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Understanding Barriers To Genetic Testing For Sickle Cell Trait: The African-American Male Perspective

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UNDERSTANDING BARRIERS TO GENETIC TESTING FOR SICKLE CELL TRAIT: THE AFRICAN-AMERICAN MALE PERSPECTIVE

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ABSTRACT

Research has shown a reluctance in African-American males to pursue testing for sickle cell trait. Few studies have tried to discern what barriers are contributing to this issue within the African-American male community. Research suggests a lack of knowledge may be the biggest contributing factor. This study hypothesized there would be a significant difference in knowledge of sickle cell trait based on educational level, age, and health beliefs. African-America male participants (N=116), ages 18 and over, completed a questionnaire assessing knowledge, risk perception, health beliefs, barriers, and motivating factors within the context of sickle cell trait. One-way and two-way analysis of variance identified age as an influential factor. Results showed a significant interaction between age and knowledge of sickle cell trait and sickle cell disease ($p = .009$). Factors including perceived discrimination, perceived risk of sickle cell trait based on parent report, and sentiments on playing sports with sickle cell trait were all influenced by age (all $p < 0.05$). Health beliefs such as having tattoos or piercings and getting annual check-ups with a primary care physician were also influenced by age (both $p < 0.02$). The most significant barrier identified was a lack of information about testing options from primary care physicians, while the largest motivating factor for testing was for personal health reasons. Findings from this study could aid genetic counselors with strategies to increase sickle cell trait testing in African-American men. Thereby, increasing awareness of sickle cell trait in the community for informative health and reproductive outlook.

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CHAPTER 1

INTRODUCTION

Sickle cell disease (SCD) is a genetic condition that was first included on newborn screening (NBS) in the 1970s and is currently included on NBS panels in all 50 United States (Pitt, 2010). A nationwide initiative for universal and population screening was implemented with the signing of the National SCD Control Act Law in 1972, which was predominately geared towards African-Americans. The goals of the law were to educate and prevent new occurrences of SCD within the population, given the increased carrier frequency of sickle cell trait (SCT) in African-Americans. The carrier frequency is 1 in 12 to 1 in 13 with the median life span for individuals with SCD being 42 in males and 48 in females (Housten, Abel, Lindsey, & King, 2016; Sickle Cell Disease: Data & Statistics, 2017; Platt et al., 1994).

Many felt anger and resentment towards the sickle cell screening initiatives, which birthed the long history of stigmatization of SCD within the African-American community. During this time-period, many African-Americans felt forced to undergo sickle cell screening for employment, health insurance, and marriage purposes, which ultimately lead to discrimination in each of those areas (Naik & Haywood, 2015). Although the laws were changed to assuage the situation, the damage had already been done (Markel, 1992). Presently, the health and social stigma of SCD may be lessened in the African-American female community due to routine testing for SCT in pregnant women (Long, Thomas, Grubs, Gettig, & Krishnamurti, 2011).

Research has shown that African-American women have a better understanding of the importance of genetic testing and counseling and are most often the participants in studies involving SCD and SCT (Gustafson, Gettig, Watt-Morse, & Krishnamurti, 2007). Participation from African-American men in these types of studies has been low, leaving researchers with only half of the complete picture on how to increase health literacy of SCD within the community (Boyd, Watkins, Price, Fleming, & DeBaun, 2005). Male participation in these studies has been described as "infrequent" and "difficult" with no literature explicitly stating why uptake, or incidence, of African-American males pursuing testing of SCT is low, making it hard for health care professionals, such as genetic counselors, to appropriately counsel at-risk couples when the male partner refuses to have testing for SCT (Gustafson et al., 2007). Emerging evidence shows there may be health related complications involved with SCT in cases of extreme heat, dehydration, and high altitudes. This signifies it may be of the upmost importance for individuals at risk for SCT to know their status for reasons other than reproductive decisions (Aloe, Krishnamurti, & Kladny, 2011).

The purpose of this study is to understand the contributing factors behind the low uptake and/or refusal of testing for SCT in African-American men. It is hypothesized that there is a significant difference in knowledge of SCT based on educational level, age, and health beliefs within the African-American male community, thereby perpetuating this reluctance to testing for SCT. There are several factors the present study will hopefully address to discern the barriers, or challenges, that exist when testing this population for SCT. This information will be useful for genetic counselors who discuss SCT with African-American men and women during the genetic counseling session.

CHAPTER 2

UNDERSTANDING BARRIERS TO GENETIC TESTING FOR SICKLE CELL TRAIT: THE AFRICAN-AMERICAN MALE PERSPECTIVE[1]

[1] Foster, S. D., Zvejnieks, D., Donald, Y., & Mazoue, C. To be submitted to the *Journal of Health Psychology*.

2.1 INTRODUCTION TO SICKLE CELL DISEASE

Sickle cell disease, SCD, is a collective term for a group of inherited single-gene disorders affecting hemoglobin, the oxygen-transporting molecule found in red blood cells. The condition results from a structural change in red blood cells that causes them to become hard and sickle shaped, which makes it difficult for hemoglobin to circulate oxygen to different tissues within the body (Jenerette & Brewer, 2010). SCD occurs when two pathogenic variants of the beta-hemoglobin gene are inherited, one from each parent. Individuals with sickle cell trait, SCT, have inherited a normal copy and a pathogenic variant of the beta-hemoglobin gene, making them carriers for SCD, one of the most common types of hemoglobinopathies (Bender & Douthitt, 2014).

SCD affects multiple body systems, with the main clinical manifestations resulting from the effects of deoxygenation, vaso-occlusion, and tissue necrosis (Jenerette & Brewer, 2010). In 1910, James B. Herrick published the first article on the disease where he observed the sickle shaped cells taken from a Grenadian dental student who displayed complications of what would become known as SCD. Several years after the initial discovery, many scientists sought out to conduct experiments to better understand the nature and etiology of the condition. By 1949, Dr. James V. Neel uncovered SCD was inherited through an autosomal recessive pattern (Frenette & Atweh, 2007). More recent studies have been geared towards understanding the pathophysiology of SCD and its prevalence in certain populations. The most commonly affected populations include individuals with sub-Saharan African, South American, Saudi Arabian, Indian, and Mediterranean origins (Gallo et al., 2010). Each SCD subtype is classified by its effect on beta-hemoglobin, with the most common form being sickle cell anemia (HbSS), which

accounts for 60-70% of all cases of SCD. Sickle cell anemia is followed in incidence by sickle-hemoglobin C (HbSC), sickle cell beta-thalassemia (HbSB+ and HbSB^{o}), and more rare forms that result from an interaction between HbS with HbD-Punjab, Hb O-Arab, and Hb E, and Hb Lepore (Serjeant, 2013; Bender & Douthitt, 2014).

2.2 CLINICAL FEATURES

With the inclusion of SCD on the mandatory newborn screening panels in all 50 United States, most babies with SCD are detected 1-2 weeks after birth (Gallo et al., 2010). Typically, symptoms appear before the first year of life, making it imperative for children with SCD to be identified, monitored, and treated before symptoms can manifest (Jenerette & Brewer, 2010). Those symptoms are characterized by "vaso-occlusive events and chronic hemolytic anemia [that] can affect multiple organs in the body including: bones, liver, kidneys, brain, eyes, and joints" (Bender & Douthitt, 2014, p.1). The first recognized clinical manifestation in infants and young children is dactylitis, which is pain or swelling of the hands and feet. Often splenomegaly occurs in younger patients due to sequestration of blood cells within the spleen, which increases the risk of bacterial infections. If two or more of these splenic attacks occur before age 2, a splenectomy is ideal followed by life-long treatment with penicillin as a prophylactic (Serjeant, 2013). Depending on the severity of chronic hemolysis, it can cause a multitude of symptoms including: anemia, jaundice, delayed growth and/or sexual maturation, and cholelithiasis. The more severe spectrum of hemolysis can result in pulmonary artery hypertension, priapism, leg ulcers, septicemia, rhabdomyolysis, and even death (Bender & Douthitt, 2014). Sickle cell anemia, HbSS, and beta-thalassemia, HbSB^{o}, are the more clinically

severe phenotypes where the onset of anemia and hemolysis develop by 6–12 months of age (Bender & Douthitt, 2014).

Since SCD was first described over 100 year ago, morbidity and mortality remain high with a median survival range estimated to be 42 years in men and 48 years in women; however, more recent articles have quoted the median age as 45–55 years old (Platt et al., 1994; Serjeant, 2013). There are many clinics and organizations geared towards caring for individuals with SCD to help properly manage the effects of the condition as they age. In the early childhood years, acute chest syndrome is the most common cause of death in children with SCD after the age of 2 with symptoms characterized by pulmonary chest pain, cough, dyspnea, and pleuritic pain of avascular necrosis (Serjeant, 2013). There is also an increased risk of hemorrhagic strokes that occur at the peak ages of 6 and 25 years old, which are associated with a poor prognosis. For adults with SCD, acute chest syndrome continues to have an effect, coupled with a risk of congestive heart failure and deteriorating renal function, possibly due to the decline in total hemoglobin after the age of 40 years old (Serjeant, 2013).

2.3 ETIOLOGY

SCD is caused by pathogenic variants in the *HBB*, hemoglobin subunit beta, gene which produces a protein called beta-globin that makes up two out of the four protein subunits of the larger adult hemoglobin protein located in red blood cells. Mutations in the *HBB* gene cause abnormal versions of beta-globin to be produced and distorts the red blood cells into sickled shaped cells that die prematurely and can lead to anemia (Bender & Douthitt, 2014). The Glu6Val pathogenic variant is the most well-known of the point mutations that leads to SCD. These cells are often hard and inflexible, making them more

susceptible to getting stuck in the small blood vessels in the body, leading to many of the disease's clinical manifestations. The beta-like globin genes are located on chromosome 11 and are expressed in the order in which they develop. During fetal development, hemoglobin F and gamma (Y) are expressed and after birth, the infant's hemoglobin makes the transition to hemoglobin alpha and hemoglobin beta. During this switch from fetal to adult hemoglobin, these disorders of the beta hemoglobin genes will begin to manifest themselves, explaining why newborns with SCD do not have immediate complications after birth (Frenette & Atweh, 2007).

2.4 INCIDENCE

Per the Center for Disease Control, SCD affects 100,000 Americans with an occurrence of 1 out of every 365 African-American children having the condition. HbSS, or sickle cell anemia, is the most common form of SCD and most often affects those of African descent. Approximately 1 in 12 to 1 in 13 African-Americans are carriers of an S beta-globin mutation (Jenerette & Brewer, 2010; Housten et al., 2016; Sickle Cell Disease: Data & Statistics, 2017; Boyd et al., 2005). Given the autosomal recessive inheritance of this condition, we can accurately predict the chance of an affected pregnancy in individuals who are known heterozygotes of an *HBB* pathogenic variant. Using the example of parents with the HbAS genotype, we know that each pregnancy has a 25% chance of having sickle cell anemia, a 50% chance of being a carrier for S beta-globin, and a 25% chance of having normal hemoglobin.

The prevalence of SCT has been well studied and may have developed as a part of a heterozygote advantage in Africa as a protective mechanism against malaria. Malaria, which is caused by the bacterium *plasmodium falciparum*, is an infectious disease that is

common in African countries. Researchers believe that heterozygotes for the sickle gene (HbAS) are protected against the harmful effects of malaria. The proposed theory is that AS heterozygotes have less red blood cells that are parasitized in their blood, meaning they are less likely to develop the more life-threatening forms of malaria, cerebral malaria, and malaria with severe anemia. AS heterozygotes often survive malarial infections while those with sickle cell anemia, HbSS, do not and are more susceptible to the lethal effects of malaria due to hyposplenism, which causes a lower clearance of infected red blood cells (Luzzatto, 2012; Grosse et al., 2011). A study by Elion et al. in 1992, stated that sickle hemoglobin, HbS, has undergone evolutionary selection at least 5 times because of its malarial protective effects (Naik & Haywood, 2015) (Figure 2.1).

2.5 NEWBORN SCREENING

Understanding more about the severity and early onset of SCD led to its inclusion onto many of the newborn screening (NBS) panels in the United States. SCD was first added to newborn screening panels in the US around the 1970s (Pitt, 2010; Benson & Therrell, 2010). The most common screening techniques include: sickle solubility testing, hemoglobin electrophoresis, high performance liquid chromatography, and isoelectric, focusing (Naik & Haywood, 2015). The follow-up and communication of NBS results for SCT and SCD vary across state, which has caused some disconnect in the disclosure of positive results to families. A study done in 2010 showed that sickle cell “stakeholders”, such as hematologists, hospitals, and families are not being properly informed of the results. Specifically, for SCT, the study reported that an average of only 37% of families were notified about positive results. This study highlighted a potential need to make

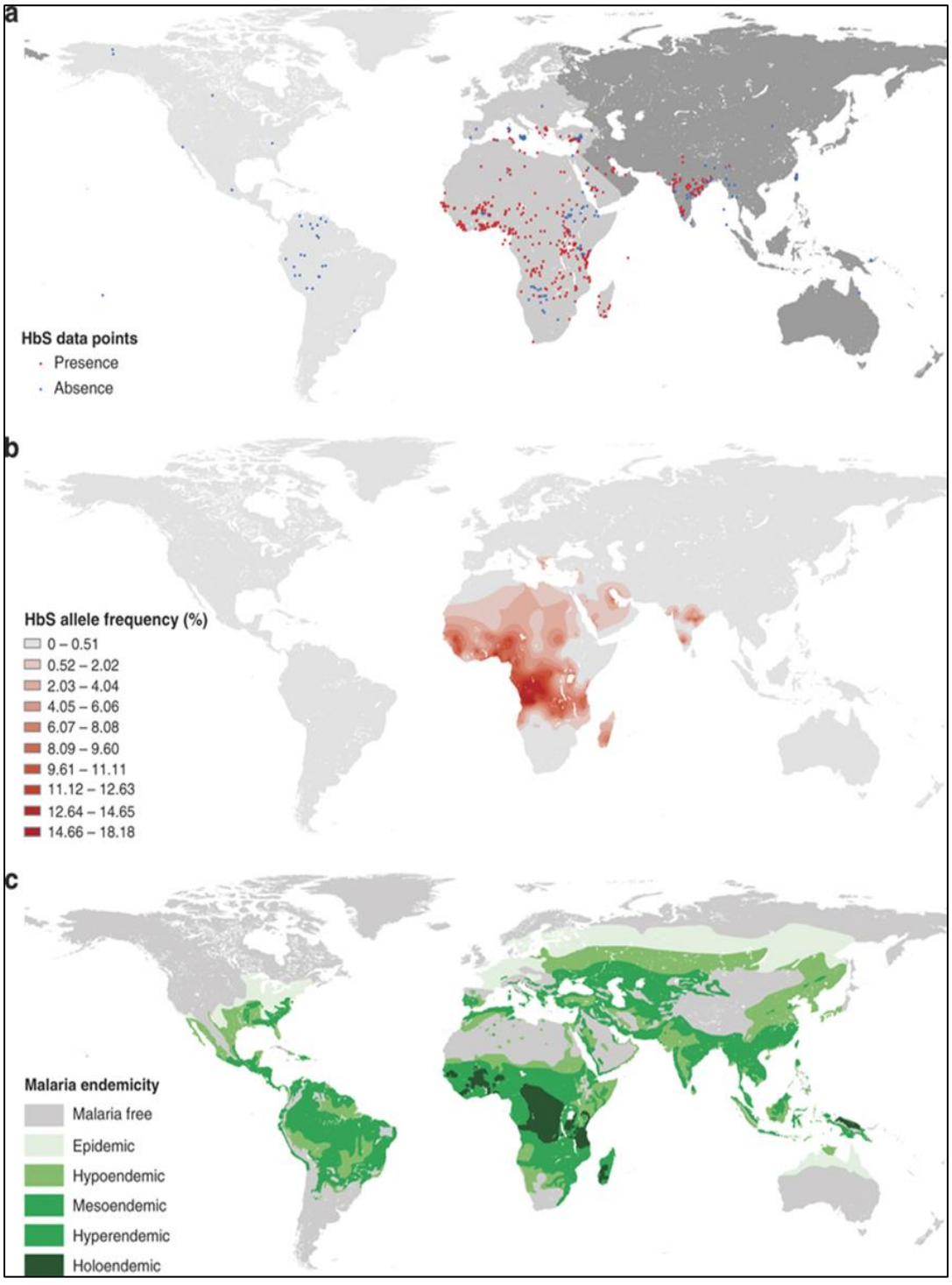


Figure 2.1 Depiction of Malaria vs. HbS Correlation

changes within the follow-up and communication of NBS results (Kavanagh, Wang, Therrell, Sprinz, & Bauchner, 2008).

2.6 POPULATION SCREENING

Prior to the late 1960s, there were hardly any established programs bringing awareness of SCD to African-American communities. In 1972, the National SCD Control Act was signed into law by President Richard Nixon to start a nationwide initiative for universal newborn screening, population screening, and counseling by providing more education and genetic counseling to patients with SCD and SCT. For most of the 1970s, this law focused on identifying at-risk individuals, research, and increasing community awareness and education (Naik & Haywood, 2015).

Ultimately, population screening became a normalized part of healthcare because many healthcare professionals began to recommend testing for SCT when status was unknown (Boyd et al., 2005). In 1972, The Department of Defense developed guidelines for mandatory and universal SCT screening for all Army, Air Force, and Navy recruits after adverse effects in individuals with SCT were observed. During that time, the adverse effects observed included sudden death and exertional rhabdomyolysis at high altitudes. Without enough evidence to support claims of the adverse effects, The Department of Defense decided to withdraw their mandatory requirement in 1985. Currently, individuals in the Navy, Air Force, and Marines are still subject to universal SCT screening; however, the Army discontinued this health measure in 1996. The same mandates were made by the National Collegiate Athletic Association, NCAA, in the late 1970s before being thrown out and reinstated in 2010 with an opt-out option (Naik & Haywood, 2015).

Although many saw the benefit of learning their SCT status, it did not take long for many to associate SCD with the black community, despite its occurrence in other diverse populations. The stigmatization of SCT carriers occurred with the national population screening efforts put forth by the National SCD Control Act in 1972. When President Nixon spoke about the initiatives, he used language that may have propagated ideals that negatively associated African-American individuals with SCD. In his speech, he stated: "This disease is especially pernicious because it strikes only blacks and no one else… these actions make it clear, I believe, the urgency with which this country is working to alleviate and arrest the suffering from this disease." (Naik & Haywood, 2015, p. 5). Many African-American men and women began to feel forced into undergoing testing for several different reasons including employment, health insurance, and even for marriage purposes. It ultimately led to discrimination in each of those areas (Naik & Haywood, 2015). The "fear" of SCD was cultivated mainly because of the eugenics movement during the advent of population screening and contributed to individuals being denied life insurance and barred from entering the U.S. Air Force Academy. It caused much anger within the community, further stigmatizing carriers of sickle cell. Although laws were changed to assuage the situation, the damage was already done (Markel, 1992).

In more recent years, one study has shown that perceived discrimination in SCD happens in all aspects of society and in healthcare professions. Most of the discrimination that has been documented is disease-based and not race-based and is associated with the presenting severity of the condition (Haywood et al., 2014). In a study by Lawrence et al. in 2014, they sought out to understand athletes' perceptions of SCT screening and examined the possibilities for discrimination. Most athletes believed being

a known SCT carrier could lead to potential discrimination, resulting in a reduction in playing time, current or future athletic opportunities, and a denial of health insurance (Haywood et al., 2014). Perhaps, the reason sickle cell stigmatization is so strong within the community stems from some of the outward physical effects of SCD like jaundiced eyes and leg ulcers (Bediako et al., 2016). Some fear the prospects of SCD and are uncomfortable with talking about it, which could contribute to at-risk individuals not wanting to know their status (Gallo et al., 2010).

2.7 SIGNIFICANCE OF THE STUDY

The health-related stigma attached to SCD is still relevant. Those with the condition are still looked upon as weaker and are discriminated against by employers and family members (Jenerette & Brewer., 2010). However, routine testing for SCT in pregnant women may have lessened the stigma in recent years, specifically in African-American women. Many studies on the subject have confirmed that although the barriers of knowledge about sickle cell testing and perceived discrimination are still present, African-American women understand the importance of genetic testing to help them determine their trait status (Long et al., 2011). In a study performed 10 years ago, the participants, who were all African-American women, discussed their health beliefs regarding genetic testing and counseling for SCD. While most of the participants gained a better understanding of the severity and benefits to screening, many still did not believe they were at risk for having a child with SCD even when their status was unknown (Gustafson et al., 2007). Although multiple efforts have been made to increase awareness and education on SCT and SCD, there is still a low uptake or acceptance for screening in the African-American population. It appears a lack of education and stigma are still

driving forces behind the low uptake given that a study in 2005 showed that overall, African- American women are poorly informed on the genetics, inheritance, and variability of SCD. The data from this study was comparable to those conducted in the late 1960s (Boyd et al., 2005).

Health literacy within the African-American community regarding SCT or SCD is still a common issue. One interesting study in 2014, set out to examine the accuracy of self-reporting in SCD juxtaposed to genetic confirmation. Their research showed that of the fifty-one individuals who reported they had SCD, only 5.9% were confirmed to have SCD, 67.2% were confirmed with SCT, 5.9% with hemoglobin C trait, and 25.5% with having normal hemoglobin. Oppositely, of the 75% of individuals who reported not having SCD, 100% were concordant with their self-report (Bean et al., 2014). These results are most striking as they support the conclusions of many other articles that highlight improving education of adults about their status is paramount, given the marked differences in the background knowledge of SCD and SCT. Bean et al. (2014) argued that education should be done within the public health or primary care setting since those healthcare professionals have more opportunities to educate the patient.

Gustafson et al.'s study showed one interesting detail, which is the current basis for the present study, where they identified that one barrier for the female participants was convincing their male partner to get screening for SCT (Gustafson et al., 2007). Of the multiple studies that have been published in the past, most have only been able to capture one half of the story since African-American male participation has been low for both undergoing testing and participating in studies involved with SCT testing. Currently, there is no literature explicitly stating the reasons why uptake continues to be low in

African-American men (Housten et al., 2016). Whenever male participation occurs, it is often described as "infrequent" and the lack of participation makes it difficult for health care professionals, such as genetic counselors, to understand the barriers in testing for SCT. The only reassuring way to decrease a mother's worry about the risk to their unborn child for SCD is to test the father (Gustafson et al., 2007). Anecdotally, some genetic counselors have noticed the reluctance of African-American men to have genetic testing as well. Using the example of African-American participation in clinical trials, many of the same reasons could be extrapolated to testing for SCT. In this study, many of the men cited their reluctance to participating stems from the past exploitation and treatment of those in the Tuskegee syphilis experiment, which may also contribute to the mistrust that many people of color have with health care professionals and to their refusal of testing for SCT (Jacobs, Rolle, Ferrans, Whitaker, & Warnecke, 2006; BeLue, Taylor-Richardson, Lin, Rivera, & Grandison, 2006).

Addressing this issue has become more relevant since the NCAA (National Collegiate Athletic Association) established their genetic screening program that included mandatory SCT screening as a part of the medical exam for all its division 1, 2, and 3 players in 2010. The decision was made after a string of African-American students died from complications of SCT in 2009. Studies were conducted that confirmed dehydration, high altitudes, low oxygen, and increased blood viscosity were some of the factors that could lead to sudden deaths in athletes with SCT (Aloe et al., 2011).

Researchers have observed an increase in sickled cells, or exertional sickling, in individuals with SCT when exposed to extreme physical activity. Exertional sickling refers to an increase in sickled cells with exertion or increased exercise, which could

potentially lead to blockages of red blood cells in the blood vessels and sudden death (Ferrari, Parker, Grubs, & Krishnamurti, 2015). The NCAA guidelines explained that students should be screened for SCT so that students and coaches are more aware of their status in case extra precautions should be taken to avoid health-related problems (Harris, Haas, Eichner, & Maron, 2012). They explained that coaches should not train students with SCT as hard; however, the Army stated that treating everyone the same would be more beneficial to avoid ostracizing those with SCT. For example, the Army recommended that everyone, not just those with SCT, should be kept cool and hydrated to avoid health issues on especially hot days (Tarini, Brooks, & Bundy, 2012).

The recent NCAA policy change on SCT has not gone without backlash from other organizations because of the ethical implications of testing student athletes. Ferrari et al. examined some of the implications of this type of health reform and stated they oppose the mandatory screening rule because organizations such as the American Society of Hematology and the Sickle Cell Disease Associations of America believe testing all athletes for SCT is unwarranted because the link to sudden death has not been confirmed. The recommendations from the article state that including genetic counseling as a part of the screening program without testing would insure student athletes are being properly educated on the natural history of SCT, its reproductive implications, along with potential health and psychosocial concerns (Ferrari et al., 2015).

Although there are laws preventing discrimination against someone because of their genetic make-up, the risk of discrimination, perceived or real, is a palpable fear in the African-American community, especially for men, whenever there is disclosure of SCT status within the military, job force, and recently in sports (Ferrari e al, 2015). Since

most individuals with SCT 1) do not experience any complications and 2) are not aware of their status and 3) are reluctant in wanting to know their status, it has become even more important for African-American men and women to know their trait status, not just for the risk of SCD in potential pregnancies but also for the potential risk it poses to their health (Harris et al., 2012). Apart from sudden cardiac death, ongoing research shows SCT may be associated with hematuria, glaucoma-post hyphema, renal disease, and splenic ischemia (Westerman et al., 2002; Kiryluk, Jadoon, Gupta, & Radhakrishnan, 2007; Harris et al., 2012; Pandey, 2015; Naik et al., 2016). The NCAA ruling has made genetic counselors more aware of some of these issues testing for SCT can pose for athletes but has not presently been studied in African-American men who are non-athletes (Aloe et al., 2011).

2.8 HYPOTHESIS

Therefore, the questions proposed in this study revolve around uncovering if African-American men more likely not to be tested for SCT and understanding the reasons why. It is hypothesized that there is a significant difference in knowledge of SCT based on educational level, age, and health beliefs within the African-American male community, thereby perpetuating this reluctance to testing for SCT. There are several areas the present study will hopefully address to discern which barriers or challenges, besides education, exist when testing for SCT. Several examples include stigma, perceived discrimination, and a fear of needles. Grosse et al., who conducted a study on African-American women, also pointed out that there is a lot of uncertainty about male partner's trait status because of a decreased risk perception along with a lack of understanding about the inheritance and natural history of SCD and SCT. The study also

cited a fear of needles as another interesting but rarely talked about barrier in the literature. This fear stems from the pain often associated with phlebotomy, which is potentially strong enough for some men to refuse testing (Gallo et al., 2010; Gustafson et al., 2007). A similar study looked at the health beliefs, barriers, and motivations that exist within African-American women, but not in men (Gustafson et al., 2007).

The current aims of this study include:

I. Assessing African-American men's knowledge and perceptions on genetic testing, risk, and health within the context of SCT
II. Understanding barriers and motivating factors in testing for SCT
III. Recognizing the practice implications for genetic counselors

Understanding these factors in African-American men, helps to gather the full picture of why this issue exists within genetic counseling and how to address these concerns appropriately. From a health literacy standpoint, it is most important to learn more about what African-American men already know and understand their perspectives on genetic testing for SCT because it is an important health issue within the community that is not discussed.

For genetic counselors, this information is important to know because it will increase awareness amongst this population that is most at risk for SCT and SCD and help to fill in gaps surrounding why this issue exists and what we as genetic counselors can do about it. Knowing more about the barriers, health beliefs, and motivating factors will assist genetic counselors in addressing some of these issues during counseling sessions. It could open a dialogue between African-American families and their genetic counselors so that they can reduce myths surrounding SCT and SCD and increase

community awareness of the potential health risks associated with SCT. Gaining the African-American male perspective will be one of the first steps in figuring out what genetic counselors can do to improve uptake of men pursuing testing so that we can give accurate risk assessment to expecting couples and reduce the anxiety that is commonly seen when there is a risk of SCD in a pregnancy. More importantly, genetic counselors could be on the forefront of informing patients about the potential risks associated with SCT for their own health.

CHAPTER 3

MATERIALS AND METHODS

3.1 POPULATION

African-American, or black, males over the age of 18 were invited to participate in the study, with the target population being 18 to 45, the most common childbearing ages. Most participants that were invited to take the survey were physically located in the greater Columbia, South Carolina area. All females and non-African American, or black male participants were excluded from participating in the study. There were no exclusions based on age, religious affiliation, educational background, or socioeconomic status.

3.2 AREAS OF RECRUITMENT

The participants were recruited from many areas within Columbia, South Carolina. The following places were provided paper surveys: James R. Clark Memorial Sickle Cell Foundation events (i.e. 12th annual SCD walk, Sickle Cell Family Day, The Giving Tree), a Friendship Baptist Church homeless group meeting, University Specialty Clinics Genetic Counseling suite at Palmetto Health-Richland Hospital System, and an African-American Shriner's group meeting. For the online survey, participants were recruited through postings of an online link to the survey from the primary investigator's Facebook page and the University of South Carolina's Facebook pages.

3.3 SURVEY MATERIALS

The protocol for this study was approved by the University of South Carolina Institutional Review Board under the title of "Understanding Barriers to Genetic Testing for SCT: The African-American Male Perspective". In the appendix, a copy of the Institutional Review Board's approval letter for protocol Pro00067778 can be found. The survey was initially created in a printed version, with an online copy being created on surveymonkey.com. For the online survey, two additional exclusion criteria questions based on gender and ethnicity were added to vet out participants who would otherwise not meet the in-person participant criteria (i.e. non-black and female individuals). They are listed below:

1. What is your gender?

a) male b) female

2. What is your ethnicity/race?

a) African-American/Black b) European-American/White c) Latino/Hispanic

All participants were instructed to read the letter of participation before continuing to the survey and were told their contribution would be anonymous, confidential, and optional.

3.4 SURVEY METHODS

Using survey methodology, a questionnaire containing themes about health literacy regarding SCD and SCT, perspectives on genetic testing, health perceptions, barriers, and motivations was formulated and distributed to all willing participants beginning on September 9, 2017. The questionnaire consisted of 31 questions with five additional demographic questions. A copy of the survey is provided in Appendix D. All participants were invited to answer Questions 1- 20 of the survey. Questions 1-9 asked

knowledge based questions regarding SCT and SCD and consisted of multiple choice and true/false answer. Questions 6-7 were utilized from a previous survey constructed for a similar study by Gustafson et al. (2007). Questions 10-18 were used to assess risk and health perception using a Likert scale. Participants were asked to rate their level of agreement with each statement (1= strongly disagree, 5= strongly agree).

Participants were then asked about their perceptions on genetic testing in questions 19-20 using yes/no answer. Only participants who answered yes to question 20 were invited to complete questions 21-24 which asked participants questions about the nature in which they underwent genetic testing for SCT. Question 21 was multiple choice while questions 22-24 were yes/no answer. All participants were invited to answer questions 25-31. Questions 25 -29 were used to assess health beliefs using yes/no answer. Questions 30 and 31 asked participants about barriers and motivating factors to genetic testing for SCT respectively. Both questions were multiple answer. Questions 32-36 were demographic, multiple-choice, questions that asked participants about their age, educational level, yearly income, relationship status, and number of children, respectively.

An incentive was offered in the form of an in-person or online drawing for gift cards ranging from $5 to $10 from various chain restaurants (i.e. McDonald's, Chic Fil-A). No more than 7 gift cards per month were given. Each participant was entered into the drawing within the same month the survey was completed in. The names and/or phone numbers were taken for the drawing and winners were either physically present to receive their gift card or they were mailed to them. All identifying information collected from the drawings was destroyed once the winners were contacted and their gift cards

were mailed. The participants were all told this information before completing the survey to establish trust and confidentiality.

3.5 DATA ANALYSIS

For the knowledge based questions detonated as the “sickle cell quiz”, answers were split into two variables with correct answers coded as 1 and incorrect answers coded as 0. If a respondent did not answer one of the knowledge based questions, their answer was coded as incorrect. There was a maximum score of 9 possible for knowledge of SCD and SCT. The grand mean knowledge scores were calculated based on group and educational level. Using SPSS Statistical Software, two-way, one-way repeated, and one-way ANOVAs was used to assess statistical differences in age, educational level, knowledge, health beliefs, and perceived risk and discrimination. Statistical differences in education level and knowledge were analyzed using an independent samples t-test.

CHAPTER 4

RESULTS

4.1 DEMOGRAPHIC INFORMATION

One hundred and sixteen of the 124 surveys collected were used in the data analysis because they were 80% or more completed. Of the participants that answered the demographic questions, all identified as black and/ or African-American (100%), were primarily between the ages of 31 to 49 (41.0%), with some college education (31.9%), married (36.2%), with no children (34.5%), and making less than $10,000 dollars a year (23.3%) (Table 4.1).

4.2 SPECIFIC AIM I: ASSESSING AFRICAN AMERICAN MEN'S KNOWLEDGE AND PERCEPTIONS ON GENETIC TESTING, RISK, AND HEALTH WITHIN THE CONTEXT OF SCT

Overall, approximately 41% of participants reported getting tested for SCT within their lifetime with the other 59% reported never being tested for SCT (Figure 4.1). Twenty-one percent reported seeing a genetic counselor for SCT testing with only 2% having refused testing within their lifetime (Figure 4.2 & Figure 4.3).

Of the 49 (42.2%) people that had SCT testing, 13 (11.2%) participants were tested at birth, which made up the largest category. Nine participants (7.8%) reported being tested in the military, and another 9 (7.8%) reported getting tested for an unknown reason that was not listed within the survey choices. The third largest category, made up

Table 4.1 Survey Demographics

Variable	**Population (n= 116)**	
	Number	**Percentage**
Age Intervals		
18-19 y	4	3.5
20-24 y	12	10.5
25-30 y	17	14.9
31-36 y	13	11.4
37-42 y	11	9.6
43-49 y	23	20.2
50+ y	34	29.8
Education Level		
Some high school	8	7.2
High school graduate	27	24.3
Some college	37	33.3
College graduate	23	20.7
Graduate/professional school	16	14.4
Yearly Income		
less than $10,000	27	25.5
$10,000- $20,000	9	8.5
$20,000- $30,000	16	15.1
$30,000- $40,000	10	9.4
$40,000- $50,000	13	12.3
$50,000- $100,000	23	21.7
$100,000 +	8	7.5
Relationship Status		
Married	42	38.9
Single	41	38
In a relationship	25	23.1
Number of Children		
0	40	36.7
1	13	11.9
2	25	22.9
3	14	12.8
4	8	7.3
5 or more	9	8.3

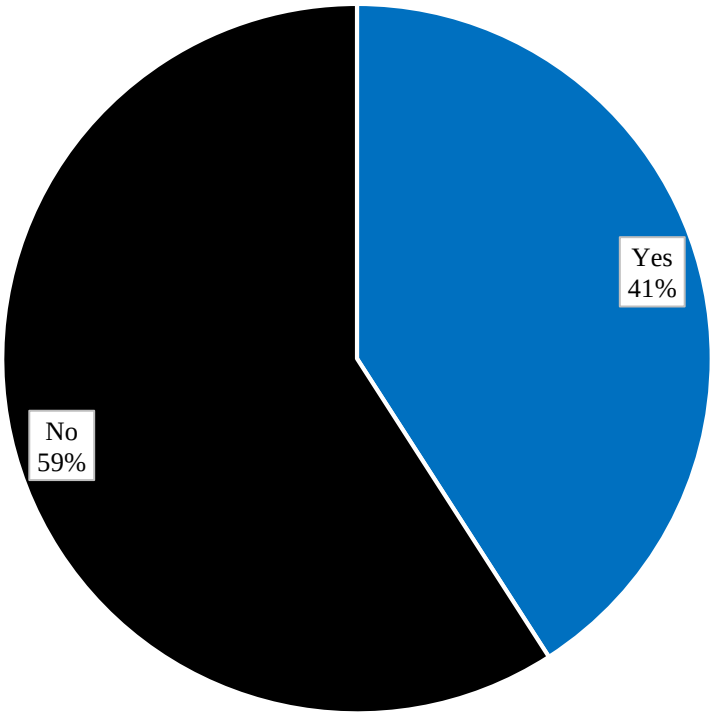


Figure 4.1 Participant Report of Testing for SCT

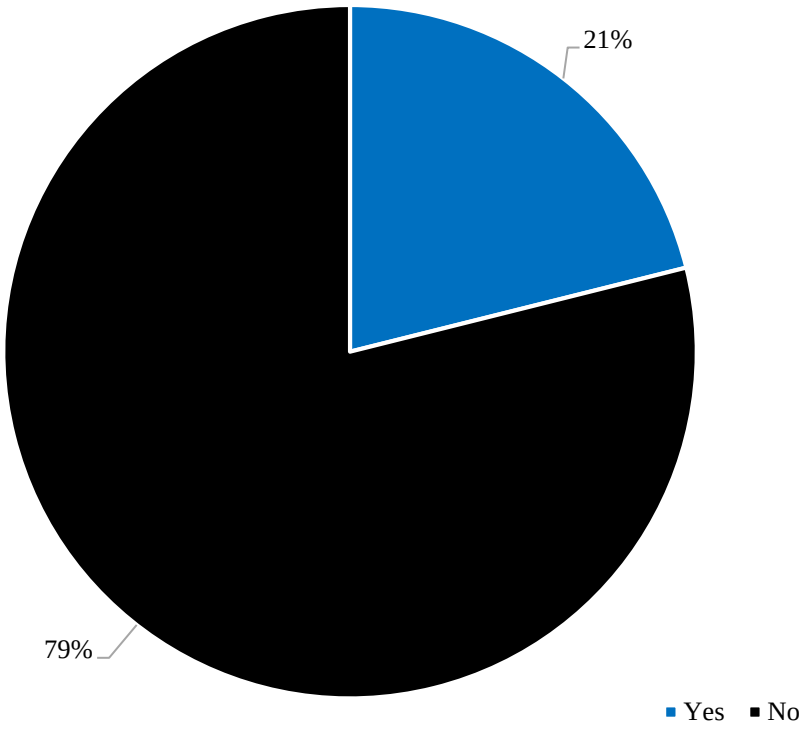


Figure 4.2 Participant Report of Seeing a Genetic Counselor for Sickle Cell Trait Testing

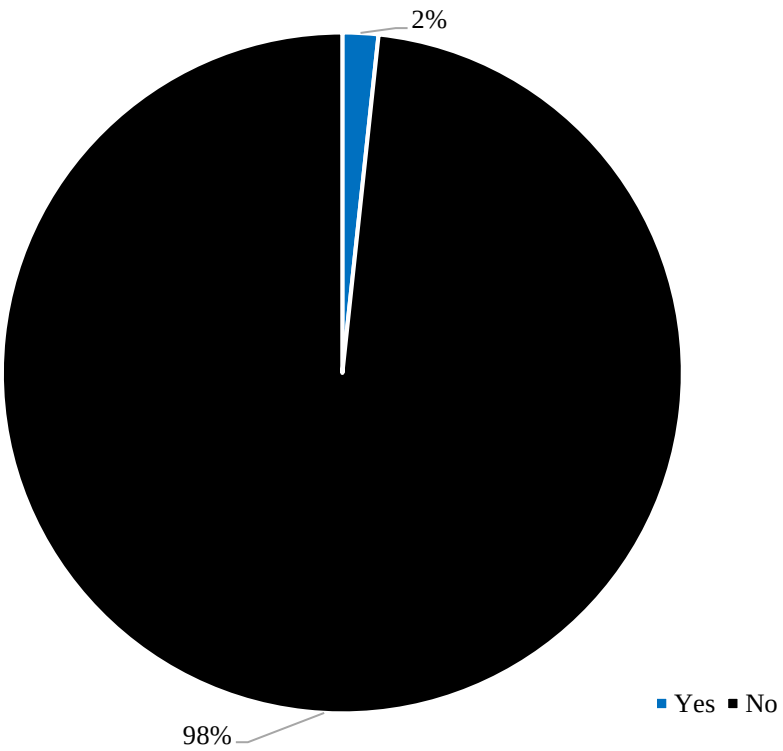


Figure 4.3 Participant Response to Refusal of Testing for Sickle Cell Trait

of 8 (6.9%) participants, reported getting testing for personal health reasons. The fourth largest category of participants stated they were tested for SCT for sports (n=6, 5.2%). The smallest category, made up of 4 (3.4%) participants, reported getting tested for SCT during their partner's pregnancy (Figure 4.4).

When asked if the participant had documentation stating their SCT status, 20 (46.5%) participants had documentation while the other 23 (53.4%) respondents did not (n=43). Thirty-two (76.2%) respondents were told they were negative for SCT, while 10 (23.8%) were told that they did have SCT (n=42). When asked if partners would be aware of their positive SCT status, 33 (80.5%) participants agreed their partner would be aware while 8 (19.5%) participants said their partners would not be aware of their positive SCT status (n=41, Figure 4.5).

On the sickle cell quiz, the grand mean knowledge score was 6.206 (69.55% correct) with a maximum of 9 correct answers possible. The 18 -19 age group scored the highest with a mean knowledge score of 7, while the 50+ age group scored the lowest with a mean knowledge score of 5.53 (Figure 4.6).Of the knowledge based questions, participants were most likely to guess incorrectly on the carrier frequency of SCT in African-Americans (31% correct). Participants were most likely to guess correctly that black people are predominantly affected by SCD (92.2% correct). Using descriptive statistics, the percent correct for each question on the sickle cell quiz were calculated and the correct answers were compiled into a table (Table 4.2).

A two-way ANOVA was conducted to look for any statistically significant differences between age and knowledge based on the total answers correct on the sickle cell quiz. Results showed there was a statistically significant negative association

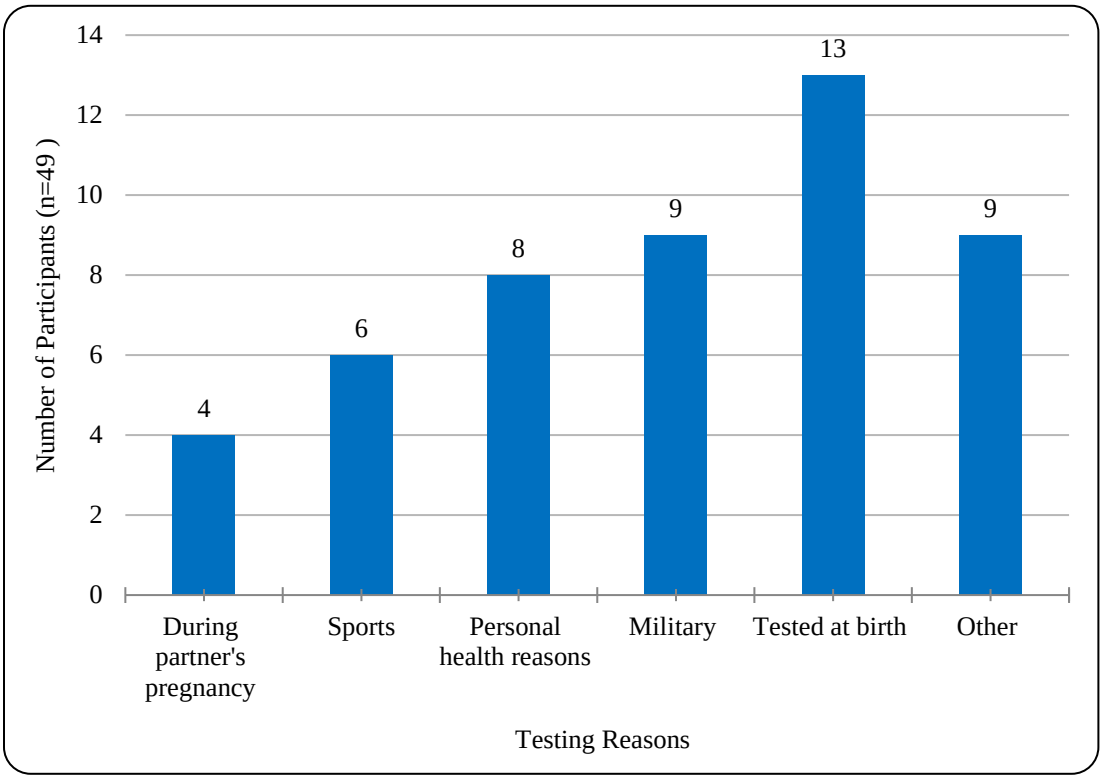


Figure 4.4 Participant's Selected Reasons for Being Tested for Sickle Cell Trait

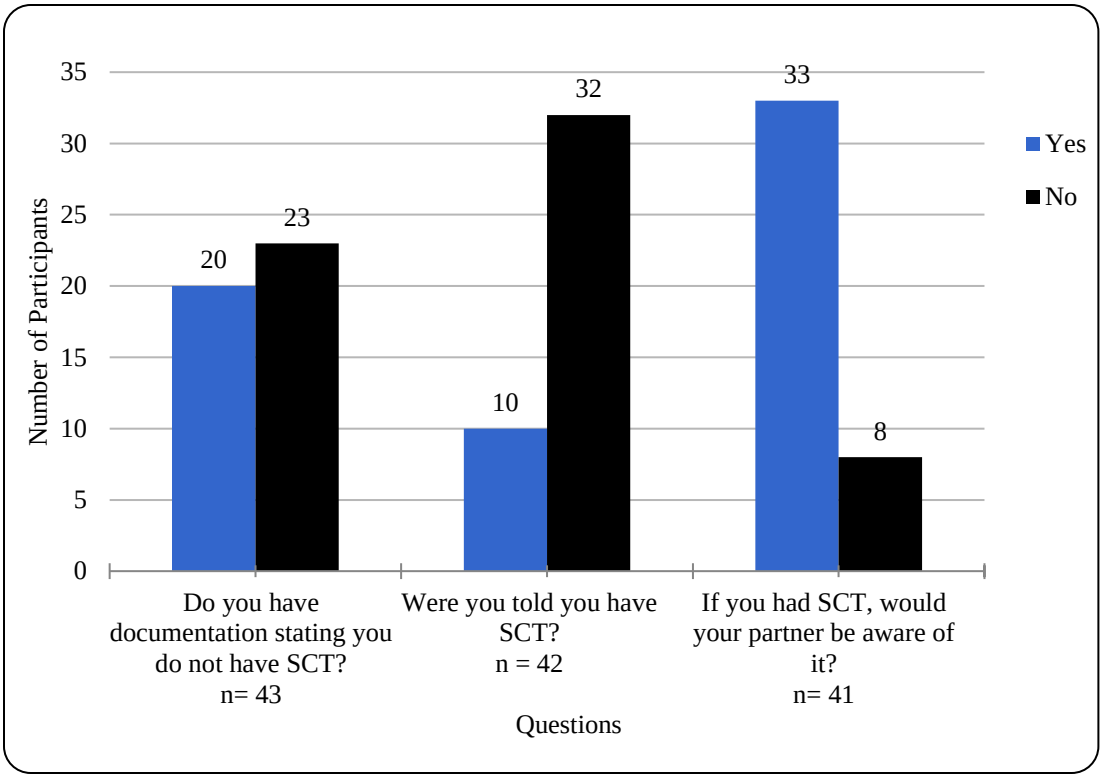


Figure 4.5 Participant Responses to Sickle Cell Trait Documentation, Status, and Partner Awareness

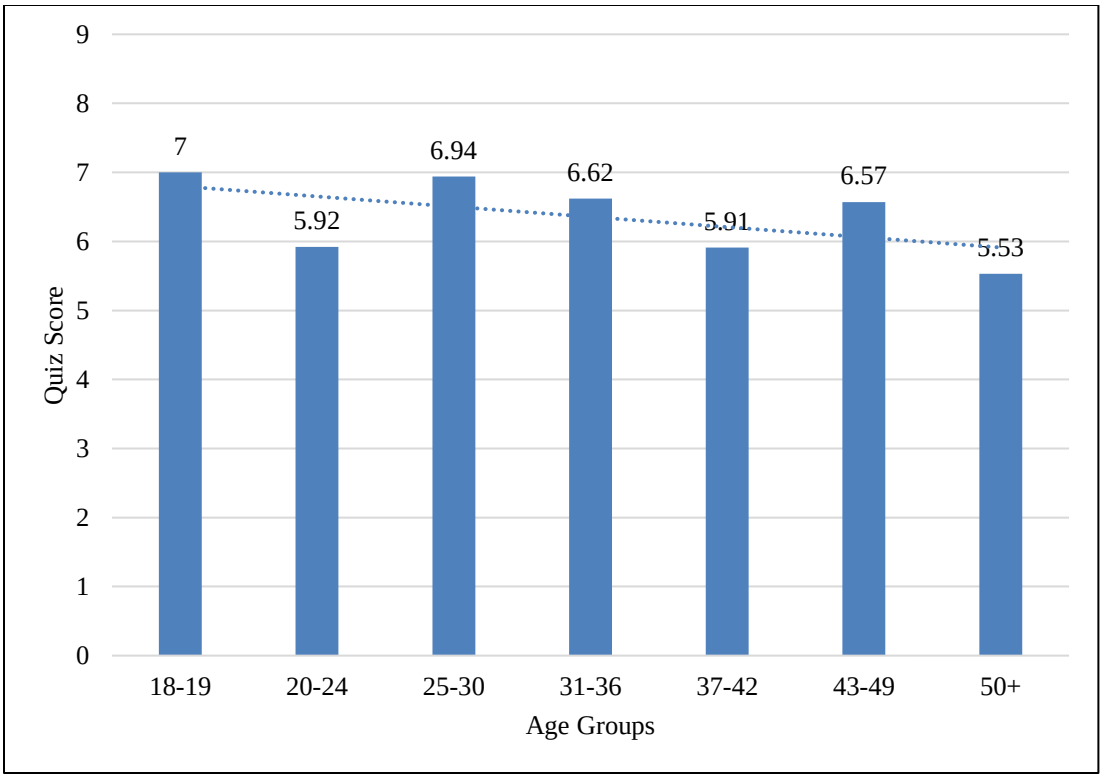


Figure 4.6 Mean Score of Total Correct on Sickle Cell Quiz Based on Age Group

Table 4.2 Chart of Knowledge Based Questions with Percent Correct Frequencies

Knowledge Based Questions:	**Answer:**	**Population (n=116) % Correct (Frequency)**
Sickle cell disease (SCD) is caused by...	b) 2 altered genes passed from parents	56% (65)
Sickle cell trait (SCT) is caused by...	c) 1 altered gene passed from the parent	64.7% (75)
Individuals with sickle cell disease...	d) all of the above	78.% (91)
Sickle cell disease most often occurs in...	c) black people	92.2% (107)
One out of every _____ African-Americans has sickle cell trait.	a) 1 out of every 12 people	31% (36)
Sickle cell disease makes red blood cells...	d) hard and sickle shaped	50% (58)
How can you tell if someone carries the gene for sickle cell trait or sickle cell disease?	c) with a blood test	81% (94)
Individuals with sickle cell trait can have symptoms under extreme conditions, such as when dehydrated or in low oxygen environments.	a) True	76.7% (89)
Sickle cell trait and sickle cell disease can be cured?	b) False	87.1% (101)

between age and knowledge of SCD and SCT, $F (6, 107) = 3.007$, $p = .009$, partial $\eta 2 = .144$ (Table B.1). To strengthen significance values, all other results were tabulated with the following adjusted age groups: 18-30, 31-49, and 50+ age groups. There was a statistically significant positive association between the newly defined age groups and the knowledge score on the sickle cell quiz, $F (2, 111) = 5.739$, $p = .004$, partial $\eta 2 = .094$ (Table B.2).

A two-way ANOVA was conducted between education level and the correct score on the sickle cell quiz. There was no statistically significant association between education and knowledge of SCD and SCT, $F (4, 106) = 1.611$, $p = .177$, partial $\eta 2 = .057$ (Table B.3). A one-way repeated measures ANOVA was conducted to determine whether there were statistically significant differences in the total correct on the sickle cell quiz based on education level. Analysis showed there were no statistically significant

differences in the mean scores on the sickle cell quiz when compared to education levels ($p > 0.05$) (Table B.4). An independent-samples t-test was run to determine if there were differences in the total correct on the sickle cell quiz between participants who had an educational level of high school or below and college level or higher. For the scores on the sickle cell quiz for each educational category, there was homogeneity of variances, as assessed by Levene's test for equality of variances ($p = .877$). There was no statistically significant difference between the two educational categories for the total correct on the sickle cell quiz (95% CI, -1.1 to .02), $t(109) = -1.899$, $p=0.60$ (Table B.5).

To assess risk and health perception of the study participants, several statistical tests were run to look for significant differences within the data. A one-way ANOVA was conducted to determine whether there were statistically significant differences in health and risk perception questions regarding SCT status and age groups. There was a statistically significant difference between the age group's answers on questions regarding perceived discrimination ($p = 0.040$), perceived risk based on parent report of SCT ($p= 0.008$), and playing sports ($p=0.016$). All other questions were not statistically significant between the age groups ($p>0.05$) (Table B.7).

Two figures were constructed with the first displaying the risk and health perception questions using box-and-whisker plot for each of the three age groups, and the second figure displaying the statistically significant questions along with their p-values (Figure 4.7, Figure 4.8, Figure 4.9, & Figure 4.10). A one-way ANOVA was conducted to determine whether there were statistically significant differences in health and risk perception questions and educational level. There were no statistically significant

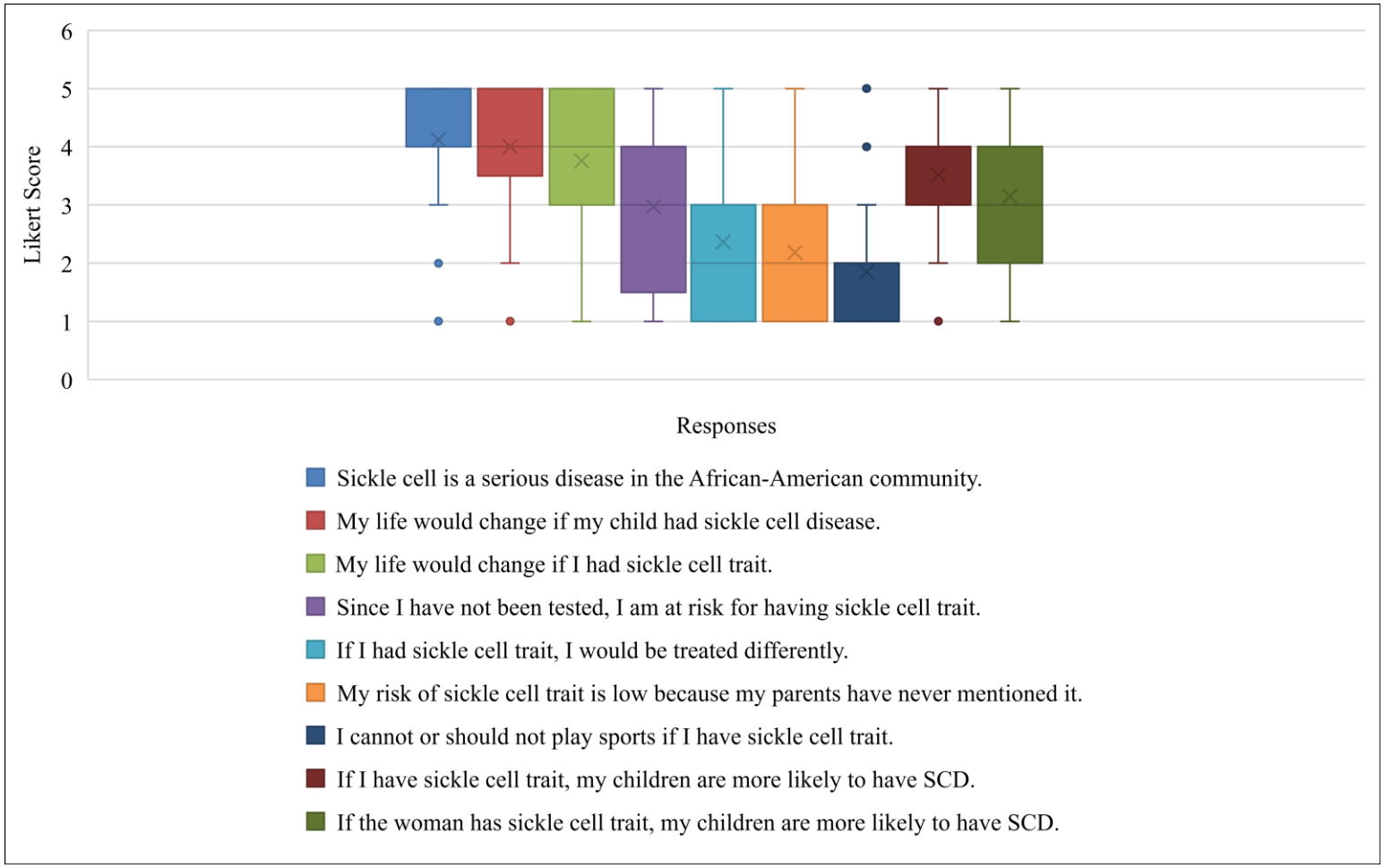


Figure 4.7 Box-and-Whisker Plot of Likert Scale Responses of Risk Perception for Sickle Cell Trait and Sickle Cell Disease in Participants Age 18 to 30

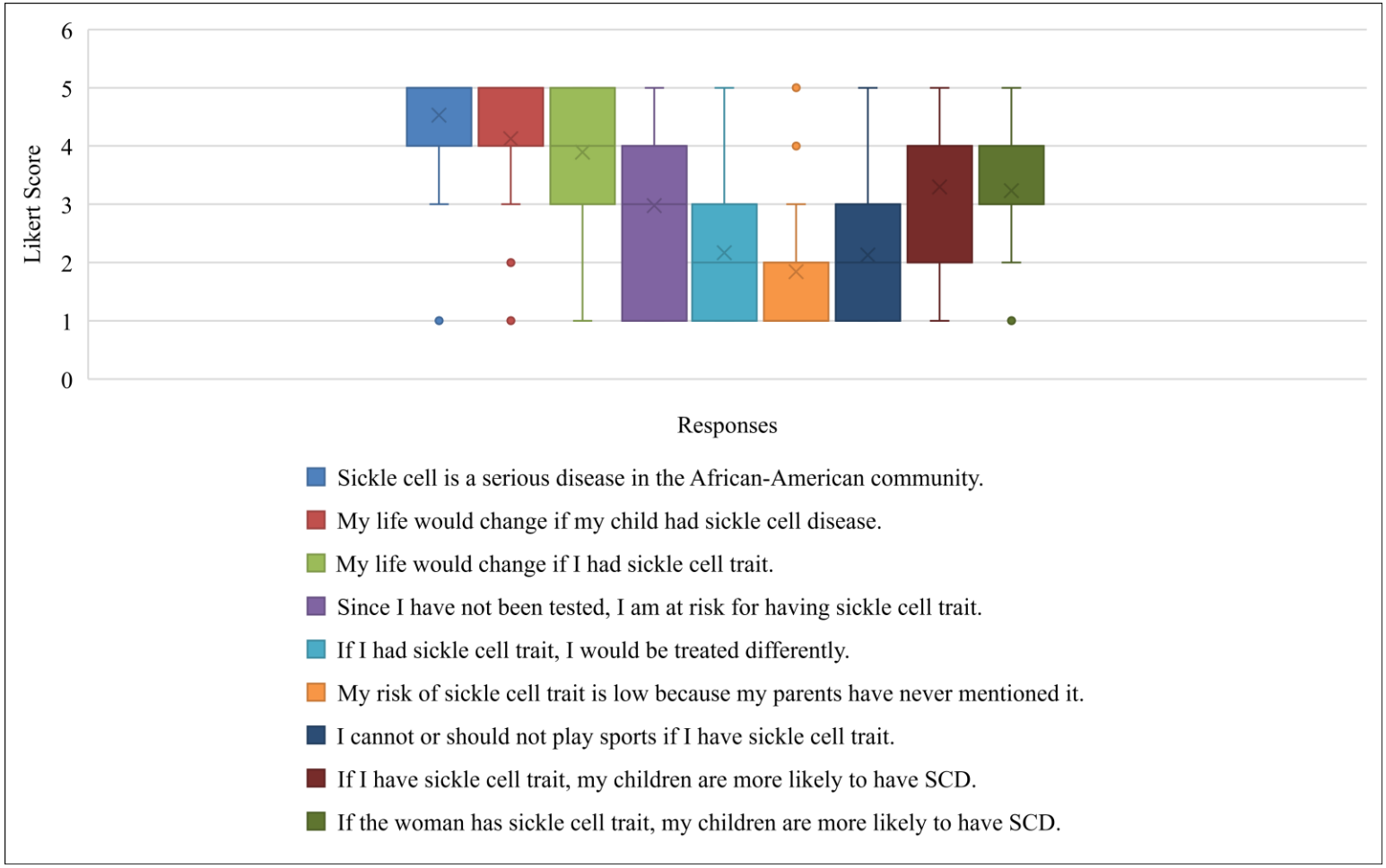


Figure 4.8 Box-and-Whisker Plot of Likert Scale Responses of Risk Perception for Sickle Cell Trait and Sickle Cell Disease in Participants Age 31 to 49

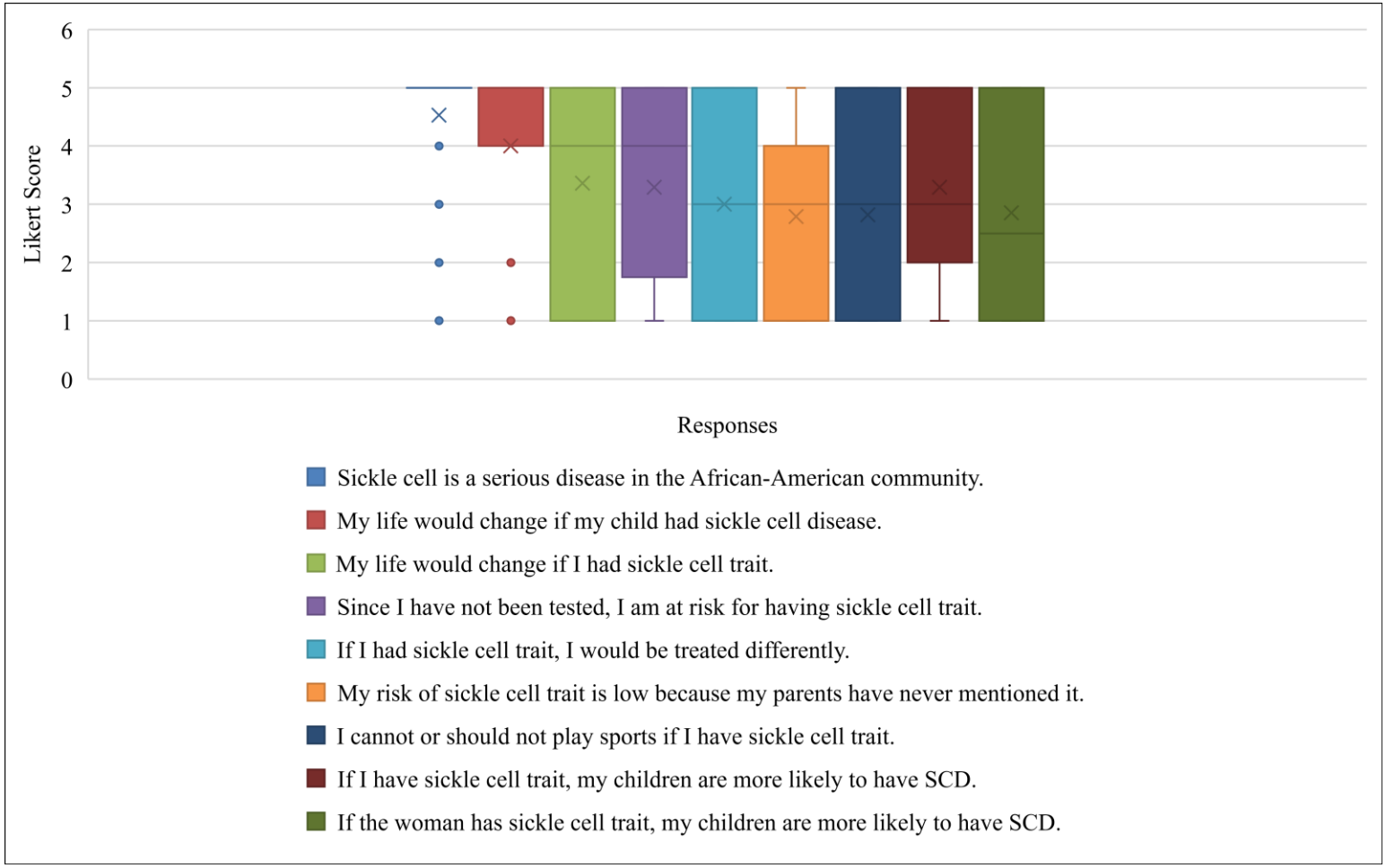


Figure 4.9 Box-and-Whisker Plot of Likert Scale Responses of Risk Perception for Sickle Cell Trait and Sickle Cell Disease in Participants Age 50+

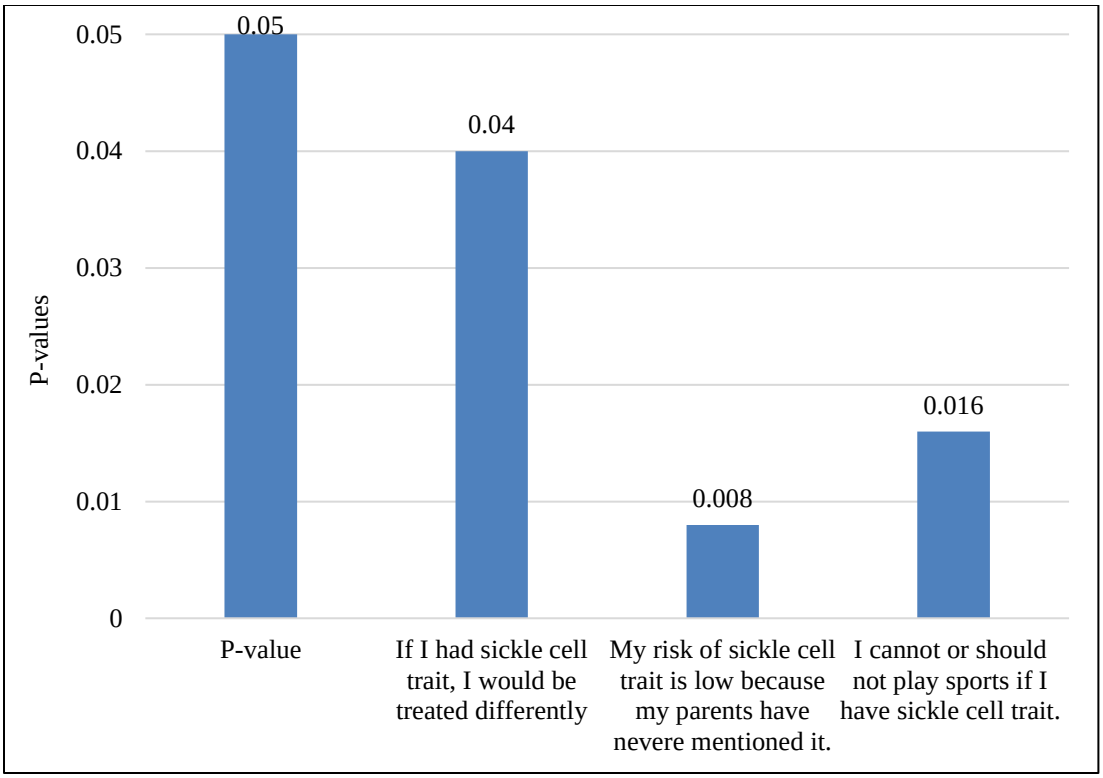


Figure 4.10 Observation of Statistically Significant Health and Risk Perception Questions about Sickle Cell Trait vs. Age

differences in the way participants answered the questions based on education level ($p>0.05$). (Table B.8).

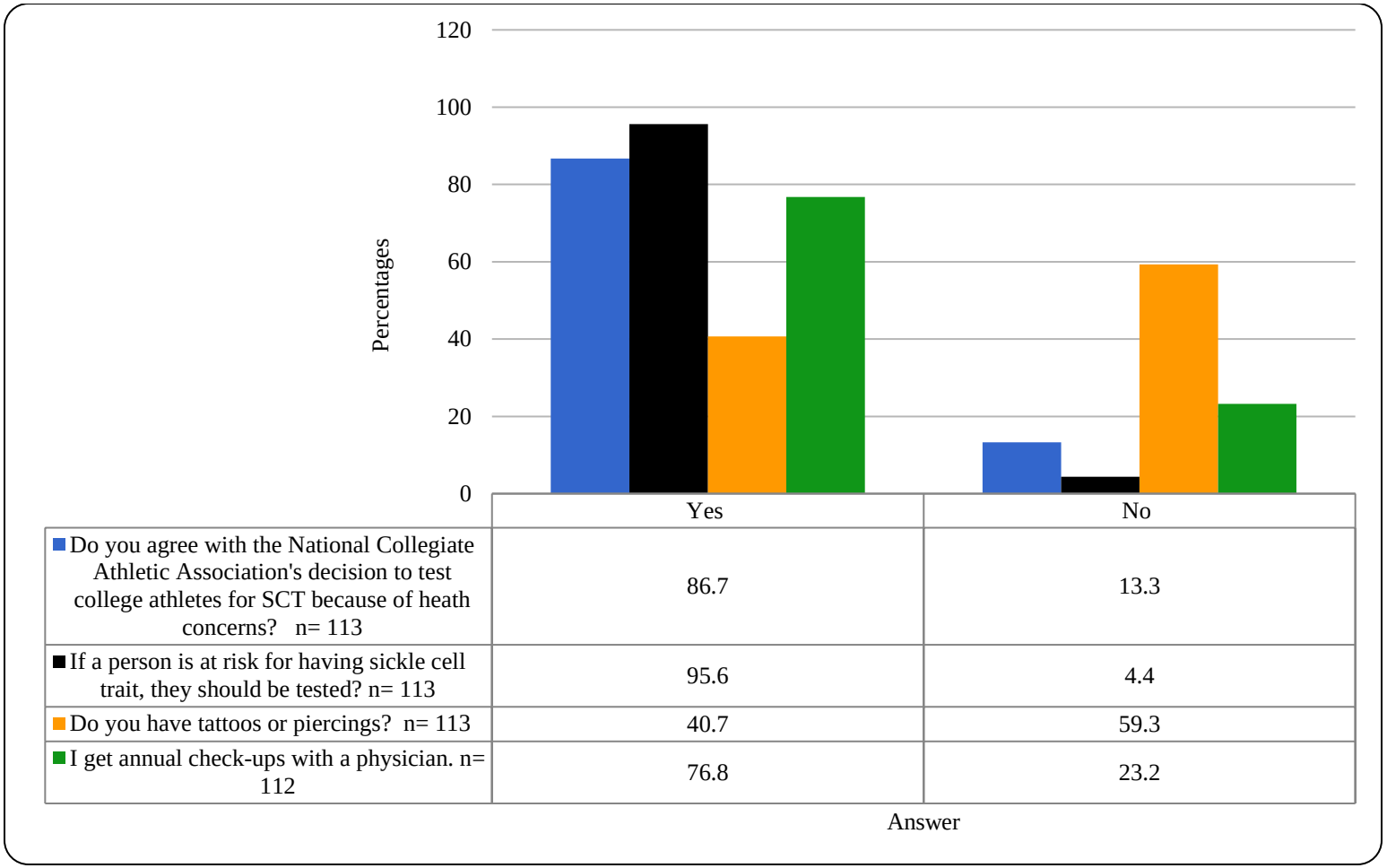
Of the over 112 participants who answered survey questions on health beliefs, 87.6% of respondents agreed with the NCAA decision to test all college athletes for SCT due to health concerns. Those same participants also agreed that any persons at risk for SCT should be tested (95.6%). Approximately 76.8% of participants reported getting annual check-ups with a physician. Overall, only 40.7% of participants reported having tattoos or piercings (Figure 4.11).

A one-way ANOVA was conducted to determine whether there were statistically significant differences in age groups and health beliefs regarding SCT and general health. There was a statistically significant difference between the groups on questions regarding having tattoos or piercings ($p = 0.019$) and getting annual check-ups with a physician ($p= 0.010$). All other questions were not statistically significant between the age groups ($p>0.05$) (Table B.9 & Figure 4.12).

Lastly, a one-way ANOVA was conducted to determine whether there were statistically significant differences in educational level and health beliefs regarding SCT and general health. Analysis showed there were no statistically significant differences in the way the participants answered based on their educational level ($p>0.05$) (Table B.10).

4.3 SPECIFIC AIM II: UNDERSTANDING BARRIERS AND MOTIVATING FACTORS IN TESTING FOR SCT

Overall, the "Not applicable" category was chosen the most by participants when asked about barriers to genetic testing (36.2%). The second largest barrier to testing was reported to be doctors never mentioning SCT testing during health visits (29.3%). The



	Yes	No
Do you agree with the National Collegiate Athletic Association's decision to test college athletes for SCT because of heath concerns? n= 113	86.7	13.3
If a person is at risk for having sickle cell trait, they should be tested? n= 113	95.6	4.4
Do you have tattoos or piercings? n= 113	40.7	59.3
I get annual check-ups with a physician. n= 112	76.8	23.2

Figure 4.11Participant Responses to Health Belief Questions Regarding Sickle Cell Trait and General Health

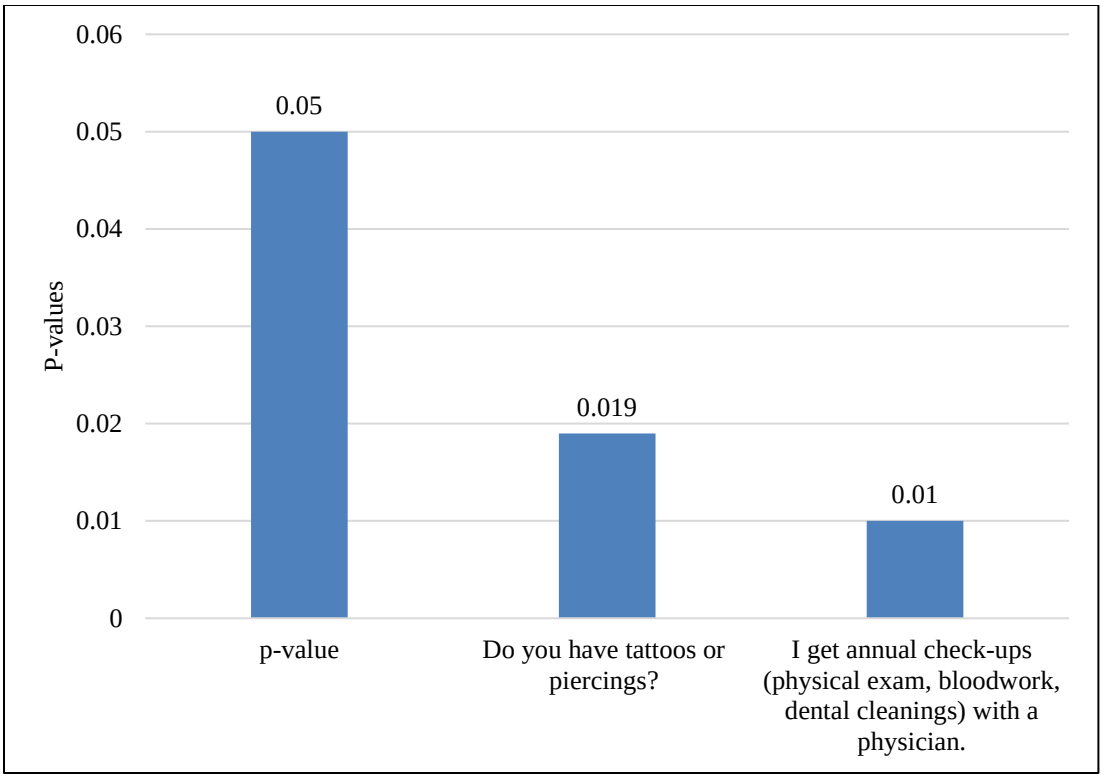


Figure 4.12 Observation of Statistically Significant Health Beliefs vs. Age

third largest barrier reported was participants not feeling they were at risk based on their family history (16.4%). Respondent reported the fourth largest barrier to testing involving cost of SCT testing (11.2%). The other 27.5% of participants chose barriers including: work time constraints (3.4%), a fear of needles (6.0%), no interest in testing (6.9%), fear of discrimination from job or insurance companies (4.3%), and distrust of SCT testing laboratories (6.9%) (Figure 4.13).

The largest motivating factor for genetic testing for SCT was reported to be respondents wanting to know their status for their own health (58.6%). The second largest motivator reported by participants was getting testing for their own or future children's health (31.9%). Approximately 23.3% of the participants when asked about motivating factors cited they had already been tested. The fourth largest motivating factor for genetic testing for SCT was being at a sickle cell health event (11.2%). The other 15.5% of participants chose motivating factors including: family history of SCT (6.9%), pressure from partner or health professional to have testing (1.7%), and positive SCT status of partner (6.9%) (Figure 4.14).

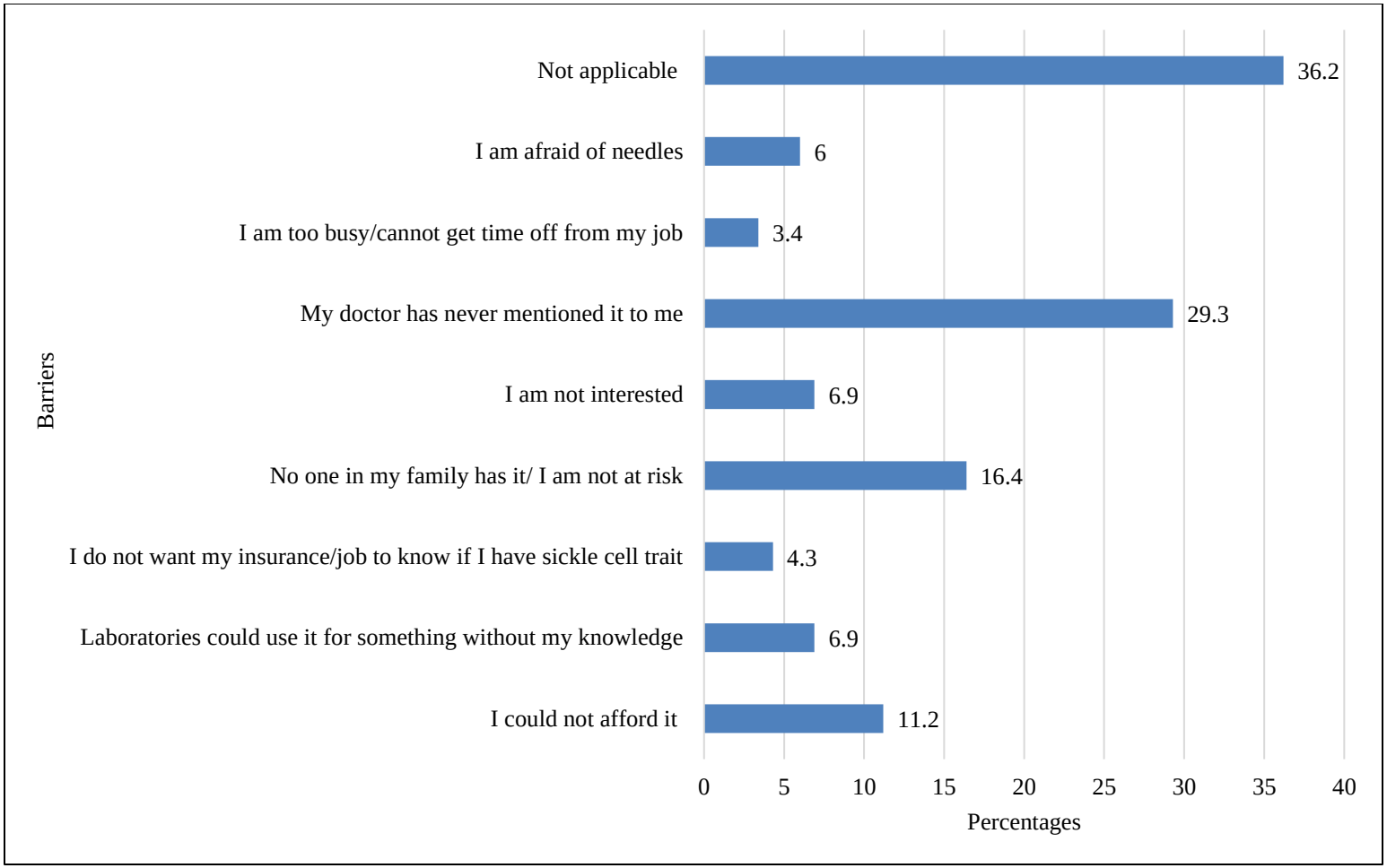


Figure 4.13 Barriers to Genetic Testing for Sickle Cell Trait

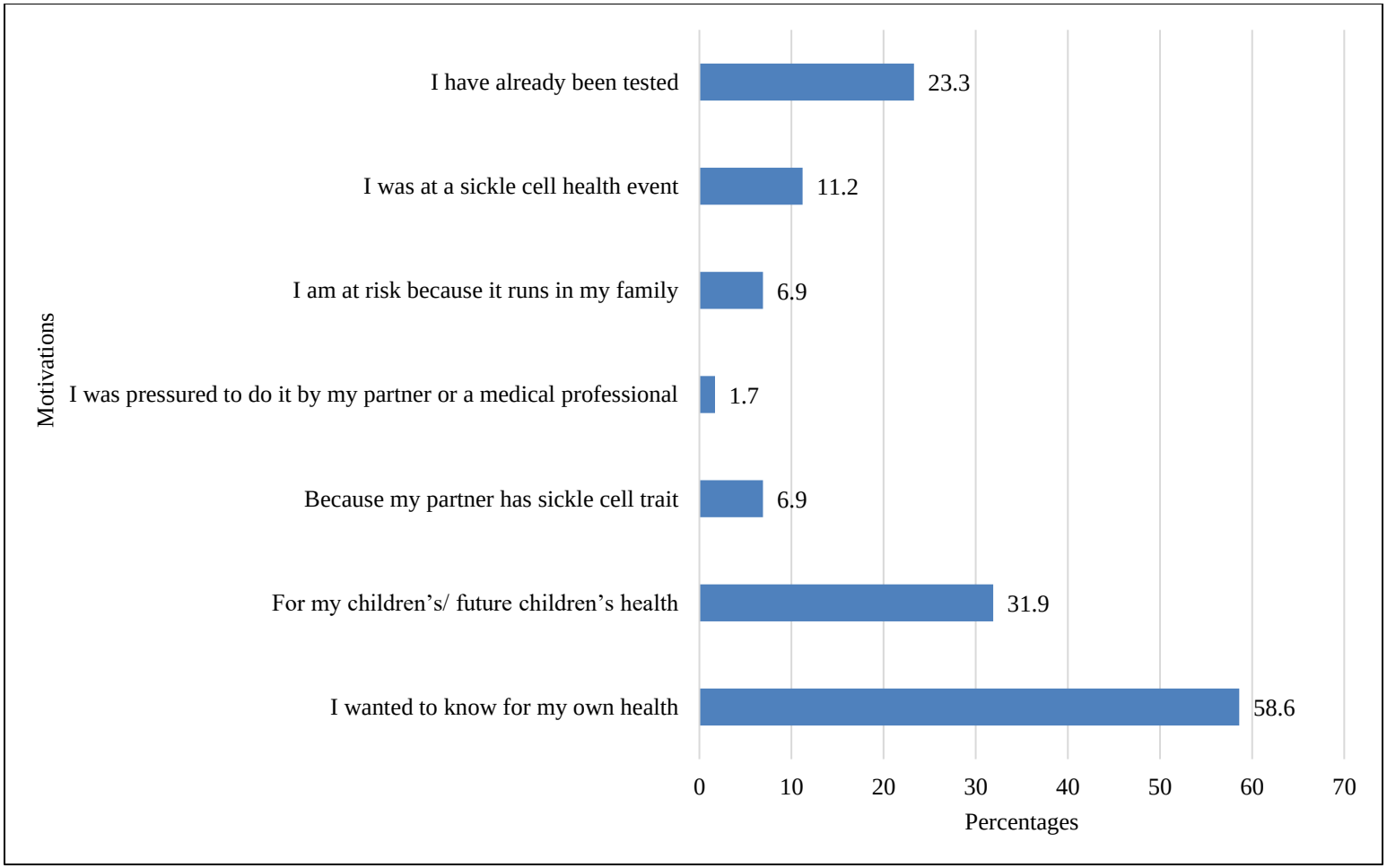


Figure 4.14 Motivating Factors to Genetic Testing for Sickle Cell Trait

CHAPTER 5

DISCUSSION

5.1 SPECIFIC AIM I: ASSESSING AFRICAN-AMERICAN MEN'S KNOWLEDGE AND PERCEPTIONS ON GENETIC TESTING, RISK, AND HEALTH WITHIN THE CONTEXT OF SCT

Based on this study, it was hypothesized that there would be statistical differences in knowledge of SCD and SCT based on age and education level. However, this research can only accept part of the hypothesis since there was only a statistically significant difference in the age groups and knowledge (Table B.1; Table B.2). There were no statistically significant differences in knowledge, risk perception, and health beliefs based on educational level (Table B.3; Table B.4; Table B.5). In Figure 4.6, there is a clear downward trend in the data showing that younger participants are more knowledgeable about SCT and SCD than their older counterparts. This could mean that healthcare professionals and educators are getting better at communicating this information than they have in the past.

When looking at the percent-correct on the "sickle cell quiz" there was one striking feature. Over 92.2% of men knew that African-American/black people are more often affected by SCD, while only 31% of men knew the national carrier frequency of SCT was 1 in 12 (Table 4.2). This highlights there may be a discordance in perceived risk versus the actual risk for SCT within this study population (Figure 4.2; Figure 4.11). It seems as if the perceived risk for these individuals is being lessened by some unknown

factor. Given that none of the participants scored a 9 out of 9 on the sickle cell quiz, perhaps this difference in perceived risk could be driven by the low health literacy on the topic that still exists within the African-American male population, thus affecting the reluctance, or refusal, to be tested for SCT, because these participants do not feel they are at a high risk for being SCT carriers.

Overall, this study showed that African-American men are more likely to have not been tested for SCT, which gives validity as to why this study was pursued (Figure 4.1). However, the data shows that African-American men may be more willing to be tested than previously thought because 98% of men in this study had never refused to be tested for SCT (Figure 4.2). A majority of those who had been tested stated that they were tested at birth or through the military, which makes sense given the historical background of SCT testing. When observing the breakdown of the reasons participants had been tested, it was interesting that participants were more likely not to have documentation stating their status. The participants in this testing cohort stated they would tell their partners their status; however, eight participants would not, or will not tell their partner about their positive status. This could be evidence that there is still stigma within the community about others knowing a person's SCT status, as there are still feelings of fear and rejection that could come from having a positive status. There is cause for concern within NBS given the low number of individuals who could produce documentation of their SCT status.

The main purpose of NBS for SCD, and ultimately SCT, was to identify and treat individuals born with SCD and to bring awareness to carriers of SCT for their own reproductive knowledge. Given that a majority of NBS programs began in the later 1980s

to early 1990s, it was expected that more participants would have known their status. This highlights that there may need to be changes made in how we report and follow-up with individuals who screen positive for SCT. In this study, a major influence uncovered was that there was a significant difference between the age groups in their perceived risk based on the parent report of SCT. This was mainly observed in the 31 to 49 age group with those participants more likely to disagree that their risk of SCT was low because their parents never mentioned their SCT status to them (Figure 4.6). The 18 to 30 and 50+ age groups were more likely to be neutral or agree with that statement respectively (Figure 4.7 & Figure 4.8). The Kavanagh et. al. study in 2010 discussed the need to make changes with how we follow-up with SCT and this present study adds to that sentiment. It is more paramount than ever for people to be aware of their status with the associated health concerns that have been discovered in recent years.

When comparing the other significant results from the Box and Whisker plots, there was a significant difference in the ages when comparing perceived discrimination with a positive test for SCT or SCD. It seems as if the 50+ age group trended more towards agreeing that they would be treated differently if they had SCT, which could be explained by those individuals being more likely to have been subjected to population screening in the early 1970s (Figure 4.8). The 18 to 30 age groups were more likely to disagree that they could not or should not play sports if they have SCT. This may be explained by these individuals being subjected to the NCAA's ruling on testing for SCT, however, more research would be needed to make this proposed correlation (Figure 4.7).

When comparing the differences between other health beliefs, results showed that age seemed to be the factor playing the most significant role (Table B.9). The questions

regarding getting annual checkups and having tattoos and/or piercings could be explained by the beliefs held by the younger versus the older participants. Older participants may be more likely to disagree with having tattoos and/or piercings and may go to the doctor more regularly than the younger men in this study (Figure 4.12).

5.2 SPECIFIC AIM II: UNDERSTADING BARRIERS AND MOTIVATING FACTORS IN TESTING FOR SCT

Overall, the barriers observed in this study highlight that over a third of participants did not feel there were any barriers stopping them from being tested for SCT. One interesting result from this section of the study shows that perhaps the men are interested in pursuing testing, but have not been approached about it from their primary healthcare provider. Normally, genetic counselors would be the individuals to order genetic testing for SCT in adult patients, but primary healthcare providers, PCP, could be another way to access testing. In Bean et. al's (2014) study, they mention this exact thought given that PCPs are more available to discuss and educate patients on SCT. It was expected that a significant barrier would be participants not feeling as if they are at risk given the observed disconnect in perceived risk versus actual risk of SCT in African-Americans (Figure 4.13).

Perhaps negative family history is driving the risk perception as well, or these participants are more knowledgeable about the inheritance of SCT and are not concerned about the risk of SCD during a pregnancy. This could explain why more participants were knowledgeable about the inheritance of SCT than they were about the inheritance of SCD on the "sickle cell quiz" (Table 4.2). With motivating factors, it was surprising that approximately 58.6% of participants were motivated to have testing for their own health

purposes. Perhaps this group of participants were more aware of the health concerns related to SCT, given that 76.7% answered that they knew about the adverse events associated with SCT. Most participants were concerned about the risk to pass on SCT to their children and cited reproductive health as another motivating factor (Figure 4.14).

5.3 SPECIFIC AIM III: RECOGNIZING THE PRACTICE IMPLICATIONS FOR GENETIC COUNSELORS

There are several different areas that seem to be more important in how to discuss testing for SCT with African-American men, which has practice implications for genetic counselors. Several of the barriers that were identified in this study could be assuaged overall by tailoring the sessions in several ways including discussions on: G.I.N.A. (Genetic Information Nondiscrimination Act) to lessen fears of discrimination at work or through insurance companies, providing logistical information about the laboratory and the testing process to decrease mistrust of SCT testing laboratories, and offering low cost ways to be tested, such as through a local Sickle Cell Disease foundation. Some other ways that genetic counselors may also want to tailor their sessions include explaining more on the proposed evolutionary mechanism behind SCT to increase knowledge about why it is so prevalent in the African-American population. Having a more developed background knowledge about the disease may lessen the feeling of being targeted to have testing that some African-Americans have expressed.

Focusing more on the potential health risks associated with SCT may also increase the uptake of SCT testing as this study shows that there may be a desire to know this information if it has an impact on their heath, or the health of their future children. Overall, 21% of individuals in this study had seen a genetic counselor for SCT. Genetic

counselors have a special skill set that could be utilized to address the physical and psychosocial barriers and motivating factors discussed within this study. Prenatal genetic counselors would have a role in educating and increasing the knowledge of their African-American patients regarding SCT and SCD. It highlights that if genetic counselors want to work to improve the uptake of genetic testing for SCT, there may have to be changes made in how African-American males are approached because they have different needs that may need to be considered as a part of the decision-making process.

5.4 LIMITATIONS

This study has several limitations. Most of the participants who participated in this study were in South Carolina and may not reflect those of African-American men in other states. Additionally, the goal of this research was to study the beliefs of males aged 18 to 45, childbearing ages, but the study design was altered as there was difficulty recruiting younger participants. Therefore, the results obtained from this study could be skewed because it included the 50+ age group, who may have different views based on the era they grew up in (i.e. during the civil rights movement when discrimination was a more common occurrence). Most of the participants were recruited from sickle cell events, which produces a major bias in the data collected from this study. It is possible that those participants were more educated on SCD and SCT themselves as they may have been carriers of SCT or had an affected family member, meaning it may not reflect the true knowledge base level of African-American men who are not a part of that cohort. This may explain why “not applicable” was the highest category chosen when asked if participants had any barriers to genetic testing for SCT.

5.5 CONCLUSION

This study set out to understand the physical and psychosocial barriers that exist within the African-American community in respect to genetic testing for SCT. Through this research, some insightful information was identified that has implications for genetic counselors who counsel patients on SCT. The knowledge obtained from this research may aid genetic counselors in increasing the uptake of testing for SCT by tailoring their sessions to focus on the needs of this group. Although there is still an issue with low health literacy in the African-American community regarding SCD and SCT, there are other confounding factors such as age, health beliefs, and perceived risk, which could influence the decision-making for this population. Genetic counselors are going to be vital in correcting and demystifying any misinformation in the community and educating individuals on the potential health concerns regarding SCT.

This study identified public health concerns stemming from the follow-up and communication of SCT results to those who screen positive for SCT. From this study, it seems the needs of those individuals are not being met and may contribute to why some African-Americans are unaware of their trait status. This study suggests a call to action should be considered towards making changes to increase awareness of SCT and SCD within the African-American community. Future research may include recreating this study in states that have similar or different methods of communicating NBS results to see how they compare to the results of this study. It may also be helpful to recreate this study in different states to see if the results are comparable between African-American men in other areas of the United States.

In conclusion, the hope is for this research to have an impact on understanding what barriers exist within the African-American male community in genetic testing for SCT and to provide representation to a population of vulnerable individuals whose voice is often too weak to be heard. This study will likely assist genetic counselors and their African-American patients with opening a dialogue that helps address their needs and increase the uptake of SCT testing in African-Americans, not just for their reproductive risk knowledge but for their overall health as well.

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APPENDIX A

INSTITUTIONAL REVIEW BOARD APPROVAL LETTER

UNIVERSITY OF
SOUTH CAROLINA

OFFICE OF RESEARCH COMPLIANCE

INSTITUTIONAL REVIEW BOARD FOR HUMAN RESEARCH
APPROVAL LETTER for EXEMPT REVIEW

Shandrea Foster
School of Medicine
Genetic Counseling
Columbia, SC 29208

Re: **Pro00067778**

This is to certify that the research study, "***Understanding Barriers to Genetic Testing for Sickle Cell Trait: The African-American Male Perspective***," was reviewed in accordance with 45 CFR 46.101(b)(**2**), the study received an exemption from Human Research Subject Regulations on **6/5/2017.** No further action or Institutional Review Board (IRB) oversight is required, as long as the study remains the same. However, the Principal Investigator must inform the Office of Research Compliance of any changes in procedures involving human subjects. Changes to the current research study could result in a reclassification of the study and further review by the IRB.

Because this study was determined to be exempt from further IRB oversight, consent document(s), if applicable, are not stamped with an expiration date.

All research related records are to be retained for at least three (3) years after termination of the study.

The Office of Research Compliance is an administrative office that supports the University of South Carolina Institutional Review Board (USC IRB). If you have questions, contact Arlene McWhorter at arlenem@sc.edu or (803) 777-7095.

Sincerely,

Lisa M. Johnson
IRB Assistant Director

APPENDIX B

SUPPLEMENTARY DATA ANALYSIS TABLES

Table B. 1 Two-way ANOVA for Sickle Cell Quiz vs. Age Groups

Tests of Between-Subjects Effects						
Dependent Variable: Total Correct on Sickle Cell Quiz						
Source	**Type III Sum of Squares**	**df**	**Mean Square**	**F**	**Sig.**	**Partial Eta Squared**
Corrected Model	34.393[a]	6	5.732	3.007	0.009	0.144
Intercept	3125.714	1	3125.714	1639.736	0	0.939
q0034	34.393	6	5.732	3.007	0.009	0.144
Error	203.967	107	1.906			
Total	4623	114				
Corrected Total	238.36	113				

Table B. 2 Two-Way ANOVA for Sickle Cell Quiz vs. Newly Defined Age Groups

Tests of Between-Subjects Effects						
Dependent Variable: Total Correct on Sickle Cell Quiz						
Source	**Type III Sum of Squares**	**df**	**Mean Square**	**F**	**Sig.**	**Partial Eta Squared**
Corrected Model	22.339[a]	2	11.17	5.739	0.004	0.094
Intercept	4239.795	1	4239.795	2178.576	0	0.952
AGE	22.339	2	11.17	5.739	0.004	0.094
Error	216.021	111	1.946			
Total	4623	114				
Corrected Total	238.36	113				

Table B. 3 Two-Way ANOVA for Sickle Cell Quiz vs. Education Level

Tests of Between-Subjects Effects						
Dependent Variable: Total Correct on Sickle Cell Quiz						
Source	**Type III Sum of Squares**	**df**	**Mean Square**	**F**	**Sig.**	**Partial Eta Squared**
Corrected Model	13.127[a]	4	3.282	1.611	0.177	0.057
Intercept	3263.928	1	3263.928	1602.481	0	0.938
q0035	13.127	4	3.282	1.611	0.177	0.057
Error	215.9	106	2.037			
Total	4481	111				
Corrected Total	229.027	110				

Table B. 4 One-Way Repeated Measures with ANOVA-Multiple Comparisons for Sickle Cell Quiz vs. Education Level

One-Way Repeated Measures with ANOVA-Multiple Comparisons						
Dependent Variable: Total Correct on Sickle Cell Quiz						
Bonferroni						
(I) Education Level:	(J) Education Level:	Mean Difference (I-J)	Std. Error	Sig.	95% Confidence Interval	
					Lower Bound	Upper Bound
Some high school	High school graduate	0.3333	0.57449	1	-1.3137	1.9804
	Some college	-0.2432	0.55646	1	-1.8386	1.3521
	College graduate	-0.4348	0.5858	1	-2.1142	1.2447
	Graduate/professional school	-0.6875	0.61798	1	-2.4592	1.0842
High school graduate	Some high school	-0.3333	0.57449	1	-1.9804	1.3137
	Some college	-0.5766	0.36123	1	-1.6122	0.4591
	College graduate	-0.7681	0.40496	0.606	-1.9291	0.3929
	Graduate/professional school	-1.0208	0.45026	0.254	-2.3117	0.2701
Some college	Some high school	0.2432	0.55646	1	-1.3521	1.8386
	High school graduate	0.5766	0.36123	1	-0.4591	1.6122
	College graduate	-0.1915	0.37895	1	-1.278	0.8949
	Graduate/professional school	-0.4443	0.42702	1	-1.6685	0.78
College graduate	Some high school	0.4348	0.5858	1	-1.2447	2.1142
	High school graduate	0.7681	0.40496	0.606	-0.3929	1.9291
	Some college	0.1915	0.37895	1	-0.8949	1.278

	Graduate/professional school	-0.2527	0.4646	1	-1.5847	1.0793
Graduate/professional school	**Some high school**	0.6875	0.61798	1	-1.0842	2.4592
	High school graduate	1.0208	0.45026	0.254	-0.2701	2.3117
	Some college	0.4443	0.42702	1	-0.78	1.6685
	College graduate	0.2527	0.4646	1	-1.0793	1.5847

Table B. 5 Independent Samples Test for Sickle Cell Quiz vs. Educational Level (High school vs. College)

Independent Samples Test						
		Levene's Test for Equality of Variances		t-test for Equality of Means		
		F	Sig.	t	df	Sig. (2-tailed)
Total Correct on Sickle Cell Quiz	Equal variances assumed	0.024	0.877	-1.899	109	0.06*
	Equal variances not assumed			-1.968	86.412	0.052

Independent Samples Test						
		t-test for Equality of Means				
		Mean Difference	Std. Error Difference	95% Confidence Interval of the Difference		
				Lower	Upper	
Total Correct on Sickle Cell Quiz	Equal variances assumed	-0.5385	0.28355	-1.10044	0.02352	
	Equal variances not assumed	-0.5385	0.27357	-1.08227	0.00535	

Table B. 6 Pearson's Product Moment Correlation for Yearly Income vs. Education Level

Pearson's Correlation			
		Education Level:	**Yearly Income:**
Education Level:	**Pearson Correlation**	1	.402**
	Sig. (2-tailed)		0
	N	111	102
Yearly Income:	**Pearson Correlation**	.402**	1
	Sig. (2-tailed)	0	
	N	102	106

Table B. 7 One-Way ANOVA for Perceived Risk and Discrimination vs. Age

ANOVA Table						
		Sum of Squares			df	Mean Square
Sickle cell is a serious disease in the African American community. * Age	**Between Groups**	**(Combined)**	3.935		2	1.967
	Within Groups	115.688			111	1.042
	Total	119.623			113	
My life would change if my child had sickle cell disease. * Age	**Between Groups**	**(Combined)**	0.447		2	0.224
	Within Groups	209.234			110	1.902
	Total	209.681			112	
My life would change if I had sickle cell trait. * Age	**Between Groups**	**(Combined)**	5.605		2	2.802
	Within Groups	242.165			110	2.202
	Total	247.77			112	
Since I have not been tested, I am at risk for having sickle cell trait. * Age	**Between Groups**	**(Combined)**	2.431		2	1.216
	Within Groups	253.007			111	2.279
	Total	255.439			113	
If I had sickle cell trait, I would be treated differently. * Age	**Between Groups**	**(Combined)**	14.146		2	7.073
	Within Groups	236.275			111	2.129

	Total	250.421		113	
My risk of sickle cell trait is low because my parents have never mentioned it. * Age	**Between Groups**	**(Combined)**	17.034	2	8.517
	Within Groups	184.335		108	1.707
	Total	201.369		110	
I cannot or should not play sports if I have sickle cell trait. * Age	**Between Groups**	**(Combined)**	16.676	2	8.338
	Within Groups	212.386		110	1.931
	Total	229.062		112	
If I had sickle cell trait, my children are more likely to have Sickle Cell Disease. * Age	**Between Groups**	**(Combined)**	1.123	2	0.562
	Within Groups	193.131		111	1.74
	Total	194.254		113	
If a woman has sickle cell trait, my children are more likely to develop SCD. * Age	**Between Groups**	**(Combined)**	3.006	2	1.503
	Within Groups	200.933		111	1.81
	Total	203.939		113	

Table B. 8 One-Way ANOVA for Health and Perceived Risk vs. Education Level

ANOVA Table							
		Sum of Squares		df	Mean Square	F	Sig.
Sickle cell is a serious disease in the African American community. * Education Level	Between Groups	(Combined)	0.053	1	0.053	0.048	0.827
	Within Groups	118.138		108	1.094		
	Total	118.191		109			
My life would change if my child had sickle cell disease. * Education Level	Between Groups	(Combined)	0.001	1	0.001	0	0.983
	Within Groups	203.669		107	1.903		
	Total	203.67		108			
My life would change if I had sickle cell trait. * Education Level	Between Groups	(Combined)	3.051	1	3.051	1.366	0.245
	Within Groups	241.167		108	2.233		
	Total	244.218		109			
Since I have not been tested, I am at risk for having sickle cell trait. * Education Level	Between Groups	(Combined)	1.491	1	1.491	0.649	0.422
	Within Groups	248.181		108	2.298		
	Total	249.673		109			
If I had sickle cell trait, I would be treated differently. * Education Level	Between Groups	(Combined)	3.759	1	3.759	1.695	0.196
	Within Groups	239.514		108	2.218		
	Total	243.273		109			
My risk of sickle cell trait is low because my parents have	Between Groups	(Combined)	1.715	1	1.715	0.889	0.348
	Within Groups	204.535		106	1.93		

never mentioned it. * Education Level	**Total**	206.25		107			
I cannot or should not play sports if I have sickle cell trait. * Education Level	**Between Groups**	**(Combined)**	0.514	1	0.514	0.254	0.615
	Within Groups	216.202		107	2.021		
	Total	216.716		108			
If I had sickle cell trait, my children are more likely to have Sickle Cell Disease. * Education Level	**Between Groups**	**(Combined)**	0.243	1	0.243	0.141	0.708
	Within Groups	186.63		108	1.728		
	Total	186.873		109			
If a woman has sickle cell trait, my children are more likely to develop SCD. * Education Level	**Between Groups**	**(Combined)**	0.013	1	0.013	0.008	0.931
	Within Groups	191.841		108	1.776		
	Total	191.855		109			

Table B. 9 One-Way ANOVA for SCT Health Beliefs vs. Age

ANOVA Table							
			Sum of Squares	df	Mean Square	F	Sig.
If yes, do you have documentation stating you do not have sickle cell trait? * Age	Between Groups	(Combined)	1.179	2	0.589	2.472	0.098
	Within Groups		9.298	39	0.238		
	Total		10.476	41			
Were you told that you have sickle cell trait? * Age	Between Groups	(Combined)	0.237	2	0.119	0.615	0.546
	Within Groups		7.324	38	0.193		
	Total		7.561	40			
If you had sickle cell trait, would your partner be aware of it? * Age	Between Groups	(Combined)	0.027	2	0.014	0.087	0.917
	Within Groups		5.748	37	0.155		
	Total		5.775	39			
Have you ever refused testing for sickle cell trait? * Age	Between Groups	(Combined)	0.016	2	0.008	0.444	0.642
	Within Groups		1.949	111	0.018		
	Total		1.965	113			
Have you or your partner ever been to a genetic counselor to test for sickle cell trait? * Age	Between Groups	(Combined)	0.445	2	0.223	1.323	0.271
	Within Groups		17.657	105	0.168		
	Total		18.102	107			

Do you agree with the National Collegiate Athletic Association's decision to test college athletes for sickle cell trait because of health concerns? * Age	**Between Groups**	**(Combined)**	0.009	2	0.004	0.038	0.963
	Within Groups		12.982	109	0.119		
	Total		12.991	111			
If a person is at risk for having sickle cell trait, they should be tested? * Age	**Between Groups**	**(Combined)**	0.033	2	0.017	0.38	0.685
	Within Groups		4.744	109	0.044		
	Total		4.777	111			
Do you have tattoos or piercings? * Age	**Between Groups**	**(Combined)**	1.883	2	0.941	4.098	0.019
	Within Groups		25.037	109	0.23		
	Total		26.92	111			
I get annual check-ups (physical exam, bloodwork, and dental cleanings) with a physician. * Age	**Between Groups**	**(Combined)**	1.629	2	0.815	4.812	0.01
	Within Groups		18.281	108	0.169		
	Total		19.91	110			

Table B. 10 One Way ANOVA for SCT Health Beliefs vs. Education Level

ANOVA Table							
	Sum of Squares		df	Mean Square		F	Sig.
If yes, do you have documentation stating you do not have sickle cell trait? * Education Level	Between Groups	(Combined)	0.354	1	0.354	1.405	0.243
	Within Groups	9.841	39	0.252			
	Total	10.195	40				
Were you told that you have sickle cell trait? * Education Level	Between Groups	(Combined)	0.07	1	0.07	0.383	0.54
	Within Groups	6.905	38	0.182			
	Total	6.975	39				
If you had sickle cell trait, would your partner be aware of it? * Education Level	Between Groups	(Combined)	0.001	1	0.001	0.004	0.952
	Within Groups	6.358	37	0.172			
	Total	6.359	38				
Have you or your partner ever been to a genetic counselor to test for sickle cell trait? * Education Level	Between Groups	(Combined)	0.407	1	0.407	2.637	0.107
	Within Groups	15.747	102	0.154			
	Total	16.154	103				
Do you agree with the National Collegiate Athletic Association's decision to test college athletes for sickle cell trait because of health concerns? * Education Level	Between Groups	(Combined)	0.007	1	0.007	0.057	0.811
	Within Groups	12.91	106	0.122			
	Total	12.917	107				

If a person is at risk for having sickle cell trait, they should be tested? * Education Level	**Between Groups**	**(Combined)**	0.097	1	0.097	2.742	0.101
	Within Groups	3.755	106	0.035			
	Total	3.852	107				
Do you have tattoos or piercings? * Education Level	**Between Groups**	**(Combined)**	0.032	1	0.032	0.129	0.72
	Within Groups	26.042	106	0.246			
	Total	26.074	107				
I get annual check-ups (physical exam, bloodwork, and dental cleanings) with a physician. * Education Level	**Between Groups**	**(Combined)**	0.038	1	0.038	0.21	0.648
	Within Groups	19.175	106	0.181			
	Total	19.213	107				

APPENDIX C

SUPPLEMENTARY FIGURES

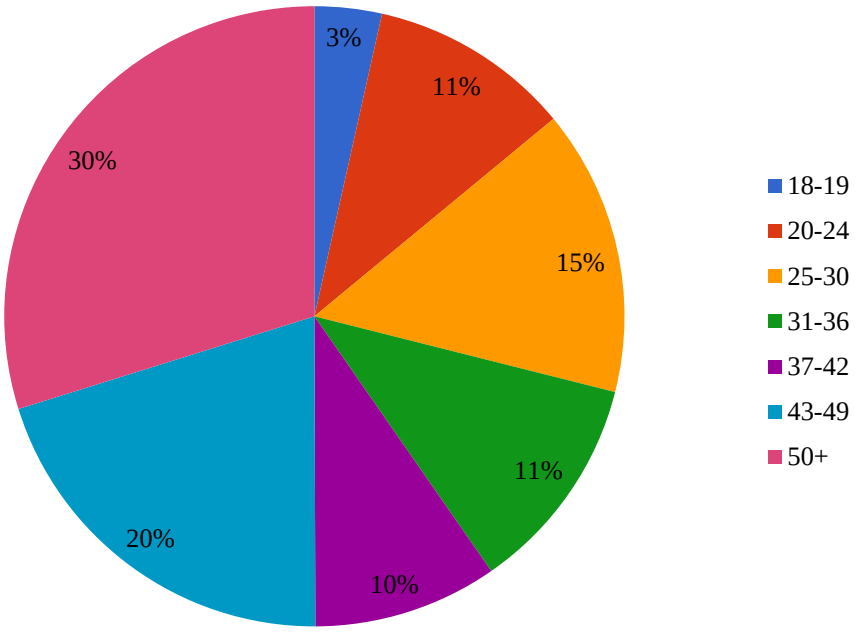

Figure C.1 Breakdown of Study Participants' Ages

APPENDIX D

STUDY QUESTIONNAIRE

Greetings,

My name is Shandrea Foster and I am a genetic counseling student in the University of South Carolina Genetic Counseling Program. You are being asked to participate in this survey because you are an 18 year, or older, African-American/black male. The purpose of this research is to gather insight into the challenges, health beliefs, and attitudes towards sickle cell trait testing within the African-American male community.

If you decide to participate, this survey will take approximately 5 to 7 minutes to complete. All survey responses collected are voluntary and anonymous, meaning no information collected can be traced back to you. If there is a question you are uncomfortable with answering, please skip it and continue on to the next question. By completing this survey, you are consenting to its use within this study and any other future academic meetings, presentations, or publications.

Each participant has the option to participate in a drawing which will include a chance to win 1 of the 2 gift cards (Chick-Fil-A, Wal-Mart, Amazon, etc.) offered every month from September to December of 2017.

To enter into the drawing, please write down your email address on the small sheet attached to the survey. Return the small slip of paper with your email address and the paper survey to the suite's front desk or to the person who presented the survey to you. Please tear this sheet away and keep it for your records. If you are a winner, look for an email with the heading titled "USC SICKLE CELL SURVEY WINNER" from this email address: shandrea.foster@gmail.com.

If you have any questions, you may contact me at shandrea.foster@gmail.com, or my faculty supervisor at Debera.Zvejnieks@uscmed.sc.edu, or us both at (803) 545-5775. If you have any questions about your rights as a participant, please contact the University of South Carolina's Office of Research Compliance at 803-777-7095.

Thank you for your consideration,
Shandrea Foster
(803) 545-5775
Shandrea.foster@gmail.com

Assessing Health Literacy

Please circle the answer you think is correct.

1. Sickle cell disease (SCD) is caused by…

a) sexual transmission b) 2 altered genes passed from parents c) 1 altered gene passed from the parent

d) a virus

2. Sickle cell trait (SCT) is caused by…

a) sexual transmission b) 2 altered genes passed from the parent c) 1 altered gene passed from the parent

d) a virus

3. Individuals with sickle cell disease…

a) have pain crises b) get more infections c) can have organ damage
d) all of the above

4. Sickle cell disease most often occurs in….

a) boys b) girls c) black people d) white people

5. One out of every _____ African-Americans has sickle cell trait.

a) 1 out of every 12 people b) 1 out of every 60 people c) 1 out of every 1000 people

6. Sickle cell disease makes red blood cells…

a) round and soft b) stiff and round c) soft and sickle shaped d) hard and sickle shaped

7. How can you tell if someone carries the gene for sickle cell trait or sickle cell disease?

a) they look sick b) they will eventually have sickle cell disease c) with a blood test
d) there is no way of e) none of the above

8. Individuals with sickle cell trait can have symptoms under extreme conditions, such as when dehydrated or in low oxygen environments. a) True b) False

9. Sickle cell trait and sickle cell disease can be cured? a) True b) False

Risk and Health Perceptions:
On a scale of 1 to 5, please rate your level of agreement with the following statement.
1= strongly disagree; 2= mostly disagree; 3= neutral; 4= mostly agree; 5= strongly agree

Questions: **Strongly Disagree 1 2 3 4 5 Strongly Agree**

10. Sickle cell is a serious disease in the African-American community. 1 2 3 4 5

11. My life would change if my child had sickle cell disease. 1 2 3 4 5

12. My life would change if I had sickle cell trait. 1 2 3 4 5

13. Since I have not been tested, I am at risk for having sickle cell trait. 1 2 3 4 5

14. If I had sickle cell trait, I would be treated differently. 1 2 3 4 5

15. My risk of sickle cell trait is low because my parents have never mentioned it 1 2 3 4 5

16. I cannot or should not play sports if I have sickle cell trait. 1 2 3 4 5

17. If I have sickle cell trait, my children are more likely to have SCD. 1 2 3 4 5

18. If the woman has sickle cell trait, my children are more likely to have SCD. 1 2 3 4 5

Healthcare Aspects and Healthcare Professionals
Please answer yes or no to the following questions.

19. Have you ever refused testing for sickle cell trait? Yes No

20. Have you ever been tested for sickle cell trait? Yes No

If you answered no to question 20, please skip questions 21, 22, 23, and 24. Then continue to question 25.
If you answered yes to question 20, please answer questions 21, 22, 23, and 24. Then continue to question 25.

21. If yes, why were you tested? **Please circle all that apply.**

During partner's pregnancy Sports Personal health reasons Military Tested at birth Other

22. If yes, do you have documentation stating you do not have sickle cell trait? Yes No

23. Were you told that you have sickle cell trait? Yes No

24. If you had sickle cell trait, would your partner be aware of it? Yes No

25. Have you or your partner ever been to a genetic counselor to test for sickle cell trait? Yes No

26. Do you agree with the National Collegiate Athletic Association's decision to test college athletes for sickle cell trait because of health concerns? Yes No

27. If a person is at risk for having sickle cell trait, they should be tested? Yes No

28. Do you have tattoos or piercings? Yes No

29. I get annual check-ups (physical exam, bloodwork, and dental cleanings) with a physician. Yes No

Barriers to Testing

30. **I refused in the past or will not get testing because: Circle all that apply**

a) I could not afford it
b) Laboratories could use it for something without my knowledge
c) I do not want my insurance/job to know if I have sickle cell trait
d) No one in my family has it/ I am not at risk
e) I am not interested
f) My doctor has never mentioned it to me
g) I am too busy/cannot get time off from my job
h) I am afraid of needles
i) Not applicable

Motivations to Testing

31**. I received genetic testing or will in the future because: Circle all that apply**

a) I wanted to know for my own health
b) For my children's/ future children's health
c) Because my partner has sickle cell trait
d) I was pressured to do it by my partner or a medical professional
e) I am at risk because it runs in my family
f) I was at a sickle cell health event
g) I have already been tested

Demographic Information of Questionnaire:
Please circle the one that most represents you.

32. Age: 18-19 20-24 25-30 31-36 37-42 43-49 50+

33. Education Level:

Some high school High school graduate Some college

College graduate Graduate/professional school

34. Yearly Income:

less than $10,000 $10,000- $20,000 $20,000- $30,000

$30,000- $40,000 $40,000- $50,000 $50,000- $100,000 $100,000 +

35. Relationship Status: Married Single In a relationship

36. Number of Children: 0 1 2 3 4 5 or more