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Paths to Tier 1 Genomics Implementation: A Survey of Chronic Disease Directors

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Abstract

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MPH, University of Connecticut, 2000

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Dissertation Submitted in Partial Fulfillment

of the Requirements for the Degree of

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Health Services

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Abstract

Although evidence is currently available for population-based genetic screening and testing of individuals and their family members for certain hereditary chronic disease conditions (Tier 1), few states have integrated these genomic applications into chronic disease prevention programs. State and territorial chronic disease directors (CDDs) could provide the leadership needed to deliver these applications in more states. The purpose of this study was to determine whether an association exists between current chronic disease genomics funding or specific state genomic activities and the level of knowledge and interests in genomics by these directors. Rogers's diffusion of innovations (DIT) theory was used to explain the current climate of state chronic disease genomics and the need for an innovation *champion* to promote these evidence-based applications both in and out of the state health departments. A nonexperimental, cross-sectional, correlational survey of CDDs ($N = 58$) was performed using the Chronic Disease Director's Survey and results were analyzed using chi-square, independent *t* test, ANOVA, logistic regression, and Pearson's correlation coefficient. Results showed CDDs knowledge of genomics is unrelated to current state funding; however, CDD knowledge and interest in genomics was associated with inclusion of genetics in cancer control and cardiovascular health action plans, Tier 1 condition education, privacy and nondiscrimination laws, Behavioral Risk Factor Surveillance System (BRFSS) genomics questions, and frequent collaborations with outside entities. These results provide clear ideas to increase CDDs knowledge and interest in chronic disease genomics and potentially impact Tier 1 genomics implementation in more states.

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Dedication

I would like to dedicate this dissertation to my family, specifically my husband, Peter, without whom I could not have accomplished this goal. This was *my* dream, something I knew I wanted to attain shortly after obtaining my master's degree and raised my hand for once I went back to work after raising my kids and felt stuck professionally; I knew I could do more. Living my dream, however, required sacrifice not only for me but for everyone who was close to me. Sacrifice came in the form of less time together, missed events, fewer finances, more chores, and having to deal with my stress level to complete this dissertation and degree. I couldn't have done it without all of them, including my kids, Patrick, Colleen, and Ana, who were always rooting me on and can now see what is possible. My example shows them that you're never too old to learn something new and positively use that knowledge to make the world a better place.

Acknowledgments

I would like to thank my chair, Dr. Ji Shen, for being the absolute best chair a student could have. I'm glad I hounded him for over two years to take me on. He was always there when I needed him and was able to back me off the ledge more than once when I was starting to get worried about progression or even if I had a clue about what I was doing. I would also like to thank Dr. Nicolette Alexander for her knowledge and expertise towards this project and for jumping into the middle of this; the transition was seamless. Furthermore, I would like to recognize Dr. Diana Naser, the Walden University Research Reviewer, for her diligent work on my study. I would also like to thank the APHA Genomics Forum policy subcommittee for allowing me access to the survey data used in this dissertation. Finally, I would like to thank all the mentors I've had throughout my life who fostered my love of science and my particular passion for genomics and its potential for better health of the human population.

I would be remiss if I didn't thank the one who actually makes this all happen. This isn't my doing, it's only by the will of God that I got through this and get through each and every day. I'm glad He's got my back.

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Chapter 1: Introduction to the Study

Introduction

The completion of the Human Genome Project in 2003 opened many doors for scientists and health professionals to identify and potentially prevent common disorders through gene analysis. Over the last decade, this progress has not only impacted clinical medicine and individual patients but has also shown the ability to reduce morbidity and mortality of susceptible populations through more personalized public health programming (Auffray et al., 2016; Cragun, Lewis, Camperlengo, & Pal, 2016). Unfortunately, translation and implementation of genomic advances has been slow, both in clinical medicine and public health (National Academies of Sciences, Engineering, 2016). Most research in this area has been focused on implementation in the clinic; however, evidence supports the use of genomic technology for population-wide chronic disease prevention. Therefore, gaining an understanding of the opportunities and challenges to public health genomic implementation is prudent at this time.

In this study, I conducted a quantitative survey of state and territorial chronic disease directors (CDDs) in the United States to examine what genomic activities are currently being achieved and determine if there is an association between these state activities and what these CDDs know or are interested about in chronic disease genomics. This study was important at this time because of recent evidence-based recommendations for screening at-risk individuals and their family members for hereditary forms of three chronic diseases; breast cancer, colon cancer, and cardiovascular disease. Identification of this group of individuals and subsequent treatment or preventative measures could

reduce the morbidity and mortality from these conditions and allow for positive social change through increased health and quality of life for those affected.

This introduction to the study will include background information leading to the current landscape, the purpose of this study and why it is important, and a description of the problem. I will also provide an explanation of the research questions and hypothesis, a description and justification of the theoretical framework, and outline the nature and significance of the study. Finally, I will define terms specific to this study and clarify the study's assumptions, scope, delimitations, and limitations.

Background

The Healthy People 2020 initiative has included genomic activities for the first time, signifying increasing evidence that family history and genetic testing can be used to promote health benefits in clinical and public health capacities (Valdez, Yoon, Qureshi, Green, & Khoury, 2010; Weir et al., 2015). State public health genomics activities have traditionally been focused on newborn screening (NBS); however, evidence and test availability has prompted recommendations for adult population screening initiatives (Green, Dotson, Bowen, Kolor, & Khoury, 2015). The expansion of public health genomics from newborn screening into chronic disease is important and timely considering the impact of new molecular technology and research advances in the field (Bowen, Kolor, Dotson, Ned, & Khoury, 2012). Pilot public health genomics programs in chronic disease have showed that advances can be made by conducting evaluations and examinations that support use of genomic information and family history for disease prevention efforts; integrating this information into existing programs; and developing

and circulating educational materials for health care providers, policy makers, and the public (St Pierre et al., 2014).

The last time a survey of state genetic activities was performed was in 2001 (Coalition of State Genetics Coordinators, 2002; Piper et al., 2001). Also at that time, Kaye et al. (2001) made very specific recommendations regarding the need for the integration of genetics into public health and how genomic activities were connected to the core functions of assessment, assurance, and policy development. These authors also provided the rationale for and details of responsibilities for a state genetics coordinator position in order manage activities and facilitate collaborations in genomics. Another analysis of the role of genetics in the provision of essential public health functions found that these programs provide for many public health obligations including diagnosing and investigating health problems and hazards in the community (NBS), mobilizing community partnerships with genetics professionals and other health care providers, and linking the population to needed personal health services (Wang & Watts, 2007).

Although these studies support the use of genomics as a public health tool outside of NBS, implementation beyond the pilot programs has been slow. In 2010, the Association of State and Territorial Health Officials (ASTHO) published a State Public Health Genomics Resource Guide outlining novel approaches for public health departments to translate genomic science into public health practice using examples from a limited collection of states with innovative programs (ASTHO, 2010). Although this toolbox was created to help other states find ways to integrate genomics into public health, the slow adoption requires investigation into why genomic services for chronic

diseases at the state level is not supported. A 2006 survey of state health officers confirmed important emerging public health functions; however, genomics was not one of them (Beitsch, Brooks, Grigg, & Menachemi, 2006). Moreover, a 2011 survey of chronic disease public health professionals about their training needs did not include any questions about genomics (Wilcox, Majestic, Ayele, Strasser, & Weaver, 2014). Until public health practitioners begin to think about genomics as a viable tool for public health prevention, implementation of state programs will likely not become a priority. This study was needed at this time to assess the current status of knowledge and interests in genomics by state CDDs and identify opportunities and challenges to increasing awareness of the importance of genomics by this group in light of the new chronic disease genomic testing recommendations.

Problem Statement

The Centers for Disease Control and Prevention (CDC) Office of Public Health Genomics (OPHG), the U.S. Preventive Services Task Force (USPSTF), and others have evaluated evidence and formulated recommendations for hereditary forms of chronic disease conditions that would benefit from patient genetic counseling, testing, and cascade screening of family members (Dotson et al., 2014). These applications are divided into a three-tier classification system with Tier 1 genomic applications having clear evidence for practical implementation (Khoury, Coates, & Evans, 2010). Initial Tier1 applications have been identified as hereditary breast and ovarian cancer (HBOC), Lynch syndrome (LS), and familial hypercholesterolemia (FH; Bowen et al., 2012).

Despite recommendations and evidence to support screening for these conditions, only a limited number of states are working in this area (Green et al., 2015).

Federal funding from the CDC OPHG and the Division of Cancer Prevention and Control (DCPC) to support state chronic disease genomics infrastructure development, surveillance activities, and implementation of evidence-based recommendations has been limited to a small number of states since 2003: Michigan, Minnesota, Oregon, Utah, Connecticut, Colorado, Georgia, and Ohio (CDC, 2016; Green et al., 2015; St Pierre et al., 2014). Furthermore, to date, most implementation strategies have been focused specifically to address initiatives in HBOC, less often on LS, and limited activity on FH. ((Laufman, Duquette, & Trepanier, 2012; Nordestgaard et al., 2013; Trivers, Rodriguez, Cox, Crane, & Duquette, 2015). Other states, such as Washington, Hawaii, Illinois, and New York, have made incremental strides in state genomics planning without CDC funding ((ASTHO, 2010; Trivers et al., 2015)).

The ability to utilize these evidence-based initiatives under the current climate could be problematic and negatively impact our public's health if citizens have limited access to these screening programs. Although studies have shown that chronic disease departments are hindered by poor collaborations, shifting goals, lack of organizational support, limited resources, alternating priorities, and competency by the public health workforce (Allen et al., 2013; Alongi, 2015), understanding and leadership by CDDs could also impact program implementation. Examining this group to evaluate their knowledge and interests in genomics is important in order to assess their ability to engage in these new technologies and support promotion of funding opportunities. Additionally,

establishing an association between certain current state genomic activities and the knowledge and interests of CDDs could provide the evidence needed for increased coordination and funding to the states.

Purpose of the Study

The purpose of this quantitative survey design was to determine whether there is an association between current state genomics funding or specific state genomic activities and the level of knowledge and interests in genomics by state CDDs. In this quantitative survey design study, I analyzed the results of a survey of CDDs in all U.S. states and territories. My intent with this study was to identify and describe particular activities or particular states that may be associated with an increased level of knowledge and interest in genomics by CDDs and which may also influence implementation of Tier 1 genetic tests at the state level.

Research Questions and Hypothesis

Research Question 1: To what extent, if any, is there an association between states that have received funding for chronic disease genomics and the level of knowledge and interests in genomics by state and territorial CDDs?

$H_0$1: There is no association between states that have received funding for chronic disease genomics and the level of knowledge and interests in genomics by state and territorial CDDs.

$H_1$1: There is an association between states that have received funding for chronic disease genomics and the level of knowledge and interests in genomics by state and territorial CDDs.

Research Question 2: To what extent, if any, is there an association between current state genomic activities and the level of knowledge and interests in genomics by state and territorial CDDs?

$H_0$2: There is no association between any current state genomic activities and the level of knowledge and interests in genomics by state and territorial CDDs.

$H_1$2: There is an association between one or more current state genomic activities and the level of knowledge and interests in genomics by state and territorial CDDs.

Current state genomic activities that were seen as having a potential impact on chronic disease public health genomics program implementation were queried. These included (a) a state genetics needs assessment, (b) a state genetics needs assessment that includes chronic disease conditions, (c) inclusion of genetics in the state public health action plan, (d) genetic educational programs, (e) genomics topics on the BRFSS, (f) analysis of state cancer registries or other vital records data to identify citizens with hereditary cancer syndromes, (g) frequency of collaborations or partnerships with outside entities related to genomics, and (h) presence of legislation and/or regulation specifically related to genomics. To determine the knowledge and interests in genomics by the CDDs, they were asked about their (a) awareness of contact information for clinical genetic services for potential referral or consultation, (b) knowledge of Tier 1 recommended conditions, (c) agreement with genomic statements, and (d) interest in the integration of genomic activities.

Theoretical Framework

In this study, I used the theoretical framework of the diffusion of innovations theory to explore the adoption of chronic disease genomics at the state level. According to Rogers (2003), adoption of new innovations in organizations can be challenging even if the advancements have clear, evidence-based rewards as in the current climate of Tier 1 recommendations. Diffusion is a process that occurs over time through communication between members of a social system and culminates with a modification of the structure and function of the social system (Rogers, 2003). The four main elements of this theory include the characteristics of the innovation itself, communication channels, time, and a social system that supports adoption (Rogers, 2003)

In this study, I examined the social system that supports adoption of chronic disease genomics, particularly looking at specific state genomic activities that may be associated with the level of knowledge and interests in genomics by CDDs. Moreover, based on Roger's (2003) theory, adoption of chronic disease genomics is ready to move into more states at this time; I will discuss this topic in more detail in Chapter 2. This adoption could be connected to genomic champions who have worked to secure funding for genomic activities in their states. My determination of whether or not there was an association of greater knowledge and interests in genomics by the CDDs in the few states that have received funding could influence the need to identify a genomic champion in each state and secure more funding for state genomic activities.

Nature of the Study

In this study, I used a nonexperimental, cross-sectional, correlational quantitative survey design to examine current state genomic activities and possible associations with the level of knowledge and interests in genomics by CDDs from all U.S. states and territories. This survey was originally developed by a subcommittee of the American Public Health Association (APHA) Genomics Forum Policy Committee (GFPC), of which I am a member, in order to determine opportunities and challenges of state CDDs in genomics and possibly create a position statement by the APHA. The survey design was chosen because it would be fairly easy to administer to the study group via e-mail web-link, be simple to develop at little or no cost, and could ask a number of pertinent questions to obtain a broad range of data (see Frankfort-Nachmias & Nachmias, 2008).

With the first half of the survey, I collected information about the presence of specific state genomic activities and in the second half inquired about knowledge and interests in genomic topics among CDDs. State genomic activities were either present (Yes), absent (No), or unknown (Don't know). One question was framed to inquire about frequency of collaborations with outside entities and was measured on a Likert scale. Knowledge and interests about genomic topics were also measured on a Likert scale; however, these results were converted to a numerical product for analysis of the *level of knowledge or interest* (e.g. Agreement with genomic statements: 1= strongly disagree through 5= strongly agree). I performed statistical analysis to determine the relationship between the variables to see if state funding or any particular activity was associated with an increased level of knowledge and interests in genomics by the CDDs.

Definition of Terms

I am providing the following definitions to ensure uniformity and understanding of these terms throughout the study:

Cascade screening: The systematic identification and testing of family members of an individual who has a particular disease of interest (Ned & Sijbrands, 2011).

Diffusion: The process by which an innovation is communicated through certain channels over time among members of a social system (Rogers, 2003).

Evidence-based medicine: Health technologies and practices supported by sound research evidence (Greenhalgh, Robert, Macfarlane, Bate, & Kyriakidou, 2004)

Genetics: The study of heredity with a focus on a specific and limited number of genes with known function in disease (Manolio, 2016).

Genomics: The study of an individual's entire genetic makeup, the genome, while also examining how the genome interacts with environmental or behavioral factors. This is especially important in the study of complex chronic diseases that affect large factions of the population (Cragun et al., 2016)

Innovation: A novel set of behaviors, routines, and ways of working, which are directed at improving health outcomes, administrative efficiency, cost-effectiveness, or user experience, and which are implemented by means of planned or coordinated action (Greenhalgh, Robert, & Bate, 2005).

Precision medicine: Tailoring medical therapies to subcategories of disease based on genomics (Ashley, 2015).

Precision public health: Providing the right intervention to the right population at the right time (Khoury, Iademarco, & Riley, 2016).

Public health genomics: The study and application of knowledge about the elements of the human genome and its functions, including interactions with the environment, in relation to health and disease in populations (Cleeren, Van der Heyden, Brand, & Van Oyen, 2011).

Scope and Delimitations

The scope of this study consisted of all state and territorial CDDs in the United States. To date, this group had not been surveyed specifically about knowledge in genomics and its connection to chronic disease nor the possible association with current state genomic activities. Each state and territory has one director and all are members of the National Association of Chronic Disease Directors (NACDD), which was my point of contact and access to this study group. The total possible participants for this study was 58, and each chronic disease director had an equal chance to participate in this study. To encourage participation, I limited the quantitative survey design to mostly closed-ended questions and took place in a 6-week timeframe.

I explored a range of theoretical models to provide a framework for this study. Consideration was given to the transformational leadership and transtheoretical models, utilization management, attribution, and complex adaptive theories. Upon discussion with my committee, I decided to use Rogers' diffusion of innovations theory to explain the current state of chronic disease public health genomics.

Assumptions

One assumption I made in this study was that the database provided for the target population by the NACDD was current and accurate. The CEO of the NACDD explained that the database is updated yearly and frequent e-mails are sent each month ensuring accuracy (J. Robitscher, personal communication, January 5, 2017). Another assumption was that the sample of survey responses is representative of the whole population of CDDs being studied. Because one question on the survey asked about what state the chronic disease director practices in, this helped determine if the study population resided in different parts of the country (heterogeneous sample) or from states that have received some type of funding for state genomic activities related to chronic disease. A final assumption was that respondents answered the questions truthfully. As this is a confidential, voluntary questionnaire, it would be more likely that these directors would be honest in their responses.

Limitations

Due to the small sample size available for the study, results may not be generalizable beyond the specific population from which the sample was drawn. Because the number of CDDs in the United States and territories is limited, a small response rate impacted the power of the analysis (< 80%) by introduction of Type II errors and not allowing for generalizability to the study population (Field, 2013). The nonresponse bias is also important; the nonresponse bias is how different or similar those who do not respond are from the whole survey population (Johnson & Wislar, 2012). Those who chose to complete the survey may have had more knowledge and interest about genomics

and vice versa. Also, because the subcommittee and I conducted this study voluntarily, there was no monetary compensation for the directors to complete the survey, which may have also impacted the response rate (Cho, Johnson, & VanGeest, 2013). This survey was a web-based instrument with a short completion time (6 weeks), which could have affected participation if the CDDs did not find the time or remember to complete it in the allotted window. A final limitation was that, although this was a confidential web-based survey, this did not assure that the chronic disease director who received the survey was the one who completed it in part or in totality.

Significance

Data from this study could provide public health leaders at the federal, state, and local levels with information as to what specific genomic activities are associated with increased levels of knowledge and interest in genomics by state CDDs. This information could be starting point for states to increase genomic activities to conform to the new Healthy People 2020 objectives, Precision Medicine Initiative, and Tier 1 genetic testing and screening recommendations (Auffray et al., 2016; Dotson et al., 2014; Weir et al., 2015). These results could also support and provide rationale for public and private funding for state genomic activities and identify states that are ready to begin genomics implementation. Finally, the results of this survey could provide a small snapshot of what is and is not being done in the states today in regards to genomics as well as highlight the level of knowledge and interest in specific genomic topics by CDDs.

Implications for Social Change

Walden University defines positive social change as "a deliberate process of creating and applying ideas, strategies, and actions to promote worth, dignity, and development of individuals, communities, organizations, institutions, cultures, and societies" (Walden University, 2016, para. 2). This definition is expected to provide a foundation for student research that will result in the betterment of human and social conditions. In the context of this study, the implications for social change relate to the rights of the public to have access to evidence-based technology that has the ability to reduce morbidity and mortality of certain hereditary diseases. Gaining an understanding of the opportunities and barriers to Tier 1 chronic disease genomics implementation could provide the "ideas, strategies, and actions" for increasing this work in more states (Walden University, 2016, para. 2). If there is a connection between particular state genomic activities and how knowledgeable and interested the CDDs are in this area, this finding could encourage funding for more state activities. This funding could provide a ripple effect of engagement for chronic disease departments, state health departments, and other stakeholders.

Summary

Individuals who work in public health have a responsibility to ensure that the communities they serve are healthy by assuring safe, accurate, and accessible chronic disease genomic services (Cragun et al., 2016). Expansion of state chronic disease genomics to a larger proportion of states is prudent at this time in light of the Healthy People 2020 objectives, the Precision Medicine Initiative, and evidence-based Tier 1

genetic testing and screening recommendations (Auffray et al., 2016; Modell, Greendale, Citrin, & Kardia, 2016; Weir et al., 2015). The purpose of this study was to identify some possible opportunities and challenges to reach that goal. This foundation will support ideas to facilitate implementation of chronic disease public health genomics in more states.

In this introduction, I presented the statement of the problem, research questions, definition of terms, scope, nature, and significance of the study, as well as study limitations. In Chapter 2, I will provide a review of the literature in the field of public health genomics from its inception to the present day and connect what is currently happening through Roger's diffusion of innovations theory. I will outline the methodology and data collection procedures in Chapter 3 and describe the data analysis and findings in Chapter 4. Finally, Chapter 5 will include my summary and discussion of the findings along with conclusions and recommendations for future studies.

Chapter 2: Literature Review

Introduction

The problem I addressed with this study was the need for public health genomic programming to move beyond traditional NBS and into the chronic disease arena in the United States and its territories (Bowen et al., 2012). Current evidence is available to encourage genomic risk assessment through the screening and testing of individuals and cascade follow-up and testing of at-risk family members for hereditary forms of breast, ovarian, and colon cancer as well as cardiovascular disease (Dotson et al., 2014). Public health practitioners, especially at the state level, are poised to be the leaders in facilitating evidence-based genomic surveillance and screening for certain hereditary chronic diseases (Green et al., 2015).

This chapter will include a narrative of the birth of public health genomics through the present day including how public health genomics ties into the core public health functions, the role of genomics in population health, a definition of precision public health, Tier 1 genetic testing recommendations, and the burden of each condition. Finally, I will describe the role of state chronic disease departments in Tier 1 implementation and the current landscape. Through my review and discussion of the literature on the framework of this study, Rogers' diffusion of innovations theory, I will demonstrate where public health genomics is today and how it is poised to move to the next level, what translation barriers exist, and how collaboration is crucial to implementation success.

The purpose of this quantitative survey design was to determine whether there is an association between current state genomics funding or specific state genomic activities and the level of knowledge and interests in genomics by state CDDs. Using this quantitative survey design, I analyzed results of a survey of CDDs in all U.S. states and territories. The intent of this study was to identify and describe particular activities or particular states that may be associated with an increased level of knowledge and interest in genomics by CDDs, which may also influence implementation of Tier 1 genetic tests at the state level.

Literature Search Strategy

I conducted a review of the literature using the Walden University Library, Google Scholar, and the World Wide Web. I searched the CINAHL, MEDLINE/PubMed, Science Direct, EBSCO, and ProQuest Central databases for peer-reviewed, English language journal articles focusing on articles of interest in the last 5 to 6 years (2010–2016). Key search terms used included *public health, genetics or genomics, chronic disease directors, knowledge of genomics, population genomics, champion,* and *diffusion of innovations.* These searches yielded 326 scholarly journal articles, four books, and two dissertations related to the topic of this study.

Theoretical Foundation

At the time of this study, very few states are doing any significant work in chronic disease public health genomics with a majority of states incorporating these activities on a limited basis and predominantly focusing on the core function of assurance (Laufman et al., 2012). Those few states who are leading the implementation of Tier 1 testing play an

important role in the diffusion of these new evidence-based public health applications by modeling activities, providing public health outcome data, and championing the cause as an opinion leader or change agent. In this literature review, I investigated how Rogers' diffusion of innovations theory could be used to explain the development of the field of chronic disease public health genomics and ways that this theory could be used with the results of this study to identify clear avenues to increase adaptation across more states throughout the country.

Rogers's Theory

Diffusion is "the process by which an innovation is communicated through certain channels over time among members of a social system" (Rogers, 2003, p. 5). The four main components are (1) the *innovation*, (2) *communication channels*, (3) *time*, and (4) the *social system*. An innovation is a new idea that will likely bring forth a certain degree of uncertainty to the social system depending on the number of alternatives available and the probability that the new innovation is superior to or enhances current practices (Rogers, 2003). Communication of the benefits of the new innovation is a two-way process that occurs over many cycles of information exchange to reach a mutual understanding (Rogers, 2003). Time is an important component of the diffusion process whether to understand the innovation decision process from first knowledge to acceptance or rejection, why certain individuals adopt the innovation earlier or later in the process, and the innovation's rate of adoption in a system (Rogers, 2003). Finally, diffusion has the capability of altering the structure or function of a social system when a

new idea is conceived, diffused, and either incorporated or rejected leading to consequences that change the social system (Rogers, 2003).

The five adopter categories are (1) *innovators*, (2) *early adopters*, (3) *early majority*, (4) *late majority*, and (5) *laggards* (Rogers, 2003). Innovators, the first to adopt a new idea into a system, actively seek new ideas, often reach outside their own locale for information and support, and are able to handle greater amounts of uncertainty (Rogers, 2003). These individuals often serve as change agents who influence others in the social system to adopt the innovation (Rogers, 2003).

Early adopters are also enthusiastic about new innovations while still being selective about what they adopt (Rogers, 2003). This group also has a great degree of opinion leadership; however, they examine both the positive and negative aspects of an innovation, so buy-in from this group is critical to adoption success (Rogers, 2003). These individuals are often consulted by others for advice and information about the innovation, serve as role models, and help decrease uncertainty by others (Rogers, 2003). The early majority adopts new innovations before the average members of the system; however, adoption comes only after lengthy deliberation (Rogers, 2003). This group is an important link to the diffusion process by connecting the enthusiastic leaders with the typical members of the group who are inclined to be more resistant to adoption (Rogers, 2003).

The late majority are skeptical and only adopt innovations due to economic or system pressures even after they have been persuaded of the utility of the new idea; most if not all of the uncertainty of adoption must be removed before this group will join

(Rogers, 2003). Finally, laggards are the last to adopt a new innovation because they traditionally live in the past, are resistant to change, and are suspicious of new ideas (Rogers, 2003). Rogers (2003) explained that by the time laggards adopt a new innovation, it may already be out of date and surpassed by a newer method.

How fast an innovation is adopted by the members of a social system is contingent on its perceived characteristics, type of innovation decision and communication channels used, inherent nature of the social system, and the extent of effort by change agents in diffusing the innovation (Rogers, 2003). The diffusion effect explains the important relationship between the rate of knowledge about an innovation and the rate of its adoption by those in the system (Rogers, 2003). As the level of innovation knowledge increases to the 20–30% range, only small amounts of adoption occur; however, once this threshold passes the *tipping point* (which can be slightly different depending on the innovation and system), the rate of adoption increases exponentially (Griliches, 1957). This threshold is often contingent on the point at which opinion leaders in a system begin to look favorably on the innovation and activate peer networks in the social system.

How Roger's Theory Has Been Used in Previous Research

According to Schon (1963)**,** resistance to change is normal and may also seem desirable in many instances to assure stability in organizations. In order to promote changes that are in the best interests of the organization and those they serve, a champion will often emerge to fight for the introduction and development of a new innovation. These champions are most often emergent leaders from within the organization who are

effective at influencing the leadership process to produce the desired change (Taylor, Cocklin, Brown, & Wilson-Evered, 2011). Champions are intrinsically motivated, energetic and enthusiastic, and committed to the cause; either the new advancement finds a champion or dies (Schon, 1963).

Previous researchers have examined the use of champions on diffusion of innovations in health care settings. A 2006 study on the implementation of the MOVE! weight-management program in the Veteran's Health Administration found that organizational readiness for change and the presence of an innovation champion were key factors in the success of this program (Weiner, Haynes-Maslow, Kahwati, Kinsinger, & Campbell, 2011). Novick et al. (2015) also found that champions who advocated for the enactment of a new model for prenatal care were instrumental in successful implementation and sustainability at group practices. Finally, a study about the adoption of the Agency of Healthcare Research Quality tools to assess pharmacy's health literacy practices also found that a change champion would have a positive impact (Shoemaker, Staub-DeLong, Wasserman, & Spranca, 2013).

Roger's Theory and Public Health Genomics

The field of public health genomics outside of NBS has seen some great successes in model states since the establishment of the Office of Public Health Genomics at the CDC in 1997, the completion of the Human Genome Project in 2003, and the formalization of the field in 2005 (Green et al., 2015; Modell et al., 2016). These model states have all received some kind of funding for genomic activities and assessments (ASTHO, 2010; CDC, 2016; Green et al., 2015; St Pierre et al., 2014; Trivers et al.,

2015) Table 1 shows the model states and their category of adoption. At this time, the field appears to be at or over the tipping point and is ready for integration of chronic disease genomics into more states (19–20) in the early majority category.

Table 1

Current Status of Chronic Disease Public Health Genomics Programming Adoption

Adoption category	% expected	*N* based on 58 states and territories	States involved
Innovators	2.5%	1–2	Michigan[a,b,c] Oregon[a,b,c]
Early Adopters	13.5%	7–8	Connecticut[a,b,c] Utah[a,b,c] Minnesota[a,b] Georgia[a,b] Colorado[c,d] Ohio[a] Washington[e]
Early Majority	34%	19–20	
Late Majority	34%	19–20	
Laggards	16%	9–10	

Note. (a) St Pierre et al., 2014, (b) Green et al., 2015 (c) CDC, 2016, (d) ASTHO, 2011a, (e) ASTHO, 2011b.

Innovativeness refers to how quickly or reluctantly an individual or system unit adopts an innovation (Rogers, 2003). New innovations are often proposed by opinion leaders, such as the OPHG, who maintain a high degree of credibility regarding the technical and theoretical aspects of the innovation (Rogers, 2003). Change agents, who are often in the early adopter category, work alongside opinion leaders to champion the

adoption of an innovation (Rogers, 2003). In this case, the change agents would champion the diffusion of chronic disease genomics at the state level.

One area that has been shown to impact an innovation's adoption is the communication and influence that occurs through social networks, peer and expert opinion, champions, and change agents (Greenhalgh et al., 2004). Currently, model states that are doing work in chronic disease public health genomics have at least one individual who devotes time to initiate agendas, develop and assess programs, seek and obtain funding, provide education, and facilitate stakeholder collaborations (ASTHO, 2010; St Pierre et al., 2014). As Rogers (2003) explained, the "presence of an innovation champion contributes to the success of an innovation in an organization" through communication of the benefits of an innovation over a period of time and numerous conversations to influence the rate of adoption (p. 414). Schon (1963) clearly stated that without a champion, new ideas will likely die from normal and somewhat desirable resistance to change. Having a genomic champion at the state level that is knowledgeable and committed to chronic disease genomics could positively impact the rate of adoption of genomics in state health departments.

Although evidence exists to support chronic disease genomics in the states, only a handful of states are working to that end. Diffusion of this new innovation and the rate of adoption by more states can be encouraged by a variety of factors such as the level of communication and influence provided to stakeholders, which includes state and territorial CDDs. Innovator and early adopter states have at least one genomics champion

who has likely impacted the rate of adoption in their states and this can be a model to increase implementation in additional states.

History of Public Heath Genomics

In 1990, the National Institutes of Health and Department of Energy launched the Human Genome Project to develop technology that could analyze DNA, map and sequence the human genome, and investigate associated ethical, legal, and social issues (National Human Genome Research Institute (NHGRI), 2015). In order to address the population health impact of the HGP, the CDC established the Office of Genetics and Disease Prevention (now the OPHG) and created a strategic plan to address the translation of genomic advances into population health (Zimmern & Khoury, 2012). Since its inception in 1997, the OPHG has involved many partners to anticipate, evaluate, and demonstrate the translation of genomics into population health practices (OPHG, 2011). A meeting in Bellagio, Italy in 2005 resulted in the formal definition of public health genomics as "a multidisciplinary field concerned with the responsible and effective translation of genome-based knowledge and technologies into health care practices to improve population health" (Bellagio Report, 2005; CDC, 2007, p. 1). Public health genomics seeks to use population data of genetic variation and environmental influences to establish evidence-based interventions for disease prevention and health improvement.

Use of genome-based knowledge for public health interventions has been around long before the term public health genomics was first defined. In 1961, Dr. Robert Guthrie developed the first NBS test to identify infants with phenylketonuria resulting in

the institution of mandatory state screening programs in 1963 (NHGRI, 2016). Today, virtually all babies born in the United States undergo NBS in state run programs to detect a variety of endocrine, metabolic, and hematologic conditions that are genetic in nature and was named one of the 10 Great Public Health Achievements of the 20th century (CDC, 2011; Ross, 2010). Universal screening of newborns highlights the ability of public health to reduce morbidity and mortality of hereditary conditions through state-run programs.

Recently, there has been increasing momentum from the national level to encourage the integration of genomics into public health programming. In December of 2010, the Office of Disease Prevention and Health Promotion included genomics in the Healthy People 2020 objectives for the first time (Office of Disease Prevention and Health Promotion, 2016). These new objectives reflect increasing evidence to support the use of genetic tests and family health history in clinical medicine and public health. The first two recommendations include (a) Women with a high familial risk of breast, ovarian, tubal, or peritoneal cancer could benefit from genetic counseling to learn more about genetic testing for the breast cancer susceptibility gene (BRCA) 1/2 mutations and post-test surgical options to reduce risk, and (b) All newly-diagnosed colorectal cancer patients should receive information regarding genetic testing to identify a hereditary form of this cancer (LS), which could benefit family members by reducing their risk of colorectal cancer caused by LS through screening and interventions.

In January 2015, President Obama announced his support for the Precision Medicine Initiative, which aims to link researchers, providers, and patients to focus

disease prevention and treatments based on individual differences in genetics, environment, and lifestyle (The White House, 2015). His $215 million, 2016 budget financing is to be a collaborative public and private investment in genomic advances, tools for managing and analyzing large sets of data while protecting patient privacy, and health information technology. This initiative is also designed to engage at least a million Americans to volunteer their health information to study health outcomes, develop new treatments, and introduce a more precise and personalized healthcare system. Of course, all of these promises are at-risk under the new administration.

How Public Health Genomics Fulfills the Core Public Health Functions

The mission of public health is "fulfilling society's interest in assuring conditions in which people can be healthy" (Institute of Medicine Committe for the Study of the Future of Public Health, 1988, p. 7). This mission is to be carried out through public and private partnerships, however, public agencies have a responsibility to assure that essential components are in place to address the mission effectively. Along with that mission are three core public health functions; assessment which includes collection and analysis of population health data, assurance of quality services to all, and policy development based on sound scientific knowledge and use of the democratic political process. Table 2 provides some examples of genomics in relation to these core public health functions.

Table 2

Role of Genomics in the Delivery of Essential Public Health Services

Core Function	Description	Examples
Assessment	Monitor Health Diagnose & Investigate	• Utilize family history or genetic testing to identify at-risk individuals[b] • Perform epidemiologic studies on the prevalence of genetic risks factors / variants within the community to determine their contribution to identified health problems[a] • Study gene-gene and gene- environment interaction[b] • Assess the availability, appropriateness, and accessibility of quality genetics resources in the community[a] • Assess the impact of genetic information and its value in improving health[a] • Research the community's and health care providers' knowledge of the use of genetics to improve health[c]
Assurance	Link to / Provide Care Assure A Competent Workforce Evaluate	• Collaborate with other public and private entities and educate public health staff and private health-care workers about the use of genetic information to improve health[a] • Incorporate genomics into the curricula of medical schools, nursing schools, and schools of public health and provide opportunities for continuing education around genomics[b] • Evaluate genomic tests, services, and information to ensure availability, efficacy, accessibility, safety, quality, and ethical practices while also enforcing the policies and standards enacted to ensure this[b] • Identify and analyze the factors that influence the impact of genetic information and the delivery, utilization and quality of genetic tests and services[a]

(table continues)

Core Function	Description	Examples
Policy Development	Inform, Educate, & Empower Mobilize Community Partnerships Develop Policies Enforce Laws	• Improve genomic literacy of the public, health care practitioners, policy makers, and other stakeholders through audience-specific educational initiatives about the integration of genomics into health promotion and disease prevention programs[b] • In collaboration with stakeholders, implement regulatory policies and guidelines for clinical applications, test implementation, use, impact, and protection of genomic information, and accessibility and quality of genomic technology[a,b] • Identify and analyze the economic, social, ethical and political implications of advances in human genetics, including the information and communications needs of stakeholders[b] • Assure insurance coverage for high risk individuals[b] • Develop, enhance and sustain partnerships with key partners[a]

Note: (a) Khoury, 2011, (b) McWalter & Gaviglio, 2015, (c) ASTHO, 2010.

The Role of Genomics in Population Heath

During the last 20 years, the OPHG at the CDC and other multidisciplinary groups have been trying to use the knowledge gained from the HGP and other scientific advances and translate this into activities for population health (Zimmern & Khoury, 2012). Beyond the ever-expanding state universal newborn screening panels, public health genomics is going to play a significant role in epidemiological studies, infectious disease, chronic disease, and environmental health (Roberts, Dolinoy, & Tarini, 2014). Implications will also be felt in areas such as biostatistics, health policy and regulation, health education, health behavior responses to genomic information, and equitable distribution of the costs and benefits from genomic discoveries and applications. The

integration of genomics into this wide variety of activities will require complex structures, processes, and collaborations by a diverse range of stakeholders to fully realize the translation of genomic findings into improved population health.

Family Health History

The original genomic tool used in medicine and public health has been the use of family health history (FHH). A large majority of chronic diseases of public health significance including diabetes, cardiovascular disease, several cancers, osteoporosis, and asthma have been shown to have a strong family history component (Yoon et al., 2002). FHH is a combination of shared genetic susceptibility, environment, and behaviors and, prior to the availability of genetic testing, chronic disease programs and clinical health practitioners have traditionally focused their efforts on the environmental and behavioral components (OPHG, 2011). Use of FHH and possible genetic screening and testing could complete a three-legged stool of disease prevention targets. Public health leaders can be effective advocates in educating others about the link between FHH and chronic disease, especially to minority groups, and can use this tool as a surveillance method to identify at-risk individuals and their family members, and evaluate the impact on population interventions (Butty et al., 2012; Khoury et al., 2011; Powell, Edleson, O'Leary, Christianson, & Henrich, 2011; Senier et al., 2015). Although FHH will continue to be a valuable primary prevention tool, issues with collection, standardization, interpretation, and integration with electronic health records exist and will need to be addressed (Bowen et al., 2012; Modell, Kardia, & Citrin, 2014; Valdez et al., 2010; Williams, 2012).

From Precision Medicine to Precision Public Health

Precision medicine, sometimes referred to as personalized medicine, is a concept that implies prevention or treatments based on individual differences (Collins & Varmus, 2015). Pharmacogenomics, the most recognized precision medicine mechanism, is aimed at providing the right drug to the right patient at the right time based on an individual patient's genetic makeup (Auffray et al., 2016). Precision public health, on the other hand, focuses on individuals within a defined population for "providing the right intervention to the right population at the right time" (Khoury, Iademarco, & Riley, 2016, p. 398). Long before advances in genomics, Rose (1985) explained that populations would be healthier and costs contained if prevention efforts were targeted at those in the population who are at greatest risk for an identified disease. Identifying and explaining why some individuals, or groups of individuals, get sick while others don't is an excellent guide to public health prevention efforts. Targeted public health screening programs not only seek to protect susceptible individuals, but try to discover and control the cause of incidence; susceptibility will not exist if causes are removed or circumvented.

The completion of the Human Genome Project has enabled a significant opportunity to practice clinical medicine and public health in a novel way. Acquiring the ability to identify disease in individuals and populations based on genetic components will permit us to target prevention efforts and treatments based on heredity and individual or population susceptibility. This has been highlighted by inclusion of genomics for the first time in the Healthy People 2020 objectives and support for the Precision Medicine Initiative. The field of public health genomics fulfills the core public health functions,

supports delivery of essential health services, and will play an active role in furthering use of genomic advancements in population health.

Tier 1 Genetic Testing Recommendations

Due to advances from the Human Genome Project and genomic applications, evidence-based recommendations are now available to move public health genomics from reducing morbidity, mortality, and disability in the newborn period to identification of genetic influences across the lifespan (Bowen et al., 2012). Although a large proportion of applications will be delivered in the clinical care setting, state public health agencies are poised to be the leaders in targeted population screening programs (Khoury et al., 2011). These leaders will be responsible for program development and implementation, delivery, assessment, reduction of potential harms, equitable access, and creation of a multidisciplinary infrastructure for program support and future use of genomic applications. Public health professionals, who are focused on population health and reduction of health disparities, can successfully gather these stakeholders without bias or another agenda.

Tier Classifications

In 2005, the CDC OPHG established the Evaluation of Genomic Applications in Practice and Prevention (EGAPP) Initiative to develop a process for an evidence-based evaluation of genomic tests in clinical medicine and public health practice (Teutsch et al., 2009). Recommendations for test readiness come from a multidisciplinary expert group of nonfederal, independent individuals who evaluate a test's (a) analytic validity (ability to accurately and reliably measure the genotype of interest); (b) clinical validity (ability

to detect or predict the disorder or phenotype of interest); (c) clinical utility (evidence of improved measurable clinical outcomes and usefulness to patient management); and (d) associated ethical, legal, and social implications before suggesting its use (Green et al., 2015; Secretary's Advisory Committe on Genetic Testing, 2000). These tests have been classified into a three tiered, color-coded system to indicate what tests are ready to be integrated into clinical care and public health practice (Dotson et al., 2014; see Table 3).

Table 3

Tier Classification System of Genomic Tests

Tier 1	Tier 2	Tier 3
Genomic and Family Health History applications have a base of synthesized evidence to support integration into practice.	Genomic and Family Health History applications have insufficient evidence to support routine implementation into practice. These applications do have the potential to provide information for informed decision making by patients and providers or for informing selective use strategies (e.g. clinical trails) through clinical or public health policy decision making.	Genomics and Family Health History applications have evidence that either results in recommendations against use OR no relevant evidence is available at this time. Tier 3 applications are not ready for routine use but may be used for clinical or population research.

Note. Adapted from Dotson, W. D., Douglas, M. P., Kolor, K., Stewart, a C., Bowen, M. S., Gwinn, M., … Khoury, M. J. (2014). Prioritizing genomic applications for action by level of evidence: A horizon-scanning method. *Clinical Pharmacology and Therapeutics*, *95*(4), 394–402. http://doi.org/10.1038/clpt.2013.226.

As of April 2016, 46 tests have been classified as Tier 1, 105 are Tier 2, and nine are in Tier 3 (OPHG, 2016c). Although most of the Tier 1 genomic tests are related to

pharmacogenomics, tests also include the 31 core newborn screening conditions and three chronic disease conditions. Applications for these conditions include cascade DNA and LDL (low-density lipoprotein) testing of relatives of patients identified with FH, diagnostic screening for LS for colorectal cancer patients and cascade screening for their family members, and risk prediction and referral to genetic counseling for BRCA testing for those with a risk of HBOC.

Classifications for these Tier 1 genomic applications are based on the recommendations of the National Institute for Health Care Excellence, EGAPP, and the U.S. Preventive Services Task Force (Evaluations of Genomic Applications in Practice and Prevention Working Group, 2009; National Institute of Health and Care Excellence, 2008; U.S. Preventative Services Task Force, 2014). All three disorders are autosomal dominant (only one defective gene is required to inherit the disease); have lifelong health implications; and use family health history to identify those at risk and cascade screening to connect to family members who could benefit from further follow-up (Bowen et al., 2012). FH also includes a rarer homozygous variant (two mutations are inherited) which increases the severity of the disorder (Nordestgaard et al., 2013).

Public Health Burden of Tier 1 Conditions

There are over two million individuals who carry the mutational genes for HBOC, LS, and FH in the United States today (George, Kovak, & Cox, 2015). Considering that cancer and heart disease are currently the top two burdens in our health care system, costing an estimated $75 and $320 billion respectively per year, finding ways to identify

and reduce the incidence of these diseases should be a key public health focus (American Cancer Society, 2016; American Heart Association, 2015). At present, these conditions are poorly identified by the healthcare system so targeted Tier 1 genetic testing programs and potential cascade screening of family members implemented through state public health departments in collaboration with health care practitioners could offer significant reduction in health risks from these diseases and their associated costs (OPHG, 2014). Table 4 displays the potential impact of Tier 1 genetic testing on the conditions identified.

Table 4

Public Health Prevalence and Burden of Tier 1 Conditions

	Condition			
	Breast cancer	Ovarian cancer	Colon cancer	Cardiovascular disease
	Most common in women[1] 2nd highest cause of cancer death[1]	5% of cancer deaths in women[1]	3rd most common in men and women[1] 3rd highest cause of death[1]	Leading cause of death in both men and women
Estimated total new adult cases 2016	246,660[1] women 2,600 men[1]	22,280[1]	95,270[1]	73.5 million in pop (31.4%)
Prevalence of mutation in new cases	2–7% [2a]	10–15%[2a]	3–5%[4]	N/A

(table continues)

	Condition			
	Breast cancer	Ovarian cancer	Colon cancer	Cardiovascular disease
Potential identification per year through Tier 1 recommendations[b]	4932–17,262 persons	2,228–3,342 persons	2858–4763 persons	N/A
Risk of disease with mutation	40–80%[3]	11–40%[3]	80%[6]	50% men – 30% women[7]
Estimated prevalence of mutation in US population	1:300–1:500 [3*]	1:300–1:500 [3*]	1:370[5]	1:250[c]–1:500[7]
Estimated US population carrying mutation[d]	648,000–1,080,000	648,000–1,080,000	875,675	600,000–1.2 million[8]
Yearly deaths	40,450 women[1] 440 men[1]	14,420[1]	49,190[1]	370,000[9]

Note: (a) Significantly higher rates in Ashkenazi Jewish population, (b) Based on calculation of total new cases and % new mutation prevalence. Does not include family members potentially identified through cascade screening, (c) Estimated higher rates in European Caucasian populations, (d) Based on estimated current U.S. population of 324 million (United States Census Bureau, 2016b).

1. (American Cancer Society, 2016), 2. (D'Andrea et al., 2016), 3. (Petrucelli, Daly, & Feldman, 2013), 4. (American Cancer Society, 2014), 5. (Hampel & De La Chapelle, 2011), 6. (Guillén-Ponce, Molina-Garrido, & Carrato, 2012), 7. (National Organization for Rare Disorders, 2016), 8. (Ned & Sijbrands, 2011), 11. (American Heart Association, 2015)

Description of Tier 1 Conditions

Hereditary breast and ovarian cancer (HBOC). HBOC syndrome is caused by mutations in the BRCA1 and BRCA2 genes, which not only cause an increased risk for breast (40–80%) and ovarian (11–40%) cancers, but also pancreatic and prostate cancers (Petrucelli et al., 2013). Mutations in BRCA 1/2 are passed in an autosomal dominant fashion and cluster in families (U.S. Preventative Services Task Force, 2014). Prognosis

of these cancers is based on when the cancer is detected, so identification of increased susceptibility through genetic testing and subsequent preventative monitoring and/or prophylactic surgery could impact the morbidity and mortality triggered by these mutations. For women with these mutations, surgery could reduce the risk of breast and ovarian cancer by 69% (U.S. Preventative Services Task Force, 2014).

Current USPSTF recommendations are to screen women for a strong family history of increased risk for harmful BRCA 1/2 mutations and those identified should be offered genetic counseling and potential BRCA 1/2 mutation testing. USPSTF gives this recommendation a 'B' rating which means that they recommend provision of this service because there is a high certainty that the net benefit of this service is moderate and a moderate certainty that the net benefit is moderate to substantial (U.S. Preventative Services Task Force, 2016). This 'B' rating is significant because this allows coverage by the Affordable Care Act (Henry J. Kaiser Foundation, 2016). Beyond the USPSTF recommendations, some authors propose that population screening for the BRCA 1/2 mutation would be cost-effective in the high-risk Ashkenazi Jewish population (D'Andrea et al., 2016).

Lynch syndrome (LS). LS, also called hereditary nonpolyposis colorectal cancer (HNPCC), is the most common cause of hereditary colon cancer accounting for 3–5% of all colorectal cancers (American Cancer Society, 2014; Guillén-Ponce et al., 2012). These individuals and their families are also at greater risk for other cancers including endometrial, ovarian, and stomach (Guillén-Ponce et al., 2012). LS is an autosomal dominant cancer syndrome caused by mutations in DNA mismatch repair (MMR) genes

MLH1, MSH2, MSH6, and PMS2, which function to correct mismatched base pairs as well as tiny insertions and deletions that occur during DNA replication (Mange et al., 2015). These corrections are needed to decrease genomic instability which occurs during DNA synthesis and mutations in these genes will lead to rapid tumor growth (Guillén-Ponce et al., 2012). Evidence also shows involvement by an epithelial cell adhesion molecule (EPCAM), which indirectly affects DNA repair by causing the MSH2 gene to be turned off (Kempers et al., 2011).

Screening for LS consists of tumor testing by either immunohistochemistry (IHC) or microsatellite instability (MSI) followed by genetic sequencing and deletion analysis of the MMR genes depending on the results of IHC and MSI (Mange et al., 2015). A hallmark of LS is the early-onset of colon cancer diagnosis (< 45 years), so prompt identification of patients, tumor testing, and cascade screening of their family members could lead to a reduction in LS-caused colorectal cancer incidence and related mortality.

At this time, evidence supports the integration of Tier 1 condition identification and prevention into clinical and public health practice. Tier 1 conditions have been shown to have genomic and family health history validation to support inclusion of Tier 1 genetic testing and screening for susceptible individuals and their family members. Tier 1 conditions include HBOC, LS, and FH, which are all contributing to the large prevalence and burden of cancer and heart disease experienced in the United States today.

Familial hypercholesterolemia (FH). FH is an autosomal co-dominant disorder expressed with abnormally high concentrations of low-density lipoprotein cholesterol (LDL-C), which increases an individuals' risk of premature coronary heart disease

(CHD) from atherosclerosis (Ned & Sijbrands, 2011). FH is caused by loss-of-function mutations in the LDL receptor (LDLR) and apolipoprotein (APOB) genes and gain-of-function mutations in the proprotein convertase-subtil-sin/kexin type 9 (PCSK9) gene (Austin, Hutter, Zimmern, & Humphries, 2004). Untreated, FH poses an approximate overall 20-fold increase in CHD from the general population, which includes a 50% CHD risk in men by age 50 and a 30% risk of CHD in women by age 60 (Marks, Thorogood, Neil, & Humphries, 2016).

Elevated LDL-C levels in affected individuals begin even before birth and those with two abnormal genes (FH homozygotes) can develop CHD very early in life and die before age 20 if left untreated (Nordestgaard et al., 2013). FH is as common as Type I diabetes, and more common than cystic fibrosis or Down's syndrome, however, it is estimated that only 1-25% of all cases are diagnosed (Knowles et al., 2014; Modell et al., 2016; Ned & Sijbrands, 2011; Nordestgaard et al., 2013). Identification of individuals at risk for FH includes high levels of cholesterol and family and/or personal history of early onset CHD. Targeted screening of these individuals for the FH mutation, subsequent treatment with lipid lowering pharmaceuticals, and a program of diet and exercise could lead to the prevention of tens of thousands of heart attacks over these individual's lifetimes.

Implementation of Tier 1 Tests: Current Landscape

In 2002, a Chronic Disease Directors' Summit was convened to begin the dialogue and develop a plan to move genomics out of NBS and into chronic disease units (Association of State and Territiorial Health Officials, 2002). These experts asked the

CDC to help states respond to the resulting information and applications from the Human Genome Project. In 2003, the CDC funded four states, Michigan, Minnesota, Oregon, and Utah in 5-year cooperative agreements to integrate genomics into their state public health programs (ASTHO, 2010; Green et al., 2015). With annual awards of between $150,000-$250,000, these states were able to use internal and external planning to integrate FHH and genetic testing results into existing genetics and chronic disease policies and programs (St Pierre et al., 2014). In addition, they formed partnerships, evaluated public data, developed workforce capacity and leadership, and established justifiable interventions using FHH, assessments, and educational curricula. After the infrastructure was built in the first round, the CDC issued new 3-year cooperative agreements to Michigan and Oregon in 2008 in order to shift the focus from capacity building to translational activities in public health genomics (ASTHO, 2010). The program focus for these agreements was on HBOC surveillance, education, and policy development in support of the USPSTF 2005 recommendations.

In 2011, the OPHG shifted state funding of genomic activities to the Division of Cancer Prevention and Control (DCPC) at the CDC to highlight the focus on cancer genomics by the Healthy People 2020 objectives. The DCPC granted $300,000 per year from 2011–2014 to Michigan, Oregon, and Georgia for HBOC activities and by the end of the three-year period, each had an established, effectively operating breast cancer genomics program (Trivers et al., 2015). Recently, Utah, Connecticut, and Colorado have been included in the support of implementation of evidence-based cancer genomics recommendations (OPHG, 2016a). Also in 2011, the Connecticut Department of Public

Health Genomics Branch received a Healthy People 2020 Action Award to facilitate cancer genomics in that state (Connecticut Department of Public Health, 2012). Other states (Ohio, Hawaii, Illinois, and Washington) have fostered state genomic activities through implementation grants, academic centers, and collaborations with outside stakeholders, but none have been funded to the extent that these model programs have (Green et al., 2015).

Examples of Cancer Genomics Translation by Model States

The following list describes some ways that model states have integrated cancer genomics into their public health programming:

- Addition of breast cancer questions to the Behavioral Risk Factor Surveillance System (BRFSS).
- Analysis of state cancer registry data and using this information to communicate and educate providers and patients about potential HBOC counseling and testing of patients and their family members.
- Development of new surveillance systems with key stakeholders and genetics clinics to evaluate uptake of HBOC genetic counseling, testing and follow-up.
- Collaboration with public health clinics to integrate HBOC risk screening into the clinical intake process.
- Analysis of and collaboration with insurance companies to assure coverage for genetic counseling and testing for HBOC.
- Education of health providers and the public about Tier 1 tests.

(Trivers et al., 2015)

National Resources for State Public Health Genomics

In 2010, ASTHO developed a *State Public Health Genomics Resource Guide* highlighting the issues, strategies, and challenges to state genomic implementation with links to appropriate resources (ASTHO, 2010). This publication also described specific activities and tactics that model states were accomplishing to meet clearly defined genomic objectives. In 2014, the OPHG published an online Genomic Application Toolkit to share the public health genomics methods of these model programs and give other states some ideas and advice for development and application of genomic programs in their own states (OPHG, 2016a, 2016b). The website explains what the Tier 1 genomic applications are, their importance to population health, and how state and local health departments can play an important role in the application of Tier 1 tests by identifying people who could benefit from testing and extending that benefit to their family members. This toolkit also provides links to implementation videos and other resources to help.

The Role of State Chronic Disease Departments in Tier 1 Implementation

Because the current Tier 1 genetic testing recommendations outside of NBS and pharmacogenomics are all identified as hereditary forms of chronic diseases, state and territorial CDDs and the personnel who work in these departments should be educated about and engaged in Tier 1 genetic testing program implementation (Zimmern & Khoury, 2012). Currently, most genomic expertise in state and territorial health departments falls within maternal and child health as it relates to NBS issues, however, there now needs to be a greater understanding in chronic disease departments about the

impact of genetics on population health and how to use evidence-based recommendations to implement new practices. Unfortunately, many chronic disease programs across the country are being reduced, are often underfunded, and are not standardized or as comprehensive as they need to be, especially as they relate to genomics (Allen et al., 2013; Maylahn, Fleming, & Birkhead, 2013). Moreover, it is agreed that a majority of public health professionals have not been educated adequately on genomics, whether through public health education or on-the-job training (Department of Health and Human Services, 2011; Marzuillo et al., 2014). It will be important for CDDs to play a leadership role in the integration of genomics into state chronic disease plans, assessment of program effectiveness, education of their workforce and the communities they serve, and initiation and facilitation of collaborations with stakeholders.

By 2010, all states had a Comprehensive Cancer Control (CCC) plan in place and one study found a significant increase in genomics-related terms in these plans from 2005-2010 (Laufman et al., 2012). These CCC plans included goals and strategies related to FHH, public and provider awareness of genetics and genomics (education), breast cancer referrals, access to genetic services, support and expansion of partnerships, development and promotion of screening (diabetes), and increased research funding (Alzheimer's disease). The increase in genomic activities could have been in response to the Healthy People 2020 goals and/or evidence-based recommendations for cancer genomics; however, this study found that genomics still hasn't grown as a priority at most state levels (Laufman et al., 2012). Although evidence-based public health genomic strategies have been shown to have a significant impact on population health, the

commitment for genomics at the state level has been slow due to a shortage of organizational leadership and support, lack of understanding, and limited resources for competing priorities (Allen et al., 2013)

Currently there a small number of model states doing work in the area of chronic disease public health genomics and most are accomplishing their goals in small increments with minimal funding. That being said, the role of state public health departments, specifically chronic disease units, in light of the Healthy People 2020 objectives, Precision Medicine Initiative, and Tier 1 recommendations can and should increase to meet these imperatives. Providing evidence of successful programming and studying models that work could help obtain funding from sources who may benefit from such integration.

Challenges and Opportunities

Implementation Barriers

Since the completion of the Human Genome Project in 2003, the fields of genetics and genomics have developed rapidly, yet translation "from bench to bedside" and more so from "bench to community" has been a slow process (Cornel, Van El, & Borry, 2014). Studies show that the translational process, from research evidence to clinical practice, is 17 years, however, only 14% of all discoveries actually make it there (Khoury et al., 2007). Calculating from the date of gene discovery of the Tier 1 tests, FH (1985), LS (1987), and HBOC (1995), places the translational timeline to clinical and public health application as 2002, 2004, and 2012 respectively (Brown & Goldstein, 1986; Krainer et al., 1997; Lynch et al., 2009; Morris, Wooding, & Grant, 2011). As was discussed

previously, clinical practice guidelines and public health recommendations have been in place for some time, yet clinical medicine and public health practitioners are slow to adopt suggested practices.

An expected reason for slow adoption is funding. Ninety-eight percent of genomic funding is in the research discovery phase and "bench to bedside" applications while < 2% is devoted to population translation and outcomes research (Khoury, Gwinn, Bowen, & Dotson, 2012; Laurence, 2012). Lack of evidence and data showing health outcomes makes it hard to advocate for genomics program funding in the states and outcome data from model states is limited. Ironically, governmental support is often based on translational research data and translational research cannot be accomplished without governmental support (Modell et al., 2014). Moreover, many state chronic disease programs have been reduced including a 57% reduction in state funding by the CDC from 2013–2014 (Allen et al., 2013; Khoury et al., 2011; Maylahn et al., 2013). These limited resources along with a lack of organizational support and leadership due to competing priorities and an opinion that genomics is a low-yield investment compared to current practices will certainly slow or hinder state adoption rates (Allen et al., 2013; Khoury et al., 2007). Without strong health data analysis to encourage these evidence-based intervention (EBI) applications, it will be difficult to encourage states to move beyond their current chronic disease practices.

Other barriers to genomic translation in state chronic disease departments is the lack of awareness and education by public health practitioners, health care providers, and the public (American Public Health Association, 2013; Williams, 2012). Many of the

model states have included genomics educational assessments and/or programs to increase knowledge and awareness about genomics and recommended health practices (St Pierre et al., 2014; Trivers et al., 2015). Reimbursement for Tier 1 recommendations and other genetic services also limits what states are willing to support (Williams, 2012). Cancer genomics is not a mandated public health program like NBS and, without a nationalized healthcare system, reimbursement will be dependent on the patient and their insurance availability (which may vary widely or depend on the recommendation level) (Bowen et al., 2012). Finally, the limitations of our current electronic health records system to collect, analyze, and store the large volumes of data could impact health outcomes data (Williams, 2012). As this is one of the goals of the Precision Medicine Initiative, this data should be easier to ascertain once a better system is in place.

The Need for Collaboration

Implementation of public health genomics is difficult and the need for collaboration within state public health departments and external stakeholders is the key to success (Genetic Alliance, 2014; OPHG, 2011). State public health departments are in a unique position to foster these collaborations and mobilize partnerships that will ensure a competent public health and clinical medicine workforce as well as assure accessible and quality genetic services (Cragun et al., 2016). Because public health departments are also the only ones who have the legal authority to collect population data in some jurisdictions, they will need to lead the accumulation of this information and tabulate health outcome figures (Maylahn et al., 2013). The public health, clinical medicine, and genetic service providers' connection needs to be especially strong to assure individual

patients and susceptible populations have the information about and access to pertinent genetic services. Other important external stakeholders reside in hospitals, academic centers, local public health organizations, and advocacy organizations (Laufman et al., 2012). Advocacy organizations are especially important because they help to establish buy-in from the public and other organizations (Modell et al., 2016).

Some states have a state genetics coordinator who's scope of practice is beyond NBS (Coalition of State Genetics Coordinators, 2007). Success in novel public health interventions implementation has been tied to strong leadership and champions who are passionate about the program (Milat, Bauman, & Redman, 2015). All of the model states who have accomplished strides in public health genomics have someone who works part or full-time in that capacity at the state level or at an associated academic university. This individual can and should be the leader of the coordination, collaboration, and communication of state public health genomic services. In addition, collaboration should occur between public health agencies at the national, state, and local level as well as regional collaboratives to share ideas and challenges (Alexander, Keehn, Kaye, & O'Leary, 2016; Bowen et al., 2012).

Translating genomic advances from the discovery phase to population implementation and subsequent improvement in health outcomes is not a small task. Many barriers exist including funding, awareness, knowledge, competing priorities, reimbursement, and lack of organizational support. In order to overcome these barriers, state public health professionals need to collaborate with other stakeholders to assure these advances are accessible to the populations they serve.

Summary and Conclusions

With fully validated and clinical practice guidelines for screening of Tier 1 tests in place, implementation of screening programs for individuals and their family members at risk for HBOC, LS, and FH is ready to be launched through collaborations by public health agencies, clinical medicine practitioners, and advocacy groups (Modell et al., 2016). Because prevention of population morbidity and mortality is a key public health endeavor, Tier 1 genetic testing and cascade screening of family members illustrates how family health history can be modifiable. State chronic disease departments will be instrumental in the delivery of these programs through the formation of strong partnerships with many different sectors of the communities they serve.

CDDs are poised to be the leaders of the dissemination and coordination of these new health promotion practices while assuring a focus on the needs of underserved populations (American Public Health Association, 2013; Senier et al., 2015). Because of this, they will need to understand and help facilitate implementation of Tier 1 genetic testing recommendations. Rogers's diffusion of innovations theory explains that communication and influence are impactful to the rate of adoption; encouraging states to have an individual who can be the champion for chronic disease genomics would help more states adopt this new innovation.

In Chapter 3, I will outline the methods that were used to perform this study. This will include a description of the instrument, participants that were studied, and procedures for data collection, coding, and analysis. The analysis will be specifically

explained through a detailed presentation of the specific variables and statistical tests to be used.

Chapter 3: Research Methods

Introduction

Following Tier 1 genetic screening and testing recommendations, leaders in state and territorial health departments will be called upon to coordinate, collaborate, and communicate these initiatives in their areas (Green et al., 2015). This is especially true for CDDs, who oversee the areas touched by these recommendations for breast cancer, colon cancer, and cardiovascular health. The main purpose of this study was to determine whether certain state genomic activities or current genomics funding is associated with the level of knowledge and interests in genomics by state and territorial CDDs. This could lead to identification of state activities which may help CDDs to be more informed and prepared to lead Tier 1 testing and screening in their states. These results may also shed light on present knowledge and interests by CDDs providing opportunities and insight on challenges for Tier 1 program implementation in the states and a baseline for future research. The purpose of this chapter will be for me to describe the research questions and hypotheses, survey instrument used, participants, study variables, procedures for data collection and coding, and specific data analysis techniques. Finally, I will also define potential threats to the validity of the study.

Research Design and Rationale

In this quantitative research study, I employed a nonexperimental, cross-sectional, correlational survey design to investigate whether there is an association between current state funding in genomics and/or specific state genomic activities and the level of knowledge and interests in genomics by current CDDs in all U.S. states and territories.

This study was initiated by a subcommittee of the APHA GFPC, of which I am a member, in order to determine the current status of state CDDs in regards to readiness for implementation of Tier 1 testing and identify opportunities and challenges to that end. The committee agreed that I could perform a secondary analysis of the results of this survey to answer the research questions about the possible connection between state genomics activities and CDDs knowledge and interests in genomics.

The survey committee also determined that a quantitative, web-based, self-administered questionnaire format would be a quick, efficient, and cost-effective design to obtain the information sought (see Frankfort-Nachmias & Nachmias, 2008). Using a quantitative, cross-sectional survey would allow me to collect important information from the study group at a single point in time and delivery via a free web-based service that would eliminate costs. The other committee members and I were conducting this study voluntarily with no outside funding. Because this is a new instrument developed by the GFPC subcommittee members, there was no known reliability or validity at the time of this study. Content validity was determined based on the expertise of the committee members and other experts in areas which were thought to impact state Tier 1 implementation. A pilot study of the survey was not performed before delivery to the CDDs.

Quantitative surveys have been used in the past to identify issues in public health services delivery (Jacobs, Dodson, Baker, Deshpande, & Brownson, 2010; Stamatakis et al., 2012; Wilcox et al., 2014). Survey response rates have also been studied showing that multiple methods (mail, Internet, telephone), incentives, and follow-up attempts can

impact response rate (Cho et al., 2013; Dillman, 2015; Millar & Dillman, 2011; Pit, Hansen, & Ewald, 2013). Moreover, the advent of personal hand-held devices also seems to be having a negative impact on the completion of surveys (Stern, Bilgen, & Dillman, 2014). Because this was a project taken on voluntarily by the GFPC subcommittee (four members with no financial support), there was no availability for incentives, mailings, or telephone calls. The subcommittee decided to deliver the survey via e-mail link to the Internet with a 6-week timeline during which two follow up e-mail requests would be made. The benefit of using the Internet includes lower costs, decreased time, and easier data entry and analysis (Ahern, 2005).

The Chronic Disease Directors Survey consisted of 16 dichotomous, Likert scale, limited contingency, and demographic questions to determine age, educational degree, and state or territory (see Appendix). Some questions included a response of "Don't know" if respondents were unable to accurately answer the question. The first half of the survey (nine total questions) contained inquiries about the extent of each state's activities in genomics. The second half of the questionnaire was comprised of questions to gauge the participants' knowledge and interests of genomic topics specifically as they relate to chronic disease (four total questions, 18 different topics). Three of these questions were based on a Likert scale and were, therefore, used to determine the *level* of knowledge and interests in genomics (1 (very poor) – 5 (very good)) rating of knowledge of Tier 1 conditions; 1(strongly disagree) – 5(strongly agree) of agreement with genomic statements and interest in genomic topics.

Methodology

Population

The selected population for a given methodology is the "aggregate of all cases that conform to some designated set of specifications" (Maruyama & Ryan, 2014, p. 231). For this study, participants were CDDs from all U.S. states, territories (Guam, Marshall Islands, Micronesia, Palau, Puerto Rico, Samoa, and U.S. Virgin Islands), and the District of Columbia ($N = 58$). This was a select group with a defined number of participants. I was provided with e-mail contact information for the directors and delivery of the survey link by the NACDD located in Atlanta, GA (NACDD, 2016b). Founded in 1988, the NACDD is a nonprofit, public health organization dedicated to supporting CDDs in each state and territory by connecting over 6,000 chronic disease practitioners to create partnerships, develop policies, implement programs, and share knowledge about chronic disease prevention and health promotion (NACDD, 2016a).

Sampling and Sampling Procedures

In this study, I employed a simple random convenience sampling of all CDDs in the United States and territories ($N = 58$), and each director had an equal chance to complete the survey. The survey was disseminated to these directors through their employee e-mail. I conducted a G*power analysis for the sample size using the *t*-test difference between two dependent means (matched pairs), two tailed, with a medium effect size (0.2), 0.05 α, and 0.80 power, which yielded a sample size of 199. As this was much greater than the total sample of the population, sample size analysis could not be used in this case (see Faul, Erdfelder, Buchner, & Lang, 2009).

Procedures for Recruitment, Participation, and Data Collection

In this study, I used data that was originally collected from a quantitative survey instrument delivered via the Qualtrics survey platform to the membership of the NACDD ($N = 58$) from February 11, 2016 through March 31, 2016(Qualtrics, 2015). Each participant had an equal chance to voluntarily complete the survey with no monetary or other compensation provided for doing so. One of the GFPC members provided access to the Qualtrics platform through the University of Michigan Medical School Information Services with a specific survey link for respondents to connect to the survey (https://umichumhs.ut1.qualtrics.com/jfe/form/SV_d6IJGrKYOzZZDb7). The other committee members and I obtained access to this dataset from this committee member.

The CEO of NACDD and the NACDD policy chair were our points of contact in the organization, authors of the cover letter, and distributors of the survey link via e-mail. The cover letter explained the purpose and importance of the survey to chronic disease public health genomics, encouraged participation, and assured confidentiality of individual results (see Frankfort-Nachmias & Nachmias, 2008). This method was employed for two reminder e-mails (sent on February 25 and March 21, 2016) during the survey period.

Instrumentation and Operationalization of Constructs

The Qualtrics survey platform allowed for coding of the survey questions within the program, automatically assigning a quantitative answer choice value to each selection within a question (first answer choice = 1, second choice = 2, etc.). The program also allowed for variable naming and assignment of a label for each question. In order to

measure the variables, I named variables by the question number (Q1, Q2, etc.) and identified those with subtopics (e.g. Q9_1, Q9_2, etc.). All of this information was easily downloaded into SPSS for analysis. When this survey was originally entered into the Qualtrics system, each question was given a number; however, after committee input, some questions were relocated on the survey and the numbers were not reordered in the Qualtrics system. Therefore, the final survey questions did not follow in numerical order, but were identified correctly in SPSS. The Appendix shows the final survey and coding scheme that I used.

In this survey, I assigned the responses for state genomic activities a number (1 = Yes, 2 = No, 3 = Don't know) for all questions except for the frequency of collaborations, which was based on a Likert scale (1 = In the past quarter, 2 = In the past year, 3 = Rarely, 4 = Never, 5 = Never but potentially in the future). Additionally, question subtopics were also coded 1–5 (Q8), 1–7 (Q9), 1–4 (Q13), and 1–9 (Q25) to correspond to each subtopic from top to bottom. Except for the question regarding awareness of contact information for clinical genetic services (Yes/No), a Likert scaling method was used to identify the varying levels of knowledge and interest in genomics by CDDs on a 5-point continuum (knowledge of Tier 1 conditions rated 1–5 (Q10); agreement with genomic statements (Q11) or interest in genomic topics (Q12) rated 1 = Strongly disagree, 2 = Disagree, 3 = Neither agree nor disagree, 4 = Agree, 5 = Strongly agree, 6=Don't know (Q11) or 6 = We already do this (Q12). Each Likert scale response was given a quantitative number to correspond with the result and this had a direct correlation to the level of knowledge and interest in genomics of each survey participant; the higher

the number, the greater level of knowledge and interest and vice versa. Responses of "Don't know" were identified and removed as outliers during initial statistical analysis and then run again to include these results to examine their potential impact.

The purpose of this study was to determine possible associations between state genomic funding as well as specific state genomic activities and the level of knowledge and interests in genomics by state and territorial CDDs. Because of this, I considered each variable potentially dependent on the other. In other words, no variable was considered as an independent or causative variable. All questions in this survey were categorical (nominal); however, Likert scaled questions (Q22, Q23, Q24, and Q25) were converted to continuous values (1–5 or 6) for statistical analysis to allow for determination of a level of knowledge, interest, or frequency of collaboration by each respondent and the study group as a whole; the higher the number, the greater the level of knowledge and interest except for levels of collaboration, which employed a reverse numbering scheme (see Appendix).

I considered multiple choice questions with a response of "Yes" (1) present and responses of "No" (2) or "Don't know" (3) were considered absent initially. "Don't know" responses for state activities were changed to (2) to analyze them as not present. These results were then returned to their original states to analyze the impact of the "Don't know" responses. Finally, the "Don't know" response for Q23 (agreement with genomic statements) was removed (* in dataset), so it would not be analyzed and impact the mean value of interest by the CDDs. Tables 5 and 6 explain the variable types and

which statistical tests that I used for each association. Finally, I tested data validity and reliability with exploratory analysis and Cronbach's alpha.

Research Question 1 Variables

I identified the following variables and subvariables for Research Question 1:

1. State funding for chronic disease genomics
2. Level of knowledge and interests in genomics by CDDs

   Subvariables:

   2a. Awareness of contact information for clinical genetic services

   2b. Knowledge of Tier 1 recommended conditions

   2c. Agreement with genomic statements

   2d. Interest in genomic activities

Table 5

Operationalization of Variables for Research Question 1

Variable pair	Question type	Variables included
State genomic funding	Categorical	Identified states with funding
Knowledge and interest	Categorical Continuous	2a 2b, 2c, 2d

Research Question 2 Variables

I identified the following variables and subvariables for Research Questions 2:

1. State genomic activities.

   Subvariables:

   1a. State genetics needs assessment

   1b. State genetics needs assessment includes chronic disease conditions

1c. Genetics in state action plan

1d. Genetics education

1e. Genomics in BRFSS

1f. Analysis of state cancer registries

1g. Genetic legislation

1h. Frequency of collaborations

2. Level of knowledge and interests in genomics by CDDs

Subvariables:

2a. Awareness of contact information for clinical genetic services

2b. Knowledge of Tier 1 recommended conditions

2c. Agreement with genomic statements

2d. Interest in genomic activities

Table 6

Operationalization of Variables for Research Question 2

Variable pairs	Question type	Variables included
State genomic activities Knowledge and interest	Categorical Categorical	1-2, 1a-2a, 1b-2a, 1c-2a, 1d-2a, 1e-2a, 1f-2a, 1g-2a
State genomic activities Knowledge and interest	Categorical Continuous	1a-2b, 1a-2c, 1b-2b, 1b-2c, 1c-2b, 1c-2c, 1d-2b, 1d-2c, 1e-2b, 1e-2c, 1f-2b, 1f-2c, 1g-2b, 1g-2c
State genomic activities Knowledge and interest	Continuous Categorical	1h-2a
State genomic activities Knowledge and interest	Continuous Continuous	1h-2b, 1h-2c

Data Analysis

All survey data were obtained through the Qualtrics survey platform and directly exported into IBM SPSS version 23.0 software to perform statistical analysis, which is the most appropriate platform for analysis of quantitative survey data (IBM, 2016). The raw data was only available to the committee and myself and first examined for and cleaned of incomplete or duplicate entries or any other potential abnormalities. Assigned variable names and numerical values were also transferred in the statistical report downloaded for data analysis. Numerical values for "Don't know" responses were changed to "2" to assure they were not included in the statistical analysis and treated as "No" (not present) initially for this study. "Don't know" values were then returned to their original numbers to see what impact, if any, they had on the analysis.

Descriptive statistics were performed first to identify means, standard deviations, and range of values for all variables (Creswell, 2009). Construction of frequency distributions looked at response patterns for all variables with nominal questions showing modes and interval questions providing median, mean, range, standard deviation, and coefficient of variation (Frankfort-Nachmias & Nachmias, 2008). Frequencies were converted into percentages for meaningful interpretation and comparison and visually displayed in the results section through tables and graphs.

Research Question 1

To what extent, if any, is there an association between states that have received funding for chronic disease genomics and the level of knowledge and interests in genomics by state and territorial CDDs?

$H_0$1: There is no association between states that have received funding for chronic disease genomics and the level of knowledge and interests in genomics by state and territorial CDDs.

$H_1$1: There is an association between states that have received funding for chronic disease genomics and the level of knowledge and interests in genomics by state and territorial CDDs.

To determine possible associations of the variables in Research Question 1, I used Chi-square analysis, the independent *t* test, and multiple linear regression. Chi-square analysis examined possible associations between current state genomic funding and whether CDDs knew how to contact genetics professionals if they needed to refer a patient or required professional consultation. The independent *t* test was used to analyze whether or not state genomic funding had a possible association with CDDs level of knowledge and interest in genomics determined on a continuous scale. Finally, multiple linear regression determined possible associations using current state genomic funding as the independent variable and the level of knowledge and interest in genomics by CDDs (determined on a continuous scale) as the dependent variable.

Research Question 2

To what extent, if any, is there an association between current state genomic activities and the level of knowledge and interests in genomics by state and territorial CDDs?

$H_0$2: There is no association between any current state genomic activities and the level of knowledge and interests in genomics by state and territorial CDDs.

$H_1$2: There is an association between one or more current state genomic activities and the level of knowledge and interests in genomics by state and territorial CDDs.

Current state genomic activities that were seen as having a potential impact on chronic disease public health genomics program implementation were queried. These included (a) a state genetics needs assessment, (b) a state genetics needs assessment that includes chronic disease conditions, (c) inclusion of genetics in the state public health action plan, (d) genetic educational programs, (e) genomics topics on the BRFSS, (f) analysis of state cancer registries or other vital records data to identify citizens with hereditary cancer syndromes, (g) frequency of collaborations or partnerships with outside entities related to genomics, and (h) presence of legislation and/or regulation specifically related to genomics. To determine the knowledge and interests in genomics by the CDDs, they were asked about their (a) awareness of contact information for clinical genetic services for potential referral or consultation, (b) knowledge of Tier 1 recommended conditions, (c) agreement with genomic statements, and (d) interest in integration of genomic activities.

The examination of possible associations between the variables for Research Question 2 was determined by Chi-square analysis (categorical/categorical), independent *t* test (categorical/continuous), multiple linear regression (continuous/categorical), and Pearson's correlation coefficient (continuous/continuous). Covariation means that "two or more phenomena vary together" (Frankfort-Nachmias & Nachmias, 2008, p. 93) and this was the basis for the analysis. The null hypothesis will either be rejected or retained based on this information. It is important to note that an association of two variables does

not show causation of why the two variables are related through cause and effect (Green & Salkind, 2011).

It could be possible to determine whether the association is directional (e.g., an increase in state genomic activities will cause an increase in knowledge and interests in genomics by CDDs) by selection of a one-tailed test, however, a two-tailed test was used to assure detection of an effect in either direction if it exists (Field, 2013). Significance of association was determined by a *p*-value of < .05 and a medium effect size (coefficient of determination) was set at 0.20 to examine the amount of variability from the relationship of the two variables. Because the sample size was already known in this secondary dataset ($N = 16$), G* Power calculations provide a power (1-β error probability) of 0.116 with 15 degrees of freedom for this study (see Faul, Erdfelder, Lang, & Buchner, 2007).

Threats to Validity

External threats to validity are indicative of the level of generalizability the results have to the population being studied (Creswell, 2009). Due to the small number of CDDs in the United States and territories ($N = 58$), it was critical to obtain as many completed surveys as possible. Sample size ultimately has an effect on the power of the analysis and significance of the results as an indication of the population being studied.

There can be a variety of reasons why the CDDs did not respond to the survey request. Dillman (2015) explains that the age of Internet surveys is similar to the days of telephone surveys when individuals were inundated with phone calls seeking information to help better understand a group of individuals or population. Today, the volume of emails an individual receives at home and at work lends itself to rapid deletion in order to

clean inboxes with only the most crucial information being saved. Completion of surveys for someone else's benefit can be a difficult undertaking especially without an incentive (even altruistic) that would encourage participation.

The most significant error that will likely have an impact on this study will be the nonresponse error from those CDDs that do not respond to the survey. Nonresponse can also be due to a variety of factors including the type of population, the data retrieval method, types of questions, and the number of attempts to get respondents to complete the survey (Frankfort-Nachmias & Nachmias, 2008). Because of the significant bias and lack of generalizability that can be introduced by nonresponders, it was important to get as many responses as possible.

Because this is a correlational study and does not show causation, internal threats to validity are not relevant in this case. Internal threats to validity pertain only to experimental studies, which this is not. Moreover, as this is a new study that has never been performed before; statistical conclusion validity is unknown at this time.

Ethical Procedures

Access to the study population of state and territorial CDDs was facilitated through contact with the CEO and policy liaison of the NACDD. Because this study was originally performed by the APHA GFPC subcommittee and not connected with any organization or university, Institutional Review Board (IRB) approval was not attained prior to the original study; it was performed as an exploratory endeavor and not for research purposes. IRB approval for secondary analysis of the data for this study was obtained through Walden University (02-17-17-0282497). The state and territorial CDDs

are all adult professionals and, therefore, are not considered a part of a vulnerable population for study.

Data for this study were anonymous and had no identifying information that could link the results to the participant who answered the survey. There was information regarding which states replied, however, this will remain confidential and no information regarding individual states' current activities or future plans will be shared. Knowing which states participate was only needed to determine the geographic regions represented and analyze activities and knowledge and interests from states who have received funding against those who have not.

Once the survey results were downloaded from the Qualtrics system by the Genomics Forum Policy subcommittee member who had access, only the other subcommittee members had visibility to these results. Data are kept on secure computers and will be destroyed after 5 years. Findings will be shared with the NAACD leadership and possibly presented in future publications in scholarly journals.

Summary

The purpose of this chapter was to outline the specific steps that occurred in order to conduct this study of state and territorial CDDs to look at whether current genomic funding or certain genomic activities in each state and territory are associated with the level of knowledge and interests in genomics by these study participants. This analysis looked at each state genomic activity to see if one or more had an impact on the level of knowledge and interests in genomics by the CDDs. Furthermore, an analysis of the level

of knowledge and interests in genomics by CDDs from the states that have received funding sought to see if there is an association between these two.

In Chapter 4, I will describe the research findings and data analysis in detail. The chapter will include the data collection procedures, analysis, descriptive statistics, and outcomes as they relate to the research questions and theoretical framework. Finally, I will provide an interpretation of the findings and how consistent they are to this study.

Chapter 4: Results

Introduction

The purpose of this study was to determine whether there is an association between any current state genomic activities or chronic disease genomics funding and the level of knowledge and interests in genomics by state and territorial CDDs. In order to establish this, I analyzed data from a survey of CDDs using various statistical analyses depending on the type of survey question asked to assess possible associations. In this chapter, I will report the results of this quantitative survey of CDDs by first describing the recruitment, time frame, and response rate for this survey before presenting baseline descriptive and demographic statistics of the sample. I will then provide basic univariate analysis to show the variables under review. Finally, I will explain the results of the statistical analysis performed to answer the research question.

Data Collection

Between February 11, and March 31, 2016, all United States and territorial CDDs ($N = 58$) were invited to participate in a voluntary survey regarding genomics. During the 6-week timeframe for the study, two reminder e-mails were sent to all potential participants on February 25 and March 21, 2016. A total of 18 surveys were completed; however, two states submitted two separate surveys, so only one from each state was selected to be in the study. I based the decision about which survey to use on the credentials of the person submitting the survey; the higher credentialed participant was presumed to be the chronic disease director. Also, one participant did not answer the question about which state or territory he or she worked in; however, I was able to use the

geolocation platform from the Qualtrics system to determine what state the response came from. I used a total of 16 completed surveys for analysis with a response rate of 27.6%.

Survey Results

The study sample of CDDs from state and territorial health departments yielded responses from 15 states and one territory and represented all geographic regions of the United States except for the South-West South Central Region (see Figure 1). Six (38%) of the 16 responses were from states previously identified as innovators and early adopters of genomics and that had been provided funding for genomic activities either currently or in the past. The largest majority of CDDs were between 51–60 years of age (40%), while 26.7% were in both the 41–50 and 31–40 age range, and one director was over 60. All respondents had obtained at least a Master of Public Health or other master's degree with six participants also attaining the level of either MD or PhD. One state did not provide information regarding age or level of education (total N=15).

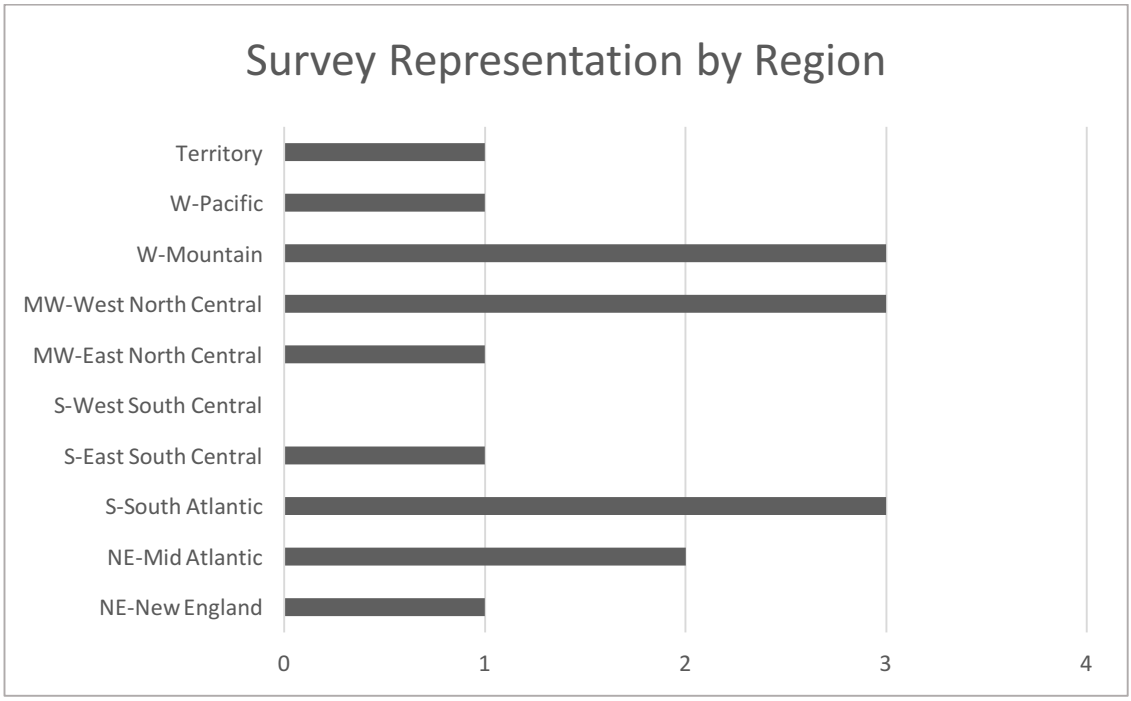

Figure 1. Survey representation by region. United States Census Bureau. (2016a). Regions and Divisions. Retrieved October 24, 2016, from http://www.census.gov/econ/census/help/geography/regions_and_divisions.html 16a).

State Activities

Overall, based on these survey results, I found that there were very few states actively engaging in genomic activities. Of the total number of questions regarding state activities, only 20–30% of respondents' states engaged in less than half of the actions (9/22) considered important in light of Tier 1 genetic testing recommendations. It is also worthy to note that most activities currently being conducted relate to breast and colon cancer and very few are focused on cardiovascular disease or FH. Furthermore, it appears that the same few states are the ones involved in these genomic activities. Figure 2 shows the greatest percentage (more than three states) of state activities being performed as reported by the CDDs.

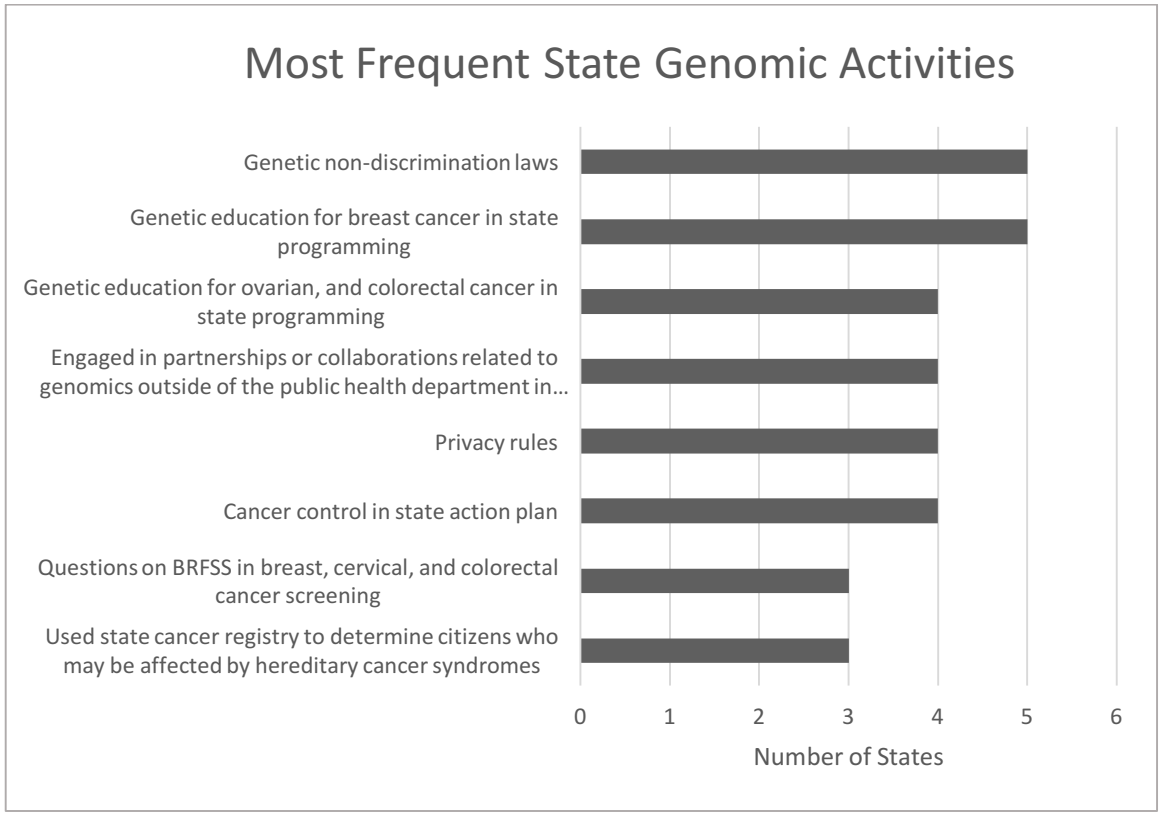

Figure 2. Most frequent state genomic activities currently occurring as reported by CDDs.

Table 7 describes frequencies and percentages of state genomic activities as reported "Yes" by the survey respondents.

Table 7

State Genomics Activities

Activity	Frequency (*N*)	Percentage (%)
Conducted genetic needs assessment	1	6
Genetic needs assessment includes action around chronic disease	1	6
Genetics in state action plan for:		
Chronic disease	2	12
Cancer control	4	25
Cardiovascular health	2	12
Genetic education integrated in:		
Breast cancer	5	31
Colorectal cancer	4	25
Ovarian cancer	4	25
Cardiovascular disease	1	6
Questions on BRFSS		
Breast/cervical cancer screening	3	19
Colorectal cancer screening	3	19
Health care access	1	6
Cancer registry analysis	3	19
Legislation/regulations		
Nondiscrimination laws	5	31
Privacy rules	4	25
Informed consent	1	6
Provision of genetic services to uninsured and low income individuals	1	6

Note. Only "Yes" results are provided in this table due to the quantity of potential answers.

Frequency of Collaborations

In the survey, CDDs were asked how often they engaged in collaborations or partnerships in relation to genomics with groups outside of the state health department. Only 20–30% of CDDs are collaborating with any regularity and it appears that the same 4–5 CDDs have been collaborating across the board. Table 8 illustrates the frequency of collaborations and partnerships occurring with each of these entities.

Table 8

Frequency of Collaborations or Partnerships Related to Genomics with Outside Entities

	In the past quarter		In the past year		Rarely		Never		Never but potentially in the future	
	N	%	*N*	%	*N*	%	*N*	%	*N*	%
Academic institutions	5	31	1	6	4	25	3	19	3	19
Primary care providers	5	31			1	6	7	44	1	6
Genetic counselors	4	25	1	6	3	19	6	38	2	13
Other clinicians	4	25	1	6	3	19	5	31	1	6
Advocacy groups	4	25			2	13	6	38	1	6
Hospitals and healthcare systems	3	19	1	6	2	13	7	44	2	13
Third party payers	3	19	1	6	3	19	6	38	1	6
Local and county health departments	1	6	1	6	3	19	8	50	2	13

Knowledge and Interest in Genomics by CDDs

Knowledge of genetics professionals. CDDs were asked if they knew how to contact genetics professionals in their state/territory if they needed genetic expertise consultation or patient referral for genetic services. Sixty-three percent acknowledged that they would be able to contact genetics professionals if the situation presented itself; the other 37% said they did not. This question did not include a response of "Don't know."

Knowledge of Tier 1 conditions. CDDs were asked to rate their level of knowledge of the recommended Tier 1 conditions: HBOC, LS, and FH. The rating scale was from 1 = very poor to 5 = very good. Knowledge was greatest in HBOC ($M = 3.13$, $SD = 1.46$), and "somewhat poor" for FH ($M = 2.50$, $SD = 1.27$) and LS ($M = 2.13$, $SD = 1.20$).

Agreement with genomic statements. CDDs were given six different genomic statements and asked about their level of agreement with each one. Responses ranged from 1 (strongly disagree) to 5 (strongly agree). A value of 6 (Don't know) was also an option; however, these results were removed when calculating the mean values to avoid skew. Table 9 represents the results from each of these statements.

Table 9

Agreement with Genomic Statements Regarding Importance of Genomics

To what extent do you agree or disagree with the following statements?	*N*	*M*	Range	*SD*
Genetic counseling and testing for hereditary cancer conditions can improve a patient's health outcomes	16	4.00	3–5	0.73
Integrating genetics into public health planning for chronic disease programming would benefit residents of our state.	16	3.88	1–5	1.09
Genetics is an important component of public health initiatives	15	3.73	3–5	0.80
Legal protections against genetic discrimination are adequate in our state.	8	3.13	2–4	0.64
As a whole, staff in the Chronic Disease Program understands how genetics relates to chronic disease	15	2.87	1–4	1.19
Citizens in our state understand how family history or genetics influences risk of chronic disease	15	2.67	1–4	1.18

Note. Results greater than 3.00 were considered positive agreement. Results of 6 (Don't know) were removed to avoid skew.

Level of interest in genomic activities. Providers were questioned on nine activities considered important to genomics integration and asked how strongly they agreed or disagreed with their existence. Responses ranged from 1 = strongly disagree to 5 = strongly agree. An option of 6 (We already do this) was included in mean value calculation as it supports agreement of the importance of an activity. Table 10 provides the results of the level of agreement for each statement.

Table 10

Level of Interest in Incorporating Genomic Activities

In my role as a CDD, I would be interested in:	*N*	*M*	Range	*SD*
Incorporating genomics into our comprehensive action plan	15	4.33	2–6	1.23
Incorporating genomics into other cancer policies and initiatives	15	4.20	1–6	1.37
Utilizing Cancer Registry data to identify high risk patients with the goal of reducing morbidity and mortality	15	3.93	3–5	0.70
Promoting or enhancing genomics awareness among medical providers	15	3.93	3–5	0.70
Incorporating cancer genomics into our state's Breast and Cervical Early Detection Program	14	3.86	2–5	0.95
Promoting or enhancing genomics awareness among the general public	15	3.73	1–5	0.96
Finding funding to hire an individual to focus on genomics and chronic disease programming	15	3.53	1–5	1.25
Incorporating ID of individuals/cascade screening for FH into Cardiac Disease Prevention Program	15	3.40	1–5	0.91
Recommending the addition of genomics questions to the BRFSS cancer modules	15	3.33	1–5	1.11

Note. Results greater than 3.00 were considered a positive interest. Results included a value of 6 (We already do this) as an indication of a positive interest.

Data Analysis Results

I performed statistical analysis to determine possible associations on all variables provided through the survey results as well as evidence of state genomic funding. Chi-square, *t* test, ANOVA, multiple regression, and Pearson's correlation were used depending on the type of variables examined. I also performed analysis in duplicate, once with the "Don't know" responses included and again after changing these results to "No." This was to acknowledge the "Don't know" responses as possible presence of an activity and then to recognize that the "Don't know" responses could mean the activity did not exist.

Research Question 1

Research Question 1: To what extent, if any, is there an association between states that have received funding for chronic disease genomics and the level of knowledge and interests in genomics by state and territorial CDDs ?

$H_0$1: There is no association between states that have received funding for chronic disease genomics and the level of knowledge and interests in genomics by state and territorial CDDs.

$H_1$1: There is an association between states that have received funding for chronic disease genomics and the level of knowledge and interests in genomics by state and territorial CDDs.

Chi-Square

Part of Research Question 1 was analyzed using Chi-square analysis to determine a possible association between state genomic funding and the level of knowledge and

interests in genomics by the directors. An assumption of the Chi-square test is that all results are independent of one another and will only provide one piece of data to one cell of the contingency table; no data will be used repeatedly (Field, 2013). Moreover, expected frequencies should be no lower than 5, which could be troublesome with a small sample size, and could have a large impact on test power.

This analysis specifically looked at whether or not a state had been identified as a funded state and if the CDD knew how to refer patients for genomic services or find expert genomic consultation in their state (Q16). Chi-square analysis showed no association with these variables ($X^2(1)=.071$, $p=.790$, phi=.067; Likelihood ratio $X^2(1)=.072$, $p=.789$, phi=.067). These results, therefore, accept the null hypothesis for Research Question 1 and indicate there is no association between these variables.

Independent *t* test

Part of Research Question 1 was also analyzed by the independent *t* test to see if funding was associated with CDDs' level of knowledge about Tier 1 conditions, interest in genomic topics, and agreement with genomic statements (Q22_1–3, Q23_1–6, Q24_1–9). The independent *t* test is a parametric test that assumes normality of the sampling distribution, outcome variables are related linearly to predictor variables, and the samples will come from a population with the same variance regardless of the level of predictor variable (Field, 2013). The final assumption is that the samples are all independent of one another. This analysis again accepted the null hypothesis for Research Question 1 and showed no association with these variables (see Table 11).

Table 11

Presence of State Genomic Funding vs. Knowledge or Interest of Genomic Topics

Knowledge or Interest	*t*	*p* value	95% CI
HBOC	*t*(14) = 1.17	.263	-.726 – 2.46
LS	*t*(14) = 2.00	.066	-.085-2.35
FH	*t*(14) = .807	.433	-.884-1.95
Citizens understand genomics and chronic disease	*t*(11) = -.693*	.502	-1.67 - .865
Staff understands genomics and chronic disease	*t*(13) = -.519	.613	-1.72 – 1.06
Genetic counseling improves outcomes	*t*(14) = .695	.499	-.557 – 1.09
Integrating genetics benefits state residents	*t*(14) = .822	.425	-.751 – 1.69
Legal protections are adequate	*t(*6) = -.685	.519	-1.53 - .858
Genetics is very important to public health	*t*(13) = .221	.829	-.879 – 1.08
Add BRFSS genomics to cancer modules	*t*(13) = .158	.877	-1.27 – 1.47
Use of cancer registry to ID at-risk individuals	*t*(13) = 1.04	.317	-.430 – 1.23
Incorporating genomics into cancer action plan	*t(*13) = 1.04	.318	-.757 – 2.16
Incorporating genomics in other cx policies	*t*(13) = 1.22	.245	-.698 – 2.50
Incorporating genomics into breast/cervical cancer early detection	*t*(12) = .406	.692	-.970 – 1.42
Incorporating ID of FH in cardiac screen	*t*(13) = .587	.567	-.803 – 1.40
Promote public awareness	*t*(13) = .183	.857	-1.08 – 1.28

(table continues)

Knowledge or Interest	*t*	*p* value	95% CI
Promote provider awareness	*t*(13) = .251	.806	.762 - .962
Funding state genetics coordinator position	*t(*13) = .572	.577	-1.11 – 1.91

Note. CI = Confidence Interval. Negative *t* values suggests the sample mean was below the hypothesized mean (see Field, 2013).
*Levine's statistic was significant; therefore, *t* statistic is from variances not assumed.

Multiple Linear Regression

Finally, part of research question one was also analyzed by multiple linear regression to examine the "flip" of the independent *t* test. This analysis looked at funding as the independent, categorical variable, and knowledge and interest in genomics by the CDDs as the dependent, continuous variable (Q22_1–3, Q23_1–6, Q24_1–9). The linear model assumes that the outcome variable is linearly related to the predictor variable, the samples have a constant variance, and are normally distributed (Field, 2013). Predictors should also be independent and uncorrelated to any external variables or linear to another predictor. This analysis provided a single association, rejecting the null hypothesis for Research Question 1, with funding and CDDs agreement with the statement "Citizens in our state understand how family history or genetics influences risk of chronic disease" $F(1,13) = 16.20$, $p = .028$.

Research Question 2

Research Question 2: To what extent, if any, is there an association between current state genomic activities and the level of knowledge and interests in genomics by state and territorial CDDs ?

$H_0$2: There is no association between any current state genomic activities and the level of knowledge and interests in genomics by state and territorial CDDs.

$H_1$2: There is an association between one or more current state genomic activities and the level of knowledge and interests in genomics by state and territorial CDDs.

Chi-Square

This analysis and looked at possible associations between state genomic activities and knowledge and interest in genomics by CDDs. Specifically, this analysis looked at presence or absence of state genomic activities (Q11, 29, 13_1–4. 8_1–5, 9_1–7, 10, 15_1–5) and whether the chronic disease director knew how to refer patients for genomic services or find expert genomic consultation in their state (Q16). The first analysis included the "Don't know" results and identified seven state genomic activities that were associated with referral and consultation knowledge by the CDDs. Table 12 depicts the results that reject the null hypothesis for Research Question 2 and are significant associations. Note that only two activities had a significant Pearson Chi-square and all other results were based on the Likelihood ratio, which is an alternative to Pearson's (Field, 2013). Also, one of the significant Pearson results (Education – Other) only had a sample size of four.

Table 12

Presence of State Activity vs. Knowledge of Genomic Referrals or Consultations- "Don't Know" Included

SGA	Pearson's Chi square	Likelihood ratio
Action plan - cancer control		$X^2(2, N = 16) = 7.71, p = .021$, phi = .60
Action plan - other		$X^2(1, N = 14) = 4.39, p = .036$, phi = - 47
Education - breast cancer		$X^2(2, N = 16) = 6.26, p = .044$, phi = .54
Education - other	$X^2(1, N = 4) = 4.00, p = .046$, phi = -1.00	$X^2(1, N = 4) = 5.55, p = .019$, phi = -1.00
Regulations - discrimination	$X^2(1, N = 16) = 4.36, p = .037$, phi = .52	$X^2(1, N = 16) = 6.01, p = .014$, phi = .52
Regulations - privacy		$X^2(1, N = 16) = 4.53, p = .033$, phi = .45
Regulations - genetic services		$X^2(2, N = 16) = 7.71, p = .021$, phi = .62

Note. SGA = State genomic activity

Table 13 provides results of the same analysis; however, the "Don't Know' results were changed to "No" in the data set. This analysis provided six different associations including two Pearson Chi-square results. The following state genomic activities were found signficantly associated with knowledge of genomic referral or consultation and reject the null hypothesis for Research Question 2.

Table 13

Presence of State Activity vs. Knowledge of Genomic Referrals or Consultations "Don't Know" Changed to "No"

SGA	Pearson's Chi square	Likelihood ratio
Action plan - cancer control		$X^2(1, N = 16) = 4.53, p = .033$, phi = .45
Education – breast cancer	$X^2(1, N = 16) = 4.36, p = .037$, phi = .52	$X^2(1, N = 16) = 6.01, p = .014$, phi = .52
Education – colorectal cancer		$X^2(1, N = 16) = 4.53, p = .033$, phi = .45
Education – ovarian cancer		$X^2(1, N = 15) = 5.03, p = .025$, phi = .49
Regulations - discrimination	$X^2(1, N = 16) = 4.36, p = .037$, phi = .52	$X^2(1, N = 16) = 6.01, p = .014$, phi = .52
Regulations - privacy		$X^2(1, N = 16) = 4.53, p = .033$, phi = .45

Note. SGA = State genomic activity

Independent *t* test

The independent *t* test was used to identify a possible association between the presence or absence of state genomic activities (Q11, 29, 13_1–4. 8_1-5, 9_1–7, 10, 15_1–5) and the level of chronic disease director knowledge of Tier 1 conditions, interest in genomic topics, and agreement with genomic statements (Q22_1–3, Q23_1–6, Q24_1–9). This test was also used to study potential associations between frequency of genomic collaborations (Q25_1–9) and whether the CDD knew how to refer patients for genomic services or find expert genomic consultation in their state (Q16). Tables 14 and 15 represent the significant associations that reject the null hypothesis for Research Question 2. Note that some results had a statistically significant Levine's statistic, which means that the sample variances may not be equal. In others, no Levine statistic was provided

and the t value was negative suggesting that the sample mean is below the hypothesized mean (Field, 2013).

Table 14

Frequency of Genomic Collaborations vs. Knowledge of Genomic Referrals or Consultations

Collaborations	*t*	*p* value	95% CI
Primary care providers	*t*(10) = -3.71*	.004	-3.16 - -0.80
Other clinicians	*t*(12) = -3.31*	.006	-2.95 - -0.61
Advocacy groups	*t*(9) = -3.68*	.005	-3.15 - -.075
Hospital/ health systems	*t*(12) = -2.76*	.018	-2.51 - -0.29
Third party payers	*t*(11) = -3.83*	.003	-2.76 - -0.75

Note. CI = Confidence Interval. All *t* values were negative and suggests the sample mean was below the hypothesized mean (Field, 2013).
*Levine's statistic was significant; therefore, *t* statistic is from variances not assumed.

Table 15

State Genomic Activities vs. Level of Knowledge or Interest of Genomic Topics "Don't Know" Changed to "No"

SGA	Knowledge or interest	*t*	*p* value	95% CI
Action plan – cardiovascular health	LS	*t*(13) = = 7.81*	.000	1.55-2.74
	FH	*t*(13) = 5.33*	.000	1.02-2.41
	Staff understands genomics and chronic disease	*t*(12) = 3.90*	.002	0.59-2.02
	Legal protections are adequate	*t*(6) = -2.47	.049	-2.56- -.009
Education – breast cancer	LS	*t*(14) = 2.97	.010	0.43-2.69

(table continues)

SGA	Knowledge or interest	*t*	*p* value	95% CI
Education – colorectal cancer	LS	*t*(14) = 3.48	.004	0.70-2.96
Education – ovarian	LS	*t*(13) = 2.38	.033	0.13-2.61
Education – cardiovascular disease	Legal protections are adequate	*t*(6) = - 2.47**	.049	-2.56 --.009
BRFSS questions– health care access	Legal protections are adequate	*t(*6) = - 2.47**	.049	-2.56 --.009
Regulations – privacy	LS	*t*14) = 2.51	.025	0.22-2.78
	Interest in FH screen	*t*(13) = 2.26	.042	.050-2.28
	Promoting provider awareness	*t*(13) = 2.31	.038	.059-1.78

Note. CI = Confidence Interval. Negative *t* values suggest the sample mean was below the hypothesized mean (see Field, 2013) SGA = State genomic activity.
*Levine's statistic was significant; therefore, *t* statistic is from variances not assumed.
** No Levine statistic was provided.

ANOVA

Analysis of variance was used to analyze the same previous set of data (presence or absence of state genomic activities (Q11, 29, 13_1–4. 8_1–5, 9_1–7, 10, 15_1–5) and the level of chronic disease director knowledge of Tier 1 conditions, interest in genomic topics, and agreement with genomic statements (Q22_1–3, Q23_1–6, Q24_1–9) only with inclusion of the "Don't know" responses. ANOVA was required to analyze this association because the factor (state genomic activity) is now in three groups. The one-way ANOVA is performed with three assumptions; the dependent variable is normally distributed, the population from which the dependent variable samples come from have equal variances, and the sample set is random and provided independent observations

(Green & Salkind, 2011). Table 16 provides the results of this analysis and significant associations that reject the null hypothesis for Research Question 2.

Table 16

State Genomic Activities vs. Level of Knowledge or Interest of Genomic Topics "Don't Know" Included

SGA	Knowledge or interest	*F*	*p* value
Genomics needs assessment	Recommend genomics in BRFSS cancer modules	$F(1,13) = 16.26$	.001
	Incorporating genomics into cancer policies	$F(1,12) = 4.82$	.048
	Promoting public awareness	$F(1,13) = 5.89$	.030
Genomics needs assessment & chronic disease	Recommend genomics in BRFSS cancer modules	$F(1,11) = 5.58$	.038
Action plan – cardiovascular health	LS	$F(2,13) = 3.85$	.049
Education – breast cancer	LS	$F(2,13) = 5.97$	.015
	FH	$F(2,13) = 13.33$	.001
	Interest in FH screen	$F(2,12) = 5.06$	.025
Education – colorectal cancer	LS	$F(2,13) = 12.61$	.001
	FH	$F(2,13) = 4.33$	.036
	Interest in FH screen	$F(2,12) = 4.71$	.031
Education – ovarian cancer	LS	$F(2,12) = 7.50$	.008
	FH	$F(2,12) = 4.12$	.043
BRFSS questions– breast/cervical cancer screening	Legal protections are adequate	$F(2,13) = 4.66$	.030

(table continues)

SGA	Knowledge or interest	*F*	*p* value
	Genetics is very important in public health	$F(2,13) = 4.02$	.044
BRFSS questions– cardiovascular health	Genetics is VIP in PH	$F(1,14) = 7.21$	.018
BRFSS questions – colorectal cancer	Legal protections are adequate	$F(2,13) = 3.97$	.045
BRFSS questions– genetic discrimination	Genetics is very important in public health	$F(1,14) = 7.21$	.018
BRFSS questions-privacy	Genetics is very important in public health	$F(1,14) = 7.21$	.018
BRFSS questions direct to consumer ads	Genetics is very important in public health	$F(1,14) = 7.21$	.018
Use of cancer registry	Legal protections are adequate	$F(2,13) = 4.01$	.044
Regulations – privacy	LS	$F(1,14) = 6.30$	.025
	Legal protections are adequate	$F(1,14) = 7.96$	.014
	Interest in FH screen	$F(1,13) = 5.07$	.042
	Promoting provider awareness	$F(1,13) = 5.33$	.038
Regulations – funding state genetics coordinator	Funding state genetics coordinator position	$F(1,13) = 5.43$	.037
Regulations – access to genetic services	Promoting provider awareness	$F(2,12) = 6.24$	.014
	Funding state genetics coordinator position	$F(2,12) = 4.03$	.046

Note. SGA = State genomic activity

Multiple Linear Regression

Multiple linear regression was used to analyze continuous and categorical variables from the survey much like the independent t-test for possible associations between categorical state genomic activities (Q11, 29, 13_1–4. 8_1–5, 9_1–7, 10, 15_1–5) and the level of chronic disease director knowledge of Tier 1 conditions, interest in genomic topics, and agreement with genomic statements (Q22_1–3, Q23_1–6, Q24_1–9). Responses of "Don't know" were included. This analysis resulted in no associations and accept the null hypothesis for Research Question 2.

This analysis was also used to identify potential associations of frequency of genomic collaborations and CDD knowledge of genomic referrals and consultations. This again, was a reverse analysis of the independent t-test performed earlier. This analysis found one association with primary care providers $F(1,15) = 7.71$, $p = .039$; the multiple correlation coefficient was 0.78. This was the only significant association to reject the null hypothesis for Research Question 2.

Pearson's Correlation Coefficient

The Pearson's correlation coefficient was used to analyze possible associations with survey questions that were both continuous (State genomic activity – Q25 – frequency of collaborations; KI - Q22_1–3, Q23_1–6, Q24_1–9 - level of CDD knowledge of Tier 1 conditions, interest in genomic topics, and agreement with genomic statements). Only collaborations in the past quarter or past year were included as they suggest more frequent partnerships. Pearson's r is significant if the two variables are linearly related (Green & Salkind, 2011). This test assumes that each variable is

normally distributed while ignoring the other variable and is normally distributed at all levels of the other variable. Another assumption is that all variables are sampled randomly and independent of one another. Table 17 presents all signifcant results from the Pearson correlation coefficient analysis that reject the null hypothesis for Research Question 2.

Table 17

Frequency of Collaborations vs. Level of Knowledge or Interest of Genomic Topics

Collaboration	Knowledge or Interest	*r*	*p* value
Academic institutions	HBOC	$r(14) = .59$	0.016
	LS	$r(14) = .54$	0.031
	Genomics into comprehensive cancer plan	$r(13) = .55$	0.034
	Genomics into other cancer policies	$r(13) = .56$	0.030
Primary care providers	LS	$r(12) = .73$	0.003
	FH	$r(12) = .59$	0.026
	ID of FH individuals and family members	$r(11) = .57$	0.044
Genetic counselors	HBOC	$r(14) = .54$	0.32
Other clinicians	LS	$r(12) = .73$	0.003
	FH	$r(12) = .58$	0.029
	Using cancer registry data	$r(11) = .57$	0.042
Advocacy groups	LS	$r(11) = .69$	0.010
Hospitals and healthcare systems	LS	$r(13) = .69$	0.021
	Using cancer registry data	$r(12) = .56$	0.038
Third party payers	LS	$r(12) = .58$	0.031
Local and county health departments	Interested in promoting or enhancing genomic awareness among medical providers	$r(12) = .59$	0.027

Note. Only responses of collaborations within the last quarter and last year were included.

Summary

Analysis of data for Research Question 1, which looked at the possible association of current state genomic funding and the level of knowledge and interest in genomics by CDDs showed only one association; CDDs agreement with the statement "Citizens in our state understand how family history or genetics influences risk of chronic disease." This single association demonstrates that current state genomic funding has very little impact on the level of knowledge and interest in genomics by CDDs.

Analysis of associations for Research Question 2 however, presence of state genomics activities and level of knowledge and interests in genomics by CDDs, showed many significant associations. Existence of cancer control action plans, breast, ovarian, and colorectal education, and regulations pertaining to non-discrimination and privacy were significantly associated with CDDs knowledge of genomic referrals for patients or consultation for themselves. These associations were intact whether "Don't know" responses were included or not.

ANOVA and independent *t* test results found that breast, ovarian, and colorectal education is associated with CDD's knowledge of LS, FH, interest in incorporating identification of individuals/cascade screening for FH in the state Cardiac Disease Prevention Program, and agreement that legal protections against genetic discrimination are adequate in their states. Questions related to the presence of genomic topics on the BRFSS were associated with CDDs agreement that genetics is an important component of public health initiatives and that legal protections against genetic discrimination are adequate in their states. Having a genetic needs assessment ($N = 1$) was associated with

interest in recommending the addition of genomics questions to the BRFSS cancer modules, incorporating genomics into other cancer polices and initiatives, and promoting or enhancing genomics awareness among the general public. Having a state action plan in cardiovascular health was associated with knowledge of LS and FH and agreement that legal protections against genetic discrimination are adequate in their states. Finally, current state regulations for genetic privacy, providing genetic services to uninsured or low income residents, and funding a state genetics coordinator position were associated with CDD's agreement that legal protections against genetic discrimination are adequate in their states, and interest in incorporating identification of individuals/cascade screening for FH in the state Cardiac Disease Prevention Program, promoting or enhancing genomics awareness among medical providers, as well as funding for a state genetics coordinator position.

Pearson's correlation discovered that CDD knowledge of Tier 1 conditions was associated with more frequent collaborations with academic institutions (HBOC, LS), other clinicians (LS, FH), genetic counselors (HBOC), and primary care providers, advocacy groups, hospitals and healthcare systems, and third party payers (LS). CDDs were more interested in incorporating genomics into the comprehensive cancer action plan and other cancer policies and initiatives when they collaborated frequently with academic institutions and would be more likely to want to incorporate the use of the state cancer registry to identify high risk patients if they collaborated with other clinicians or hospitals and healthcare systems. Lastly, CDDs would be interested in incorporating identification of individuals/cascade screening for FH into their state Cardiac Disease

Prevention Program if they collaborated with primary care providers and were interested in promoting or enhancing genomic awareness among medical providers if they connected with local and county health departments.

In this results chapter, I provided specifics about the data collection used in this study, results of the statistical analysis, and a summary of the findings of the survey of state and territorial CDDs. These findings showed a number of significant associations with specific state genomic activies and the level of knowledge and interest in genomics by state and territorial CDDs. In the final chapter of this dissertation, I will provide an interpretation of the findings, limitations of the study, recommendations for further research, and implications for social change.

Chapter 5: Discussion, Conclusions and Recommendations

Introduction

Given the potential that genomic technologies have for identifying individuals and their families at risk of heritable chronic disease and targeting public health prevention efforts, I have been puzzled at the slow uptake of these interventions. As state public health organizations are essential in the national effort to promote the use of genomic information to reduce morbidity and mortality and save lives (Green et al., 2015), examining what might be impacting this slow integration could shed light on future steps. In order to do this, I performed a secondary analysis of a survey of state and territorial CDDs about current state genomic activities and their level of knowledge and interest in genomic topics believing CDD's familiarity with genomics could impact their leadership in this area. To date, only a small number of states have received funding to incorporate genomics into their chronic disease programming, so one purpose of this study was to see if this funding was associated with an increased level of knowledge and interest in genomics by CDDs. The second purpose of this study was to examine if any current state genomic activities were linked to the level of knowledge and interest in genomics by these CDDs.

Based on the results of this survey, it appears that state chronic disease genomics funding has almost no impact on the level of knowledge and interest in genomics by the CDDs. There were, however, many significant associations with respect to specific state genomic activities and CDDs' knowledge and interest. State activities that were associated with a higher level of knowledge and interest by CDDs include a state

genomics needs assessment (n = 1); genetics inclusion in cancer control and cardiovascular health action plans; breast, ovarian, and colorectal cancer genetics education; and state laws or regulations pertaining to genetic nondiscrimination, privacy, providing genetic services to uninsured or low-income residents, and funding a state genetics coordinator position. Inclusion of genomics on BRFSS topics related to breast/cervical cancer screening, cardiovascular health, colorectal cancer, genetic discrimination, and privacy were also associated with a higher level of knowledge and interest in genomics by the CDDs. Finally, frequent collaborations (in the past quarter/year) with outside entities, mainly academic institutions, primary care providers, and other clinicians were associated with greater levels of knowledge and interest, especially for knowledge of Tier 1 conditions and particularly LS.

Interpretation of Findings

The framework from the literature on Rogers' diffusion of innovations theory indicated that disseminating, implementing, and sustaining new innovations depends largely on sufficient knowledge of the innovation to progress through the phases of adoption (Rogers, 2003). According to this theory, individuals' knowledge can impact their attitudes which, in turn, influence their decision to adopt and implement an innovation. Rogers also explained that those who adopt later in the process will require a longer innovation-decision period. This is particularly important as more states adopt chronic disease genomics programming. Therefore, pinpointing associations between specific activities and a greater knowledge or interest in genomics by CDDs could help

identify actions that might be associated with this desired outcome, chronic disease genomics implementation.

However, initially in this study I examined the potential association of state genomic funding and the level of knowledge and interest in genomics by the CDDs. An individual would imagine that states with chronic disease genomics funding would have more state activities and CDDs with a greater interest in genomic integration. Six (38%) of the 16 surveys received were from states identified as funded for chronic disease genomic activities; however, statistical analysis did not show a significant association with funding and the level of knowledge and interest in genomics by the CDDs of those states. As I was looking at an association and could not show causality in this study, one conclusion that can be made is that the greater knowledge and interest in genomics was driven by the CDD(s) themselves and was not related in any way to the funding provided to individual states. There were, in fact, some states that did not receive funding and had little or no state genomic activities, yet the CDDs were more knowledgeable and interested than others.

This also leads into the prior described theory that the genomics champion in the funded states is influential in integrating genomics into state chronic disease programs (Schon, 1963; Taylor et al., 2011). Rogers (2003) is clear that this champion is instrumental to the success of an innovation and has a positive impact on the rate of adoption. As these funded states often have at least one person, the champion, seeking this funding, driving the work, and producing evaluative data, this model seems like it would be conducive to increased knowledge and interest in genomics by the CDDs.

Because the statistical analysis showed no association, a possible conclusion is that those CDDs with increased knowledge and interest in genomics could, themselves, become that genomics champion that is integral to adoption of this innovation in their states.

In this study, I identified that there are, in fact, specific state genomic activities that are associated with greater knowledge and interest in genomics by state and territorial CDDs. Educational programs related to the Tier 1 conditions were associated with greater knowledge and interests by the CDDs; however, this was only for LS and FH. Interestingly, HBOC was not associated with increased knowledge and interest in HBOC. This mismatch is likely due to the small sample size; however, using educational endeavors to increase knowledge in genomics is not a new concept (Khoury, Gwinn, Dotson, & Schully, 2012; Talwar, Tseng, Foster, Xu, & Chen, 2016).

The BRFSS is used to survey U.S. residents concerning health-related risk behaviors, chronic diseases, and prevention measure utilization (CDC, 2017). Responses to questions on the BRFSS are used by CDDs and others as one of the indicators of state and selected metropolitan-level chronic diseases and risk factors that impact public health (Holt et al., 2015). This system is not only used for surveillance but for prioritizing and evaluating public health interventions. The fact that BRFSS genomics-related questions were found to be significantly associated with greater knowledge and interest in genomics by the CDDs is, therefore, potentially meaningful if CDDs translate that interest into leadership to prioritizing genomics into chronic disease programming.

One of the six components of successful public health program implementation has been found to be collaborations and partnerships with public and private entities

(Frieden, 2014). In this study, I found that CDDs who collaborated with academic institutions and medical professionals significantly increased their knowledge and interest in genomics. Academic institutions are often where genetics professionals (geneticists, genetic counselors) and researchers are based and these individuals can provide a wealth of knowledge in this area. The fact that collaborations with genetic counselors was not determined to be a significant factor in increased knowledge and interest could be because of the limited number of these professional nationwide (Wicklund & Trepanier, 2014).

Using Rogers' Theory to Diffuse Chronic Disease Genomics in More States

Although prevention is usually cheaper than treatments, new prevention efforts in public health frequently diffuse slowly due to the delay in observance of clear health outcomes and perception of the relative advantage by public health leadership (Rogers 2002). One factor impacting the rate of adoption is the complexity, or perceived difficulty of and innovation among members of a social system. Increasing the knowledge and interest in chronic disease genomics by CDDs will impact their perception of the complexity of these applications and help them gain a greater understanding of the potential health impact to their populations. In this study, I have identified specific genomics activities currently being performed by states that are having an impact on this knowledge and interest by CDDs. When trying to identify ways to diffuse chronic disease genomics in more states, these activities would be the ones to start with.

Rogers (2002) also mentions the importance of the early adopters to the diffusion process and their role as opinion leaders of the social system. The next segment in the

diffusion of chronic disease genomics, the early majority, will be looking towards these states for modelling, information, and advice. Based on the current landscape, I had previously identified seven early adopter states; however, results of this survey show that there are other states that are already doing work in this area and could also be instrumental opinion leaders and models on how to do this work without funding specific to chronic disease genomics.

Limitations of the Study

As I previously described in the Introduction, one of the limitations of this study was the small sample size of the study group and subsequent limited number of CDDs (N = 16) who responded to the survey. The total response correlates to published survey response rates; however, had the APHA GFPC subcommittee decided to extend the survey timeframe to longer than 6 weeks or sought alternative follow-up contact (such as phone calls or mailings), this may or may not have had an impact on the number of final responses received. I cannot know whether a delivery of this survey at an alternate time or under different circumstances would impact the survey response rate. It is known that limited datasets can impact study power and external validity (generalizability; Field, 2013); nevertheless, my analysis was able to find significant associations that can be informative to the study topic.

Another threat to external validity was not knowing who actually filled out the survey. Although this was a confidential survey, there was no way to prove that the CDD was the one who answered all or some of the questions. Moreover, there may be external conditions such as personal stress, other work deadlines, topic knowledge, no

compensation, or the short survey time frame that might have impacted their desire to participate.

According to Shadish, Cook, and Campbell (2002), one threat to internal validity is ambiguous temporal precedence. What this means is that, based on the study design, causality cannot be determined and that a researcher cannot conclude with certainty which variable causes another; the researcher is only showing that the variables have an association to one another. This often occurs in situations involving ongoing processes that interact with one another and, in turn, be affected by one another (Trochim, 2006). This phenomenon was evident in the discussion about state genomic funding and the level of knowledge and interests in genomics by the CDDs in this study. The fact that genomics funding was not significantly associated with CDD knowledge and interest means that the higher knowledge and interest is occurring regardless and, at this point in time, is not causing an increase in funding for genomics implementation. Finally, there is always the possibility that a third variable that was not explored in this study could be the causative agent for the results seen (Trochim, 2006).

Recommendations

Because state and territorial CDDs will likely be instrumental leaders in the success of chronic disease genomics implementation, further research of this group is warranted. Although the results of this study showed some significant associations, had there been a larger sample size, findings that were not significant at the .05 level could have been statistically significant (Rudestam & Newton, 2015). It would be prudent to perform both further quantitative as well as qualitative studies of this group to gather a

more detailed profile of what they know and what they feel they need to know in order to be familiar enough with public health genomics; chronic disease genomics programming; identification and contact with collaborators, partners, and stakeholders; and possible implications to lead these efforts. Gaining an understanding of what CDDs see as potential barriers to implementation or why some CDDs have greater knowledge without funding or activities would also be important.

Another area to examine would be to study current state genomic activities in depth to find out exactly what states are doing what, who the champions are, and what, if any, partnerships are being formed within the states or among the states. This could be instrumental in determining who the next 19–20 states are that might be more successful in implementing chronic disease genomics programming based on Rogers' theory (early majority). The states that are ready to move forward could be the focus of educational interventions, financial support, and collaborative efforts. This investigation could be performed with a more extensive state activity survey or possible phone interview.

Assessing organizational readiness for chronic disease genomics implementation by state health departments could identify other areas that may hinder genomics implementation (Stamatakis et al., 2012). State health departments will have a greater impact on population health if they are effectively run, have adequate resources, competent staff, and utilize evidence-based decision making (Alongi, 2015; Maylahn et al., 2013). Innovation adoption doesn't occur, however, without individual and organizational changes based on clear effectiveness and cost effectiveness of the innovation (Greenhalgh et al., 2004), so continued research and dissemination of the

benefits of Tier 1 genomic applications will be important. Finally, investigating organizational innovativeness and support structure for genomics implementation (Oishi et al., 2015) could help identify avenues for adoption in more states.

St Pierre et al. (2014) explained that chronic disease genomics implementation will have a greater chance of success if leadership capacity is developed, incorporated into population-based assessment, surveillance, and disease prevention programming, and genomic education is provided to public health and healthcare practitioners, policy makers, and the public. Of course, all of these endeavors require funding. One of the problems in the current climate is that most funding for genomic research is in the discovery phase (T1) and very little is provided towards implementation (T4;Glasgow et al., 2012; Schully, Benedicto, Gillanders, Wang, & Khoury, 2011). More research must be done to show the benefits of chronic disease genomics programming for state and territorial populations to encourage funding to that end.

Implications for Social Change

It could be argued that inheritance of genetic mutations that predispose an individual to an increased susceptibility of certain chronic diseases would eventually impact their feelings of health and well-being. The manifestation of these conditions, of course, is not only due to these genetic mutations, but also impacted by many other social, environmental, cultural, economic, and political circumstances. Public health professionals have a responsibility to safeguard the health of people and the communities in which they live by working to "assure the conditions in which people can be healthy" (APHA, 2017, para. 3). Social change can, and will, happen when public health

professionals improve population health through program and policy implementation that targets vulnerable communities and other stakeholders while also developing and delivering education to assure understanding of potential effects (Godwin & Heymann, 2015).

The overarching goal of my research in this study and beyond is to find opportunities and barriers to genomic technology implementation in healthcare and public health. The results of this study showed some opportunities to increased knowledge and interest in genomics by CDDs and putting those in place in more states could lead to an increase in chronic disease genomics programming. That would lead to greater access to identification of individuals at risk of these conditions and possible prevention or reduction of manifestations. Once an individual has been documented, this could then have a ripple effect of identification for their family members, communities as well as whole populations and have a larger impact on morbidity and mortality from these hereditary conditions.

Conclusion

The completion of the Human Genome Project in 2003 has allowed public health to practice in a more personalized and precise manner and has also been shown to improve health outcomes (Auffray et al., 2016). Evidence exists for population screening of affected individuals and their family members for three common chronic diseases with a known hereditary component, yet only a small number of states are currently doing any work in this area (St Pierre et al., 2014). State and territorial CDDs are in a very

influential position to lead chronic disease genomic programming in their states and it is important to identify ways to help them reach that goal.

Rogers' diffusion of innovations theory demonstrates that adoption of chronic disease genomics programming is ready to move into more states. Rogers' (2003) theory also explains that this next group (early majority) is an important link in the diffusion process, between the risk-takers and the skeptics, but may take somewhat longer to adopt these processes. Determination of what states will be involved in this next phase will be critical to overall chronic disease genomic adoption success. The results of this study could identify some of those next 19–20 states, particularly those who already have CDDs with a greater knowledge and interest in genomics, to address integration of particular activities (as I found in this study) or focus funding.

In order to fulfill the promise of precision medicine through genomics, more research needs to be done to understand what is hindering the translation of this promise into reality. Unfortunately, existing knowledge is limited and implementation continues to be slow (Manolio et al., 2013; Roberts, Kennedy, Chambers, & Khoury, 2017). Increased integration of evidence-based genomic applications, such as Tier 1 chronic disease conditions, to diverse populations will increase the empirical evidence needed to show the impact this technology can have on population health. State and territorial CDDs can and should be the leaders in that endeavor.

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Appendix: The Chronic Disease Director's Survey with Coding

Chronic Disease Director Survey

Q11 Has your state/territory conducted a genetics needs assessment?

- ❍ Yes (1)
- ❍ No (2)
- ❍ I Don't Know (3)

Q29 Did the genetics needs assessment include any action around genomics in chronic disease?

- ❍ Yes (1)
- ❍ No (2)
- ❍ I don't know (3)

Q13 Is genetics included in your state action plans for:

	Yes (1)	No (2)	Don't Know (3)
Chronic Disease (1)	❍	❍	❍
Cancer Control (2)	❍	❍	❍
Cardiovascular Health (3)	❍	❍	❍
Other (e.g. asthma, arthritis, Alzheimer's); please specify (4)	❍	❍	❍

Q8 Has your state integrated genetics education into programming for any of the following conditions:

	Yes (1)	No (2)	Don't Know (3)
Breast Cancer (1)	❍	❍	❍
Colorectal Cancer (2)	❍	❍	❍
Ovarian Cancer (3)	❍	❍	❍
Cardiovascular Disease (4)	❍	❍	❍
Other (please specify) (5)	❍	❍	❍

Q9 Do you currently include genomics-related questions on the following topics in the BRFSS?

	Yes (1)	No (2)	Don't Know (3)
Breast and Cervical Cancer Screening (1)	❍	❍	❍
Cardiovascular Health (2)	❍	❍	❍
Colorectal Cancer Screening (3)	❍	❍	❍
Health Care Access (4)	❍	❍	❍
Genetic discrimination (5)	❍	❍	❍
Privacy (6)	❍	❍	❍
Direct-to-Consumer Advertising (7)	❍	❍	❍

Q10 Has your state/territory analyzed state cancer registry or other vital records data to determine the number of citizens who might be affected by hereditary cancer syndromes?

- ❍ Yes (1)
- ❍ No (2)
- ❍ Don't Know (3)

Q26 If yes, when was the most recent year you analyzed these records and for what diseases/conditions?

Q25 How often do you engage in collaboration or partnership related to genomics with the following groups?

	In the past quarter (1)	In the past year (2)	Rarely (3)	Never (4)	Never but Potentially in the Future (5)
Academic Institutions (1)	❍	❍	❍	❍	❍
Primary care providers (2)	❍	❍	❍	❍	❍
Genetic Counselors (3)	❍	❍	❍	❍	❍
Other clinicians (4)	❍	❍	❍	❍	❍
Advocacy Groups (5)	❍	❍	❍	❍	❍
Hospitals and healthcare systems (6)	❍	❍	❍	❍	❍
Third party payers (7)	❍	❍	❍	❍	❍
Local and county health departments (8)	❍	❍	❍	❍	❍
Other groups (please indicate) (9)	❍	❍	❍	❍	❍

Q15 Does your state/territory have legislation and/or regulations specifically related to genetics, such as:

	Yes (1)	No (2)	Don't Know (3)
Non-discrimination laws (1)	❍	❍	❍
Privacy rules (2)	❍	❍	❍
Informed consent (3)	❍	❍	❍
Funding a State Genetics Coordinator position (4)	❍	❍	❍
Providing genetic services to uninsured or low-income residents (5)	❍	❍	❍

Q16 If you needed to refer patients to clinicians for genetic services, or if you wanted to consult someone with expertise in genetics, would you know how to contact or locate genetics professionals in your state/territory?

- ❍ Yes (1)
- ❍ No (2)

Q22 On a scale of 1-5 (with 1 very poor and 5 very good, how would you rate your knowledge of:

	1 (1)	2 (2)	3 (3)	4 (4)	5 (5)
Hereditary Breast/Ovarian Cancer Syndrome (1)	❍	❍	❍	❍	❍
Lynch Syndrome (2)	❍	❍	❍	❍	❍
Familial Hypercholesterolemia (3)	❍	❍	❍	❍	❍

Q23 To what extent do you agree or disagree with the following statements?

	Strongly Disagree (1)	Disagree (2)	Neither Agree nor Disagree (3)	Agree (4)	Strongly Agree (5)	Don't Know (6)
Citizens in our state understand how family history or genetics influences risk of chronic disease. (1)	❍	❍	❍	❍	❍	❍
As a whole, staff in the Chronic Disease Program understands how genetics relates to chronic disease. (2)	❍	❍	❍	❍	❍	❍
Genetic counseling and testing for hereditary cancer conditions can improve a patient's health outcomes. (3)	❍	❍	❍	❍	❍	❍
Integrating genetics into public health planning for chronic disease programming would benefit residents of our state. (4)	❍	❍	❍	❍	❍	❍
Legal protections against genetic discrimination are adequate in our state. (5)	❍	❍	❍	❍	❍	❍
Genetics is an important component of public health initiatives. (6)	❍	❍	❍	❍	❍	❍

Q24 In my role as a chronic disease director (CDD), I would be interested in:

	Strongly Disagree (1)	Disagree (2)	Neither Agree nor Disagree (3)	Agree (4)	Strongly Agree (5)	We already do this (6)
Recommending the addition of genomics questions to the BRFSS cancer modules (1)	❍	❍	❍	❍	❍	❍
Utilizing Cancer Registry data to identify high risk patients with the goal of reducing morbidity and mortality (2)	❍	❍	❍	❍	❍	❍
Incorporating genomics into our comprehensive cancer action plan. (3)	❍	❍	❍	❍	❍	❍
Incorporating genomics into other cancer policies and initiatives. (4)	❍	❍	❍	❍	❍	❍
Incorporating cancer genomics into our state's Breast and Cervical Cancer Early Detection Program. (5)	❍	❍	❍	❍	❍	❍
Incorporating identification of individuals/cascade screening for Familial Hypercholesterolemia (FH) into our state's Cardiac Disease Prevention Program. (6)	❍	❍	❍	❍	❍	❍
Promoting or enhancing genomics awareness among the general public. (7)	❍	❍	❍	❍	❍	❍
Promoting or enhancing genomics awareness among medical providers (8)	❍	❍	❍	❍	❍	❍
Finding funding to hire an individual to focus on genomics and chronic disease programming (9)	❍	❍	❍	❍	❍	❍

What state or territory do you work with?

- Alabama (1)
- Alaska (2)
- Arizona (3)
- Arkansas (4)
- California (5)
- Colorado (6)
- Connecticut (7)
- Delaware (8)
- Florida (9)
- Georgia (10)
- Hawaii (11)
- Idaho (12)
- Illinois (13)
- Indiana (14)
- Iowa (15)
- Kansas (16)
- Kentucky (17)
- Louisiana (18)
- Maine (19)
- Maryland (20)
- Massachusetts (21)
- Michigan (22)
- Minnesota (23)
- Mississippi (24)
- Missouri (25)
- Montana (26)
- Nebraska (27)
- Nevada (28)
- New Hampshire (29)
- New Jersey (30)
- New Mexico (31)
- New York (32)
- North Carolina (33)
- North Dakota (34)
- Ohio (35)
- Oklahoma (36)
- Oregon (37)
- Pennsylvania (38)
- Rhode Island (39)
- South Carolina (40)
- South Dakota (41)
- Tennessee (42)
- Texas (43)
- Utah (44)
- Vermont (45)
- Virginia (46)
- Washington (47)

- West Virginia (48)
- Wisconsin (49)
- Wyoming (50)
- Puerto Rico (51)
- Guam (52)
- Northern Marianas (53)
- United States Virgin Islands (54)
- American Samoa (55)

Q18 Please indicate your age

- 21-30 (1)
- 31-40 (2)
- 41-50 (3)
- 51-60 (4)
- 60+ (5)

Q20 Please indicate any degrees or board certifications you hold (i.e. MD, MPH, MBA, PhD, BA, etc.)