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# Utilization Of Genetic Screening Practices By Primary Care Providers For Individuals With Increased Risks For Alzheimer'S Disease

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Graduate Committee Approval

The Graduate Committee of Cindy Renee' Quinn, Mallory Stockstill, Lucy Barnes, Jalisha Brown, and Angela Reeves
hereby approves this research project as meeting partial fulfillment of the requirements for the Degree of Master of Science in Nursing.

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**Dedication**

This research is dedicated to GOD, my loving husband Antione,my beautiful daughter Bayleigh and my family. Without you'll, there would be no me. God, you have heard my cries and prayers, although a fall short of your glory each and everyday, you continue to pick up and put me back on the right path. To my husband Antione, thank you for your unconditional love, sacrifices, and words of encouragement. To Bayleigh, thank you for being the light on my darkest days as well giving me the motivation to strive for greater. I hope that someday you will follow in my footsteps. Family, thank you for your endless support throughout this past year. This has been one of the most challenging, yet rewarding years of my life. We did it!! With Love, Jalisha Brown

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Thank you,
Mallory Stockstill
Angela Reeves
Cindy Renee Quinn
Jalisha Brown
Lucy Barnes

**Utilization of Genetic Screening by Primary Care Providers for Individuals with Increased Risks for Alzheimer's Disease**

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***Abstract***

The purpose of this study was to determine if health care providers in the primary care setting were identifying individuals with increased risks for Alzheimer's disease and if the utilization of genetic screening related to the disease was being offered to those individuals. Extensive research into the genetic etiology of Alzheimer's disease has proven that some genetic factors are causative and increase a person's risk of developing the disease. The need for further comprehensive assessments for those with increased risk of developing Alzheimer's disease, such as genetic testing, is imperative in identifying the disease-causing gene mutations associated with the disease. These risk factors include increased age, family history, diabetes, hypertension, obesity, smoking, depression, cognitive inactivity, physical inactivity, low education, and specific genetic markers (APOE-e4). Focusing efforts in the primary care setting on identifying patients who are at increased risk of developing the disease prior to the clinical onset allows for the utilization of genetic screening. Identifying cognitively healthy individuals ages 50-75, who are at increased risk of developing Alzheimer's disease-related to their age or genetic variations in genes, will aid providers with diagnosis and clarification of risk for those individuals and their families. The current study addressed these issues with an emphasis on health promotion which has the potential of long term benefits of extending

longevity, enhancing the quality of life, and reducing health care cost.

Research was conducted to determine if genetic screening was being utilized in the primary care setting for individuals who are at increased risk for developing Alzheimer's disease. This was done by utilization of a quantitative survey. Strict caution was taken once approval was granted by the Institutional Review Board at Mississippi University for Women, this was to ensure participant privacy and anonymity as data was collected. The researchers provided questions in the form of a survey utilizing SurveyMonkey, Inc. which included demographic data, the determination of provider assessments, the evidence of genetic screening being offered, the cognitive assessments used, the reason genetic screening is not utilized, and the follow-up care provided for those who choose genetic screening. The study had 20 respondents in this sample, which was also considered a limitation in the research. Based on this sample of respondents and upon completion of the data, the study determined that genetic testing was not being utilized by primary care providers in the state of Mississippi for individuals with increased risk for developing Alzheimer's Disease.

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# CHAPTER 1

## Introduction

The purpose of this study was to determine if health care providers in the primary care setting were identifying individuals with increased risks for the disease, and to determine if the utilization of genetic screening related to Alzheimer's disease was being offered to those individuals. Extensive research into the genetic etiology of Alzheimer's disease has proven that some genetic factors are causative and increase a person's risk of developing the disease. The need for further comprehensive assessments for individuals with increased risks of developing Alzheimer's disease, such as genetic testing, is imperative in identifying the disease-causing gene mutations associated with the disease. These risk factors include increased age, family history, diabetes, hypertension, obesity, smoking, depression, cognitive inactivity, physical inactivity, low education, and specific genetic markers (APOE-e4). Focusing efforts in the primary care setting on identifying patients who are at increased risk of developing the disease prior to the clinical onset allows for the utilization of genetic screening. Identifying cognitively healthy individuals ages 50-75, who are at increased risk of developing Alzheimer's disease related to their age or genetic variations in genes, aids providers in diagnosis and clarification of risk for those individuals and their families. The current study addressed these issues with an emphasis on health promotion that has the potential of long term benefits of extending longevity, enhancing the quality of life, and reducing health care cost.

Alzheimer’s disease is a significant health problem across the United States. There is no standardization in the screening protocol for dementia and its progression. Alzheimer’s disease and other types of dementia do not limit themselves to one particular





population or socio-economic group. The disease does not discriminate among culture or ethnicity. Alzheimer’s disease is an irreversible, progressive brain disorder that slowly destroys memory and thinking skills and, eventually, the ability to carry out the simplest tasks (NIA, 2017). Alzheimer’s is ultimately ta fatal form of dementia. It is the sixth leading cause of death in the United States, accounting for almost 4% of all deaths in 2014 (CDC, 2017). The number of Alzheimer's deaths has increased because of the growing population of older adults. The death rate for Alzheimer’s disease increased 55% from 1999 to 2014. In 2014, over 93,500 deaths across all 50 states and the District of Columbia occurred due to Alzheimer's disease. Deaths attributed to Alzheimer's disease increased among adults 75 years or older. It is estimated that by 2050, 16 million people will be diagnosed with Alzheimer's Disease (CDC, 2017).

In 1906, Dr. Alois Alzheimer noticed changes in the brain tissue of a woman who has died from an unusual mental illness. Her symptoms included memory loss, language problems, and unpredictable behavior. After the patient’s death Dr. Alzheimer examined her brain and found a multitude of abnormal clumps (now referred to as amyloid plaques)and tangled bundles of fibers (now known as neurofibrillary, or tau tangles). The complex disease he discovered became known as Alzheimer’s disease. Dr. Alzheimer concluded that given the complexity of the disease, it was unlikely that one drug or single

specific intervention would successfully treat the problem. Current treatments focus on the management of disease progression and helping people maintain mental function, manage behavioral symptoms, and slowing the progression of specific problems, such as memory loss. Researchers hope to develop therapies targeting specific genetic,



molecular, and cellular mechanisms so that the actual underlying cause of the disease can be stopped or prevented (NIA, 2017). Research has proven that changes in the brain related to the disease process can be present years before symptoms appear. Evidence targets other disease processes that may contribute to Alzheimer's disease, such as heart disease, high blood pressure, and high cholesterol. Early detection of genetic markers that are known to be present in Alzheimer's disease could provide the opportunity to make changes that could delay or stop the disease progression. Modifiable risk factors such as exercise, diet, and cognitive challenges have been shown to slow or even possibly arrest the disease process (CDC, 2017).

**Statement of the Problem**

Alzheimer's disease is a progressive neurological illness that continues to affect thousands of individuals annually. Scientists have identified three genetic markers with rare variations related to Alzheimer's disease, along with other genetic markers that are known to suggest increased risk; however, these do not guarantee that a person will develop the disease. Investigators worldwide are working to identify additional genes responsible for causing Alzheimer’s disease. As more effective treatment becomes available, genetic profiling may become a valuable risk assessment tool for broader use.

Genetic screening for APOE-e4, the most influential risk gene, is included in some clinical trials to identify participants at high risk for the disease (Alzheimer's Association, 2019). Risk factors for the disease include not only age and family history, but also diabetes, hypertension, obesity, smoking, depression, cognitive inactivity, physical



inactivity, and low education. These factors are critical components in identifying the disease-causing gene mutations associated with Alzheimer's disease. The problem of interest in this study was based on whether or not genetic screening was utilized in the primary care setting for patients with an increased risk of Alzheimer's disease. As the population ages, the prevalence of the disease will continue to rise. The cost of treatment, long-term care, and caregiver's health maintenance will also continue to rise. This cost could potentially put additional strain on the health care system.

It was predicted that providers in the primary care setting would be confronted with barriers to genetic screening. The barriers included: cost to the patient; insurance coverage; education of the population regarding the effects of the disease; lack of time to educate the patient in the clinical setting; and lack of staff education and training related to the disease. Review of literature revealed numerous studies that demonstrated the early screening of patients with a family history, along with other risk factors such as diabetes, hypertension, obesity, smoking, depression, cognitive inactivity, physical inactivity, low education, and specific genetic markers (APOE-e4) could promote

lifestyle changes that could alter the progression of the disease. Early intervention could be the key to controlling this disease and future research.

**Statement of the Purpose**

The purpose of this study was to determine if genetic screening was being utilized in the primary care setting for individuals who were at increased risk for developing Alzheimer's disease. The risk factors which include increased age, family history,



diabetes, hypertension, obesity, smoking, depression, cognitive inactivity, physical inactivity, low education, specific genetic markers (APOE-e4) that predispose individuals to Alzheimer's disease. These risk factors can appropriately be identified in the primary care setting. The present study allowed insight into the utilization of genetic screening to identify at-risk individuals, and if genetic screening concerning Alzheimer's disease was provided in the primary care setting. Early detection of risk factors related to Alzheimer's disease would provide the opportunity for patients and providers to reduce the prevalence and complications of this disease. Early identification of individuals at risk for Alzheimer's disease, coupled with genetic screening, would decrease the burden of effects on concerned individuals and their family members. Continued education related to genetic screening and Alzheimer's disease is needed for both health care providers and individuals with increased risks of the illness like the following: age, family history, diabetes, hypertension, obesity, smoking, depression, cognitive inactivity, low education, and specific genetic markers (APOE-e4) to stay aware of current treatments and resources available. Research related to genetic testing and Alzheimer's

disease will help to develop an open dialogue among providers and increase awareness of the disease and the availability of genetic screening.

**Significance of the Study**

The significance of this study has proved to be useful to primary care providers, as well as neurologists and psychiatric-mental health professionals who diagnose, treat, and manage patients with Alzheimer's disease. While there are no preventive strategies or cures for Alzheimer's disease currently, timely diagnosis is essential for effective



management. People are living longer, but the quality of life has diminished significantly in the 21st century. Early detection allows patients and their families the option to modify risk factors related to diabetes, hypertension, obesity, smoking, depression, cognitive inactivity, physical inactivity, low education, and specific genetic markers (APOE-e4). This also allows the patient to receive symptomatic treatment to support lifestyle modifications, such as exercise, diet, and cognitive exercises. Early detection also provides the patient the opportunity to make plans for the future. Extensive research into the genetic etiology of Alzheimer's disease has proven that some genetic factors are causative and increase a person's risk for the disease. The need for further comprehensive geriatric assessments, such as genetic testing, is a critical component in identifying the disease-causing gene mutation associated with Alzheimer's disease. Focusing efforts in the primary care setting on identifying patients who are at increased risk of developing the disease prior to the clinical onset allows for the utilization of genetic screening. Identifying cognitively healthy individuals ages 50-75, who are at high risk for developing Alzheimer's disease related to their age or genetic variations in

genes, will aid providers with diagnosis and clarification of risk for those individuals and their families.

**Conceptual Framework**

Nola Pender's Health Promotion Model was chosen as the theoretical model for the current research project because Nola Pender's model promotes prevention as a means of total health and wellness. She developed her Health Promotion Model, often abbreviated HPM, after seeing professionals intervene once a patient developed an acute



or chronic illness. She became convinced that the prevention of problems could improve a patient's quality of life and the promotion of healthy lifestyles could save health care dollars. Nola Pender's framework focuses on the positive aspects of health. The initial version of the model appeared in 1982, and a revision of the model in 1996 was based on changing theoretical perspectives and empirical findings (Petiprin, 2016). Nola Pender incorporated psychological educational nursing concepts, and theories to formulate the Health Promotion Model. Pender's model stated that people have the capacity for reflective self-awareness, including assessment of their competencies. Self-awareness would motivate a positive change and create a desire for the individual to change his habits once he is aware of how negative behavior would impact his health. Pender's model emphasizes the conceptual propositions that barriers place constraints on the commitment to action. These barriers would include the cost of healthier food choices, association with individuals that use tobacco products and exposure to second-hand smoke, or the lack of wellness visits at the appropriate intervals. For example, an individual who is a smoker and obese is educated on risk factors for Alzheimer's disease.

With the knowledge provided that smoking and poor food choices increase the individual's risk in the development and progression of the disease, it could be inferred that the individual would elect to discontinue these activities as an effort to promote his health. Pender's model is based on the concept that education will increase a patient's desire to make healthier lifestyle choices and become healthier in body and mind. Once the benefits of making lifestyle changes are perceived and barriers removed, the individual would then commit to a plan of action to achieve a health-promoting lifestyle.



Pender's model concerning Alzheimer's testing in the primary care setting, emphasizes that health promotion would allow the choice of wellness rather than aggressive disease. Every patient with increased risk factors, should have the opportunity to choose genetic testing. Genetic testing would enable each patient to have further education on the disease. Genetic counseling, support groups, and online training would be considered part of genetic testing according to Pender's concept.

Pender's Model would allow everyone to individually determine if socio-cultural, economic, or political conditions could affect their health status. Health care providers must be educated in the benefits of early detection so that the general public can be knowledgeably informed. Primary care clinics could also raise awareness about healthy behaviors for the general public. Raising awareness would include health fairs, newsletters, and mass media communications to promote community involvement and knowledge of the benefits of genetic testing and Alzheimer's prevention. The current study utilized a survey that was sent via email and social media sites that include primary care providers. The survey was to determine if primary care providers were offering the

option of genetic screening to patients with an increased risk of Alzheimer's disease. The Health Promotion Model was selected to guide this research because of the model's focus on wellness-related health promotion.

**Research Questions**

Two research questions were developed to guide data collection regarding the utilization of genetic screening by primary care providers for individuals with increased risks for Alzheimer's disease.



1. Are patients who are at increased risk of developing Alzheimer's disease being identified by primary care providers in the primary care setting?
2. Are primary care providers utilizing genetic screening for patients who are at increased risk of developing Alzheimer's disease?

**Definition of Terms**

For this study, several terms were defined as they apply to this study. The theoretical and operational definitions follow, respectively.

**Patient**

*Theoretical.* Tabor's defines a patient as one who receives medical care

*Operational.* This study defines a patient as a person that is at an increased risk of developing Alzheimer's disease.

**Increased Risk Factors**

*Theoretical.* Tabors define risk factors as an environmental, chemical, psychological, physiological, or genetic element that predisposes someone to the development of the disease.

*Operational.* This study defines increased risk factors of Alzheimer's disease based on guidelines determined by the Alzheimer's Association. These increased risk factors include not only age and family history, but also diabetes, hypertension, obesity, smoking, depression, cognitive inactivity, physical inactivity, low education, and specific genetic markers (APOE-e4). The study



identifies these factors as placing patients at increased risk of developing Alzheimer's disease.

**Alzheimer's Disease**

*Theoretical.* Tabor's defines Alzheimer's disease as a chronic, progressive, degenerative cognitive disorder that causes deterioration in thinking and everyday functioning among older adults.

*Operational*. This study defines Alzheimer's disease as a chronic, cognitive disease that causes debilitating dysfunction to the patient and their family.

**Primary care providers**

*Theoretical.* Tabor's defines the primary care provider as a professional who gives health care services or an institution that supervises the rendering of such services.

*Operational.* This study defines the primary care provider as a Doctor of Medicine, Doctor of Osteopathic Medicine, physician's assistant, or a nurse practitioner in a clinical setting who provides health care to patients targeted in this study.

**Primary care setting**

*Theoretical.* Medicine.Net defines a Primary health care setting as a patient's leading source/setting for regular medical care, ideally providing continuity and integration of health care services.



*Operational.* This study defines primary health care as the setting in which the patient shares concerns about the need for genetic screening for Alzheimer's disease.

**Genetic Screening**

*Theoretical.* The National Cancer Institute defines genetic screening as testing designed to identify individuals in a given population who are at higher risk of having or developing a particular disorder or carrying a gene for a specific disease.

*Operational.* This study defines genetic screening as a blood test used to help providers in the primary care setting in Mississippi identify individuals who are at increased risk of developing Alzheimer's disease.

## Assumptions

For this study, the following assumptions were made.

1. The researchers assumed health care providers, in the primary care setting (physicians, nurse practitioners, and physician assistants), were to be adequate and equivalent educational and professional providers that would be practicing with a valid, unencumbered license and within a proficient or expert competency level.



2. The researchers assumed that the sample population of primary care providers assessed a uniform number of individuals that could be considered at risk for Alzheimer’s disease and were familiar with risk factors for Alzheimer's disease.

3. The researchers assumed that the information obtained from this study could be obtained by a survey provided through Survey Monkey, a reliable web-based survey building company.

## Summary of Background

The explanation for the current study was discussed in this chapter. Researchers sought to determine if providers in the primary care setting were identifying individuals/ patients with increased risk of developing Alzheimer's Disease. These individuals were

defined as those with risk factors such as age and family history, diabetes, hypertension, obesity, smoking, depression, cognitive inactivity, physical inactivity, and low education. All of these are crucial elements in identifying the disease-causing gene mutations associated with Alzheimer's disease. The researchers also sought to determine if primary care providers were offering and utilizing genetic screening for those individuals/patients that were identified with positive risk factors for developing Alzheimer's. After reviewing the literature associated with this ongoing health crisis, it was revealed by numerous studies that early testing of patients with a family history or other predispositions to the disease, such as hypertension, diabetes, obesity, smoking, depression, cognitive inactivity, physical inactivity, low education, and specific genetic markers (APOE-e4), could invoke lifestyle changes, such as diet, physical exercise, and cognitive exercise could modify the progression of the disease. It was predicted that early



intervention would be a substantial component in controlling this disease. By directing energy toward identifying patients in primary care who are at high risk for developing Alzheimer's before the clinical onset of the disease, preferably cognitively healthy individuals ages 50-75 with positive risk factors, then the option of genetic testing can be pursued. These genetic variations can later be further identified and aid providers with definitive diagnosis and interpretation of risk for those individuals and their families. Sharing the information collected from this study with primary care providers and the public could prove to be the key to early detection and prevention of this disease.

## CHAPTER II

## Literature Review

Identification of individuals who are at risk of developing Alzheimer's disease is of utmost importance. Early diagnosis allows for tailored counseling and care of patients and their caregivers. Providing symptomatic treatment and support also aids patients and family members in planning for the future. The purpose of this study was to determine if health care providers in the primary care setting were identifying individuals with increased risks for the disease and if genetic screening related to Alzheimer's disease is being utilized in the primary care setting. Extensive research into the genetic etiology of Alzheimer's disease has proven that some genetic factors are causative and increase a person's risk of developing the disease. The need for further comprehensive assessments,

such as genetic testing, for those with increased risk factors such as not only age and family history, but also diabetes, hypertension, obesity, smoking, depression, cognitive inactivity, physical inactivity, low education, and specific genetic markers (APOE-e4) is a critical component in identifying the disease-causing gene mutations associated with Alzheimer's. By focusing efforts identifying patients in the primary care setting who are at increased risk for developing the disease before clinical onset, opens the door for genetic testing. Determining cognitively healthy individuals ages 50-75, who are at increased risk of developing Alzheimer's disease-related to their age or genetic variations in genes, will aid providers with diagnosis and clarification of risk for those individuals and their families. This chapter introduces the theoretical framework utilized in this research, and also presents a review of research as a reference to the current study.





**Theoretical Framework**

Theories are an essential guide to the nursing profession. Nursing theories impact the way providers care for patients and maintain the integrity of the profession. Nola Pender, a pioneer in the nursing profession, made a significant contribution to the nursing profession as the founder of the Health Promotion Model in 1982. Pender's theory/ model is used universally for research, education, and practice. Through revisions and evidenced-based practice, this theory has evolved throughout the years to become the basis for preventive health measures and the critical function of nurses in helping patients prevent illness by promoting well-being and healthy lifestyles. Health promotion is essential for many reasons. The rising costs of healthcare and improving patient

outcomes have become of increasing interest not only to health care workers but also to the general public. The health promotion model focuses on helping individuals achieve higher levels of well-being by providing positive resources to help patients make behavior-specific changes. Research has proven with the implementation of the health promotion model; patients can prevent illness through their behavior. In this paper, Pender's Health Promotion Theory is thoroughly defined, and a thorough critique of research studies utilizing this model will indicate the usefulness of the theory in various populations and disease processes.

Nola Pender defines health as not just being free of disease but includes measures taken to promote good health, the individual's self-perception, and lifestyle. Pender's definition of health is the actualization of inherent and acquired human potential through goal-directed behavior, competent self-care, and satisfying relationship with others while



adjustments are made as needed to maintain structural integrity and harmony with relevant environments (Pender, Murdaugh, & Parsons, 2011). Her definition of health is the basis of the Health Promotion Model. Pender's theory/ model offers a holistic view of the patient, assessing the patient's history and self-perception to allow providers to intervene and form a plan of care accordingly. The theory is meant to serve as a guide to explore complex biopsychosocial processes and motivate individuals to engage in behavior that is directed toward the promotion of health (Pender, Murdaugh, & Parsons, 2011). This guide allows the health promotion theory to be applied to all populations and makes this theory an excellent guide for the current study.

Nurse practitioners and primary care providers play key roles in developing and providing resources for innovative prevention and health promotion programs. The primary emphasis on health promotion has the potential long-term benefits of extending longevity, enhancing the quality of life, and reducing health care costs (Pender & Pender, 1980). The current study addressed these issues, with a focus on determining if genetic screening is being provided in the primary care setting for those with increased risk of Alzheimer's disease. Adding this focus in the primary care setting can allow for early detection and identification of those with increased risks of developing Alzheimer's. Identification of at-risk individuals provides for timely diagnosis and the availability of genetic testing. Early detection can allow both patients and their families to receive effective management, treatment, and support. It can also provide those with increased risks of Alzheimer's disease the opportunity to plan for the future. Utilization of the health promotion model will promote wellness through the use of genetic screening for



Alzheimer's disease, allowing individuals the choice of wellness by providing genetic counseling, support groups, and other resources designated for this disease.

**Review of Literature**

Brodaty, Connors, Loy, Teixeira-Pinto, Stocks, Gunn, Mate, and Pond (2016.) performed a cluster-randomized trial that examined the effectiveness of a peer-led intervention for general practitioners to improve the assessment and management of dementia. The study compared the General Practitioner Assessment of Cognition (GPCOG) and the Mini-Mental State Examination (MMSE) in terms of their ability to detect dementia in primary care. Identifying individuals at risk for developing dementia and Alzheimer’s is often

overlooked in the primary care setting. While a timely diagnosis of the disease is vital for effective management, treatment, and support for patients and their families, the pressure of time constraints in the primary care setting often makes it difficult to assess cognitive decline in this setting adequately. The MMSE, one of the most widely used instruments to test for cognitive deficits, takes approximately ten minutes to perform and makes it impractical to use in the primary care setting. While the GPCOG, also a tool for cognitive impairment, was developed to assess cognitive deficits rapidly and only takes 4-6 minutes to perform. Data on the effectiveness of this screening tool in the primary care setting is limited. In this study, a sample of 2,028 patients was used to compare the GPCOG and the MMSE in their terms of ability to detect cognitive deficits such as dementia and Alzheimer's disease within the primary care setting. The Cambridge Examination for Mental Disorders of the Elderly Cognitive Scale-Revised (CAMCOG)



was used as an index for likely dementia. No theoretical framework was identified for this study.

Brodaty, et al. hypothesized that the GPCOG would perform as well as the MMSE to detect deficits in cognitive function or dementia. The study questioned if the GPCOG could be utilized in the primary care setting, in place of the time-consuming MMSE with comparable results. The sample included elderly adults from the primary care population aged 75 years or older that had visited their general practitioner in the last 24 months. Those with neurological disease, psychotic symptoms, developmental disability, substance abuse, progressive malignancy, or an illness that would prevent the

patient from completing the study were excluded. Written informed consent was obtained from all patients, general practitioners, and informants to participate. Each participant completed a detailed cognitive screen by trained research nurses, and each screening was achieved by utilizing a home visit or a visit to the general practitioner's office. Four instruments were utilized in the baseline cognitive screen. These four instruments included the GPCOG, the MMSE, Geriatric Depression Scale (GDS), and the CAMCOG. The CAMCOG, which uses diagnostic criteria such as memory, language, attention, perception, praxis, and executive function to test each patient's cognitive function. The CAMCOG has 59 items covering seven domains and includes the MMSE as a subtest. The highest possible score on the CAMCOG is 104, with anything <79 is indicative of dementia. This instrument was administered first. The GPOG and GDS followed. The GDS was used to determine if depression influenced the participant's performance of the other tools being used in the study. A 15 item measure was utilized



on this scale. A score >6 indicative of possible depression, with >10 indicating definite depression. The GPCOG consists of two sections; the first section is the patient cognitive examination that is scored 0-9. A score of <5 indicates cognitive impairment, and a score of >8 indicates impairment is unlikely. The second section is an interview with the patient's informant. This section is only given when the patient scores between 5-8 on the first section. The interview section is scored 0-6. A higher score is indicative of better cognition and function. These two scores are totaled to produce the GPCOG-total, a measure of 15. A cut-point score of 10/11 is likely indicative of cognitive impairment. Of the 2,028 participants, 1,717 (84.7%) completed all three relevant instruments

(CAMCOG, GPCOG, MMSE) administered in the study. Of the 1,717 participants, 126 (7.3%) met the criteria for likely dementia by the CAMCOG screening. The MMSE and GPCOG score on these participants were lower than the participants were older than those who did not meet these criteria. An informant interview for the GPCOG was required by 409 of the 1,717 participants. Analysis of the study's results was assisted by McNemar's test to compare sensitivity, specificity, and misclassification rates of the GPCOG and the MMSE. Receiver operating curves (ROC) and Delong's non-parametric test was used to allow comparison of the GPCOG and MMSE, including the area under the curve (AUC). Using published cut-points (10/11 for the GCPOG, and 23/24 for the MMSE) the GCPOG had greater sensitivity than the MMSE in detection of dementia (0.79 vs. 0.51, $p < 0.01$), though lower specificity (0.92 vs. 0.97, $P < 0.01$). The results were consistent despite the GPCOG's shorter administration time, indicating the GPCOG performs similarly to the MMSE in detecting likely dementia. The GPCOG and MMSE



showed similar sensitivities and specificities across the range of test scores using the ROC analysis. These results suggest possible advantages of using the GPCOG as a triage test to identify the increased risk of dementia and Alzheimer's.

Despite limitations within the study, it confirms the GPCOG performs similarly to the MMSE. These results make the study very relevant to the current research study for several reasons. Given the MMSE’s administration time is often impractical in the primary care setting, the findings of the GPCOG’s performance provides a viable alternative for dementia screening. Time investment is a significant limitation in providing cognitive screening to the elderly. Requiring less time to administer, utilizing

the GPCOG in the primary care setting would increase the likelihood of dementia screening being provided. Positive screening would indicate the need for further in-depth assessments by the primary care provider or a referral for further evaluation or management.

The current study will help determine if individuals at increased risk of Alzheimer's disease are being identified and if the utilization of genetic testing is being provided in the primary care setting for individuals with increased risk for Alzheimer's disease. Baseline assessments of dementia and cognitive impairment are critical for further genetic testing of individuals at risk. The ability to provide improved risk assessments followed up with genetic risk testing will aid in diagnosis and potentially clarify risks for the patient and family members affected by the disease. Nola Pender's Health Promotion Model will help guide the current study based on the theory's concept and emphasis on health promotion. This model can be a crucial influence for primary



care providers in utilizing genetic testing for individuals and the family members of those who are at increased risk for Alzheimer's disease.

Evenson, Hoyme, Haugen-Rogers, Larson, and Puumala conducted and reported results of their study, in which they sought to learn about the perceptions of outpatient internal medicine physicians and their patients regarding genetic testing, to help to inform the development of the integration of genetic testing into primary care. As part of a program that will progressively integrate genetic testing into primary care, this study's results will serve as baseline data about these patients and physicians, which will be referred to in future research studies. Genetic testing is becoming a part of primary care,

and the perceptions of both providers and patients are essential to program design and development. Evenson et al. did not indicate a theoretical foundation for their research study.

While Evenson et al. did not clearly state their research questions or hypotheses in the article, the purpose of the research focused on learning more about perceptions of internal medicine physicians and their patients regarding issues related to genetic testing in primary care settings. Questions motivating the research study included: What is the knowledge level of internal medicine physicians about genetic testing in primary care; what is the confidence level of these providers about communication with their patients about genetic testing; how do internal medicine physicians feel about the roles and uses of genetic counselors in primary care; what is the knowledge level of patients of internal medicine physicians about genetic testing; what are patients' perceptions of the usefulness of genetic testing; what are patients' concerns about genetic testing?



These same questions were utilized in their survey to clarify the scope of the research. This research study took place at 13 primary health clinics in three Midwestern states: North Dakota, South Dakota, and Minnesota (Evenson et al., 2016). Due to this study being funded by the University of South Dakota-Sanford School of Medicine, the clinics chosen were part of a program they used to integrate genetic testing into primary care. The physicians and patients surveyed came from these 13 clinics. The researchers prepared surveys to allow them to obtain relevant information. Content experts first approved surveys. After patients visited their internal medicine physicians at one of these 13 clinics, they were given the survey to take home, complete, and send back. For patient

surveys, 1,000 were distributed. Only 14% of the patients responded. All 62 of the internal medicine physicians at these 13 clinics received web-based questionnaires. A total of 42% ($N$ = 26) of the physicians responded. The researchers gathered descriptive data about the respondents. Data reported that the patients included age, education, sex, race, marital status, location, and income. For the physicians, descriptive data included sex, age, years in practice as a physician, and whether or not they had ordered genetic testing in the past six months or ever. Patient surveys consisted of statements about their knowledge of genetic testing, their openness to it, their concerns about it, and their perceptions of its value or benefit. Physician surveys consisted of statements related to knowledge of genetic testing and genetic counselors in primary care. Perceptions of its relevance and the physician's level of confidence in communicating about genetic testing with patients were also included. Case studies were also part of the physician questionnaires.



Patients reported knowledge, interest, and openness to genetic testing in primary care, as well as concerns about privacy; many patients would prefer to hear about their results from a genetic counselor than their physician. Physicians expressed immense knowledge of genetic testing but lower levels of confidence about communicating with patients about it. Implications of this research contribute to the need for ongoing research as genetic testing becomes more integrated with primary care; the authors encouraged further research.

Caselli, Langbaum, Marchant, Lindor, Hunt, Henslen, Dueck, and Robert conducted a study to determine perceptions from a public viewpoint on genetic testing for

Alzheimer's disease. They sought to answer questions based on the desire for genetic testing, the reaction to results once testing was concluded, and the knowledge of those results concerning the disease process. Alzheimer's disease is very prevalent, and it is believed to increase the susceptibility of inheritance if positive for the apolipoprotein E (APOE) genotype. Carrying this genetic makeup poses the most considerable risk of the development of the disease. However, with no known cure for this disease, providers have difficulty with the management of pre-symptomatic testing of Alzheimer's and the proper way to disclose this information to patients. While direct-to- consumer marketing can be a positive resource to some of the population, screening, counseling, and follow-up with consumers is unlikely to occur once purchase is made. However, if genetic testing for Alzheimer's disease was provided in a primary care clinic with a trusted health care provider on individuals who are likely to seek testing, then screening, counseling, and follow-up visits could be scheduled and carried out. The researchers designed a



questionnaire to assess the following: "the desire for preclinical testing in the absence of effective interventions, the possible reactions to such information, and how well the results of such testing would be understood." The researchers developed a questionnaire containing a series of yes/no and multiple-choice questions. Portions of this questionnaire determined the patient's demographics. This included age, sex, race, education, a first degree relative with Alzheimer's disease and other family affected, caregiver experience, and if the individual resided in an area where they were not raised.

The questionnaire proceeded to ask questions about disease perception. It determined the percentage of individuals who found genetic testing to be essential and

separated those who were only in agreement when it was covered by insurance. It determined the individuals' support system, and it revealed how those who hypothetically tested positive would react. These options included: live a healthier life, invest in long term care insurance, spend all money for pleasure or consider suicide. The survey also asked two multiple-choice questions to determine the knowledge of the respondent concerning their potential positive genetic results. The researchers proceeded to post the survey on the Alzheimer's Prevention Registry website. The survey was available online with site visitors who registered on the website between November 1, 2012-June 30, 2013. Those registered with the Alzheimer's Prevention Registry must be over the age of 18 and interested in the development of new research for Alzheimer's disease for personal knowledge development or to be a potential candidate in future research studies related to Alzheimer's disease. A total of 4,036 surveyors participated in the research study. The researchers used descriptive statistics that included the evaluation of continuous and



categorical data. The continuous data were analyzed using unpaired t-tests and univariate regression and presented using mean +/- standard deviation. The questions on the survey were defined as the independent variable. The participants were defined as the dependent variable. The survey included the opinion of genetic testing, isolated those interested in genetic testing, and determined the reaction of the results from the genetic testing. The two multiple-choice questions to determine the participants' knowledge of presymptomatic genetic testing of Alzheimer's disease for each of the previously mentioned demographic variables were analyzed using this method. Also, the categorical data were analyzed using chi-square and multivariate analysis and presented using

frequency. This information included the eight demographic variables that were evaluated using odds ratios and 95% confidence intervals. Once the data was analyzed, the researchers determined a strong desire for pre-symptomatic genetic testing for Alzheimer's disease among individuals. Of the participants, the mean age was 45-80, while the mean education was 12-20 years. Eighty-two-point, one percent of participants were women, and well over half the respondents reported having a first degree relative with an Alzheimer's diagnosis. Seventy-eight-point three percent of the surveyors believed they were at a higher than average risk of developing Alzheimer's disease at some point in their life. The survey asked participants their most significant disease related fear, and 82% of participants selected Alzheimer's disease over a heart attack, stroke, or cancer. With knowledge of no cure for the disease, 70.4% of participants felt genetic testing being offered was imperative. However, 80.8% of participants would elect to have presymptomatic genetic testing if insurance coverage was offered, while only



58.7% would elect for testing with a minimum of $100 out of pocket cost. The participants were also asked how they would respond to possible positive genetic results. Of those asked, 90.5% stated they would commit to a healthier lifestyle, which correlated with the individual's education level. In contrast, 76.3% of individuals would seek to obtain long term care insurance if they had not already, which correlated with age and male gender. Of the surveyors, 18.4% responded to these possible results by choosing to live the rest of their life to the fullest and spend all of their earned money, which correlated mostly with the racial background of white, non-Hispanic individuals. The remaining 11.6% stated they would "seriously consider suicide" if they received positive

genetic results that correlated with education. Lastly, the multiple-choice questions were listed to assess the knowledge of the individuals. The researchers asked about the outcome of an APOE e4 Alzheimer gene carrier with no current signs and symptoms of Alzheimer's disease. Of the participants, 86.9% were able to correctly identify that this carrier was at an increased risk for Alzheimer's disease. Still, it did not currently determine the existence of the disease in the individual. However, of the participants, only 32.6% were able to determine the existence or increased risk for diagnosis of Alzheimer's disease when associated with a positive biomarker test, such as an amyloid PET scan, and mild cognitive delay. From this study, the researchers determined that much more education regarding presymptomatic genetic testing for Alzheimer's disease is needed. The researchers also determined that those who had a positive family history of Alzheimer's disease were more fearful of the development of the disease themselves and were more likely to proceed with genetic testing. However, most individuals stated t



they would be willing to talk with someone, such as a health care provider or family member if they were to test positive for the APOE genetic marker. The researchers determined that psychological evaluation and genetic counseling should be ensured before the information is revealed to individuals who chose to participate in any genetic testing, especially those that currently are without a cure like Alzheimer's disease. They also determined that genetic testing can be beneficial for some, such as easing worry in those who are low risk or allowing those that are of high risk to participate in a healthier lifestyle as a preventative measure. A recommendation for future research within this article would be to expound on the compliance of positive lifestyle changes concerning

prognostic testing for this disease. This article supports the student nurse practitioners' research on The Utilization of Genetic Testing in the Primary Care Setting for Individuals with Increased Risk of Alzheimer's Disease through determining the interest of the public population in genetic testing for Alzheimer's disease. It determines the desire for testing and assesses the public understanding of the testing. With such an increase in direct to consumer genetic testing offered without any follow-up care, genetic counseling, or provider recommendations on a disease process that has limited therapeutic options and no cure, this study supports the idea that this genetic testing should be offered in a primary care setting where the provider can disclose results and can offer memory enhancing medications, genetic and psychiatric counseling, and emotional support.

Huang, Huston, and Perri conducted a research study that sought information regarding the preferences of consumers in the United States regarding predictive genetic testing for Alzheimer's disease (AD), to help to inform health-care providers,



policymakers, and developers of genetic testing. In the United States, Alzheimer's disease is currently the sixth most common cause of death. With the population aging and the prevalence of Alzheimer’s disease increasing, researchers need to gain knowledge about the disease and patient’s perceptions and attitudes about genetic testing (Huang et al., 2014). Specifically, learning how individuals decide on whether or not to undergo predictive genetic testing for Alzheimer’s disease will help health-care providers and policymakers. Huang et al. referred to this study as a “rating conjoint study,” based on Lancaster’s consumer theory. This theory serves as the framework of this study using conjoint analysis, where an individual appreciates consuming a good that is made up of

various characteristics or attributes (Huang et al., 2014). Huang et al. (2014) stated their research question for the study clearly. What characteristics of predictive genetic tests for Alzheimer's disease do consumers in the United States find to be the most important when deciding whether or not to undergo this predictive testing? Hypotheses were not articulated in the article. In this study, the three factors that were rated and analyzed were predictive value, treatment availability, and anonymity.

Through Qualtrics, the researchers distributed an anonymous online survey using conjoint analysis to a panel from the general population (Huang et al., 2014). The survey went through three rounds of pre-testing before the final survey phase of the study. The only inclusion criterion was that respondents had to be adults between the ages of 18 and 64. Twelve scenarios regarding predictive genetic testing for Alzheimer's disease were presented to the respondents, who rated the scenarios by answering 17 questions on an 11-point scale. They also answered open-ended questions about what dollar amount they



would be willing to pay for the genetic testing related to Alzheimer's disease in each scenario. The scenarios each contained different aspects of the three variables: predictive value of the genetic test, treatment availability for Alzheimer's disease, and anonymity. Over four days, a total of 295 participants responded online. The researchers collected these responses ($N = 295$). Descriptive statistics regarding the respondents indicated that their mean age was 44; 86% of the respondents were white; 39% held at least a bachelor's degree; 53% had annual household incomes of $50,000 or less; there were equal numbers of male and female respondents. Regarding Alzheimer's disease, in this sample, 15% of

respondents had a family member with the diagnosis of Alzheimer's disease, and 16% had served as caregivers for someone with the disease.

Huang et al. analyzed the results of the survey and found that the most important of the three factors was accuracy, rated to be of top importance by 64% of the respondents. Treatment availability and anonymity represented 21% and 15% of the respondents' preference ratings. The most preferred scenario was a genetic test for Alzheimer's disease that would be 100% accurate, with a cure available for Alzheimer's disease and anonymous test results. Respondents indicated that they would accept a test that had 80% accuracy if a cure for the disease was available. Regarding their willingness to pay for the test, 12.9% indicated that they would not be willing to pay for any of the scenarios. Of those respondents who did indicate a willingness to pay, they noted that they would pay $100 for the test in the most preferred situation. All scenarios with 100% accuracy had the highest willingness-to-pay results, while those scenarios with as low as 40% accuracy always showed the lower willingness-to-pay results.



Implications of the study include the importance of certainty of predictive genetic tests to consumers. The authors suggested future research studies using samples that are more representative of the general population, as well as future research on consumer preferences as treatment options and predictive value of genetic testing for Alzheimer's disease continue to improve (Huang et al., 2014).

This research identifies the preferences of genetic testing related to Alzheimer's disease and recognizes the benefits of utilizing this testing in the primary care setting. It adds to the knowledge base about patients' perceptions and values regarding genetic

testing. This consumer research study helps give an insight into how patients in the current study will perceive testing.

Wollam, Weinstein, Saxton, Morrow, Fowler, Suever-Erickson, Roecklein, and Erickson performed a neuropsychological evaluation and consensus to determine if the genetic risks of processing multiple risk alleles associated with Alzheimer's disease can be a predictor of increased risk of late-life cognitive impairment. Alzheimer's disease (AD), being the most common cause of irreversible dementia, continues to grow in prevalence and public health impact, according to both the National Institute of Health, and the National Institute of Aging, and Human Services Department. Research on the genetic etiology of Alzheimer's disease has provided knowledge of some genetic factors that are causative and increase the risk of the disease. While family history is a significant risk factor for Alzheimer's disease, other genetic variants have been identified as risk factors for cognitive decline. These variants include Brain-Derived Neurotrophic Factor (BDNF), Catechol-O-Methyltransferase (COMT), and Apolipoprotein E (APOE).



In this study, with a sample of 95 older adults, a genetic risk score was constructed based on the accumulation of these genetic alleles/variants. Identification of these variants in the primary care setting will provide improved significant risk estimates to individuals based on the presence of one or more susceptibility gene alleles. No theoretical framework was identified for this study.

Wollam, et al. identified the hypothesis for this study by the prediction that a higher genetic risk score would correspond to an increased risk of cognitive impairment. By studying target gene variants (genotypes), candidates with increased genetic risks

allowed for a genetic risk score to be assigned. APOE, which identifies a clear link to Alzheimer's Disease; BDNF and COMT, which both have essential roles in brain functions in late adulthood, were targeted to investigate how multiple gene influences can be aggregated into a single risk profile to predict the prevalence that could predispose individuals to Alzheimer's Disease. The testing of the combination of these three high-risk genotypes would be predictive of late-life cognitive impairment.

The study utilized data and sampling methods that were collected as part of more extensive research examining the utility of providing cognitive testing in older adults in primary care physician (PCP) offices. Risk factors for late-onset Alzheimer' Disease identified by the Genome-Wide Association Studies (GWAS) were used to guide this study. The study was conducted in the offices of eleven primary care providers in greater Pittsburgh and surrounding areas. The parental study included 109 participants that were recruited based on the criteria of being 65 years or older, no medical chart diagnosis of dementia, no acute illness, and permanent residence was not a nursing home facility. Of



the 109 Participants that provided consent through the University of Pittsburgh Institutional Review Board before beginning the study, five were excluded initially. The exclusions were related to incorrectly recorded data and allelic frequency differences between races. Nine additional exclusions were made after the study began, related to missing genotype information from one or more genes.

Each participant completed a comprehensive neuropsychological test panel assessing five cognitive domains: memory, executive function, spatial ability, language, and attention/ psychomotor speed. Three expert neuropsychologists made a

determination of the cognitive status. Cognitive status was classified as normal, mild, cognitive impairment (MCI), or dementia. These classifications were based on the University of Pittsburgh Alzheimer's Disease Research Center. With the final diagnosis taking into account, cognitive test scores, as well as demographics, functional, behavioral, and medical information. Adjudications were conducted blind to the study group status of the parent study. Genomic DNA was collected from each participant using the Orange-DNA Self Collection Kit and processed according to laboratory protocol from Orange-DNA. Each individual participant was genotyped for BDNF (rs6265), COMT (rs4680), and APOE( ApoE2, ApoE3, ApoE4). Individual genotype group analysis was also performed. These particular groups consisted of BDNF genotype group-Val homozygotes, Valheterozygotes, or Met homozygotes; COMT- Val homozygotes, Val heterozygotes, or Met homozygotes; and APOE- E2/E3, E2/E4, E3/E#, E3/E4, or E4/E4. The genetic score risk was computed by summing up the presence or absence of each genotype. A "0" was assigned for each genotype with



minimally associated genetic risks with cognitive deficits or a "1" for acknowledged association with cognitive deficits. A risk score was summarized from these three-component risk genotypes. The sum with genetic risk values scored between 0 and 3 was assigned. The value of "0", the absence of any genetic risk genotypes represent the lowest genetic risk category for the decline of cognitive status; "1" represents testing positive for one risk genotype, "2" represents the positive test for two risk genotypes, and "3" represents possession of all three risk genotypes. The number "3" is also representative of the highest genetic risk category for the decline in cognitive status.

Demographic variables, such as age, gender, and years of education, were also included in this analysis.

Analysis of the collected data determined that the hypothesis was statistically supported. Hierarchical logistic regression analyses were used to determine if any of the three genotypes were individually predictive of cognitive status. A genetic risk score was created to represent the accumulation of risk genotypes, such as BDNF, COMT, And APOE. The demographic variates were used to isolate the effects of the genotypes and to test whether the risk score predicted the presence of late-life cognitive impairment. The conclusions were, after controlling variations from age, gender, and education, the APOE carrier status was predictive of an increased risk of cognitive impairment (OR=3.561, P=0.032, and 95% CI=1.116,11.365). Neither BDNF or COMT genotype was related to cognitive status (BDNF, OR=1.149, P=0.755, 95% CI=0.479, 2.759; COMT OR= 1.074, P=0.808, and 95% CI=0.606, 1.903). The results were consistent with the hypothesis; the higher the genetic risk score significantly predicted, the higher the risk of having



cognitive impairment. The study proved a nearly 4-fold increase of cognitive impairment for sample participants with increased genetic risk scores and individual gene polymorphisms as covariates (OR=3.824, P=0.013). These findings support the use of a single genetic risk score to represent the accumulation of genetic influences affecting cognitive and brain health concerning Alzheimer's disease.

The study is relevant to the current research study for several reasons. The effectiveness of genetic risk scores and the potential to use these scores as predictors in cognitive impairment and Alzheimer’s disease will help providers to identify and refer

individuals who are at risk. The current study will help determine if the utilization of genetic testing is being provided in the primary care setting for individuals with increased risk for Alzheimer's disease. The ability to provide improved risk assessments to individuals based on the presence of one or more multiple genotypes will aid in diagnosis and potentially clarify risks for the patient and family members affected by the disease. Nola Pender's Health Promotion Model will help guide the current study based on the theory's concept and emphasis on health promotion. This model can be a crucial influence for primary care providers in utilizing genetic testing for individuals and the family members of those who are at increased risk for Alzheimer's disease.

Shinya Tasaki, Chris Gaiteri, Vladislav A. Petyuk, Katherine D. Blizinsky, Philip L. De Jager, Aron S. Buchman & David A. Bennett conducted a research study to access genetic influence on motor impairment through a case study of older adults and established critical molecular pathways that could mediate the risk. The study was motivated by the increasing evidence of Alzheimer's disease, which impacted late-life in



both cognitive and motor functions. Genetic variation was claimed to have a significant impact on the development of Alzheimer's dementia as well as declined cognitive. The data examined for this study were brain multi-omics, including DNA methylation, transcriptome, histone acetylation (H3K9AC), and targeted proteomics and diverse neuropathology. The source of the data for this research, as stated, was gathered from a total of 552 aged individuals who had at least four out of five omics measurements from DLPFC. No specific theoretical framework was adopted for this study. Research questions and hypotheses for the study are identified. The questions that were formulated

in this research were: What is the genetic risk for Alzheimer's dementia influenced motor functions in older adults? What are the biologic factors linking the genetic risk for developing Alzheimer's dementia with motor impairment molecules and brain pathologies explaining most genetic effects? The researchers sought to establish the relationships between genetic risk variants for AD and motor functioning in older adults.

This study was carried out in the Rush Alzheimer's Disease Center (RADC) in Chicago and employed two longitudinal, community-based aging studies that contained various harmonized data measures, which are collectively termed as ROSMAP. The ongoing studies that the research targeted the older population shows that there is a high genetic risk for AD variants and motor functioning. The researchers employed a sample of more than 3,600 older persons who agreed to annual brain donation evaluation as well as motor testing. A total of 1885 participants completed the genotyping and motor assessments by March 2018. A certified board of neuropathologists reviewed and approved the brain autopsies. The study excluded individuals who had cognitive



impairments but without dementia. Several parameters included motor functions, Parkinsonism score, motor score, neuropathology indices, omics measures, and PRS. On the motor assessment function, two related phenotypes validated in previous studies and without correlation were examined. The global Parkinsonism signs were estimated within a score of 0 to 100 across each domain. Neuropathology indices were also taken and described in supplementary methods and a complete list of brain pathologies assessed. Omics measurements for 7,159,943 single-nucleotide polymorphisms in 2093 subjects were evaluated. Lastly, the PRS generation of variants was identified based on

genome-wide association study data from IGAP and GWAS. The dependent variable was the Alzheimer's dementia motor deficits, while the independent was the genetic factors assessed by the researchers. As stated in the study, “the statistical analyses done involved linear or logistic regression models for testing the association for continuous or categorical outcomes, respectively.” Demographic variables were dropped with the use of a linear model, leaving residuals for use in establishing the association. The residuals were left to test the association at age, sex, years of education, and the first three genotyping principal genotyping components (PCs), giving an account for possible stratification within the population. Other analyses were bioinformatic analyses for evaluation of the AD-PRS effect on motor function. The analysis showed that there were associations within global Parkinsonism and global motor scores when AD-PRS was based on IGAP at an SNP threshold of $p<0.5$. PD-PRSs was also calculated to examine whether the genetic risk for PD explained variances of motor dysfunction in older adults. In this study, they did not correlate with motor scores but moderate with Parkinson's



disease. The overall results implied that Alzheimer’s disease influenced motor functions in older adults meaning that cognitive and motor impairment share at least a part in the genetic architecture. Testing for possible biological factors connecting to genetic risk for Alzheimer’s disease with motor impairment showed that brain pathologies explain most of the genetic effects. The results for this study are useful to the bodies identified by the researchers in that they will be employed in gauging whether they corresponded to those of past studies in the same area. Further suggestions from this study are to establish whether there are genes responsible for impairment lying outside of genes supported by

genome-wide significant loci. Various factors that are assessed through the use of multiple research tools make this study beneficial to the current research. The research provides a strong basis for building research as it allows for an in-depth insight into how the research was conducted. The study shared evidence that the evidence of Alzheimer's disease caused impacts in late-life in both cognitive and motor functions. Genetic variation was proven to have a significant effect on the development of Alzheimer's dementia as well as declined cognitive function. This finding can prove to be valuable information for the current study to assess the genetic influence on motor impairment in older adults. Yokoyama, Bonham, Sears, Klein, Karydas, Kramer, Miller, and Coppola conducted a study to determine the reliability of an assessment that observed multi-variant heritable probability on a group of phenotypically heterogeneous individuals with a current Alzheimer's disease diagnosis. This research study was conducted based on population growth and the medical advances that have allowed individuals to live longer lives. However, as the population ages, the incidence of Alzheimer's disease increases.



Because of the increase in Alzheimer's diagnoses, identifying the individuals at most significant risk for the development of this neurodegenerative disease is imperative. The researchers divided Alzheimer's disease into two categories: the patients who present with amnestic clinical findings and the patients who present with atypical clinical results. Those with amnestic syndromes will present with common findings of progressive memory loss and cognitive impairment. The individuals with atypical clinical outcomes will present with posterior cortical atrophy that affects visual processing, primary aphasia that affects repetition of words and sentences, and dysexecutive/behavioral

symptoms that affects the emotional and reasoning ability of the individual. Due to the variation of the disease process, determining genetic variants has proven difficult for genetic analysis. However, it is estimated that the heritability of Alzheimer's disease is around 74%, with the e4 allele of apolipoprotein E (APOE) being the most significant risk factor of the potential diagnosis. This information inspired the researchers to view the disease from a multi-locus approach in order to "increase the ability to identify individuals at highest risk for Alzheimer's disease syndrome." The investigation that took place in this research study involved two approaches to determine the polygenic risk or probability of the presence of heterogeneous Alzheimer's disease. One approach created and reviewed the credibility to predict Alzheimer's disease through the use of a multi-marker genomic risk score. A discovery associate study had the purpose of reproducing previous findings related to the disease and observing other variations associated with the risk of disease in participants. The second approach used a decision tree analysis that presented the variables and relations between the variables to calculate the result. This



calculation allowed them to determine demographic risk factors and hereditary risk factors that could lead to the development of Alzheimer's disease. The researchers sought to determine the factors and the interactions between variables that allowed them to make an educated prediction on individuals who will develop Alzheimer's disease. Because other research studies determined the APOE genetic marker was the primary genetic contributor of the disease, the research studies sought to confirm or deny this finding and determine other significant genetic markers as well.

This research study is a quantitative, quasi-experimental study. The dependent variable in this study was identified as the participants of the research study, while the independent variable was the genetic analysis. It was conducted through the first selecting participants. The individuals ranged from 65-101 years old and agreed to be subject to genotype analysis. The individuals evaluated could not be related and were made up of Caucasian males (n=216) and females (n=232). Informed consent was received from all participants, and approval was granted from the UCSF Institutional Review Board. The participants then underwent a clinical assessment and were evaluated through a neurological exam, a cognitive assessment, and assessment of any medical history. Participants in this study had a partner, such as a friend, spouse, or child that was assessed to determine functional capabilities and was their source of support throughout the study. The team consisting of a neurologist, neuropsychologist, and a nurse established diagnoses through referring to "consensus criteria for Alzheimer's disease." Atypical Alzheimer's disease diagnoses were also determined, such as logopenic variant PPA, PCA syndrome, primary executive Alzheimer's disease, vascular disease, and



dementia with Lewy bodies. Controls in this study were identified as the Mini-Mental State score of at least 26 or scoring of 0 on the Clinical Dementia Rating Scale. Controls also included the participants to have no report of the decline of cognition within the previous year and could not have shown evidence of a neurodegenerative disorder when observed by the neurological team. If any of these was observed, then the individuals were excluded from the study.

DNA samples were extracted from the participants through a peripheral blood sample. These samples were genotyped along with 75 variants to determine the association with an increased risk of Alzheimer's disease. The 75 variants were determined through observation of previous research studies used to assess the utility of genes involved in "neurodegenerative disease, neurodevelopment, social function, behavior, neuropsychiatry, language on diseases like Alzheimer's disease and frontotemporal dementia." The risk scoring was ranked by p-value. The single-nucleotide genetic variations were removed where the alleles presented with the non-random linked association. The unlinked markers were used to create a scoring set. This scoring set algorithm was implemented using a valid genotype/phenotype analysis toolset to determine its predictability. This process was done to determine both the discovery and validation cohorts. The decision tree analysis was then determined using stopping rules. These included "when subgroup totals were less than 10, when a significance value corresponding to a multiple-testing corrected x^2 test greater than p=0.01 was reached, or when a three-way interaction was reached." These results allowed for the analysis of control groups and all participants. There was also an analysis of control



groups compared with atypical Alzheimer's disease and separately with amnesic Alzheimer's disease. Added predictors consisted of gender, age, and all genetic variants with random association alleles. These predictors allowed for genetic variations to be determined and linked to the development of Alzheimer's disease.

Outcomes of the study determined that the best predictor of Alzheimer's disease was found by repeatedly generating variants until a risk score panel was developed that

was composed of 17 of those variants. When the APOE genetic marker for Alzheimer's disease was evaluated independently; it did not reveal a significant prediction value when compared with the controls. However, in the discovery cohort, the 17-marker risk score identified the clinical significance of the AUC genotype. It concluded that this marker was superior to determine the genetic susceptibility of AD than the APOE alone. In the validation cohort, this did not provide the same outcomes. In this cohort, there were no other genetic variants that determined susceptibility better than the APOE genetic marker. When analyzing all cases of Alzheimer's disease among the participants, being a carrier of the APOE genetic marker was the "first differentiator of cases from controls." Other predictions of genetic risk were determined to be greater than or equal to 77 years old, being a carrier for any of the minor alleles, and being homozygous for the dominant allele, ATP2C2, that controls language traits, such as dyslexia. Because this model used these predicting factors, it was able to diagnose Alzheimer's disease 87% of the time correctly. This study determined that the APOE genotype was the superior predictor of Alzheimer's disease. It also suggested that "phenotype variability in the disease complicates simple genetic risk modeling, particularly when co-morbidities are



suspected," meaning those with atypical Alzheimer's disease were more challenging to predict. However, other studies have been conducted to suggest that atypical Alzheimer's disease is more heritable than amnesic Alzheimer's disease. The researchers recommended a whole-genome association study in the future with more varied participants with specific atypical phenotypes. This study could determine differently genetic susceptibilities specific to Alzheimer's disease. They also suggested, "phenotypic

specificity in studies of amnestic Alzheimer's disease may also provide statistical power to identify risk factors of small effect size."

This research study is beneficial in the APRN students' research project on Alzheimer's' disease and genetic testing. It allows for a more genetic viewpoint on the idea of genetic testing for Alzheimer's disease.There are so many variations of genetic make-up concerning this disease that it is imperative to determine which of these variants will provide the most reliable outcome of an individual's genetic risk. This research study delves into the scientific side of genetic testing and displays the probability of which genetic marker best determines the development of Alzheimer's disease. In determining the specific variation of Alzheimer's disease, whether atypical or amnestic, a physician can determine which genetic testing should be done. APOE genotype is the best predictor of risk for genetic susceptibility among individuals; however, considering the variation of the disease process, amnestic and atypical Alzheimer's disease can have "differential genetic risk factors which can account inaccuracy of the traditional polygenic scoring method." Determining those at highest risk of the disease through being informed on the specific type or variants of genetic markers can allow earlier i



identification and intervention. This data creates the opportunity to provide clinical understanding and provide support before the development of symptoms of Alzheimer's disease.

A detailed review of literature specific to research related to Alzheimer's disease and genetic screening serves the current study well as a guide to promoting the need for further research in this area. The proven research from these studies can be used to direct

strategies to help improve the quality of life for individuals and their families who are or will be affected by Alzheimer's disease. Primary emphasis on health promotion that includes extending longevity, enhancing the quality of life, and reducing health care cost. The current study addresses these issues, with a focus on determining if genetic screening is being provided in the primary care setting for those with increased risks of Alzheimer's disease. Identification and early detection of Alzheimer's disease provide the opportunity for genetic screening that can provide patients and their families the opportunity to receive effective management, treatment, and support.

# CHAPTER III

## Introduction

Because of the increased occurrence of Alzheimer's disease within the United States, the researchers sought to determine if the utilization of genetic testing was being provided in the primary care setting for individuals with increased age, family history, diabetes, hypertension, obesity, smoking, depression, cognitive inactivity, physical inactivity, low education, and specific genetic markers (APOE-e4). These factors place

patients at an increased risk for developing Alzheimer's disease. Because Alzheimer's disease carries such a strong genetic correlation, the researchers hoped to bring awareness to the importance of genetic testing and encourage providers to consider the avenue of genetic testing in their practice in order to allow for preparation and health promotion among the at-risk population. The researchers sought to determine if genetic testing was being conducted by primary health care providers throughout the state of Mississippi.

**Design of the Study**

A quantitative survey was used as the design in this study, and was appropriate for the study to determine if primary care providers were utilizing genetic testing on individuals with an increased risk of developing Alzheimer's disease based on Alzheimer's Association guidelines. Alzheimer's Association guidelines determine risk factors for the disease to include not only age and family history, but also diabetes, hypertension, obesity, smoking, depression, cognitive inactivity, physical inactivity, low education, and certain genetic markers (APOE-
e4).





**Setting of the Study**

The survey was sent electronically to clinics of various settings across the state of Mississippi. These various settings included primary care clinics, urgent care clinics, and specialty clinics. Primary care providers of different credentials were displayed, including Doctors of Medicine, Doctors of Osteopathic Medicine, Nurse Practitioners, or Physician Assistants. The participants were selected at random and represented both

urban and rural areas of the state of Mississippi. The survey was also shared on a private Facebook group of Mississippi Advanced Practice Registered Nurses consisting of 1,700 members.

**Population and Sample**

A convenience sample was utilized for this study. The population of the study included men and women of all ages that presented with increased risk factors for developing Alzheimer's disease based on the guidelines put forth by the Alzheimer's Association. This population was relevant to the study in that age alone does not define the risk for the development of this disease. Alzheimer's Association guidelines state risk factors for the disease include increased age, family history, diabetes, hypertension, obesity, smoking, depression, cognitive inactivity, physical inactivity, low education, and certain genetic markers (APOE-e4). Because of the active genetic link related to Alzheimer's disease, individuals with a family history of the disease are at an increased risk of exhibiting symptoms of Alzheimer's disease at some point in their life. This survey determined if genetic screening was being offered to these individuals by their primary care providers. This information allowed education opportunities for providers



and promoted the maintenance of cognitive health for maximum longevity among patients. Electronic surveys were sent out to varying primary care providers through a reputable survey building website.

**Methods of Data Collection**

After obtaining consent from Mississippi University for Women's Institutional Review Board, the researchers distributed surveys with face validity to primary care

providers across the state of Mississippi. The researchers created a survey using a reliable survey building website, Survey Monkey, Inc. (see Appendix B). These surveys served as data collection tools and assisted in determining if providers were able to recognize those at risk for developing Alzheimer's disease based on the Alzheimer's Association guidelines. The survey also asked questions to determine if those at risk were being educated and offered genetic screening. The survey began by stating the intent of the survey and obtaining the consent to participate. It followed with a statement of privacy, ensuring that no information regarding participants or affiliated clinics would be revealed. The survey was used to collect demographic data to determine the credentials of the primary care provider (nurse practitioner, physician's assistant, or medical doctor) and their specified area of employment. It followed with questions to determine if primary care providers were assessing the patient for risk factors that would place them at an increased risk of developing Alzheimer's disease. It then determined if genetic screening was being offered to the individuals that were at an increased risk. These surveys were sent electronically to primary care providers across the state of Mississippi. The survey was also shared on a private Facebook group of Mississippi



Advanced Practice Registered Nurses consisting of 1,700 members. The surveys did not reveal any protected health information or have any patient identifiers. There were also no participant identifiers as not to skew the research study.

**Methods of Data Analysis**

Upon completion, data collected was deciphered, evaluated, and compiled. The website, Survey Monkey, Inc., initially compiled the data. The data was then released to a

reputable statistician who analyzed and parsed the data using chi-square tests to determine significance. The results were transferred and displayed using varied forms of charts and graphs. All data and research results were shared with the Mississippi Alzheimer's Association research group in hopes of educating providers on the importance of genetic screening for this disease.

**Instrumentation**

The electronic survey was completed using Survey Monkey, Inc., a reliable survey building website. It compiled all the data and preserved it securely until the collection process was completed. The survey consisted of a series of questions developed by the researchers. Questions included demographic data, provider assessments, evidence regarding genetic screening being offered, specific cognitive assessment utilized, reasons genetic screening was not utilized, and the selection of follow-up care provided. The questions were answered by each provider participating in the survey. The surveys were sent electronically to various primary health care providers across the state of Mississippi to determine whether genetic screening was being offered to individuals considered at-risk for developing Alzheimer's disease.



The survey consisted of multiple-choice questions, with three of those being "select all that apply" questions. The survey began by stating a right to privacy and ensuring that no information regarding participants or affiliated clinics would be revealed through participation in this survey. Questions 1 and 2 determined the demographic data, including the credentials of the primary health care provider (Doctor of Medicine, Doctor of Osteopathic Medicine, Nurse Practitioner, or Physician Assistant) and the type of

clinic of affiliation (primary care, urgent care, or specialty clinic). Questions 3 and 4 were related to the research questions and determined if providers were aware of Alzheimer's related risk factors and if they were routinely assessing these risk factors. Question 5 and 6 determined whether the provider was utilizing cognitive testing for patients at risk for cognitive impairment and specified which cognitive tests were being conducted, such as the Mini-Mental State Exam (MMSE), the Mini-Cog test, the General Practitioner Assessment of Cognition test (GPCOG), or other cognitive tests. Question 7 determined if the provider was aware of the research regarding genetic screening for Alzheimer's disease. Question 8 was related to the research questions and determined if genetic screening was being offered or utilized in individuals at an increased risk for developing Alzheimer's disease. Question 9 was a "select all that apply" question and provided the reason genetic screening was not utilized, including time constraints, insurance coverage, or patient refusal. Lastly, question 10 was a "select all that apply" question and assisted in determining provider follow up plan of care which included neurology referral, social services consult, medication for dementia, and/or genetic counseling. Those who chose to participate in this study had their answers submitted



anonymously, and the data was compiled, evaluated, and analyzed to determine the significance of the research study.

**Protection of Human Subjects**

The study was performed once permission was granted from the Institutional Review Board of Mississippi University for Women. Human Subjects were utilized in the conduction of this study through the completion of a survey questionnaire. The survey

determined if genetic screening was being offered to at-risk individuals by primary care providers throughout the state of Mississippi. Strict caution was taken to maintain participant privacy and anonymity as data was being collected. No identifying indicators were revealed, including the participant name or affiliated clinic name. The researchers utilized a reliable, non-traceable survey building website, Survey Monkey, Inc. The data collected was stored in a secure location until analyzed and destroyed once the analysis was concluded. Once the account on Survey Monkey, Inc. was deactivated, all obtained data information was also destroyed. This policy can be located on the Survey Monkey, Inc. website. The data obtained was only used for this research project and was not shared with any affiliated or non-affiliated persons or organizations.

**Summary**

In conclusion, the researchers hoped to provide education on the importance of genetic screening in individuals identified with an increased risk of developing Alzheimer's disease. This research was conducted through surveys distributed across the state of Mississippi to primary health care providers. This allowed the researchers to gather data and determine if individuals were being evaluated and assessed to determine the presence



of risk factors that placed them at a higher risk of developing Alzheimer's disease. It also allowed the researchers to determine if genetic screening was being offered to those at-risk individuals and identified the most common reasons why genetic screening was not offered. Identifying these individuals and promoting genetic screening will allow for yearly follow up, genetic counseling, early detection of cognitive decline, and early intervention.

# CHAPTER IV

## Presentation of Findings

Alzheimer's disease is a progressive neurological illness that affects thousands of individuals every year. The problem was determining whether genetic screening was being fully utilized in the primary care setting for patients that exhibit increased risk factors which include increased age, family history, diabetes, hypertension, obesity, smoking, depression, cognitive inactivity, physical inactivity, low education, specific

genetic markers (APOE-e4) that predispose individuals to Alzheimer's disease. Identifying individuals with increased risk factors is essential because early intervention could provide a solution to controlling this disease and providing statistics for future research. The purpose of this study was to determine if genetic screening was being utilized in the primary care setting for individuals who are at increased risk for developing Alzheimer's disease. The study sought to determine if signs of Alzheimer's disease were recognized quickly in the primary care setting. Surveys were distributed to various primary health care providers across the state of Mississippi to determine whether genetic screening was being offered to individuals considered at-risk for developing Alzheimer's disease. The researchers created a survey using a reliable survey building website, Survey Monkey, Inc. (see Appendix B). These surveys served as data collection tools and assisted in determining if providers were able to recognize those at risk for developing Alzheimer's disease based on the Alzheimer's Association guidelines, and if those patients were being offered genetic screening. The data was collected from the surveys and entered in an Excel document. Information obtained from this study will be





used to help primary providers identify and promote awareness of the signs and risk factors of Alzheimer's disease in their patients.

**Participant Characteristics**

Data for the study were obtained from Survey Monkey, Inc. a reliable survey building website. The research tool was distributed via social media professional groups

that targeted primary care providers, physician assistants, nurse practitioners, and physicians. The survey was made available to hundreds of health care providers across the state of Mississippi. The sample respondents included 20 participants. Of the 20 participants, 18 participants indicated they were nurse practitioners. The other 2 respondents indicated that they were either a family nurse practitioner or a nurse practitioner student.

**Findings**

The study concluded with a summation of findings that identified if patients with increased risk for Alzheimer's disease were being identified in the primary care setting and if primary care providers are utilizing genetic screening for high risk patients for Alzheimer's disease. The research tool provided included ten questions. The questions consisted of type of care provider, area of practice, awareness of risk factors, how often did the provider assess for risk factors, utilization of cognitive testing, tests used, reasons for not utilizing genetic screening, and what follow up plan of care was used for patients exhibited signs of Alzheimer's disease.

When reviewing the data of the study population, the title of care provider was assessed first (See Figure 1). Of the 20 participants, 90% of the care providers indicated



that they were nurse practitioners (n=18) and 10% indicated they were either a family nurse practitioner or a nurse practitioner student (n=2). The next question asked the participant what his or her area of practice was; 65% (n=13) responded that they were in primary practice, 20% in primary care (n=4), and 15% (n=3) indicated that they practiced in another setting, which included the emergency room, nursing homes, and inpatient

behavioral health & addiction (See Figure 2). Of the 20 participants, 80% (n=16) responded that they were aware of the risk factors that predispose patients to developing Alzheimer's disease, whereas 20% (n=4) said that they were not aware of these risk factors (See Figure 3). The next question asked the care provider if they routinely assessed or risk factors for Alzheimer's disease. The survey showed that 70% of the respondents (n=14) indicated that they routinely assessed risk factors for Alzheimer's disease (age, family history, diabetes, hypertension, obesity, smoking, depression, cognitive inactivity, physical inactivity, and low education) and 30% (n=6) said they do not (See Figure 4). When asked if the care provider utilized cognitive testing for individuals with increased risk for cognitive impairment or Alzheimer's disease, most respondents (70% (n=14)) indicated they utilize cognitive testing for individuals with increased risk for cognitive impairment or Alzheimer's disease and 30% (n=6) said that they do not utilize cognitive testing (See Figure 5). The next question asked participants what tests they utilized for testing individuals with increased risk of Alzheimer's disease . Most respondents (65%; N=13) indicated using the MMSE for cognitive testing, either alone or in combination with GDS. The percentage of providers who used the Mini-Mental State Exam (MMSE) test was 50% (n=10), the Mini-Mental State Exam +



Geriatric Depression Scale (MMSE + GDS) test was 15% (n=3), the Cambridge Cognition Exam (CAMCOG) test was 5% (n=1), and No Test Used was 30% (n=6) (See Figure 6). Only 50% of the respondents (n=10) were aware of research regarding genetic screening and Alzheimer's disease, and none of the respondents used genetic screening for individuals with increased risk for Alzheimer's disease. Providers were then asked for

reasons for not utilizing genetic screening for individuals with increased risk for Alzheimer's disease. Those citing insurance coverage concerns were 45% (n=9), insurance coverage + time restraints 20% (n=4), patient refusal 5% (n=1), time restraint concerns 10% (n=2), and other reasons 20% (n=4) (See Figure 7). The most common reason for not utilizing genetic screening for individuals with increased risk for Alzheimer's disease was Insurance Coverage, 65% (n=13). Of the respondents citing insurance coverage as the reason for not utilizing genetic screening, 4 respondents also cited time restraints. Time restraints was the second leading reason cited for not using genetic screening 30% (n=6). Respondents citing other reasons included: 1 being unaware of genetic testing and 3 indicating their area of practice was the reason (Emergency Room, Orthopedics, and 1 did not specify). The respondents next indicated that if the patient screening determined risk factors that predispose the patient to Alzheimer's disease, the follow up plan of care would include the following and all that would apply. Follow up plan of care reported was neurology at 65% (n=13), education related to Alzheimer's disease and plan of care at 45% (n=9), medication therapy at 40% (n=8), lifestyle modification at 30% (n=6), social services consult at 20% (n=4), genetic screening/counseling at 10% (n=2), and Alzheimer's support group at 10% (n=2). Most



(70%) respondents selected more than one item from the follow up plans of care list. Below is a bar graph that summarizes the actual plans of care indicated by respondents (See Figure 8). The final question asked providers if the patient screening determines risk factors that predispose the patient to developing Alzheimer's disease, as well as what his or her follow up plan of care would include. The responses were the following: none

indicated at 10% (n=2), neurology referral at 20% (n=4), social services consult at 5% (n=1), education related to Alzheimer's disease and plan of care (education) at 5% (n=1), neurology referral + medication therapy at 5% (n=1), neurology referral + medication therapy + education at 10% (n=2), neurology referral + medication therapy + lifestyle modification at 5% (n=1), social services + medication therapy + lifestyle modification at 5% (n=1), medication therapy + support group + education at 5% (n=1), genetic screening + education + lifestyle modification at 5% (n=1), neurology referral + education + genetic screening at 5% (n=1), neurology referral + education + support group at 5% (n=1), neurology referral + education + social services consult at 5% (n=1), neurology referral + lifestyle modification at 5% (n=1), neurology referral + education + lifestyle modification at 5% (n=1) (See Figure 9).

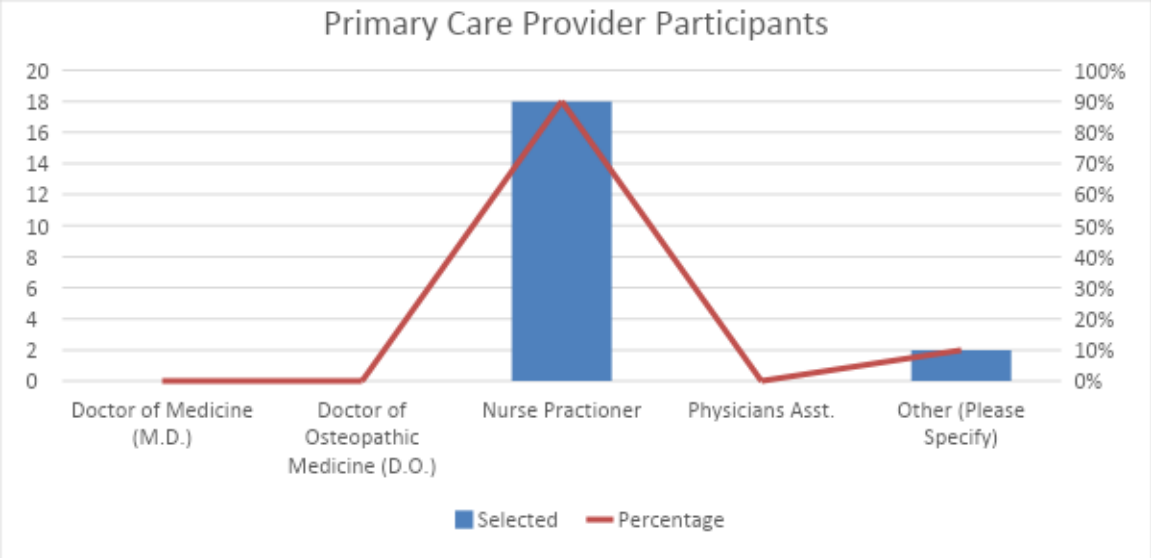


*Figure 1*, Primary Care Providers

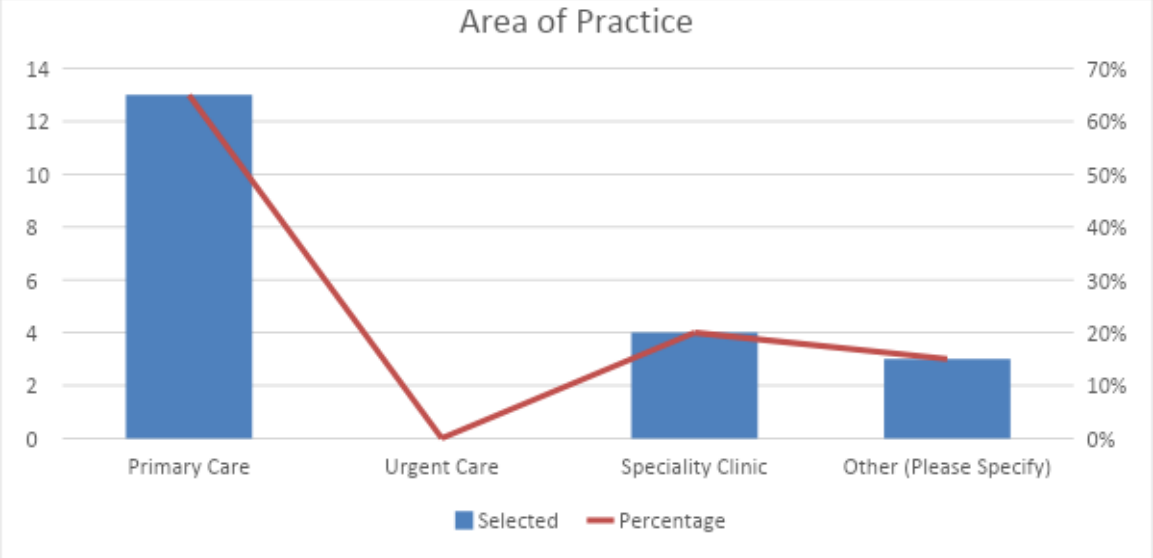


*Figure 2*, Area of Practice

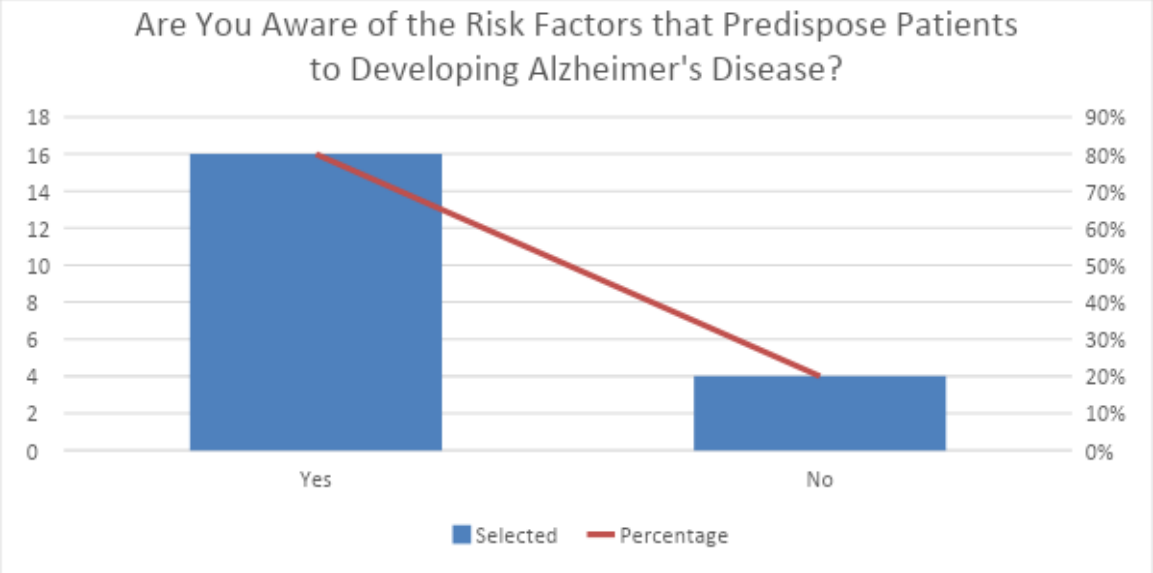


*Figure 3*, Aware of Risk Factors

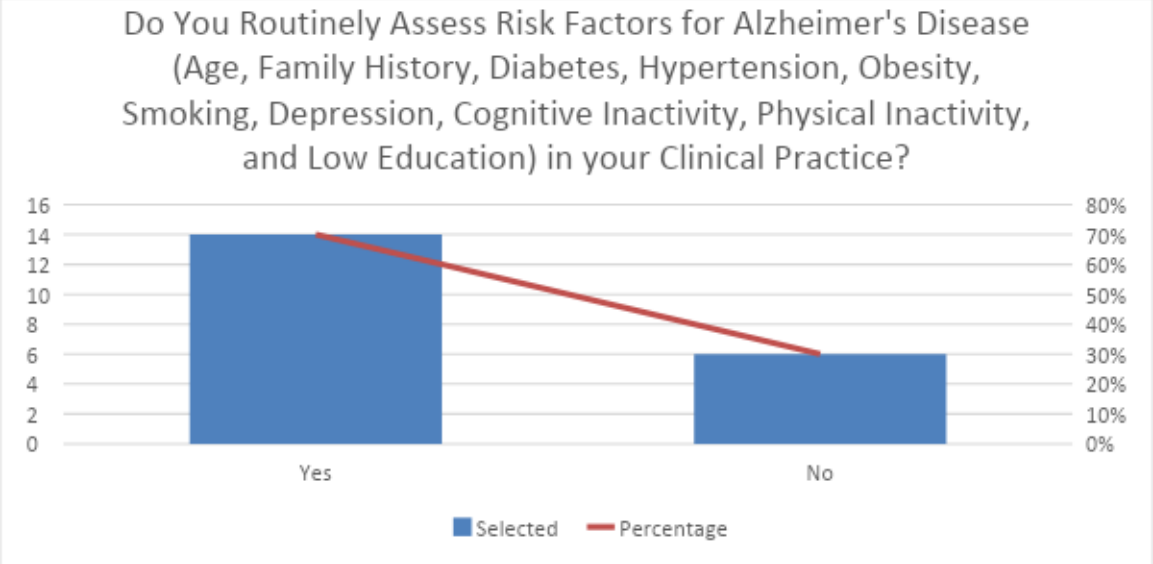


*Figure 4*, Routinely Assess

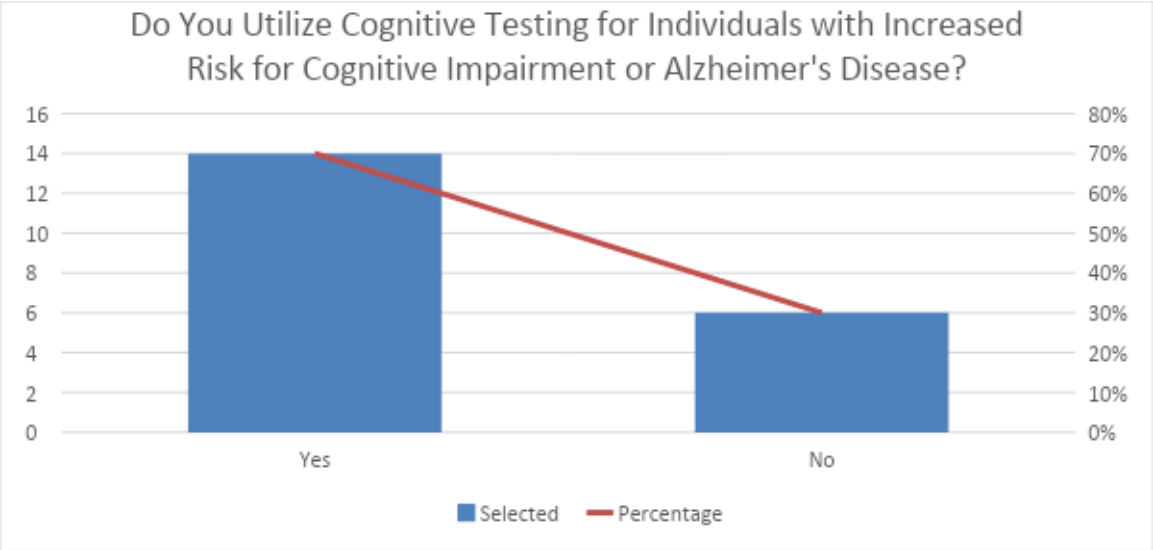


*Figure 5*, Utilize Cognitive Testing

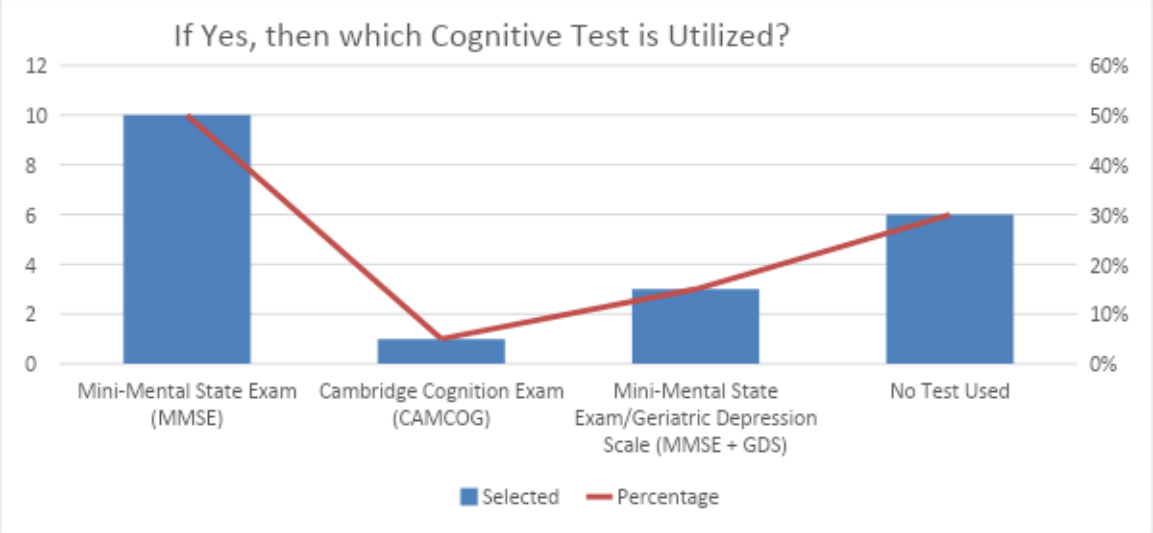


*Figure 6*, Cognitive Test Utilized

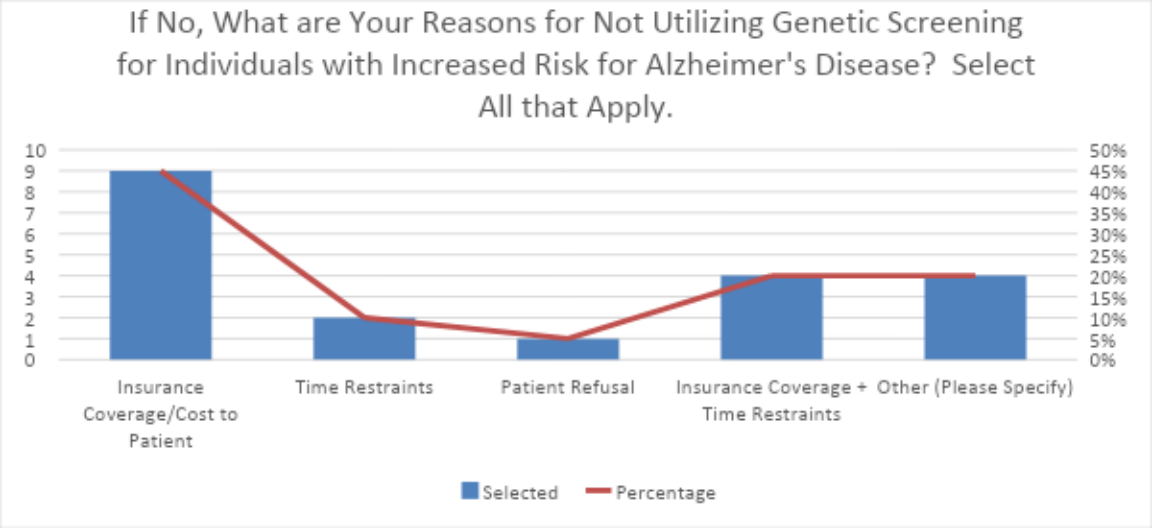


*Figure 7*, Reasons for Not Utilizing Genetic Screening

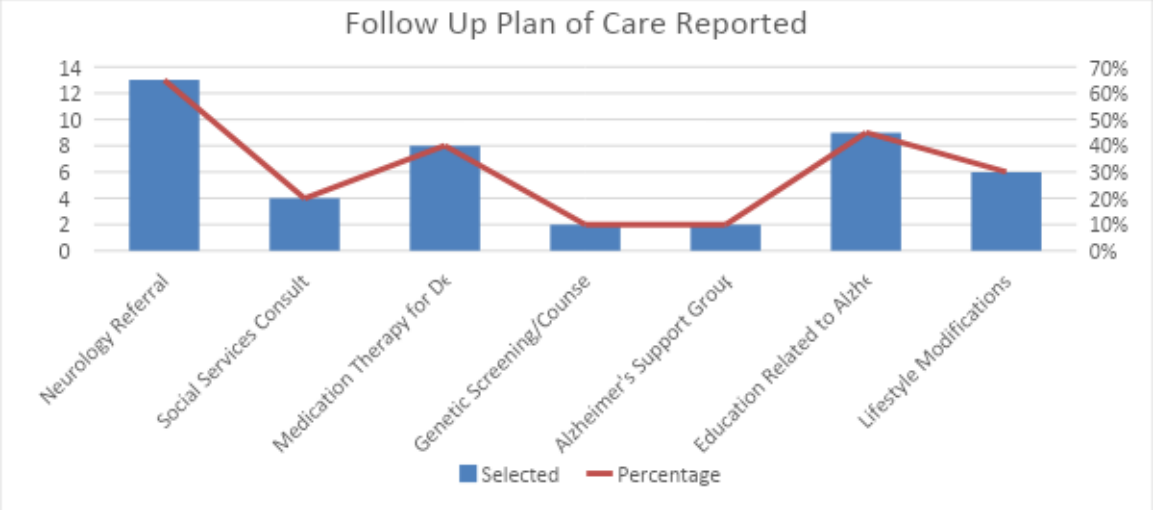


*Figure 8*, Follow Up Plan of Care

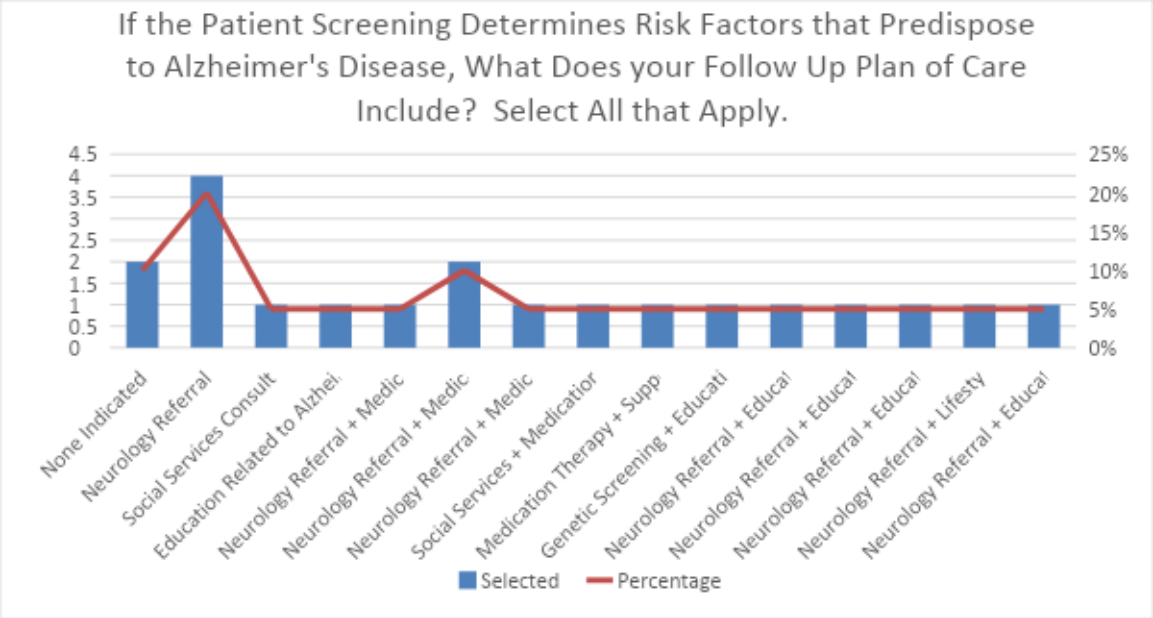


*Figure 9*, Risk Factors Follow Up

**Statistical Findings**

The purpose of this study was to determine if genetic screening was being utilized in the primary care setting for individuals who are at increased risk for developing Alzheimer's disease. The research study produced 20 respondents in this sample. Eighteen of the 20 participants (90%) identified as being Nurse Practitioners. The other two respondents indicated that they were Family Nurse Practitioners or Nurse Practitioner students. When respondents indicated their area of practice, 65% (N=13) responded that they were in primary practice, 20% in primary care (N=4), and 15% (N=3) indicated that they practiced in another setting, which included the ER, Nursing homes, and Inpatient Behavioral Health & Addiction.

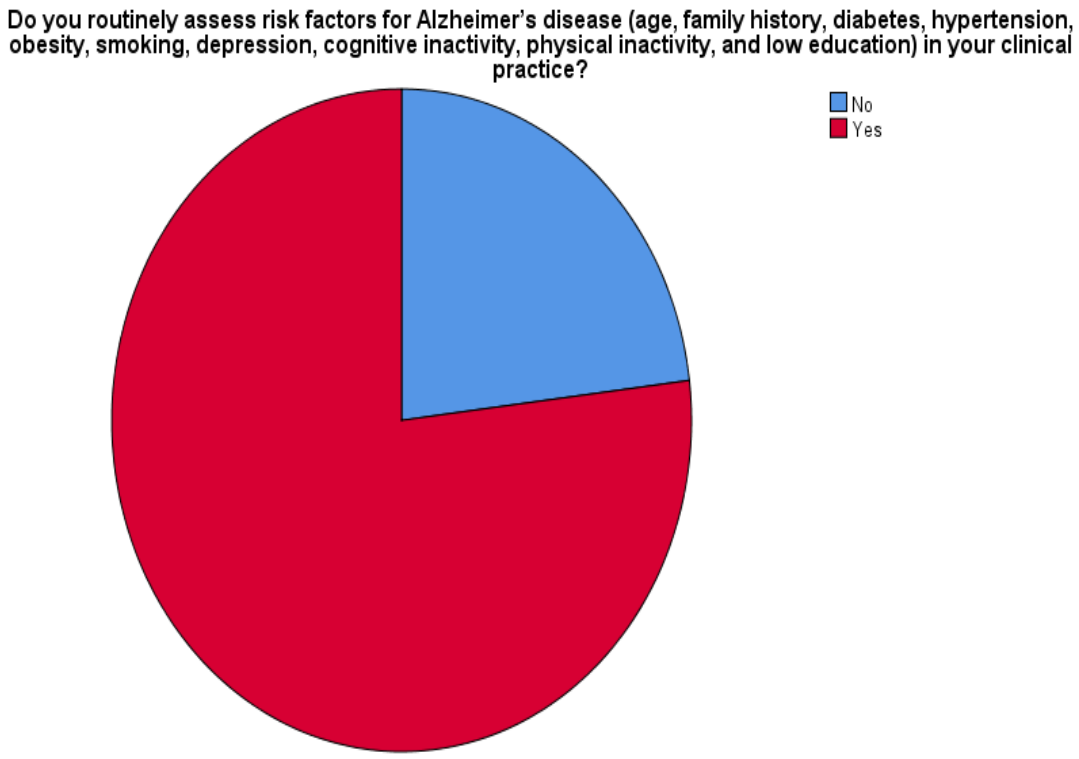


*Figure 10*, Patients at High Risk Being Identified

**Research Question 2**

Are primary care providers utilizing genetic screening for patients who are at increased risk of developing Alzheimer's disease? The answer to this question, based on this sample of respondents is no. None of the 13 primary care providers utilized genetic screening for individuals with increased risk for Alzheimer's disease.

**Reasons for not utilizing genetic screening for individuals with increased risk for Alzheimer's disease**

| | | Frequency | Percent | Valid Percent | Cumulative Percent |
|---|---|---|---|---|---|
| Valid | Insurance Coverage | 6 | 46.2 | 46.2 | 46.2 |
| | Time Restraints | 1 | 7.7 | 7.7 | 53.8 |
| | Patient Refusal | 1 | 7.7 | 7.7 | 61.5 |
| | Insurance Coverage + Time Restraints | 4 | 30.8 | 30.8 | 92.4 |
| | Other- Unaware of test | 1 | 7.7 | 7.7 | 100.0 |
| | Total | 13 | 100.0 | 100.0 | |

*Table 1*. patient or provider time restraints 38.5% (n=5) (See Table 1 & Figure 11).

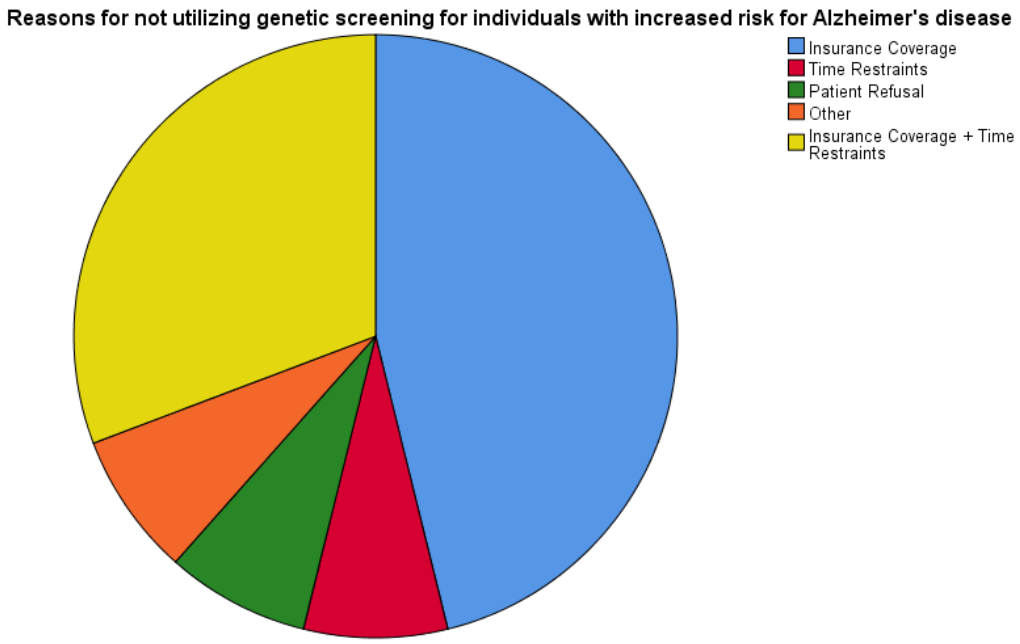


*Figure 11*, Reasons for Not Utilizing Genetic Screening

**Summary**

In total, 20 surveys were collected from participants across the state of Mississippi. The research study addressed the research questions set for this study. It was sought to determine if genetic screening was being utilized in the primary care setting for individuals who are at increased risk for developing Alzheimer's disease. The study revealed that primary care providers are aware of patients that exhibit signs for high risk of developing Alzheimer's disease. The second research question was also addressed when respondents were asked if they were utilizing genetic screening for those with increased risks of Alzheimer's disease. Only 50% of the respondents (N=10)

acknowledged they were aware of the research regarding genetic screening and the disease. There was no data collected that confirmed genetic screening was being offered or utilized for this population.

# CHAPTER V

## Summary and Conclusions

Alzheimer's disease is an irreversible, progressive disease of the brain that slowly begins with the destruction of memory and cognition and eventually leads to the inability to perform necessary activities of daily living. This disorder of the brain is a substantial health problem and the disease occurrence is increasing with the aging population. Unfortunately, there is no standard screening protocol for the diagnosis and progression of the disease. It is the sixth leading cause of death in the United States, accounting for almost 4% of all deaths in 2014 (CDC, 2017). Current disease management focuses on preserving memory and cognition and managing behavioral symptoms. The researchers sought to determine if providers in the primary care setting were identifying individuals/patients at increased risk of developing Alzheimer's. Factors that increased the risk of Alzheimer's disease was defined to include advanced age, family history, diabetes, hypertension, obesity, smoking, depression, cognitive inactivity, physical inactivity, and low education. The researchers also sought to determine if primary care providers were offering and utilizing genetic screening for those individuals/patients who were identified with positive risk factors for developing Alzheimer's. The researchers used Nola Pender's Health Promotion Model (HPM) as the theoretical framework to guide this study.

After reviewing the literature associated with this ongoing health problem, it was revealed by numerous studies that early testing of patients with a family history or other

predispositions to the disease, such as hypertension or diabetes, could invoke lifestyle changes that could modify the progression of the disease. The researchers determined that early intervention could be a substantial component in controlling this disease by identifying patients in primary care who are at high risk for developing Alzheimer's before the clinical onset of the disease, and those with positive risk factors. These genetic variations can later be further identified and aid providers with definitive diagnosis and interpretation of risk for those individuals and their families.

The e4 allele of apolipoprotein E (APOE) gene poses the most substantial risk concerning the development of the disease. It is included in many of the clinical trials and research studies discussed in the review of literature, such as Tasaki et al., where the study determined that the APOE genotype was the superior predictor of Alzheimer's disease. Along with this, Yokoyama et al. also reiterated the strong genetic correlation to Alzheimer’s disease and further supported the APOE genotype as being the most significant risk factor of the potential diagnosis. In this research study patients underwent cognitive testing prior to genetic testing and had to score at least a 26 on the Mini-Mental State Exam score or a 0 on the Clinical Dementia Rating Scale. Genetic testing was then done to test for the APOE gene, and the patients were followed closely with weekly cognitive testing to investigate mental decline. This research provided improved risk assessments on individuals based on the presence of the APOE genotype aided them in diagnosis and potentially clarifying risks for the patient and family members affected by the disease. The current research project sought to exemplify the strong genetic correlation in the review of literature in order to support the importance of genetic t

testing. While this importance was conveyed, there were no primary care providers within the current research study that stated the utilization of genetic testing. Evanson et al. actually found in the research study that physicians expressed immense knowledge of genetic testing but lower levels of confidence about communicating that knowledge with their patients. The current research study did determine lack of knowledge of genetic testing related to Alzheimer's disease as a reason genetic testing was not performed or offered. Along with this, the study by Brodaty et al. revealed time investment as a significant limitation in providing cognitive screening to the elderly. This was also true in the current research study performed. A large portion of responses expressed time constraints as a reason for not utilizing or performing genetic testing on individuals at risk for developing Alzheimer's disease.

In this study, the researchers used a quantitative survey as the design for data collection. Strict caution was taken once approval was granted by the Institutional Review Board at Mississippi University for Women to ensure participant privacy and anonymity as data was collected. The researchers provided questions in the survey using SurveyMonkey, Inc. This survey included demographic data, the determination of provider assessments, the evidence of genetic screening offered, the cognitive assessments used, the reason genetic screening was not utilized, and the follow-up care provided for those who chose genetic screening. The sample size was 20. Upon completion of the data collection, the researchers determined that genetic testing was not being utilized by primary care providers in the state of Mississippi. The summary and

discussion of the findings along with implications, limitations, and recommendations will be discussed for the remainder of the chapter.

**Interpretation of Findings and Conclusions**

The purpose of this study was to determine if genetic screening was being utilized in the primary care setting for individuals at increased risk of developing Alzheimer's disease. The researchers had 20 respondents in this sample. Eighteen of the 20 participants (90%) identified as being Nurse Practitioners. The other two respondents indicated that they were Family Nurse Practitioners or Nurse Practitioner students. When respondents indicated their area of practice, 65% (N=13) responded that they were in primary practice, 20% in primary care (N=4), and 15% (N=3) indicated that they practiced in another setting, which included the ER, nursing homes, and inpatient behavioral health & addiction. When the respondents were asked if they were aware of genetic screening for Alzheimer's disease, only 50% of the respondents (N=10) acknowledged they were aware of the research regarding genetic screening and Alzheimer's disease. When questioned if the risk factors of Alzheimer's disease were routinely assessed, including age, family history, diabetes, hypertension, obesity, smoking, depression, cognitive inactivity, physical inactivity, and low education, 70% of the respondents (N=14) indicated that they did routinely assess for these risk factors. The participants were questioned if they utilized cognitive testing for individuals with increased risk for cognitive impairment or Alzheimer's disease, and most of the respondents (70%, N=14) indicated that they did utilize cognitive testing routinely. Of the respondents who utilized cognitive testing, (65%; N=13), the Mini-Mental State Exam

(MMSE) was selected as the primary assessment for cognitive testing. The respondents replied that they utilized the MMSE either alone or in combination with the Geriatric Depression Scale. When the research question was asked to determine whether genetic screening was offered or utilized in patients with an increased risk of developing Alzheimer's, none of the respondents stated utilization of genetic screening for individuals with increased risk for Alzheimer's disease. When questioned about the reasoning of not utilizing genetic testing, the respondents 65% (N=13) indicated insurance coverage prevented them from offering genetic testing. Of the respondents stating insurance coverage as a reason not to utilize genetic screening, four respondents also stated time restraints as a limitation. Time restraints were the second leading reason cited for not using genetic screening (30%; N=6). Four respondents cited other reasons genetic testing was not utilized, including one respondent unaware of genetic testing, and three respondents in an area of practice where it was not considered applicable (ER, Orthopedics, and one did not specify). According to the results of this survey, patients who are at increased risk of developing Alzheimer's disease are being identified and screened. Thirteen of the respondents work in a primary care setting. Of these, 76.9% (N=10) routinely assessed risk factors for Alzheimer's disease. Of those who routinely assessed risk factors of Alzheimer's disease, eighty percent (N=10) utilized cognitive testing for these individuals at increased risk for developing Alzheimer's Disease. Genetic testing was not being offered or utilized within the primary care setting to individuals at risk for developing Alzheimer's. While most primary care providers were identifying and screening these individuals at increased risk for developing Alzheimer's, none of the 13

primary care providers who participated in the survey offered or utilized genetic screening for these identified individuals. Of the 13 primary care providers identified, the key reasons chosen for not utilizing genetic screening was lack of insurance coverage (76.9%; N=10) and patient/provider time restraints (38.5%; N=5). To determine the follow up plan of care once risk factors had been identified,, the respondents were provided with a question in the format of selecting all that apply. The following options were listed: neurology referral, social services consult, education related to Alzheimer's and plan of care, lifestyle modifications, genetic counseling, and medication therapy. Seventy percent of respondents selected a combination of the listed options. Two of the respondents selected that they would not include any of the options in the at-risk patient's follow-up plan of care. Four respondents (20%) selected a neurology referral alone, while one respondent (5%) selected only a social services consult. One respondent (5%) opted to utilize only education regarding Alzheimer's disease. Two respondents (10%) selected a combination of medication therapy, education regarding Alzheimer's disease, and neurology referral as their preferred follow-up plan of care. One respondent (5%) selected a neurology referral and medication as a desired plan of care while another respondent (5%) selected neurology referral, medication therapy, in addition to lifestyle modifications. One respondent (5%) chose to utilize social services, medication, and lifestyle modifications, while another respondent (5%) relied on medication, genetic counseling, and education regarding the disease process. Another respondent (5%) selected genetic screening, medication, and lifestyle modifications, while a separate respondent (5%) utilized genetic screening and medication but preferred genetic

counseling over lifestyle modifications. Another (5%) respondent opted for a neurology referral, education regarding respondent Alzheimer's, and lifestyle modifications while another respondent (5%) only selected a neurology referral and education. The final respondent (5%) selected a neurology referral and a social service consult regarding the treatment plan of care of an individual identified as at increased risk of developing Alzheimer's. Overall, 13 of the 20 respondents selected either alone or in combination to refer to their patient identified to be at an increased risk of developing Alzheimer's to neurology for management of the disease process. While none of the survey participants offered or utilized genetic screening to individuals at risk for developing Alzheimer's, two of the respondents did include genetic screening along with a combination of other options in their proposed plan of care.

There were two research questions addressed in this study. Are patients at increased risk for developing Alzheimer's disease being identified by providers in the primary care setting; and are primary care providers utilizing genetic screening for patients at increased risk for developing Alzheimer's disease? When reviewing the current practices for screening patients of patients with increased risk, seventy-nine percent of providers routinely assessed risk factors for Alzheimer's disease. Eighty percent of the respondents identified as utilizing cognitive testing. None of the respondents offered genetic testing for the patients at increased risk for developing Alzheimer's. This study highlighted the lack of genetic testing as being utilized.

The findings of the current researchers were similar to those of Brodaty et al. in that both studies suggest that cognitive testing is beneficial for a timely diagnosis of Alzheimer's disease.

**Limitations**

The limitations of the study which were identified prior to data collection were only a small geographical area was studied, a small sample size being collected, and limited distribution of the study was distributed via social media outlets to distribute to the appropriate audience, this limitation only included feedback from Nurse Practitioners which limited access to gather and collect data. The research questions only addressed primary care providers, neglecting those that work in specialty areas such as neurology, and psychology that are also capable of utilizing genetic testing, thereby limiting the range of responses. The reliability of survey data was also considered a limitation not due to the respondents providing accurate honest answers. Lack of confidence in the subject area could also limit the study to biased responses.

The major limitation on the research study was a global pandemic. COVID-19 created numerous limitations in health care and prevented researchers from hands on collection of data. Alzheimer's disease is the most common cause of dementia. This disease occurs worldwide and affects all ethnic groups. The findings concluded in this study were that patients who are at increased risk of developing Alzheimer's disease are being identified and screened, and over half of the respondents identified as routinely assessing risk factors. Those who regularly evaluate risk factors (80%) utilize cognitive testing for the individuals at increased risk. Only 50% of the respondents (N=10) were

aware of research regarding genetic screening. The researchers concluded that none of the respondents used or offered genetic screening for individuals with increased risk for Alzheimer's disease. The primary reason for not using or offering genetic screening for individuals with increased risk for Alzheimer's disease as insurance coverage.

**Implications and Recommendations**

It is estimated by 2050, 16 million people will be diagnosed with Alzheimer's (CDC, 2017). It is the sixth leading cause of death in the United States (CDC,2017). Modifiable risk factors such as diet, and cognitive challenges have been shown to slow the disease process (CDC, 2017). Medical management can improve the quality of life for individuals living with Alzheimer's disease and their caregivers. There is currently no known cure for Alzheimer's disease. In order to provide education on modifiable risk factors, the providers must be aware and up to date on genetic screenings.

Our research project revealed that although Primary Care Providers in Mississippi are aware of the availability of genetic screening, very few are utilizing genetic testing for patients with increased risk for Alzheimer's disease. The most common reason for not implementing genetic screening for their patients with risk factors for Alzheimer's was insurance coverage, followed by time restraints. This study implies that even though the awareness is there, the resources are not utilized. With the results of this research study, providers can explore opportunities to implement better screening and utilization of existing resources into their practices. This survey was specifically designed to test the knowledge and utilization of the available testing for patients at an increased risk of developing Alzheimer's. With early knowledge, the primary care provider could

implement lifestyle changes that could prolong the patient's quality of life. The nursing theory used in this study shows that a patient will do better if they know to do so. Increasing knowledge not only increases awareness for the patient but the provider, as well.

Recommendations that arise from the results of this research study include increasing awareness of the benefits of genetic testing for patients with increased risk of developing Alzheimer's Disease as well as increasing the sample size and the geographical area. Mass marketing on Social media and television could encourage patients to start asking questions about their care, and the options they have. When patients are included in their plan of care, they are better to adhere to treatment options, and make those lifestyle changes to prevent or slow the progression of developing the disease. Future research could expand their sample size by doing paper surveys, as well as face to face interviews to obtain feedback. Educating Primary Care Providers and the public would increase the demand for such testing. Education plays a significant factor in the lack of use of any resource. Education on the benefits of early intervention for those at-risk patients, for their families, as well as the Primary Care Provider, would also increase the usage of this resource. Recommendations for Advanced Nursing Practice is the implementation of a guideline for screening would be beneficial. For example, in the electronic medical record (EMR), there could be screening questions that target cognitive decline. There could be standardized steps to implement in the early stages of decline. More focus on family history should be a talking point with the patient. Family history could be a reliable indicator of the need for early detection by genetic screening.

Insurance Coverage was the most common factor that prevented the utilization of genetic screening. If providers utilize genetic testing more frequently, insurance companies will begin to include it in their covered benefits. Also, lobby for insurance coverage for genetic screening as a preventative service to decrease the patient's rate of decline, which, in turn, reduces insurance pay-out.

Recommendation for future research can be done to assess the provider willingness to utilize genetic testing if education was more readily available, and insurance coverage was not an issue. If all of the stated barriers were removed, what would be the patient's increased benefit? Would their quality of life be improved? Would the patient change anything about their lifestyle? Would mental health be more of a priority in the routine care of patients? These questions could be the starting points for future discussions.

**Summary**

This study was conducted to provide education on the importance of genetic screening in individuals identified with an increased risk of developing Alzheimer's disease. The study was conducted through a quantitative survey distributed to health care providers across the state of Mississippi. This allowed researchers to gather data and determine if individuals who were at increased risk of developing Alzheimer's disease were being identified and offered genetic screening. While the study revealed most primary care providers were identifying and screening these individuals who were at increased risk for developing Alzheimer's, none of the providers that participated in the survey offered or utilized genetic screening for these identified individuals. The study also provided insight to the most common reasons why genetic screening was not offered,

which was found to be time constraints and insurance coverage. Studies have been conducted to prove the identification of individuals with increased risks of Alzheimer's disease and the promotion of genetic screening in the primary care setting would allow for yearly follow up, genetic counseling, early detection of cognitive decline, and early intervention. There are many limitations to this study that can be improved upon and corrected for future research. This study provides a substantial foundation and realization that although risk factors are being identified in the primary care setting, genetic screening is not being offered. Utilization of this data, along with further research will be beneficial to promote the importance of early intervention for those at risk of Alzheimer's disease.

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Appendix A

The W Mississippi University for Women
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March 30, 2020

Dr. Carey McCarter
College of Nursing and Health Sciences
1100 College St. W-910
Columbus, MS 39701

Dear Dr. McCarter:

I am pleased to inform you that the members of the Institutional Review Board (IRB) have reviewed the following proposed research and have approved it as submitted:

| | |
|---|---|
| **Name of Study:** | Utilization of Genetic Screening by Primary Care Providers for Individuals with Increased Risks for Alzheimer's Disease. |
| **Research Faculty/Advisor:** | Carey McCarter |
| **Investigators:** | MSN Students |

I wish you much success in your research.

Sincerely,

Scott Tollison

Scott Tollison, Ph.D.
Provost and Vice President for Academic Affairs

ST/tc

pc: Irene Pintado, Institutional Review Board Chairman

Survey

Instructions to participants are as follows:

- By completing this survey, you are giving consent to participate.
- This survey is both a demographic and data collection survey to determine if genetic testing is utilized in the primary care setting for individuals with an increased risk of Alzheimer's disease.
- Participants will remain anonymous.
- No participant shall identify their employment facility.

Please select the most appropriate answer to the following questions:

1. Please select which best applies.
   A. Doctor of Medicine (M.D.)
   B. Doctor of Osteopathic Medicine (D.O.)
   C. Nurse Practitioner
   D. Physician's Assistant
   E. Other:_____

2. Please select which best describes your area of practice.
   A. Primary Care Clinic
   B. Urgent Care Clinic
   C. Specialty Care Clinic / Other:_____

3. Are you aware of the risk factors that predispose patients to Alzheimer's disease?
   A. Yes
   B. No

4. Do you routinely assess risk factors for Alzheimer's Disease (age, family history, diabetes, hypertension, obesity, smoking, depression, cognitive inactivity, physical inactivity, and low education) in your clinical practice?
   A. Yes
   B. No

5. Do you utilize cognitive testing for individuals with increased risk for cognitive impairment or Alzheimer's disease?
   A. Yes
   B. No

6. If yes, which cognitive test is utilized?
   A. MMSE
   B. CAMCOG
   C. GPCOG
   D. Geriatric Depression Scale (GDS)
   E. Other

7. Are you aware of research regarding genetic screening and Alzheimer's Disease?
   A. Yes
   B. No

8. Do you offer/ utilize genetic screening for individuals with increased risk for Alzheimer's Disease?
   A. Yes
   B. No

9. If no, what are your reasons for not utilizing genetic screening for individuals with increased risk for Alzheimer's disease? Select all that apply.
   A. Insurance coverage/ Cost to patient
   B. Time restraints
   C. Patient refusal

10. If the patient screening determines risk factors that predispose to Alzheimer's disease, does your follow up plan of care include: Select all that apply.
    1. Neurology referral
    2. Social services consult
    3. Medication for dementia
    4. Genetic screening/counseling
    5. Alzheimer’s support group
    6. Education related to Alzheimer’s disease and treatment.

APPENDIX C

**Descriptive Statistics**

There were 20 respondents in this sample. Eighteen of the 20 participants (90%) indicated being Nurse Practitioners. The other two respondents indicated that they were FNP or NP students.

**Professional title**

| | | Frequency | Percent | Valid Percent | Cumulative Percent |
|---|---|---|---|---|---|
| Valid | Nurse Practitioner | 18 | 90.0 | 90.0 | 90.0 |
| | FNP/NP Student | 2 | 10.0 | 10.0 | 100.0 |
| | Total | 20 | 100.0 | 100.0 | |

When indicated the area of practice, 65% (N=13)responded that they were in Primary practice, 20% in primary care (N=4), and 15% (N=3) indicated that they practiced in another setting, which included the ER, Nursing homes, and Inpatient Behavioral Health & Addiction.

**Are you aware of the risk factors that predispose patients to developing Alzheimer's disease?**

| | | Frequency | Percent | Valid Percent | Cumulative Percent |
|---|---|---|---|---|---|
| Valid | No | 4 | 20.0 | 20.0 | 20.0 |
| | Yes | 16 | 80.0 | 80.0 | 100.0 |
| | Total | 20 | 100.0 | 100.0 | |

70% of the respondents (N=14) indicated that they routinely assessed risk factors for Alzheimer's disease (age, family history, diabetes, hypertension, obesity, smoking, depression, cognitive inactivity, physical inactivity, and low education)

**Do you routinely assess risk factors for Alzheimer's disease (age, family history, diabetes, hypertension, obesity, smoking, depression, cognitive inactivity, physical inactivity, and low education) in your clinical practice?**

| | | Frequency | Percent | Valid Percent | Cumulative Percent |
|---|---|---|---|---|---|
| Valid | No | 6 | 30.0 | 30.0 | 30.0 |
| | Yes | 14 | 70.0 | 70.0 | 100.0 |
| | Total | 20 | 100.0 | 100.0 | |

Again, most of the respondents (70%, N=14)) indicated that they utilize cognitive testing for individuals with increased risk for cognitive impairment or Alzheimer's disease.

**Do you utilize cognitive testing for individuals with increased risk for cognitive impairment or Alzheimer's disease?**

| | | Frequency | Percent | Valid Percent | Cumulative Percent |
|---|---|---|---|---|---|
| Valid | No | 6 | 30.0 | 30.0 | 30.0 |
| | Yes | 14 | 70.0 | 70.0 | 100.0 |
| | Total | 20 | 100.0 | 100.0 | |

Most respondents (65%; N=13) indicated using the MMSE for cognitive testing, either alone or in combination with GDS.

**Test utilized**

| | | Frequency | Percent | Valid Percent | Cumulative Percent |
|---|---|---|---|---|---|
| Valid | No test used | 6 | 30.0 | 30.0 | 30.0 |
| | MMSE | 10 | 50.0 | 50.0 | 80.0 |
| | MMSE + GDS | 3 | 15.0 | 15.0 | 95.0 |
| | CAMCOG | 1 | 5.0 | 5.0 | 100.0 |
| | Total | 20 | 100.0 | 100.0 | |

Only 50% of the respondents (N=10) were aware of research regarding genetic screening and Alzheimer's disease, and **none** of the respondents used genetic screening for individuals with increased risk for Alzheimer's disease.

The most common reason for not utilizing genetic screening for individuals with increased risk for Alzheimer's disease was Insurance Coverage, 65% (N=13) of respondents indicating that this was a reason. Of the respondents citing insurance coverage as a reason not to utilize genetic screening, four respondents also cited time restraints. Time restraints was the second leading reason cited for not using genetic screening (30%; N=6). Four respondents cited other reasons, including one being unaware of genetic testing and three indicated their area of practice (ER, Orthopedics, one did not specify).

**Reasons for not utilizing genetic screening for individuals with increased risk for Alzheimer's disease**

| | | Frequency | Percent | Valid Percent | Cumulative Percent |
|---|---|---|---|---|---|
| Valid | Insurance Coverage | 9 | 45.0 | 45.0 | 45.0 |
| | Time Restraints | 2 | 10.0 | 10.0 | 55.0 |
| | Patient Refusal | 1 | 5.0 | 5.0 | 60.0 |
| | Insurance Coverage + Time Restraints | 4 | 20.0 | 20.0 | 80.0 |
| | Other | 4 | 20.0 | 20.0 | 100.0 |
| | | | | | |
| | Total | 20 | 100.0 | 100.0 | |

The respondents indicated that if the patient screening determine risk factors that predispose to Alzheimer's disease, the follow up plan of care would include the following:

| Follow up plan of care reported | Frequency | Percentage |
|---|---|---|
| Neurology | 13 | 65.0 |
| Education related to Alzheimer's disease and plan of care | 9 | 45.0 |
| Medication Therapy | 8 | 40.0 |
| Lifestyle modification | 6 | 30.0 |
| Social Services Consult | 4 | 20.0 |
| Genetic Screening/Counseling | 2 | 10.0 |
| Alzheimer's Support Group | 2 | 10.0 |

Most (70%) of respondents selected more than one item from the follow up plans of care list. Below is a table that summarizes the actual plans of care indicated by respondents.

**If the patient screening determines risk factors that predispose to Alzheimer's disease, does your follow up plan of care include:**

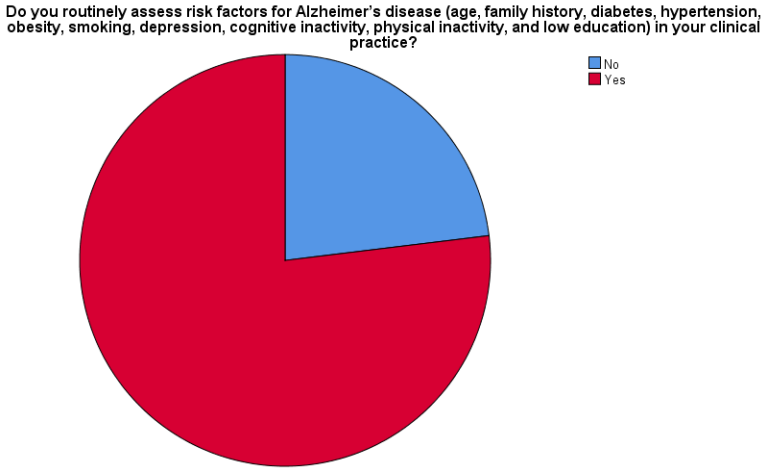
| | | Frequency | Percent | Valid Percent | Cumulative Percent |
|---|---|---|---|---|---|
| Valid | None indicated | 2 | 10.0 | 10.0 | 10.0 |
| | Neurology referral | 4 | 20.0 | 20.0 | 30.0 |
| | Social Services Consult | 1 | 5.0 | 5.0 | 35.0 |
| | Education related to Alzheimer's disease and plan of care (Education) | 1 | 5.0 | 5.0 | 40.0 |
| | Neurology referral + Medication Therapy | 1 | 5.0 | 5.0 | 45.0 |
| | Neurology referral +Medication Therapy +Education | 2 | 10.0 | 10.0 | 55.0 |
| | Neurology referral + Medication Therapy + Lifestyle Modification | 1 | 5.0 | 5.0 | 60.0 |
| | Social Services + Medication Therapy+ Lifestyle modification | 1 | 5.0 | 5.0 | 65.0 |
| | Medication Therapy +Support Group +Education | 1 | 5.0 | 5.0 | 70.0 |
| | Genetic Screening +Education +Lifestyle Modification | 1 | 5.0 | 5.0 | 75.0 |
| | Neurology Referral +Education +Genetic Screening | 1 | 5.0 | 5.0 | 80.0 |
| | Neurology Referral + Education + Support Group | 1 | 5.0 | 5.0 | 85.0 |
| | Neurology referral +Social Services Consult | 1 | 5.0 | 5.0 | 90.0 |

| | | | | | |
|---|---|---|---|---|---|
| | Neurology Referral + Lifestyle modification | 1 | 5.0 | 5.0 | 95.0 |
| | Neurology Referral+ Education +Lifestyle modification | 1 | 5.0 | 5.0 | 100.0 |
| | Total | 20 | 100.0 | 100.0 | |

**Research Questions:**

**1. Are patients who are at increased risk of developing Alzheimer's disease being identified by providers in the primary care setting?**

According to the results of this survey, patients who are increased risk of developing Alzheimer's disease are being identified and screened. Thirteen of the respondents work in a primary care setting. Of these, 76.9% (N=10) routinely assess risk factors for Alzheimer's disease. Of those that routinely assess risk factors of Alzheimer's disease, 80% (N=10), utilize cognitive testing for these individuals at increased risk.

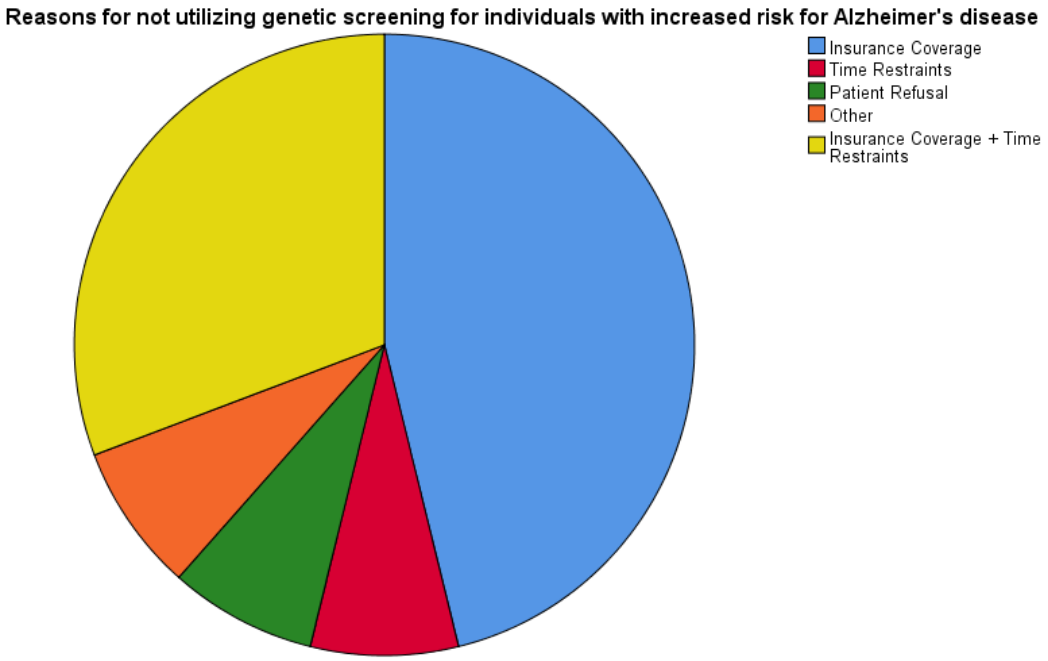



**2. Are primary care providers utilizing genetic screening for patients who are at increased risk of developing Alzheimer's disease?**

The answer to this question, based on this sample of respondents is no. None of the 13 primary care providers offers/utilized genetic screening for individuals with increased risk for Alzheimer's disease. For this subset of the sample, the primary reasons given for not using genetic screening in Insurance Coverage (76.9%; N=10) and Patient/Provider time restraints (38.5%; N=5).

**Reasons for not utilizing genetic screening for individuals with increased risk for Alzheimer's disease**

| | | Frequency | Percent | Valid Percent | Cumulative Percent |
|---|---|---|---|---|---|
| Valid | Insurance Coverage | 6 | 46.2 | 46.2 | 46.2 |
| | Time Restraints | 1 | 7.7 | 7.7 | 53.8 |
| | Patient Refusal | 1 | 7.7 | 7.7 | 61.5 |
| | Insurance Coverage + Time Restraints | 4 | 30.8 | 30.8 | 92.4 |
| | Other- Unaware of test | 1 | 7.7 | 7.7 | 100.0 |
| | Total | 13 | 100.0 | 100.0 | |


Reasons for not utilizing genetic screening for individuals with increased risk for Alzheimer's disease
Insurance Coverage
Time Restraints
Patient Refusal
Other
Insurance Coverage + Time Restraints