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# Perceived Health Status, Health Values and Health Goals as Influences on Individual Pursuit of DTC Genome Testing: Implications for Healthcare of Ill & Healthy

Carolyn Marie Dachs Farrell  
*Clemson University*

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PERCEIVED HEALTH STATUS, HEALTH VALUES AND HEALTH GOALS AS  
INFLUENCES ON INDIVIDUAL PURSUIT OF DTC GENOME TESTING:  
IMPLICATIONS FOR HEALTHCARE OF ILL & HEALTHY

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A Dissertation  
Presented to  
the Graduate School of  
Clemson University

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In Partial Fulfillment  
of the Requirements for the Degree  
Doctor of Philosophy  
Healthcare Genetics

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by  
Carolyn Marie Dachs Farrell  
December 2014

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Accepted by:  
Dr. Bonnie Holaday, Committee Co-Chair  
Dr. Kelly Smith, Committee Co-Chair  
Dr. James P. Evans  
Dr. Janet Craig

## ABSTRACT

This dissertation examines an individual's perceived health status, and health values, as influences on self-initiated health action, specifically the pursuit of Direct-to-consumer (DTC) genome testing. Motivation for this independent health action may also have implications for other individual health behaviors, including the potential for positive changes that are lasting rather than temporary. Health Capability, the conceptual framework, supports individual health values and goals, ability to access valued healthcare services and includes measures of health status. However, Health Capability lacks an operationalized mapping of its major concepts. This dissertation proposes and evaluates an original concept mapping and relationships.

An overview of this dissertation and components is presented in Chapter one. Chapter two addresses the genomics and healthcare landscape related to independent individual pursuit of Direct-to-consumer (DTC) genome testing. The Health Capability framework is initially presented in this chapter, together with the research hypotheses, targeted literature review of key concepts and the proposed concept mapping with relationships. Chapter three describes the Health Capability conceptual framework, its adaptation and extension for the dissertation and its potential for use in health promotion and prevention research. A systematic review of the literature on perceived health status is discussed in chapter four, as well as a targeted review of approaches to concept measurement and most commonly used instruments.

Chapter Five presents the dissertation study. This research involves 1455 self-initiated Direct-to-consumer (DTC) genome testing users who represent a naturally

occurring phenomenon and provide a unique population to study the impact of an individual's subjective perceived health status, personal health values and health goals. Participants completed surveys as part of the Impact of Personal Genomics (PGen) study. These participants were classified into four mutually exclusive health status groups based on individual perception of health and a healthcare system perspective of health status (existence of a medical diagnosis). These groups were analyzed for the following: 1) discrepancies in health status, 2) differences in reasons for genome testing, perceived risk and health values, and 3) potential relationships among variables. Results confirmed discrepancies between individual and biomedical health status. One group may represent the “worried well”. Interest in health information was high (98–99%) across all groups, as was health value of genome test results. Two items distinguished all groups (interest in pharmacogenomics information and learning risk for other diseases ( $p < .001$ )). When groups differed, perceived rather than biomedical health status was often involved, yet both factors demonstrated influences (variable dependent). Risk perception was moderately correlated (.301) with health status group, yet clearly does not equate with perceived health. Persons with medical diagnoses and self-rated not good health scored highest for items relating to immediate personal health and for family. Study findings are consistent with individual perceived health status and health values as significant factors influencing self-initiated health action (DTC genome testing).

Chapter six further interprets research results as they relate to the hypotheses and to future research plans. Implications of the study results for revision and extension of the Health Capability concept mapping and framework are also discussed. These



include: 1) supporting health status and values as principle concepts; 2) supporting addition of a measure of individual (subjective) perceived health status; 3) representing resources outside the healthcare system; and, 4) exploring the possibility of a personal need or value, as a “trigger” to action. These are areas of planned further research relevant to personalized healthcare, effective clinical practice, a collaborative healthcare model and meaningful policy development.

## DEDICATION

My genetics/genomics research, professional practice, teaching and involvement in guidelines and policy initiatives are dedicated to the following: 1) promoting informed and responsible integration of genetics and genomics into healthcare; 2) advocating for individual health values and goals, personal empowerment, and individual involvement and responsibility in health-related decisions and priorities; 3) promoting research involving healthy persons to foster understanding of their health goals as well as explore strategies for health promotion and prevention; and, 4) fostering timely healthcare policy development responsive and adaptable to advances in genomics with application at individual as well as public health levels.

## ACKNOWLEDGMENTS

I would like to express my deepest appreciation to my Committee Chair, Dr. Bonnie Holaday, whose mentoring, support, creativity, internal strength and steadiness has meant a great deal to my professional and personal development. I also want to express my sincere appreciation to Dr. Kelly Smith, who accepted responsibility as Co-Chair, and whose fresh and candid perspective provided dimension and clarity, complimented by a philosophical sense of humor. I also feel very fortunate to have had the support, mentoring and editorial expertise of Dr. James Evans, with whom I could discuss and debate genomics at levels ranging from clinical applications, to evolving genomic sequencing technology, to Direct-to-Consumer testing and issues with personalized healthcare. Rounding out my stellar Committee, I valued Janet Craig's expertise in healthcare administration, policy and challenging on practical issues. The fruits of their time, support and constructive guidance will remain with me forever.

Appreciation for an essential element must be acknowledged. After twenty years of searching for a doctoral program where I could take my passion for clinical genetics to the next level to promote education of healthcare practitioners, genetics/genomics services to individuals and families, and creative approaches to policy development, I found the interdisciplinary Ph.D. in Healthcare Genetics program in the School of Nursing at Clemson. I feel privileged to be graduating from this program and University.

I want to express my sincere appreciation to the Robert Wood Johnson Foundation Executive Nurse Leader Program. I had the honor and privilege of being selected for a three-year fellowship in 2001. The experience, including the faculty,

Executive Board, nationally recognized leaders, my peer group leader and executive mentors, and the support, constructive dialogue and friendship of my cohort (several in particular), created fertile ground for my professional (and personal) growth and are a cherished part of my being. These experiences and perspectives were life-changing.

Last, but certainly not least, I feel blessed to have the love and support of my family and friends. Thinking of them warms my heart. To my daughters, who have become remarkable women, thank you for your support, encouragement, patience and caring. Finally, to my husband, a depth of gratitude and love beyond what words can capture, for all that, for the daily coffee service to the library, but also for your enthusiastic support and participation as my first-line critical reader bringing professional skills and personal abilities that were invaluable.

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## CHAPTER I

### INTRODUCTION & PURPOSE OF THE RESEARCH

#### **Background**

An estimated one million persons have self-initiated Direct-to-Consumer (DTC) genome testing touted for its abilities to identify potential health risks and ancestral heritage (Genetics & Public Policy Center, 2011; Wojcicki, 2013). Users of this testing span adults of all ages, with health status varying from having medical diagnoses to healthy persons. Why do consumers want and pursue this testing despite cautions about its limitations and the potential harms if results are misinterpreted (American College of Medical Genetics and Genomics, 2013; American College of Medical Genetics and Genomics (ACMG), 2012; Clarke, 2013; Hesse, Arora, & Khoury, 2012)? DTC studies to date of individuals' motivations have limitations and leave many questions unanswered (Bloss, Darst, Topol, & Schork, 2011; Facio et al., 2011; Goldsmith, Jackson, O'Connor, & Skirton, 2012; Gollust et al., 2011; D. J. Kaufman, Bollinger, Dvoskin, & Scott, 2012). Akin to studies of mortality and morbidity where self-rated health status was a predictor of health outcomes and reflected actual individual health (Idler & Benyamini, 1997; Miilunpalo, Vuori, Oja, Pasanen, & Urponen, 1997; Okosun, Choi, Matamoros, & Dever, 2001), does perceived health status as well as health values influence individual health needs, goals and motivations on a grander scale (and throughout the lifespan), regardless of health status or values as determined by healthcare system standards? If so, this could and should change the face of healthcare.

From another perspective of this question, consider persons who present to their healthcare provider with health concerns yet have no diagnosable condition. Are they the “worried well” (Garfield, 2006), are they cases illustrating discrepancies between health status as perceived by the individual versus healthcare system assessment criteria (Kivinen, Halonen, Eronen, & Nissinen, 1998; Okosun et al., 2001), and/or are they proactive prevention-oriented persons whose needs are not being met in the context of a disease-oriented biomedical model (Juengst, Settersten Jr, Fishman, & McGowan, 2012; Personalized Medicine Coalition, 2014)? Are these various groups of persons also included among DTC genome testing users, hoping to get some answers or information important to their health functioning? The purpose of the dissertation study is to examine individual perceived health status (in contrast to health status based on healthcare system criteria) and individual health values as factors that distinguish and influence (motivate) individual health needs, goals and actions in persons who independently pursue DTC genome testing.

### **Overview and Integration of Chapters, Content and Study**

The background, current status, challenges and implications of genomic testing (including the DTC aspect) for individuals and the healthcare system are presented in Chapter II. My unique research study (Chapter V) required exploration of a healthcare model that embodies study concepts and philosophy, bridges the gap between the traditional biomedical model and respect for individual health values and goals, allows for independent individual choices and actions (e.g., DTC genome testing), and supports examination of subjective individual health influences and new strategies for effective

healthcare services and health promotion. The Health Capability conceptual framework (Ruger, 2010) fits these criteria and is described in Chapter III, which also presents an early version of the proposed conceptual process model (since this chapter represents an article submitted for journal publication in the Fall of 2013). The current, detailed concept mapping proposal for operationalizing Health Capability, a need recognized by its author (Ruger, 2010), includes designation of relationships, corresponding hypotheses and concepts covered in literature review, and is explained in Chapter II. A systematic review of the literature on perceived health status is described and discussed in Chapter IV, together with concept measurement approaches and instruments, unifying the Health Capability model (i.e., author's suggested consideration of the SF-36 instrument), the research study (i.e., survey question used to capture this concept) and future potential research and practice applications. The dissertation research study (Chapter V) examines health status from the unique perspective of combining an individual's perceived health status (dichotomized) with a biomedical assignment of health status (presence/absence of diagnosis/es), thus creating mutually exclusive health status groups for analysis of differences related to subjective health factors (i.e., individual health values, reasons/motivations for testing, perception of health risk). The study population is 1,455 self-initiated DTC genome testing users who completed surveys as part of the large scale NIH funded Personalized Genomics (PGen) study (R. Green & Roberts, 2012b). This sample represents a rare, naturally occurring virgin sample of persons who pursued broad-based health-related genome testing without a healthcare practitioners' order or as part of an approved research study.

Results of this dissertation research have current and future substantive relevance to genome testing, healthcare models and health-related applications across individuals, populations, ages, ethnicities, and all clinical practices, as discussed in Chapter VI. Beyond the genomics context, this study offers real potential to gain new, previously unrecognized or misinterpreted, insights into subjective human factors and health values that may contribute to, or even trump, biomedical factors as influences on individual health-related needs, actions, receptivity to information and interventions, and implementation as well as endurance of health-related behaviors. These factors are important to effective and efficient allocation, prioritizing and development of healthcare services, resources and policy.

### **Objectives of the Dissertation**

Specific objectives of this study include the following:

- 1) Representation of the realities and controversies of the genomic healthcare environment, genomic testing and motivations of Direct-to-Consumer (DTC) genome testing users (Chapter II);
- 2) Identification and description of a healthcare conceptual framework (Health Capability), philosophically aligned with individual health values and goals, and compatible with examining personal factors influencing individual pursuit of genome testing (Chapters II and III);
- 3) Development of a concept mapping for the Health Capability model to represent the principle concepts (including health status and health values, as described in the literature review), their proposed relationships, and added elements philosophically consistent with Health Capability and relevant to the dissertation research (i.e., access to resources outside the healthcare system, e.g., DTC laboratories) (Chapter II: current concept mapping; Chapter III: earlier version as submitted with an article for publication);

- 4) Investigation of the concept of perceived health status, how it is defined and operationalized, and the nature, types, strengths and limitations of approaches and instruments used to measure, represent, interpret and apply this concept (Chapter IV);
- 5) Description of the original research categorizing and characterizing DTC genome testing users by health status groups based on an individual's perceived health versus a medical perspective of health status (Chapter V);
- 6) Description and comparison of relationships/associations between health status categories of DTC users and the following: 1) reasons (motivations) for genome testing, 2) socio-demographic factors, 3) perception of health risk, and 4) health value of genomic information (Chapter V);
- 7) Initial exploration to identify and present previously unknown individual reasons/motivations for pursuit of genome testing, based on answers to open text questions (Chapter V);
- 8) Evaluation of research findings and dissertation processes, including suggested modifications related to Health Capability and the proposed concept mapping, strategy(s) for choice and use of perceived health status measures, and discussion of implications for clinical practice and future research (Chapter VI).

## CHAPTER II

### GENOMICS HEALTHCARE ENVIRONMENT, CONCEPTUAL FRAMEWORK FOR RESEARCH AND LITERATURE REVIEW

#### **Background: The Genomics & Healthcare Environment**

Biomedical advances engendered by the Human Genome Project (HGP), spanning whole genome sequencing (WGS), evolving technology and bioinformatics, have opened new horizons for healthcare. These include the following: 1) diagnosis of previously unknown or complex disorders (ACMG, 2012; Gahl et al., 2011); 2) assessment of common conditions such as cancer for risk, treatment and prognosis (e.g., colon cancer) (National Comprehensive Cancer Network (NCCN), 2014); 3) identification of pharmacogenetic variation important to drug therapy; risk screening for early identification, risk management and prevention (Botkin et al., 2010; Do et al., 2011; Kwon et al., 2010); and, 4) genome panels, e.g., related to neuromuscular and cardiovascular conditions (AmbryGenetics, 2012; Marian, 2012). Genomic factors can now be identified in association with low but increased risk for complex and common disorders impacting large numbers of people (Offit, 2011).

Genome testing offers a unique opportunity for personalized DNA-based health risk assessment with test results promising information potentially impacting individual health, risk management and prevention strategies (Berg, Khoury, & Evans, 2011). These capabilities, advances and potentials are not only published in the scientific literature, e.g., DTC testing that identified a previously unsuspected mutation having clinical significance (Personalized Medicine Coalition, 2011; M. E. Roberts, Riegert-

Johnson, & Thomas, 2011), but are a daily part of mass media news (Grogan, 2014), online information (Landro, 2013), social networking, and marketing efforts (ScottH, 2013) raising consumer awareness and interest in genome testing. Genomic testing could conceivably be so powerful an influence as to transform medicine and healthcare (Evans & Green, 2009).

### **Issues and Controversies: Healthcare Model, Policies and Individuals**

Consistent with the biomedical model and healthcare policies, recommendations advise limiting whole genome testing to patients assessed by a healthcare provider, meeting eligibility criteria for a specific diagnosis or targeted genetic risk, and based on guidelines (American College of Medical Genetics and Genomics, 2013; American College of Medical Genetics and Genomics (ACMG), 2012; Botkin et al., 2010; Graves, Peshkin, Luta, Tuong, & Schwartz, 2011; Skirton, Goldsmith, Jackson, & O'Connor, 2012). Third party payers' policies for genetic testing are limited, targeting specific conditions or approaches for medically justified exceptions. Coverage does not include full genome testing for identification of risks (BlueCross BlueShield, 2014; UnitedHealthcare, 2014). These professional guidelines and coverage policies reflect concerns for potential harms to patients, lack of evidence to support clinical benefit beyond targeted applications or that risk translates to disease, and decisions based on economic healthcare realities (e.g., limited resources).

Consumers committed to genome testing must take the initiative to obtain this testing on their own. Direct-to-Consumer (DTC) genomic testing (NHGRI, 2012) is available to anyone, without involvement of a licensed practitioner, via DTC labs and the



Internet (GPPC, 2011). Until recently (May, 2014), despite Federal Drug Administration cautions and prohibitions (Gutierrez, 2013), a few DTC labs offered consumers health-related genome testing and information (for as little as \$99 at one lab that included interpretative reports and raw genomic data) (23andMe, 2014b), as an option among “packages” such as ancestry and paternity testing (Clarke, 2013; Hensley, 2013; Wagner, 2013). This latter lab stipulates that it no longer provides health reports at the present time (although raw genomic data is still generated). The ultimate fate of individual DTC health-related genome testing, in general, is yet to be determined. Individual independent use of DTC genome testing is highly controversial (Caulfield, 2011; Evans & Khoury, 2013; R. C. Green & Farahany, 2014; Personalized Medicine Coalition, 2011). Issues span healthcare, ethics and individual rights (Rivkin & Grossman, 2013), consumer knowledge and related needs (Hahn et al., 2010), public health, laboratory reporting of risk (Bloss, Topol, & Schork, 2012; Imai, Kricka, & Fortina, 2011), evidence (or lack) for action ability based on genomic profiles (Janssens et al., 2008; Janssens, Wilde, & van Langen, 2011), provider readiness for integration of genomics (Evans & Green, 2009; Frueh, Greely, Green, Hogarth, & Siegel, 2011; Li, 2011; McBride et al., 2009), and gaps in policy, standards and regulatory arenas (Caulfield & McGuire, 2012; Frueh et al., 2011; Hauskeller, 2011; Kontos & Viswanath, 2011). Meanwhile, evidence mounts supporting the unique value of genomic information and the integration of risk testing into health assessment and healthcare based on its potential for broader applications to health promotion (Ashley et al., 2010; Botkin et al., 2010; Korf, 2012; Manolio et al., 2013).

## **Summary & Impact on Dissertation Study**

A definitive gap exists between what genome testing is authorized, regulated and deemed of clinical utility by the healthcare system (Palomaki et al., 2010) and is covered by insurance policies (Graf, Needham, Teed, & Brown, 2013), versus what is of possible individual value and personal utility (Bunnik, Schermer, & Janssens, 2011; Grosse, McBride, Evans, & Khoury, 2009), is/has been accessible via DTC laboratories (Genetics & Public Policy Center, 2011; Graf et al., 2013) and has some professional and advocacy group support (Caulfield, 2011; Personalized Medicine Coalition, 2014; The Lancet Editorial, 2012). Although the consumers' ability to obtain DTC health-related genomic testing in the future might appear to be diminishing (23andMe, 2014b; FDA, 2014; Gutierrez, 2014; National Human Genome Research Institute, 2014), the final policies are yet to be determined. This is dependent upon not only U.S. FDA regulatory authority (National Human Genome Research Institute, 2014; Shuren, 2010; Wagner, 2013), but also policies in process, potential constitutional challenges (Rivkin & Grossman, 2013), and laboratory-related decisions at international levels. This situation, however, does not change the very relevant questions posed in this dissertation research, nor more importantly, the broader knowledge that may be gained from this singular population and study of self-initiated DTC genome testing users.

## **Framing the Research: Questions & Hypotheses**

### **Research Questions**

*What motivates people to seek genome testing independent of their healthcare providers? Why do they pursue this controversial testing? Are they the “worried well” (Garfield, 2006), are they cases illustrating discrepancies between health status as perceived by individuals versus healthcare system assessment criteria (Kivinen et al., 1998; Okosun et al., 2001) and/or are they proactive prevention-oriented persons whose needs are not met in the context of a disease-oriented biomedical model (Juengst et al., 2012; Personalized Medicine Coalition, 2014)?*

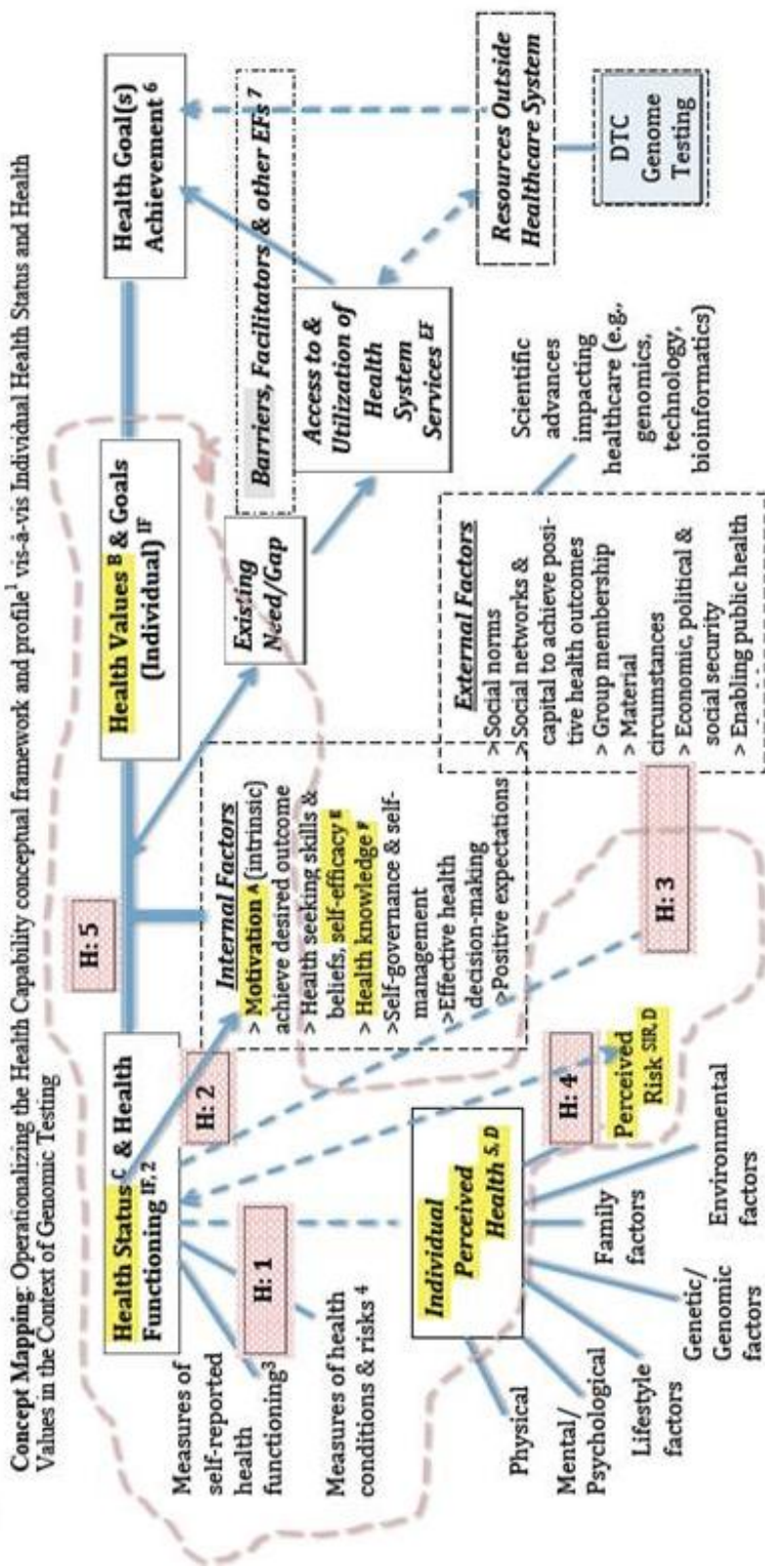
*Do individually perceived health status and individual health values impact health needs, motivations and actions in persons who independently pursue genomic testing and information, regardless of health status and values determined by healthcare system and biomedical criteria? Is there personal health value to genomic information beyond bio-medically defined and determined use & value?*

### **Research Hypotheses** (See Figure 2.1 for hypotheses and relationships)

Hypothesis 1: Discrepancies exist between an individual’s perceived general health status and health status as defined by healthcare system measures (existence of a medical diagnosis).

Hypothesis 2 (null): There will be no difference in motivation(s) for pursuit of genome testing between health status groups stratified by individual perception of health and healthcare system measures (existence of a medical diagnosis).

Figure 2.1: Concept Mapping: Operationalizing the Health Capability conceptual framework



**KEY:** Relationships: Arrow = dependent/independent; Lines: Double = interrelated (non-dependent), Single = concept/factor components; Dashed = unique/unknown; H = Research Question/Hypothesis focus; H: 3 cannot be depicted simply: it is the relationship between socio-demographic factors & health status (Ruger, 2010)

<sup>1</sup> Health status & health functioning can be viewed from two perspectives – as assessed & measured per the healthcare system, but also as self-perceived by the individual

<sup>2</sup> Measures per healthcare system instruments/methods (e.g. SF-36, mental & physical functioning)

<sup>3</sup> Measures per healthcare system assessments: biomedical markers, diagnoses & risk factors (behavioral & lifestyle)

<sup>4</sup> Measure added per this author as dimension of health status, interrelated with health values & goals, and influencing individual potential needs or gaps to health goal achievement

<sup>5</sup> Requires principles/constructs of Health Agency (individual ability to achieve valued health goals & act as own agent) & Health Functioning (relates to outcome of action for health)

<sup>6</sup> Barriers exist across all direct and indirect paths to Health Goal Achievement

<sup>7</sup> Self-interpreted/reported measures of perceived risk (including <sup>1</sup>, <sup>3</sup> & personally derived from any source), <sup>1</sup> Internal factor, <sup>2</sup> External factor, <sup>3</sup> <sup>4</sup> <sup>5</sup> Concepts in Literature Review.

Hypothesis 3 (null): There will be no difference in socio-demographic factors between health status groups stratified by individual perception of health and healthcare system measures (existence of a medical diagnosis).

Hypothesis 4 (null): There will be no difference in perception of risk between health status groups stratified by individual perception of health and healthcare system measures (existence of a medical diagnosis).

Hypothesis 5 (null): There will be no difference in health value of genomic information between health status groups stratified by individual perception of health and healthcare system measures (existence of a medical diagnosis).

### **Research Rationale and Justification**

By categorizing health status as a combination of an individual's perceived health and health status using healthcare system measures (i.e., existence of a medical diagnosis), and examining the identified subjective health-related variables, associations between health status groups and their distinguishing factors may be identified. This study has the potential to contribute new knowledge and understanding about human motivations in seeking and using health-related services such as DTC, as well as impacting and committing to health-related behaviors, relevant to clinical practice and research. The study findings may contribute to informing future strategies for screening and assessment, especially for individuals who challenge the healthcare system, such as the apparently healthy (e.g., the worried well, versus the early ill, versus the prevention-oriented). Implications, however, extend to other areas of identified need in healthcare. These include facilitating practitioner and individual discussions and decisions related to

individual health priorities (McGuire, Diaz, Wang, & Hilsenbeck, 2009), approaches to risk assessment and management, targeted interventions and options, effective allocation of healthcare services and resources (e.g., avoiding treatments, procedures and expenditures that do not have individual health value) (Ruger, 2010), and integrating measures for evaluation of healthcare services and outcomes relating to both clinical utility and personal utility since both impact services and policy development (Bunnik et al., 2011).

The dissertation study's unique dataset of 1,455 Personalized Genomics study survey respondents (R. Green & Roberts, 2012b), all who self-initiated DTC genomic testing, serve as a natural, uncontrolled population to learn about subjective individual factors potentially influencing independent pursuit and interest in using health-related testing and services (i.e., genomic testing), but also potentially contributing to informing and advancing healthcare in a broader realm, as noted above.

### **Conceptual Framework: Health Capability**

A healthcare model and conceptual framework is needed whose tenets acknowledge individual health values and goals, and include primary concepts of the current dissertation study. This model must support inclusion of individual (subjective) criteria in determining health goal achievement, allow for individual access to health-related resources beyond the traditional healthcare system, foster health promotion and prevention on an individual level, and define health values and successful outcomes by other than economic measures. A model integrating these dimensions has been recommended previously (J. P. Sturmberg, O'Halloran, & Martin, 2012). Given an

empowered public, evolving advances in bioinformatics and technology, a healthcare system expected to meet the health needs of individuals and the general public, and economic realities, any meaningful model must also be systems-oriented and supportive of communication, collaboration, shared responsibility in decision-making and policy development involving all stakeholders (Li, 2011; Joachim P Sturmberg, 2007).

The Health Capability conceptual framework meets these stipulated criteria. This healthcare model is discussed in detail in Chapter III. In this chapter, a conceptual mapping (see Figure 2.1) is proposed, presented and discussed as the basis for organizing key concepts, representing their relationships and identifying the focus of the literature reviewed later in this chapter.

### **Concepts, Relationships and Study Foci**

The principle tenets and concepts of the Health Capability conceptual framework include individual health values, health status and health functioning, and health goal achievement (Ruger, 2010) (see Figure 2.1). Their relationships were not specified in the original conceptual framework, or subsequently. Thus, consistent with current study hypotheses, these concepts are depicted as interrelated, being joined by a heavy solid line. A Health Capability profile provided the internal and external influencing factors (Ruger, 2010). The major categories of which are summarized in Table 2.1. These internal and external factors are listed within overlapping hashed boxes in Figure 2.1, thus representing a postulated dynamic interplay. The area within the red-hashed irregular borderline identifies the current study focus. Solid blue lines represent connections

**Table 2.1: Health Capability Profile\*: Influencing Factors.**

<b>INTERNAL</b>	<b>EXTERNAL</b>
A. Health status & health functioning	A. Social norms
B. Health Knowledge	B. Social networks and social capital
C. Health seeking skills and beliefs, self-efficacy	C. Group memberships
D. Health values and goals	D. Material circumstances
E. Self-management; ability achieve health outcome	E. Economic, political & social security
F. Effective health decision-making	F. Utilization/Access to health services
G. Intrinsic motivation to achieve health outcomes	G. Enabling public health & health sys
H. Positive expectation: achieving health outcomes	<i>*Adapted from Ruger, 2010, p 45-46.</i>

between the concepts, factors or components that were identified by the author. Hashed blue lines represent connections that include a factor or component proposed in my current study. Hypotheses, denoted by an “H” and the hypothesis number, are in small red boxes positioned with the corresponding concepts and relationship. Based on Hypothesis 1, the proposed influence of individual perceived health (highlighted in yellow in the text box in the left middle of Figure 2.1) has been added and connected by a blue-hashed line to Health Status. Similar, for Hypothesis 4, the propose influence and connection between Health Status and Personal Measures of Risk (highlighted in yellow) is represented by a hashed line. Hypothesis 2 and Hypothesis 5 are represented by solid lines between concepts and factors. Hypothesis 3 represents an association between Health Status and components involving selected external factors (socio-demographic based on survey data). Resources available outside the traditional healthcare system are represented as an extension (hashed line) of existing resources, on the right side of Figure 2.1. This component, specifically the DTC genome testing box, is not contained within a red-hashed border but instead is highlighted in blue (lower right of Figure 2.1) since it defines the primary eligibility criteria for study participants and identifies that all pursued



this route. Study concepts, highlighted in yellow, are numbered and correspond to the literature review later in this chapter.

### **Conceptual Framework Adaptations**

Two conceptual framework adaptations were done for this study for the purposes of operationalization and model extension (i.e., genomics and prevention). First, health status and health functioning, represented in Health Capability collectively as one entity within internal factors, is reflected in my proposed Concept Mapping (Figure 2.1) as having two facets. The first facet measures, as designated by Ruger (Ruger, 2010) and represented in Figure 2.1: 1) self-reported health functioning (e.g., as measured by the SF-36, mental functioning and physical functioning (McHorney, Ware John E, Lu, & Sherbourne, 1994; Ware, 2009)), and 2) measures of health conditions (e.g., biomedical markers, diagnoses, diseases including mental health) and extrinsic risk factors (e.g., smoking, exercise, diet, drug use, obesity). Although these two types of measures recognize the existence of both physical and mental health, and are “self-reported”, the instruments and measures offered are from the perspective and criteria of the healthcare system. One represents a measure of health functioning (not health status). The other set of measures is based on established medical tests, measurements and guidelines, including representing (and limiting) risk factors to a medical model context and its expected health-related behaviors. In contrast, the proposed model addresses a gap in understanding of the individual perspective of (their) health status (highlighted in yellow, and connected by a blue-hashed line to Health Status), regardless of that as assessed or assigned by healthcare system processes and measures. Not to incorporate such

information seems contrary to the Health Capability framework's primary tenet of commitment to individual health, values and goals.

The second modification is a re-framing of health values as a primary concept, as opposed to simply representing and limiting it under internal factors (see Table 2.1). This proposed approach allows for: 1) examining and comparing the interpretation of health status based on a biomedical model and criteria versus an individual's perceived health status, and 2) examining a direct relationship between an individual's health status and health values as potential motivators of health action/behavior toward achievement of health goals.

### **Review of Literature**

The goal of this literature review is to: 1) provide the background, current status and relevant perspectives of the dissertation study's primary concepts; and, 2) identify and discuss the conflicts and gaps in the existing research, literature and perspectives. The concepts are presented in the order of historical development and the evolution of this dissertation. These are represented in Figure 2.1 by superscripts in uppercase letters adjacent to the corresponding highlighted concepts. The literature review also provided the basis for operationalizing the key concepts into measurable, delineated constructs.

#### **Motivation (and Genomic Testing)** (*Figure 2.1, Superscript A*)

Motivation, an internal factor of Health Capability, has been the subject of several DTC genomic testing studies to gain an understanding about the individual's interest and reasons for pursuit of genome testing. The systematic review of the literature, including

qualitative and quantitative studies of DTC users, identified the following themes, reasons and desires for individual pursuit of genome testing: curiosity, health-related (e.g., disease risk, risk factors), specific disease related (e.g., personal diagnosis; condition in the family), health promotion, gaining information about oneself, genealogy, helping others, contributing to research, and recreation-related (Goldsmith et al., 2012; Gollust et al., 2011; Su, Howard, & Borry, 2011).

However, the findings of these studies are limited for multiple reasons spanning the nature and extent of the sample, the population or data source, the study structure, the survey or instrument, and inherent or researcher biases. Specific reasons include the following: 1) study populations involving small and/or convenience samples (Bloss et al., 2010; McBride et al., 2009; Su et al., 2011); 2) participants who may not represent the stated population (i.e., candidates for genome testing as opposed to actual users, or persons who pursued testing because it was free in contrast to self-initiated genome testing users who pay for testing) (Kaphingst et al., 2012; D. Kaufman, Murphy, Scott, & Hudson, 2008); 3) participants not representative of the general population (e.g., persons having an advanced science background or profession, working in the healthcare industry, or the majority having a college education) (Bloss et al., 2012; Su et al., 2011); 4) discrepancies based on the source (Goldsmith et al., 2012) of data (e.g., participant answers in conjunction with the labs they used versus postings on outside sites) (Su et al., 2011); 5) limited percent of survey responders (Bernhardt et al., 2012); 6) survey-designated answer options for pursuit of testing (Facio et al., 2011); and, 7) potential researcher biases, as with interpretation of written motivations (Goldsmith et al., 2012;

Su et al., 2011). For example, a participant's stated reason for testing is interest in knowing one's genome, and that may be classified as curiosity; however, the underlying motivation was concern, hope and curiosity to gain information that might explain existing, or undiagnosed symptoms.

There is a dearth of large-scale genomic testing studies related to motivation (and self-efficacy) (Keller et al., 2010), most notably involving actual DTC genomic risk testing users who self-initiated testing (Bloss et al., 2010), with limited exceptions. However, the Personalized Genomics (PGen) study of 1648 self-initiated DTC genome testing users (R. Green & Roberts, 2012b) found that motivation for DTC genome testing varied by self-reported health status (R. C. Green et al., 2013) with interest including not only risk identification, but also confirmation and etiology of an existing diagnosis (Meisel et al., 2013). Another large-scale study, the Scripps Genomic Health Initiative (SGHI), found that 82% of participants would want to know risk findings for non-preventable conditions (Bloss et al., 2010), although this was not part of the study. This is consistent with results and expectations of another study (Michie, Henderson, Garrett, & Corbie-Smith, 2011). Other studies supported health promotion, adopting a healthier lifestyle, and ability to take responsibility for future health as motivators for seeking and using genomic information (Cherkas, Harris, Levinson, Spector, & Prainsack, 2010; McGowan, Fishman, & Lambrix, 2010).

### **Summary**

Although the findings of DTC genomic testing research has contributed to initial understandings related to motivation, studies are few in number with significant

limitations. Specifically these studies: 1) cannot be presumed to represent individuals who pursue DTC “on their own”; 2) cannot adequately inform an understanding of health values, perception of health and risk, and health needs as motivators for genome testing (or future health behaviors); and, 3) are compromised in their ability to inform clinical practice, prevention strategies and policy directions (Bloss et al., 2010; Goldsmith et al., 2012; Goldsmith, Jackson, O'Connor, & Skirton, 2013).

### **Health Values** (*Figure 2.1, Superscript B*)

The gap in understanding individual interest in, and motivation for, pursuit of genomic testing may involve even more basic human factors, such as individual health values. A representative definition of human values involves “... an enduring belief that a specific mode of conduct or end-state of existence is personally or socially preferable to an opposite or converse mode of conduct or end-state of existence” (Rokeach, 1973). This same author purposely avoided use of the terms "ought", "should", and "conceptions of the desirable", discussed many types of values, yet health values was not included.

A systematic review of the literature was performed using PubMed, PsychINFO, CINAHL and Cochrane databases. Since PubMed Medical Subject Headings (MeSH) does not include individual health values, a three-step process was used to identify relevant records. First, the term health values were searched as a keyword, identifying 107, 859 records. Next, a search using the MeSH terms individual or personal resulted in 7,939,611 records. Third, a search stipulating the union of these two sets resulted in 64,430 records. In order to identify records specific to genomic information and testing, a series of searches was done using these, as well as five other genomic-related MeSH

terms. By combining this result with that for individual health values, 337 records were identified (limited to humans). Six additional records were identified through other sources, i.e., a university library resource expert and references cited within a health values-related text, for a total of 343 records. These records were screened by title and/or abstract content. Exclusion criteria included the following: records lacking mention, or the essence, of health values, individual health perspective or personal value of genomic/genetic information. From a complementary perspective, the inclusion criteria retained records that used these previously stated terms, or their essence, as well as research involving focus groups and qualitative studies of individual health views. This resulted in a final selection of 30 records for full review, including qualitative and quantitative studies, perspective articles and commentaries. Several articles or studies did not actually address individual health values or used values from a different context, such as equating it with knowledge, and these were excluded. Corresponding searches of PsychINFO, CINAHL and Cochrane databases did not identify any further records.

Individual Perspective: Very few studies addressed individual health values as a subjective personal concept with qualitative aspects (e.g., what the subjective elements are, their relative importance), and their relationship to interest in or pursuit of genetic testing. Studies of relevance typically used a survey or targeted focus groups for specific disorders or conditions, identifying value or belief themes that influenced interest in genetic testing or information (Doukas, Fetters, Coyne, & McCullough, 2000; McBride et al., 2009; Pivetti, Montali, & Simonetti, 2012). Although these studies spanned very different populations, from men of various ethnicities aged 18-90 related to prostate

cancer risk and gene testing, to 20 pregnant women and new mothers related to uptake of prenatal genetic testing (PGT), the identified themes and reasoning were similar in nature. The essence of themes were the following: 1) usefulness and benefits of information (e.g., for making choices, preparing, family planning, taking action such as changing diet or exercise; 2) interest in and concerns about susceptibility, risks and consequences; 3) beliefs about barriers; and, 4) issues of morality and trust (Doukas et al., 2000; McBride et al., 2009; Pivetti et al., 2012). These studies were few in number, limited to specific conditions, have small sample sizes and/or only partially address the issue of individual health values; however, they also attested to this concept's influence and the need for further research.

Cultural and Societal Influences: The cultural and societal influence on values and perceptions was also evident. What is perceived as important to an individual or community group may be of little value by healthcare system standards, and vice-versa (e.g., knowledge of risk prior to symptoms versus reduction in mortality). This perpetuates a frustrating, costly, resource draining and ineffective cycle for practitioners, patients, educators and policy makers. Studies, such as those involving Fragile X genetic screening of children in the Colorado schools' special education classes, found that interest in genetic testing reflects cultural beliefs regarding the importance of genetic information and expectations about future possibilities (Nelkin, 1996). The author also posits that the media fosters a public impression of social meaning to genetic testing that it may support individual needs. Whether or not one agrees with this position, there is no question about the existence of marketing related to the value and potential of

genetic/genomic information and testing, an increasing public awareness and interest, and the healthcare view of its limited clinical utility (Ducournau, Gourraud, Rial-Sebbag, Bulle, & Cambon-Thomsen, 2011; Jordens, Kerridge, & Samuel, 2009).

Healthcare System Perspective & Issues: The literature review identified one relevant large scale study involving 12 focus groups and 97 healthcare providers (i.e., genetic counselors, physicians and nurses in four different geographic areas) that demonstrated discrepancies and conflicts encompassing sixteen major ethical and professional domains (Veach, Bartels, & LeRoy, 2001). Value-associated conflicts were among the second and third most common issues, spanning intrapersonal (professional with self), interpersonal (between professional and the patient, or a colleague), and extra-personal (among patient family members) relationships. It is noteworthy that all examples of values' conflicts (5) provided by the study authors had a moral basis. Yet the common occurrence of conflicts in the broader spectrum of health-related values, especially between healthcare providers and patients as well as healthcare providers with each other, supported the relevance and need for studies to examine individual health values. Such information relates to the provision of meaningful healthcare services, prioritization and allocation of healthcare resources, expectations for patient health behaviors and outcomes, support for individual healthcare values, and the development of effective policy (Ruger, 2010; J. P. Sturmberg et al., 2012).

Health Values as an Economic Measure: An economic perspective of health value was a common component among the identified studies. In healthcare, value is often defined as an outcome/cost ratio, such as cost-effectiveness (CE) or incremental



cost-effectiveness ratio (ICER) (Kwon et al., 2010; Woolf, Lewin, Marks, Fielding, & Sanchez, 2009). CE may be based upon the achievement of significant differences in outcomes, such as morbidity, mortality or health benefits, often within a given time frame, related to a specific treatment, strategy or technology (Tilburt et al., 2011). An economic view of health value also exists in the prevention context. Two examples are the assignment of a CE threshold of \$50,000/year of life gained in the context of breast cancer treatment and testing (Kwon et al., 2010), and individual willingness to pay for services (i.e., genetic counseling, information and testing services) based on the contingent valuation method (Eden et al., 2013). Individual values and quality of life (QOL) factors are typically not included (Tilburt et al., 2011). A quasi-economic view of health values, captured by the idea of worth or tangible value of genetic/genomic information from the framework of exchange, gift and genetic responsibility theories, was demonstrated in a study of 752 donors of specimens for genetic research (Michie et al., 2011). This study's results, despite the focus on altruistic motivation while respecting a monetary value of donations, distinctly supported the existence of underlying individual health motivations and expected personal health value for participants. Of donors, 70.8% wanted to find out information about their own health, although informed consent specified that was not an intended use for the specimens. Donors also spoke of their specimens in the context of diagnosis rather than research use, with an expectation of results, consistent with a personal value to their genetic information.

Summary & Discussion: The studies reinforced the existence and influence of underlying individual health values, the expectation and/or desire for personal

information, the potential (or perceived) value of genomic information to the individual, and the necessity for further research to gain a better understanding of future applications in healthcare. Existing gaps include the individual (patient, researcher, system) subjectivity in defining and operationalizing health values, its relationship with other personal health components (e.g., perceived health status), and its contribution to an individual's motivations, decisions, actions and expectations for healthcare information and rights of self-determination, including the pursuit and use of genomic testing (Nyrhinen, Hietala, Puukka, & Leino-Kilpi, 2009). There was also a distinct gap in understanding and perspective involving individual health values related to clinical utility versus personal utility (Bollinger, Scott, Dvoskin, & Kaufman, 2012; Bunnik et al., 2011; Grosse et al., 2009). A practical question arises about the use of alternate definitions and measurements for health value, specifically focusing on individual subjective health values, that may better delineate personally relevant influences, how they interrelate with other subjective personal health components (e.g., perceived health status), individual needs and health goals, as well as the value and personal utility of genomic information as it relates to individual health choices and behaviors.

### **Perceived Health Status** (*Figure 2.1, Superscript C*)

A systematic review of the literature was performed concerning the individual's perceived health status and this is addressed in Chapter IV. The review also includes review of literature and discussion about the two main methods used to capture this construct, specifically a single question or a measurement instrument. The focus in this section is restricted to relevant foundational and contextual information.

The healthcare system traditionally has viewed health status using the biomedical model, focusing on the presence or absence of symptoms, illness or increased risk as defined by the presence of objective scientific evidence to make a diagnosis, justify intervention and determine treatment (Borrett, 2013). Many situations do not meet these standards. The need for examining health and health status from the subjective individual perspective is recognized, yet the healthcare system is struggling with how to do this, what models to use, what values to include and the mechanisms to use for evaluation (Loughlin et al., 2013).

Studies that include an individual's perspective of health status typically involve the use of a survey or questionnaire that limits capturing this construct to a single qualitative, yet quantitatively measured, item or focus for analysis. For example, one study of DTC users of genomic testing asked participants to rate their (overall) health using a 5-point Likert-type scale ranging from very poor to very good (Bloss et al., 2010), while a study of African-American males focused on cardiovascular disease examining health status, but from the context of health abilities (and functioning) using a health practices scale (DeCuir, 2007). Health status and health values were frequently framed in the context of health risks, risk likelihoods and binary situations (Wang et al., 2009). These risk and binary approaches lack appreciation of the broader and qualitative dimensions of health values and individual freedoms, and foster an all-or-nothing stance that minimizes the potential contribution of research in multifactorial and complex common disorders, which includes predictive genomic testing (Boenink & van der Burg, 2010).

Although the search focus was health status, the majority of records identified and reviewed were disease or risk focused, involving quantitative or mixed methods studies that included measures of perceived health status based on the study focus or need. The most frequently used instrument (8/19; 42%) for evaluating perceived health status was the SF-36 (Footit & Anderson, 2012; Funk et al., 1997; Gartsman, Brinker, Khan, & Karahan, 1998; Hunter, McKee, Black, & Sanderson, 1995; Lindsay, Smith, Hanlon, & Wheatley, 2001; Mattera et al., 2000; McHorney et al., 1994; Ware, 1976, 2009). This instrument has been suggested for measuring health status and health functioning in the Health Capability conceptual framework (Ruger, 2010). The SF-36 encompasses 36 questions, spanning eight scales (physical functioning; role-physical; bodily pain; general health; vitality; social functioning; role-emotional; and mental health), plus the perceived general health status item. The repeated use of this measure across more than 4,000 studies has supported its validity and reliability (Ware, 2009). This is addressed, together with other instruments in common use, in Chapter IV.

These instruments have limitations related to the following: 1) the limited number of questions, reflecting the inability to cover all desired health status related content areas; and, 2) the survey structure in that questions provide all answer choices, without open ended questions, thereby precluding the ability to capture unique individual subjective concerns or issues. These issues may alter the understanding of an individual's perceived health status, and their contribution to influencing health related needs and actions, such as the decision to pursue genomic testing. Based on this search,

no instrument was identified that encompassed an individual's perception of health status, and subjective personal health values, concerns and goals.

## **Discussion**

The need for further research to examine and foster understanding about the individual's reasons for independent pursuit of genome testing was demonstrated by the results of a literature review (Goldsmith et al., 2012) attesting to gaps, biases, lack of actual users and likely underrepresentation of persons pursuing DTC genome testing. These findings also lend support to the current study's proposal that an individual's perception of health is a complex construct composed of many components, differentially relating on an individual level to motivation, health values and goals, and health behaviors. The elements identified, yet not combined or examined for potential interactions in a research study thus leaving unanswered questions, include the following: physical and/or psychological, i.e., disorders or symptoms (Heshka, Palleschi, Howley, Wilson, & Wells, 2008); perceived and actual health risks, such as for multifactorial conditions (Zeggini et al., 2008); familial conditions and genetic aspects (Heshka et al., 2008; O'Neill et al., 2009); environmental influences, as with Parkinson's disease (Do et al., 2011); lifestyle choices, such as smoking (Janssens et al., 2011; Saudny, Cao, & Egeland, 2012); and, individual goals, such as health promotion (Juengst et al., 2012; Lea, Skirton, Read, & Williams, 2011). Awareness of these components and possible interactions challenge previous research findings based on simplifying and restricting health status to representation as a biomedical, or single entity.

These health-related components are noted in the Concept Mapping, Figure 2.1, as comprising Individual Perceived Health Status. This study incorporates only the perceived risk component (see following). The other components will be examined in future research.

### **Risk Perception** (*Figure 2.1, Superscript D*)

Perception of health risk is a factor associated with individual pursuit of genomic testing (Gollust et al., 2011). Most studies use the terms perception of risk, risk perception and perception of health risk synonymously without defining these terms, but instead imply their meanings based on the study context, population focus and/or operationalization of items measured (Hahn et al., 2010; D. J. Kaufman et al., 2012; Lerman, Rimer, & Engstrom, 1991; Lipkus, 2007; McAllister, 2003; van Maarle, Stouthard, & Bonsel, 2003; Watson et al., 1999). For example, perception of risk was used as disorder specific for breast cancer (Watson et al., 1999) and for Alzheimer's disease (among others) (Heshka et al., 2008). The single focus of risk perception in these studies can be viewed as a strength for statistical analysis, yet it is also a limitation if perception of risk includes qualitative components not sought, measured or recognized as important to individual health values and goals.

Philosophical Origins and Measures of Risk: Two fundamental views of risk operate in parallel based on cognitive psychology and neuroscience. There is the “analytic system” that uses algorithms and norms, and the “experiential system” based on individual experiences, affect (emotions) and values (Slovic, Finucane, Peters, & MacGregor, 2004). The former, consistent with a biomedical scientific model, reflects

objective, reasoned and logical processes that can be measured, quantified and controlled. The latter reflects individual health values, subjectivity, human dynamics and personal goals, consistent with valuing genomic information and mapping back to the Health Capability concepts and principles.

Risk perception incorporates “a situation or an event where something of human value (including humans themselves) is at stake and where the outcome is uncertain” (Rosa, 1998), a risk target (self vs. others) (Sjöberg, 2000), and “how concerned we are about the consequences” (Sjöberg, Moen, & Rundmo, 2004). Three key features emerged, all subjective in nature, being: assessment of probability, an undesired event/entity, and concern about the impact of the outcome for self. Individual perception and interpretation of health risk is paramount. This is not confined to a single, static, isolated risk factor.

Defining Individual Perception of (Perceived) Health Risk: For the purpose of the current study, individual *risk perception* is defined as: *Individual recognition or appreciation of real or believed vulnerability, compromised health status and/or likelihood of a disease(s), disorder(s) or condition(s) (specific or in general), as either increased relative to others, or existing to an extent beyond that acceptable to that individual* (BusinessDictionary.com, 2012; Dearborn & McCullough, 2009; Gregory & Mendelsohn, 1993; Griffith, 2011; Lloyd et al., 1996; McAllister, 2003; "Risk Communications Bibliography," 2012; Rosa, 1998; Slovic, 1987; Walker, Mertz, Kalten, & Flynn, 2003; Watson et al., 1999). This definition facilitates the operationalization of various potential individual influences contributing to an individual's perception of risk

(which parallel components relevant to an individual's perception of health, as noted previously).

Genetics/Genomic Context: Perception of risk, from a genetic perspective, has been operationally defined and measured in different ways (Lipkus, 2007). These included the following: quantification of risk for a specific disease, e.g., cancer, using numeric measures (comparative; binary; personal; population percentage of risk) (Kelly et al., 2007); measuring risk likelihood for selected diseases, e.g., as with a Likert-type scale (Lerman et al., 1991); assessing the extent or category of risk (e.g., present/absent; high/moderate/low); and, accuracy of individual recall for disease specific numeric risk, test result or biochemical screening values (Kaphingst et al., 2012; D. J. Kaufman et al., 2012; van Maarle et al., 2003).

Risk perception research in genetics has examined this phenomenon in participants, primarily after receiving the results of genetic testing (Heshka et al., 2008; D. J. Kaufman et al., 2012). A review of the literature on the perceived risk, psychological and behavioral impacts of genetic testing identified 35 articles and 30 studies (Heshka et al., 2008). No differences were found in risk perception between gene mutation carriers and non-carriers 12 months post-test, postulating one explanation for the lack of differences as possible differences in risk perception between these two groups. A gap in research exists related to the examination and understanding of perceived health and risk before genome testing, its potential influence on individual health values, expected results and pursuit of testing, and the impact on health-related behavior. Although this has been studied to a limited extent, the focus on risk perception



has been disease/condition specific or based on case scenarios, as opposed to actual personal results and experience (D. J. Kaufman et al., 2012).

**Concepts Assumed in the Current Study** (*Figure 2.1, Superscripts E and F*)

Self-efficacy (Superscript E): This concept, among Health Capability's internal factors (see Table 2.1), includes coping behavior, the extent of energy expenditure, and sustaining in the face of obstacles and aversive experiences, are supported by behavioral change theory (Bandura, 1977). Self-efficacy is defined as "...the judgments people hold about their capacity to master specific tasks and to cope with challenging situations (Oles et al., 2013) (p.48). Self-efficacy has relevance to the well-established health-related and prevention theories including health belief, health locus of control and health capability (Janz & Bcker, 1984; Norman, 1995; Rosenstock, 1974; Ruger, 2010). Studies involving self-initiated testing and genetic risk concerns support the concept of self-efficacy as relevant to the pursuit and/or use of genomic testing (Grispen, Ronda, Dinant, de Vries, & van der Weijden, 2011; McGowan et al., 2010). Thus, persons whose health goal was to secure genome testing, and who independently accomplished this testing despite traditional healthcare system barriers, all exhibited self-confidence and self-efficacy.

Health Knowledge (Superscript F): Defining and measuring the concept of health knowledge, an internal factor of Health Capability, was included in several genetics studies (Haga et al., 2013; Leighton, Valverde, & Bernhardt, 2012). This poses a myriad of challenges, not the least of which is determining the relevant focus. The gap in research addressed by this study relates to individual perceived health status, health values, reasons for pursuit of genome testing and perception of health risk. These

subjective concepts do not require, or necessarily correlate with, genetics knowledge. Genetics knowledge will not be examined in the current study. As further justification for this decision, previous studies of DTC genome testing users has consistently shown participants to tend to have higher levels of education than the general population, to tend to understand that genome testing results relate to potential risks not outright diagnoses, and that discussion with healthcare providers is recommended before taking any health-related action. These studies also demonstrate inherent biases, such as the nature of the questions posed, interpretation of answers and scoring, use of accuracy as a surrogate for knowledge, the nature of knowledgeable comparison groups, and assumed gain in knowledge if research educational materials are accessed (Keller et al., 2010; Leighton et al., 2012; van Maarle et al., 2003).

### **Summary and Relationship to Other Chapters**

This chapter described the genomics and healthcare environment, the increasing use and evolving potential of genome testing, and the issues and challenges related to individual pursuit of this testing. Research questions and hypotheses address the components potentially influencing individual self-initiated pursuit of DTC genome testing, from the perspective of an individual's subjective perception of their health versus health status based on healthcare system parameters. Other possible subjective influences include motivating elements, individual health values and goals, and perceived risk. The concept mapping proposed in this chapter operationalizes the conceptual framework, Health Capability. Relevant concepts and components of the Health Capability model related to the study are presented, along with corresponding literature

reviews. These concepts include motivation, health values, perceived risk and perceived health status. Perceived health status is the main concept based on literature reviews. The concept map is examined along with approaches and instruments to measure this concept. This background and processes lead to the study presented in Chapter V. The DTC genomic testing research supports the importance of concepts essential to this dissertation research study and to Health Capability. However, if and how these concepts and components interrelate, influence health-related priorities and decisions, impact health action and behavior, and compare with traditional perspectives of health status have not been examined. Data from the PGen study of DTC genome testing users (R. Green & Roberts, 2012b), forms the basis for this study. It offers an opportunity to address these questions in self-initiated users with no input from healthcare providers or researcher-based influences prior to their decision to pursue genome testing. The broader implications of this research are addressed in Chapter VI.

## CHAPTER III

### PERSONALIZED GENOME RISK TESTING: EXAMINING A MODEL TO INFORM AND AMELIORATE THE DICHOTOMY FACING HEALTHCARE

#### **Introduction & Background**

This chapter is an article submitted for review and publication to the *Journal of Health Services Research & Policy*. The following pages are copied from the PDF file, exactly as submitted in compliance with journal requirements.

The content of this article complements and extends information on the Health Capability conceptual framework. This model encompasses the concepts and prioritization of individual health values and goals, from both a philosophical and a practical basis. These are integral to this study. The concepts/constructs of perceived health status, health values (and goals), health functioning and health agency relate directly to the users of Direct-to-Consumer (DTC) genomic testing. In the Health Capability profile, internal and external health-related factors are recognized as influences on the individual at different system levels (individual, family/community, healthcare systems, and societal/governmental). However, a concept map of these relationships does not exist. This article includes a proposed structure and relationship of essential concepts that became the Concept Mapping presented in Chapter II. This included an expansion of some components for enhanced health-related applications, such as the context of genomic testing in healthy persons. Components of this map were tested in the study (Chapter V).

## **Personalized Genome Risk Testing: Examining a Model to Inform and Ameliorate the Dichotomy Facing Healthcare**

### **Abstract**

Technologic advances with the Human Genome Project (HGP) permit sequencing of essentially the entire human genome. Although practitioner-ordered targeted genome testing is an increasing component of healthcare, individuals can independently access whole genome testing through direct-to-consumer (DTC) labs for a multitude of risk/susceptibility factors with potential value for their health, as well as make self-directed risk management choices with varying understanding of results. This situation clearly demonstrates gaps, deficiencies and issues within the healthcare system, its models and policies. A model is needed that integrates biomedical and values-based models, but also fosters prevention and respect for individual health values. This article proposes extension of the Health Capability conceptual framework,<sup>1</sup> with its underlying principles prioritizing individual health values and goals, to prevention and health promotion vis-à-vis genomic risk testing. This model is amenable to the changing healthcare environment, advances in biotechnology and collaborative decision-making. It also fosters examination and implementation of practical, strategic and cost effective approaches to healthcare research, interventions and policy development, uniquely including an individual focus extending to public health contexts.

### **Introduction: competing realities of whole genome risk testing**

Technologic advances with the Human Genome Project (HGP) permit sequencing of essentially the entire human genome. Targeted genome testing has proven medical value for diagnosis (e.g., cardiovascular diseases), treatment decisions (e.g., colon cancer) and identification of familial risk mutations (e.g., BRCA). Consistent with the biomedical model and policies internationally, recommendations advise limiting whole genome testing to patients assessed by a healthcare provider, meeting eligibility criteria for a specific diagnosis or targeted genetic risk, and based on guidelines.<sup>2,3</sup> In contrast, Direct-to-Consumer (DTC) laboratories offer whole genome testing for a

multitude of ever-increasing risk/susceptibility factors with potential value to individuals for health information and risk management choices.<sup>4</sup> Thus, a definitive gap exists between what is deemed of clinical utility by the healthcare system and covered by insurance policy, versus what is accessible to the general public for increasingly affordable rates via the Internet and DTC laboratories.<sup>5,4</sup> DTC allows individuals to circumvent the controls (and protections) of the healthcare system – taking health, prevention, symptom evaluation and healthcare into their own hands.<sup>6</sup> The extent of DTC testing uptake by consumers, motivations for testing, and if information is adequately understood, interpreted and used for health behavior modification are among poorly understood issues due to limited research, convenience or poorly-representative samples, and built-in (albeit unintentional) research biases or assumptions.<sup>7</sup> This author argues that given genome risk testing availability through over 20 DTC labs,<sup>4</sup> a public with limited genomics expertise, and that whole genome testing is becoming integral to mainstream healthcare,<sup>2,8</sup> the salient question is not “if” genomic testing should be available DTC. Rather, the operative question is: “What are the issues and barriers impeding, and critical to, time-sensitive, responsible and effective integration into healthcare practice, research and policy?” This paper confronts values and structure inherent to the healthcare system that warrant re-examination based on advances in biotechnology particularly whole genome risk testing and DTC, explores use of a relatively new healthcare model that prioritizes individual health values and social justice, and proposes model adaptation and strategies for application to research, practice and policy development.

#### **The healthcare system and genome risk testing perspectives: alternate realities?**

*Prevention:* Whole genome testing for risk identification relates to screening and prevention. In the biomedical model, screening is offered to at-risk populations (e.g., prenatal, newborn) for specific risk factors, using scientifically reliable tests that are cost-effective and have clinical utility (ameliorate or prevent disease). Whole genome risk testing does not appear to meet these criteria – all persons would be included, testing would involve the entire genome, analytic validity and clinical utility varies with the nature of testing and gene findings, and it is not cost effective.

Furthermore, WGS will result in variants of uncertain significance (VUS) and incidental findings, thus also supporting limiting clinical testing.<sup>9</sup> Alternatively, consider that DTC genome testing: (1) has identified previously unsuspected mutations having clinical significance,<sup>10</sup> (2) uses next generation technology with proven analytic validity, and clinical utility for certain conditions,<sup>4</sup> (3) incurs no cost for testing to the healthcare system (i.e., individual self-pay), (4) may be of individual value for personal health decisions, and (5) is among other DTC tests (i.e., pregnancy, HIV). Additionally, WGS has been analogized to newborn screening – which is universally accepted and recommended despite varying extent of tests, and risk of false positives or uncertainties; it is also an area ripe for genome screening despite ethical concerns.<sup>11</sup>

*Research:* Per information readily available on National Institutes of Health (NIH) websites, funding is more often for quantitative research, whereas qualitative and prevention research is limited. Reviewer criteria are consistent with a biomedical model. The “gold standard” for research is the randomized control trial (RCT) and large population-based studies, yet these approaches are not practical or meaningful for much qualitative and innovative prevention research.<sup>12</sup> Lack of statistical significance may be due to limitations in capturing qualitative influences or prevention outcomes (e.g., reduced disease incidence or morbidity requires more time), or data analysis issues (e.g., measurements at population levels when relevance relates to individuals or sub-groups). Justification for prevention research also poses special challenges with lack of background data – it cannot exist without funding: a Catch 22 situation.

*Policy:* Although healthcare guidelines and policies for genetic testing and coverage exist,<sup>2,3,5</sup> these are limited in scope and there is no effective regulatory approach or legal constraint for much of DTC genome testing.<sup>13</sup> Whole genome risk testing epitomizes the healthcare system’s stymie with “... reconciling innovation and healthcare agendas... The current ‘downstream’ health services research & policy approach to innovation misses the mark on one crucial point... how to promote the design of innovations that are likely to be more valuable than others”.<sup>14</sup> (p.251)

*Value:* Value is often defined as an outcome/cost ratio, such as cost-effectiveness (CE) or incremental cost-effectiveness ratio (ICER). CE may be based upon achievement of significant differences in outcomes, such as morbidity, mortality or health benefits related to a specific treatment, strategy or technology.<sup>15</sup> This bespeaks a primarily economic perspective, its underlying values and a given time frame for the measure(s) – usually condition specific. In a prevention context, the CE threshold is usually \$50,000/year of life gained.<sup>16</sup> Individual values and quality of life (QOL) factors are typically not included.<sup>15</sup> A practical question arises as to whether use of alternate definitions and measurements for "value", such as individual health values, may better delineate the impact of health promotion behaviors and prevention. Related research, including DTC genomic risk testing, may shed light on more effective allocation of healthcare resources (e.g., avoiding expenditures if testing is not a health value) and policy development (e.g., targeting newly identified responsive risk sub-groups).

DTC genome testing challenges the healthcare system structure and functionality. Consumers directly receive simultaneous information for numerous health conditions, risk factors and, despite disclaimers, some findings consistent with a medical diagnosis. Based on the extent, growth and public interest in genome testing, it is unlikely that future policies or laws will re-capture control or prohibit DTC access. Is genome testing and information solely a medical resource or is it an individual right? There is no healthcare model (except aspects of newborn screening, HIV or pregnancy testing), no uniform approach, and no comprehensive controlling policy(s) – for whole genome risk testing.<sup>13</sup>

#### **Theoretical/conceptual models in healthcare**

Recent journal publications have tackled the subject of healthcare models in an effort to address recognized problems, deficiencies and issues in the current healthcare system.<sup>17-21</sup> The discussions involve primarily the traditional biomedical model versus a value-based medicine (VBM) model. The *biomedical model*, characterized by a disease/illness focus with the physician as



the knowledgeable caregiver, is based on and prioritizes a scientific, randomized clinical trials, population focused, evidence-based, cost-benefit philosophy. *Values-based medical models (VBM)* are grounded in psychology, integrating the impact of individual values on behaviors and health outcomes. Although VBM values are consistent with prevention, they may have unrecognized biases. They retain an illness focus, physician led context, goals reflecting and measuring traditional healthcare values and concepts, and an evidence-based philosophy.<sup>19</sup> *The Genomic Medicine Model (GMM)* is based on the biomedical model yet integrates individual values, (genetic) risk identification and prevention.<sup>21</sup> However, whole genome risk screening is not endorsed since potential harms outweigh benefits. Meanwhile, consumers, patient advocacy organizations, biotechnology companies and others that prioritize individual values and rights promote a responsible, informed individual freedom and choice regarding genome testing.<sup>10</sup> The need for a new healthcare model that integrates the biomedical model, individual values, collaborative decisions, technologic advances, policy development, prevention and health promotion is globally recognized.<sup>21-23</sup>

### **Health Capability conceptual framework**

Health Capability is a conceptual systems model<sup>1</sup> (Figure 1) based on Amartya Sen's Capability Theory presented in 1984, together with its Aristotelian origins and respect for individual autonomy, social justice, public health and economics. Health Capability has two essential principles – individual health and well-being, and health agency. This individual context and prioritization forms the basis for extension to population health – the converse of biomedical and VBM models.

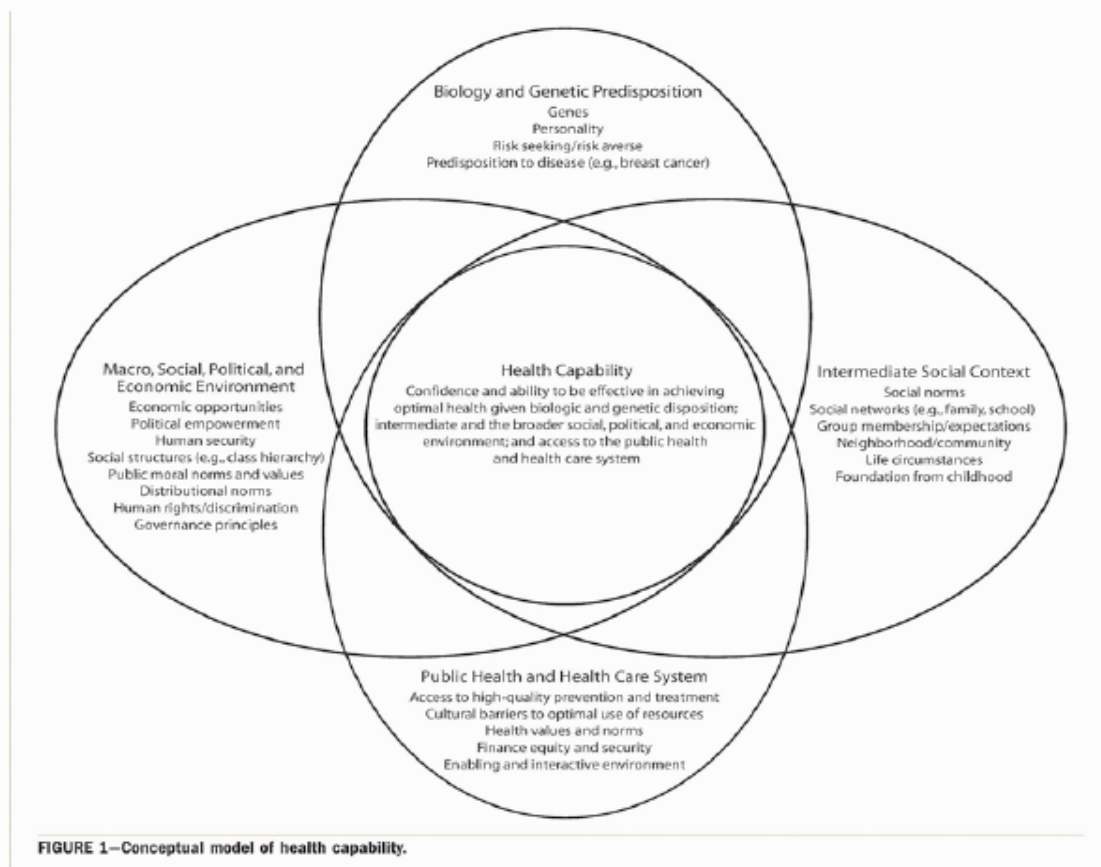
*Health* is an abstract, complex and multi-faceted concept. As often occurs in literature, Health Capability uses health as a given entity without defining, yet captures it as the reality that "people seek good health".<sup>1 (p.242)</sup> Integral to this premise exists the reality of challenges to good health (i.e., risks) and the objective to maintain or promote health. Factors are identified comprising the Health Capability Profile:<sup>1</sup> *Internal* (individual health status, health seeking skills, self-efficacy, health

Figure 3.1: Conceptual model of health capability.

Figure 1  
*Personalized Genome Risk Testing: Examining a Model to Inform and Ameliorate the Dichotomy Facing Healthcare*

17 September 2013

Figure 1: Conceptual model of health capability<sup>1</sup>



1. Ruger JP. Health capability: conceptualization and operationalization. *Am J Public Health*. 2010; 100: p47.

knowledge, health values and goals, self-management, effective decision-making, motivation) and *External* (social networks, memberships, political and economic factors, utilization and access to services). Considering Health Capability, genomic risk factors can be perceived as a threat to good health, triggering a health need and value of genomic risk testing. Ability to accomplish this goal is influenced by *Internal* and *External* factors (see Figure 2).

The principles of *individual health* and *well-being* embrace respect for individual health goals and values. *Health functioning* is seen as the outcome of action to maintain or improve health.<sup>1</sup> Outcomes encompass an interactive, multidimensional process with mutually understood goals wherein achievement is individually defined. The dimensions and components embodied in health functioning are numerous, attesting to flexibility yet complexity, with ability to focus and operationalize for clinical and research purposes.

*Health Agency*, a construct unique to Health Capability, establishes commitment to supporting ability for individuals to access and achieve personal health-related goals as agents of their own health, respecting their individual role & responsibility in health choices (agency freedom).<sup>1</sup> In contrast to biomedical and VBM models, this tenet can support individual choice to pursue genomic risk testing. Health agency recognizes motivation and self-efficacy as influences in attaining health goals. Self-efficacy studies support its: (a) impact on initiation of coping behavior and extent of effort expended despite barriers, (b) association with psychological and behavioral factors in predicting persons likely to seek genetic tests, and (c) self-rated competency in healthcare system use.<sup>24</sup> These studies directly relate to examining genomic information and pursuit of genomic risk testing as an individual value and goal, and potential influence of results on health behaviors.

#### **Health Capability: research and unique concepts**

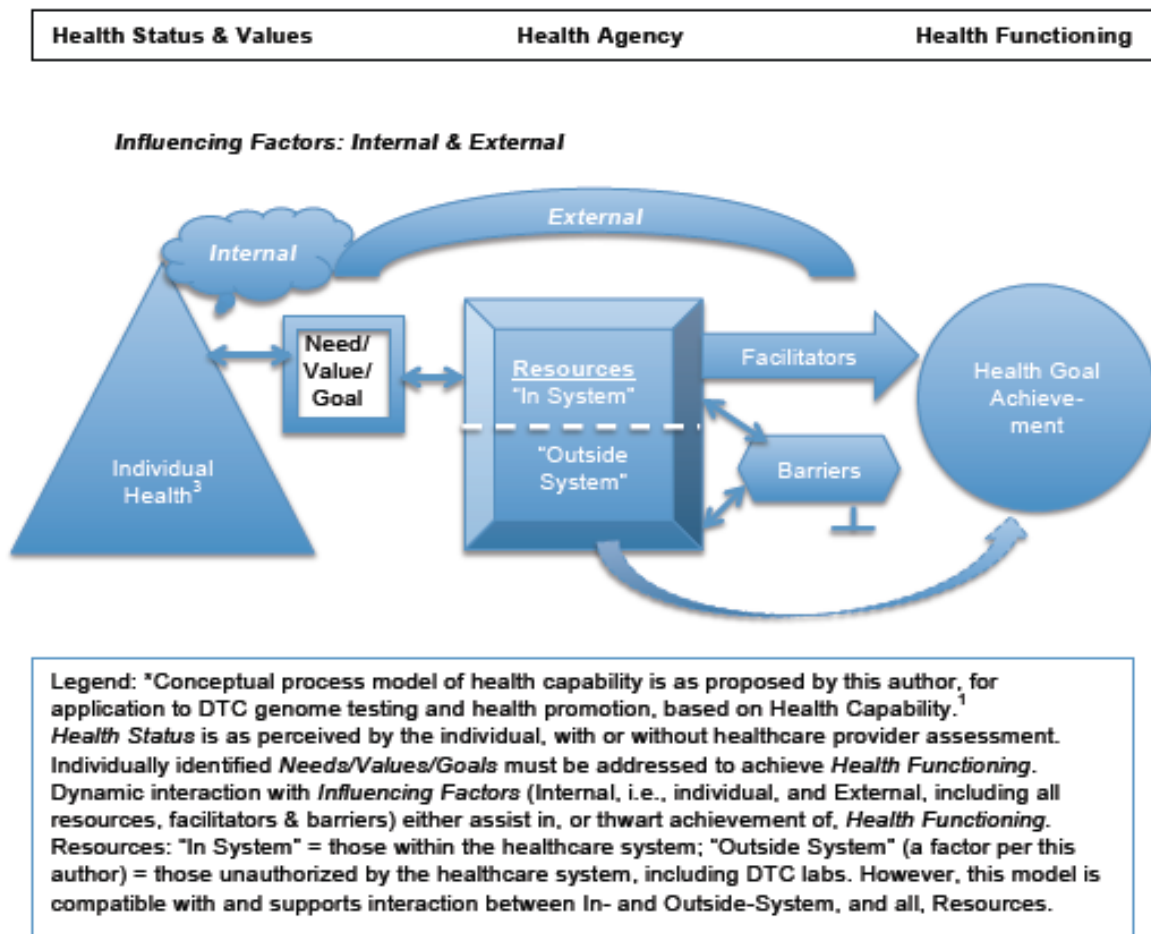
Health Capability has had limited research opportunity, yet evidence exists to support its potential, transferability and fruitfulness. Extending Capability theory to chronic disease research,<sup>25</sup> personal

Figure 3.2: Conceptual process model of health capability.

Figure 2  
*Personalized Genome Risk Testing: Examining a Model to Inform and Ameliorate the Dichotomy Facing Healthcare*

2 October 2013

**Figure 2: Conceptual process model\* of health capability: identifying individual health status, values & goals toward achieving health functioning**



resources and social environment were demonstrated to impact patient capability with diet and physical exercise, thus supporting the essence of health agency (and health functioning).

*Health Agency* factors can be operationalized to genomics. Social support is demonstrated in that the only “legitimate” method to obtain genomic risk testing is through a licensed healthcare provider,<sup>2</sup> or grant supported research. In contrast, another key element of health agency is ability for individuals to act as their own agent of health. This principle underscores the need for research to understand individual health values and goals related to genomic risk testing – particularly via the DTC route – and its relevance to health behaviors in order to enlighten imminent clinical practice applications and healthcare policy decisions.

In Health Capability, justice is not synonymous with equality. In contrast to equality of services, extent of such or outcomes of interventions, justice is evaluated vis-à-vis equity of access and healthcare system support to achieve individual goals. This view offers unique opportunities for economic and public health research.

Health Capability’s multidisciplinary basis, in particular economics, offers distinct strengths. Beyond typical cost-benefit analyses, Health Capability recognizes alternate methodologies and economic models for quantitative and qualitative analyses, such as examination of healthcare and personal costs for insured versus uninsured.<sup>28</sup> The influence of health insurance coverage and policies has immediate relevance for genomic risk testing. Current policies (Medicare, third party payers, limited disease-specific) do not cover full genome testing for early identification of potential risk.<sup>5</sup> Instead, policy development is primarily reactionary – driven by numbers (e.g., of affected, or population-size), outcomes evidence and cost/benefit analyses.<sup>16</sup> This approach is not amenable to timely evaluation, prioritizing individual health values, or integrating innovations and advances in biotechnology, as with whole genome risk testing.



**Health Capability: a work in progress**

Health Capability's conceptual framework has a disease/illness origin; however, it is adaptable to incorporate health promotion and prevention compatible with evolving directions in genomic testing and healthcare. The proposed model (Figure 2) provides a conceptual interpretation of the process, principles, concepts and interactions involved in Health Capability – from its implied starting framework of individual health status and health values to achievement of health goals (i.e., Health Functioning). This model uniquely includes formal recognition of resources outside the traditional healthcare system (i.e., DTC labs), yet fosters interaction among resources. It also supports important directions in genomic healthcare – movement beyond a disease/illness model; alternate service access and delivery environments; respect for individual health values and goals; evolving genomic testing and bioinformatics; partnership and shared responsibility between consumers and practitioners; and a multisystem collaborative approach for health promotion and prevention.<sup>27</sup>

**Discussion**

The biomedical healthcare model, its scientific basis and “gold standard” of RCTs is well-respected, yet publications note deficiencies<sup>12</sup>. A new model is needed in this era of genomic testing, personalized healthcare, bioinformatics, virtual capabilities and evolving service access and delivery environments.<sup>8, 27</sup> The ideal model will include mechanisms for qualitative components, recognition of individual values, prevention, new methods for cost-benefit analysis, collaborative partnerships and joint decision-making supportive of research, clinical application and policy development.<sup>1, 14, 28, 29</sup> This genomics environment and DTC risk testing challenge the healthcare system to not only re-examine the concept of value, but to change its view and definition of populations.<sup>8, 15, 17</sup> Scientific, prevention and values-based perspectives do not preclude a symbiotic compatibility. To the contrary, a systems approach validates complementarity and its dynamic potential.

Health Capability embodies systems and complexity thinking philosophies (Figure 1). Systems and complexity theories recognize multi-level subsystems and expect dynamic, flexible and holistic (versus analytic or reductionist) approaches that recognize the need to align a model with real behaviors.<sup>28, 30</sup> Interaction among subsystems, including the individual, fosters respect for all with evolving knowledge and learning contributing to understanding and unified visions and goals. These theories also include goal rather than detail orientation, recognition of non-linearity, and the influence of time and its irreversibility.<sup>28, 30</sup> Genomic risk testing typifies the environment conducive to these theories – ever-evolving technologies plus stakeholders at varying levels spanning multiple disciplines, agencies, payers, industry and government(s), as well as philosophies and values. Health capability's systems orientation (Figure 1) and complexity not only allows for functional decomposition (of components), measurement and analysis in research and clinical settings,<sup>30</sup> but on a grander scale, supports constructive dialogue among stakeholders (e.g., summits), problem-solving and coordinated policy development.

An individual values oriented healthcare system is depicted by Sturmberg using a vortex analogy.<sup>29</sup> The ideal (functional) captures seamless, dynamic interacting system(s) funneling to the vortex (goal) of individual values. In contrast, the current healthcare system in Australia (not unlike the USA) represents decision-making as a dichotomy of parallel, periodically intersecting yet separate subsystems with limited interactions lacking balance and with competing goals (vortices). The disparate goals lead to independent actions and pathways by the individual – as with DTC genomic risk testing – again underscoring the need for a realistic healthcare model.

The Health Capability paradigm respects the biomedical model and responsible decision-making in healthcare including biologic, scientific and economic considerations, yet does so by positing an alternate perspective (and ontology) on individual values and social justice. The humane approach of commitment to individual health values and goals allows for moving beyond viewing the individual as the impetus for or object of action, or the need to instruct, inform or guide the "patient"

based on established healthcare system norms, guidelines or values. Rather, if the individual perceives genomic risk as compromising to health, then healthcare system recognition of this health value will impact clinical care. This individual perception is consistent with the concept of ecstatic temporality – where the past, present and future are experienced in an overlapping, non-linear reality with the individual seeing one's own (limitless) possibilities, but which are compromised by illness (or risk).<sup>18</sup> Further, distinct from research to date, one could examine individual health values not as singular entity or factor, but instead as part of a complex individual subsystem comprised of identifiable components that can be defined, measured and interpreted (as per this author's research in process).

Ability to meld biomedical and values-based philosophies is supported in discussion of Heidegger's hermeneutic philosophy in that including a humane framework allowing for questioning about the impact of individual values (and subjective experience) does not question or disrupt, but rather compliments, the scientific framework.<sup>18</sup> However, biomedical and VBM models pose limitations for genomic risk testing research since both prioritize science and biologic factors as the key influence on results, thus essentially eliminating or minimizing possible individual influences.

Health Capability's view of justice – valuing individual health goals and supporting equity of access – offers unique opportunities for research to measure its impact clinically, economically and practically. This viewpoint does not mean blind deference to individual whims; it requires that practitioners educate the individual/patient to promote informed choices, consistent with the Hippocratic foundations of physician ethical values of beneficence, non-maleficence and autonomy. Thus, the real challenge is changing the values, structure and what is rewarded within the greater healthcare system. Health and prevention are touted and valued as the ideal. In reality, however, this mantra also provides a marketable strategy for broader initiatives and the greater economic benefits based in disease and illness – drug research, new pharmacologics, diagnostic technologies, treatments and medical care. Genomic risk testing requires a prevention and health



promotion perspective, respect for individual health goals, alternate access to health services, new measures of health value, new criteria for research funding, new strategies for policy development, and a new healthcare model to facilitate these processes.

### **Conclusion**

This article proposes and discusses extension of the Health Capability conceptual framework to fill a recognized gap and urgent need in existing healthcare models based on an evolving and complex healthcare system, as exemplified by the challenges, potentials and issues with whole genomic risk testing.

Current practices, guidelines and policies for full genome risk testing are limited, based on traditional healthcare models and values, are primarily reactive as opposed to proactive and allow (or inadvertently foster) independent consumer action and testing. This creates a dichotomy of healthcare and services, poses risks, and may actually impede future genomics research, understanding and responsible integration into healthcare. A new model that integrates biomedical, values-based and prevention models is needed, but will pose challenges to existing healthcare system values, structure, economics and policies. This model will also impact business, technology and academic priorities. Meanwhile, the genomics and bioinformatics explosion continues offering real promise for affordable individual and public health. Without a viable model promoting proactive, responsible integration of genomic risk testing into mainstream healthcare, DTC testing will likely increase while medically directed evaluations, treatments and responsible health promotion – as well as policy development and examining new measures of health value, healthcare services and cost-effectiveness – are further compromised and stymied.

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## CHAPTER IV

### PERCEIVED HEALTH STATUS AS A MEASURE FOR ASSESSMENT AND MANAGEMENT OF HEALTHY PERSONS: A REVIEW OF THE LITERATURE

#### **Introduction & Background**

This chapter is an article submitted for review and publication to the journal *Research and Theory in Nursing Practice*. The following pages are copied from the PDF file, exactly as submitted in compliance with journal requirements and author guidelines.

This article is a systematic review of the literature pertaining to the concept of perceived health status. It also includes a focused and detailed presentation of approaches and instruments to measure this concept. Knowledge of this concept, approaches used to determine its influence, and measurement instruments are essential to this dissertation and the Health Capability paradigm. This knowledge is relevant to operationalizing perceived health status, as well as to the data analysis and interpretation of the study results (Chapter V). The research results will contribute to understanding the dimensions and influence of perceived health status, its role in the Health Capability (Chapter III), and its relevance to clinical practice and research. The dissertation study results related to self-perceived health status (as well as health values, Chapter II) will also contribute to the further development of a functional/operationalized model for Health Capability, to assessing appropriate methods and instruments for measuring these concepts, and possibly to broader applications across traditional and evolving healthcare (Chapter VI).

## Perceived Health Status as an Assessment Tool

### Abstract

Health status, an individual's general health including personal qualitative and quantitative factors without targeting a specific health problem, is a universal concept integral to healthcare and individual well-being. Existing measures of health status are framed from bio-physiologic, illness and public health frameworks. This leaves gaps in understanding influences of individual perceived health status on health needs, values, goals and behaviors that impact clinical practice decisions and care of healthy persons. This article presents results of a systematic literature review of individual perception of health status, including study foci, concept operationalization, measurement instruments, and limitations and gaps in practice and research. Strategies are offered for healthcare integration of perceived health status. Databases reviewed include: CINAHL, PubMed, EBM Review and PsychINFO. Health status assessment, encompassing an individual's perceived health status, has immediate and long-range relevance for universal nursing research, theory and practice, even extending to prevention, health promotion and integrating genomic information.

*Keywords:* health status, perceived health status, health status measure, conceptual model, prevention, genomic healthcare

## Perceived Health Status as an Assessment Tool

### Introduction

Health status, an individual's general health condition taking into account various qualitative and quantitative aspects of their life without targeting a specific disease or problem (Gartsman, Brinker, Khan, & Karahan, 1998), is a universal concept integral to healthcare and with known clinical utility (Ruggieri, Elkin, & Chute, 2000). "Perception, the intuitive knowing that takes into account prior knowledge and the input of senses, influences how a person behaves, makes decisions, and thinks about him/herself" (Footitt & Anderson, 2012). An individual's perception of their health status is an important component of the initial evaluation of patients by healthcare providers that contributes to guiding decisions including testing, intervention, treatment and evaluation of outcomes (Di Giulio, 2014; Funk et al., 1997; Thier, 1992). Given the globally recognized need to view health beyond illness and disease (World Health Organization, 1948), healthcare systems internationally recognize the need for, and have committed to, fostering individual and public health through risk reduction, prevention and health promotion initiatives (Khoury et al., 2009; Kumpusalo, Pekkarinen, Neittaanmäki, Penttilä, & Halonen, 1992; NINR, 2011; US Department of Health & Human Services, 2014).

Health status assessment including risk identification, individual needs and health promotion is consistent with the holistic philosophies of Nursing as well as this discipline's complementary contributions to an integrated healthcare system (Glennister, 2011; Lau, Hartman, & Ware, 1986; Rosenstock, 1974). Perceived health status contributes significantly to an individual's health behaviors and health outcomes (Bruce & Fries, 2003b; Di Giulio, 2014; Ruger, 2010). Integrating perceived health status into clinical health assessment and nursing research supports the National Institute of Nursing Research's (NINR) bio-behavioral, interdisciplinary and health promotion goals in support of "individuals to become guardians of



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their own well-being” (NINR, 2011). It offers Nursing an opportunity to further the Science and take a role in innovative research at the growing interface with prevention, health promotion and evolving healthcare, as with advances in genomics (Genomic Nursing State of the Science Advisory Panel et al., 2013). Despite the recognized value of individual perception of health status, neither this measure nor dimensions to capture its relevant subjective individual perspectives, are incorporated into routine healthcare or Nursing practices (Di Giulio, 2014).

The purpose of this article is to present an examination of perceived health status from the individual’s perspective, targeting healthy persons, as a concept and tool for assessment, understanding and predicting individual health, health values and health behaviors for use in research and clinical contexts. This article includes: (1) a review of the literature on an individual’s perceived health status, including a review of the theoretical basis of this concept, a discussion of research findings, and gaps related to healthy persons and prevention; (2) a review of instruments used to measure perceived health status, as well as their foci, strengths and limitations; and, (3) potential strategies for integrating the concept of perceived health status in healthy persons with evolving healthcare directions in genomics, nursing and prevention.

### Background

Perception of health status is a well-recognized subjective phenomenon, comprised of several domains of health, including sociocultural and socio-psychological factors not accessible to an external observer that reflect actual individual health (Miilunpalo, Vuori, Oja, Pasanen, & Urponen, 1997; Okosun, Choi, Matamoros, & Dever, 2001). Perceived individual health status is a powerful measure of individual mortality, survival and global (i.e., general) health status, and is a significant independent health factor (Idler & Benyamini, 1997). Self-rated health is complementary to, and captures something beyond, traditional biologic indicators of health



## Perceived Health Status as an Assessment Tool

assessment, recognizing the complex interaction of physical, psychological, social and personal factors, plus the weighting of values and preferences, that impact individual perceptions of health status, health outcomes and healthcare utilization (Houssein, McKenna, & Scott, 1997; Hunt et al., 1980; Idler & Benyamini, 1997). These same factors pose gaps in current knowledge, research and application to practice, and cannot be adequately captured by existing biomedical or psychological measures (Bentzen & Christiansen, 1995; Di Giulio, 2014; Houssein et al., 1997).

Discrepancies exist in perceived health status as rated by individuals versus ratings by healthcare practitioners (Idler & Benyamini, 1997; Kivinen, Halonen, Eronen, & Nissinen, 1998; Okosun et al., 2001). Individual perception of health, such as personally unacceptable health-related changes, may influence when healthcare services are sought and the choice of intervention, as well as the associated motivations, health goals and values, and decisions impacting health action or behaviors (Hunter, McKee, Black, & Sanderson, 1995). These discrepancies also raise questions about deficiencies with existing instruments in measuring and capturing perceived health status and its dimensions. Discrepancies in perceived health status between individuals and their providers contribute to compromised healthcare, poor individual-provider relationships, sub-optimal compliance in healthcare regimens, and independent individual health actions [i.e., to address personal health concerns, undiagnosed symptoms, risk and prevention information needs, or health promotion (Landro, 2013)]. Although the need for examining health and health status from the subjective individual perspective is recognized (Mäilunpalo et al., 1997), the healthcare system is struggling with how, what models, what values and what mechanisms to use for evaluation (Loughlin et al., 2013).

The present day realities and potentials of genomic healthcare and genome testing add another layer of complexity to perceived health status. A multidisciplinary workshop convened

## Perceived Health Status as an Assessment Tool

by the NIH and Centers for Disease Control and Prevention to review the scientific basis of personal genomics (i.e., genome testing of individuals, including the direct-to-consumer route) in risk assessment and prevention recognized that “The clinical validity and utility of personal genomics is a moving target with rapidly developing discoveries but little translation research to close the gap between discoveries and health impact” (Koch, 2012). One recommended domain to target was “. . . 5) assessing how the concept of personal utility can affect health benefits, costs, and risks by developing appropriate metrics for evaluation”, ultimately stating that to fulfill the promise of personal genomics, a rigorous multidisciplinary research agenda is needed (Khoury et al., 2009).

### Methods

A systematic literature review of perceived health status was performed that used PRISMA guidelines (Liberati et al., 2009) and included searches of PubMed, PsychINFO, CINAHL and Cochrane databases. The initial PubMed search identified 20,808 records. Based on the goals that perceived health status relate to individuals, overall health status, identification of measurement instruments, and application for research and/or clinical contexts, inclusion criteria were: (1) perceived health status, (2) individual (or self-perceived) health status, (3) perceived general (or overall) health status, (4) instrument/measurement of perceived health status, (5) quantitative or qualitative research, or a systematic review of the literature, and (6) English language. The time frame of publications spanned 1976 to 2014 in order to capture origins of the concept and instruments, as well as all potentially relevant studies. Using the inclusion criteria, 120 records were identified.

Despite the search parameter stipulating perception of general health status, most identified records related health status to a specific disease, illness or the elderly. This raised the

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possibility that some records could have been missed due to database keyword limitations. Thus, alternate terms that might identify perceived health status – in general and in healthy persons – were explored using several resources (i.e., library expert, health-related articles and books, and references cited in previously identified articles). An additional 25 records were identified, for a total of 144 after removal of duplicates. Abstracts of these records were screened, leaving 48 records for full text review. The exclusion criteria eliminated studies that: (1) were disease or condition specific, unless a stated goal of the study included assessment measuring perceived health status, (2) had a primary economic focus, (3) used a surrogate as the primary measure of perceived health status (e.g., quality of life, health values), (4) involved and focused on a study-specific measurement instrument, (5) assessed the accuracy of individual perception of health status, (6) targeted pregnant, mentally compromised, or child (under age 13) populations, or (7) were non-English or were inaccessible publications (e.g., an older article, not available electronically and at significant cost).

## Results

The results of the systematic literature review targeting perceived health status that is presented and discussed in this article reflect themes relevant to Nursing, interdisciplinary healthcare practices, models and research. The themes of studies of perceived health status, as categorized by this reviewer, are addressed in the following sections: (1) the Concept, and its operationalization in healthcare research and clinical settings, as well as by comparison between individual and provider perspectives; (2) Clinical applications, including specific (types of) conditions, interventions, outcomes and populations; and, (3) Measures of perceived health status, including instruments, use and assessment.

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### **Perceived Health Status: Concept and Conceptual Framework**

#### **Concept**

Perceived health status originated as self-rated health in a psychosocial epidemiologic research context, evolving as perceived health in the medical context (Idler & Benyamini, 1997). The review of literature contributed to characterizing the concept and construct of perceived health status based on how it was operationalized, measured and interpreted, rather than being defined in the studies or identified as having a specific theoretical basis. Perceived health status was usually a secondary or complementary consideration, yet its unique contribution and utility for both research and clinical use was recognized (Di Giulio, 2014). Since perceived health status is recognized as a complex concept having several health domains, reflecting subjectivity in how it is defined, and posing challenges in measurement and interpretation, some studies represented perceived health status interchangeably with a related concept or by a surrogate measure. The surrogate measure, such as health values, health functioning or quality of life, was considered to encompass the essence of perceived health status from a specific health or general life context, and could be applied in various ways, e.g., as a one-time measure or as a measure pre- and post-intervention (Fu & Kattan, 2006; Hunter et al., 1995; Larson, C, Gustafson, & Batalden, 1996; Lichtenstein & Thomas 1987; Shmueli, 1999) (See Tables 1 & 2). These surrogate measures, however, also represent complex concepts with their own inherent potential biases, depending upon operationalization. An underlying theoretical framework or context was often implied by the study design, focus, methodology or measure of perceived health status.

#### **Biomedical model/context**

When perceived health status was included in research studies or clinical settings, it was typically one part of an overall health status evaluation from a medical context. Although the



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**Table 1: Perceived Health Status: Clinical Applications Studies**

Author, Year	Sample	Purpose [Focus]	Instrument/Methodology	Findings
Benyamini, 1999	Review of 19 studies related to self-rated health	To amend, extend terms and update the summary table of mortality studies by Idler (1997) related to self-rated health status [Mortality]	Review of Literature (Medline).	Findings consistent with previous report (Idler, 1997), supporting the association between poor rating of self-health and increased risk of mortality (only 2 studies showed no effect).
DiGiulio, 2014	83 Italian cardiology centers included their 1,465 outpatients (with chronic HF, NYHA classes II-III, with coronary and non-coronary etiology), randomized in the GISSI-HF trial in an observational outcome study	To assess the independent prognostic value of health status measured with the KCCQ on mortality and hospital admissions over a period of 3.3 years [Mortality & morbidity]	Kansas City Cardiomyopathy Questionnaire (KCCQ) (23 items; 5 domains) administered at baseline	When stratified by pre-defined criteria, lower KCCQ scores (<25) compared to best scores (>75) were predictive of mortality, but not of hospital admissions risk. Lower KCCQ scores discriminated risk between NYHA II and III classes.
Funk, 1997	180 (of 228) patients with primary untreated head & neck cancer (HNC), from the Dept. of Otolaryngology, U Iowa College of Medicine, underwent pretreatment & 6 month follow-up evaluation.	To investigate the pre- and post-treatment global health status of HNC patients compared to age matched, U.S. population norms, using a self-administered general health status survey. [Condition-specific morbidity]	Medical Outcomes Study 36-Item Short-Form Health Survey (SF-36), including physical health (PCS) & mental health (MCS) component summary scores.	In HNC patients aged 45-54, 55-64 and 65-74, pre-treatment PCS & MCS scores were significantly worse (all 8, 5 of 8, and MHS scores, in respective groups) compared to age-matched norms (p < .05). HNC patients had worse physical scores at 6 months post-treatment, but showed significant improvement in pain and mental health scores (p = .049). Self-rated health is useful for global health assessment in this population.
Gartsman, 1998	544 patients (age 18-86: 322 men; 222 females) with 5 different common shoulder conditions	To evaluate patients with shoulder conditions to establish norms for SF-36 scales & summary measures, and to compare results with other medical disease specific SF-36 data. [Condition specific morbidity & norms]	SF-36 (including subscales & Physical & Mental Health Component Summary)	Patients had statistically significant decrease in health, specifically Physical function, Role-Physical, Bodily Pain, Social function, Role-emotional & PCS. Individual perception of general health ranked in severity as for persons with several major medical conditions.

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Griep, 1997	171 healthy persons, ages 53-86	To assess the general health status via the SENIEUR protocol and to assess the nutritional status via anthropometric measures [Morbidity & early detection in elderly]	SENIEUR questionnaire (for ageing studies in the European Community): focus on health problems, unhealthy habits, physical exam & lab tests	Significant association between age, general health and dental state with diminished odour perception. Odour perception is important to nutritional state; diminished odour perception may be early indicator of physical deterioration.
Hunter, 1995	217 (of 258) British men aged 55 years or older, and known to have reported mild, moderate, or severe lower urinary tract symptoms	To determine the extent that urinary symptoms, and associated bothersomeness, interfere with activities of daily living (ADL) & affect health status & quality of life [Condition specific morbidity]	SF-36	Most men reported no interference with ADL. Statistically significant decrease in scores of physical role, social functioning, mental health and general health perception with increasing symptom severity. The association between health status and bothersomeness was more marked than for symptom scores.
Idler, 1997	Review of 27 U.S. & international community studies of self-rated health & mortality	To collect & appraise published studies of self-rated health and mortality. [Mortality]	Literature search of studies of self-rated health status (using single or multiple measures) related to mortality	Overall, the predicted effect of self-ratings was the rule, supporting that measuring self-rated health was relatively insensitive to variations in different instrument questions. Self-rated health had an effect independent of covariates.
Kivinen, 1998	470 initially (90% of invited) Finnish men aged 70-89 years (mean 76.2). Analysis on those living (365).	To study self-rated & physician-rated health in older Finnish men (having a high prevalence of coronary heart disease) to analyze concordance of the two methods & examine factors explaining differences [Comparison of perception of health status: individual versus healthcare provider]	Survey/Questionnaire. Health Status: Individual: One 4-option opinion question (ranging from feeling very healthy to not healthy). Physicians judged subjects by 5 categories (very good to very poor). Survey also included 12 activities of daily living & 30 symptom items.	Self-rated Health: Very, Moderately or Not healthy in 17%, 47% & 37%, respectively. Significant difference between self-rated & physician-rated health; only 36% congruence. Of men rated as healthy by physicians, only 25% considered self as healthy. Congruence better for poor health category. The older the man, the more likely physician rated health as poor.

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Kulik, 1987	110 college students: 67 healthy (23 males; 44 females) taking introductory psychology & 43 student volunteers (21 males; 22 females) considered ill (awaiting treatment for minor conditions, e.g., flu, strep throat, at the student health center).	To determine: (1) the effect of acute illness on unrealistic optimism & on perceptions of preventability of negative life events, and (2) whether ill individuals are relatively more or less interested in receiving risk-reduction information for health & non-health problems. [Comparison of perceived health status: ill versus healthy persons]	Form asked participants to assess and compare their risk & preventability assessments (with others of same age & sex) for 6 health (e.g., VD, tooth decay, lung cancer, heart attack, ulcer) & 4 non-health (divorce, suicide, car accident, mugging) negative events. Participants also completed the Health Opinion Survey as to preference for active versus inactive role in the health care process.	As a whole, participants believed their risk for health & non-health conditions was less than average. The healthy showed optimistic bias in 9/10 events. There was no main effect for health status; yet there was a significant health status x problem interaction: illness was associated with perception of greater risk for other health problems. Ill students tended to see health problems less preventable. The more at health risk individuals felt, the more interested in prevention information, but not so for the ill.
Larson, 1996	167 acute myocardial infarction patients from 3 US community hospitals	To determine if there is an association between meeting patients' information needs and their overall satisfaction with care & their general health status outcomes. [Patient satisfaction, health status & outcomes]	Two Study-specific questionnaires (with some items based on the SF-36): (1) patient satisfaction at two weeks post discharge, and (2) clinical status, health status & extent of help needed & received at eight weeks post discharge	Meeting patient information needs positively & significantly associated with patient satisfaction & general health status (evaluated by biomedical measures & psychosocial functioning, except for one quality of life question).
Lichtenstein, 1987	Sample of Medicare beneficiaries	To: (1) examine changes over time in self-reported perceived & functional health for an elderly population, & (2) be able to draw inferences about the appropriateness of each measure for administrative and/or research situations. [Comparison of health measures]	Examination of two health status measures: perceived health status and functional health	The perceived health status measure is more suitable for descriptive studies of health in elderly. Functional health status measures are more relevant to clinical use and studies examining the relationship between an individual's physical health status and subsequent behavior.

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Lindsay, 2001	183 patients awaiting elective coronary artery bypass grafting (CABG) recruited 1 month before expected date of operation.	To assess: (1) health status, level of social support and presence of coronary artery disease (CAD) risk factors before and after CABG, (2) symptom relief 12 months postop; & (3) association between pre-op health status & recurrence of symptoms [Condition specific morbidity & intervention assessment]	Preoperative assessment included severity of symptoms, coronary artery disease risk factors, the SF-36, and social activities questionnaire.	Angina & breathlessness completely relieved in 55% & 36% of patients, respectively, and significantly reduced in those with residual symptoms. Patients with low SF-36 scores & low social network scores pre-op were less likely to be relieved of symptoms. Health status scores pre-op were lower across all 8 domains compared to other reported CAD patients & the general population. Duration of exercise, METS and chest pain during exercise ECG test are significantly associated with patients' GHP & PF. However, wide variability on individual level, and all non-invasive measures only explained 14% or less of variation in PF & GHP.
Mattera, 2000	195 (of 238) consecutive patients (68% male, mean age 55.6 ± 11.1 years) with known or suspected ischemic heart disease referred for exercise testing with myocardial perfusion imaging	To determine association between several cardio-vascular noninvasive testing measures (e.g., exercise duration, ECG, myocardial perfusion) with patient reported physical functioning (PF) & general health perception (GHP)	Medical Outcomes Study Short Form (SF-36)	Subjective health assessments are valid health status indicators in middle-aged populations, and can be used in cohort studies & population health monitoring. Self-reported chronic disease or disability, and use of physician services, were weaker predictors of mortality than subjective health assessments. Stability of perceived health status was relatively high.
Miilumpalo, 1997	Finnish residents (from an industrial & 2 rural towns): 1340 men & 1500 women (77% of total), aged 35-63* at study onset. (Since mortality rate was low in subjects < 35, they were excluded).	To assess: (1) the value of various self-reported health assessments, specifically via association with annual outpatient physician visits and age-specific mortality; & (2) stability of perceived global health status over time [Mortality, morbidity & change over time]	Postal Questionnaire (1980): 120 structured questions spanning SES, health behaviors & functional capacity (health status measured by one Likert-type item). Follow-up at 1-year: to obtain use of physician services & verify stability of respondents' perceived health status (88% response rate).	Subjective health assessments are valid health status indicators in middle-aged populations, and can be used in cohort studies & population health monitoring. Self-reported chronic disease or disability, and use of physician services, were weaker predictors of mortality than subjective health assessments. Stability of perceived health status was relatively high.



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<p>Okosun, 2001</p>	<p>Data from 10,298 surveys from the Third National Health and Nutrition Examination Survey (NHANES III): included Blacks, Whites &amp; Hispanics, ages 17-90, considered healthy (absence of common medical problems, e.g., stroke, diabetes, hypertension, musculoskeletal, thyroid, respiratory, depressive &amp; psychological disorders).</p>	<p>To examine: (1) the relationship between obesity and self-rated health, and (2) the degree of agreement between patients' self-rated health status and physicians' impression of patients' health [Population &amp; condition specific morbidity; Comparison of perceived health status: individual versus healthcare provider]</p>	<p>NHANES III Survey of the National Center for Health Statistics. Self-rated general health was based on one 5-point Likert-type question (ranging from excellent to poor). Physicians' rating of impression of subjects' health was based on 2 factors: the same question &amp; scale as subjects, as well as the results of physical examination.</p>	<p>In men &amp; women, significant association between obesity &amp; self-rated health. Subjects reporting excellent health decreased with increasing level of obesity in all 3 ethnic groups. Compared to whites, blacks &amp; Hispanics were at increased odds of reduced self-rated health. Concordance between self-reported health and physicians' impressions was poor.</p>
<p>Schiffman, 2001</p>	<p>76 patients, age &gt;18 years, from the uveitis clinic of the NIH Warren G Magnuson Clinical Center</p>	<p>To examine patients with uveitis as to: (1) whether they have lower general health status &amp; visual functioning scores than normal subjects, and (2) the association between uveitis severity and visual functioning &amp; general health status. [Condition specific morbidity]</p>	<p>The SF-36 (PCS &amp; MCS) &amp; the National Eye Institute Visual Functioning Questionnaire (NEI VFQ-25)</p>	<p>Uveitis has a significant impact on both general health and visual functioning; both were markedly lower than reference groups. Although there is an association between these two measures, they are complementary in that each provided distinct information.</p>

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**Table 2: Perceived Health Status: Studies Examining Measures & Instruments (also see Table 3)**

<b>Author, Year</b>	<b>Sample</b>	<b>Purpose</b>	<b>Instrument/ Methodology</b>	<b>Results/Findings</b>
Bardsley, 1993	284 (of 378) randomly selected diabetic outpatients attending a hospital diabetes service. Candidates were sent questionnaires.	To compare 3 approaches to measuring health status in diabetic patients & to assess instruments for clarity & ease of completion, distribution of scores, correlation with assessment by a trained interviewer & association with measures of the impact of diabetes, i.e., metabolic control	Three instruments: the Nottingham Health Profile (NHP); 4 categories of an Anglicized version of the Sickness Impact Profile (SIP) aka the Functional Limitations Profile (FLP); & the Positive Well-being (PWB) scale. A subset of 21 persons was selected for home interview by a trained professional (blinded to results).	NHP & FLP results suggested general validity in patients with diabetes, yet more so for the limited number with significant health problems involving pain or disability. The PWB scale did not identify these findings (as expected), yet identified factors relevant to lifestyle & well-being.
Bentzen, 1995	2263 male & female heads of households who participated in the Danish Health Study 1982-83.	To examine whether a current health scale produces reliable and valid measures of self perceived health status	Survey for Danish study adapted from Rand's Health Insurance Study (aka SF-36).	The scale has acceptable reliability & validity properties; it is preferable to a single item measure of general health status. However, current health had low loading with health perception (physical, mental, social).
Bergner, 1981	Successive field trials: 696 persons (80% of invitees) from a random sample of prepaid group practice enrollees; 199 persons (77% of invited) from a family medicine clinic who considered self as sick; 2000 people from an Alabama study	Final development & assessment of the reliability and validity of a behaviorally based measure of health status. Also to compare 3 methods of administration.	Sickness Impact Profile (SIP)	High test-retest reliability & internal consistency. Analysis supports clinical, discriminant & convergent validity. Relationship between SIP & clinical criterion measures ranged moderate to high. Self-administered forms that are understood may be more valid than with interviewer.
Brazier, 1993	Postal survey of 1582 people (of 1980; 83% response rate; 95% completion rate), aged 16-74, randomly selected from two general practices in Sheffield, UK	To test a recently developed single index measure of general health, the EuroQol, & examine it for validity & sensitivity by comparing to the SF-36 (Short Form) Health Survey Instrument	EuroQol Questionnaire (EQ)	Evidence supported construct validity of EQ dimension responses & the derived total EQ health score (distinguished between groups). EQ response & total scores agreed with the UK SF-36 profile scores. EQ was less sensitive to low levels of perceived ill health & across the range of health states. EQ revised.

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Chern, 2000	4,225 employees (of an original 14,331), age 18-64, from two sectors (one state agency, one private company) enrolled in three health plans of BlueCross / BlueShield of Virginia	To test the stability & validity of health status measurement in a working population.	SF-36: the original eight scale and a modified five scale short-form health survey. Distributed by mail.	The revised 5-scale model was highly stable over time in this relatively healthy population. Their perception of health relates to "physical health status", represented by physical functioning or role limitations.
DeCuir, 2007	92 adolescent Black American males (age 13-18, without known cardiovascular disease) from two sites in Tennessee & Louisiana.	To describe the health barriers, health perceptions (health self-efficacy & health status) & CV risk factors reported by Black American adolescent males and the influence of these variables on health promotion behaviors	Concepts (& Measures): Health barriers (question as to 3 major things preventing healthy lifestyle changes); Self-efficacy (12-item survey/Self-Efficacy Scale); Perceived health status (25-item Status-Rated Abilities for Health Practices Scale). Also used a 40-item Adolescent Health Promotion Scale and a 49-item Youth Risk Behavior Survey	Four categories of reported perceived health barriers: diet, environment, finances & other. Perceived health status and self-efficacy levels were moderately high, yet reported health promotion behaviors noted only moderate participation. These adolescents had physical health findings or family history suggestive of higher risk for hypertension, obesity & cardiovascular disease.
Essink-Bot, 1996	63 renal dialysis patients	To investigate the feasibility, internal structure & psychometric characteristics (internal consistency, test-retest reliability, construct validity) of two widely used generic health status measures for use with renal dialysis patients by comparison with each other & related established scales	Instrument foci: The Nottingham Health Profile (NHP) & the Sickness Impact Profile (SIP). For analysis purposes, other instruments included: the State-Trait Anxiety Inventory (STAI), the Self-rating Depression Scale (SDS-Zung), the Karnofsky Performance Scale, the Activities of Daily Life (ADL) & the Index of Well-Being (IWB).	NHP scales showed slightly higher feasibility & internal consistency than SIP (mean $\alpha=0.67$ & $\alpha=0.65$ , respectively). NHP demonstrated test-retest reliability, but somewhat weaker inter-correlation than SIP. SIP loaded more as a measure of physical functioning; NHP was more a mental factor measure (perceived health), excepting physical mobility. NHP & SIP correlated as expected with the five other measures.
Footitt, 2012	328 community-living adults (ages $\geq 65$ years)	To explore associations between perceived wellness and health-related quality of life, comorbidities and modifiable lifestyle factors	Perceived Wellness Survey and the 36-Item Short Form of the Medical Outcomes Study	Positive associations were found between perception of wellness and health-related quality of life. General health (perceived), vitality and mental health had strongest and statistically significant association with perceived wellness. The study found perceived wellness to be a multidimensional construct in older adults.

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Fu, 2006	20,428 people, age 18 & older (average 45.4; females 52%; 29% minority) from the 2003 US Medical Expenditure Panel Survey (MEPS), maintained by the Agency for Healthcare Research & Quality (AHRQ) that provides representative estimates of healthcare utilization, expenditures, payment sources & insurance coverage	To document whether there are racial & ethnic differences in individuals' perception of their general health status assessed by preference-based measures (Health-related quality of life; HRQoL) and assess the five attributes and scores of EQ-5D	EuroQol (EQ 5D), part of the MEPS Survey, including the HRQoL measure of preference for a state of health (3 choices related to problem: no, some, or severe) based on five attributes: mobility, usual activities, self care, pain/discomfort & anxiety/depression. Scoring: 0 = dead; 1 = perfect health	Majority had no problems with the 5 attributes. 49% rated self in full health. Pain scale had most with problems (41%); self-care had least (4%). Blacks & Hispanics less likely to answer no problems than Whites; Asians more likely to answer full health. Perception of HRQoL differed by race & ethnicity, yet differences in objective measures lessened with control of education & income, & disappeared if medical conditions & functional limitations were considered.
Gephardt, 2001	Two samples: 205 individuals (99 males; 106 females; ages 13-87; mean 42.3) of the general Dutch population; and 286 Students (107 males; 179 females; ages 18-57; mean 25.2)	To examine the psychometric properties of a Dutch translation of the Health Hardiness Inventory (HHI) and to determine relationships between health hardiness scales and self-reported health & preventive health behavior.	HHI Questionnaire - 3 scales (35 items): health commitment (10), health challenge (7) & health control (18, including a perceived health competence subset of 8 items). Revised (RHHD): Health Value (HV; 6 items); Internal Health Locus of Control (IHLLOC; 5); External Locus of Control (EHLLOC; 7); Perceived Health Competence (PHC; 6).	Preventive health behavior was related to (& independently predicted by) higher HV & PHC scores. In general population, self-reported health was not related to HV. Construct validity of RHHD was supported by the relationship between health hardiness and preventive health behavior & health.
Houssein, 1997	200 consecutive rheumatoid arthritis (RA) patients (148 females; 52 males; mean age 58.9 years)	Evaluate the relationship of a "generic health status measure" (the Nottingham Health Profile) to the clinical, laboratory & radiological changes in the EULAR (European League Against Rheumatism) core data set for RA.	Nottingham Health Profile (NHP)	Mean scores on NHP for RA patients were higher than for a random population & for a sample of patients with other common diseases. NHP scores had significant correlation with Health Assessment Questionnaire scores (another validated health status instrument) & other benefits, i.e. disability focus, correlation with RA disease activity & providing information complementary to conventional biological measures.

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<p>Hunt, 1980</p>	<p>167 persons (of a convenience sample of 213) from the Dept. of Community Health, Nottingham University. Four groups: (A) 50 men, fit &amp; active; (B) 28 people, presumed healthy (no medical diagnosis); (C) 49 people, most with some disability; (D) 86 people with at least 1 chronic disease (&amp; likely to have symptoms).</p>	<p>Development &amp; testing of a health status instrument for construct validity, targeting its six health profile packages by observing its power to discriminate between elderly groups differing in health status.</p>	<p>Nottingham Health Profile (NHP) with its 6 packages (see Table 4) – interviewer administered at subject's home; plus a short mental status test</p>	<p>Statistically significant differences between groups on all 6 packages. NHP capable of discriminating between groups: low scorers had better health; high scorers had greater number &amp; severity of perceived problems. Age, sex &amp; marital status were not significant overall. Most persons affirmed few statements (median 4, of possible 38). In these elderly, perceived health status fit with objective (clinical) health status.</p>
<p>Jenkinson, 1993</p>	<p>13,042 randomly selected subjects, ages 18-64 years, drawn from computerized registers of the family health services authorities for Berkshire, Buckinghamshire, Northamptonshire &amp; Oxfordshire.</p>	<p>To gain population norms (scores for the 8 dimensions) for the short form 36 health survey questionnaire (SF-36) in a large community sample &amp; explore the questionnaire's internal consistency &amp; validity.</p>	<p>Postal survey via booklet containing the SF-36 and several other items concerned with lifestyles &amp; illness</p>	<p>Response rate of 72% (9332); sample confirmed representative of population. High internal consistency of the different questionnaire dimensions. Normative data (age, sex, social class) consistent with previous studies. SF36 is a potentially valuable tool in medical research, with recognized limitations.</p>
<p>Kumpusalo, 1992</p>	<p>427 men and 366 women (total 80% participation), ages 20-64, from the Finnish Healthy Village Study</p>	<p>To present &amp; discuss the dimensions of health status based on principal component factor analysis of the Health profile</p>	<p>Comprehensive health examination, including a health profile questionnaire with 90 health status items (e.g., self-rated health &amp; functional status, social activities, sick leaves, perceived health &amp; emotional well-being) &amp; more than 120 physical, psychological &amp; social health status variables. Physical health status examination included 13 items (e.g., weight, height, BMI, HR, BP, skin folds) and 20 blood &amp; urine tests.</p>	<p>Based on minimum correlation values, 37 health status variables chosen for factor analysis. Identified 6 health status dimensions (physical functioning, emotional state, perceived health, anthropometric state &amp; BP, social functioning, &amp; biochemical state), accounting for 46% of total variance. Health profile analysis by sex revealed the same dimensions but different order: men ranked self-rated general health more closely with physical function; women with perceived health.</p>

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<p>Pincus, 1999</p>	<p>688 consecutive patients (51, mean age all patients; mean differed by disease type) with rheumatic diseases (162 RA, 114 fibromyalgia, 63 osteoarthritis, 34 systemic lupus erythematosus, 20 vasculitis, 16 scleroderma, 18 psoriatic arthritis, 261 other) completed questionnaire weekly for 2 years</p>	<p>To develop components of a multidimensional Health Assessment Questionnaire (MDHAQ) by adding new items in the existing HAQ instrument (i.e., advanced activities of daily living) to overcome “floor effects” of patients with normal scores despite functional limitations &amp; to screen for psychological distress</p>	<p>Multidimensional Health Assessment Questionnaire (MDHAQ): the Health Assessment Questionnaire (HAQ) + 6 advanced ADL item &amp; 4 items for psychological distress</p>	<p>MDHAQ items have good test-retest reliability &amp; face validity. Normal scores were reported by 23% &amp; 16% of patients by MHAQ &amp; HAQ, respectively. In contrast, fewer than 5% had normal score on MDHAQ (based on added advanced ADL items), thus supporting reduction of the floor effect. Scores for psychological items correlated significantly with advanced ADL scores; also with education, more so than with age or duration of disease.</p>
<p>Salaffi, 2002</p>	<p>78 consecutive patients with RA (55 women; 23 men; mean age 56 years, range 19–78) from care facilities of the Department of Rheumatology of Ancona</p>	<p>To compare the responsiveness of disease-specific, generic and preference-based instruments to changes in articular status and perceived health in patients with rheumatoid arthritis (RA)</p>	<p>Arthritis Impact Measurement Scale 2, the AIMS2, the Medical Outcome Study Short Form Health Survey (SF-36), and preference-based instruments (rating scale, RS and time tradeoff, TTO)</p>	<p>Using 3 European Rheumatology (EULAR) criteria as external indicators of improvement/response, mean change scores in generic &amp; specific health status instruments and utility measures were significantly related to response category. AIMS2 subscales (physical function, pain, psychological function &amp; social interaction) were slightly more responsive than those of SF-36. Physical &amp; pain dimensions were most sensitive for measuring change over one year. For utility, RS scores were significantly more responsive to detecting changes in preferences.</p>



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<p>Shmueli, 1999</p>	<p>1958 (of 2,030) individuals, aged 45-75 years (mean age 58; men 47%; average educational level 11 years), representing the Israeli Jewish urban population (800,000).</p>	<p>To: (1) explore the relationship between two measures of health status and a health-value scale (quality of life) in the general population, and (2) assign health values to various health states indicated by the measures of health.</p>	<p>National Survey (conducted face-to-face by the Gertner Institute) on health issues &amp; included: (1) the SF-36 profile, with its 8 health status scales &amp; 2 summary components, (2) a five-category individual perception of general health question, &amp; (3) a question rating quality of life (as a measure of health value). Final sample was 1958 for study of the HQL &amp; EVGFP relationship, and 1918 for the HQL &amp; SF-36 relationship.</p>	<p>Main outcome: assigning health values (scores) to subjective health-status. The general health scale had highest correlation with HQL. General health &amp; vitality were the main determinants of health value. The SF-36 sub-scales accounted for &gt;50% of variation in HQL; their average correlated (0.67) best with HQL. Lower health status related to greater variation in health values (scores).</p>
<p>Stuecki, 1996</p>	<p>52 (of 106 eligible) consecutive patients undergoing hip arthroplasty (mean age 60 years; 67% female; 89% white)</p>	<p>To analyze the problems of interpreting change scores of ordinal health status measures for clinical research or practice</p>	<p>SF-36, physical ability scale</p>	<p>Items ranked by level of difficulty &amp; baseline score. Ranking same by SF-36 &amp; Rasch analysis. Using Rasch &amp; an interval scale, items of moderate difficulty posed challenges in interpretation (inflated numeric gains for patients with moderate disability) Using patient perception of level of change in function showed important variation of numerical gains with baseline.</p>
<p>Wang, 2013</p>	<p>Data from the 2009 Family Core component of the National Health Interview Survey (NHIS) of non-institutionalized U.S. citizens, including over-sampling of minorities. Inclusion criteria: adults, seen by healthcare professional within the last two weeks before the survey</p>	<p>To examine health care access disparities with regard to health status and presence of functional limitations (as a common measure of disability and multi-morbidity), after controlling for individual race/ethnicity, insurance status and income in the U.S.</p>	<p>The 2009 Family Core component of the National Health Interview Survey (NHIS). Examined six measures of access to care in the twelve months prior to the interview. Access measures included: no usual place of care, unable to get or delayed medical care, unable to get dental or mental healthcare, unable to get prescription drugs.</p>	<p>People with functional limitations and/or perceived health status as poor/fair experience greater barriers to access. Insurance status (none, as well as type) was the single factor associated with all six measures of access to care. Disparities among racial/ethnic groups in most access indicators &amp; income levels became statistically insignificant after accounting for individual health status measures.</p>

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term perceived health status tends to imply a focus on health, typically the context of studies was a specific disease or type of condition (see Table 1). The health status assessment primarily involved objective healthcare system measures, typically physical (e.g., examination, weight, BMI), functional (e.g., ability to carry out activities of daily living), bio-physiologic (e.g., comorbidities, angina or dyspnea scale), or condition-specific medical tests (e.g., EKG) (Di Giulio, 2014; Larson et al., 1996; Lindsay, Smith, Hanlon, & Wheatley, 2001; Mattera et al., 2000) (see Table 1). Perceived health status was measured either by a single Likert-type question (for the individual and/or healthcare professional) to categorize perceived health, and/or was measured using an instrument befitting the nature of the study (i.e., perceived general/overall health status pre- and post intervention, disease-associated manifestations, functional status), typically based on established and tested instruments (discussed later in Instrument section). Thus, although not stated, the underlying theoretical framework was consistent with a biomedical model.

Research assessing commonalities and differences in how perceived health status is viewed, interpreted and influenced was also consistent with a biomedical model, as demonstrated in studies comparing perceived health status as rated by individuals versus by their healthcare practitioners (Kivinen et al., 1998; Okosun et al., 2001) (see Table 1). One study involving sick and elderly persons found ratings of perceived health status, based on a single Likert-type question, to be similar between patients and providers (Kivinen et al., 1998). Despite this finding of similarity in ratings, another study determined that the nature of demographic and health status factors, such as older age, morbidity and increasing extent of illness, was associated with higher correlations between ratings of perceived health status by individuals and their providers (Hunt et al., 1980). Other studies demonstrated significant discrepancies in perceived health status between individuals and their physicians, as measured by one Likert-type question,



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even becoming more disparate with better individual health (Idler & Benyamini, 1997; Okosun et al., 2001) (see Table 1). One of these studies included a second question for physicians to consider physical examination results in their specific rating of perceived health status (Okosun et al., 2001), thus supporting a biomedical context.

Another source of discrepancy in perceived health status, targeting potential subjective views of morbidity and age related expectations, was associated with an illness/disease framework. An obesity study assessed agreement between physicians' rating and individuals' rating of their health status using the same single 5-point measure, demonstrating that individual perceived health status correlated with the extent of obesity and that concordance with physicians' ratings was poor (k values of  $< .40$ ) and decreased with increasing obesity level (Okosun et al., 2001). Research studies in elderly populations also identified low congruence between self-rated health and physicians' rating of individual health related to older age and overall (general) health. A study of Finnish men at risk for coronary heart disease found only 36% congruence that became poorer with increasing patient age (Kivinen et al., 1998), and an English community study noted rating discrepancies as most evident in overall health status with individual perceived health status as being realistic (Hunt et al., 1980).

### **Value-associated models/context**

**Economic.** Several studies directly or indirectly supported use of perceived health status as a measure for assessing economic value, such as costs and/or predicting allocation of healthcare system resources (e.g., physician contacts, healthcare costs) (Miiilunpalo et al., 1997). Self-rated global (general) health status was found to provide a simple and cost-effective measure for determining economic health value based on a systematic review of the literature (Idler & Benyamini, 1997). This earlier literature review, representing utilization of perceived

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health status in economic and/or policy perspectives, was also identified in the current literature search (Fu & Kattan, 2006; Shmueli, 1999) (see Table 2). The merit of economic applications of perceived health status was also evident in studies by secondary aims and discussion. Although the purpose of a study of over 20,000 responses to a Medical Expenditure Panel Survey was to examine the knowledge gap and whether differences exist in the perceptions of health status based on ethnic or racial ancestry (Fu & Kattan, 2006), the researchers also noted that preference-based scores are important to cost-effectiveness in the context of quality-adjusted life year (QALY) analysis where this measure is increasingly becoming part of clinical trials and health economic evaluations. Other studies used health status as an outcomes measure meaningful to utilization of resources since usual biomedical measures could not capture patient changes (i.e., associated with common shoulder problems) (Gartsman et al., 1998), or for creating health status utility scores for the purpose of economic evaluations (Shmueli, 1999).

**Functional/Sociologic.** In a clinical context, perceived health status and health values may determine the nature and content of assessments, impacting access to care and policies authorizing healthcare services. In a study examining data from the 2009 Family Core component of the National Health Interview Survey, perceived health status was operationalized as health functioning and morbidities, with results identifying that persons having a worse health status encountered the greatest barriers to healthcare, across all six dimensions of access (T. Wang, Shi, Nie, & Zhu, 2013) (see Table 2). Although these findings are important to public health for the most ill or disabled persons, functional measures of health status contribute little to understanding of relatively healthy, functional persons.

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### Summary

Research studies support the relevance of the concept of perceived health status and its use in assessment of individual health for practical purposes, including understanding of individual factors not captured in routine questions or surveys, to assist in treatment decisions, and to facilitate coordinated plans for medical management (Hunt et al., 1980). This research also demonstrates that discrepancies exist in perceived health status as rated by individuals in contrast to their healthcare providers, and that self-rated health and perceptions are more aligned with actual health outcomes, attesting to the complexity, subjective nature, and distinct value of this concept. Perceived health status was also measured by, or used as, a surrogate for other measures including health values and health functioning. These studies demonstrated benefit for economic evaluation and utilization of healthcare resources that are important in the current healthcare environment. Thus, although typically not stated, the nature of perceived health status studies to date exemplify primarily a biomedical or economic, or to a lesser extent, a functional or sociologic theoretical/conceptual framework. These models and their definitional basis for perceived health status impact decisions integral to aspects of research and clinical practice (Fu & Kattan, 2006; Kwon et al., 2010; Nguyen et al., 2012), potentially limiting or biasing the nature of data sought by researchers and used by clinicians (e.g., a disease and/or dysfunction focus) and the associated interpretations (lacking information about good health or relevant subjective individual factors). These recognitions about perceived health status, health values, the influence of underlying healthcare models and the distinct contribution of an individual's perception of their health status signal the existence of gaps in, and the need for research to further, understanding and utilization of individual subjective health information at the interface

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with health priorities, healthcare decision-making, utilization of healthcare resources, quality assurance and respect for individuals (Barr, 1995; Di Giulio, 2014).

### Perceived Health Status: Clinical Applications

The literature review of perceived health status identified quantitative or mixed methods studies primarily having a disease, illness or risk focus, targeting the elderly, age-specific mortality and/or chronic disease/morbidity, and establishing validity with these variables over time (Benyamini & Idler, 1999; Idler & Benyamini, 1997; Lichtenstein & Thomas 1987; Miilunpalo et al., 1997) (see Table 1). In two complementary systematic reviews focused on mortality, all but four of forty-six studies demonstrated self-rated health as having an independent effect when all covariates were entered; self-rated health reliably predicted survival even controlling for known risk factors (Benyamini & Idler, 1999; Idler & Benyamini, 1997).

### Overall Health

In most studies involving health status assessment, the context was disease or illness primarily focusing on health status itself represented by bio-medical, quantifiable, objective health status measures (Di Giulio, 2014; Larson et al., 1996; Lindsay et al., 2001; Mattera et al., 2000) (see Table 1). Perception of health status was generally a secondary or complementary consideration. This scenario was exemplified by the only Nursing conducted study identified by the current literature search (Di Giulio, 2014) that targeted the mortality and hospital admissions of heart failure patients. However, findings included: (1) the unique contribution of individual perception of health status, (2) the utility of this concept for both research and clinical use, and (3) limitations of patient health classification based on physician's perceptions. This latter issue was consistent with another study where the healthcare provider assessment was used as an objective "gold standard" to evaluate accuracy or value of individual perception of health status

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(Okosun et al., 2001) (see Table 1). Perceived (overall) health status was also found to be a useful measure in the elderly, for early identification of physical health deterioration (Griep et al., 1997) and for descriptive health studies, in contrast to functional health status that was more relevant to physical health and related behaviors (Lichtenstein & Thomas 1987) (see Table 1).

## Pre- and Post-Treatment

Perception of health status has been incorporated into pre- and post-treatment or intervention research, typically through use of an existing instrument. A study of head and neck cancer patients used The Medical Outcomes Study 36-item short form health survey (SF-36) (see Table 3) and found that perceived health status before treatment was significantly worse than age-matched controls in the general population, that it involved both physical and mental health components, and that the latter component was most significant in the older age group (Funk et al., 1997) (see Table 1). These results were interpreted to support the importance of baseline perceived health status assessment in order to improve the understanding of outcomes post-treatment in these patients. Perceived health status was repeatedly proven to have significant prognostic and outcomes value (Di Giulio, 2014; Idler & Benyamini, 1997).

## Disease Populations

Several studies integrated perceived health status as a method for evaluation of illness/disease populations, consistently demonstrating ability to distinguish subgroups (e.g., cardiology patients, patients with urinary tract symptoms) and identify relevant qualitative factors (e.g., related to quality of life) for the purposes of treatment, type of intervention and/or prognosis (Di Giulio, 2014; Hunter et al., 1995). Other studies compared one illness/disease population with another, or with the general population, as a strategy to establish a tangible measure for healthcare practices and decisions (Gartsman et al., 1998; Lindsay et al., 2001). For



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example, the SF-36 was used for self-assessment of individuals with five different shoulder conditions, demonstrating a significant decrease in health status compared to the general population, specifically related to physical functioning and elements of the physical health components score (Gartsman et al., 1998) (see Tables 1 & 3). The authors noted that although the SF-36 does not directly measure shoulder function, findings support its use in this population based on: (1) its ability to identify significantly lower physical functioning scores; (2) its value as a baseline and outcomes measure in this population posing challenges to justify interventions and capture differences; and, (3) the fact that the perception of health status in their patients ranked comparable to scores associated with several major medical conditions.

## Specific Populations

Individual self-rated (perceived) health status has been used to assess differences between and within populations, as with race, ethnicity and common health conditions (Fu & Kattan, 2006; Okosun et al., 2001) (see Tables 1 & 2). A study of obesity and self-rated health found a statistically significant association between these two variables ( $P < .05$ ), significant differences in self-rated health status (lower) when comparing Blacks and Hispanics to Whites, and a tendency for the reporting of health status as “excellent” to decrease with increasing obesity in all three ethnic groups (Okosun et al., 2001). Another study examined self-reported health status and quality of life in different racial and ethnic groups using data from the national Medical Expenditure Panel Survey (MEPS) (Fu & Kattan, 2006). Although almost half (49%) of individuals rated themselves as in full health, with the majority (59-96%) lacking health problems for any of five measured attributes, differences in perceived health status were seen based on race and ethnicity. Blacks and Hispanics were less likely to answer that they had no problems compared to Whites; Asians were more likely to answer that they had full health. In

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**Table 3: Instruments & Measures of Individual Perceived Health Status**

NAME	EuroQoL: European Quality of Life <sup>1</sup>	HAQ (Health Assessment Questionnaire), MHAQ (modified HAQ) & MDHAQ (multidimensional HAQ) <sup>2</sup>	NHP: The Nottingham Health Profile <sup>3</sup>	SF-36: The Medical Outcomes Study 36-item Short Form health survey) <sup>4</sup>	SIP: Sickness Impact Profile <sup>5</sup> (also known as AIMS)
Purpose/ Function	Measure quality of life & to create an index value for each health state.	Evaluate & measure functional health status	Measure & assess subjective health status	Measure of perceived general health status	Behaviorally based index of health for impact of sickness & dysfunction
Components & foci	Health status, attitudes, perceived satisfaction, values & general well-being. EQ-5D (part of MEPS) preference-based HRQoL measure: health care quality & health status measures (5 attributes: mobility, self care, usual activities, pain/ discomfort, anxiety/ depression)	Full HAQ: 5 Dimensions (disability; discomfort; drug toxicity; dollars/cost; death) “The HAQ” (short form): Disability Index (DI), Pain scale & Global health scale. <i>Functional health status categories.</i> Eight sections: dressing, arising, eating, walking, hygiene, reach, grip & activities	Six health profile sections: Physical mobility (8); Pain (8); Sleep (5); Emotional reactions (9); Social isolation (5); & Energy level (3)	Physical Health: Physical Functioning (PF; 10), Role-Physical (RP; 4), Bodily Pain (BP; 2), General Health (GH; 5) & Mental Health: Vitality (VT; 4), Social Functioning (SF; 2), Role-Emotional (RE; 3) & Mental Health (MH; 5)	Statements about health dysfunction in 12 areas of activity
Instrument: # of Questions, Format & Scoring	Each attribute is reflected by a question with 3 possible responses (problem: no, some, or severe). Combinations result in 243 possible health states, each with assigned scores. State of health on scale of 0 (dead) to 1 (perfect health).	Full: 20 questions (2-3 questions/section). Scoring 0-3 (without difficulty to unable to do) for each item. Section scoring based on worst score. Global health & Pain are measured by a visual analog scale (0-100: very well to very poor)	38 Yes/No questions (representing departures from normal functioning). Scored 0-100 (no problems to problems with all listed: for each section).	36 questions. Likert-type (5-point): strongly agree to strongly disagree. 8 scales scored 0-100. Low scores mean has problem; high scores mean no problems or normal functioning. Two Component Summary measures: standardized mean = 50, SD = 10	136 items: categories (5 independent; 3 physical; 4 psychological).

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Strengths & Unique features	Used worldwide. Over-sampled for ethnic and racial minorities to enhance precision of these estimates. Provide nationally representative estimates of healthcare utilization, expenditures, sources of payment, and insurance coverage for US civilian non-institutionalized population.	Disability (DI) & pain scales used for long term outcomes.	Disability focus. Items represent actual words of lay people in describing impact of illness. Ease of use.	Can use eight subscales, or the two major components: Physical health (PHC) and Mental health (MHC)	Overall, category, and dimension scores may be calculated. Main foci are dysfunction & sickness.
Validity & Reliability	Well-validated. Best for major morbidity conditions.	Hundreds of publications attesting to validity & reliability	Evidence supports convergent & construct validity (can discriminate between health status groups).	Tested extensively; found to be an acceptable measurement instrument in various populations.	High test-retest reliability & internal consistency. Evidence supports convergent & discriminant validity; also clinical validity.
Comments/ Issues/Limits	Single index measure. HRQoL measure of preference for a state of health. Designed & recommended for use in cost-utility analyses in many national guidelines.	Used worldwide. HAQ focus is functional status (disability), e.g., arthritis. Other derivatives: Modified HAQ & Clinical HAQ	Focus on health functioning, e.g., arthritis. Initially used in study of elderly.	Profile measure. Items written by experts or researchers, then tested in patients. Sensitivity to specific conditions varies. Studies using the 2 component scores have inconsistent results.	Applicable for outcomes evaluation, and across all types & extents of illness, & demographics. Does not assess positive function.

KEY of Instruments: <sup>1</sup>EuroQoL = European Quality of Life (Brazier, Jones, & Kind, 1993; EuroQoL Group); <sup>2</sup>HAQ = Stanford Health Assessment Questionnaire (and its two of its many derivatives; others includes: CLINHAQ & Modified CLINHAQ = Clinical Health Assessment Questionnaire) (Bruce & Fries, 2003a, 2003b; Fries, Spitz, Kraines, & Holman, 1980; Pincus, Swearingen, & Wolfe, 1999); <sup>3</sup>NHP = Nottingham Health Profile (Hunt et al., 1980); <sup>4</sup>SF-36 = Medical Outcomes Study 36-item Short Form health survey (McHorney, Ware John E, Lu, & Sherbourne, 1994; Ware, 1976, 2009; Ware, Kosinski, & Gandek, 2003; Ware & Sherbourne, 1992); <sup>5</sup>SIP = Sickness Impact Profile (Bergner, Bobbitt, Carter, & Gilson, 1981). Other sources: (Fu & Kattan, 2006; Houssein, McKenna, & Scott, 1997; Jenkinson, Coulter, & Wright, 1993)



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contrast to the previous study (Okosun et al., 2001), race and ethnicity differences disappeared when medical conditions and function/activity limitations were considered (Fu & Kattan, 2006).

### Healthy Individuals

There were very few studies that examined healthy individuals and their perceived health status (see Table 1). Two complementary reviews of the literature in the late 1990's with a focus of perceived health status and mortality identified only two (Benyamini & Idler, 1999; Idler & Benyamini, 1997). One study was noteworthy in demonstrating a strong self-rated health effect (adjusted OR of 19.6) for poor versus excellent health, thus supporting the reasonable consideration that perceived health status captures some unique aspect beyond medical history. These two reviews found that only three studies included some consideration of ancestry, family history or longevity despite the recognized likely impact on perceived health status (Benyamini & Idler, 1999; Idler & Benyamini, 1997). The current literature review identified only a few additional studies of perceived health status that included healthy populations (see Tables 1 & 2), spanning the following range of research populations and purposes: (1) the elderly – one study to predict health deterioration, another examining ability to discriminate between elderly health status groups (Griep et al., 1997; Hunt et al., 1980); (2) minority groups to evaluate use and suitability of different measurement instruments related to race and ethnicity (Fu & Kattan, 2006); (3) working age persons – one study to compare the SF-36 instrument and a modified shorter version for use in this population, the other study to examine findings for prediction of healthcare services use and mortality (Chern, Wan, & Pyles, 2000; Miilunpalo et al., 1997); and, (4) sick and healthy college students to explore differences in perceived health status and influences on future expectations for health (Kulik & Mahler, 1987). No studies were identified with the primary purpose of studying healthy persons to improve understanding of their

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perceived health status, health values and goals, motivating factors, and/or factors influencing interest in risk reduction, prevention or health promotion.

### Summary

Existing research and clinical studies demonstrate consistent findings supportive of the unique, statistically significant association between individual perceived health status and outcomes related to survival, mortality, the elderly and general health, even after accounting for covariates that affect health (Idler & Benyamini, 1997; Okosun et al., 2001). These studies also support the use of perceived health status in different disease populations and public health contexts. The framework of identified studies is consistent with a biomedical model and operationalization of perceived health status focusing on symptoms, illness or increased risk as defined by the presence of objective scientific evidence to make a diagnosis, justify intervention, and determine treatment and policy (Borrett, 2013). Even the few studies identified as targeting prevention addressed either specific diseases, common public health problems, populations considered at higher risk, or addressed allocation of resources using economic parameters.

The perceived health status research discussed above also demonstrates the potential influence of ethnic and racial differences on self-rated (perceived) health, yet other researchers note the limited extent of culturally derived individual health values studies (Grandes et al., 2008). Other factors may contribute to findings of racial and ethnic differences associated with perceived health status, such as varying cultural views of personal health, differences in study populations, the nature of survey instruments, and/or the approach to data analysis and its interpretation (e.g., combining low frequency minority groups). Thus, existing studies depict a significant gap and the need for research and knowledge of perceived health status in healthy,

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minority and different racial/ethnic populations, as well as from the perspective of prevention, including motivations such as risk reduction and intended health behaviors.

### Measures & Instruments

Perceived health status represents a concept and construct that is subjective in nature, thus posing not only a significant challenge to define it, but also to measure it. Essentially two distinct approaches exist to measure and evaluate perceived health status, in the form of an instrument or as a single question. This raises questions about which approach is better, what measurements and instruments currently exist, and criteria for selecting one instrument over another. Logical and analytic concerns include whether a questionnaire will be understood, if its length compromises completion or if brevity compromises the validity, and whether a single item can measure perceived health status. Validity, reliability and utility issues apply to either approach. These considerations are addressed in this section.

### Approaches To Measurement

**Single Item Measure versus Instrument.** Perceived health status was operationalized and measured in one of two ways: (1) as a single question for individual rating of health status, e.g., in general, compared to others, or using a 5-point Likert-type scale (Idler & Benyamini, 1997; Kivinen et al., 1998; Shmueli, 1999), or (2) by an instrument(s) having a number of items/questions, either designed for a specific study (DeCuir, 2007; Kumpusalo et al., 1992; Salaffi, Stancati, & Carotti, 2002) or an existing instrument (Benyamini & Idler, 1999; Bergner, Bobbitt, Carter, & Gilson, 1981; Brazier, Jones, & Kind, 1993; Bruce & Fries, 2003b; Fries, Spitz, Kraines, & Holman, 1980; Hunt et al., 1980; Pincus, Swearingen, & Wolfe, 1999; Ware, 1976, 2009; Ware, Kosinski, & Gandek, 2003) (see Tables 2 & 3).

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In the late 1990's, two complementary reviews of the literature targeting self-rated health status in relation to mortality identified a total of 46 national and international studies (Benyamini & Idler, 1999; Idler & Benyamini, 1997). The authors examined self-rated health from three contexts: (1) a single self-rated item; (2) other health status variables; and, (3) other covariates. These latter variables and covariates collected and examined by the various researchers included: self-reported chronic or medical conditions, physical functioning or disability, mental/cognitive factors, pain, symptoms, and medications, as well as socio-demographics and practitioner derived objective health measures (e.g., height, weight, BMI, or targeted disease measures). Use of a single question to capture global (general) health status, regardless of how it was phrased, demonstrated consistent predictive effects (Benyamini & Idler, 1999; Idler & Benyamini, 1997). On the other hand, there are limitations to using a single question as the basis for representing perceived health status. There is wide variation on an individual basis that poses difficulty for interpretation (Ware & Sherbourne, 1992). A single item also precludes the ability to examine and measure the potential influence of other existing subjective factors on perceived health and health behavior, e.g., health values, locus of control, perceived health competence and interest in prevention (Gebhardt, van der Doef, & Paul, 2001).

### **Instruments**

The review of literature identified that studies using an instrument for perceived health status assessment typically used one of five, although a few developed study-specific questionnaires (Di Giulio, 2014; Griep et al., 1997) (see Table 1). The commonly used and validated instruments included: the EuroQol (European Quality of Life; EQ), the Health Assessment Questionnaire (HAQ; or one of its versions), the Nottingham Health Profile (NHP), the Medical Outcomes Study 36-item short form health survey (SF-36), and the Sickness Impact



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Profile (SIP) (Bergner et al., 1981; Brazier et al., 1993; Bruce & Fries, 2003b; Fries et al., 1980; Hunt et al., 1980; Pincus et al., 1999; Ware, 1976, 2009; Ware et al., 2003) (see Table 3). These five instruments were developed as measures of generic health (stated as not disease-specific) in conjunction with study purposes, incorporating content to address the health concepts and issues of concern. These factors represent both the strengths and the limitations related to approaches to assess general and perceived health status. Several studies involved the comparison of different methods or instruments for health status assessment, as for targeting a specific disease, condition or population (Bardsley, Astell, McCallum, & Home, 1993; Pincus et al., 1999).

### **Medical Outcomes Study 36-item short form health survey (SF-36)** (see Table 3)

The SF-36 was the most frequently used instrument (12/33; 36.4%) identified in the current literature review evaluating perceived health status (Bentzen & Christiansen, 1995; Chern et al., 2000; Footit & Anderson, 2012; Funk et al., 1997; Gartsman et al., 1998; Hunter et al., 1995; Larson et al., 1996; Lindsay et al., 2001; Mattera et al., 2000; Salaffi et al., 2002; Schiffman, Jacobsen, & Whitcup, 2001; Stucki, Katz, Johannesson, & Liang, 1996; Ware, 1976, 2009). The SF-36 is composed of 36 items (in eight scales) selected from previously established and validated instruments including the Health Perceptions Questionnaire (HPQ) (Ware, 1976), health status surveys associated with the Health Insurance Experiment (HIE) and the full length Medical Outcomes Study (MOS) instrument. It conceptualizes and measures health as comprised of physical and mental dimensions, but also integrates social functioning (Stewart, Hays, & Ware John E, 1992; Ware & Sherbourne, 1992).

As background, the HPQ evaluation process is relevant to understanding the selection process employed for SF-36 items. The HPQ was developed as a general health rating instrument with 36 statements of opinions spanning eight physical, mental and functional health

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perception scales. It was evaluated in approximately 2,000 adults from the general population through five field tests, with return rates of 37% for mailed questionnaires and from 67% to 95% for personally delivered questionnaires versus an interviewer approach (Ware, 1976). Factor analysis was performed with retention requiring both a high loading (>40%) and highest correlation (discriminant validity) with a specific scale. Thirty-two items met this criteria; discriminant validity was almost 99%. Six of the eight scales were balanced with positively and negatively worded items; scale scores approached that of a normal distribution. Although test-retest reliability coefficients for scale scores ranged from 0.41 to 0.86 across the different field tests, with the lowest associated with a disadvantaged population and the highest correlation and stability associated with the general health items/scale, overall ability for use was supported. The eight scales could be grouped, based on the primary source of reliable variance in their scores, into three types of perceptions about health – past/present, future, and sick role propensity, with consistent variance in scores across all field tests. Ware indicated this finding as noteworthy for support of construct validity, its potential contribution to understanding the meaning of scores, and to possible reduction in the number of scales or items, yet also recognizing the complexity of measuring and interpreting scores in that items were not scale or construct specific. Another foundational basis for the SF-36 was the extensive development and refinement of health status surveys associated with the Health Insurance Experiment (HIE) that ultimately raised questions about the potential use, and effective construction, of these types of survey instruments for sick and older populations (Ware John E, 1992). The Medical Outcomes Study (MOS) of adults (from three major U.S. cities) with chronic and psychiatric conditions was ultimately used to address these questions through development of standardized instruments of generic health using scales and items from the earlier studies.

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The SF-36 was based on 3,445 MOS study patients ranging in age from 18 to 98 years of age who completed a 245-item questionnaire; they were also examined as twenty-four subgroups differing in socio-demographic backgrounds, diagnosis and disease severity (McHorney, Ware John E, Lu, & Sherbourne, 1994). The completion rate across the eight scales ranged from 75% to 98% (median of 91.5), with the lower rates among disadvantaged persons; however, scale scores were computed successfully for 89.9 – 100%. Across the 24 subgroups, the full range of possible health states was observed, except in ten. Internal consistency of items within scales exceeded the minimum standard of 0.40 for all items (except one); it was also exceeded across the scales 97% of the time. Item discriminant validity was supported since correlation between an item and its hypothesized scale was greater than its correlation with other scales 99.5% of the time. The scaling success of patients across different groups ranged from 35-100% (mean of 92%), varying by subgroup and concept, and with the lowest percent occurring with small groups, and groups of persons with lower education, diagnosis of CHF, diagnosis of MI, and psychiatric or complicated medical conditions. Each scale surpassed minimum reliability standards of 0.5- 0.7 for groups; the individual standard of 0.9 was only met for the Physical functioning (PF) & Mental Health (MH) scales. Floor effects were most notable for the role disability and social functioning (SF) scales based on the diagnosis (in 41 – 61%), and ceiling effects were notable for persons who were younger, free of disabling conditions and not poverty status (20 – 24%), and for persons with milder medical conditions (60 - 65%).

A study examined the SF-36 scales and items, in comparison to the full length MOS instrument and the earlier SF-20 shorter version, noting the SF-36 as distinguished by: (1) the physical functioning scale retained all items from the full version, thus tripling the number of levels over the SF-20; (2) the role functioning (physical) scale contains a subset of the full MOS

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and modified items, enhancing representation of role limitations, and physical and mental health factors; (3) the bodily pain scale added one item beyond the SF-20 relating to pain interference with normal activity; (4) the social functioning scale retained an improved version of the SF-20 question, plus adding one about interference due to physical or emotional problems; (5) the mental health scale retained the SF-20 five items (the MHI-5; Mental Health Inventory) whose summary score was highly correlated (0.95) with the full length 38 item MHI; (6) the vitality scale added four new items with proven discriminant validity (from the MHI of the HIE, and based on the national NHANES survey) to improve ability to discern subjective well-being (Sherbourne, 1992); and, (7) the general health perceptions scale includes the single 5-point Likert-type item for perceived general health, four modified SF-20 items that correlate ( $r = 0.96$ ) with the 22-item General Health Rating Index (GHRI) derived from the HPQ and reduces redundancy (Stewart & Ware, 1992; Ware & Sherbourne, 1992), and a sixth item about change in general health in the past year. In studies over the years, most items demonstrate meaningful correlations ( $> 0.40$ ) with their hypothesized scales (McHorney et al., 1994; Ware, 2009).

Use of the SF-36 measure in over 4,000 studies, including internationally, has supported its validity and reliability (Ware, 2009). The SF-36's eight subscales can be captured by two summary measures: the physical health (PHC) and mental health (MHC) components; however, studies have not yet validated these two components. On the other hand, the sub-scale measures have generally been shown to exceed reliability standards and correspond to expected and relevant subscale components, typically achieving 80-90% of their empirical validity in studies involving physical and mental health criteria (Ware, 2009). The SF-36 appears to be more sensitive than other existing instruments for measuring perceived health status in persons with less severe health conditions (Houssein et al., 1997). This assessment is also supported by a



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study of healthy older adults that compared the SF-36 to a commonly used and tested wellness instrument (the Perceived Wellness Survey) with results identifying a significant correlation between perceived wellness and general health (Footitt & Anderson, 2012).

A study of over 2,000 urban Jews in the general population compared two measures of perceived health status: the SF-36 (the Medical Outcomes Study 36-item short form health survey) as a measure of health functioning, and a single 5-point subjective rating question of one's health-related quality of life (Shmueli, 1999). Health values (operationalized as numeric scores, not as a qualitative concept) were assigned to the subjective health status rating (i.e., "good" versus "poor" general health equaled a health "value" of 76-81, or 45-61, respectively). Standard deviations (SD) were quite large, especially with reported poorer health. The eight SF-36 scales (see Table 3) demonstrated a linear relationship to health value (in this context), whereas the PCS (physical component scale) and the MCS (mental component scale) did not. Of practical importance, the researchers noted that a simple average of the eight sub-scales best correlated (0.67) with Health Quality of Life (HQL), and accounted for greater than 50% of the variation in HQL. The scales that measured general health (the scale most highly correlated with HQL), vitality and physical functioning were the main determinants of health value, with a mean HQL rating of 71.3 and SD of 19.2. These results affirmed the utility of perceived health status measures, yet pointed out issues with the different bases of scoring and how that may impact data analysis and interpretation. The authors also noted that "clinical validity of health-status measures leads to different structures than those obtained from validation with respect to subjective evaluation of quality of life" (Shmueli, 1999) (p124).

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### EuroQol

This instrument, originally representing six health dimensions with the potential to identify 216 health states and composed of items selected by the EQ Group from previously established generic measures of health, was tested in a population of 1582 patients as a non-disease specific instrument for two purposes: (1) describing health quality of life, and (2) to generate a single index value for each health state (Brazier et al., 1993) (see Tables 2 & 3). As a method to evaluate the EuroQol, it was compared to the United Kingdom's version (language Anglicized) of the SF-36, a tested instrument in widespread use (Ware & Sherbourne, 1992). Dimensions/Categories, as well as the general nature of items, of the EQ and UK SF-36 were determined to be comparable (see Table 3), yet the nature of items and answer choices (i.e., limited to a few choices and/or a visual analog scale for the EQ, versus a Likert scale for the UK SF-36) and scoring (i.e., as a single index value versus a range of scores, for the EQ versus UK SF-36, respectively) differed. The EQ demonstrated construct validity through testing of hypotheses related to age ( $p < .05$  or  $< .01$ , across all dimensions except anxiety and depression, as proposed), sex (significant only for anxiety and depression), and use of services (significant for persons with health problems). Convergent and discriminatory validity was assessed by comparison of EQ and UK SF-36 scores. The EQ had significantly lower means than the UK SF-36 on each dimension, although the dimensions displayed some correlation and the correlation coefficient for the total scores of the EQ and UK SF-36 was significant ( $p < .01$ ). Related to the EQ's 216 potential health states, ten (10) accounted for 95% of the observations and 171 (of 216; 79%) health states never occurred. As Brazier noted, discrimination between different levels of health found the EQ scores (versus the UK SF-36) to have: (1) a more skewed distribution (in part due to limited scoring categories, rather than a range of possible scores); (2)

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a greater ceiling effect (limited ability to determine differences in persons who were generally healthy), as occurred in 95% and in 81% of persons in the EQ functional and emotional dimensions, respectively (versus 37-72% and 2% in the SF-36, respectively); and, (3) similar “floor” effects, relating to the less ill (Brazier et al., 1993). Based on identified issues, modifications were made to the EQ, reducing the ceiling effect for the total score to 54%. A unique aspect of the EuroQol is its design as an index measure, in contrast to a health profile as with the UK SF-36, thus supporting use of EuroQoL (EQ-5D) as a preference-based quality of life measure with ability to distinguish between populations, e.g., ethnic, racial differences (Fu & Kattan, 2006), and its use for value and cost-utility analyses (Houssein et al., 1997). The EuroQol has been criticized as difficult to use, crude and less responsive to change in health status than the SF-36, yet the SF-36 was noted to lack adequate reliability and responsiveness for clinical studies use, e.g., for specific disease such as rheumatoid arthritis (Brazier et al., 1993; Houssein et al., 1997).

### Sickness Impact Profile (SIP)

The SIP was designed to examine perceived health status from the perspective of illness and dysfunction, with the intent to measure intervention-based changes over time or in different groups for clinical, programming and policy purposes (Bergner et al., 1981) (see Tables 2 & 3). The instrument was tested in a succession of large field trials spanning three years including over 3,000 persons (inpatients, outpatients, home patients, general public) from a combination of group practice enrollees, a family medicine clinic and a large sample from another study. The SIP was administered in one of three ways: by interviewer, by interviewer delivered self-administered, and by mailed self-administered. Validity, assessed by the relationship between SIP scores and three types of measures, demonstrated ability to discriminate among subgroups

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and correlations between criterion measures. Clinical validity, assessed by correlations between SIP scores and clinical measures, ranged between 0.41 to  $-0.84$ . Test-retest reliability ( $r = 0.92$ ) and internal consistency ( $r = 0.94$ ) were high. Limitations of SIP are that it does not assess levels of positive functioning, it does better for higher levels of illness, and while it directly assesses illness, this is not the case for dysfunction assessment. The study also found that either method of self-administration produced higher mean scores than interviewer administered, and that the interviewer administered showed higher correlations with the sickness and dysfunction measures. These data support greater validity with self-administered measures, and the likely best approach as a combination with an available professional resource. Compared to the NHP, SIP was more a measure of physical functioning (Essink-Bot, Krabbe, van Agt, & Bonsel, 1996).

### Health Assessment Questionnaire (HAQ)

The HAQ is a clinically based instrument, originally published in 1980 and developed using a composite of items/components from other existing instruments for use in multiple illnesses yet primarily for rheumatoid arthritis patients, to measure health-related quality of life vis-à-vis health functioning and activities of daily living (Bruce & Fries, 2003b; Fries et al., 1980; Pincus et al., 1999) (see Tables 2 & 3). The HAQ presently in common use is a short version (two-page) composed of the original unchanged Disability Index (DI), Pain scale and Global Health scale, while other HAQ components have been added, edited and/or evolved, as needed. Comparison of the HAQ global health scale with other clinical measures such as the EuroQol “Torrance thermometer” for quality of life demonstrated a significantly high correlation ( $p < .001$ ) consistent with convergent validity of similar constructs (Bruce & Fries, 2003b). Reliability has been demonstrated through assessments including item-total correlations and factor analyses of the HAQ in its original form, its modifications (i.e., the modified,



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multidimensional and clinical versions; see Table 3), and adaptations in over 60 different languages or dialects (Bruce & Fries, 2003a, 2003b; Pincus et al., 1999). Reproducibility has been demonstrated by test-retest correlations (ranging from 0.87 to 0.99) and by comparisons between self and interviewer administration (correlations ranging 0.85 to 0.95) (Bruce & Fries, 2003a). The HAQ, promulgated as a generic measure of health despite its historical emphasis related to rheumatology, has proven to be amenable to adaptations and international use as well as uniquely including dollar/cost, drug and death related components among its health items. Limitations of the HAQ relate to measures of sensory or psychiatric disability, or patient satisfaction, as well as ceiling and floor effects associated with constraints of the concept of disability (i.e., if someone measures at an extreme initially, a change will be difficult to capture, or will be missed). The HAQ format is recognized for its ease of use and application to various diseases, health problems and disabilities, yet as with the SF-36 and the AIMS (SIP), it has been criticized for limitations related to the patient's views on outcomes (Houssein et al., 1997).

## Nottingham Health Profile (NHP)

The NHP was developed as an instrument to capture the subjective view of health status, with component questions related to health functioning and departures from normal (Hunt et al., 1980) (see Tables 2 & 3). A study of four groups of elderly persons, ranging from healthy (without a medical diagnosis) to having diagnosed medical problems, supported convergent validity in that the NHP could distinguish between these groups and that the number of affirming statements was significantly different among groups (likelihood of occurring by chance was  $< 0.001$ ) (Hunt et al., 1980). The study also found that there was no association between the number of affirmative statements and the perceived severity of that item. It was not possible to determine whether perceived health status related to the presence of symptoms or an actual

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diagnosis. The author concluded that congruence between consumers and provider perspectives may be more similar than expected; yet further study using appropriate instruments is indicated. In another study, the NHP was found to be less sensitive than the SF-36 in less severe disease, but not less sensitive related to severe conditions or with the elderly (Houssein et al., 1997). In comparison to the SIP, the NHP showed slightly higher feasibility, internal consistency and was more a mental (perceived) health measure than of physical functioning (Essink-Bot et al., 1996) (see Tables 2 & 3). Other noted strengths of the NHP are its representation of the impact of illness in actual words of lay people and its ease of completion, thus facilitating its use and accuracy in interpretation by users (Houssein et al., 1997).

## Other Instrument-related Issues

It is relevant to mention that a study examining standardized nomenclature to represent health concepts (e.g., health status) in two instruments, the Clinical Health Assessment Questionnaire (CLINHAQ) and the Modified Health Assessment Questionnaire (MHAQ), identified significant differences between their overall representational ability based on computerized medical language systems (i.e., the Unified Medical Language System (UMLS) versus SNOMED) (Ruggieri et al., 2000). These authors concluded that health-related concepts should be grounded in a universally agreed upon conceptual model, in particular related to the domains of functional health status and health status assessment, since it forms the basis for evaluating outcomes, impact of disease and allocation of resources. Health concepts and vocabulary also contribute to the structure and focus of research, instruments and interpretation (Bruce & Fries, 2003a), as well as perspectives on health values, health promotion and prevention initiatives.

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### Summary and Assessment

Two approaches are typically used to measure and examine perceived health status, either a single question or an instrument compatible with the study purpose and goals. Despite the potential concern of limitations in interpretation of individual perceived health status based on the answer to a single question, research supports its significant value. Alternatively, use of various instruments developed to measure health status (see Table 3) have repeatedly demonstrated validity and reliability that is important to meaningful clinical and research applications (Miilunpalo et al., 1997).

The existing research reviewed spanned use of various common instruments attesting to their applicability as measures of generic health, their tested and demonstrated validity and reliability, and their relative ease of use and time to complete. This raises questions as to how a researcher or clinician determines which approach or instrument to use. The SF-36 is appropriate for use as a measure of general health, for evaluation in pre- and post intervention studies, and for examining health-related constructs, health and illness behavior (Ware, 1976). The distinguishing characteristics of the SF-36 are its relatively better ability (compared to the other instruments) to measure healthy states (although noted to have ceiling effects), its extended use and testing, and its historical record as the standard for comparison and evaluation of other instruments. However, the SF-36 two summary component scores have yet unproven validity although its eight subscale scores are validated. The EuroQol is relevant for use in morbid populations. It is unique in its purposeful design as a single measure of health or quality of life, making it especially useful in the context of decision-making for healthcare interventions, cost-effectiveness and/or policy determinations. The HAQ, although historically associated with rheumatologic disorders, is a measure of generic health, having many components that correlate

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with the SF-36 as well as being used extensively, having several versions and adapted in numerous languages. The distinguishing characteristics of the HAQ involve its disability focus and the inclusion of dollar/cost and drug/toxicity components. The strengths of the Sickness Impact Profile (SIP) relate to measuring and assessing high-level illness, disability and physical functioning. The NHP strengths pertain to its user-derived lay terminology, ease of completion, a mental (emotional) health mindfulness, and relevance to morbid and elderly populations. This information can be useful to guide clinicians and researchers in selection, use or possible modification of a health status measure – be that a single question or existing instrument.

Despite the strengths, research testing, and demonstrated validity and reliability of these approaches and instruments, gaps and limitations exist to varying extents related to inherent design, conceptual basis and/or applied orientations including: (1) a typically illness, disease, mortality and/or dysfunction focus, typically consistent with a biomedical model, regardless if stated as such; (2) the nature of concept operationalization, approach to measurement and survey structure that may limit individual ability to answer in an informative or meaningful way (Houssein et al., 1997); (3) the use of surrogate measures for perceived health status and the surrogate's complexity (e.g., health values) (Fu & Kattan, 2006; Hunter et al., 1995; Shmueli, 1999); (4) healthcare system values determination based on outcomes (defined by the researcher or clinician) or economic criteria; (5) approaches to data analysis that may result in bias, such as differences in scoring of answers or combining/eliminating groups or persons for the sake of statistical significance (Miilunpalo et al., 1997); and, (6) the study or instrument absence of recognized potentially influencing factors, such as family history (Idler & Benyamini, 1997). Thus, although assessment of perceived health status and specific approach or instrument selection must be capable of capturing data consistent with the study purpose or intended clinical



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use (Lichtenstein & Thomas 1987), researchers and clinicians cannot lose sight of capturing important qualitative, potentially influencing, subjective elements that would otherwise be missed. There is a distinct absence of questions/items with a healthy orientation.

### Discussion

Perceived health status is a complex concept/construct comprised of physical, psychosocial, environmental and personal factors. Research supports that an individual's perceived health status contributes a unique dimension to predicting mortality and morbidity, and to evaluating the outcomes of healthcare interventions, yet gaps exist in understanding the subjective personal factors integral to this individual perception of health and the associated health values, motivations and basis for decisions and actions. In a review of literature on perceived health status in the late 1990's, Idler identified needs and suggested opportunities for research including: (1) study outcomes other than mortality, e.g., morbidity; (2) study special populations, other than the aged; (3) use a qualitative approach, e.g., what respondents capture in their own words; (4) study the cognitive/cultural processes associated with individual judgments; and, (5) pursue new approaches to measurement instruments (Idler & Benyamini, 1997). These remain as current and even more relevant needs now.

Research targeting assessment of perceived health status among its main purposes has focused primarily from the perspective of illness, with little application or exploration of perceived health status in healthy persons. The lack of perceived health status studies in healthy persons may be related to limited funding for prevention research and/or study determined priorities (e.g., the outcome of alternate treatment approaches for a disease), the focus of reporting, alternate views of perceived health or related health concepts (e.g., perceived risk, or health values as economic valuation), as well as limitations due to database designated search

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terms and author-designated keywords. For example, the explosion in genomics has led to questions and research about motivations of healthy Direct-to-Consumer (DTC) genome testing users, with some studies including a question or component about perceived health status that could potentially contribute to understanding this concept from a healthy person perspective (Bloss et al., 2010; Green & Roberts, 2012; Kaufman, Bollinger, Dvoskin, & Scott, 2012); however, these studies were not identified by any of the current literature searches.

Clinical and research use of perceived health status to date, and instruments or approaches in use, are based on a biomedical, disease-oriented and symptomatology model (Lindsay et al., 2001). Although the biomedical model has definitive clinical and research strengths from the perspective of disease, it is reductionist in the context of complex healthcare issues, prevention and subjective individual factors impacting health (Sturmberg, 2007). This model is not conducive to exploring a qualitative, subjective or interaction-based individual perspective of health status or health values in healthy or perceived vulnerable persons (Kulik & Mahler, 1987). In the biomedical model, prevention efforts generally target health risks and risk likelihood, yet in binary, disease/non-disease situations (Krugman, 2013; C. Wang et al., 2009). These approaches support a disease risk-focused discourse lacking appreciation for broader health values (e.g., those of the individual), recognition of individual freedoms (choices) and the potential influence of other contextual variables (e.g., age, sex, education, family history). Viewing health from the perspective of illness and threats (risks) leads to a tendency to obscure or minimize the potential contribution of research involving more complex common and multifactorial disorders, and fosters an all-or-nothing stance about the value of health and prevention, such as with pre-symptomatic risk identification that is becoming a reality through genomic testing (Boenink & van der Burg, 2010). It also thwarts examining healthy individuals'

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perceived health status in the context of prevention and health promotion efforts. This is necessary to understand personal priorities, needs, motivations and factors relevant to personal utility and health behaviors, otherwise compromising the potential for integration of this knowledge into Nursing strategies for research, routine care, health promotion, risk identification and prevention, including access to new technologies and healthcare resources.

Potential strategies for addressing this gap in knowledge about the subjective factors integral to an individual's perceived health status, and assessing its impact on individual health, health values, behaviors and outcomes, include: (1) exploration of healthcare models that integrate the concept of perceived health status, prioritize individual health values and goals, and recognize health promotion and prevention at the individual level; (2) pursuit of nursing research to assess the impact of integrating perceived health status measures into research and routine clinical practice including for healthy persons; and, (3) exploring various instruments for measuring perceived health status, including existing, modified or newly developed.

### **A Model for Consideration**

The Health Capability conceptual framework is a potential model for nursing research and clinical use. Health Capability is an interdisciplinary systems model (Ruger, 2010) (see Figure 1) based on Capability Theory (Sen, 1985) with Aristotelian origins and respect for individual autonomy, social justice, public health and economics. Health Capability has two essential principles: (1) individual health and well-being, and (2) health agency, embracing respect for individual health values and goals. This individual context and prioritization forms the basis for extension from the individual level to population health, as opposed to the converse of biomedical and values-base medical (VBM) models. This context is relevant to study findings that individual perceptions of health can be very different from clinician expectations based on

test results and objective data (Mattera et al., 2000). *Health* is captured as the reality that “people seek good health” (Ruger, 2010) (p.242). Integral to this premise exists the reality of challenges to good health (i.e., medical problems, health risks) and the objective to maintain or promote health. This conceptual framework identifies subsystems and proposes the Health Capability profile denoting internal and external factors that interrelate with health values and goals, and includes the constructs of health status and health functioning (Ruger, 2010) (see Figure 1). Self-rated health, as discussed in this paper, also reflects the presence or absence of external and internal resources (factors) that can attenuate decline in health (Idler & Benyamini, 1997). The Health Capability model suggests consideration of the SF-36 instrument for measurement of health status (vis-à-vis health functioning). Thus, this model supports perceived health status as an essential concept, yet its assessment and interpretation may be limited by the same constraints identified in the literature review if measurement is based solely on traditionally used measures. Integrating the perspective of a Nursing model compatible with the concept of perceived health status may offer a meaningful research opportunity.

### **Instrument and Measurement Strategies**

Two strategies, representing opportunities for Nursing study and application in clinical practice consistent with Nursing’s commitment to holistic healthcare, health promotion, prevention and qualitative research, involve minor modifications to existing methodologies. Specifically, given the established validity and value of a single Likert-type question about one’s perceived health status, yet the recognized absence or limitations of assessing perceived health status in healthy persons, a single open text question could be added to the initial intake assessment by simple inquiry about the person’s current health need, desired goal and how they “see” that being addressed. This subjective and open information could serve as the entree for



Figure 4.1: Conceptual Model of Health Capability.

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Figure 1: Conceptual Model of Health Capability

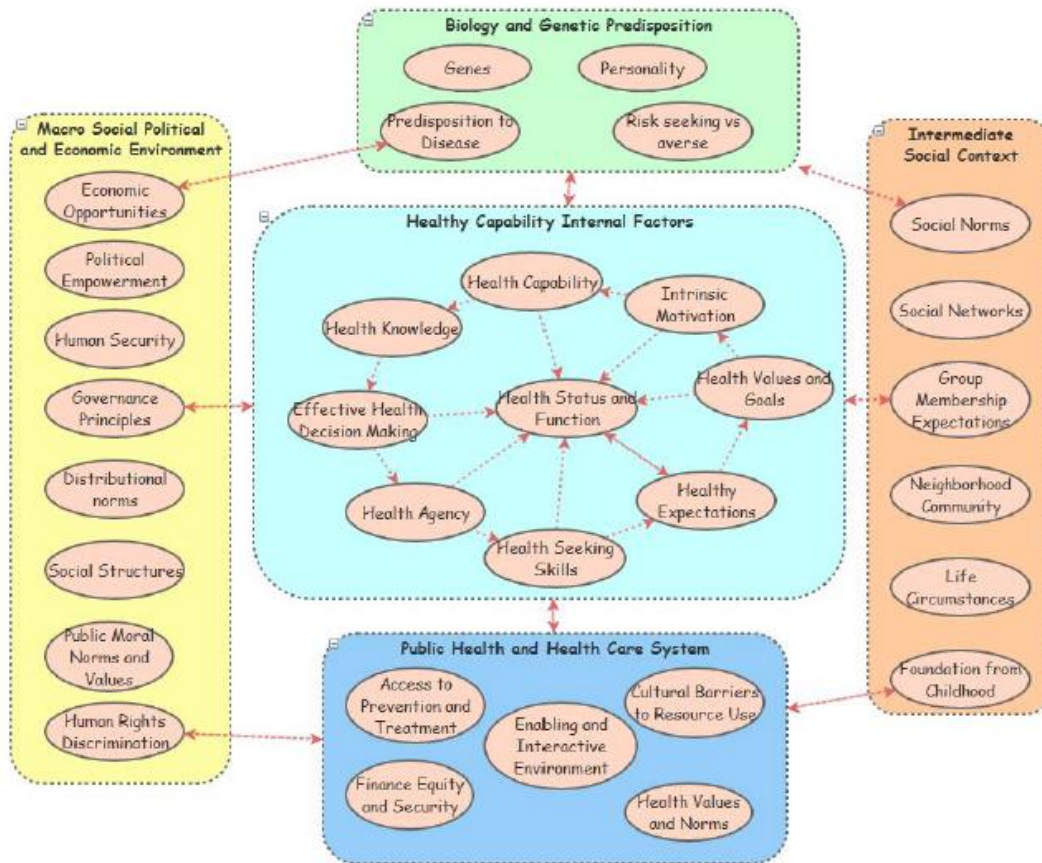


Fig. 1 - Conceptual Model of Health Capability -2 [Source]

<http://insightmaker.com/insight/779>

From Jennifer Prah Ruger (2010) Health Capability Conceptualization and Operationalization

Am J Public Health 100 p41-49 available from SSRN <http://ssrn.com/abstract=15080>

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nursing discussion with patients/individuals, prioritizing assessment and jointly determined intervention. It also could be explored in nursing research, for example, by comparing the content of text answers with the single perceived health status answer. In this way, otherwise unknown factors may be identified.

Second, an instrument is needed that can meaningfully assess healthy persons, from the basis of an appropriate model, whether that involve modification of an existing instrument (e.g., the SF-36, or other in Table 3) or development of a new one that also integrates relevant previously tested and validated questions. This instrument must include items that can detect health differences in relatively healthy persons, thus eliminating the ceiling effect with current instruments where health improvement poses challenges to recognition, or where results potentially bias interpretation in a skewed direction (e.g., that the person is healthy and does not need healthcare services). Tackling this type of clinical and/or research initiative is consistent with Nursing philosophies that include health promotion and prevention. Instrument testing and/or development also offer Nursing a potential opportunity for interdisciplinary collaboration with an aligned discipline that may contribute complementary conceptual perspectives and items. For example, in the literature review process about perceived health status, one article compared the concepts of health and wellness, the latter coming from a behavioral, counseling and education philosophy (Greenberg, 1985). Although beyond the scope of this paper, it is noteworthy that the two concepts include some of the same elements, and that wellness is often measured using the (validated) Perceived Wellness Survey (PWS) (Adams, Bezner, & Steinhardt, 1997), thus opening the possibility to examine use of selected health related items.

A review of the literature presented a discussion of existing research, foci and instruments addressing the concept/construct of perceived health status. Gaps and needs were

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identified spanning clinical, research and public health arenas. Potential strategies were presented for consideration, including a proposed systems-based interdisciplinary healthcare model that integrates the construct of health status and associated measures, and suggestions for instrument development and/or modification for concept measurement. These considerations reflect sincere commitment to individual health perspectives, values and goals (including personal utility of healthcare related services), incorporate dimensions of biomedical, Nursing and behavioral models, and offer opportunities for examining clinical, theoretical and research applications. It is incumbent upon the Nursing profession to be well-prepared for evolving challenges and the changing paradigm for healthcare, such as posed by the explosion of informatics, technology and genomics, as well as to utilize Nursing's track record and inherent strengths in interdisciplinary and international collaborations to foster respect for individual health values, prevention and health promotion in research, education, clinical practice and policy initiatives (Badzek, Henaghan, Turner, & Monsen, 2013; Colthart, 2010; Genomic Nursing State of the Science Advisory Panel et al., 2013; Goldsmith, Jackson, O'Connor, & Skirton, 2012; Greco, Tinley, & Seibert, 2011).

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## CHAPTER V

### INDIVIDUAL PERCEIVED HEALTH STATUS AND HEALTH VALUES: FACTORS INFLUENCING SELF-INITIATED GENOME TESTING AND HEALTH ACTION

**Authors:** Carolyn D Farrell, PhD(c), MS, Clemson University; Kevin Eng, PhD, Roswell Park Cancer Institute; Bonnie Holaday, PhD, Clemson University; J. Scott Roberts, PhD, University of Michigan School of Public Health and Robert C Green, MD, MPH, Brigham and Women's Hospital, Harvard Medical School, for the PGen Study Group

**Corresponding Author:**

Carolyn D Farrell, 84 Carriage Circle, Buffalo, NY, 14221

(716) 308-3592, (716) 688-2002 [fax], [cmfarre@g.clemson.edu](mailto:cmfarre@g.clemson.edu)

## Abstract

**Purpose:** To: 1) examine health status, categorized by both individual perceived health and biomedical health (medical diagnosis), for discrepancies and to characterize group differences, and 2) assess individual perceived health status and health values as influencing self-initiated genome testing. **Methods:** 1464 direct-to-consumer (DTC) genome testing users, participants in the Impact of Personal Genomics (PGen) study, were classified into four mutually exclusive health status groups. Groups were analyzed on reasons for pursuit of testing, individual risk perception and health values by Chi-square ( $\chi^2$ ), t-tests, ANOVA and correlations. **Results:** All four categories were represented, confirming discrepancies between individual and biomedical health status. One group may represent the “worried well”. Interest in health-related information was high (98–99%) across all groups. Two items distinguished all groups (interest in pharmacogenomics information and learning risk for other diseases ( $p < .001$ ), while some items distinguished one group. When groups differed, perceived rather than biomedical health status was often involved, yet both factors demonstrated influences (variable dependent). Risk perception was moderately correlated (.301) with health status group. **Conclusions:** Individual perceived health status and health values are significant factors influencing self-initiated health action (pursuit of genomic testing), suggesting benefit of integration and complementarity in effective healthcare practice, research, models and policy.

**Keywords: [5]**

Perceived health status, individual health values, personalized healthcare, risk perception, genomic testing

**Introduction**

Through 2014, an estimated one million persons have pursued direct-to-consumer (DTC) genome testing, providing them with health-related results<sup>1,2</sup>. Despite recent FDA regulatory action currently restricting DTC labs from marketing and providing health-related genomic testing<sup>3,4</sup>, final authoritative determinations are yet to be made<sup>5</sup>. Nonetheless, official positions are unlikely to thwart public interest, demands and expectations regarding genomics in healthcare<sup>6-8</sup>. Motivations for individual pursuit of genome testing identified through research span curiosity, desire for information about health (risks), support for research, genealogy, and fun/entertainment<sup>9-13</sup>. These results are limited, however, by factors including recruited or convenience populations, small sample size, anticipated versus actual genome testing, example cases versus actual results, equating genetics knowledge with ability to understand or use genomic information appropriately, use of surrogate measures for subjective concepts (e.g., absence of medical diagnosis as individual good health) and the nature of survey questions<sup>9,13-17</sup>. This latter issue regarding questions potentially compromises findings and interpretations in ways not previously appreciated or readily apparent, e.g., what questions are posed (albeit objective); use of technical or culturally-influenced terminology; limited, duplicitous or confusing answer choices; and researcher/clinician

over-simplification or misunderstanding of participant text responses. Thus perpetuating that “we do not know what we do not know”<sup>18</sup>.

Important unanswered questions and gaps in knowledge remain. Do individuals who pursue DTC genome testing deem this information essential to their health and health management decisions? Do they represent the “worried well”<sup>19</sup>, or proactive prevention-oriented persons, neither deemed needy of over-burdened healthcare services? And perhaps, of most relevance, “Does individual perceived health status (regardless of biomedical health status) and personal health values act as drivers influencing individual pursuit of genome testing or value (personal utility<sup>20</sup>) of genomic information? Answers to these questions extend well beyond the genomics context.

Perceived health status, although recognized as a powerful predictive factor in studies of mortality and morbidity<sup>21-24</sup>, has seen limited exploration in the context of prevention and health promotion research. In spite of its potential importance, it is essentially treated as an ancillary or complementary item/measure<sup>17,24</sup> rather than as an integral, and possibly even determinative, factor in individual health priorities and decisions. Perceived health status, by its subjective nature, must encompass some internal personal measure(s) for interpretation of one’s health (and/or risks)<sup>25</sup> resulting in perceived gradations of acceptable, unacceptable or choice to ignore. Depending upon that assessment, an individual may determine whether needs exist and if so, whether or not to take action. This process also implies the existence of personal health values (distinct from those of healthcare, e.g., cost/benefit)<sup>26</sup> and goals that influence motivations to action. Thus, individual health-related actions can be viewed as part of an



interactive process with perceived health status and personal health values and goals, consistent with the conceptual basis (Health Capability<sup>27</sup>) of the current study.

The population studied here represents a rare, naturally occurring sample of both medically diagnosed as well as healthy persons (not as controls), unaffected by the influence of clinicians, researchers or a controlled study prior to independently deciding to pursue genome testing, who subsequently enroll in the PGen study<sup>28</sup>. This study sought to explore the influence of subjective individual factors, combined with and distinguished from traditional healthcare contexts, specifically perceived health status, health values, risk perception<sup>17,29-32</sup> and reasons for pursuit of genome testing. This knowledge relates not only to DTC users, but potentially contributes to identifying previously unrecognized individual motivations and values that impact many health behaviors, as well as fostering individual-practitioner partnership and respect in healthcare priorities and decisions, and more effective use of healthcare resources.

## **Materials and Methods**

### **Study Design**

This study examined survey data from persons who independently pursued direct-to-consumer (DTC) genome testing and voluntarily participated in the National Institutes of Health funded Impact of Personal Genomics (PGen) Study<sup>33</sup>. The PGen initiative developed three surveys using items (e.g., multiple-choice, Likert-type) from existing genetics research instruments with previously demonstrated reliability and validity<sup>14-16,34-39</sup>. Items spanned domains and content relevant to the current study and its conceptual framework (Health Capability)<sup>27</sup>, including but not limited to motivations and

expectations (for genome testing), risk perception, socio-demographics, personal utility, individual health values, individual medical history, family history and health-related behaviors. Surveys were completed in 2012, two weeks prior to pursuit of genome testing (BL), and two weeks (2W) and six months (6M) after testing. This study, approved by the PGen Review Board, Harvard Medical School, and the Clemson University IRB, utilized de-identified study-specific data provided in an SPSS format, primarily from the BL survey with relevant items solely in the 2W survey.

### **Participants & Survey Dissemination**

Potential participants were invited via the health-based social networking site *PatientsLikeMe* and a banner on the Pathway Genomics website. Personal genome testing (SNP analysis), including provision of health-related results, was pursued through either the 23andMe or Pathway Genomics laboratory<sup>40,41</sup>. The original PGen population consisted of 1,838 consenting persons, with 1,648 eligible after eliminating partial or late received surveys; 1,464 persons completed the second survey and were eligible for this study. They were categorized into four mutually exclusive health status groups (the independent variable) based on dichotomizing perceived health and medical diagnosis status. Participants included males and females, ages 19-94, of different races, ethnicities and educational, socioeconomic and occupational backgrounds.

### **Data Analysis**

Statistical power was assessed based on the number of eligible PGen participants, as well as the numbers anticipated for the current study's groups. This was determined to

be sufficient for the intended data analyses, with a power of .8 and at a 0.05 level of significance<sup>42</sup>. It is relevant to note that the study variables are qualitative and subjective in nature, were assessed primarily by Likert-type scale items (with 3-5 choices), and were suspected to be in an interrelated, interdependent relationship (consistent with this study's conceptual framework). Descriptive statistics (mean; standard deviation) and frequency analyses were done to characterize health status groups (see Table 5.1). Groups were compared on socio-demographic items, reasons for pursuit of genome testing, risk perception and individual health values using Chi-square ( $\chi^2$ ), t-tests and/or analysis of variance (ANOVA) (depending on whether these dependent variables were categorical or continuous). Statistically significant differences identified by ANOVA were subjected to independent t-tests between two-group combinations to identify the source/s of significant difference. Original PGen scoring of variables was maintained, or if modified, variable direction and construct integrity was preserved. Operationalization of measures is addressed in the Results sections. Limited qualitative data was examined by query of study-relevant text answers for targeted terms (keywords) to identify and quantify existence of factors that could impact health status groups and/or interpretation of results (e.g., mention of undiagnosed health problem, or diagnosis not included in the survey).

## **Results**

### **Health Status Groups**

Respondents were classified into four mutually exclusive health status groups by dichotomizing perceived health status and health status based on medical diagnosis.

**Table 5.1. Characteristics of PGen study participants<sup>1</sup> included in health status groups.**

Health Status Groups Total N = 1455 <sup>A</sup>	Diagnosed ILL N=213		Med. Managed N=950		Healthy N=292		p-value
	N	(%)	N	(%)	N	(%)	
<b>Characteristics of PGen sample<sup>1</sup></b>							
<b>Number of Medical Conditions<sup>2</sup></b>							
None	0		0		292		N/A
One or more	213		950		0		
Range # Conditions (Mean)	1-10	(1.999)	1-9	(2.750)	N/A	(0)	0.000
<b>Perceived Health Status<sup>3</sup></b>							
Good	0		950		292		N/A
Not Good	213		0		0		
<b>Age, years</b>							
Mean (SD)	51.2	(14.8)	50.1	(15.3)	37.5	(12.1)	0.000
% Under 35 (% ≥65)	16.9	(19.2)	22.6	(20.0)	50.0	(4.8)	
Range	21-91		19-94		19-76		
<b>Gender</b>							
Female	148	69.5%	580	61.1%	162	55.5%	0.006
<b>Education</b>							
< College Degree	76	35.7%	197	20.7%	44	15.1%	0.001
College Degree	75	35.2%	398	41.9%	132	45.2%	
> College Degree	62	29.2%	355	37.3%	116	39.7%	
<b>Income</b>							
< \$100,000	155	72.8%	500	53.5%	138	47.6%	0.000
> \$100,000	54	25.4%	435	44.9%	152	52.4%	
<b>Race/Ethnicity (frequency &gt;5%)</b>							
White	194	91.1%	868	91.4%	246	84.2%	0.002
Hispanic	11	5.2%	50	5.3%	18	6.2%	0.824
<b>Adopted</b>							
Yes	12	5.6%	46	4.8%	16	5.5%	0.842
<b>Biologic Children</b>							
Yes	116	54.5%	529	55.7%	108	37.0%	0.000
<b>Marital Status</b>							
Single	40	18.8%	152	16.0%	84	28.8%	0.201
Married	109	51.2%	544	57.3%	141	48.3%	
Widow/Divorce/Separated	45	21.1%	122	12.8%	18	6.2%	
Live tog/Long term relation	19	8.9%	132	13.9%	49	6.8%	
<b>Role in Own Healthcare Decisions<sup>4</sup></b>							
I have sole/main <sup>4</sup> final decis	123	57.7%	610	64.2%	206	70.6%	0.027
MD & I share responsibility	76	35.7%	290	30.5%	69	23.6%	
MD makes main/sole <sup>4</sup> decis	14	6.6%	50	5.3%	17	5.8%	

**KEY:** Abbreviations: tog = together; decis = decision

<sup>1,A</sup> Impact of Personal Genomics (PGen) study. N=1464 eligible for second survey (2W). The current study excluded 9 records (2 missing data for group assignment & 7 from a Health in Question health status group too small for analysis) N/A = not applicable (these variables defined 4 mutually exclusive Health Status groups, including Health in Question<sup>A</sup>)

<sup>2</sup> Refers to 14 survey-specified medical conditions (types of disorders), with individual totals ranging 0 -14 (actual highest total was 10). Variable was dichotomized to either None (no medical diagnosis), or One or more medical diagnoses.

<sup>3</sup> Likert-type scale choices were dichotomized: Good = good, very good & excellent; Not good = fair & poor.

<sup>4</sup> Survey question offered five choices. Reduced to three values for data analysis (sole and main choices were combined).

Diagnosis related to presence or absence of any of fourteen survey-specified types of conditions: arthritis, asthma, cancer, chronic kidney disease, diabetes, eye, gastrointestinal, heart, high cholesterol, lupus, mental illness, neurological, obesity and psoriasis. Perceived health status was dichotomized as: good (good, very good, excellent) or not good (fair, poor). Of 1,464 eligible surveys, two lacking answers for perceived health status were eliminated, as were another seven noted among the following health status groups, leaving 1,455 persons:

- Diagnosed Ill (DI): 213; one or more diagnoses (mean 4.40; range 1-10) and perceived health as not good.
- Medically Managed (MM): 950; one or more diagnoses (mean 2.75; range 1-9) and perceived health as good.
- Healthy (H): 292; without a diagnosis and perceived health as good.
- Health in Question (HQ): 7; without a diagnosis and perceived health as not good (eliminated due to small size; see Discussion).

Diagnoses types between the DI and MM groups were not statistically different except for arthritis, eye, gastrointestinal, obesity and neurological conditions ( $p < 0.05$ ), and kidney disease ( $p = 0.05$ ). Socio-demographic characteristics, noting significant differences on several variables, are summarized in Table 5.1. T-tests clarified most differences involved the Healthy group. The DI and MM groups had similar mean ages (51.2 and 50.1, respectively, compared to 37.5 for the Healthy) and percent having biologic children (54.5 and 55.7%, respectively, compared to 37% for the H). Sex was only significantly different ( $p = 0.006$ ) between DI and H groups (69.5% versus 55.5% female, respectively). Participants were primarily White, 5-6% Hispanic, and minorities

represented fewer than 5% (African-Americans, Hawaiian/Pacific Islanders, Asians, American Indians). Income was significantly different ( $p < 0.001$ ) by t-test comparisons involving the DI group. Individual decision-making role with healthcare providers demonstrated the majority, regardless of group, shared in responsibility; however, the DI and H groups were significantly different ( $p = 0.034$ ).

### **Reasons for Pursuit of Genome Testing**

Importance of twelve specifically queried reasons for pursuit of genome testing is summarized in Table 5.2, including five identified as significantly different. All other items, except one, were uniformly rated as important. Finding out about personal response to different medications distinguished all three groups ( $p < 0.001$  for both DI group comparisons;  $p = 0.037$  for MM with H). T-tests identified the DI group as the source of most between group significant differences ( $p < 0.01$  for personal risk for disease, creating a better plan for the future, and test seeming fun/entertaining). The Healthy group was the source of significant difference for only importance of health condition risk information for children ( $p < 0.01$ , with either DI or MM).

A text response to reasons for pursuit of genome testing was queried for keywords including “undiag...”, “health problem”, “I have”, “unknown” and “health concern” to identify frequencies of participants expressing desire/hope to get information about undiagnosed health problems or diagnoses beyond those in the survey. Of 1,352 respondents, twenty-eight instances were identified (see Table 5.2). This information would have changed health status group assignment for two cases (from H to MM).

Table 5.2: Reasons for pursuit of genome testing.

Reasons Pursued Testing	Health Status groups: (Total N = 1455)	Significance <sup>1</sup>	Dx ILL N=213 N (%)	Med. Managed N=950 N (%)	Healthy N=292 N (%)
1. Curiosity					
Important		0.572	211 (99.1)	934 (98.3)	288 (99.0)
2. Personal Risk for Specific Disease <sup>2</sup>					
Important		<b>0.012</b>	<b>207 (97.2)</b>	<b>866 (91.3)<sup>A</sup></b>	<b>266 (91.1)<sup>A</sup></b>
3. Learn Genetic Make-up without Physician					
Important		0.455	132 (62.0)	586 (61.7)	168 (57.5)
4. Desire to Improve Health					
Important		0.059	190 (89.2)	805 (84.8)	238 (81.5)
5. Find Out Response Type to Different Meds					
Important		<b>0.000</b>	<b>191 (89.7)</b>	<b>717 (75.6)</b>	<b>202 (69.4)</b>
6. Create Better Plan for Future					
Important		0.014	186 (87.3)	<b>747 (78.7)<sup>B</sup></b>	<b>228 (78.4)<sup>B</sup></b>
7. Personal Interest in Genetics in General					
Important		0.972	198 (93.0)	887 (93.4)	271 (93.1)
8. Seemed Fun & Entertaining					
Important		<b>0.001</b>	<b>142 (66.7)</b>	<b>731 (77.0)<sup>C</sup></b>	<b>236 (81.1)<sup>C</sup></b>
9. Family Members Using Personal Genomics <sup>3</sup>					
Important		0.226	41 (19.2) [35.0] <sup>3</sup>	237 (24.9) [39.6] <sup>3</sup>	71 (24.3) [39.7] <sup>3</sup>
10. Desire Learn More: Limited Family Info <sup>3</sup>					
Important		0.159	166 (77.9) [85.6] <sup>3</sup>	662 (69.7) [78.3] <sup>3</sup>	190 (65.1) [72.7] <sup>3</sup>
12. Want Health Risk Info for Children <sup>3</sup>					
Important		<b>0.003</b>	<b>140 (65.7) [82.4]<sup>A,B</sup></b>	<b>666 (70.1) [82.8]<sup>A,B</sup></b>	<b>197 (67.5) [75.5]<sup>3</sup></b>
Q6 Text: # Targeted Reasons Pursued Testing <sup>4</sup>			13	15	0

**KEY & NOTES**

- <sup>1</sup> Chi Square (3x2) & Pearson (asymptotic, 2-tailed). **BOLD** = Significant. If significant, t- tests done to confirm significance and source(s).
  - <sup>2</sup> Separately, Q6 text answers in some records include mention of a condition/health problem(s) of concern other than those specified in the survey.
  - <sup>3</sup> Second [bracketed] percent represents deleting "not applicable" from Group N. However, other issues exist; used original (see Discussion).
  - <sup>4</sup> Records with text answers using term(s) indicating hope/reason as getting information about undiagnosed, or recently diagnosed, health problem.
- Highlight = Group(s) that is (are) source of statistically significant differences  
 Highlight = MM and H with similar frequencies; not statistically different. <sup>A</sup> p = 0.996; <sup>B</sup> p = 0.991; <sup>C</sup> p = 0.357  
 Highlight = DI and MM with similar frequencies; not statistically different. <sup>B</sup> p = 0.989
- NOTE: Missing entries for items 1-12 were treated as not interested (only 1 record in MM and/or H groups for some questions).



## **Risk Perception**

Risk perception analysis encompassed two approaches. Frequencies (percentages) of perceived increased risk to develop each of twelve survey-specified conditions is presented in Figure 5.1. Another twelve conditions are not included due to poor response rate (< 40%). Figure 5.1 illustrates vertically, by increasing depth of color of health status group columns, an apparent trend in perceived risk being lowest in the Healthy, in the mid-ranges in the Medically Managed, and highest in the Diagnosed Ill. In contrast, cluster patterns across health status groups are represented horizontally in Figure 5.1 (bracketed, left side), e.g., three conditions at the bottom and four at the top demonstrating parallels in highest perceived risks across all three groups.

Second, risk perception was captured as a single variable representing the total number of survey-specified conditions (0-24) that each individual noted as being at perceived higher than average chance to develop (survey language). This type of additive approach has been used previously in research<sup>43</sup>. Group means were significantly different ( $p < 0.001$ ). The means (and range of conditions) were: Healthy 1.61 (0-8), Medically Managed 3.01 (0-17) and Diagnosed Ill 4.36 (0-13). Between group comparisons by condition, presented in Table 5.3, demonstrate the Healthy group as significantly different from the DI for all nine conditions and from the MM on seven conditions. Comparison between the two groups with medical diagnoses (DI and MM) demonstrated significant differences relating to colorectal and lung cancer, diabetes and heart disease, despite similar mean ages. Among persons indicating no increased risk for any of the 24 conditions, 92.4% indicated their perceived health status as good. In

**Figure 5.1: Percentage of Individuals within Health Status Groups at Perceived Above-Average Chance to Develop Survey-Specified Conditions.**

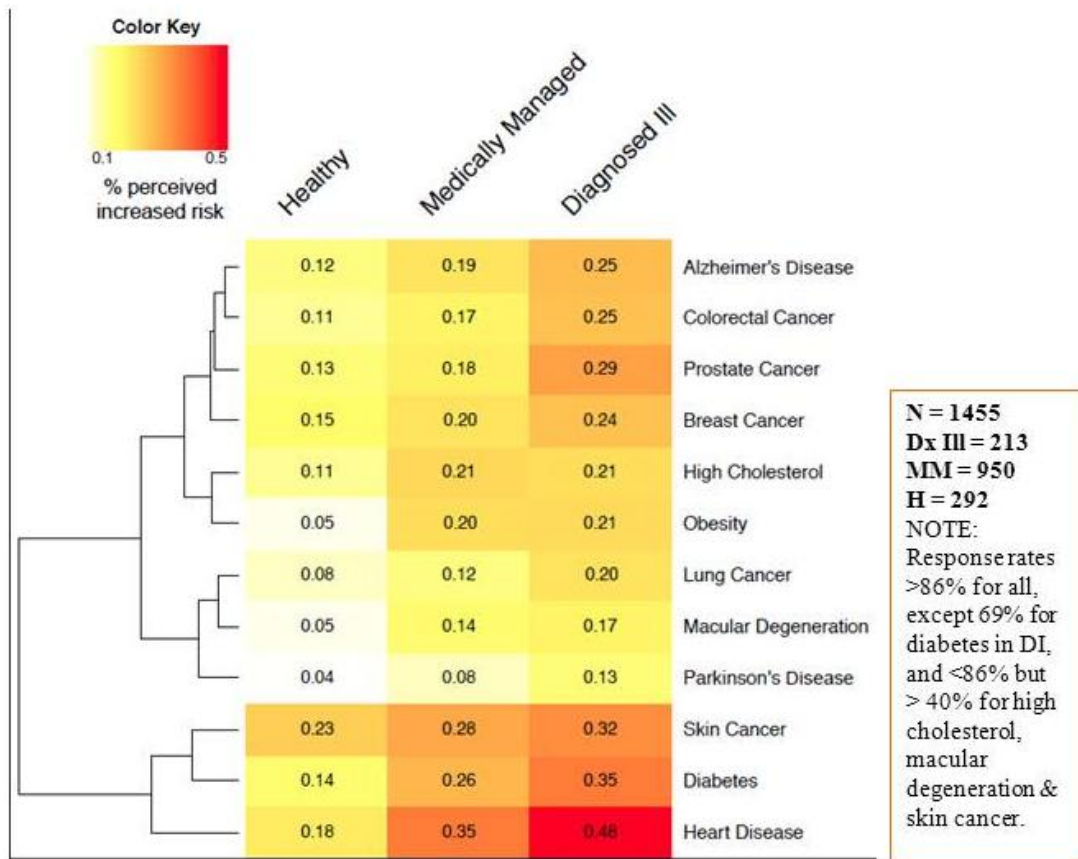


Table 5.3: Individuals at perceived above-average chance of developing survey-specified conditions by health status group.

Survey Conditions	DIAGNOSED ILL N = 213 <sup>A</sup> # (%) at increased risk	Significance** ILL with MEDICALLY MANAGED	MEDICALLY MANAGED N = 950 <sup>A</sup> # (%) at increased risk	Significance** MEDICALLY MANAGED with HEALTHY	HEALTHY N = 292 <sup>A</sup> # (%) at increased risk	Significance** HEALTHY with DIAGNOSED ILL
<i>Alzheimer's</i> <sup>B</sup>	54 (25.3)	.059	182 (19.1)	.007	34 (12.0)	.000
<i>Breast cancer</i> <sup>C</sup>	36 (24.3) <sup>A</sup>	.210	114 (19.6) <sup>A</sup>	.158	25 (15.4) <sup>A</sup>	.041
<i>Prostate cancer</i> <sup>C</sup>	19 (29.2) <sup>A</sup>	.053	66 (17.8) <sup>A</sup>	.133	17 (13.0) <sup>A</sup>	.007
<i>Colorectal cancer</i> <sup>D</sup>	53 (25.0)	.017	162 (17.0)	.007	31 (11.0)	.000
<i>Lung cancer</i> <sup>D</sup>	42 (19.7)	.010	115 (12.1)	.041	23 (8.0)	.000
<i>Diabetes</i> <sup>D</sup>	74 (34.7)	.004	250 (26.3)	.000	40 (14.0)	.000
<i>Heart disease</i> <sup>D</sup>	103 (48.3)	.000	329 (34.6)	.000	53 (18.0)	.000
<i>Obesity</i> <sup>B</sup>	45 (21.1)	.079	193 (20.3)	.000	16 (5.0)	.000
<i>Parkinson's</i> <sup>B</sup>	28 (13.1)	.053	79 (8.3)	.023	13 (4.0)	.001

**KEY & NOTES:**

<sup>A</sup> Group N, except breast & prostate cancer (N = relevant sex). Female: DI = 148, MM = 581, H = 162. Male: DI = 65; MM = 369; H = 130.

\*\* Significance is based on Pearson Chi Square.

• Highlighted Orange blocks represent between group statistically significant differences.

<sup>B</sup> H is significantly different from DI and from MM.

<sup>C</sup> H is significantly different from only DI.

<sup>D</sup> Statistically significant differences between AND across ALL Groups

• Mean number of conditions by Group: Diagnosed Ill = 4.36; Medically Managed = 3.01; Healthy = 1.61

• Scoring of perceived risk for each condition was dichotomous: 1 = above average chance to develop; 0 = average or lower than average chance. Maximum possible individual score was 24. Range of individual scores was 0-17.

• Response rate  $\geq$  86% (except 69% for obesity in the DI group) for these Table 3 (first nine) survey-specified conditions.

• Missing entries considered at average or less than average risk since respondents had indicated above-average risk for other conditions.

contrast, 62.5% of persons having no perceived increased risk had one or more medical diagnoses.

To potentially identify if risk perception was differentially associated with perceived health status, with health status based on existing medical diagnosis, or with the combined health status, Pearson correlations were done. Perceived risk was weakly correlated with medical diagnosis status (.272), was moderately correlated with health status group (.301) and was moderately correlated (.326) with perceived health status. This suggests a greater influence by perceived health status. All correlations were statistically significant at the  $p = .01$  level (two-tailed).

### **Health Values**

Consistent with existing literature, individual health values were operationalized in this study by items (ten pre-testing and six post-testing) encompassing participant indicated importance, interest, consideration or value (in the context of genomic testing or information)<sup>27,44</sup>. Table 5.4 summarizes the results of answers pre-testing, including details of the four achieving statistical significance. The DI group was significantly different from both the MM and H groups on all four items. The MM and H groups were similar on two of the four, as well as being within 1.5% of each other on three other items. The Healthy group was significantly different from both the DI and MM groups on one item. All three health status groups were significantly different regarding interest in learning information about personal response to drugs and indicating interest in learning risk for other disease/s. Two items not achieving statistically significant

Table 5.4: Personal Factors Considered, of Interest & Valued in Deciding About Pursuit of Genomic Testing.

<i>Factors Considered &amp; Questions</i>	Significance (2-tailed)	Diagnosed Ill (N=213) N (%)	Medically Managed (N=950) N (%)	Healthy (N=292) N (%)
1. How well results predict if I get a particular disease Considered	0.020*	187 (87.8%)	755 (79.5%)	234 (80.1%)
2. Whether I can take health actions from results Considered	0.143	200 (93.9%) <sup>A</sup>	850 (89.5%) <sup>A</sup>	263 (90.1%)
3. Other factors considered? (Text response?) Yes	0.066	38 (17.8%)	137 (14.4%)	31 (10.6%)
4. What I learn can help reduce my risk of getting sick Agree	0.035*	191 (89.7%) <sup>A, B</sup>	783 (82.4%) <sup>A</sup>	244 (83.6%) <sup>B</sup>
5. Learning info: Risk disease/health problem Interested	0.143	211 (99.1%)	935 (98.4%)	283 (96.9%)
6. Learning info: Response to Drugs Interested	0.000*	207 (97.2%) <sup>C</sup>	870 (91.6%) <sup>C</sup>	249 (85.3%) <sup>C</sup>
7. Learning info: Carrier status (pre-pregnancy plans) Interested	0.000*	113 (53.1%)	532 (56.0%)	200 (68.5%) <sup>C</sup>
8. Learning info: Ancestry Interested	0.978	205 (96.2%)	913 (96.1%)	280 (95.9%)
9. Learning info: Traits <sup>1</sup> Interested	0.804	85 (39.9%)	645 (67.9%)	191 (65.4%)
10. Learning risk: other disease. (Text response?) Yes (N= 390)	0.000*	90 (42.3) <sup>C</sup>	254 (26.7) <sup>C</sup>	46 (15.8) <sup>C</sup>
<b>Q6 Text: Reasons for Pursuit Genome Testing<sup>2</sup> (N=155)</b>				

**KEY:**

#1-2: dichotomized (Considered/Not). #3 & #10: Text answer present (Yes/No). #4: Agree=2; Disagree=1; Neutral=0. #5-9: dichotomized (Interested/Not).

\*BOLD = Statistically significant by t-testing (2-tailed; equal variance assumed – as conservative approach).

Highlight orange = source of statistically significant difference/s (either the single group, or all three)

Highlight green = No statistically significant difference between groups. Bonferroni & Scheffe tests suggest combining groups into a subset.

<sup>A</sup> Statistically significant difference between DI & MM by T-test ( $p \leq 0.01$ )

<sup>B</sup> Statistically significant difference between DI & H by T-test ( $p \leq 0.05$ )

<sup>C</sup> Statistically significant difference between highlighted group/s in each 2-group combination by T-test ( $p \leq 0.005$ )

<sup>1</sup> Missing 515 entries.

<sup>2</sup> Q6 Text answers include term(s) indicating...



differences (see Table 5.4) had uniformly high levels of interest (89.5-99%). The value of obtaining information about traits could not be assessed reliably because of missing data (30.8% - 59.2%).

Six post-testing items inquired about genome results in relation to individual health value. Results on Likert-type items (five scored 1-5, lowest for not at all or strongly disagree; highest for extremely or strongly agree) identified value for all items across all health status groups, and a pattern of highest to lowest scores from DI, to MM, to H. Overall, means ranged from 3.46 (between neutral and somewhat agree) in the Healthy group (for results helping to decrease risk of getting sick), to 4.16 in the DI group for results being valuable. This latter item had uniformly high agreement across all three groups (means 4.07 and 3.98 for MM and H, respectively). Statistically significant differences were identified by t-tests on two items: 1) importance of information as influencing future health management, between the DI and H groups ( $p = 0.019$ ) and, 2) ability to use the information to improve health ( $p < 0.001$ ) between all groups.

## **Discussion**

This large-scale study of self-initiated DTC genome testing users demonstrated that categorizing individuals by health status based on both perceived health and existence of a medical diagnosis created the four hypothesized groups, each having clinically meaningful differences. This supported not only the previously established influence of perceived health status related to existing medical conditions and morbidity<sup>21,22</sup>, but contributed new awareness of its involvement in the health-related choices of healthy persons.

## **Health Status Groups**

The groups were characterized to identify distinguishing characteristics and answer the research questions. The Healthy group represents individuals who generally do not come to the attention of the healthcare system, and the discrepant health status groups were also of interest.

An unexpected finding was that there were only seven persons in the Health in Question group. This group, who had no diagnosis but indicated their health as not good, could represent the “worried well”<sup>19</sup>, persons lacking diagnosis for an existing health problem, and/or be hoping for some type of specific information. The low number could be explained if larger numbers exist but are subsumed into other groups, e.g., into the MM or DI because they have a co-existing survey-specified diagnosis. This consideration is supported by the 28 persons with diagnostic concerns identified through analysis of text answers to reasons for pursuit of genome testing; however, none were in the HQ group. Mitigating factors include limited query, attribution of concerns to others (e.g., relatives, thus were not counted), not articulating this concern in their text answer, or non-response. It could also be that an existing medical diagnosis was their highest priority and/or that other concerns are addressed during regular healthcare visits.

Both the Diagnosed Ill and Medically Managed groups have medical diagnoses yet sort into different groups, attesting to the influence of perceived health status. Although group differences could be ascribed to differences in the mean number of diagnoses for the DI and MM groups (4.40 versus 2.75, respectively), in both groups all persons had medical diagnoses, the range in number of conditions was similar (nine



versus ten, respectively) and their mean ages were similar. It is possible that the nature/type of diagnoses differed between these groups, or that perceived risk contributed to perceived health.

### **Perceived Risk**

Risk Perception, a concept explored in prior DTC genetic testing studies as a motivating factor or concern<sup>12,17,45</sup>, was examined in relationship to perceived health status in the current study. Risk perception was found to be significantly different across health status groups (see Table 5.3), being greatest in the DI group. From the distribution of groups and conditions in Figure 5.1, it appears that both perceived health status and medically defined health status contribute to risk perception. Two study findings suggest that perceived health status may be an initial, greater and/or at least meaningful influence for pursuit of genomic testing. The Healthy group, with perceived good health and no medical diagnosis, had the lowest perceived risk. This makes sense and does not necessarily imply the influence of perceived health status. However, the fact that the MM group had a significantly lower perceived risk than the DI group supports the potential influence of perceived health status (good in MM). The Health in Question group, albeit small, were youngest (mean age 30.3) and were without a survey-specified medical diagnosis, yet rated their health as fair or poor and had the highest mean (4.57) for conditions at perceived increased risk. These persons were not identified by text query of reasons for pursuit of genome testing, yet cursory examination of related text fields identified statements such as: "...interested in Genomics, and think it is a good idea given my medical history..." and "I chose ... genetic testing because of some health

issues I am experiencing”. Entries also included mention of desire for risk identification, condition-specific or prevention-related information. A qualitative study involving this HQ group and examination of all study population text answers may identify others in this group, shed light on this perplexing issue and contribute to understanding health factors relevant to effective and individualized healthcare.

Since the nature of the groups examined the combined effect of both health status parameters, in relation to perceived risk, correlations were done to examine these parameters separately. Perceived risk was weakly correlated (.272) with medical diagnosis status alone, whereas perceived health status alone or combined with medical diagnosis status demonstrated moderate correlations (.301 and .326, respectively). These weak or moderate correlations are consistent with other findings. Of the persons (33) indicating the highest number of conditions at increased risk (10-17), the majority (23) had only one to three medical diagnoses. Of the persons (65) with the highest number of medical diagnoses (7-10), only one indicated more than eight risk conditions. These results support that perceived risk and perceived health status likely influence each other (are positively correlated), yet also indicate that neither equates with nor substitutes for the importance of integrating the other. Further studies may be helpful in clarifying risk perception relevant to these health status groups, and perceived health status alone, such as disorder-specific perceived liability/risk, positive family history and/or the results of genomic testing.

Finally, it is noteworthy that regardless of health status group, the major targets of perceived increased risk were heart disease, diabetes, skin cancer (melanoma), and breast

and prostate cancer. These concerns may reflect positive family history, personal experience, genome laboratory information, public education, the influence of mass media, or interest in common diseases<sup>11</sup>. This knowledge has relevance for clinicians, researchers, personalized healthcare and genomic testing laboratories, relating to readiness and timing of educational efforts, addressing screening and management of risk factors, and discussion with potential pursuit of genomic testing.

### **Reasons for Pursuit of Genome Testing & Health Values**

Across all three health status groups, at least 69% of participants assigned importance to ten of twelve reasons for pursuit of genome testing (see Table 5.2); over 81% indicated the desire to improve health as important. The reasons posed to participants for assessment of their interest were consistent with those identified in other DTC genome testing studies, as was the finding that the majority assigned importance to the desire to improve health<sup>10,12,46</sup>. This response implies a perceived or expected connection between genomic information and improving health. The lowest levels of importance were associated with family members using personal genomics, and learning about genetic make-up without physician involvement. The former suggests that family pressures were not a major issue (although relevant for over half), and while the latter is consistent with previous research demonstrating individual interest in collaborating with healthcare providers<sup>11</sup>, 58-62% of participants rated this item as important. Possible explanations include the inherent nature of persons who independently pursue genome testing, public concerns about genetic discrimination (e.g., if genomic information gets into their health records)<sup>47</sup>, or that genomic information is valued and cannot be obtained

through their healthcare provider. Patients and providers could benefit from open discussion to foster better understanding of health values, goals, concerns and utility of genomic information toward personalized and more effective healthcare<sup>48</sup>.

The Diagnosed Ill and Medically Managed groups, despite having a medical diagnosis and age in common, demonstrated statistically significant differences on four of five reasons for pursuit of genome testing (see Table 5.2). These items related to immediate medical risks (specific disease, drug response) or future planning (for self or family). In contrast, the Medically Managed and Healthy groups both with perceived good health, who were statistically different in age, income and diagnostic status, were not significantly different on importance for three (of the five reasons) for pursuit of testing. These reasons involved future plans, identifying disease risk and entertainment value, suggesting less immediate perceived health needs.

The most dramatic finding was that all three groups were significantly different ( $p < 0.001$ ) on the two survey questions about importance of finding out about personal response type to different medications (see Table 5.2 and Table 5.4). These results demonstrate internal consistency, and also support potential clinical relevance for this study's health status categorization. The DI group had the highest percent interested in drug response information, followed by the MM and then the Healthy group; however, even that lowest encompassed 85.3%. The DI's highest level of importance may be because that they have the most to gain if genomic information helps in personalizing more effective treatment or avoiding drug-related complications. This reasoning suggests a greater influence of diagnostic health status, as opposed to perceived health, for

individual importance of learning their drug response type; however, this would not explain why the MM group had a lesser percentage than the DI. These results, and the lower percent of importance for the H group, support the influence of both medical diagnosis and perceived health. This assessment is also supported by the significant difference ( $p < 0.001$ ) across all three groups for interest in learning about the risk for other diseases. Overall, these findings demonstrate the personal value and importance that individuals place on personalized drug-related information<sup>17</sup>, providing an insight relevant to healthcare providers, policy developers and genome testing laboratories.

The Diagnosed Ill group also assigned the highest importance to receiving information about personal risk for a specific disease, and creating a better plan for the future (see Table 5.2). The desire for genomic information about an existing disease may be surprising, yet is supported by another study<sup>49</sup>, although the reasons are not clear. Do these people hope that genomic information will clarify or assign cause for their condition, foster targeted treatment to ameliorate disease, and/or have prognostic value? Or does this simply reflect a fundamental desire of people to value “answers”. These potential reasons could interrelate with importance of creating a better plan for the future. The DI group (e.g., if facing mortality) may view genomic information as important to family, consistent with their assigned importance to genomic information for their children (82.4%).

Finally, the MM and H groups also had a high percent assigning importance to personal risk for disease (see Table 5.2). Participants across all groups indicated creating a better plan for the future as important, yet the reasoning, values or goals behind those

similar choices may be very different. This information may be useful to clinicians. For example, for the DI group, planning for the future may warrant discussion of DNA banking, whereas, for the perceived healthy groups, there may be a desire for early risk identification, maintaining and promoting health, the issue of personal utility or value, potential for motivating health behaviors, and/or implementing prevention measures.

Values Pre-testing: Although there was no specific health values question, ten values-related items allowed some capture of this concept with inferences (see Table 5.4). Interestingly, the nature of these value results paralleled the group findings for reasons for pursuit of genome testing. In four out of five items, the DI group was significantly different from the others. The one exception was the Healthy group for value (higher) of carrier status information, possibly due to the questionnaire's parenthetical reference to pre-pregnancy planning and the group's significantly younger (<0.001) mean age. Both the Healthy and Medically Managed groups were similar (within 1.2% of each other) and had lower frequencies (yet were 79.5–83.6%) than the Diagnosed Ill group for considering how well results predict disease and agreement that what they (will) learn from genome testing can help reduce the risk of getting sick. This again seemed to depict a tendency for the DI group to be interested in their disease state, treatment and avoiding illness.

Post-testing: The direct question of whether genome testing was valuable scored highest across all groups, with no statistically significant difference. All groups also indicated all values-related items as important; however, groups demonstrated

statistically significant differences by t-tests involving genomic information as influencing future health management and using it to improve health.

In summary, the study findings support personal health values as relevant to pursuers of genome testing, and genomic information as having personal health value. Despite this uniformity, the distinctive health status groups created in this study allowed identification of statistically significant differences on noted values items based on perceived health status (where groups with medical diagnoses differed). Although results indicate genomic results have value, without further qualitative study, there is no way to confirm assumed meaning or associated personal translation into health actions.

### **Study Issues and Limitations**

The two surveys used for this study were based on validated instruments and measures with demonstrated reliability, yet issues that limited results or interpretation were the following: 1) the surveys were lengthy, potentially explaining the attrition between the first and second surveys; 2) perceived redundancy or complexity in certain sections, e.g., the second set of twelve risk related conditions with a less than 40% response rate; however, it was apparent that some participants neglected to answer items when their perceived risk was not increased; 3) potential overlapping answer options, e.g., as encountered by persons without children, when offered both not applicable and not interested regarding interest in testing for children; 4) confusing terminology or phraseology—either technical, colloquial or with more than one possible interpretation, e.g., “genomic information can help reduce the risk of getting sick” and “learning about



‘traits’”; and, 5) interpretation of multiple choice or text answers by limited analysis or assumed meaning, i.e., curiosity had different meanings.

Study specific issues arose. Both surveys included questions relevant to examining this study’s subjective concepts (i.e., perceived health status, risk perception and health values); however, these items were not repeated thus precluding pre- and post-testing comparison. The study design did not have a control population; all study participants had pursued genome testing. Data analysis was limited in that most variables were categorical and postulated to be in an interdependent relationship (as opposed to independent/dependent). Health status groups, created by dichotomizing perceived health status and existence of a medical diagnosis, may have restricted numbers in the Health in Question group (that was too small to include in analysis).

Despite these considerations, the unique nature of this naturally occurring population of persons who independently pursued genome testing, without involvement of their healthcare providers or researchers, allowed examination of subjective health-related factors as potential influences on self-initiated individual health actions (i.e., DTC genome testing).

## **Conclusions**

Perceived health, alone or in combination with medical diagnostic status, is able to distinguish or is correlated with overall personal health status, risk perception and health values. This knowledge may facilitate integration of individual health perception and health values into healthcare in order to collaboratively assess, prioritize and provide personalized, efficient and cost-effective healthcare services. This approach is

compatible with the Health Capability framework (basis for this study) and other healthcare models that recognize individual health values, ability for personal choices and responsible support of independent consumer actions including genomic testing on behalf of their health needs and goals.

Existing perceived health status research is primarily structured from an illness or biomedical framework, with outcomes measures such as delay in mortality, reduced morbidity, improved functional status or cost/benefit ratios. Little research exists involving healthy persons (except as controls) who seek health promotion services<sup>21,22,50,51</sup>. These persons may be viewed as the worried well and/or as draining limited healthcare resources<sup>19</sup>. On the other hand, consistent with public education efforts promoting healthy behaviors, the value of screening and recognizing risk factors, these people may represent persons interested in maintaining and promoting health and/or having legitimate health problems that do not meet diagnostic criteria.

Perceived health status is integral to overall health status. It may differentially influence healthy persons, as opposed to persons as they age or health declines when biomedical status may become more urgent and motivate action. This reasoning could also support health-related actions of relatively healthy persons as they face acute illness or anticipated health risk. This study population included 20% healthy persons, most of who would not otherwise have come to the attention of the healthcare system. What motivates these persons, as well as the other 80% with medical diagnoses, was explored in the context of personal health perception, values and self-initiated health action vis-à-vis genomic testing in hopes of informing gaps in the current healthcare system.

Perhaps we are not seeing some worthwhile outcomes (e.g., health behavior change) because we are focused on the quantitative outcomes (delay of mortality, lesser morbidity or measurable functional status) rather than the goals and values of the individuals and the behaviors they deem worthwhile. Perhaps we could get better outcomes by measuring an individual's valued outcomes, and at the same time make more efficient use of human and economic healthcare resources<sup>52</sup>.

Despite study limitations, genomic information is supported as both an individual health value and as having perceived value for improved individual perspective on personal health status, perceived control of health, ability to decrease the risk of getting sick and the ability to improve health and influence future health management. Research attests to the fact that users of DTC genome testing desire to discuss and share test results with their healthcare providers<sup>11,17</sup>. So why do we not take advantage of this interest to work collaboratively in partnership with our patients and the healthy public in decisions about genome testing? Why do we indirectly support and promote individual independent pursuit of this testing?

The underlying message of this study's findings is that practitioners and patients could likely benefit from integrating three simple questions into routine clinical practice. These questions are the following: 1) "In general, how would you rate your health?" Then ask, "Why?" 2) "What is your greatest health concern, need or value?" And, 3) "What is your health goal?" From this, the practitioner can collaboratively determine an effective and practical plan. As far as genomics, that would be the next question, as genomic screening for risk becomes part of routine healthcare practices. Personalized

healthcare integrating genome testing for everyone is on the horizon whether that is via the DTC route, an indirect path or new approaches in conjunction with healthcare providers.

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## CHAPTER VI

### EXTENDING THE HEALTH CAPABILITY CONCEPTUAL FRAMEWORK: INDIVIDUAL PERCEIVED HEALTH STATUS AND HEALTH VALUES

#### **Introduction**

The main research question addressed the motivations for individual independent pursuit of genome testing. The hypotheses tested whether individual perceived health status (including perceived risk) and personal health values were influencing factors. Chapter II provided the literature review and discussion of these concepts. Health Capability, the conceptual framework, provided the basis for the proposed concept mapping and relationships. The rationale and relevance of this model for this research was presented in Chapter III. The major tenets of this framework included individual health values, goals and health agency (supporting individual choice), the existence of internal and external influencing factors consistent with research concepts and variables, and its adaptability for extension to prevention and health promotion for DTC genome testing. Perceived health status, the key concept/construct for this research, was addressed in Chapter IV in a systematic review of the literature. Also included in this literature review was a discussion of instruments and approaches used to capture and measure this concept/construct. Chapter V presented the research study, methodology, results and discussion, limitations and identified areas of need for future research and clinical applications.

Chapter VI will present the impact of the study findings related to research hypotheses and enhancing and modifying the proposed Health Capability concept

mapping. Plans for additional research to clarify and characterize components of perceived health status will also be presented. Finally, based on the outcomes of this study, the need for future research will be presented.

## **Background**

This dissertation research study examined health status, dichotomized by perceived health status and a biomedical view of health status (existence of a medical diagnosis), in DTC genome testing users. The primary hypothesis was that discrepancies exist between the individual and biomedical view of health status. This hypothesis was supported by the results of categorizing participants into health status groups that confirmed the existence of the proposed four mutually exclusive health status groups. These groups included the two discrepant groups, specifically persons with one or more medical diagnoses who rated their health as good to excellent, and healthy persons who rated their health as poor or fair. This latter group supported existence of the “worried well” (Garfield, 2006). A healthy group was also identified, as persons without a medical diagnosis and rating their self-health as good to excellent. Each of these groups raises questions as to differences in the reasons (motivations) for pursuit of genome testing, health values and distinguishing characteristics. Findings also have relevance for prioritization in clinical practice, individual health behaviors, cost-effective healthcare services and research.

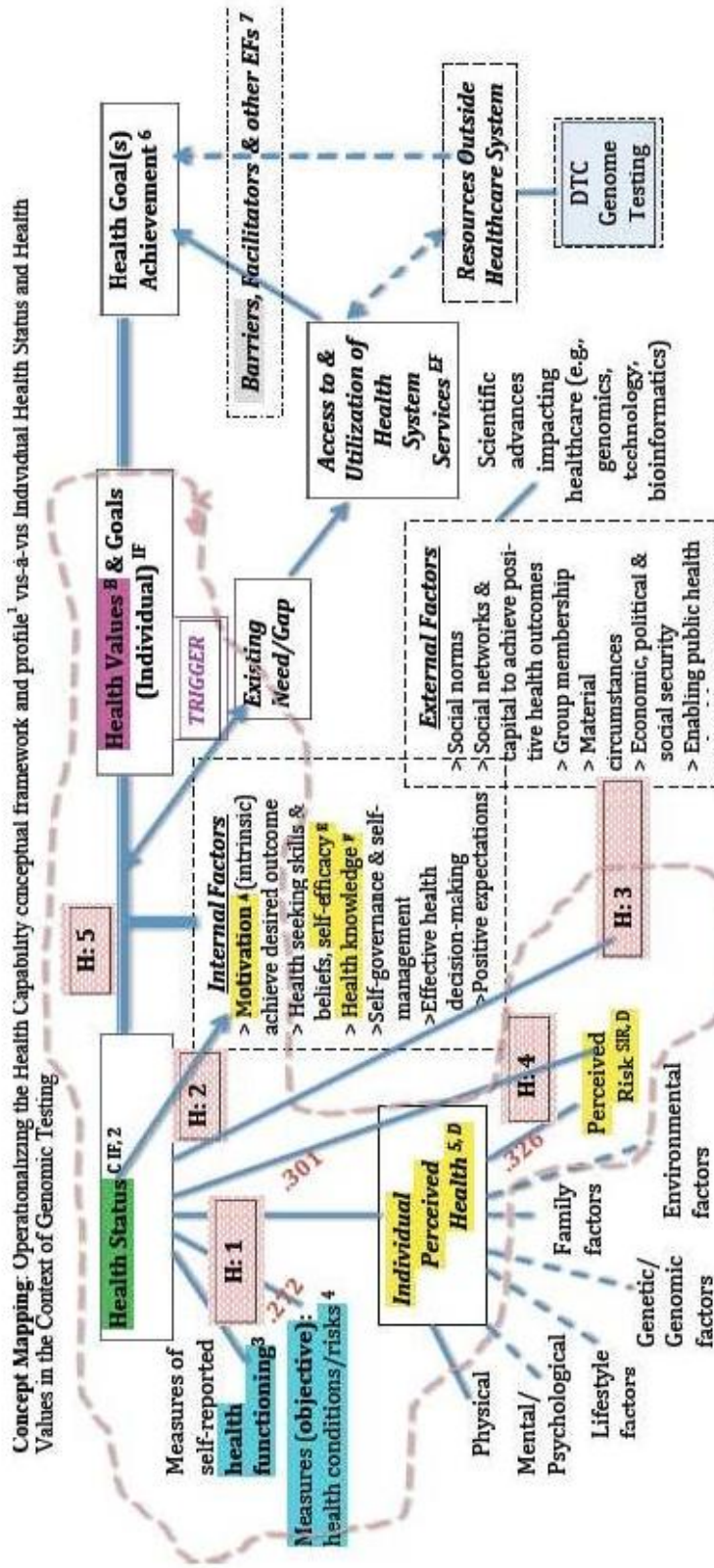
The Health Capability model (Ruger, 2010) was used as the conceptual framework for this study. This is a relatively young paradigm without an operational model or measurement instrument and little applied clinical research. The unique

contribution and potential for the Health Capability framework is its commitment to individual health values, support for individual health goal achievement, research at the individual level with extension from there to the population level, and its adaptability for application to prevention and independent individual action in pursuit of health goals (i.e., genome testing).

### **Health Capability: Proposed Concept Mapping—Revised**

In Chapter II, concept mapping was proposed to operationalize Health Capability (Ruger, 2010) for this study and evaluate its use for this dissertation research. It was also intended that this model be applicable to other potential clinical and research use in the context of a theoretical framework committed to physician (practitioner)—patient (individual) partnership, recognition of individual health values and goals, and support for prevention at the individual as well as public health levels. The original concept mapping has been updated and revised based on the results of the dissertation research (see Figure 6.1, including Pearson correlation values and highlighting of study concepts as discussed below). Although the research hypotheses did not include one specific to Health Capability, the fact that this model served as the framework for study concepts, for proposed and tested relationships, and for adaptation for prevention and health promotion vis-à-vis DTC genome testing, coupled with the study findings (presented in Chapter Five and discussed further in this chapter), is consistent with initial assessment and support for its use and further study in DTC genome testing research.

Figure 6.1: Concept Mapping: Operationalizing the Health Capability conceptual framework



**KEY: Relationships:** Arrow = dependent/independent; Lines: Double = interrelated (non-dependent), Single = concept/factor components; Dashed = unique/unknown; H = Research Question/Hypothesis focus; H: 3 cannot be depicted simply: it is the relationship between socio-demographic factors & health status

<sup>1</sup> (Ruger, 2010)

<sup>2</sup> Health status & health functioning can be viewed from two perspectives – as assessed & measured per the healthcare system, but also as self-perceived by the individual

<sup>3</sup> Measures per healthcare system instruments/methods (e.g., SF-36, mental & physical functioning)

<sup>4</sup> Measures per healthcare system assessments: biomedical markers, diagnoses & risk factors (behavioral & lifestyle)

<sup>5</sup> Measure added per this author as dimension of health status: interrelated with health values & goals, and influencing individual potential needs or gaps to health goal achievement

<sup>6</sup> Requires principles/constructs of Health Agency (individual ability to achieve valued health goals & act as own agent) & Health Functioning (relates to outcome of action for health)

<sup>7</sup> Barriers exist across all direct and indirect paths to Health Goal Achievement

<sup>8</sup> Self-interpreted/reported measures of perceived risk (including <sup>2</sup> & <sup>4</sup> & personally derived from any source); # Internal factor; # External factor; # Concepts in Literature Review.

## Health Status

This concept previously included “health functioning” in its title since both terms were included in the Health Capability profile (Ruger, 2010). However, health functioning reflects only limited measures of health status, usually in relation to disability or the ability to perform activities of daily living (Lachman & Agrigoroaei, 2010; Schiffman, Jacobsen, & Whitcup, 2001). Thus, Health Status (green highlight) was modified to encompass three measures of health (see Figure 6.1): the two components (blue highlight) specified in the Health Capability profile (Ruger, 2010), plus the addition of perceived health status (yellow highlight).

1. **Functional**: Maintained as a component of health status, including its measurement (e.g., by the SF-36 (Ware, 2009; Ware & Sherbourne, 1992)).
2. **Health Conditions and Risks (objective)**: Maintained as a component of health status, including its measures (e.g., vital signs, EKG, CBC, screening, etc.). The term objective is included in order to distinguish this from the individual (subjective, perceived health) component. The Pearson correlation coefficient (.272), based on results of this research, has been added to the relationship line between Health Status (the construct, represented by the research Health Status groups) and the biomedical measure of health status (existence of a medical diagnosis). Although the correlation analysis was found to be statistically significant, the large study population (N = 1455) may have contributed to inflating its apparent impact since the correlation value itself (.272) is considered only weakly positive.
3. **Individual Perceived Health (subjective)**: This component was added as part of the proposed concept mapping prior to the completion of the dissertation research. It is consistent with Health Capability’s philosophical recognition of an individual’s view and values for their health, as well as its respect for an individual’s assessment of their health as evidenced by inclusion of measures of self-reported health functioning. However, measures of functional health status and biomedical health status cannot capture the totality of an individual’s perceived health, what is of personal relevance and value, and what impacts actions and outcomes. Studies of individual perceived health

have demonstrated its significant and unique contributions in outcome studies of mortality and morbidity (Benyamini & Idler, 1999; Idler & Benyamini, 1997). However, there is little research from a prevention focus, and the limited research that exists is population and disease/illness oriented (Chern, Wan, & Pyles, 2000). This was discussed in the review of literature in Chapter Four. Assessment of health status from the individual perspective, and awareness of personal health values, has been recommended as an integral component for routine healthcare practice essential to the ability to evaluate meaningful outcomes (Barr, 1995). Thus, based on the literature review, previous studies and the current study results that support the distinct nature of individual perceived health status (yellow), and its association with the overall construct of Health Status (green), the relationship line between them was changed to solid (from dashed) and the relevant correlation values were added.

### **Perceived Health Status (Hypothesis 1)**

Perceived health status, the subject of literature review as presented and discussed in Chapter IV, was captured, measured and validated in research either by a single Likert-type item or a measurement instrument (Benyamini & Idler, 1999; Idler & Benyamini, 1997; Leung, Luo, So, & Quan, 2007). An instrument to assess perceived and targeted health status is frequently used in studies involving disease, mortality, morbidity, disability or interventions (Bardsley, Astell, McCallum, & Home, 1993; Salaffi, Stancati, & Carotti, 2002). The single item is preferred for brevity and where an overall perspective of perceived health status will achieve the study's need or intent. Based on the PGen survey and database, perceived health was represented by a single item in the dissertation research. It was used to create Health Status groups combining both the subjective individual perception of health with an objective biomedical perspective of health (existence of medical diagnosis). Hypothesis 1, that discrepancies existed between these two perspectives, was confirmed by identification of participants within all four



health status categories. This result supported the integration of perceived health status as a separate, contributory subdomain of Health Status.

### **Perceived Health Status: Proposed Components**

Perceived health status was proposed to be comprised of seven subdomains (components; see Figure 6.1) based on existing genetics, genomics and perceived risk research and literature (Cameron, Sherman, Marteau, & Brown, 2009; Carere et al., in press; R. Green & Roberts, 2012b; Heshka et al., 2008; D. J. Kaufman et al., 2012; Orom et al., 2012; Valdez, Yoon, Qureshi, Green, & Khoury, 2010). The subdomain of primary focus, in conjunction with the examination of individual health status and personal health values, was perceived risk. This component is discussed below, as are variables relating to limited components of other perceived health status subdomains, together with the implications based on the study findings. These subdomains are:

1. Physical Factors: Discussed below
2. Mental/Psychological Factors: Relate to mental, emotional or psychological health status, including any existing medical condition that the individual views as compromising to his/her health
3. Familial Factors: Relates to health or risk status based on ancestry, ethnicity, race, condition or potential medical problem that the individual views as impacting (or potentially) his/her health risk (e.g., a condition that tends to run in the family)
4. Genetic Factors: Relates to existence (per individual report) of a known specific genetic disease, mutation, or genomic risk associated with a mutation in oneself or one's relative
5. Environmental Factors: Includes exogenous risk factors over which the individual has little or no personal control, such as those existing in one's

place of employment, geographic area, and/or as reportedly prescribed or medically necessary (e.g., per one's healthcare provider)

6. Lifestyle Factors: Includes behavioral factors over which the individual has personal control, such as smoking, alcohol consumption, use of illegal, non-prescription or self-initiated drugs (e.g., not prescribed by one's healthcare provider)
7. Perceived Risk Factors: Discussed below

Physical factors: In the proposed concept mapping, physical factors related to physical health status, including any existing medical condition that the individual viewed as impacting or compromising to his/her health. This subdomain is incorporated in all three measures of health status (functional, biomedical and perceived). The focus discussed here is limited to assessments and comments in the context of perceived health status. Physical health is known to impact one's perceived health, yet physical health status is also related to biomedical health and existing diagnosis, and functional health status, both of which also contribute to quality of life considerations (Lachman & Agrigoroaei, 2010). This study was limited to the examination of biomedical health captured as the existence (or not) of one or more medical diagnoses, and was compared with and noted to have distinctions from, an individual's perceived health status. To what extent perception of physical health status contributed to one's overall perceived personal health status is not known and could not be characterized/quantified by this study. However, given the finding that the health status groups with medical diagnoses had significantly different (higher) perceived risks (number of conditions at increased risk), and that previous research supported the importance of functional health (Haseen, Adhikari, & Soonthorndhada, 2010; Lachman & Agrigoroaei, 2010), it may be that

perceived physical health is more relevant when it is perceived as compromised or urgent (as opposed to when one is healthy) (Mann, de Ridder, & Fujita, 2013). In this dissertation, related to Hypothesis 3, age was examined in association with health status groups, and separately, with individual perceived health status. Statistically significant differences between health status groups supported rejection of the null Hypothesis 3 that there would be no difference. However, significant differences were limited to combinations with the Healthy group, whose mean age was 37.5, in contrast to 51.2 and 50.1 in the Diagnosed Ill and Medically Managed groups, respectively. These results are consistent with other studies that demonstrate conflicting results related to age and pursuit of DTC genome testing (Bloss et al., 2010; Cherkas et al., 2010). It is also noteworthy that in the overall study population, individual age was not correlated with perceived health status. This latter finding lent support to the position that physical health status may be sufficiently addressed by the combination of functional and biomedical health status measures. Further, if one's physical health is not compromised or of concern, it may be that its influence or contribution to perceived health status essentially becomes negligible. This would be relevant for future research to understand individual motivations for health actions.

Perceived Risk (Hypothesis 4): Based on the previous literature review and studies incorporating perceived risk (or perception of risk) related to genomics (Acheson et al., 2010; Heshka et al., 2008; D. J. Kaufman et al., 2012; McBride et al., 2009; Sjoberg, 2000; Tarr et al., 2014) performed to develop Health Capability, the concept was defined as individual recognition or appreciation of real or believed vulnerability,

compromised health status and/or likelihood of a disease(s), disorder(s) or condition(s) (specific or in general), as either increased relative to others, or existing to an extent beyond that acceptable to that individual. This definition reflects the individual's subjective perspective and values, consistent with Health Capability and the nature of survey data used in this dissertation research. Perceived risk was represented in the dissertation on an individual level as the sum of perceived risks (likelihood to develop) for twenty-four survey-specified conditions. Rejection of the null Hypothesis 4 that projected no significant difference between health status groups was supported by the finding of significant differences between health status groups by t-tests. Separate correlation analyses of perceived risk identified the highest correlation (.326) with perceived health status, the lowest (.272) with biomedical health status, and a middle value (.301) with the combined health status groups. These results supported a relationship between perceived risk and individual health status, both from objective criteria and subjective individual perspectives, warranting inclusion of both as relevant to individual health status and assessment.

Research studies have examined and confirmed perceived risk as an interest, reason or motivator for individual pursuit of DTC genetic or genomic testing (Goldsmith et al., 2012; Gollust et al., 2011; David Kaufman, Bollinger, Devaney, & Scott, 2010). Another study determined that persons who pursued DTC genome testing were most interested in the condition or diagnosis they had (Meisel et al., 2013). However, perceived risk has not been studied as to its relative contribution among other personal (subjective) motivators for pursuit of DTC genome testing, or as to its association with

one's combined perceived and medical diagnostic health status (as supported in the current study). This study found perceived risk to be among the highest for the same medical conditions (i.e., heart disease, diabetes, skin cancer, breast cancer, prostate cancer, and high cholesterol), across all health status groups, ranging lowest in the Healthy group, to intermediate in the Medically Managed group, to highest in the Diagnosed Ill group. These conditions are consistent with risk concerns identified in prior DTC genome testing studies [e.g., heart disease (Gollust et al., 2011)], and accounted for risk concerns in a minimum of 13%, and a maximum of 48%, of persons within the health status groups. Whether or not concern about a single condition, or a collective sum of perceived risks, is a significant contributor to pursuit of DTC genome testing remains uncertain. Furthermore, this issue raises the question of a threshold of tolerance for risk or need for information, that influences health-related action (i.e., DTC genome testing). Studies are needed to address these potential influences, as well as to explore the possibility of triggering events, needs or perceptions.

An important realization that derived from this dissertation process and the outcomes of the research related to perceived risk versus perceived health. It is noteworthy that, with the exception of perceived risk, the sub-domains of individual perceived health proposed in the concept mapping all related to both perceived health and to perceived risk (components as identified by the literature review). However, since the correlation between them is at most a moderate positive (.326), they are neither mutually exclusive nor would these results suggest the use of one as more important or as a surrogate for the other. This is an important finding relevant to appreciating the gaps and

potential biases in the healthcare literature, research and clinical practice. A primarily risk focus reflects a disease/illness model and minimization or lack of health related freedoms and choices if one is deemed healthy (i.e., not sick) (Boenink & van der Burg, 2010). Perceived health is typically not the focus or included in research. If it is included, it is generally disease focused or captured by a single question, reported as general frequencies with little other application (Barr, 1995; Carere et al., in press; Lichtenstein & Thomas 1987; Miilunpalo et al., 1997). Thus, not only does this indicate a gap in research and understanding of the potential impact or influence of perceived health status, but it also raises awareness and questions related to the evaluation of and healthcare priorities for healthy, or seemingly healthy, persons. This includes the need for further study of the “worried well”, and the extent of their numbers that may be obscured within other health status categories or non-responses. It also includes the goals and values of healthy persons, with the potential for adjusting healthcare priorities, allocation and type of services, and contributing to resource-effective health promotion. These issues were discussed in Chapter V.

### **Reasons for Pursuit of Genome Testing and Socio-demographics** (Hypotheses 2 & 3)

The reasons for pursuit of genome testing, associated with Hypothesis 2, interrelate with health values as well as health goals, and are addressed in the Discussion section. Socio-demographic characteristics, and support for or rejection of Hypothesis 3, are considered as relevant in the following sections.

## **Health Values** (Hypothesis 5)

Health values, addressed in the review of literature included in Chapter II, were operationalized in the dissertation research using PGen survey Likert-type questions (from both the pre-testing and the two week post-testing surveys) relating to items of individual health importance, interest, consideration and/or value. The study results confirmed an association between Health Values and Health Status groups. Two of the five health values items (importance of genome results for future health management, and for health improvement) were significantly different by t-tests between health status groups. This finding supports rejection of the null Hypothesis 5 that there would be no difference in health value of genomic information between health status groups. The results indicate that there is a distinctive contribution from (subjective) perceived health as opposed to the (objective) existence of a medical diagnosis (i.e., if unsupported, all persons with a medical diagnosis, or all persons with perceived good health, would not be significantly different). On the other hand, the fact that all health status groups: 1) were not significantly different on three of the five items; 2) all rated all items as having at least some value/importance (means ranging from 3.46 to 4.16, with 3.0 being neutral and 5.0 being strongly agree); and, 3) uniformly rated the highest for genome results as being valuable to their health [consistent with previous research (McBride et al., 2009)], supported the relevance of health values to pursuit of genome testing, and potentially to other self-initiated health behaviors. The comparison of pre-testing expected health value of genome test results with post-testing individual value based on actual results was not possible since the same measures were not repeated. However, the findings do contribute

to supporting individual health values as a primary component of the proposed operationalization of Health Capability, and its applicability to DTC genome testing research.

Future study of the combined contributions and effects of individual perceived health and health values on individual health goals and behaviors is supported by the current results, as well as other research (Shmueli, 1999). A future study will examine an individual's perceived health status combined with health values (i.e., the items that differentiated health status groups in the current study), in relation to an individual's intended behavior change as stated at two weeks post testing compared with their stated health behaviors as reported at six months.

### **Study Limitations**

One limitation of this study is that the correlation coefficient between overall Health Status and Individual Perceived Health was based solely on the health status groups for the former, and the limited measure of perceived risk for the latter. Despite this limitation, a relationship was demonstrated (see Figure 6.1; the line between them has been changed to solid & non-directional).

Limitations also exist related to the fact that this study involves secondary analysis of data from the original PGen research surveys. Multiple-choice questions were framed from, and/or provided answer options supported by, results of previous DTC genome testing research studies; however, that approach did not allow for other possible individual responses. Another limitation in the current study was the inability to evaluate individual health values pre-testing since that concept and related questions were not



included in the pre-testing survey. Despite these limitations, the PGen survey questions and data did include questions in which relevant individual answers and qualitative data could be captured. Targeted questions with text answers were queried for keywords that confirmed, on a preliminary basis, the existence of different individual perspectives related to reasons for testing and interpretation of terminology used (e.g., curiosity). These subjective text answers coupled with the ability for a mixed qualitative and quantitative approach to data analysis provided a unique dimension not included in existing research (Bloss et al., 2010; Goldsmith et al., 2012).

Generalizability was also a relevant issue from two perspectives. The current study population, participants in the PGen study, demonstrated socio-demographic characteristics consistent with previous DTC genome testing users research (Bloss et al., 2010; Goldsmith et al., 2012; McGuire et al., 2009). Thus, study results are likely generalizable to other DTC genome testing users. On the other hand, this study population, consistent with the previously cited studies, is different from, and is not generalizable to, the general population. Specifically, these cited studies of DTC users have found that participants tend to have higher levels of education; have higher incomes; be Caucasian; be professionals, scientists, in academia or employed in mid to upper level jobs; and indicate their perceived health as good. Importantly, however, as the cost of DTC genome testing decreases, public awareness increases, marketing and new genome related testing services are offered (e.g., prenatal sex typing), NIH funded research exists for new approaches to early risk identification and/or health promotion using genomic screening (e.g., in newborns), FDA regulatory authority has gaps in controls of DTC

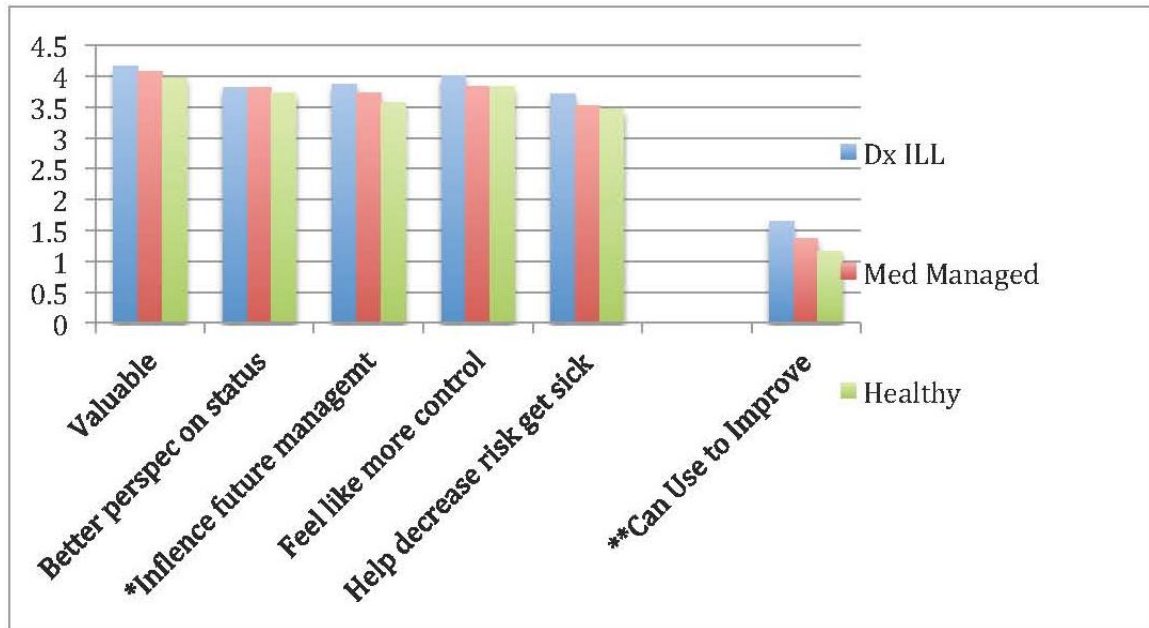
laboratory tests, and/or genome testing (or screening) further demonstrates clinical utility, there could be a significant increase in the number of individuals pursuing genome testing, as well as the socio-demographics of DTC testing users.

### **Discussion and Conclusions**

Perceived health status of the DTC genome testing users in this study spanned all five Likert-scale choices from excellent to poor, with the majority (1,235/1,455) rating their health as good to excellent, regardless of medical diagnostic status. However, by sorting the study population into four mutually exclusive health status groups based on inclusion of perceived health status, distinguishing characteristics emerged. Persons with a biomedical diagnosis and perceived poor or fair health expressed higher levels of interest for genomic information of immediate personal relevance, or for their family, i.e., about their current medical condition, other health risks and response type to different medications. In contrast, study respondents indicating their health as good to excellent, whether or not they had a medical diagnosis, expressed greater interest in genomic information related to future health, risks and ability for prevention (although also interested in the previously noted health information). Thus, perceived health status was associated with the type of genomic information sought, the percent indicating an item as important, and interest in immediate versus future use.

Health values were supported as important to the DTC genome testing study participants. All three health status groups assigned the highest scores to genomic information as being valuable to health (see Figure 6.2). In addition, group means on all other health values items were in the direction of having value, with some items

**Figure 6.2: Genome Test Results—Value To Respondents’ Health.**



KEY: \*\*Statistical significance. Set of bars at right is based on maximum of 3.

extending into the range of extremely important/valuable. Three observations are noteworthy. The highest mean scores for all health values items occurred in the Diagnosed Ill group. This finding suggests that persons with medical diagnoses and self-rated fair or poor health may perceive the greatest potential for genomic information to offer some help for their current health. Second, all groups scored similarly (high) related to genomic information as providing a better perspective on their health status and for feeling like they had more control of their health. This supports that DTC genome testing users tend to value an active role in health related decisions and ability to take charge of their health. Third, all respondents indicated, in a positive direction, that genomic information can be used to improve their health. Whether or not that is accurate

in the context of clinical utility, it is seen as valuable for personal utility. This information has direct relevance to healthcare providers, prioritization of services, appreciation for what is meaningful to consumers, targeting health education (and needs) and collaborative decision-making.

This study's survey requested participants to indicate the importance of twelve reasons for pursuit of DTC genome testing, based on motivations identified by previous research as discussed in Chapter II. Some of these reasons overlap into the realm of individual health values and health goals. For example, close to 100% of respondents in all health status groups indicated that their pursuit of genome testing considered obtaining (the goal of) personal health information (see Table 5.2, Chapter V). This goal is further supported by the relatively high percent among all groups indicating the desire for genomic information about their response type to medications.

The research Hypothesis 2 (null), stating that there would be no difference between health status groups related to reasons for pursuit for genome testing, was rejected for some reasons (e.g., finding out response type to different medications) and was supported for others (e.g., curiosity, and family members using personal genomics). An important consideration, however, is that on several reasons for pursuit of genome testing (e.g., genomic information related to personal risk for disease, for future children and the desire to improve health) over two-thirds of persons in all groups scored the reasons as of interest. These findings not only indicate an explanation for apparent non-significant differences between groups (since the majority in all groups were interested), but suggest that DTC genome testing users are interested in goals associated with

prevention and health promotion. It is also relevant to note that although more than 90% in each group responded that curiosity was involved, with that reason potentially viewed as having no clinically relevant health value or goal (and similar regarding pursuit of testing as fun and entertaining), an initial review of text answers of reasons for pursuit of genome testing attributed very different uses or meanings to curiosity. This study finding highlights a dimension important to clinicians and researchers concerning the structure, content and selection of survey questions, but also their interpretations.

The results of this study demonstrated that perceived health status distinguished health status groups and made a significant difference in individual reasons for seeking DTC genome testing, goals for genomic information and in risk perception. Perceived health status served as a complement to, and at times evidenced a higher correlation, than biomedical health status (e.g., for perceived risk status). The importance of individual health values was also demonstrated in self-initiated genome testing users. These findings lend support for use and testing of the Health Capability conceptual framework as a model for integrating individual health values and goals into healthcare services, delivery and policy directions. This study's results contribute to furthering understanding of the influence of perceived health status and its potential for application in the context of early identification of risk, prevention and health promotion. The modified concept mapping provides a basis for further study of individually relevant health concepts and factors that may influence health behaviors, such as self-initiated health action (e.g., DTC genome testing, or exercise program). It also facilitates the ability to test for existence of triggers, a threshold effect, or factors impacting readiness for health information and

commitment to healthy behaviors. This knowledge has the potential for more effective strategies for provision of healthcare and use of its limited resources.

### **Future Research**

The findings of this study indicate the need for research in three areas. First, there is a need for research and strategies spanning beyond traditional models of healthcare to incorporate prevention, as well as integrating subjective individual perceived health, personal health values and goals, and motivating factors impacting adherence to health behaviors (Breckenridge & Eichler, 2013; Mann et al., 2013; Ruger, 2010). This requires a theoretical or conceptual healthcare framework, such as Health Capability, that philosophically and practically integrates these principles. The model requires a systems perspective, yet commitment to the individual, to facilitate and recognize contributions and perspectives from all stakeholders while respecting cost-effective and health promotion oriented healthcare (J. P. Sturmberg et al., 2012).

Second, based on the established ability to represent, measure and integrate perceived health status by a single Likert-type subjective item, it is reasonable to consider the possibility of a similar approach for perceived risk. Although typically perceived risk is posed in the context of specific disease or risks, one or two overall measures of perceived risk may serve as an effective screening or triaging measure. For example, asking the patient/respondent what, in general, they consider their level of health risk to be as compared to others of similar age and sex, akin to previously posed survey questions (R. Green & Roberts, 2012a; D. J. Kaufman et al., 2012). Secondly, a question needs to be posed for the individual to consider the condition for which they feel they are

at greatest risk, then rating that by a Likert-type item or numeric measure. Working from the focus of this dissertation, the next step would be to combine these two (or three) values to represent perceived health status. In combination with, and contrast to medical health status, these groups and variables could be examined related to the potential impact on self-initiated health action and behavior. This approach has two immediately relevant benefits. Clinically, there is the potential to efficiently identify the existence of some condition of high-risk concern to the individual, with the possibility to intervene in a more timely and effective manner. From a research context, this approach may provide a method to test and compare the separate contributions of other subjective personal health/risk factors versus a combined measure.

Third, in addition to the constructs, concepts and relationships proposed, examined and modified, there remains an over-riding question relevant to healthy persons, whether they are persons without a medical diagnosis or they are persons with a medical diagnosis but with perceived good to excellent health. That question is “Why now?” Why have these persons, spanning significantly different ages, diagnostic status and income levels chosen to pursue self-initiated genome testing when they did? This author is proposing, and adding to the concept mapping, a “triggering” event (or threshold effect). This would be something subjective, beyond the combined interaction of medical health status, perceived health status, and individual health values and goals—that provides the impetus for action in healthy persons, or apparently non-acute or not medically necessitated situations. This dimension of perceived health status is the intended direction of future research. These events are proposed to include factors such

as the individual's age (e.g., being the same age as the parent when diagnosed), exposure to a perceived health risk (e.g., with potential concern for cancer), change in health status (i.e., newly diagnosed medical condition), and other personal factors of which we likely do not know what we do not know (Costello, 2014). In addition, concepts from other disciplines may be relevant to incorporate into an interdisciplinary approach to research, such as self-regulation and self-realization (Mann et al., 2013; Miquelon & Vallerand, 2008), in the context of motivating factors for self-initiated pursuit of health-related information, as with genome testing.

Further research is also planned to examine the other proposed subdomains (components) of perceived health (and/or risk), their association with and potential contribution to differentially influencing perceived health status, and if/how these factors are individually prioritized related to health values. These issues have relevance to individualizing, prioritizing and effecting quality and cost-effective healthcare services and policy, collaborating with consumers, and promoting health at individual and public health level.



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## APPENDICES

Appendix A

Letter of Permission for Use of Figure 3.1 (Chapter III):  
Conceptual Model of Health Capability



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## Appendix B

### PGen Data Distribution Agreement

PGen Data Distribution Agreement

Version date: September 12, 2013

#### **The Impact of Personal Genomics (PGen) Study**

#### **Data Distribution Agreement**

The undersigned parties hereby enter into this Data Distribution Agreement (DDA) as of the date specified on the final page hereof. For correspondences related to this agreement please contact:

Sarah Kalia  
[skalia@genetics.med.harvard.edu](mailto:skalia@genetics.med.harvard.edu)  
617-264-5881

#### **INTRODUCTION**

The NIH-funded Impact of Personal Genomics (PGen) Study is a longitudinal survey of consumers of two personal genetic testing companies, 23andMe and Pathway Genomics, to gather empirical data about the characteristics of these consumers and the psychological, behavioral, and health impact associated with personal genetics services. Data may also include participants' personal genetic risk information, provided with participants' consent.

The undersigned parties entering into this DDA include the following:

- Recipient(s): the researcher(s) and institution or other entity receiving access to the PGen Study Data.
  - If the individual requesting data access is a student or trainee, he or she must identify a faculty mentor to be the responsible party. The student/trainee and faculty mentor must co-sign this document as Recipients.
- Joint Principal Investigator Dr. Robert C. Green at Brigham and Women's Hospital and Harvard Medical School, and Joint Principal Investigator Dr. J. Scott Roberts at the University of Michigan School of Public Health
- Co-Investigators from 23andMe and Pathway Genomics

To protect the confidentiality and privacy of PGen participants, Recipients granted access to study data must adhere to the requirements of this DDA. Failure to comply with this DDA could result in its termination and denial of further access to PGen Study Data, and may leave violators liable to legal action, for instance on the part of PGen participants, corporate research partners, or the U.S. Government.

#### **TERMS AND CONDITIONS**

**1. Data.** Data including but not limited to any and all information derived from survey materials, genetic risk information and any and all data derived from statistical analyses linking genetic data with other study data will be kept confidential and will not be shared with anyone other than the Recipient(s) authorized in writing. "Data" refers to any and all study data, either obtained directly from PGen participants or obtained from third parties as authorized by the participants, with oversight by the Partners Human Research Committee.

**2. Research Project.** This data will be used by the Recipient(s) only in the below Research Project (list name of Research Project and objective of research project below):

---

**2.1** If any aspect of the Research project e.g. data analysis is to be performed by any entity other than the Recipient(s) such entity is to be named below:

---

**3. Non-transferability.** The Data specified in this agreement should not be distributed and transferred to any other individuals or entities unspecified in this agreement regardless of its intended use. A separate agreement would need to be obtained and approved by both Principal Investigators and by at least one representative from each of the genetic testing companies, 23andMe and Pathway Genomics.

**4. Collaboration.** The Recipient agrees not to publish: any comparative data of 23andMe and Pathway Genomics resulting from the PGen Study, any company-specific data, or any data from which company-specific data could be inferred, without prior written permission from the Principal Investigators and Co-Investigators from both 23andMe and Pathway Genomics. The Recipient agrees not to make any public statements that compare the companies based on data that is derived from the PGen Study dataset.

**5. Review of Analyses.** At the discretion of the Principal Investigators, analyses of PGen Data performed by the Recipient(s) may be subject to review by the PGen Study team prior to submission for publication.

**6. Manuscript Review.** The Recipient agrees that any scientific publication of PGen Data will be reviewed and approved in writing by the Principal Investigators, and shared with Co-Investigators from 23andMe and Pathway Genomics for comment prior to submission for publication.

**7. Acknowledgements.** The Recipient must acknowledge the Data gathered by the PGen Study staff in any and all oral and written presentations, disclosures, and publications resulting from any and all analyses of Data, by including “for the PGen Study Group” and the PGen Study grant number **HG005092** at the end of the author list or in an Acknowledgements section.

**8. Anonymity.** The Recipient agrees that Data will not be used, either alone or in conjunction with any other information, in any effort to determine the individual identities of any of the participants from whom Data were obtained or derived.

**9. Security.** The Recipient should be aware of computer and data security, and must make sure that the information is protected with industry standard data protection practices.

**REQUIRED SIGNATURES**

\_\_\_\_\_  
Recipient Name & Title (Faculty only)

\_\_\_\_\_  
Date

\_\_\_\_\_  
Recipient Signature

*Carolyn D Farrell*

\_\_\_\_\_  
Student Co-signature (if applicable)

10/21/13

Robert C. Green, MD, MPH

\_\_\_\_\_  
Joint Principal Investigator Name & Title

\_\_\_\_\_  
Date

*Robert C Green*

\_\_\_\_\_  
Joint Principal Investigator Signature

10/21/13

J. Scott Roberts, PhD

\_\_\_\_\_  
Joint Principal Investigator Name & Title

\_\_\_\_\_  
Date

*J. Scott Roberts*

\_\_\_\_\_  
Joint Principal Investigator Signature

Joanna Mountain, Senior Director of Research  
23andMe Co-Investigator Name & Title

10/16/2013  
Date

*Joanna Mountain*

\_\_\_\_\_  
23andMe Co-Investigator Signature

Ben Thumander, Contractor  
23andMe Authorized Signatory Name & Title

10-16-2013  
Date

*Ben Thumander*

\_\_\_\_\_  
23andMe Authorized Signatory Signature

Approved as to Form  
23andMe Legal Dept.

By *JM*

James PLATJE, CEO  
Pathway Genomics Co-Investigator Name & Title

10-7-2013  
Date

*James Platje*  
\_\_\_\_\_  
Pathway Genomics Co-Investigator Signature

Appendix C

Institutional Review Board (IRB) Communication of Exemption



Carolyn Farrell <cmfarre@g.clemson.edu>

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**Carolyn Farrell's research study entitled "DTC Genomic Testing Users: Perspectives for Health Care Based on Individual Values, Perceptions of Risk, Risk Factors and Health Status"**

9 messages

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Laura Moll <LMOLL@clemson.edu>  
To: "cmfarre@g.clemson.edu" <cmfarre@g.clemson.edu>  
Cc: Bonnie Holaday <HOLADAY@clemson.edu>

Wed, Mar 12, 2014 at 4:54 PM

Dear Carolyn,

The Clemson University Office of Research Compliance (ORC) has determined that the project identified above **does not involve human subjects** as defined in the Federal regulations governing the protection of human subjects in research [45 CFR 46.102(f)] and is, therefore, **not subject to IRB review**.

As per our conversation this afternoon and , at this time, this project will not involve either "intervention or interaction" with living individuals, or the collection or use of "identifiable private information" about living individuals. Therefore, IRB review is not required.

Please contact this office again if there are any changes to this project that might bring it under the purview of the IRB. It is the responsibility of the ORC to determine whether any specific research project falls within the definition of research with human subjects, as provided by Federal regulations and institutional policy.

Thank you for contacting us to check on whether your project required IRB review and approval.

Good luck with this project and please feel free to contact me if you have any questions.

Best,

Laura :-)

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Laura A. Moll, M.A., CIP  
IRB Administrator  
Office of Research Compliance  
223 Brackett Hall  
Clemson University  
Clemson, SC 29634-5704

lmoll@clemson.edu  
Phone: 864-656-6460  
Fax: 864-656-4475

IRB email: [irb@clemson.edu](mailto:irb@clemson.edu)

[www.clemson.edu/research/compliance/irb/](http://www.clemson.edu/research/compliance/irb/)



SafeZone

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**Carolyn Farrell** <[cmfarre@g.clemson.edu](mailto:cmfarre@g.clemson.edu)>  
To: Laura Moll <[LMOLL@clemson.edu](mailto:LMOLL@clemson.edu)>  
Cc: Bonnie Holaday <[HOLADAY@clemson.edu](mailto:HOLADAY@clemson.edu)>

Wed, Mar 12, 2014 at 5:55 PM

Dear Ms. Moll (Laura)  
Thank for your time and expert assessment on behalf of the Clemson IRB office, and this written confirmation of the determination that IRB review is not indicated/necessary for my proposed PhD research.

I also appreciate your kind offer of future assistance.  
Best to you too,  
Carolyn

[Quoted text hidden]

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Carolyn D Farrell, PhD(c), MS, RN, WHNP-BC, CGC  
Clemson University, Healthcare Genetics PhD Program  
(716) 308-3592 (cell)

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**Bonnie Holaday** <[HOLADAY@clemson.edu](mailto:HOLADAY@clemson.edu)>  
To: Laura Moll <[LMOLL@clemson.edu](mailto:LMOLL@clemson.edu)>, "[cmfarre@g.clemson.edu](mailto:cmfarre@g.clemson.edu)" <[cmfarre@g.clemson.edu](mailto:cmfarre@g.clemson.edu)>

Wed, Mar 12, 2014 at 8:37 PM

Thanks Laura!

Bonnie

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**From:** Laura Moll  
**Sent:** Wednesday, March 12, 2014 4:55 PM  
**To:** [cmfarre@g.clemson.edu](mailto:cmfarre@g.clemson.edu)  
**Cc:** Bonnie Holaday  
**Subject:** Carolyn Farrell's research study entitled "DTC Genomic Testing Users: Perspectives for Health Care Based on Individual Values, Perceptions of Risk, Risk Factors and Health Status"

[Quoted text hidden]

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**Bonnie Holaday** <[HOLADAY@clemson.edu](mailto:HOLADAY@clemson.edu)>  
To: Carolyn Farrell <[cmfarre@g.clemson.edu](mailto:cmfarre@g.clemson.edu)>

Wed, Mar 12, 2014 at 8:38 PM

That was good news. State this in your methods section