health initiatives.^[29]

The impact of these layers of regulation is uneven across both jurisdictions and sectors. In some jurisdictions (eg, Victoria) closely related State and Commonwealth regimes exist simultaneously, while in others (eg, Tasmania) only the 'National Privacy Principles' apply. There is a screaming need for a consistent, national approach to wind back the 'balkanisation' of Australian privacy law.^[30]

Digital storage poses many privacy risks to health records, not the least of which is the *scale* of risk. Digital storage within a network creates more points of access to data, while, in comparison to an act of intrusion into a paper registry, one unauthorised 'hack' into a computer database, or one error by a data administrator, may compromise the records of a much larger number of people, as well as *more data* relating to each individual (Carter 1999).^[31]

The heartland of privacy protection is the distinction – found in statutory privacy principles – between the 'primary' purpose(s) for which the information can be collected or generated (eg, clinical care) and other, 'secondary' purposes for which it could potentially be used and disclosed (eg, research). When combined with audit trails, these restrictions upon use and disclosure for 'secondary purposes' provide a framework for protecting against both 'function creep' (the gradual use of information for additional, 'secondary purposes'), and the improper accessing and use of information by authenticated users.^[32] Current government initiatives for electronic health information systems, such as HealthConnect, are being 'sold' to the public on the basis that patients will be able to choose whether their data participates in the network, and to control who has access to it within the network (Commonwealth of Australia 2000, pp.77, 121, 127, 164). Ideally, patient concerns about the privacy of genetic information, and other categories of sensitive information, might be met by a structured, variegated system of protocols for partitioning 'sensitive' categories of data and for controlling when and to whom different defined categories of data could travel, in accordance with consumer perceptions of their relevance and sensitivity. In reality, clinical and administrative requirements are likely to place limits on this 'ideal'. While the architecture of current initiatives remains fluid, one possibility is that patients could choose from a limited, graded 'menu' of options that protect the privacy of defined categories of data in

different ways. Facilitating consumer choice and a degree of control, and balancing it with the demand for 'multi-function' health records, remains a major challenge in the development of on-line systems. An appropriate resolution to this tension is central to the success of on-line health information systems such as HealthConnect, and to the protection of genetic health information on-line.

Conclusion

I have argued in this paper that, while genetic information may be sensitive, the privacy issues raised by the on-line storage of genetic data in a clinical context are risks shared with other categories of sensitive health information. This is not to suggest that *none* of the privacy challenges associated with genetic information are unique or justify 'special' laws. For example, the fact that genetic information is both predictive in character and hence inherently personal, and yet divulges information about genetic kin, may lead to difficult ethical and legal conflicts. Where genetic testing has revealed or may reveal that a sibling, son or daughter faces a substantial and quantifiable risk of serious disease or disability, in circumstances where earlier interventions could alleviate or reduce the harm suffered, there may be strong moral arguments for breaching confidentiality (Leung 2000).^[33] The fact that genetic testing information is sensitive does not dilute its importance to third parties at risk of avoidable harm. Balanced against this are the privacy interests of the index case in protecting his or her intimate information, and public health justifications supporting confidentiality in genetic counselling contexts.

These and other privacy issues, however, are not a function of digital storage. Furthermore, given the current complexity of health privacy law and future trends in clinical medicine, special genetic privacy laws should be carefully considered, lest they inadvertently fragment the information base upon which clinical care increasingly relies. In the on-line context, a generic solution that respects patient concerns about what information is sensitive, and permits patients themselves to determine the level of protection accorded various categories of information, should remain the central priority.

References

Annas G, Glantz L and Roche, P (1994). 'Drafting the Genetic Privacy Act: Science, policy and practical considerations'. *Journal of Law, Medicine and Ethics*; 23: 360-366.

Australian Institute of Health and Welfare (2001). *Health Expenditure Bulletin*, no 17, September 2001, viewed 22 March 2002, <<http://www.aihw.gov.au/publications/hwe/heb17/index.html>>.

Australian Law Reform Commission and Australian Health Ethics Committee (2001). *Protection of human genetic information*. Issues Paper 26, Australian Law Reform Commission and Australian Health Ethics Committee.

Ayres D, Horvath D and Kidd M (2001). 'Information management implementation co-ordination group and the NSW Government's action plan for health'. *HealthCover*, August-September: 11-17.

Baum F (1998). *The new public health: an Australian perspective*. Oxford University Press, Melbourne.

Bell D and Bennett B (2001). 'Genetic secrets and the family'. *Medical Law Review*; 9: 130-161.

Callahan D (1996). 'The genetic revolution'. In: D Thomasma and T Kushner (eds), *Birth to death: science and bioethics*. Cambridge University Press, Cambridge, 13-33.

Carter M (1999). 'Protecting consumers' interests in their health records'. *Australian Health Law Bulletin*, 8(2): 13-16.

Chalmers D (1999). 'The challenges of human genetics'. In: I Freckelton and K Petersen (eds), *Controversies in health law*. The Federation Press, Sydney, NSW, 202-239.

Charlesworth M (1989). *Life, death, genes and ethics: biotechnology and bioethics*, ABC Books, Crows Nest, NSW.

Commonwealth of Australia (2000). *A health information network for Australia: report to health ministers by the National Electronic Health Records Taskforce*. Viewed 21 March 2002 <http://www.health.gov.au/healthonline/ehr_rep.htm>.

Commonwealth of Australia (2000). 'Report to the Electronic Health Records Taskforce: the benefits and difficulties of introducing a national approach to electronic health records in Australia' (appendix to *A health information network for Australia: report to health ministers by the National Electronic Health Records Taskforce*). Viewed 21 March 2002 <http://www.health.gov.au/healthonline/ehr_rep.htm>.

Department of Health (NSW) (2000a). *Report of the NSW Health Council – a better health system for NSW* (the 'Menadue Report'). New South Wales Health Council, Department of Health, Sydney.

Department of Health (NSW) (2000b). *Specialised testing for genetic disorders: Part 1 – recommendations for service provision*. Department of Health (NSW), Sydney.

Department of Health (NSW) (2001). *Guidelines for newborn screening* (Departmental circular 2001/45). Department of Health (NSW), Sydney.

Department of Human Services (Victoria). Public Health Division (1996). *Genetic services in Victoria: a discussion paper*. Viewed 28 March 2002 <<http://www.dhs.vic.gov.au/phb/hdev/genetics/contents.htm>>.

Department of Human Services (Victoria). Aged Community and Mental Health Division (2000). *Going forward: primary care partnerships*. Viewed 21 March 2002 <<http://www.dhs.vic.gov.au/acmh/ph/pcp/forward/index.htm>>.

Department of Human Services (Victoria) (2001a). *Primary care partnerships: better access to services – a policy and operational framework*. Viewed 21 March 2002 <<http://hnb.dhs.vic.gov.au/acmh/phkb.nsf>> (indexed under 'Primary Care Partnerships').

Department of Human Services (Victoria) (2001b). *Primary care partnerships: information management strategic directions*. Viewed 21 March 2002 <<http://hnb.dhs.vic.gov.au/acmh/phkb.nsf>> (indexed under 'Primary Care Partnerships').

Dever G (1997). *Improving outcomes in public health practice: strategy and methods*. Aspen Publishers Inc., Gaithersburg, Maryland.

Douglas R (2001). 'Disease control in the information era'. *Med J Aust*; 174: 241-243.

Emery J and Hayflick S (2001). 'The challenge of integrating genetic medicine into primary care'. *BMJ*; 322: 1027-1030.

Godwin J, Hamblin J, Patterson D and Buchanan D (1993). *Australian HIV/AIDS Legal Guide*, 2nd edition. The Federation Press, Sydney.

Goldman J and Hudson Z (2000). 'Virtually exposed: privacy and e-health'. *Health Affairs*; 19: 140-148.

Goraya A and Scambler G (1998). 'From old to new public health: role tensions and contradictions'. *Critical Public Health*; 8: 141-151.

Gostin L (1995). 'Genetic privacy'. *Journal of Law, Medicine and Ethics*; 23: 320-330.

Hammond W, Pollard D and Straube J (1998). 'Managing healthcare: a view of tomorrow'. In: B Cesnik, A McCray and JR Sherrer (eds), *MedInfo '98 (proceedings of the 9th World Congress on Medical Informatics)*. IOS Press, Amsterdam, 26-30.

Harper A, Holman D and Dawes V (1994). *The health of population*, 2nd edition.

Churchill Livingstone, Melbourne.

Health Insurance Commission, undated. *The HIC and public key infrastructure (PKI)* (information brochure supplied by the Commission).

Hetzl D (2001). 'Death, disease and diversity in Australia, 1951 to 2000'. *Med J Aust*; 174: 21-24.

Karjala DS (1992). 'A legal research agenda for the human genome initiative'. *Jurimetrics Journal*; 32: 121-222.

Kirby R (1999). 'From public health to population health: epidemiological yardsticks for perinatal care'. *J Perinatol*; 19: S16-S20.

Korf BR (1996). *Human genetics: a problem-based approach*. Blackwell Scientific, Cambridge, Massachusetts.

Leung W-C (2000). 'Results of genetic testing: when confidentiality conflicts with a duty to warn relatives'. *BMJ*; 321: 1464-1466.

Magnusson R (1995). 'Specific consent, fiduciary standards and the use of human tissue for sensitive diagnostic tests and in research. *Journal of Law and Medicine*; 2: 206-232.

Magnusson R (1996). 'Australian HIV/AIDS legislation: a review for doctors'. *ANZ J Med*; 26: 396-406.

Magnusson R (2002). 'Data linkage, health research and privacy: regulating data flows in Australia's health information system'. *Sydney Law Review*; 24: 5-55.

Mount C, Kelman C, Smith L et al (2000). 'An integrated electronic health record and information system for Australia?'. *Med J Aust*; 172: 25-30.

Murray T (1997). 'Genetic exceptionalism and 'future diaries': is genetic information different from other medical information?'. In: M Rothstein (ed.), *Genetic secrets: protecting privacy and confidentiality in the genetic era*. Yale University Press, New Haven, Connecticut, 60-73.

National Health and Medical Research Council (2000). *Ethical aspects of human genetic testing: an information paper*. NHMRC, Canberra.

NHMRC – see National Health and Medical Research Council.

Office of the Federal Privacy Commissioner (2001). *Guidelines on privacy in the private health sector*. Viewed 28 March 2002 <<http://www.privacy.gov.au/health/guidelines/index.html#1>>.

Richardson J (1998). 'Economic evaluation of health promotion: friend or foe?'. *Aust NZJ Public Health*; 22: 247-253.

Schwartz R (1996). 'Genetic knowledge: some legal and ethical questions'. In: D Thomsma and T Kushner (eds), *Birth to death: science and bioethics*. Cambridge University Press, Cambridge, 21-34.

Smith G (2000). *Human Rights and Biomedicine*. Kluwer Law International, The Hague.

Skene L (1998). 'Patients' rights or family responsibilities? Two approaches to genetic testing'. *Medical Law Review*; 6: 1-41.

Trent R (2002). 'Genetics'. *Med J Aust*; 176: 18.

Wexler N (1992). 'Clairvoyance and caution'. In: D Kevles and L Hood (eds), *The code of codes: scientific and social issues in the human genome project*. Harvard University Press, Cambridge, Massachusetts, 211-243.

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[1] The development of HealthConnect is being coordinated by the National Health Information Management Advisory Council (NHIMAC), established in July 1998 by Australian Health Ministers, and bringing together consumers and representatives of Australian governments and the private health sector. Following on from its November 1999 report, *Health Online*, the National Electronic Health Records Taskforce was established by Health Ministers through the Council. The Taskforce's July 2000 report, *A Health Information Network for Australia*, proposed key features of a network of electronic health records, although the architecture currently remains fluid (Commonwealth of Australia 2000).

[2] The National Electronic Health Records Taskforce quotes studies to the effect that three consumers per 1000 admitted to hospital will die and one will suffer long-term disability due to adverse drug events, and that up to 70% of such events may be prevented by improved information systems (Commonwealth of Australia 2000, p.57; Report to the Electronic Health Records Taskforce, *The Benefits and Difficulties of Introducing a National Approach to Electronic Health Records in Australia* April 2000, p.22).

[3] Recently, production of the Medicare number became a prerequisite to the receipt of pharmaceutical benefits (in addition to Medicare benefits): see *National Health Amendment (Improved Monitoring of Entitlements to Pharmaceutical Benefits) Act 2000* (Cth).

[4] Specifically, digital certificates will ensure *authentication* (the identity of the message sender), *data integrity* (non-alteration of data between sending and receiving), *non-repudiation* (assurance that the sender cannot dispute creating and sending the message) and *confidentiality*, in respect of data transfers between subscribers to the relevant certificate policies. There are two types of digital certificates: Healthcare Location Certificates, and Healthcare Individual Certificates (Health Insurance Commission, undated).

[5] These include the secure transmission of referrals from doctors to specialists and to hospitals, secure discharge summaries from hospitals to doctors, secure transmission of pathology results to doctors, secure on-line claiming for doctors and pharmacists, and secure access to the Australian Organ Donor Register and Australian Childhood Immunisation Register (Health Insurance Commission, undated).

[6] The record would include the 'consumer's details, their needs as identified or assessed, what services are planned and when they are required, relevant data collected (for example, test results) and summary reports from each contact with the consumer. The record should also have the capability to notify providers and consumers when actions, such as service reviews, scheduled appointments or proposed remedial action, are required and to notify consumers when scheduled self-management activity should occur' (Department of Human Services (Victoria) 2001b, pp.30-31).

[7] PoCCS would record admission history, vital signs and clinical measurements, and could be used to generate clinical pathways and care plans, to order tests, and to generate discharge referrals and clinical reports (Ayres, Horvath and Kidd 2001, pp.14-15).

[8] In Australia, as in other developed countries, there is strong pressure towards cost savings and efficiency gains in the health sector. Spending on health care is slowly increasing, and the budget impact is expected to be magnified in future by the health needs of a growing, aged population (Commonwealth of Australia 2000, p.11). Through the 1990s, total spending on health services (government and non-government) grew at an average annual rate of 4%, reaching \$53.7 billion in 1999/2000 (Australian Institute of Health and Welfare 2001).

[9] Mount, Kelman and Smith argue that the health information system:
(a) should serve the health needs of both the individual and the nation.
(b) should enable the monitoring of trends, and facilitate health administration and management.
(c) should improve the efficiency of health service delivery, both personal care and public health services.
(d) should build from a primary care and population health base.
(e) should meet privacy and confidentiality requirements.
(f) should be developed intentionally rather than accidentally, in a coordinated rather than fragmented manner (Mount, Kelman and Smith 2000).

[10] The WHO defines health as 'a state of complete physical, mental, and social well-being and not merely the absence of disease or infirmity', viewed 28 March, 2002, <<http://www.who.int/aboutwho/en/definition.html>>

[11] See *Privacy Act 1988* (Cth), Schedule 3, National Privacy Principle 2.1(a); Office of the Federal Privacy Commissioner 2001, pp.14-15.

[12] See *Privacy Act 1988* (Cth), Schedule 3, National Privacy Principle 2.1; *Health Records Act 2001* (Vic), Schedule 1, Health Privacy Principle 2.2; *Privacy and Personal*

Information Protection Act 1988 (NSW) ss 17-19; Health Records (Privacy and Access) Act 1997 (ACT) s 5 Privacy Principles 9-10.

[13] In New South Wales, for example, genetics laboratories have been funded by Health Department and Area Health Service allocations under a consortium model where each laboratory 'specialised' in a small number of genetic disorders (Department of Health (NSW) 2000b, para 1). In Victoria, the Victorian Clinical Genetics Service (within the Murdoch Institute), a centralised, State-funded service, provides an over-arching, coordinating role in the provision of clinical genetics services to all hospitals, clinics and primary care providers employing clinical geneticists, genetic counsellors and scientists.

[14] The Human Genetics Society of Australasia maintains a database of those genetic tests currently being offered in Australia, and by which organisation within each State: <http://www.hgsa.com.au/> (refer to 'DNA Testing Laboratories').

[15] <http://www.hgsa.com.au/> (refer to 'HSGA Policies').

[16] More specifically, the Department states that: 'Evidence based pathways should be developed which provide rigorous and robust criteria for offering tests. Issues to be considered include testing capabilities, criteria for test availability, sensitivity and specificity of tests, costs and benefits, risk categories for access, numbers in the community for whom testing is appropriate, pretest genetic counselling requirements, quality control measures in laboratories and recommended throughput, and whether it is appropriate to offer a particular test from one or more laboratories' (Department of Health (NSW) 2000b, para 4.6).

[17] The following discussion draws on verified data from an interview the author conducted with Professor David Danks, former Director, Victorian Clinical Genetics Service and Murdoch Institute, Royal Children's Hospital, Melbourne, 27 June 1994.

[18] Newborn screening using 'Guthrie spots' has long been carried out by States and Territories ('ALRC and AHEC' 2001, pp.258-260). The NSW Newborn Screening Program, based at the New Children's Hospital, Westmead, Sydney, screens newborns for phenylketonuria, primary congenital hypothyroidism, cystic fibrosis, galactosaemia, and more than 20 rarer metabolic disorders. Of the 87,000 babies born each year in NSW, around 90 are diagnosed with one of these genetic conditions (Department of Health (NSW) 2001).

[19] In New South Wales, for example, genetic neonatal cystic fibrosis screening occurs under the *Public Health Act*. The current screen tests for the 12-15 commonest mutations causing CF. The Delta F508 mutations account for about 85% of cases.

[20] See *Duncan v Medical Disciplinary Committee* [1986] 1 NZLR 513, at 521; *Slater v Bissett* (1986) 69 ACTR 25, at 28-29; *Brown v Brooks*, unreported, Supreme Court of New South Wales, 18 August 1988, McLelland J (duty of consulting clinical nurse).

[21] Most but not all of these provisions apply to public sector-held health information. See *Health Administration Act 1982 (NSW) s 22; Public Health Act 1991 (NSW) s 75; Mental Health Act 1990 (NSW) s 289; Health Services Act 1988 (Vic) s 141* (also applies to private hospitals); *Mental Health Act 1986 (Vic) s 120A; Health Services Act 1991 (Qld) s 63; Health Act 1937 (Qld) ss 100E, 100FO, 100I; Public and Environmental Health Act 1987 (SA) s 42; South Australian Health Commission Act 1976 (SA) s 64.*

[22] See *Public Health Act 1991 (NSW) s 17; Health Act 1958 (Vic) s 128; HIV/AIDS Preventive Measures Act 1993 (Tas) ss 17-19.* For a more detailed review, see Magnusson 1996, pp 399-402.

[23] *Privacy Act 1988 (Cth) s 14.*

[24] *Privacy Act 1988 (Cth) Schedule 3* (the 'National Privacy Principles', introduced by the *Privacy Amendment (Private Sector) Act 2000 (Cth)*).

[25] *Privacy and Personal Information Protection Act 1998 (NSW); Health Records Act 2001 (Vic).*

[26] *Information Privacy Act 2000 (Vic).*

[27] *Health Records (Privacy and Access) Act 1997 (ACT); Health Records Act 2001 (Vic).*

[28] See 'HealthConnect Fact Sheet 2: Privacy, Confidentiality and Security', July 2001, viewed 28 March 2002, <http://www.health.gov.au/healthonline/hc_2.htm>

[29] A recent information sheet, 'Towards a National Health Information Privacy

Framework', states that this legislation may determine 'how consumer consent should operate in HealthConnect (including consent of what information is transferred, to whom, and for what purposes), the responsibilities and obligations of providers, the purpose for which the information can be used, and governance arrangements'.

[30] To quote NSW Privacy Commissioner Chris Puplic, speaking to the First National Privacy Conference, Melbourne, 26 November 2001.

[31] Other examples include Kaiser Permanente, a managed care organisation in the United States, which provides online services to members, which mistakenly sent 838 e-mail messages to the wrong recipients, some of which contained sensitive information (Goldman and Hudson 2000, p.141).

[32] This is perhaps the most serious risk within on-line health information systems. Recent examples include the case of an employee of the Health Insurance Commission detected browsing the records of women who had had IVF treatment, and Asian women: see 'Health, Privacy and the New Technology', *The Law Report*, ABC Radio, 27 February 2001; *Lodkowski v Comcare*, unreported, Federal Court of Australia, Goldberg J, 5 March 1998 (disclosure by an employee of the Health Insurance Commission that the applicant had had a termination of pregnancy prior to marriage).

[33] Leung (2000) presents a case study featuring a man with a recently diagnosed autosomal recessive inheritance, treatable in the presymptomatic stage, who refused permission to disclose his diagnosis to his siblings.



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