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INTRAFAMILIAL DISCLOSURE OF GENETIC RISK INFORMATION FOR HEREDITARY BREAST CANCER: A COMMUNICATIONS FRAMEWORK?

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There is currently little guidance for health professionals regarding intrafamilial communication of genetic risk for hereditary breast cancer caused by a mutation in the BRCA 1 or 2 genes. A number of national laws address risk communication, but virtually none address intrafamilial communication in a meaningful way. Non-governmental bodies have, to various extents, noted or addressed intrafamilial communication, but there is no harmonization. Importantly, basic considerations are not well-developed, including (i) who is the family, (ii) what is genetic information, and (iii) how is the obligation or responsibility to communicate genetic information characterized. If genetic risk information is to be viewed as something more than "individual", frameworks must be developed to aid health professionals and patients in ensuring that others in the family who need the information receive it.

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The simple fact is that there is very little guidance for health professionals when it comes to intrafamilial communication of genetic risk for hereditary breast cancer caused by a mutation in the BRCA 1 or 2 genes. National laws set forth pieces of the puzzle, but virtually none directly address intrafamilial communication. International and national organizations shed a little more light on the subject, but again fail to provide more than cursory guidance. The current system of communication depends primarily on the health care professional who is tasked with explaining the results of a genetic test or genetic risk assessment to a patient. However, this system is often insufficient to meet the needs of patients and their families. Furthermore, the increasing accessibility of genetic information will only further exacerbate the effects of a lack of communications framework.

This paper seeks to discuss a few basic questions that are inherent in intrafamilial communication of genetic risk. First, who is the family? If patients are supposed to spread this information, who should they be telling? Second, what is the information patients are supposed to provide? Given the importance of these questions there is surprisingly little consensus on their answers. What are the normative frameworks saying on these questions?

If we look to the law for an answer, it will generally be at the national level. Unfortunately, no country has legislated definitions of 'family' and 'genetic information' in the context of intrafamilial communication. However, there are examples from laws with similar subject matters that can be enlightening even for intrafamilial communication of genetic risk information. Beyond the law, national and international organizations have addressed these issues as well. Over the past years, countries such as the United States and Australia have sought to legislate these terms.

1. FAMILY AND GENETIC INFORMATION

(i) Legislated Responses

United States

In the United States, the recently enacted *Genetic Information Nondiscrimination Act* prohibits the use of genetic information by third parties for employment or health insurance decisions. For families, the law protects the genetic information of relatives up to and including the 4th degree.[1] Married spouses and adopted children are also included, but this is the extent of the non-biological relations included in the definition.[2]

The same law also defines genetic information—somewhat broadly—to include information about a patient's genetic test, genetic tests of family members and the "manifestation of a disease or disorder in the family members."[3]

Australia

Australia adopted a set of guidelines to address *physician* communication of genetic information to relatives.[4] In these guidelines, "genetic relative" is defined more restrictively than "family member" in the US law: only relatives up to the 3rd degree may be informed and there is no inclusion of legal relations such as spouses or adopted children. However, this law also recognizes and supports that these relatives might inform others in the family, thereby causing the information to reach a wider audience than just the 3rd degree relatives and lessening the burden on the patient.

Conversely, these guidelines' provisions for genetic information are perhaps more broad than the US law: "genetic information is gained

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from a range of sources (eg clinical examination, DNA testing and chromosome studies, newborn screening, family history) and may confirm a condition that is clinically apparent, or be predictive of the likelihood of an individual developing or carrying a mutated gene causing a condition."[5] However, it must be noted that this is not a "definition" but rather "the context to determine whether the use of genetic-related information requires any special handling or protection."

Canada

Canada, at the federal and provincial levels, has no law defining family or genetic information that is closely related to the circumstances of intrafamilial communication. Various family law regimes define family, but those definitions vary based upon the specific context of the law. For example, federally the *Divorce Act* gives a broad definition of children of the marriage, including a child for whom one or both spouses stand in place of a parent.[6] The regulations of the *Immigration and Refugee Protection Act* define family as spouse or common-law partner and dependent children.[7] Provincial legislation has not clarified the matter any further: "child," "spouse" and "parent" have been defined within numerous child and family laws, but these laws are often silent as to who else can be considered as a family member.[8]

When it comes to genetic information, Canadian law provides even less guidance. Indeed, the term is not specifically defined in current law, although statutes governing health or medical information might incorporate (implicitly or explicitly) genetic information.[9]

(ii) International, Regional and National Norms

We can look to sources other than governments for a clue as to how family and genetic information might be used in the context of intrafamilial communication. A number of bodies have addressed the issue, but much like governments they lack harmonization.

On one end, the European Commission has adopted a narrow definition of family by acknowledging that genetic testing is of consequence to blood relatives such as offspring.[10] On the other, both the French National Consultative Ethics Committee and German Society of Human Genetics have adopted a broad interpretation of family to include the extended family as well as legal relatives such as spouses.[11]

The definition of genetic information has also been addressed by various organizations. UNESCO has adopted a *very* narrow view of genetic information as "information about heritable characteristics of individuals obtained by analysis of nucleic acids or by other scientific analysis."[12] Others have taken a more encompassing position.

The Council of Europe broadly states that "the expression 'genetic data' refers to all data, of whatever type, concerning the hereditary characteristics of an individual or concerning the pattern of inheritance of such characteristics within a related group of individuals."[13] This is potentially broader than standards in both the United States and Australia.

2. PERSONAL RESPONSIBILITY

Having definitions might set the stage for a communications framework. Another important question is how to go about increasing disclosure rates of genetic risk for breast cancer within

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families. Certainly, it is not uncommon under the present system of communication (though it is a stretch to call it a "system") for patients to inform family members of risk following a genetic test,[14] but there is no uniformity in how this is done and there are still patients who refuse to disclose.

(i) Legislated responses

Although a number of organizations have addressed intrafamilial communication of genetic risk information, there is a nearly complete absence of national-level law aimed at this. The one exception is France, which adopted a law in 2004 to create a framework for disclosure of this information to families.[15] However, this law does not create a system of obligations and sanctions. Rather, it provides patients with an anonymous method for disclosing genetic test results to family if they did not wish to do so personally. Participation in the system is voluntary, and patients are not penalized for refusing to disclose. Unfortunately, there is no evidence to date on how well this system has worked. Such data could provide incentive for other countries to develop similar mechanisms for disseminating genetic risk information and thereby create a more uniform international network.

(ii) International and national norms

The bulk of guidance on intrafamilial communication of genetic risks comes from national and international bodies that have expertise in medico-ethical issues. The majority consensus of these documents is that patients have the primary responsibility for disclosing risks to others in their families.

The American Medical Association places the burden of communicating genetic information on patients, although physicians are encouraged to educate patients about the risks of not providing risk information to family.[16] However, there is no duty, express or implied, for patients to actually disclose the information, just a prohibition against physicians doing so without patients' consent. A similar statement has been made by the American Society of Human Genetics, asking that patients be made aware of the implications of a test result to family, but not obliging them in any way to actually disclose the results to family.[17]

Others have gone further to recognize a duty of patients to communication genetic information. Groups in both France and Germany have noted a *moral* obligation of patients to disclose genetic information.[18] In Germany, this obligation extends to relatives and spouses, and France includes those who would benefit from the information—a potentially larger group of individuals than family alone.

A middle route has been to mark the importance of this information to family without going so far as creating a moral (or personal) obligation to disclose the information. The Nuffield Council on Bioethics places primary responsibility for communicating the information with patients, although it would permit physicians to disclose without the consent of patients in limited circumstances.[19] The policy appeals to patients' feelings of obligation in a more subtle way: patients "acting responsibly would normally wish to communicate important genetic information to other family members who may have an interest in that information…."[20] In other words, if you do not disclose this information to your family, you are irresponsible.

3. DISCUSSION

For a communication framework to be functional, it has to have consistent and well-

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reasoned definitions of basic terms, including "family" and "genetic information." What becomes clear from an examination of legislation and current national and international norms is a lack of agreement on what these terms mean. Of course, this might in part be due to intrafamilial communication not being addressed in the law. In addition, many of those guidelines that set forth obligations and rights with regard to intrafamilial communication fail to provide definitions to clarify who and what are to be included in disclosures. However, the breadth of usage in law and policy gives us at least a clue as to how these terms should be defined. Because the concern here is not the protection of information against use by third parties, but rather the dissemination of information to those who might benefit from it, a broad definition of both family and genetic information is preferable.

Moving from basic definitions to a mechanism for disclosure, most policy statements addressing the issue at the very least encourage patients to communicate risk information to family members (these policies are not limited to breast cancer risk). This is necessary because, with limited exceptions, legal and ethical restrictions prevent health professionals from providing this information directly. However, these matters have been discussed for over a decade, and there is still little guidance for health professionals and patients.

Although patients are expected to communicate genetic risk information, delineation of an *obligation* to do so in limited circumstances has been slow in coming. This perspective has been legislated in France, but its effectiveness is as yet unknown. A number of organizations and policymaking bodies assert that patients hold the responsibility (moral or otherwise) to protect the health of family members, at least when it comes to genetic risk, but their recommendations have not been implemented on a larger scale or in any manner that would increase their effectiveness.

All of these laws or guidelines fail to provide detail as to how families should be informed, how health professionals should convey the information to patients, and how patients should resolve internal conflicts regarding the disclosure. What we are left with is a set of diverse (though not conflicting) norms and an absence of beneficial guidance on the particulars of intrafamilial communication.

One factor that might impede the development of such detailed guidance is the nature of genetic information. For one, its provision without effective clinical intervention and support could provoke anxiety in family members who receive it. Second, the fear of genetic discrimination by governments, employers and insurers and of stigmatization by friends and family[21] might have prevented a more honest national and international discussion of the benefits of this information being shared rather than held in the strictest of confidence. Indeed, it is not unheard of that family history, a predictor of genetic risk, is used to discriminate.[22] This type of action has not helped allay the fears of discrimination, although the number of laws prohibiting discriminatory use of genetic information is growing.

Finally, not all genetic information is of the same value. In this review, we refer to disclosing information indicating the presence of a familial mutation in the BRCA 1 or 2 genes. Although continued debate surrounds the lifetime risk conferred by these mutations, studies have estimated the risk to be as high as in the range of 56% to 87%. In the realm of hereditary breast cancer, there are other genes that play a role in cancer development, but their risk is either less well defined or of lesser total value than that of the BRCA1/2 genes. Therefore deciding when it

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would be appropriate to disclose risk related information may be largely dependent on the context of its relative clinical impact. This in itself, might be another impediment to the development of appropriate guidance.

Presently, it is becoming increasingly difficult for patients and health professionals navigate their way when it comes to intrafamilial communication of genetic risk for hereditary breast cancer—this is true for many other genetic conditions as well. If, as has been written, "the very nature of genetic information, as both individual and universal, now mandates its treatment as familial,"[23] frameworks must be developed that recognize this fact and ensure that those who most need the information are provided it. And if, as the Nuffield Council on Bioethics has inferred, patients who do not share genetic risk information with family act irresponsibly, we must provide them with the tools to help them act differently.

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