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WARNING PATIENTS' RELATIVES OF GENETIC RISKS: POLICY APPROACHES

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As an increasing number of genetic tests for specific early- and late-onset disorders move from research to the clinical setting, health care professionals are faced with new challenges or, alternatively, with novel twists on age-old ethical dilemmas. A finding that an individual carries a deleterious mutation can indicate that his or her relatives are at an increased risk of being affected by the same genetic disorder.[1]

Though most patients share such risk information with their family once aware of its broader implications, some individuals refuse to do so.[2] This places the health care professional in a quandary, particularly when the disorder in question is serious and preventable or treatable. On one hand, the health care professional owes the patient a duty to keep the information secret, and on the other, he or she has a duty to act for the benefit of others, including the family members who have a significant interest in knowing that they may be at risk. The potential benefit of this knowledge for the patient's relatives therefore calls into question the duty of confidentiality of the health care professional.

This edition of GenEdit will briefly examine the legal and ethical duties of health care professionals, and the policy approaches that have been adopted at the international, regional and national levels to address the dilemma created by a patient's refusal to share relevant genetic risk information with those who are at risk.

A. The Health Care Professional's Legal and Ethical Duty of Confidentiality

The duty of confidentiality has been a fundamental element of medical ethics since antiquity. Based on values of autonomy, respect for personal integrity and trust, its importance continues to be emphasised in the codes of ethics of various health professions.[3]

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The 2004 *CMA Code of Ethics* for example states that physicians must "[p]rotect the personal health information of [their] patients" and may disclose such information only with their patients' consent or as provided for by law.[4]

The legal duty of confidentiality has its source in a number of privacy protection statutes, health laws, regulations governing health care institutions and professionals, as well as common law rules and civil law principles. Briefly stated, the legal duty entails that health care professionals must keep information they receive in the context of a therapeutic relationship in confidence, unless the patient consents to its disclosure. Confidentiality is not, however, absolute. Exceptions to the general rule authorise health care professionals to disclose confidential information to third parties in limited circumstances. These vary across jurisdictions, but generally include situations in which there are paramount reasons for breaching confidentiality, such as when disclosure can limit or prevent a serious and imminent harm to the health or safety of an individual or a group of individuals. In North America, exceptions have been recognised in cases of suspected child abuse, risk due to an infectious disease, and the utterance of threats of physical harm towards third parties.

While a small number of jurisdictions have enacted laws that specifically address genetic information and its disclosure to family members, in most countries, the processing of this type of personal information is governed by general privacy or personal data protection statutes. These statutes provide valuable protection for individual interests in the confidentiality of all types of sensitive data, including, by definition, genetic information. However, they fail to take into account its familial nature. Family members are considered in the same manner as other third parties (such as employers, financial institutions, or one's neighbour) and therefore subject to the same rules.

The statutory exceptions that authorise the non-consensual disclosure of personal information for purposes related to the protection of a third party are limited in scope and may apply only exceptionally to genetic risk information.

In many Canadian provinces for example, disclosure is not authorised unless the risk is imminent, results from an emergency situation, or constitutes a risk of serious injury or death. As a result, the disclosure of genetic risk information to family members may be prohibited even if there is a highly probable, serious and preventable risk to their health. A number of genetic-specific statutes, as well as policies, guidelines and recommendations developed by professional and other organizations on the subject of privacy and genetic information attempt to strike a balance between the rights of patients and family members.

B. Possible approaches

There are at least five different approaches possible when attempting to solve this dilemma. A first approach is based on the current, atomistic model of health care and adopts a rule of strict confidentiality. In order to maintain the inherent trust in the health care professional-patient relationship, the professional cannot disclose information about the patient to a third party, including the patient's family members, without his or her consent. The moral obligation to disclose genetic risks, if any, is that of the patient. The professional's ethical duty is fulfilled when he or she informs the patient of the relevance of the genetic information for family members, and attempts to persuade him or her of the importance of sharing it with those at risk.[5] In addition, the professional can provide tools such as documentation to assist the patient in communicating with relatives.[6]

This approach, adopted by the European Parliament,[7] the French National Consultative Ethics Committee for Health and Life Sciences,[8] the American Society of Clinical Oncology,[9] the Council on Ethical and Judicial Affairs of the American Medical Association,[10] and the Canadian Cystic Fibrosis Foundation[11] among others,

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preserves the therapeutic relationship and protects the family's interests, particularly where sufficient time is allowed for effective counselling.

It however presumes close family ties and functional relationships, an assumption that does not apply to all families.

A second approach, based on the principle of mutuality, characterises genetic risk information as familial information.[12] Though the relevant relationship may be, as in the first model, that of the individual patient and the health care professional, the shared nature of genetic information entails that the patient is not entitled to confidence. Under this approach, the health care professional would have a broad ethical duty to warn the patient's family members of genetic risks if the patient refuses to do so himself or herself. If this ethical duty becomes a standard of practice, it could evolve into a positive legal duty, rather than a permissive exception to confidentiality. This approach would have significant implications for the therapeutic relationship, as it would fundamentally change the health care professional's role. Brought to its ultimate conclusion, it could also lead to the position that the patient is the family rather than the individual seeking care, which would have considerable legal ramifications.

The World Health Organization tends toward this approach. It specifically recognises that "genetic information may affect an entire family," and recommends that "[t]he provision of genetic information to relatives about the family so as to learn their own genetic risk should be possible, especially when a serious burden can be avoided." [13] However, it also recognises that the primary moral duty of informing family members of the existence of a genetic risk does not rest with the health care professionals, as it recommends to health care professionals that they inform their patients that it is their (the patients') responsibility to communicate to their blood relatives that they may be at risk.

The third approach is based on a modification of the implicit contract that governs the physician-patient relationship. It involves informing the patient, prior to testing, that in the presence of certain circumstances the health care professional will disclose relevant genetic risk information to family members regardless of the patient's intentions. The presumption of confidentiality would stand, but would be subject to clear exceptions. The patient would have the option to accept these conditions and undergo testing as planned, to search a health care professional whose policies meet the patient's expectations or to forego testing entirely. One could argue that the therapeutic relationship would not be threatened if the expectations of the professional and the patient are clearly established from the outset.[14] Though the patient's autonomy seems to be respected in this approach, his choice may be theoretical only given the limited number of professionals available in many regions and the often limited knowledge of the complexity of genetic risks among health care professionals. In practice then, this approach may on the contrary be coercive, leaving patients with the perception that they have no real choice.[15]

The fourth approach reflects an intermediary position: as a general rule, the patient's right to confidentiality must be respected; however, the non-consensual disclosure of confidential information may be considered ethically permissible in exceptional circumstances. A number of guidelines, policies and recommendations espouse this approach.[16] The seriousness of the potential harm, its preventability, and the necessity of disclosure are key elements of the justification for disclosure.

For example, the World Medical Association's position is that exceptions to medical secrecy are justifiable only if the disclosure of information could avert a serious harm and if it is limited to relevant genetic information. Even in such cases, breaching patient confidentiality is a last resort, as all efforts must have been made to convince the patient to share the information himself/herself.[17]

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Other policies require additional elements such as a high probability of harm, a high likelihood that the information will be used to avert harm, the absence of any reasonable alternatives to non-consensual disclosure and a balance of potential harms and benefits that favours disclosure.

While most guidelines and policies leave the interpretation and application of these criteria to health care professionals, some jurisdictions require the approval of an ethics committee[18] or of a competent agency or authority.[19]

This intermediary approach would not impose a legal obligation on health care professionals to warn family members. It would simply provide an ethical justification for doing so, as well as a defence against a possible complaint for breach of confidentiality. It has the advantage of protecting the therapeutic relationship and the patient's expectation of confidentiality while recognising that in exceptional circumstances, the needs of family members may be given precedence.

The fifth and final approach is also based on the acknowledgment of an exception to the general rule of confidentiality. As in the fourth approach, a health care professional could breach the recalcitrant patient's right to confidentiality when in the presence of a foreseeable and highly likely risk of serious yet preventable harm to identifiable individuals. In this case however, the disclosure would be made to an independent organisation or agency that would then transfer the relevant information to each family member's physician if one is known, or to a health care professional who would contact the family members and provide guidance for follow up care. France has adopted a variation of this approach when it revised its *Bioethics Laws* in 2004. It seems to give the concerned patient the legal responsibility of warning family members of genetic risks, but implements a procedure that involves the physician. When a patient is diagnosed with a serious anomaly for which a treatment or preventive measure exists, the physician must provide the patient with a written and verbal explanation of the potential harm to which non-disclosure of the genetic risk could expose family members.

The patient then has two options: he can disclose the information himself or avail himself of the "procedure for medical information of a familial nature." If he chooses the latter option, he gives the physician the names, addresses and degree of familial relation of each family member. The physician sends the relevant information to the "Agence de la biomédecine" who then informs the family members through a second physician of the availability of familial medical information that could be relevant to their health.[20] This approach has the advantage of recognising both the value of medical confidentiality and the significance of genetic risk information for family members, while not placing health care professionals in the difficult position of contacting individuals with whom they have no professional relationship to inform them of a health risk. To that extent, it adopts a strategy that has proven effective for decades in public health: the reporting of risk and subsequent provision of information to at-risk individuals by specialist third parties. It however carries the disadvantage of requiring the creation of elaborate administrative processes.

C. Discussion: Balancing Interests in the Protection of Confidentiality and in the Prevention of Harm

These various policy approaches attempt to establish a balance between two morally valuable social interests: the protection of medical confidentiality and the prevention of harm. Though the duty of confidentiality is not absolute, it is a cornerstone of the health care professional-patient relationship. It is essential to creating the climate of trust required in a therapeutic relationship,[21] it respects the moral integrity of patients and their right to control information about themselves, and it complies with the implicit undertakings of health care professionals whose professions promote confidentiality through their codes of ethics.[22] Ultimately, confidentiality benefits society because it leads to better health and to the prevention of disease.[23] As such, it cannot be set aside lightly.

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The prevention of harm and suffering associated with genetic disorders also benefits society and, in some circumstances, may justify breaching confidentiality. Genetic disorders can have a significant impact on the lives of individuals, including premature death, chronic pain or physical ailment, severe psychological or behavioural impairment, as well as social ramifications and the financial stress linked to ill-health.

They can also affect the lives of those who are close to the afflicted individual, potentially creating social and familial hardships. When such serious harms are highly likely and preventable, the disclosure of a genetic risk can clearly benefit at-risk family members.

However, not all genetic disorders share these characteristics.

- not all genetic disorders are equally serious. Some disorders may, in their mild forms, present symptoms that do not create serious impairment or to which affected individuals can adapt.[24] The evaluation of the magnitude of harm is a complicated matter firstly because variable expressivity and penetrance entail that it is difficult in many cases to predict how a genetic disorder will affect an individual's health. In addition, the characterisation of the effects of a disorder is highly subjective since perception of disease, impairment and quality of life varies greatly among individuals, families and even geneticists.[25] The perception and the actual impact of a disorder on the lives of those who are affected depend on a number of factors, including one's economic, social and cultural environment. Psychological harm is also very difficult to evaluate, particularly in the absence of knowledge about an individual's personal circumstances.

- the probability of harm associated with genetic disorders is highly variable. At one end of the spectrum, the risk of transmission of monogenic diseases is clear and may be as high as 50%, while at the other end, the risk of transmission of common complex disorders is difficult to predict and is much lower. In the latter cases, genetics is but one factor in the materialisation of the disorder.

- most genetic disorders are not currently preventable or treatable. In some cases, the available preventive measures are of limited efficacy or carry their own risk of harm because they are invasive or can entail side effects. These must be taken into account in evaluating the benefit of disclosure. However, even if a disorder is not preventable, disclosing the existence of a genetic risk may foster the autonomy of at-risk family members.

In addition, the non-consensual disclosure of genetic risk information can be harmful at various levels:

- at an individual and societal level, it could have a considerable negative impact on trust in geneticists and health care professionals in general, which would diminish professionals' ability to provide adequate care and to conduct prevention activities;[26]

- disclosing genetic risk information to a patient's family members after the patient has expressly refused to do so would show utmost disregard for his or her autonomy and moral integrity, and could lead to mental and emotional distress for the patient as well as the family members;[27]

- it could have repercussions on the patient's relationship with his or her close relatives, particularly if it brings to light sensitive issues;[28]

- it could infringe on the family member's autonomy if he or she prefers not to know of the risk;[29] and

- if disclosure became a standard of care, it would fundamentally change the professional's role, making the therapeutic relationship "subservient to a more diffuse public health obligation, benefiting an unspecified number of nonpatient relatives."[30]

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Given the variability of genetic conditions and the numerous factors that can have an impact on the ramifications of the disclosure of a genetic risk, it is not possible to develop precise rules or policies of general application. These cases necessarily involve a measure of professional judgement. In our opinion, health care professionals should not be imposed a positive duty to warn.

The primary ethical responsibility to warn family members of a genetic risk should rest with the patients, since they are in the best position to communicate with their relatives. However, if a patient refuses to do so, health care professionals should have the legal and ethical discretion to inform the patient's family members of the genetic risk. This option should be available as a last resort only. Before a breach of confidentiality is contemplated, health care professionals should be required to ensure that all other options have been explored, namely by considering whether the risk information could possibly be communicated without revealing any personal information, informing patients prior to testing of the potential significance of results for family members, attempting to convince patients of the need to share the information with at-risk relatives through appropriate counselling before and after testing, and offering assistance to patients who are uncomfortable with communicating the information. Taking these steps may eliminate in many cases the need to resort to breaching confidentiality.

Finally, fostering the acceptance of genetic information as normal medical information of particular importance for family members may lead to a shift in values from individual autonomy to principles such as reciprocity, mutuality and solidarity. [31] This would ensure that in the future, genetic risk may be freely shared within families in the ethos of mutuality and solidarity.

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[5] B.M. Knoppers, "Genetic Information and the Family: Are We our Brother's Keeper?" (2002) 20:2 Trends Biotech 85 at 85.
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[15] K. Offit *et al.*, "The 'Duty to Warn' a Patient's Family Members About Hereditary Disease Risks" (2004) 292:12 JAMA 1469 at 1472.
[16] This is the essence of the World Health Organization's recommendations discussed above. Regionally, the Council of Europe and the European Commission have adopted a similar approach: Council of Europe, *Recommendation R(97)5 of the Committee of Ministers to Member States on the Protection of Medical Data,* (1997), online: Council of Europe <http://cm.coe.int/ta/rec/1997/97r5.html> (date accessed: 25 July 2005); Directorate-General for Research, European Commission, 25 Recommendations on the ethical, legal and social implications of genetic Testing (Brussels: European Commission, 2004). At the national level, see for example, Australian Law Reform Commission and Australian Health Ethics Committee, *Essentially Yours: The Protection of Human Genetic Information in Australia* (Sydney: Australian Law Reform Commission, 2003), recommendation 21-1; British Medical Association, *Human Genetics: Choice and Responsibility* (Oxford: Oxford University Press, 1998) at 73; Nuffield Council on Bioethics, *Mental Disorders and Genetics* (London: Nuffield Council on Bioethics, 1998) at 50-51; Science Council of Canada, *Genetics in Canadian Health Care: Report 42* (Ottawa: Minister of Supply and Services, 1991) at 72-73; President's Committee for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research: *Screening and Counseling for Genetic Conditions: The Ethical, Social and Legal Implications of Genetic Screening, Counseling, and Education Programs* (Washington, D.C.: Government Printing Office, 1991); U.S., Institute of Medicine, Committee on Assessing Genetic Risks, *Assessing Genetic Risks: Implications for Health and Social Policy* (Washington, D.C.: Institute of Medicine, 1994); American Society of Human Genetics, "ASHG Statement: Professional Disclosure of Familial Genetic Information" (1998) 62 Am. J. Hum. Genet. 474 at 474.
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[20] *Loi n° 2004-800 du 6 août 2004,* J.O., 7 August 2004, ss. 4-5. The law lacks clarity with regard to the issue of legal responsibility. Though the law seems to impose on the patient a legal duty to inform his or her family members, it does not clarify whether this duty lies in civil law or in criminal law. The sanctions that would apply in the event of a failure to comply with the law are currently unclear. Moreover, the law does not specify

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the extent of the initial physician's responsibility. In the first option, must the physician verify that the patient has indeed informed the family members? In the second option, would the physician be liable in the event that he or she failed to ensure that the patient provided complete information, or failed to transmit the information to the Agence? These questions are not addressed in the Act.

[21] B.A. Brody *et al.*, *Medical Ethics : Analysis of the Issues Raised by the Codes, Opinions and Statements* (Washington, D.C.: Bureau of National Affairs, 2001) at 177.

[22] *Ibid.* at 178; P. deCruz, *Comparative Health Care Law* (London: Cavendish Publishing, 2001) at 51-53.

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[30] Offit *et al.*, *supra* note 16 at 1472.

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