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Bringing the Social into Genetics: The Psychosocial Genetics Risk Assessment and Management Framework (PG-RAM)

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Abstract As increasing numbers of people are identified at risk for multi-factorial diseases, questions of how to assess, communicate and manage genetic risk will be critical from health services and policy perspectives. However, there is currently no evidence-based genetic risk assessment and management framework to assist policy makers, clinicians and other stakeholders. A comprehensive psychosocial framework for risk assessment and management has been developed in the context of security hazards or threats. In an adaptation of that model, we present the Psychosocial Genetics Risk Assessment and Management framework (PG-RAM). It offers principles to enhance the integration of evidence-based best practices into genetics health services, as well as to identify issues, knowledge and gaps. The framework identifies the core elements of the situation, effects, population and interventions, all spanning several phases of genetic disorders. The framework provides an excellent starting point for knowledge syntheses in the context of genetic risk and could serve as the conceptual basis for practical tool development to guide healthcare professionals and decision makers in preparing for and responding to the psychosocial aspects of genetic risk.

Keywords Genetics · Psychosocial · Risk management · Risk assessment

Introduction

The traditional focus of medical genetics has been single-gene disorders such as Huntington disease (HD) or chromosomal disorders such as Down syndrome (DS), accompanied by a focus on non-directive counseling regarding decision-making around genetic testing reproductive issues (Khoury 2003). The sequencing of the human genome, however, will allow identification of genetic variants at multiple loci

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that increase or decrease risks for a variety of common diseases (Khoury 2003). There is evidence that several common diseases (e.g., cancer, heart disease, diabetes) have important hereditary influences, and "it is expected that predictive genetic tests will be available for as many as a dozen common conditions, allowing individuals... to learn their individual susceptibilities and to take steps to reduce those risks for which interventions are or will be available" (Collins and McKusick 2001; p. 543).

The extension of 'risk' to larger portions of society presents many societal and health system questions (Khoury 2003; Wilson 2006). There are frameworks for the assessment of genetic tests (Haddow and Palomaki 2004) and clinical guidelines are emerging in relation to some patient groups (McIntosh et al. 2004; Trepanier et al. 2006). However, while an extensive literature documents psychosocial effects from a largely clinical or individual patient perspective (Heshka et al. 2008), the wider social aspects of genetic risk information have not been so fully considered in policy deliberations.

In a very different context, Lemyre and colleagues proposed a comprehensive psychosocial risk assessment and management (P-RAM) framework for terrorist threats and attacks (Lemyre et al. 2008). Since the psychosocial aspects of terrorism are often overlooked in current terrorism preparation and planning efforts, such a tool reminds actors of the direct and indirect effects of any intervention on behaviors and cognitions. Too often, detection, containment and short-term consequence management of the physical hazard are emphasized and wider or longer-term consequences neglected (Lemyre et al. 2008). Similarly, genetic health services also tend to focus on the detection and management of genetic risk, to the exclusion of a myriad of longer-term psychosocial effects (Heshka et al. 2008). We suggest the P-RAM framework might provide a useful platform for the consideration of the broader psychosocial aspects of genetic risk. We briefly describe the P-RAM and then consider how it might be adapted to the assessment and management of the psychosocial aspects of genetic risk.

Psychosocial Risk Assessment and Management Framework (P-RAM)—A Brief Overview

The P-RAM framework for security threats and events adopts a multilevel approach to risk management. It stresses the dynamic interaction between aspects of the individual, community and society, as well as aspects of the disaster event itself as mediators of the psychosocial responses to a crisis event. Event characteristics (e.g., suddenness of event) interact with preexisting individual factors (e.g., previous experience with crisis events), as well as community and societal factors (e.g., organizational structures and resources) to increase or decrease negative psychosocial effects. Notably, the P-RAM framework recognizes distinct time phases of threats and events (e.g., preparedness and planning, threat, impact and recovery) which may necessitate different risk management responses (Lemyre et al. 2008). The framework represents a shift from reactive to proactive emergency management: Understanding the ways individuals perceive and respond to a threat prior to the occurrence of an event could illuminate policy approaches that would promote preparedness and foster resilience among individuals and communities.

Key Elements of the P-RAM Framework

At its simplest level, the P-RAM framework includes three key elements (situation, population and intervention) that influence the risk of positive or negative psychosocial outcomes (effects). Recall that event characteristics and individual and community factors are also thought to influence the psychosocial outcomes that follow a terrorist threat or attack. They may be risk factors (e.g., lack of prior experience with the event) or protective factors (e.g., availability of coping resources). The following provides a brief description of the four key elements of the P-RAM framework. More detailed descriptions can be found in Lemyre et al. (2008).

Element 1: The **situation** element describes aspects of the hazard (real and perceived), its vector and the agent. Situational characteristics might include type of hazard, duration of attack or number of casualties. These characteristics mediate the severity of subsequent psychosocial effects. The situation is postulated to be dynamic; therefore, the types of psychosocial effects and required interventions will change over the event timeline. In the P-RAM model, the event timeline is conceptualized as a series of continuous phases including preparedness and planning, threat, warning, impact, rescue, recovery and reconstruction. This conceptualization allows psychosocial effects and interventions to be identified for each phase in the event timeline.

Element 2: The **population** element refers to the individuals, groups or communities targeted. The framework suggests that populations will vary in their degree of vulnerability to psychosocial effects. For example, the P-RAM framework identifies children, the elderly and first responders as potentially vulnerable subgroups that may require specific psychosocial interventions.

Element 3: **Effects** refer to both 'normal' (e.g., adverse effects, protective behaviors) and 'abnormal' (e.g., identified by DSM-IV or other effects such as family violence) psychosocial reactions. Normal psychosocial adverse effects are further categorized as behavioral (e.g., increased use of alcohol), cognitive (e.g., disbelief or poor concentration), spiritual (e.g., changes in one's belief in God), emotional (e.g., anxiety, fear, shock), social (e.g., social withdrawal) and physical (e.g., sleeping difficulties, headaches). Normal psychosocial benefits, such as resilience, altruism, or a greater sense of self worth, are also possible.

Element 4: The **interventions** element includes both psychosocial interventions, whose aim is to prevent negative psychosocial effects and promote active coping, as well as bioenvironmental interventions, whose aim is to protect public health (e.g., minimize exposure). The P-RAM framework includes interventions at the individual, organizational and community levels. Each of these is further divided into interventions related to risk communication, education, social support and professional counseling.

P-RAM summary The P-RAM framework offers a distinct and structured paradigm for anticipating and responding to the psychosocial effects of threats and events. Currently, the research team is testing several practical tools based on the framework (e.g., risk communication tutorials) to guide decision makers and first responders in preparing for and responding to the psychosocial aspects of a terrorist event. Because of its multi-level, evidence-based approach, we suggest that the P-RAM tool can be adapted for use in genetics health services and policy-making.

Psychosocial Aspects of Genetics

Following the P-RAM framework, the PG-RAM framework adopts a multilevel approach to genetic-risk management. It recognizes the dynamic interaction between aspects of the individual, community and society, as well as aspects of the genetic risk itself as mediators or moderators of the psychosocial response to genetic risk. Table 1 displays some of the risk factors that might affect the psychosocial outcomes in response to genetic risk. Event characteristics (e.g., disease severity, controllability of the risk) interact with preexisting individual factors (e.g., coping style, experience with disease), as well as community and societal factors (e.g., norms about disability) to increase or decrease negative psychosocial effects. The PG-RAM framework also recognizes that the risk management response to genetic risk (e.g., genetic counseling) may affect psychosocial outcomes in both positive (e.g., improved risk understanding) and negative (e.g., family tension) ways. Like the management of terrorism risk, the PG-RAM framework also delineates several temporal phases of genetic risk (e.g., pre-symptomatic phase to illness phase). The time phase is notable since different interventions may be required at different phases of the risk experience (Lemyre et al.

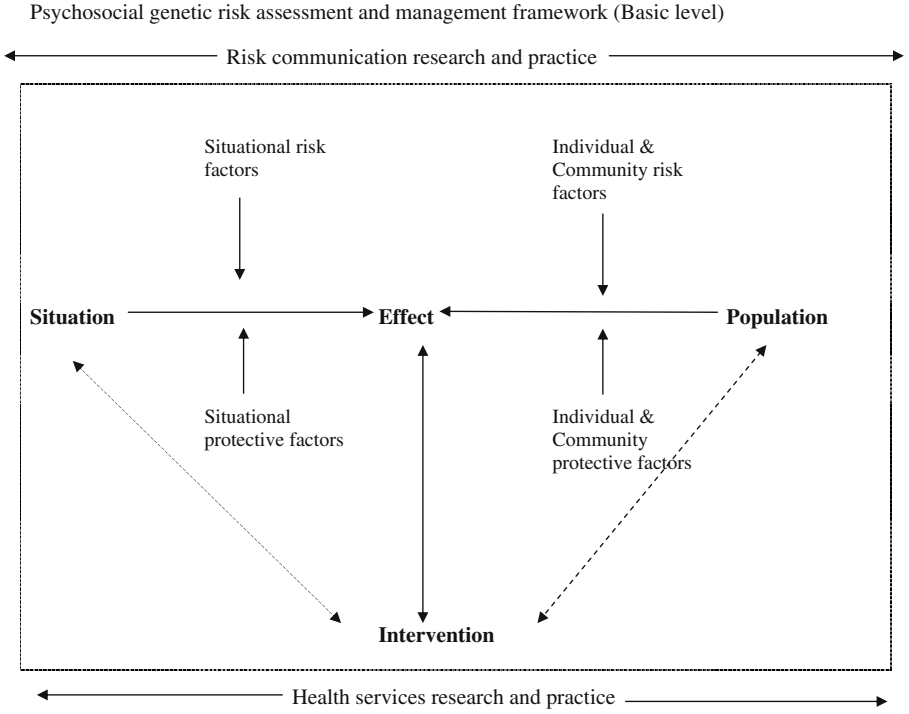
Table 1 Multilevel risk factors in the assessment of psychosocial responses to genetic risk

Event (i.e., risk) features	Individual characteristics	Family characteristics	Community/societal features
-Amount of uncertainty reduction	-Health status	-Communication style of family (open/closed)	-Availability of community resources (e.g., support groups)
-Test utility (e.g., likelihood of disease following positive test result)	-Need for cognition	-Geographic and/or Emotional closeness	
-Disease severity	-Tolerance for ambiguity	-Number of affected relatives	-Social norms regarding disability
-Possibility of control	-Demograhpics (age, gender, education)	-Pattern of disease expression in family	-Social norms about responsibility for health
-Method of discovering one's risk	-Prior experience with genetic illness		-Access to testing services
	-Availability of social support or other coping resources		

2008). As in the P-RAM framework for terrorist threats, the PG-RAM framework represents a proactive approach to the management of the psychosocial effects of genetic risk. The framework can assist in the development of genetic risk management strategies not only by identifying psychological and behavioral impacts of such risk, but also by informing the design of interventions aimed at improving coping strategies and responses to a genetic health threat. This is important since the current standard of care in genetics health services includes many components for which there is little to no evidence (either of benefit or harm) (Wilson 2006).

Key Elements of the PG-RAM Framework

The PG-RAM framework is illustrated in its most minimal form in Fig. 1. At this basic level, there are three major elements—situation, population and intervention—that influence genetic psychosocial outcomes (effects). This framework emphasizes a key relationship between psychosocial effects and psychosocial interventions, and also between the population(s) and the intervention and between the situation and the intervention. As such, the PG-RAM framework recognizes that different populations (e.g., at risk children, family physicians) may require different interventions (e.g., professional counseling, risk communication instruction). It also



Adapted from Lemyre et al. (2005)

Fig. 1 Psychosocial genetic risk assessment and management framework (Basic level). Adapted from Lemyre et al. (2008)

allows for different interventions depending on characteristics of the situation (e.g., disease severity). Note that both risk communication and health services (research and practice) are assumed to be essential and ongoing throughout the entire genetic risk event. It is also assumed that the outcomes of risk communication and health services will themselves influence the psychosocial outcomes (e.g., risk communication should influence genetic risk perception). The following sections will describe the key elements in more detail and review some empirical evidence in each area.

The Situation

The situation element describes aspects of the genetic risk (real and perceived) and the subsequent availability of genetic testing. Important characteristics of the risk might include disease severity, whether something can be done about the risk (e.g., surgery, drug management), the amount of uncertainty reduction provided by the genetic test, as well as the availability of genetic counseling or other genetic health services. These factors mediate the expected psychosocial effects by acting as risk or protective factors. For example, if a genetic test does not reduce the amount of perceived uncertainty associated with the genetic risk (e.g., inconclusive results of BRCA 1 or 2 testing), negative psychosocial effects might follow testing (e.g., elevated levels of stress) (Baum et al. 1997). If, however, there is some possibility of control following testing (e.g., colonoscopy in the case of inherited colon cancer), positive psychosocial effects might be expected (e.g., adherence to colon cancer screening guidelines) (Hadley et al. 2004).

There are two particularly notable aspects of the situation element in the PG-RAM framework: 1) Perceived genetic risk, and 2) Genetic testing interest and uptake. First, subjective genetic risk perception is itself an important characteristic of the situation element since it can affect a variety of psychosocial outcomes such as testing uptake, psychological distress, controllability beliefs and protective health behaviors such as screening (Cameron and Diefenbach 2001; Codori et al. 1999; Heshka et al. 2008; Lerman et al. 2002). There is now a large literature on genetic risk perception for a variety of disorders, although measurement problems and inconsistent research findings make it difficult to pinpoint the exact effect of perceived risk on a variety of psychosocial outcomes (Croyle and Lerman 1999). There are also gaps in our knowledge about perceived risk, particularly in those at risk people who decline testing. For example, Binedell and Soldan (1997) noted that little is known about those who decline or never request testing for Huntington disease (HD), even though they are in the majority. Thus, we have little knowledge about how these individuals perceive and cope with their risk or whether they need (or have access to) a variety of genetic health services (e.g., testing, family or group counseling, or other forms of therapy). This is a good example of necessary health services research identified in Fig. 1.

Qualitative risk perception research has identified several psychosocial impacts of genetic risk information. For example, Hallowell (1999) interviewed women at risk for breast and ovarian cancer and found that risk was largely infused with moral meaning. Women felt they had a responsibility to determine their own genetic risk, encourage other family members to do the same and engage in some form of risk management. Other research demonstrates that risk perception is not static; rather, at

certain life junctures, genetic risk became salient (e.g., nearing the age of onset of a relative with cancer) (Kenen et al. 2003). Cox and McKellin (1999) also found that risk for HD became salient at critical moments (e.g., a diagnosis of HD in a parent or sibling), but otherwise was accorded less importance in everyday life. These investigations of risk perception are enlightening because they highlight a variety of psychosocial aspects of living with genetic risk. Thus, the PG-RAM framework suggests a thorough exploration of genetic risk perception in risk management responses. The National Society of Genetic Counselors concurs, highlighting the importance of exploring perceived cancer risk in its counseling guidelines (Trepanier et al. 2006).

A second important aspect of the situation element is the availability of genetic testing subsequent to the discovery one is at risk for a genetic disorder. A host of demographic, psychological and social variables have been identified as influencing the decision, or at least the intent, to have a predictive genetic test. In the case of HD, for example, planning for the future, reducing uncertainty and informing children were among the most common reasons given for testing (Meiser and Dunn 2000). Interest in BRCA1 or 2 testing was related to perceived risk, with those perceiving a higher risk more likely to intend to be tested (Lerman et al. 2002). Commonly cited reasons for BRCA testing also include wanting to know one's children's risk and to increase protective screening behaviors (Struewing et al. 1995). Family history of the disease was a predictor of testing interest both for BRCA (Donovan and Tucker 2000) and prostate cancer (Cormier et al. 2002). High levels of cancer-related worry predicted intention for colorectal cancer testing (Codori et al. 1999) and BRCA1 (Cameron and Diefenbach 2001). Comparatively, disease-specific distress appears to *deter* testing interest for HD (Lerman et al. 2002).

Predictive genetic testing is a fairly recent medical option, and these (largely) descriptive studies are useful in providing an overview of the area and in identifying key predictor and outcome variables. Research on genetic testing interest is also informative since it is used to guide the evolvement of genetic services and to specify health policy and funding allocations (Bottorff et al. 2003). In the PG-RAM framework, this research is crucial for preparedness and planning of genetics health services. Also important in this regard is the public's knowledge and attitude towards genetic testing.

The public's factual knowledge of heredity and genetic testing is limited (Henderson and Maguire 1998; Singer et al. 1998). However, virtually all studies report high levels of interest in having a genetic test, whether using population-based, general public samples (Morren et al. 2007; Sanderson et al. 2005) or high-risk samples (Cormier et al. 2002; Struewing et al. 1995). Testing interest was high for virtually all types of genetic tests, whether Huntington disease, breast/ovarian cancer, colon cancer and even bipolar disorder, for which no predictive test currently exists (Etchegary 2004; Jones et al. 2002; Sanderson et al. 2005). Actual test uptake in at risk samples has deviated, however, from early studies of testing interest. For example, uptake rates for genetic tests are higher when an effective treatment or prevention strategy exists (Marteau and Croyle 1998): On average, the uptake rate for HD (for which no cure or prevention exists) is roughly 10–15%; for breast cancer (for which there may be some possibility of prevention or treatment), it is roughly 50%; for familial adenomatous polyposis (FAP; for which there is effective treatment), the uptake rate is roughly 80%.

There is generally a positive attitude towards genetic testing. The Eurobarometer survey on biotechnology (Gaskell et al. 2000) found strong public support for genetic testing across virtually all of Europe. Within Canada, the overarching response to biotechnology was "cautiously optimistic" (Sheehy et al. 1998); applications involving health (e.g., genetic testing for disease) were more acceptable than cosmetic applications (e.g., improving food's taste). Our more recent research also found positive attitudes towards genomics, particularly genetic testing to determine disease risk (Etchegary et al. 2010). Attitudinal research such as this is informative as it provides a snapshot of current public opinion and identifies public attitudes and values, which must be considered by policymakers as they grapple with regulatory issues in this area.

As the foregoeing suggests, there are a number of important characteristics that must be considered in the situational element of the PG-RAM framework. It is also suggested that these characteristics may change as the situation unfolds, and as such, the type of psychosocial effects and required interventions will also evolve over the crisis timeline. In the context of genetic risk, there are also a variety of temporal phases, each with their own psychosocial effects and demands.

Expanding the Concept of Time

Street and Soldan (1998) provided a conceptual framework of the range of psychosocial issues faced by families with genetic illnesses based on Rolland's classic work in chronic illness. Rolland (1994) identified five elements in relation to chronic illness that present a variety of psycho-social demands: onset, course (i.e., timeline), outcome, incapacitation and amount of uncertainty. The second of these elements, time phase (course) of the illness (including the crisis, chronic, and terminal phases), is not sufficient to account for the *pre-illness phase* of some genetic illnesses (Street and Soldan 1998). This phase is especially relevant for at risk individuals and for test candidates with a positive genetic test result since no physical manifestations of the disease are observable during this phase.

Subsequent to Street and Soldan's (1998) framework, Rolland (1999) later distinguished between the *pre-symptomatic* (1) pre-crisis, (2) crisis, and (3) chronic phases in genetic illness. The former refers to life before a genetic test is available or even considered by family members. At this time, however, families could already know about the history of genetic illness in the family and members can have strong beliefs about their own vulnerability to the illness. The pre-symptomatic *crisis* phase begins for many families when a predictive test actually becomes available or when members actively consider taking the test. This phase extends to the entire decision making process and subsequent to the test. Finally, Rolland (1999) suggested that the pre-symptomatic *chronic* phase is similar in many respects to the chronic phase of living with chronic illness, and can extend over a person's lifetime.

In later writing, Rolland and Williams (2005) clarified the timeline of genetic disorders. Rolland and Williams distinguished the nonsymptomatic and postclinical onset phases of genetic illness, each with subphases and attendant psychosocial demands of its own. Figure 2 displays the time phases of genetic disorders (adapted from Rolland & Williams). The Nonsymptomatic Awareness phase generally

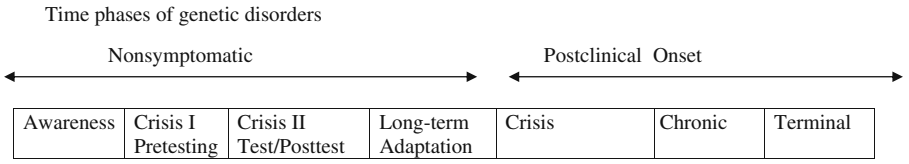


Fig. 2 Time phases of genetic disorders. Adapted from Rolland and Williams 2005

includes some knowledge of genetic risk in the family, but there has been no active consideration of testing by family members. However, some family members may be concerned about future illness, termed anticipatory loss. In this phase, the major psychosocial demand may be living in fear of future illness.

In contrast, in Crisis Phase I (Pretesting), family members actively consider testing and the psychosocial ramifications of this decision for themselves and other members of the family. Rolland and Williams (2005) suggest that the decision-making in this phase may not be a one-time event. People may decide to postpone testing, in which case they move back to the awareness phase. Crisis Phase II (Testing and post-testing) includes the testing and the early posttest period. In this phase, there are a variety of psychosocial outcomes and challenges to be met by families:

> (1) acknowledging and accepting the "permanence" of the genetic knowledge and its implications, (2) grieving losses or changes in personal or family identity, (3) creating meaning about the genetic information that preserves a family's sense of mastery and competency, and (4) developing family flexibility in the face of future uncertainty and loss to maximize preservation of key life cycle goals (Rolland and Williams 2005; p. 13).

Finally, the Nonsymptomatic Long-term adaptation phase incorporates the timespan between a positive test result and clinical onset of the illness. This phase can be short or it can extend until the end of life if the genetic illness does not emerge. Like the other phases, a variety of psychosocial outcomes and challenges attend this phase (e.g., minimizing relationship skews between affected and unaffected family members.

Similar to Lemyre et al.'s (2008) P-RAM framework for public safety threats, distinguishing the time phases of genetic disorders encourages a longitudinal view that constructs genetic risk as an ongoing process with landmarks, transitions and evolving psychosocial demands. This view fits well with the PG-RAM framework for genetic risk. Notably, the concepts of nonsymptomatic and clinical onset phases have implications for health services delivery and represent a shift in current genetic risk management practices. As Rolland and Williams (2005) noted, psychosocial interventions are typically offered at the crisis phase of clinical symptom onset. However, a longitudinal view of genetic disorders:

> ...suggests the value of periodic assessment and reevaluation of individuals, couples and families in relation to both the nonsymptomatic and symptomatic phases of a condition. The time phases and transition points can inform the timing of psychosocial consultations. p. 18.

The Effects

Following the P-RAM, effects are categorized into normal and abnormal psychosocial effects that occur as a result of genetic risk: 1) normal psychosocial effects, which include both adverse effects and positive reactions, and 2) abnormal psychosocial effects, including those disorders identified by DSM-IV and other effects such as family breakdowns. Lemyre et al. (2008) further categorize normal psychosocial effects as behavioral, emotional, cognitive, social, physical and spiritual. We follow this classification in the PG-RAM as well, adding family effects as a category. Table 2 displays possible normal and abnormal psychosocial effects identified in the PG-RAM.

The genetics literature is replete with studies and reviews documenting the psychological impact of genetic risk information for a variety of disorders (Broadstock et al. 2000), notably inherited cancers (Cameron and Diefenbach 2001; Codori et al. 2005; Heshka et al. 2008) and HD (Meiser and Dunn 2000). Regardless of type of disorder, the findings were very similar: Few negative psychological effects (e.g., increased anxiety or depression) were observed following genetic testing in both carriers and noncarriers, at least in the short term (Heshka et al. 2008). Even in HD, a fatal genetic disorder, anticipated psychiatric problems (e.g., suicide) have rarely materialized (Almqvist et al. 1999). However, very small subgroups of the tested population (HD and inherited cancers) sometimes show short-term increases in post-test distress.

Meiser (2005) reviewed empirically-derived risk factors for psychological distress subsequent to genetic testing. These included: high pretest levels of distress, prior history of depression, individual coping style (e.g., information monitors more likely to be distressed while awaiting results) and having lost a relative to hereditary cancer among others. As Meiser noted, these findings highlight the importance of pre-test counseling to encourage test candidates to anticipate emotional reactions to test results. Additionally, screening for risk factors could be incorporated into clinical practice, thereby identifying those test candidates that may need additional support following testing (Broadstock et al. 2000; Meiser 2005).

The focus of extant literature is on the clinical outcomes of testing with far less on the behavioral and cognitive outcomes (Heshka et al. 2008; Meiser 2005). A recent meta-analysis and systematic review concluded that while prospective studies reported improvements in risk accuracy following counseling, controlled trials showed no difference in the level of perceived risk post-counseling (Braithwaite et al. 2006). Meta-analysis of controlled trials did show increases in knowledge of cancer genetics, however.

Regarding behavioral outcomes, Meiser (2005) concluded that the majority of carriers and non-carriers do adopt recommended screening and preventive behaviors following genetic testing for BRCA 1 or 2, HNPCC and FAP (i.e., breast and colon cancer mutations, respectively). However, a more recent systematic review found that behavioral outcomes were investigated in only a few studies and small effects were observed regarding breast self exam, clinical breast exam and mammography (Braithwaite et al. 2006). Very few studies explored posttest behavior in non-carriers. Lerman et al. (2000) found no changes in breast cancer screening uptake in non-carriers subsequent to BRCA testing. One study found a minority of non-carriers

Table 2 PG-RAM framework of psychosocial effect categories (adapted from Lemyre et al. 2008)

Normal Psychosocial Effects									Abnormal Psychosocial Effects
Behavioral	Family	Emotional	Cognitive	Social	Physical	Spiritual	Psychosocial Benefits	Disorders	Others
Increased use of tobacco or alcohol	Changing roles for family members (e.g., from spouse to caregiver)	Anxiety/fear	Risk perception	Alienation	Sleep disturbance	Finding meaning in illness	Resiliency	Acute stress disorder	Effects not listed in DSM-IV such as family breakups
Adherence to screening advice (e.g., mammography)	Family identity	Feeling stigmatized	Genetics knowledge	Social withdrawal	Change in appetite	Changes in one's belief in God	Adaptive coping	Suicide attempts	
Prophylactic surgery	Communication among members	Shock	Disease knowledge	Increased conflict within relationships	Headaches		Family cohesion	Major depression	
Life choices (e.g., marriage, reproduction, career)	Pre-selection of family member thought to be the one to develop the family illness	Guilt or blame	Coping appraisal	Impaired capacity to work	Somatic complaints		Greater appreciation for life	Generalized anxiety disorder	
Symptom watching		Anger	Beliefs about luck or fate	School impairment	Gastro-intestinal problems				
		Depression							
		Helplessness							

who was not reassured by the negative test result and expressed a need for continued careful screening (Lim et al. 2004). One woman in their sample had even undergone prophylactic mastectomy. Thus, there may be a subgroup of non-carriers who is not reassured by negative results raising the question of appropriate interventions for this group.

We have provided only brief descriptions of the emotional, cognitive and behavioral effects of genetic-risk information. Table 2 displays other important effect categories in the PG-RAM (e.g., social, family). We note there is virtually no literature on the physical or spiritual outcomes following genetic testing, and very limited literature on the social and family effects. Use of the PG-RAM framework allows the identification of knowledge gaps such as these.

The Population

In the PG-RAM, the population element specifies important subgroups for whom genetic risk may have varying psychosocial effects. Test candidates, both testing positive and negative, are obvious important populations, and the bulk of extant literature focused on these populations. However, current literature has noticeable gaps in knowledge about certain populations: 1) at risk people who decline the offer of testing or who have no contact with genetics clinics; 2) individuals who request testing, but are denied for failing to meet eligibility requirements (Lee et al. 2005); 3) test candidates whose results are inconclusive or whose mutation is of unknown clinical significance (Meiser 2005), and 4) spouses and children of test candidates. Other important populations include family physicians, who increasingly are being asked about genetic testing by their patients and generally decide who is referred to specialist genetic services (Wilson 2006). Limited research shows that general practitioners do not feel knowledgeable about most genetic disorders, raising the question of whether risk communication education might be necessary for this group (Bottorff et al. 2005).

The family as an important subgroup is also identified. Despite the obvious implications of genetic risk for all family members, very few studies adopt a family perspective from which to study the psychosocial impacts of genetic risk (Rolland and Williams 2005).

Interventions

The final element in the PG-RAM framework aims to identify interventions for various levels of the population that prevent negative psychosocial outcomes (e.g., anxiety) and encourage positive psychosocial responses (e.g., increase health protective behaviors, support family communication). The PG-RAM framework suggests four primary levels of psychosocial interventions: the individual, family, organizational (e.g., healthcare professionals) and society. As in the P-RAM, these categories may be subdivided into psychosocial interventions related to: 1) risk communication, 2) education, 3) social support, and 4) professional counseling.

There are few intervention studies in the genetic risk context, and most have been at the individual level in the context of genetic counseling (Braithwaite et al. 2006; Lee et al. 2005). Both reviews concluded that genetic counseling improved

knowledge of cancer genetics without an adverse effect on worry or anxiety; some evidence suggested that counseling improved perceived risk of cancer. However, there are theoretical and methodological problems with the relatively few interventions to be found in existing literature. Braithwaite et al. (2006) noted that the specific components of a genetic counseling intervention were not fully described in some studies, making it difficult to precisely describe and evaluate interventions. They also noted that more than half of the interventions reviewed were evaluated in a single clinic with only a few healthcare professionals involved in their delivery, thus limiting external validity.

These interventions were all at the individual level, and there are obvious gaps in our knowledge at this level. We are unaware of any psychosocial interventions at the family, healthcare professional or community levels; however, interventions appear to be necessary. For example, Paling (2003) noted that while almost every physician will need to communicate about risk with his or her patients, few physicians had any risk communication training. Risk communication is paramount in the context of genetic disorders, and family physicians are normally responsible for referring their patients to specialist genetics services (Wilson 2006). However, there is very little research which investigates the attitudes, knowledge and practices of general practitioners in the context of genetic risk. The little work that is available, however, suggests that physicians in general practice have limited knowledge about genetics and have low levels of confidence in their ability to provide genetics health services (Bottorff et al. 2003; Emery et al. 1999). However, Canadian physicians and nurses reported being involved in caring for people at risk for hereditary adult onset disease, and perceived important roles for their professions in providing genetics health services (Bottorff et al. 2003). Thus, educational interventions may be required for these healthcare professionals, and in Canada, some research is addressing this issue. For example, problem-based learning modules on hereditary cancer have been developed for use by family physicians in small group formats (Blaine and Carroll 2002). Generally speaking, however, there are very few interventions for healthcare professionals in the context of genetics.

We noted that interventions might also be required at the family and community levels. However, there are serious gaps in our knowledge in these areas which must be closed before interventions can be developed. For example, how does the public perceive at risk individuals? With what effects? What is the effect of genetic testing on the family system? How do families communicate about genetic risk? What sort of family interventions might be necessary? This type of knowledge synthesis is identified by the PG-RAM framework as necessary for the evidenced-based design of interventions (at any level).

Potential Applications of the PG-RAM Framework

While there are great expectations of the ability of genome-based technologies to promote individualized health care and effective disease prevention through improved risk prediction, the evidence base at the health system level has centered around a clinical model in which individual informed decision making is the central goal. Systematic reviews suggest that the risk of negative psychological consequen-

ces for individuals who decide to have genetic tests is very low (Heshka et al. 2008). However, the clinical model, which focuses on deliberation and individualization of decision-making, is inadequate as for examining the effects of applying genome-based technologies at a population level. Examples of population-oriented genome-based technologies include newborn screening programs (McCabe and McCabe 2008), direct-to-consumer marketing of genetic tests (McGuire et al. 2007), and the systematic application of family history taking in complex disease risk assessment (Qureshi et al. 2009). These examples share two important characteristics, namely that the genome technology in question is applied more or less uniformly to an entire target population and that the resources to support individualized deliberation and informed decision making are limited or absent. There is limited direct empirical evidence of the psychological impact of these emerging applications of genome technology, and it is unclear whether findings generated in clinical contexts can be extrapolated to the population context.

These issues will be explored as part of a 5 year research program funded by the Canadian Institutes of Health Research (CIHR) Emerging Team in Genomics in Screening (Lead PI: Wilson, Co-Is: Lemyre, Etchegary, along with other team members). This research program includes examination of newborn screening and family history taking, as well as public engagement with emerging genome technologies, in which the PG-RAM framework will be applied to generate empirical evidence of its usefulness in practice. The goal is to generate robust evidence to guide policy and practice decisions about the risks and benefits of population-based genomics interventions beyond clinical outcome measures.

Conclusion

As more and more people are identified at risk for multi-factorial diseases and direct-to-consumer genetic tests continued to be marketed to the public, questions of how to assess, communicate and manage genetic risk will be critical from both health services and policy perspectives. However, there is currently no evidence-based genetic risk assessment and management framework to assist planners, policy makers, clinicians and other stakeholders. Although adapted from a very different risk context (terrorism), the PG-RAM framework offers a mechanism to enhance the integration of evidence-based best practices into genetics health services, as well as to identify issues, knowledge and gaps. The framework takes a holistic view of the psychosocial elements of genetic risk, and identified the core elements of the situation, effects, population and interventions. The framework is flexible and accommodates a variety of risk and protective factors for psychosocial outcomes, different cultures and relevant populations, as well as a variety of interventions, all spanning several phases of genetic disorders. As with the P-RAM, the framework could provide the conceptual basis for practical tool development to guide healthcare professionals and decision makers in preparing for and responding to the psychosocial aspects of genetic risk. However, the PG-RAM promotes identification of knowledge gaps that must be addressed prior to intervention research or practical tool development. Thus, the PG-RAM framework provides an excellent starting point for required knowledge syntheses in the context of genetic risk. In presenting

the PG-RAM framework for consideration, we hope to encourage evidence-based genetics health care. That is, the conscientious use of current best evidence in decision-making about the new genetics at the clinical, administrative and policy-making levels.

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