review

Educating the general public about multifactorial genetic disease: applying a theory-based framework to understand current public knowledge

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The present article describes the application of a theory-based framework to understand current public knowledge of genetic risk factors of multifactorial diseases. The main innovative aspect is the application of E. M. Rogers' knowledge framework which distinguishes three types of knowledge: "awareness knowledge," "how-to knowledge," and "principles knowledge." We argue that distinguishing these types of knowledge allows for a more sophisticated overview of the general public. To illustrate the application of Rogers' framework, we performed a literature review of current public knowledge of genetic risk factors of multifactorial genetic diseases. Relevant articles were identified by searching the Pubmed, Web of Science, Embase, CINAHL, ERIC, and PsycINFO databases from January 1990 until January 2007 and by performing reference list and author searches. Although this review showed that current public knowledge is limited, it also showed that the knowledge framework may be a useful tool for assessing different types of public knowledge and pinpointing flaws or caveats in public knowledge with more precision and subsequently develop public health campaigns to remedy such flaws. Implications for genetic education are discussed. ***Genet Med* 2008:10(4):251–258.**

Key Words: *general public, genetic risk, awareness, knowledge, multifactorial disease*

As genetic science evolves, genetic predispositions to medical conditions are increasingly better understood. The completion of the Human Genome Project in 2003 has vastly increased professional knowledge of genetics.[1] The discovery of so-called polymorphisms (i.e., genetic variants) has broadened the scope of genetic risks from single-gene disorders to multifactorial diseases.[2] These advances in the understanding of genetic predispositions to disease have several implications for public health promotion[3]; the most obvious being whether and how to use genetic information to promote public health.[4] Investigating public understanding of genetic information is therefore an important dimension of the translation of professional genetic knowledge into public health benefits.[5]

Because knowledge is considered one of the prerequisites of health behavior,[6–8] a necessary first step in public health promotion involves increasing the general public's knowledge of the genetic risk factors of multifactorial diseases.[1] E. M. Rogers' conceptualization of knowledge distinguishes three types of increasingly complex knowledge: "awareness knowledge," which refers to knowledge about the existence of an innovation; "how-to knowledge," which is practical knowledge concerning the proper use of an innovation; and "principles knowledge," or knowledge of the underlying theoretical principles of the innovation.[9] Although increasing awareness knowledge is an important first step, public health programs should not neglect the other two forms of knowledge. Indeed, if how-to knowledge is not acquired before or during the adoption of the innovation, discontinuance is highly likely.[9]

Although a considerable body of literature has shown the value of this knowledge framework in understanding public knowledge of technical innovations,[10,11] there is relatively scarce research on the value of this framework for the diffusion of new knowledge or ideas. We propose that Rogers' knowledge framework is a valuable asset to the diffusion of (professional) knowledge of genetic risk factors for two reasons. First, the distinction between awareness, how-to, and principles knowledge presents a sophisticated overview of public knowledge of genetic risk factors. Second, it allows us to pinpoint caveats in the public's knowledge with more precision and adapt future public health campaigns to remedy them with appropriate strategies.

In the case of diffusing professional knowledge of genetic risk factors, awareness knowledge refers to simply knowing that there are genetic risk factors of a particular disease. This type of knowledge may act as a motivator to acquire how-to and principles knowledge.[9] For instance, knowing of the existence of genetic risk factors can motivate individuals to assess

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Disclosure: The authors declare no conflict of interest.

Submitted for publication October 18, 2007.

Accepted for publication January 10, 2008.

DOI: 10.1097/GIM.0b013e31816b4ffd

Table 1
Inclusion and exclusion criteria

Inclusion criteria	Exclusion criteria
Studies should focus on awareness, how-to, or principles knowledge of genetic risk factors of multifactorial diseases	Examined knowledge of inheritance and genetics in general
Participants should be representative of the general population without being specifically educated about genetic risk factors by professionals	Participants are individuals who know they have a genetic predisposition or have attended genetic counseling or other education about genetic risk factors
Studies should describe original articles published in a peer-reviewed journal	Concerned editorials, commentaries, book reviews, bibliographies, resources, or policy documents
	Reported secondary data analysis or only reviewed other studies

their family history or opt for genetic counseling and testing.[12] Knowing of the existence of genetic risk factors of a certain disease, however, does not guarantee adequate use of professional knowledge of genetic risk factors in decision-making concerning preventive behavior (e.g., assessing family history or changing unhealthy lifestyles). To this end, how-to knowledge is needed. How-to knowledge refers to practical knowledge of genetic risk factors, such as knowing how genetic risk factors influence the overall risk of developing the disease (e.g., knowing that there is no one-on-one relationship between genetic risk and disease development).

In contrast, principles knowledge refers to theoretical knowledge of genetic risk factors, such as knowledge of the underlying working mechanisms through which genetic risk factors affect disease development (e.g., knowing that certain polymorphisms create a genetic predisposition that interacts with other factors to develop the disease). Although adequate decision-making can occur without proper principles knowledge, the risk of falsely using perceived knowledge of genetic risk factors in decision-making is substantially higher without it.[9] For instance, a flawed or speculative understanding of how a disease can be genetic if the disease has not yet been observed in the family (as could be the case in recessive disorders or certain polymorphisms) has been shown to adversely affect health behavior.[13,14]

The general public thus needs to know that polymorphisms interact with other genes and environmental factors, such as lifestyle, to cause the development of the disease.[15] Understanding the complexity of gene-environment interactions (i.e., how and why genetic risk factors affect the development of multifactorial diseases) may prevent feelings of fatalism caused by unchangeable genetic predispositions and increase motivation to change unhealthy lifestyles, both in the absence, but especially in the presence of a genetic predisposition. Despite the importance of such knowledge for adequate preventive behavior, little research has focused on the general public's knowledge of genetic risk factors and how and why these factors affect health.[16] However, future health promotion programs intended to increase public knowledge of genetic risk factors for multifactorial diseases are likely to be unsuccessful if they disregard the public's prior knowledge.[17]

We propose in this article that applying Rogers' knowledge framework to map current public knowledge of genetic risk factors of multifactorial diseases will provide us with valuable insights on the structure of public knowledge as well as guide and assist future research and public education about multifactorial genetic disease. To illustrate the application of Rogers' framework, we performed a literature review of the general public's current knowledge of genetic risk factors of multifactorial diseases. The value of Rogers' knowledge framework will be discussed in light of the results of this review.

MATERIALS AND METHODS

Search strategy

Literature searches were conducted in the Pubmed, Web of Science, Embase, CINAHL, ERIC, and PsycINFO databases from January 1990 until January 2007. The following keywords were exhaustively combined: *awareness, knowledge, general public, general population, community, lay understanding, genetic risk, familial risk, multifactorial disease*, and *genetic risk factors*. Further, we also performed reference list and author searches. Table 1 specifies the inclusion criteria used to identify relevant studies.

Study selection

The selection process was completed in three separate phases. During the first phase, articles were selected or excluded based on their titles only. Studies selected for further review and those that could not be excluded without doubt proceeded to the second phase of the review process. Articles judged relevant based on their abstracts were included in the present review; those deemed irrelevant were excluded. In the third and final phase of the selection process, the remaining articles were exhaustively reviewed for content.

We included articles that examined awareness, how-to, or principles knowledge of genetic risk factors of multifactorial diseases among the general public. Articles were excluded if they examined knowledge of genetics in general or knowledge of genetic risk factors among individuals attending genetic counseling clinics. From an initial sample of 1174 eligible articles, we selected 20 for the present review. Because of the heterogeneity of the studies, we opted against pooling the data and thus against a meta-analysis of these studies. Relevant data were abstracted and summarized in Tables 2 and 3.

Table 2
Studies examining the general public's awareness knowledge of genetic risk factors of multifactorial diseases

Study	Country	Design	Sample	Disease	Outcome
Ackermann et al.[19]	Germany	Quantitative	2108 women of 23 gynecological outpatient services	Cervical cancer	93.3% identified genetic risk factors of cervical cancer
Bottorff et al.[43]	Canada	Quantitative	761 women without family history and 260 women with a family history	Breast cancer	Approximately 80% of each group was aware of the existence of breast cancer genes
Bruno et al.[44]	Italy	Quantitative	677 healthy women with or without a family history	Breast cancer	77% self-reported having heard about the existence of breast cancer genes. Women with and without a family history did not differ with regards to awareness of the existence of breast cancer genes
Hunt et al.[45]	England	Qualitative	61 men and women from middle and working class were selected from a large cross-sectional survey and subsequently interviewed	Heart disease	More than two thirds spontaneously mentioned genetic risk factors
Keighley et al.[46]	21 European countries	Quantitative	20,710 individuals over 16 years of age representative of general population in Europe	Colorectal cancer	54% was aware of the genetic risk factor of colorectal cancer
Mogilner et al.[20]	The United States	Quantitative	354 female patients of Mount Sinai Medical Center	Breast cancer	Overall, approximately 50% was aware of *BRCA1/2* genes. Participants of African-American descent (19%) and those who only completed elementary school (13%) were relatively unaware compared with whites (68%) and those who completed graduate school (71%)
Morris et al.[47]	The United States	Quantitative	2353 individuals contacted in a national survey	Four environmentally induced conditions	Public awareness of genetic risk factors of environmentally induced conditions was limited; 32% is unaware of genetic risk factors of environmentally induced conditions
Tambor et al.[21]	The United States	Quantitative	473 women without an increased risk of breast cancer	Breast cancer	51% was aware of the existence of breast cancer genes. Lower-educated individuals (less than high school) were 2.5 times less likely to be aware of genetic risk factors
Van den Nieuwenhoff et al.[22]	The Netherlands	Quantitative	4117 individuals representative of the general population	Inherited high cholesterol	50% was aware of inherited high cholesterol. Higher-educated individuals were more aware than lower-educated individuals (OR = 1.73, $P < 0.001$). Women were more likely to be aware than men (OR = 1.40, $p < 0.001$).
Waller et al.[18]	Great Britain	Quantitative	1940 individuals representative of the British population	Cervical cancer	17.6% reported genetic factors as playing a role in the development of cervical cancer. Higher-educated individuals were more aware of genetics as a risk factor (23.3%) than lower-educated individuals (11.4%). Women were more aware than men (19.9% vs. 14.5%)
Wardle et al.[48]	Great Britain	Quantitative	3693 adults representative of the British population	Cancer	Awareness of genetic risk factor was relatively low compared with awareness of other risk factors: approximately 50% was aware of the genetic risk factor of breast cancer to approximately 22% for lung cancer
Welkenhuysen et al.[49]	Belgium	Quantitative	329 women from Flemish Belgium	Breast cancer	79% reported being aware of hereditary breast cancer

Table 3
Studies examining the general public's how-to and principles knowledge of genetic risk factors of multifactorial diseases

Study	Country	Design	Sample	Disease	Outcome
Bates et al.[24]	The United States	Qualitative	108 individuals from urban, suburban, and rural communities in Georgia were interviewed	Heart disease	Overall, participants perceived genetic predisposition to mean heightened but not absolute risk and result in an increased risk of becoming sick
Bottorff et al.[43]	Canada	Quantitative	761 women without family history and 260 women with a family history	Breast cancer	Knowledge was relatively limited (mean = 4 on a 0–7 scale). Women with a family history did not show superior knowledge compared with women in the general public
De Vries et al.[28]	The Netherlands	Quantitative	457 Dutch adults	Cancer	Although the majority of participants recognized that genetic risk factors increase, but do not determine cancer development, they overestimated the role of these factors (52%) and how they cause the development of cancer. Higher-educated individuals had more knowledge of how genetic risk factors cause cancer than lower-educated individuals
Donovan and Tucker[23]	The United States	Quantitative	220 women awaiting routine medical services	Breast cancer	Knowledge about how genetic risk factors affect risk (mean = 7 on a 0–14 scale) was less than knowledge about other risk factors (mean =12 on a 0–18 scale). Whites had more knowledge than African Americans (7.7 vs. 7.0, $P = 0.02$).
Henneman et al.[29]	The Netherlands	Quantitative	817 individuals from a Dutch consumer panel	Not specified	Most of the participants had adequate knowledge of genetic risk factors. Higher-educated individuals and men had more knowledge than lower-educated individuals and women
Inoue et al.[25]	Japan	Quantitative	1355 individuals representative of general population	Cancer	Participants accurately perceived genetic predisposition as resulting in heightened risk of developing cancer
McMenamin et al.[27]	Ireland	Quantitative	1250 women and 1105 men from the general population	Breast cancer	The majority of the participants accurately reported increased risk due to genetic risk factors (77% of the men and 92% of the women)
Mesters et al.[26]	The Netherlands	Qualitative	40 women and 9 men were interviewed	Cancer	Although participants recognized the increased risk due to genetic risk factors, their knowledge of the working mechanisms of genetic factors was (highly) inadequate

RESULTS

Awareness knowledge of genetic risk factors

According to Rogers' suggestion, the first phase of public health education should focus on increasing awareness of the existence of the genetic risk factors of multifactorial diseases. Several studies have examined public awareness of the genetic risk factors of many multifactorial diseases in a wide variety of countries (Table 2). Overall, these studies suggest that the general public is reasonably aware of the genetic risk factors of multifactorial diseases, with approximately 59% (range, 17.6–93.3%) of the sample being aware of the existence of genetic risk factors, although much lower[18] and much higher[19] proportions have also been observed.

Most studies, however, showed differences in public awareness of the genetic risk factors for different types of cancer. For instance, on average, approximately 60% was aware of the genetic risk factors for breast cancer, whereas some studies observed that only approximately 20% was aware of the genetic risk factors of cervical cancer. Because we were not able to identify more than one study examining public awareness of heart disease, environmentally induced conditions, and hypercholesterolemia, we were unable to draw any conclusions regarding public awareness of these conditions.

Several studies observed that awareness of genetic risk factors of multifactorial diseases is highly dependent on sociodemographic factors. For instance, individuals with low educational levels were less aware of genetic risk factors compared with highly educated individuals.[18,20–22] African Americans were less likely to be aware of genetic risk factors than whites,[20] and women were more aware than men.[18,22]

The studies reviewed here show a mixed view of public awareness of genetic risk factors of multifactorial diseases, with little over half the population being aware of the existence of such risk factors. The overall picture may, however, be somewhat poorer, because being aware of genetic risks factors does not guarantee a clear understanding of how they influence health.

How-to knowledge of genetic risk factors

Although public knowledge of genetics has been relatively little investigated,[16] a few studies have examined the public's knowledge of the genetic risk factors of multifactorial diseases (Table 3). With regards to how-to knowledge, the central question concerns whether the general public has practical knowledge of genetic risk factors and how they influence the risk of development of multifactorial disease. The few studies examining this type of knowledge show that the general public's knowledge seems limited. For instance, Donovan and Tucker[23] observed that the public has limited knowledge on how genetic risk factors influence health, especially when compared with other risk factors. However, the general public does seem to understand that having a genetic predisposition implies heightened, but not absolute risk.[24–27]

In line with the studies on the general public's awareness of genetic risk factors, these studies seem to suggest that knowledge of how the genetic risk factors of multifactorial diseases affect health is largely dependent on sociodemographic factors, such as ethnicity,[23] gender,[27] and level of education.[28,29]

Principles knowledge of genetic risk factors

Principles knowledge is the most complex form of knowledge, focusing on theoretical issues such as why genetic risk factors affect the development of multifactorial diseases in the way they do. Only one study has examined the public's principles knowledge: Mesters et al.[26] observed that although participants recognized the increased risk owing to genetic risk factors, their knowledge of the working mechanisms of genetic factors was (highly) inadequate. Because participants associated cancer with faulty cells, they were unable to distinguish between the genetic aspect of cancer and the "faulty cell." Mesters et al. concluded that the general public's principles knowledge is largely insufficient and superficial.

DISCUSSION

The present review shows that the general public has limited awareness knowledge of genetic risk factors as a cause of multifactorial disease and even less knowledge of how and why these factors affect health. On average, just over half the research sample was aware of the existence of genetic risk factors of multifactorial diseases (i.e., awareness knowledge). Furthermore, although the public seems to understand that having a genetic predisposition means heightened and not absolute risk, they seem to have limited how-to and principles knowledge of genetic risk factors.

We should note that the studies we reviewed were not conducted with Rogers' framework of knowledge in mind. Our analysis of the potential utility of the knowledge framework is thus limited to the pooled data. Although most studies did distinguish awareness from general knowledge, we argue that distinguishing knowledge into awareness, how-to, and principles knowledge allows for a more detailed picture of the general public's knowledge. The distinction between awareness, (practical) how-to, and (theoretical) principles knowledge depicts a knowledge continuum representing an increasingly complex form of knowledge. This continuum, ranging from being aware of the existence at one end to knowing the underlying mechanisms of genetics at the other end, allows us to better map public knowledge and enables us to set a threshold amount of knowledge the public ought to possess to accurately process new information. Unfortunately, we were not able to identify such a threshold, partly owing to the fact that the original studies were not conducted with the Rogers' framework in mind. Future research examining the general public's knowledge of genetic risk factors using Rogers' knowledge framework will be able to do just that. Moreover, the knowledge framework will also allow us to pinpoint flaws or caveats in public knowledge with more precision and subsequently develop public health campaigns to remedy such flaws. Indeed, our results suggest that the public's how-to knowledge is limited, whereas the public's principles knowledge is largely insufficient; a finding that would not have been observed using only the awareness-knowledge distinction.

Moreover, recent research has shown that individuals implicitly make a distinction between practical how-to knowledge and theoretical principles knowledge.[30,31] Several studies have shown that individuals were only interested in the consequences of genetic risk factors and how to manage them, and were not at all interested in the underlying genetic principles.[30] Similarly, the public has no need for detailed information on the working mechanism of DNA, but is interested in more practical knowledge of the role of genetics in the development of cancer (e.g., identifying the signs of a genetic predisposition to cancer).[31] Such general lack of interest in the theoretical mechanisms of genetic risk factors until such knowledge is relevant[26] (e.g., when diagnosed with a family history) may also explain why the general public's principles knowledge is largely insufficient. More importantly, however, these issues suggest that the knowledge framework is a useful tool to understand the general public's knowledge of genetic risk factors.

Perhaps unsurprisingly, awareness, how-to, and principles knowledge seem to be largely dependent on sociodemographic factors, such as ethnicity, gender, and level of education. Ethnic minorities and lower-educated individuals seem to have less knowledge than whites and higher-educated individuals.[32]

Although these results do not add to the existing literature, they do highlight the resulting caveat in current public health promotion practices. More specifically, although the dissemination of genetic information to the general public has received considerable attention in the last decade,[3] the present results on the relationship between sociodemographics and knowledge suggest that professional knowledge on genetic predispositions may not yet have been equally disseminated. Our results suggest that the diffusion of genetic knowledge should especially focus on ethnic minorities and the lower-educated segment of the population, because it is these which seem to lag behind.

The translation of genetic knowledge from the professional to the layman, however, "goes beyond mere dissemination and/or diffusion."[33] Knowledge translation is a complex process, which is, at least partly, due to the complexity and incompleteness of genetic knowledge itself. Indeed, even among health educators[34] and medical experts[35–37] knowledge of genetics is surprisingly low. Consequently, it may be argued that the observed differences in knowledge between lower- and higher-educated individuals may not only be due to incomplete dissemination, but also to the complexity of the disseminated information. Indeed, lower-educated individuals have more difficulty understanding genetic information than higher-educated individuals,[38] which can also be seen from our results. Moreover, this perceived difficulty of genetic information may discourage lower-educated individuals from even attempting to process genetic messages.

Because our review suggests that ethnic minorities and lower-educated individuals are the least knowledgeable and have more difficulties understanding genetic information, we suggest special attention be paid to both the dissemination of genetic knowledge to these segments and their abilities to understand genetic information. One possibility would be to simplify the information without changing its meaning. However, because other factors (e.g., the relevance of the information) beyond its complexity may also influence behavior or decision-making, we first need to understand the different ways by which the public processes such information.[33]

Recent research in this area suggests that the public has at least some understanding of genetics in general. For instance, Lanie et al.[39] observed that the public seems to understand to some extent the gene-environment interaction underlying the development of multifactorial diseases. Alarmingly, however, they also observed general confusion about basic terms such as "genes" and "genetics," and argued that such confusion may adversely impact public understanding of more complex genetic messages (also see the 2002 World Health Organization report on *Genomics and World Health*[2]). These misconceptions may also explain our finding that the public's how-to and principles knowledge is limited and superficial. Indeed, research has shown that misconceptions about basic genetic terms may lead to incorrect processing of new information about genetic risk factors for multifactorial diseases, especially when individuals are unaware of their misconceptions.[40,41]

These issues raise the question of whether informing the general public about genetic risk factors of multifactorial diseases is an effective strategy in promoting public health. Although knowledge of such factors can potentially serve as a cue to action or otherwise motivate individuals to engage in preventive behavior, it may not contribute to preventive behavior over and above knowledge of other risk factors in high-risk individuals.[42] Furthermore, for previously unaware individuals, information on the existence of genetic risk factors actually decreases perceived susceptibility, and as a result, leads to lower motivation to engage in preventive behavior (unpublished data, 2007). In promoting public health, one should therefore not focus on increasing awareness knowledge alone, which may produce adverse effects, but at the same time increase understanding of how and why genetic risks influence the development of multifactorial diseases.

Unfortunately, our results suggest that increasing awareness knowledge is the outcome of recent public health campaigns. In other words, although the general public has reasonable awareness knowledge, they lack adequate how-to and principles knowledge. On the one hand, previous campaigns may have neglected the more complex how-to and principles knowledge. On the other hand, these campaigns may have tried to address such knowledge, but failed because of existing misconceptions about genetics. Both explanations suggest that future campaigns should first determine the target group's prior knowledge, and then build upon it to increase how-to and principles knowledge, ensuring accurate interpretation of the new information. Although the public may be primarily interested in how-to knowledge,[30,31] we do suggest not neglecting principles knowledge. Indeed, Seemann[50] suggested that education should not only focus on the *know-how* of a subject, but also on the *know-why*. For present purposes, having accurate knowledge of the underlying mechanisms or principles knowledge substantially increases, but is not necessary to, the proper use of how-to knowledge in decision-making processes and preventive behavior.[9] Moreover, individuals seem interested in principles knowledge when they deem it personally relevant[26] and should thus be available. Accurate principles knowledge may also allow for the transfer of this knowledge to other health issues.

Advances in genetic science have provided public health promoters with information about genetic aspects of multifactorial diseases and consequently enabled them to use this information in educating the general public. However, public health promotion has a long way to go concerning public understanding of genetic risk factors and why these factors increase, but do not determine, the risk of developing multifactorial diseases. For now, we may need to mark time and ask ourselves how to effectively educate the general public about the genetic risk factors of multifactorial disease. On the one hand, the complexity of genetic information may lead to misunderstanding and misconceptions which, in turn, may result in the incorrect processing of new information. On the other hand, learning of the existence of genetic risk factors may not only have no effect on preventive behavior, it may indeed have

adverse effects. Furthermore, too much media attention to genetic risk factors can lead to an overestimation of the impact of such factors,[28] and consequently to an underestimation of the impact of lifestyle on disease development. Such misconceptions may reduce the public's motivation to change their lifestyles in response to information about genetic risk factors.

To conclude, the knowledge framework as suggested by E. M. Rogers proved to be a useful tool in broadening our insight into public knowledge of genetic risk factors. It allowed us to differentiate between knowledge, which grows more complex by degree, in gradual stages. Future research may use this knowledge framework to provide a more detailed overview of the general public's knowledge of genetic risk factors. As genetic knowledge was lower among ethnic minorities and the lower-educated segment of the population, we suggest detailed attention be paid to both the dissemination of genetic knowledge to these segments and their abilities to understand genetic information. However, before using genetic information in public health promotion, we need to understand how such information is processed, how it influences subsequent decision-making, and whether it effectively motivates individuals to engage in preventive behavior. Once we have identified the processes through which knowledge of genetic risk factors successfully influences preventive behavior, information about genetic risk factors, as determined by Rogers' knowledge framework, will be a valuable asset to the public health promotion repertoire.

ACKNOWLEDGMENTS

This study was financially supported by Maastricht University. It was performed at the School for Public Health and Primary Care (Caphri), a part of the Netherlands School of Primary Care Research (CaRe), which was acknowledged by the Royal Netherlands Academy of Arts and Sciences (KNAW) in 1995. We thank Janaica Grispen for valuable comments on an earlier draft of this article.

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