ASSESSING THE ATTITUDES AND BELIEFS OF AFRICAN-AMERICANS TOWARD NEWBORN SCREENING AND SICKLE CELL DISEASE

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Each year 15-20 infants with sickle cell disease and 600 infants with trait are born in western Pennsylvania with no significant decrease in annual incidence. A 2006 study by Children's Hospital of Pittsburgh's Comprehensive Hemoglobinopathy Program surveying African-American women in the prenatal setting found that these women have a high perception of the severity of sickle cell disease and the importance of sickle cell trait screening but a low perceived susceptibility to sickle cell disease. The current research was designed as a qualitative follow-up study to assess African-American community members' attitudes and knowledge of sickle cell, prenatal testing, and newborn screening, to characterize barriers to education and awareness of newborn screening and sickle cell, and to determine if a community-based intervention could be developed to improve awareness of these topics. Four focus groups were conducted with 35 participants at the Kingsley Association in a predominantly African-American community of Pittsburgh. Participants were recruited from the Healthy Black Family Project. Transcripts were analyzed using thematic analysis and demographic information was compiled from a pre-discussion survey. Qualitative analysis has demonstrated that participants fall into one of three knowledge categories: the unaware, those with accurate but incomplete information, and those with misinformation. Participants have an understanding of sickle cell disease course. However, inheritance of sickle cell and the personal risk to have children or family members with the condition is not well understood. Participants have knowledge of the methods and indications for prenatal testing and value prenatal testing for the opportunities for choice and awareness. Risks of prenatal testing were identified as miscarriage as well as personal and family stress. Newborn screening was believed to be beneficial for preparation and treatment. Barriers to education and awareness of sickle cell and newborn screening were classified as personal, familial, and societal. The public health significance of this work is the identification of community members who are eager to decrease the prevalence of sickle cell in their community and the potential to design community discussion groups which address genetics topics and provide African-Americans with the tools to communicate with family and physicians about risk for sickle cell.

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PREFACE

I would like to sincerely thank my thesis committee members for their willingness to give of their time and their constant support in the formation and completion of this project. I have had a wonderful experience working with the Center for Minority Health and would like to especially thank Dr. Stephen Thomas for allowing me the opportunity to be a researcher with the Healthy Black Family Project. His enthusiasm for the project and community gave me great inspiration. I am forever grateful to Betsy Gettig and Robin Grubs for their guidance with this project and for providing a listening ear whenever the task seemed overwhelming. I want to thank Dr. Krishnamurti for helping to shape the direction of this project and for the financial resources to complete it.

A big thank you to the genetic counseling students who have come before me: Vinaya Murphy, Kristen Vogel, and Beth Dudley. You built the relationship with the community so this research could be carried forward. I would have never been able to accomplish this project without the friendship, love, and support of my comrade Leah Slattery. We kept each other going through these two years and were fortunate to share many laughs. You are truly a friend for life. Thank you to my family, Mom, Dad, and Evan. I am fortunate to have a family as wonderful as you. Thank you Tim for being my daily support and for always making me feel loved and appreciated. I cannot wait for the next step.

1.0 INTRODUCTION

The Center for Minority Health at the University of Pittsburgh's Graduate School of Public Health is devoted to eliminating racial and ethnic health care disparities in the Pittsburgh area and to providing the tools for individuals to make informed decisions regarding both their own health and the health of their families. Stephen B. Thomas, Director of the Center for Minority Health, along with a team of public health educators have established a mission based on the goals set forth by US Department of Health and Human Services Healthy People 2010 project: to eliminate ethnic and racial health disparities by the year 2010. This mission has historically focused on seven core health issues including: cardiovascular disease, diabetes, hypertension, HIV and AIDS, infant mortality, immunizations, and mental health. The Healthy Black Family Project is an extension of this mission with goals to reduce the rates of hypertension and diabetes in the African-American populations of Pittsburgh. The project was established by the Center for Minority Health in 2003 and is housed within the Kingsley Center and Hosanna House of East Liberty and Wilkinsburg, respectively. These primarily African-American Pittsburgh communities have been the focus of this intervention project but have also provided the energy to fuel a project which aims to help individuals change their lifestyle behaviors.

Through the Healthy Black Family Project, a community based health intervention project, African-Americans living in the Pittsburgh area have the opportunity to participate in physical activity, smoking cessation, stress management, diabetes support group, and nutrition classes at no charge. Instructors provide not only the leadership for classes but also the motivation and positive reinforcement necessary to make these lifestyle changes. Additionally, a team of genetic counseling students invite members to learn more about their family health history and the relationship between familial diseases and individual health through the Family Health History Initiative. Members meet with a genetic counseling student to construct a three generation pedigree and then engage in a discussion about their individual health risks based on information gathered in the family health history. At the conclusion of the family health history, an individual is invited to participate in the Minority Research Recruitment Database. Informed consent allows the Center for Minority Health to contact the individual in the event future research studies become available and he/she would be eligible to participate.

To date, three genetic counseling students have completed thesis projects analyzing the impact of the Family Health History Initiative. Kristen Vogel examined the characteristics of individuals who complete the family health history and choose to participate in the Minority Research Recruitment Database (2005). Results demonstrated that women and those without health insurance were more likely to enroll (Vogel, 2005). Analysis was also able to identify the importance of incentives as motivation for participation in the database and distrust as the main barrier to enrollment (Vogel, 2005). Research by Vinaya Murthy demonstrated that HBFP participants' perceptions of colon cancer and heart disease risk were more likely to become accurate following the family health history session (2005). This analysis identified the family history as a valuable tool in identifying at-risk individuals and promoting accurate risk perceptions (Murthy, 2005). In addition to improving risk perception for chronic disease, the family health history was shown to increase self-reported physical activity of HBFP participants in research performed by Beth Dudley (2006). These prior projects have illustrated the effectiveness of the family history in increasing accuracy of health knowledge, changing lifestyle behaviors, and informing strategies for minority research recruitment.

The overall goals of the Healthy Black Family Project have focused on health conditions and health issues where a racial or ethnic disparity is evident. For example, epidemiological data on individuals residing in Allegheny County demonstrate that African-Americans are twice as likely to die from complications related to diabetes than Caucasians (Hunte, 2002). The Healthy Black Family Project has developed a strategic plan to help prevent diabetes in those at risk and improve the health of those suffering with the condition through physical activity, dietary management, and the improvement of other lifestyle behaviors. This targeted design has continued to make significant strides in lessening the impact of these common, multifactorial diseases among those residing in Pittsburgh communities. However, there has been a significant lack of attention focused on alleviating the burden of sickle cell, a single gene disorder, in the African-American populations of Pittsburgh. However, as the Healthy Black Family Project grows in its scope and ability to provide services, sickle cell disease will be a new opportunity where the project may target an intervention to decrease the burden of disease among African-Americans residing in the Pittsburgh area.

Through the aid of the Healthy Black Family Project and the Comprehensive Hemoglobinopathy Program of Children's Hospital of Pittsburgh, this project examined how the African-American community of Pittsburgh perceives sickle cell disease, sickle cell trait, individual and familial risk to pass on the condition, prenatal testing, and newborn screening. In addition, this project attempted to address the community's perceived barriers to education and awareness of sickle cell disease and newborn screening.

The Comprehensive Hemoglobinopathy Program of Children's Hospital of Pittsburgh is responsible for following infants with abnormal hemoglobinopathy newborn screen results who reside in the 32 counties identified as Western Pennsylvania. Within this region, newborn screening annually detects approximately 600 infants with sickle cell trait and 12-15 infants with sickle cell disease. The majority of these children with sickle cell trait and disease are of African-American ethnicity, and despite the efforts of the Hemoglobinopathy Program, the level of sickle cell education and awareness in this community is not sufficient. Information collected by the Hemoglobinopathy Program has demonstrated that a significant number of those families found to have a child with sickle cell trait do not elect genetic counseling through Children's Hospital of Pittsburgh (Kladny, 2005). Though a portion of these families may seek education elsewhere or receive genetic counseling via telephone, it is assumed that a majority do not receive the information necessary to understand the risks to future children and grandchildren to inherit sickle cell trait or disease (Kladny, 2005).

A previous study by the Hemoglobinopathy Program of the University of Pittsburgh employed the use of surveys to assess the impact of sickle cell education on the acceptance of genetic screening for sickle cell trait in African-American women in the prenatal setting. Results demonstrated that a brief educational intervention on sickle cell increases knowledge of sickle cell disease and the acceptance of carrier screening. Survey results also showed that African-American women of childbearing age have a high perception of the severity of sickle cell disease and the importance of sickle cell trait screening but a low perception of susceptibility to sickle cell disease and barriers to sickle cell trait screening. At the conclusion of this work, recommendations were made to incorporate the results of this study into focus groups with

African-American community members in order to learn how to improve sickle cell education and awareness (Gustafson, 2006).

The intent of this project was to accomplish the recommendations set-forth by this previous study, to provide insight into the attitudes and beliefs of Pittsburgh's African-American population toward sickle cell disease, sickle cell trait, and newborn screening, and to offer potential strategies to improve the communication between health care providers and families on the inheritance and screening practices of this condition. Future projects can use this information to develop community interventions where accurate information is disseminated, families are benefited, and the disease burden of this important genetic condition is significantly reduced.

2.0 SPECIFIC AIMS

The specific aims of this study are as follows: To assess African-American community members' 1.) knowledge and perceived risk of sickle cell 2.) attitudes toward and knowledge of prenatal testing 3.) attitudes toward and knowledge of newborn screening 4.) barriers to receiving information on newborn screening and sickle cell. Additionally, this study will attempt to determine if a community-based intervention could be developed to improve communication about sickle cell and newborn screening.

3.0 BACKGROUND AND SIGNIFICANCE

3.1 SICKLE CELL DISEASE

Sickle cell disease is an autosomal recessive genetic condition that results in red blood cells with a characteristic sickle shape. Individuals with this condition can experience hemolysis, anemia, pain episodes, swelling, and vascular occlusion potentially leading to ischemic attacks and organ damage (Richer, 2005; Wethers, 2000, "Part II"; Wilson, 2003). Sickle cell disease is a pan-ethnic condition with the highest prevalence among those of African, Mediterranean, Middle Eastern, Indian, Caribbean, and Central and South American descent (Wethers, 2000, "Part I"; Wilson et al, 2003). Every 1 in 12 African-Americans is a carrier for sickle cell trait, and the disease affects every 1 in 375 African-Americans (Richer, 2005; Wethers, 2000, "Part I").

3.1.1 MOLECULAR GENETICS AND PATHOGENESIS

Mutations in the *HBB* gene are responsible for causing the known forms of sickle cell disease including hemoglobin SS disease, hemoglobin SC disease, hemoglobin SD disease, and the two forms of sickle beta-thalassemia, HbSβ+ and HbSβ_0 (Richer, 2005; Wethers, 2000, "Part I"; Wilson et al, 2003). The *HBB* gene encodes the beta-globin chains of hemoglobin, an oxygen-carrying protein composed of two alpha and two beta chain subunits found within red blood cells. This type of hemoglobin is known as hemoglobin A. Mutations in the *HBB* gene lead to an altered beta-globin chain and resulting structural change to the protein conformation of hemoglobin. In HbSS disease, a mutation in the sixth codon of exon 1 in the *HBB* gene is responsible for creating hemoglobin S. This mutation results in the replacement of glutamic acid with valine in the amino acid chain. Two of these HbS mutations are required to cause the phenotypic features of sickle cell disease (Ballas, 2002; Wilson et al, 2003). HbSS disease represents 60-70% of all sickle cell anemia (Richer, 2005). However, additional mutations responsible for HbC, HbD, or beta-thalassemia can also cause the features of sickle cell disease in the presence of an HbS mutation (Richer, 2005).

The expressivity of clinical features in individuals with sickle cell disease is difficult to predict. However, individuals with HbSS and HbSβ_0 have the most severe disease expression (Wilson et al, 2003). In comparison to HbSβ_0, where there is a complete inability to produce a beta-globin chain, those individuals with HbSβ+ produce a smaller quantity of beta-globin chains and therefore, have a milder disease course (Wethers, 2000, "Part I").

Hemoglobin S and other abnormal beta-globin chains ultimately create a brittle consistency that is easily lysed but is also more capable of adhering to the endothelial cells of the vascular system. The pathogenic process begins when the sickled hemoglobin is polymerized as a result of deoxygenation. The red blood cell then becomes dehydrated and deformed (See Figure 1). It is this red blood cell that can adhere to the vascular endothelium. As this process occurs, cell signaling factors produced by platelets bridge the gap between receptors on the endothelium and the sickled red blood cell. The adherence of sickled red blood cells to the endothelium has been shown to be responsible for the vaso-occlusion, pain crises, and strokes that can occur in individuals with sickle cell disease (Ballas, 2002; Wilson et al, 2003).

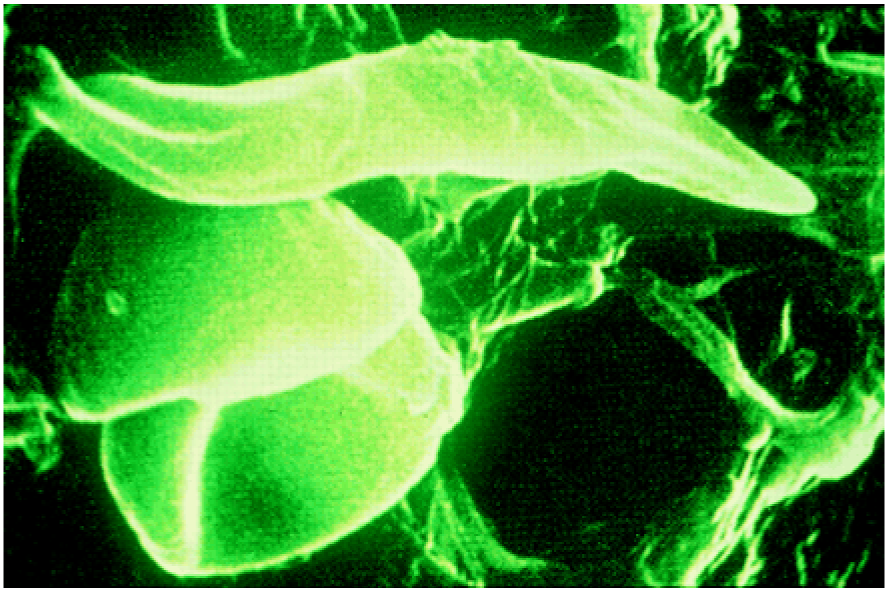

Figure 1. Sickled Cell
(Laird 2006)

3.1.2 CLINICAL COURSE OF SICKLE CELL DISEASE

The severity of the clinical course in sickle cell disease is variable among affected individuals (Wilson et al, 2003). The onset of symptoms usually occurs within the first two years of life with a presentation of body pain and swelling of the hands and feet called dactylitis (Claster & Vichinsky, 2003). The pain episodes of sickle cell disease can vary in frequency and severity. Beginning at a young age and continuing through adulthood, these pain episodes can prevent individuals from attending school and work and interfere with daily life (Claster & Vichinsky, 2003). Children and adults must be closely monitored for infection and sepsis. Infection may be marked by pain, swelling, and fever (Claster & Vichinsky, 2003). Even low-grade temperatures must be considered when evaluating an individual for infection (Claster & Vichinsky, 2003). These infections most often include Salmonella species and *Staphylococcus aureus* (Claster & Vichinsky, 2003). Human parvovirus B19 infection is responsible for approximately 80% of aplastic crises, when red blood cell production is significantly reduced in the bone marrow (Claster & Vichinsky, 2003).

The hemolysis that occurs due to the sickled shape of blood cells can lead to chronic anemia, jaundice, and delays in growth while increased red blood cell adherence to the endothelium increases the likelihood of vaso-occlusion and organ damage. Red blood cells can become trapped in the spleen causing decreased hemoglobin concentration and enlargement of the spleen (Claster & Vichinsky, 2003). This process called splenic sequestration can increase damage to the splenic tissue and potential for infection (Claster & Vichinsky, 2003). As sickled red blood cells accumulate in the endothelium of the cerebral arteries, there is a potential for brain injury (Claster & Vichinsky, 2003). Strokes usually occur without warning but can also be preceded by headaches or loss of coordination (Claster & Vichinsky, 2003). Approximately 10% of children with sickle cell disease will experience a stroke before age 18 (Claster & Vichinsky, 2003). Children between 4 and 6 years of age are at highest risk for stroke (Wethers, 2000, "Part II"). Vaso-occlusion can also cause damage to the genitourinary system. Renal acidosis, damage to the distal renal tubule, renal medullary carcinoma, proteinuria, and renal failure may occur (Claster & Vichinsky, 2003; Wethers, 2000, "Part II"). Males are at risk to experience vaso-occlusion in the form of priapism where obstruction of the venous drainage can cause a long-lasting and painful erection (Claster & Vichinsky, 2003; Wethers, 2000, "Part II").

Patients with sickle cell disease may experience acute chest syndrome, a condition characterized by fever, chest pain, and/or difficulty breathing. The appearance of a new pulmonary infiltrate on radiological studies of the chest is evidence for a diagnosis of acute chest syndrome and can be caused by infection and/or infarction. Both bacteria and viruses may be the infectious agents responsible. However, the most common cause of acute chest syndrome is a fat emboli from an infarction of the long bone which travels to the lung (Claster & Vichinsky, 2003; Wethers, 2000, “Part II”).

Individuals with sickle cell disease have a shortened lifespan in comparison to their healthy counterparts. However, cohort studies following children with sickle cell disease from birth to age 18 years have demonstrated an increase in the mean age at death and a decrease in childhood mortality. Infection has been identified as the number one cause of death in sickle cell patients, but studies are demonstrating a decrease in mortality rates due to infection. This increase in survival and decrease in mortality due to infection are attributed to early identification of disease through newborn screening and early intervention through prophylactic antibiotics (Quinn et al, 2004).

3.1.3 INHERITANCE AND RECURRENCE RISK

Sickle cell disease is inherited in an autosomal recessive manner (Richer, 2005). In the majority of cases, affected individuals have parents who are obligate heterozygotes or carriers for sickle cell trait. Individuals with sickle cell trait do not experience health effects as a result of their carrier status. The parents of an affected child who are known carriers of sickle cell trait have a 25% chance to have a second pregnancy with sickle cell disease, a 25% chance to have an unaffected child, and a 50% chance to have a child who is a carrier of sickle cell trait. All children of an affected individual will inherit a sickle cell mutation from this parent.

3.1.4 DIAGNOSIS

A sickle cell disease diagnosis can be achieved through hemoglobin electrophoresis, isoelectric focusing, high performance liquid chromatography, and/or molecular testing. These tests have comparable accuracy, but isoelectric focusing and high performance liquid chromatography are the most common methods performed to achieve an initial diagnosis. Automated isoelectric focusing provides a high degree of resolution when determining what types of hemoglobin are present. Liquid chromatography is useful for separating out proteins even at low concentrations, but this technique is limited by its inability to detect $HbS\beta_0$ (Richer, 2005; Wethers, 2000, "Part I").

Solubility tests such as Sickledex should never be used in the diagnosis of sickle cell disease. This test can identify the presence of sickled cells but is unable to determine which hemoglobin variants are present. Newborns have inaccurate solubility test results due to the predominance of fetal hemoglobin present at birth. Solubility testing has also been shown to be inaccurate in individuals with severe anemia (Wethers, 2000, "Part I").

Clinical molecular genetic testing is available for the *HBB* gene through targeted mutation analysis and sequencing (Richer, 2005). A detection rate of approximately 99% is reported by laboratories providing testing. Molecular testing can be used to confirm the diagnosis of sickle cell disease, test carriers, and perform prenatal diagnosis with a known mutation.

3.1.5 MANAGEMENT OF SICKLE CELL DISEASE

Sickle cell disease requires a team of physicians for the management of clinical features. For patients, pain is often the most difficult feature to manage because of the potential for physicians' misperceptions regarding the cause of pain and fear of addiction to pain medication (Wethers, 2000, "Part II"). Pain can be due to vaso-occlusion, acute chest syndrome, avascular necrosis, and other orthopedic problems (Claster & Vichinsky, 2003). Pain crises often require hospitalization and IV narcotic pain medication. Prophylactic hydroxyurea therapy has been shown to lessen the incidence and severity of pain episodes and incidence of acute chest syndrome in adults with sickle cell disease. There is limited data on the use of this therapy in children. Adequate hydration, oral analgesics, and avoidance of extreme heat and cold have been shown to alleviate the occurrence of pain crises. Often a pain management plan is instituted to prevent delay in the control of pain when a patient seeks care at a hospital emergency room (Claster & Vichinsky, 2003; Wethers, 2000, "Part II").

Prophylactic antibiotics should be started as soon as a diagnosis of sickle cell disease is established. Preferably, this occurs as soon as possible after a positive newborn screening result. Providing prophylactic penicillin to infants has been shown to reduce mortality from pneumococcal sepsis. Proper immunizations should be adhered to in a timely manner for these children. The influenza vaccine is also strongly encouraged (Claster & Vichinsky, 2003; Wethers, 2000, "Part II").

Management guidelines are in place for acute chest syndrome, enlarged spleen, and stroke. Sickle cell disease patients are often followed closely by pulmonology specialists for acute chest syndrome and pulmonary hypertension. Treatment for acute chest syndrome may require antibiotics, pain medication, and oxygen. Those individuals with pulmonary hypertension can experience cardiac and respiratory complications. Oxygen therapy, hydroxyurea treatment, vasodilation, and/or anti-coagulants may be beneficial to prevent progression of this condition where there is potential for sudden death. An enlarged spleen can but does not have to follow an infection in sickle cell patients. Treatment may include penicillin, transfusion, and/or splenectomy depending on the severity of the condition. The strokes experienced by individuals with sickle cell can be both silent and catastrophic events. Symptoms of headache, hemiparesis, seizures, or changes in coordination or personality may be indicative of a stroke. Due to the high risk of a second stroke, these patients are most often put on a preventative transfusion protocol after confirmation of the initial stroke through MRI (Claster & Vichinsky, 2003; Wethers, 2000, “Part II”).

3.2 NEWBORN SCREENING

3.2.1 HISTORY OF NEWBORN SCREENING

Newborn screening is a specific panel of tests performed on a blood sample obtained from the newborn in the first few hours of life. Newborn screening has a history beginning in the early 1960s when Dr. Robert Guthrie developed a simple and effective test to screen newborns for phenylketonuria (Pass, 2000). Phenylketonuria is a genetic disease caused by the body's inability to metabolize the amino acid phenylalanine into tyrosine. The essential enzyme phenylalanine hydroxylase is rendered non-functional due to mutations in the *PAH* gene. When left untreated, phenylketonuria causes an accumulation of phenylalanine in the brain and ultimately, mental retardation. The testing process implemented by Dr. Guthrie was designed to take a sample of blood from the infant's heel which was then placed on a piece of filter paper (Pass, 2000). The sample could be analyzed using bacterial inhibition techniques applied directly to the filter paper (Pass, 2000). It was a fast, efficient, and sensitive method of determining if an infant was affected by phenylketonuria (Pass, 2000). Positive results indicated the need to administer restrictive dietary therapy which could prevent accumulation of phenylalanine and stop the process leading to mental retardation (Pass, 2000).

Newborn screening for phenylketonuria was eventually adopted by all states in order to attempt to eliminate this preventable form of mental retardation (Therrell, 2006). Mandatory newborn screening programs required states to pay for laboratory fees and medical follow-up for those children with positive results (Therrell, 2006). However, the screening program for phenylketonuria was believed to be cost-effective for states because it decreased the need to pay for the institutionalization of individuals with mental retardation resulting from this genetic condition (Therrell, 2006).

3.2.2 CURRENT STATUS OF NEWBORN SCREENING IN THE UNITED STATES

In general, the newborn screening process can be broken down into five components: screening of the newborn, follow-up, diagnostic testing, disease management, and evaluation. The screening of newborns begins with obtaining parental informed consent in some states and then a heel stick blood sample is spotted onto filter paper or Guthrie card. This card is then sent to either a state sponsored or commercial lab where automated testing is typically performed. In most states, follow-up is pursued only if the infant has a screening result which falls outside the normal range (See Figure 2). Negative results are reported to the pediatrician and do not require the infant to be evaluated. Follow-up of abnormal newborn screen results includes locating the infant and then contacting the family to inform them of the result.

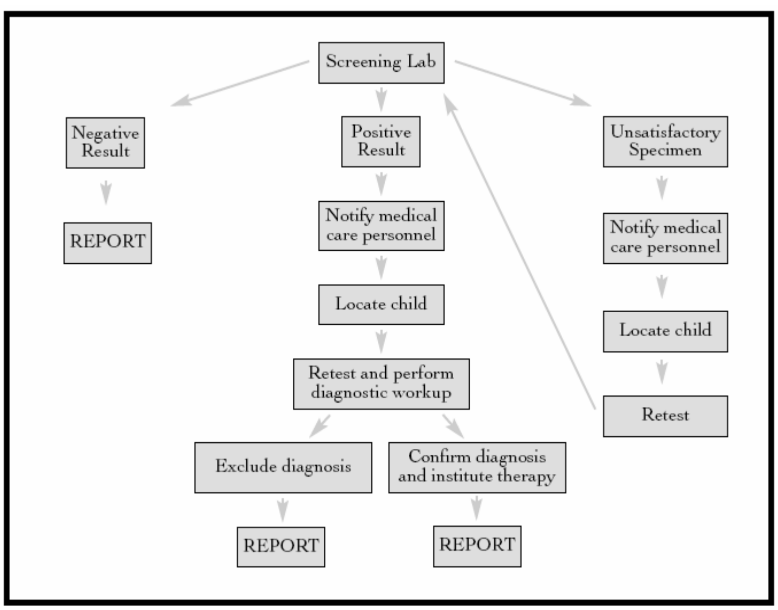

Figure 2. Follow-up Process for Newborn Screening Results (Pass, 2000)

Appointments are established with a newborn screening center or pediatrician within about two days following the result. Upon evaluating the infant at this appointment, further diagnostic testing is often pursued. Newborn screening provides indication of laboratory values outside the normal range but cannot always be diagnostic. Further testing often includes DNA testing and/or more specialized laboratory tests. Once a formal diagnosis has been established, the infant is typically followed by their pediatrician for management of the condition. Management can include treatment, dietary and nutrition counseling, genetic counseling to discuss the inheritance and recurrence risk of the condition, and addressing the psychosocial needs of the family. The newborn screening program must be continuously evaluated to ensure it is meeting the needs of the families who are affected by a positive diagnosis and that the child's care is coordinated as he/she ages (Kaye, 2006; Pass, 2000).

There is currently no federal law overseeing newborn screening in the United States which leads to a lack of standardization and consistency from state to state (Therrell, 2006). The choice of which genetic conditions are screened for and how these screening programs are regulated is controlled by individual state legislation (Therrell, 2006). Currently, all states screen for sickle cell disease and trait except New Hampshire where screening is required but has not been completely implemented (Kaye, 2006). Only twenty states have regulations which include consent requirements and each of these states differs in what requirements parents must fulfill to opt in or out of screening (Therrell, 2006). Additionally, only twenty states have regulations which require newborn screening program healthcare professionals to provide education to parents prior to the screening process (Therrell, 2006). The method of parental education may be oral, written, or both, and the timing of education in some states can come well after the testing process has occurred (Therrell, 2006). Fifty programs have developed a newborn screening pamphlet which in the majority of cases describes how the blood sample will be obtained, what conditions are being screened for, and how results will be reported (Therrell, 2006). Less than half of newborn screening program pamphlets discuss the possibility of a false-positive result, the limitations of testing, and when results will be available (Therrell, 2006). Examination of the readability and user-friendliness of these newborn screening brochures has demonstrated that 92% are written above an eighth grade reading level which has been established as the average reading level of a US adult (Arnold, 2006). Additionally, 81% of brochures required improvement in helping parents to identify the essential information (Arnold, 2006). A study surveying the 52 newborn screening programs in the United States and its territories regarding genetic counseling following a positive diagnostic test found that 76% of programs routinely offer counseling to families of affected infants (Farrell, 2001). Even more striking, only about 50% of programs offer genetic counseling after diagnostic testing reveals a false-positive newborn screen result or an infant is identified as a carrier (Farrell, 2001).

The follow-up appointment for infants with an abnormal newborn screen result also lacks standardization in the information which is communicated to parents. A 2005 study examining the content of communication between pediatric residents and parents following a newborn screen result for sickle cell trait demonstrated many missed opportunities by physicians to share key information. This study analyzed conversations for key content regarding sickle cell disease and trait, early placement of good news regarding sickle cell trait in comparison to sickle cell disease, and the ratio of individualized to general information. Results demonstrated that only 8.5% of transcripts contained the key content thought to be necessary for parental understanding and only 27% provided reassuring news about sickle cell carrier status in relation to sickle cell disease. Approximately 22% of physician statements were in regards to sickle cell disease which the newborn did not have and 50% of transcripts included misinformation about the risk of sudden death in those with sickle cell trait (Farrell, 2005).

There is evidence to suggest that general practitioners and pediatricians who manage abnormal newborn screen results do not feel competent in this role. A randomized mailed survey of these physicians found that 89% of pediatricians and 44% of general practitioners had managed an abnormal newborn screen result within the past five years (Kemper, 2006). Approximately 56% of these physicians would prefer newborn screening centers to provide the initial evaluation of the infant and a majority of general practitioners stated that they did not feel competent discussing the conditions found on the newborn screening panel (Kemper, 2006). Interestingly, only 64% of pediatricians felt it was necessary to refer newborns with sickle cell trait for genetic counseling while 81% believed it was necessary to refer newborns who are carriers for cystic fibrosis (Kemper, 2006). This could be explained by a lack of understanding regarding the autosomal recessive inheritance of both of these conditions. In situations where an infant is a carrier for sickle cell or cystic fibrosis, it would be important to discuss parental carrier screening and the possibility of having a child affected with disease if both parents are found to be carriers. These results also indicate pediatricians are unfamiliar with the ACTion sheets developed by the American College of Medical Genetics to provide a step by step process of how to manage an abnormal screening result for each condition on the panel (Kemper, 2006). Current ACTion sheets explicitly state that newborns with sickle cell trait should be offered genetic counseling (Kemper, 2006).

3.2.3 QUALITATIVE ANALYSIS OF PARENTAL UNDERSTANDING OF NEWBORN SCREENING

A qualitative analysis of parental understanding of newborn screening and the informed consent process was conducted in the United Kingdom in 2005. A total of 47 parents who had participated in the newborn screening program were interviewed over the telephone or attended a focus group. Their responses were classified within three categories: information for parents, degree of parental choice, and recording parents' decisions. A majority of parents stated they did not recall receiving information about newborn screening before the blood sample was obtained but admitted that the time following the birth was chaotic. The parent may not have been focusing on the informed consent process. Parents also stated they did not feel there was an emphasis placed on the seriousness of the possible screening results during the communication exchange with a physician or nurse. However, the majority opinion was that it is unnecessary to provide detailed information about the genetic conditions prior to the test results. This information can overload a new parent and cause needless worry. Participants felt newborn screening should be routine, and the process of informed consent would provide justification for screening and decrease the number of parental refusals. Parents felt that verbal consent to screening was sufficient and written consent would increase the number of parental refusals (Hargreaves, 2005).

A 2003 study conducted twelve focus groups including 102 participants to examine the attitudes, beliefs, and concerns of parents regarding newborn screening. Participants included both Caucasian and African-American parents from in and around the Chicago, Illinois area. Specifically, seven focus groups were composed entirely of African-American participants. Focus group questioning centered on newborn screening for treatable (phenylketonuria) and untreatable (Duchenne muscular dystrophy) conditions. Researchers found very few individuals who could accurately describe newborn screening. In four separate focus groups, there were zero participants who could describe newborn screening. In each of three focus groups, one respondent was able to supply information regarding the type of conditions being screened for and the need to provide treatment. Participants expressed confusion over the difference between newborn screening and the testing of infants for drugs, alcohol, jaundice, and infection. Importantly, only one individual out of 102 participants mentioned newborn screening as a method to test for sickle cell disease. The majority of individuals felt newborn screening is beneficial and should be mandatory. The opinion that young, unknowledgeable parents may refuse screening and ultimately hurt their children through late detection of disease appeared throughout conversations. When asked to describe their concerns about newborn screening, participants were fearful that they would not be told the true purpose for which their child's sample would be used. Parents also expressed concern regarding using a child as a guinea pig and the government obtaining blood samples for cloning. An overarching theme among participants was the need for parental education throughout the newborn screening process. Many parents supported the idea of providing this education during the prenatal period (Campbell, 2003).

The newborn screening communication process was further explored in a 2006 qualitative study of newborn screening knowledge and awareness among parents and health care providers. This study recruited an ethnically diverse sample of 51 parents and 78 health care providers from Louisiana, New Mexico, and Maryland for 22 focus groups. Approximately 43% of participants were African-American. In support of previous studies, a majority of parents expressed a lack of knowledge about what newborn screening is and what it tests for. This study found similar confusion among parents about the difference between newborn screening and testing for environmental exposures. Very few parents were aware of when and how they would be contacted about results of screening, and many did not know retesting was a possibility. This study found that parents were not concerned about the consent process and felt screening is mandatory so consent is not necessary. Parents desired information about newborn screening in the third trimester from their health care provider instead of after the delivery. They requested a pamphlet of reading material to add to and reinforce the communication process. For these parents, the essential information provided to parents should be brief and include the mandatory nature of screening, the benefits of screening, the potential for necessary retesting, and how/when parents would be contacted (Davis, 2006). A 2006 qualitative study of the communication process between health care providers and parents on the topic of the newborn hearing screen supported these findings and identified a need to begin the communication process before birth and to provide supplemental reading material for parents to return to for more information (Arnold, 2006, "Infant hearing").

3.2.4 NEWBORN SCREENING FOR SICKLE CELL DISEASE

The first statewide newborn screening program for sickle cell disease was initiated in New York in 1975. In subsequent years, other states followed due to the availability of government funding through the Congressional National Sickle Cell Anemia Control Act. This legislation provided funding for sickle cell screening of all children under the age of 7 and those individuals in their reproductive years. However, it was not until 1986 that studies were able to show the benefit of taking prophylactic penicillin to prevent infection and early death in children with sickle cell disease. This direct evidence of how early identification of affected children could allow for beneficial treatment and decreased morbidity and mortality caused greater acceptance of hemoglobinopathy screening in other states. Pennsylvania adopted newborn screening for sickle cell in 1990. By 1996, the American Academy of Pediatrics recommended that every state implement targeted newborn screening for hemoglobinopathies for at-risk newborns (Olney, 2000). Currently, all states screen for hemoglobinopathies.

Newborn screening for hemoglobinopathies including sickle cell disease is performed by isoelectric focusing in the majority of screening programs. There are some programs that use high performance liquid chromatography as well. If a result is not in the normal range, testing of that sample is repeated using the blood specimen from the original Guthrie card. Confirmatory diagnostic testing must follow an abnormal screening result and should be completed before the infant is six weeks of age. The diagnostic testing procedure is described in Table 1. In those with sickle cell disease, prophylactic penicillin must be administered by two to three months of age (Pass, 2000).

Table 1. Diagnostic Testing for Sickle Cell Disease (Pass, 2000)

SICKLE CELL DISEASE (FS, FSC, FSA, FSOther)	DIAGNOSTIC EVALUATION
Core Tests and Procedures	Hemoglobin separation (by at least 2 complementary methods) Isoelectric focusing HPLC Cellulose acetate electrophoresis Citrate agar electrophoresis Serial CBC, MCV, reticulocyte counts
Supplemental Tests and Procedures	Examination of peripheral blood smear Family studies: Hb separation, CBC, MCV, Hb A_2, Hb F and/or DNA analysis on parents DNA analysis of β globin genes Hb A_2 (column chromatography or HPLC) Hb F (alkali denaturation, RID, or HPLC) Cellular distribution of Hb F (e.g, Kleihauer-Betke or immunofluorescence)

3.2.5 KNOWLEDGE AND PERCEPTION OF SICKLE CELL AMONG AFRICAN-AMERICANS

A limited number of studies have been conducted to examine African-Americans' knowledge and perception of sickle cell disease, sickle cell trait, carrier detection, and newborn screening. Past research has demonstrated a significant lack of awareness regarding the difference between disease and trait status and how trait status increases the chance to have a child with disease (Midence, 1994; Treadwell, 2006; Wright, 1994). Specifically, one study of 147 African-Americans between 18-50 years of age found that 31% knew if they were carriers for sickle cell trait (Wright, 1994). A majority (73%) did identify that sickle cell is a genetic disease (Wright, 1994). Misconceptions of how one inherits sickle cell disease have also been documented among the African-American community. Interestingly, a study of individuals with sickle cell disease found that 23% believed sickle cell trait could turn into sickle cell disease (Midence, 1994).

A 2006 study was undertaken to look more closely at African-American community members' knowledge and misconceptions of sickle cell (Treadwell, 2006). Three focus groups and 282 surveys were conducted in a metropolitan area of California (Treadwell, 2006). Focus group participants were asked to identify barriers to follow-up counseling for sickle cell trait detected by newborn screening, to describe their understanding of sickle cell disease, and to suggest possible solutions for poor follow-up rates for sickle cell trait counseling (Treadwell, 2006). Common themes among community members who participated in these focus groups included limited visibility of sickle cell disease and trait and the need to use media to promote awareness (Treadwell, 2006). Participants discussed the stigma associated with any type of disease in the African-American community and the need for health care professionals to have compassion and cultural sensitivity when discussing sickle cell (Treadwell, 2006).

Surveys were designed to determine where individuals receive their information about sickle cell, what individuals currently know about sickle cell disease and trait, if individuals know their trait status, and the effectiveness of different sources of information in improving knowledge about sickle cell (Treadwell, 2006). Among respondents, 86% could identify that sickle cell causes serious health problems, and 91% stated that sickle cell disease is most prevalent among African-Americans (Treadwell, 2006). Approximately 86% of respondents knew that sickle cell disease is inherited from both parents, and slightly fewer respondents were able to correctly identify the reproductive risks of a sickle cell trait carrier (Treadwell, 2006). Eighty-one percent of individuals believed if you have sickle cell trait you have a chance to have a child with disease, and 78% of individuals believed a child with sickle cell trait would be at risk to have a child with disease in the future (Treadwell, 2006). Of survey participants, only 15.9% knew their trait status and of those individuals, 53% learned their trait status through discussion with family members (Treadwell, 2006). The greatest majority of individuals receive their information about sickle cell disease and trait from friends and acquaintances (Treadwell, 2006). Respondents who received information from friends and family were three times more likely to know their trait status (Treadwell, 2006). These findings emphasize the benefit of family discussion about sickle cell and the risk within a family.

3.2.6 IMPACT OF GENETIC COUNSELING FOR SICKLE CELL DISEASE

Genetic counseling for sickle cell disease and trait can occur in a variety of situations. Couples or individuals may be referred to a genetic counselor to discuss carrier screening to learn of their trait status before pregnancy occurs. Others may be referred for prenatal genetic counseling after a pregnancy has occurred to perform parental carrier screening and/or prenatal diagnosis through chorionic villus sampling or amniocentesis. For many individuals or families, the first encounter with genetic counseling for sickle cell comes after an abnormal newborn screen result. This result often requires follow-up testing to determine whether a child has sickle cell disease or trait. Genetic counseling can provide families with information about genetic testing for parents and additional family members, the risk of recurrence for sickle cell disease and/or trait, the clinical picture of sickle cell disease, and information about resources and support organizations for the family.

Research examining the impact of genetic counseling for sickle cell on the reproductive decisions of couples found that risk information did not have a significant impact on the number of affected children born to these couples. This study followed 35 couples at risk to have a child with sickle cell disease. The couples received genetic counseling after each member of the couple was found to be a carrier of sickle cell trait. Of these couples, 40% did not believe the information would have an impact on their childbearing plans while 37% believed the information would influence reproductive decisions. Of these 35 couples, 25 couples were able to be followed for between 1 and 120 months. The study found that these 25 couples had 13 children with sickle cell disease prior to receiving counseling and 10 children with sickle cell disease after genetic counseling occurred. The authors attribute this lack of significant decrease in the number of sickle cell disease births to the relative ineffectiveness of family planning intentions and the intense desire to have children despite knowing the risks for disease (Neal-Cooper, 1988).

Studies have also been conducted to examine how to improve the uptake of genetic counseling following an abnormal newborn screen result for sickle cell trait and ultimately, increase parental knowledge about risk for sickle disease in the family. Prenatal genetic counseling for sickle cell disease and trait has been shown to improve the uptake of genetic counseling following an abnormal newborn screen result for sickle cell trait. Yang et al demonstrated that women who received a brief description of sickle cell disease in the prenatal period were more likely to attend follow-up genetic counseling for identification of sickle cell trait on newborn screening in comparison to women who did not receive prenatal education (2000). However, prenatal education did not reduce anxiety levels significantly among women who received an abnormal newborn screen result (Yang, 2000).

A study conducted at the University of Pittsburgh found that implementing an intensive follow-up protocol for infants with an abnormal hemoglobinopathy trait identified by newborn screening increased the acceptance rate of genetic counseling (Kladny, 2005). Follow-up protocol included letters, telephone calls, educational videos, and the possibility of providing genetic counseling over the phone or in person (Kladny, 2005). Of the 52% of families who were reached by telephone, 92% accepted genetic counseling over the phone and 12% scheduled an appointment for genetic counseling in person (Kladny, 2005). Prior to the implementation of the follow-up program, only 5.3% of those families reached by telephone received genetic counseling at that time (Kladny, 2005). These strategies improve the uptake of genetic counseling services for families who receive an abnormal newborn screening result for sickle cell trait. However, there still remains a significant portion of families and parents who do not receive information about the inheritance of sickle cell disease.

3.3 AFRICAN-AMERICANS AND GENETIC TESTING

3.3.1 AFRICAN-AMERICANS' ATTITUDES TOWARD GENETIC TESTING

African-Americans' attitudes and perceptions of genetic testing for the purpose of clinical diagnosis and management has been examined among individuals in clinical, educational, and community settings. Despite the location from which subjects are recruited, participants articulate both benefits and risks of genetic testing. Participants frequently express concern for possible misuse of information gathered from genetic testing but also identified the benefit of using genetic test results for prevention (Catz, 2005; Kessler, 2005; Laskey, 2003).

In a study of attitudes toward genetics among minority populations, eight focus groups were conducted with 55 individuals recruited from community health centers (Catz, 2005). Of these participants, fifteen were African-American and composed two focus groups (Catz, 2005). The greatest percentage (35%) of minority participants could not provide any information when asked what they had heard about genetic testing (Catz, 2005). When asked more specifically about newborn screening, 45% were not aware that it is routinely performed (Catz, 2005). Minority participants stated the main benefit of genetic testing is that it can lead to prevention or preparation for disease (Catz, 2005). However, participants also mentioned concerns for the unethical use of testing, anxiety from test results, and false reassurance from negative results (Catz, 2005). Newborn screening was viewed as a method to prepare for a child's health problems (Catz, 2005). However, African-Americans and other minority participants expressed practical concerns for the outcome of genetic testing through newborn screening including concerns about insurability, financial burdens, parenting responsibilities for a sick child, and the potential to test positive for a disease with no cure (Catz, 2005).

A study by Zimmerman et al also examined African-Americans' attitudes toward genetics in comparison to those of Caucasians through surveys in the community setting (2006). Results demonstrated a majority of both African-Americans and Caucasians believe that sickle cell disease is a genetic condition (Zimmerman, 2006). In comparison to Caucasians, African-Americans were more likely to believe genetic testing will lead to racial discrimination (Zimmerman, 2006). Additionally, African-Americans were more likely than Caucasians to believe that all pregnant women should have prenatal genetic tests (Zimmerman, 2006).

In contrast to the Catz et al. study which examined attitudes toward genetics among minorities with a range of educational levels, a study by Laskey et al. pursued similar questioning among African-American college students in a premedical program. These undergraduates also stated that genetic testing could benefit individuals/families by allowing for prevention or preparation (Laskey, 2003). However, the group's overall concerns about the risks of genetic testing were more theoretical in comparison to the responses in the Catz et al study. Concerns about genetic testing included genetic discrimination, increasing abortion rates, eugenics, and breach of confidentiality (Laskey, 2003).

3.4 QUALITATIVE RESEARCH

3.4.1 QUALITATIVE RESEARCH METHODS

Some qualitative research methods attempt to provide a comprehensive description of an event in everyday terms of those events (Sandelowski, 2000). In contrast to quantitative methods, qualitative research is often a more interpretive approach where knowledge is gained from observation (Beeson, 1997; Sandelowski, 2000). Quantitative analysis uses descriptive statistics to summarize a finding but this form of analysis is limited by the pre-selection of variables to be studied which are based on the assumptions of the researchers (Sandelowski, 2000). Qualitative research opens the door for the unanticipated responses of participants by allowing for free description instead of confining individuals to items on a survey (Sandelowski, 2000). Beeson describes the depth of qualitative research method when she states, "…Instead of attempting to prove or refute a hypothesis, or posing alternatives and offering to discover which one is most accurate, you formulate your research question in such a way that you can reveal whatever processes are actually occurring. Your research question should reflect this goal by asking not simply what people do, but why they do it, or how they make sense of a particular situation- what it means to them" (1997). This process allows researchers to find facts and the meanings participants attach to those facts (Sandelowski, 2000).

Qualitative research methods are often divided into two frameworks: dependent on a theoretical position and independent of a theoretical position (Braun, 2006). The types of analyses which are tied to a theoretical position may have limited variability in how they are performed (Braun, 2006). These include interpretative phenomenological analysis, conversation analysis, grounded theory, discourse analysis, and narrative analysis (Braun, 2006). Thematic analysis is a qualitative method independent of theory, and it is this flexibility which provides for the wide range of use for this research tool (Braun, 2006).

3.4.2 THEMATIC ANALYSIS

Thematic analysis is a qualitative research method for identifying, analyzing, and reporting the patterns or themes within the data collected (Braun, 2006). This method is most often used when the research project is attempting to examine a topic where little is known or where the views of participants on a topic have not been well documented (Braun, 2006). Thematic analysis provides room for the unanticipated responses of participants. The established themes of this analysis are crucial to obtaining a rich description of the data. A theme is defined as capturing or representing a patterned response or meaning within the data (Braun, 2006). However, it is important not to fall into the trap of believing repetition of a theme means it is more critical to understanding the topic than other themes. There is no established rule stating that an idea has to be displayed in a certain percentage of the data for it to be considered a theme (Braun, 2006). However, it is essential for a theme to capture something important to answering the overall research question (Braun, 2006). The researcher must take a very active role when analyzing the data according to thematic analysis so as to identify patterns or themes, select those of interest, and report them accurately (Braun, 2006).

Before embarking on thematic analysis, researchers must decide their approach to coding and the level at which themes will be identified. Thematic analysis is often conducted in either an inductive or deductive manner. If an inductive approach is performed, then data is coded without trying to adhere to a pre-existing coding system (Braun, 2006). The preconceptions of the researcher do not play a strong role in this form of analysis and themes are created based on what is found in the data (Braun, 2006). In contrast, a deductive approach focuses on coding according to currently existing theories or previous analyses on the research question (Braun, 2006). In general, this type of analysis may not capture the overall picture or description provided by participants (Braun, 2006). After an approach to coding has been determined, it is important to assess whether or not it will be appropriate to identify themes at an explicit or interpretative level (Braun, 2006). Explicit themes capture the surface meaning of what a subject or participant has written or said while interpretative themes attempt to examine the underlying ideas, assumptions, and conceptualizations shaping the surface theme (Braun, 2006). When an approach and level to coding has been determined, the process of data analysis can begin. This process often consists of phases including becoming familiar with the data, generating initial codes, searching for themes, reviewing themes, defining and naming themes, and producing the final report (Braun, 2006). At the conclusion, the goal of this approach is to tell an overall story of the data (Braun, 2006).

Table 2. Phases of Thematic Analysis (Braun, 2006)

Phase	Description of the process
1. Familiarizing yourself with your data:	Transcribing data (if necessary), reading and re-reading the data, noting down initial ideas.
2. Generating initial codes:	Coding interesting features of the data in a systematic fashion across the entire data set, collating data relevant to each code.
3. Searching for themes:	Collating codes into potential themes, gathering all data relevant to each potential theme.
4. Reviewing themes:	Checking if the themes work in relation to the coded extracts (Level 1) and the entire data set (Level 2), generating a thematic 'map' of the analysis.
5. Defining and naming themes:	Ongoing analysis to refine the specifics of each theme, and the overall story the analysis tells, generating clear definitions and names for each theme.
6. Producing the report:	The final opportunity for analysis. Selection of vivid, compelling extract examples, final analysis of selected extracts, relating back of the analysis to the research question and literature, producing a scholarly report of the analysis.

4.0 MATERIALS AND METHODS

The study design was approved by the University of Pittsburgh's Institutional Review Board (IRB) on November 16, 2006 (Replications of IRB Approval letters for protocol # 0610018 can be found in Appendix A).

4.1.1 PARTICIPANT RECRUITMENT

This study was designed to recruit African-Americans from Pittsburgh, Pennsylvania who are members of the Healthy Black Family Project (HBFP), a community based health promotion and disease prevention program sponsored by the Center for Minority Health of the University of Pittsburgh. Both males and females over the age of 18 years were recruited. Individuals who are members of the HBFP typically reside in the predominately African-American communities of Pittsburgh. These communities are defined as the Health Empowerment Zone (See Table 3 and Figure 3) by the Center for Minority Health. The HBFP is strategically housed within this zone at the Kingsley Association. The Kingsley Association is a community recreation and meeting center located in East Liberty, Pennsylvania which supports the efforts of the HBFP and permits use of its facilities for physical fitness classes, community discussions, and support group meetings.

Table 3. Health Empowerment Zone

Zip Code	Neighborhood
15147	Penn Hills
15206	Lincoln, Lemington, Belmar, East Liberty, Larimer, Garfield
15207	Glen Hills
15208	Point Breeze North, Homewood South, Homewood North, Homewood West
15213	Terrace Village, Upper Hill
15219	Crawford Roberts, Terrace Village, Middle Hill, Bedford Dwellings, Upper Hill
15221	Homewood North, East Hills, Wilkinsburg
15224	Garfield

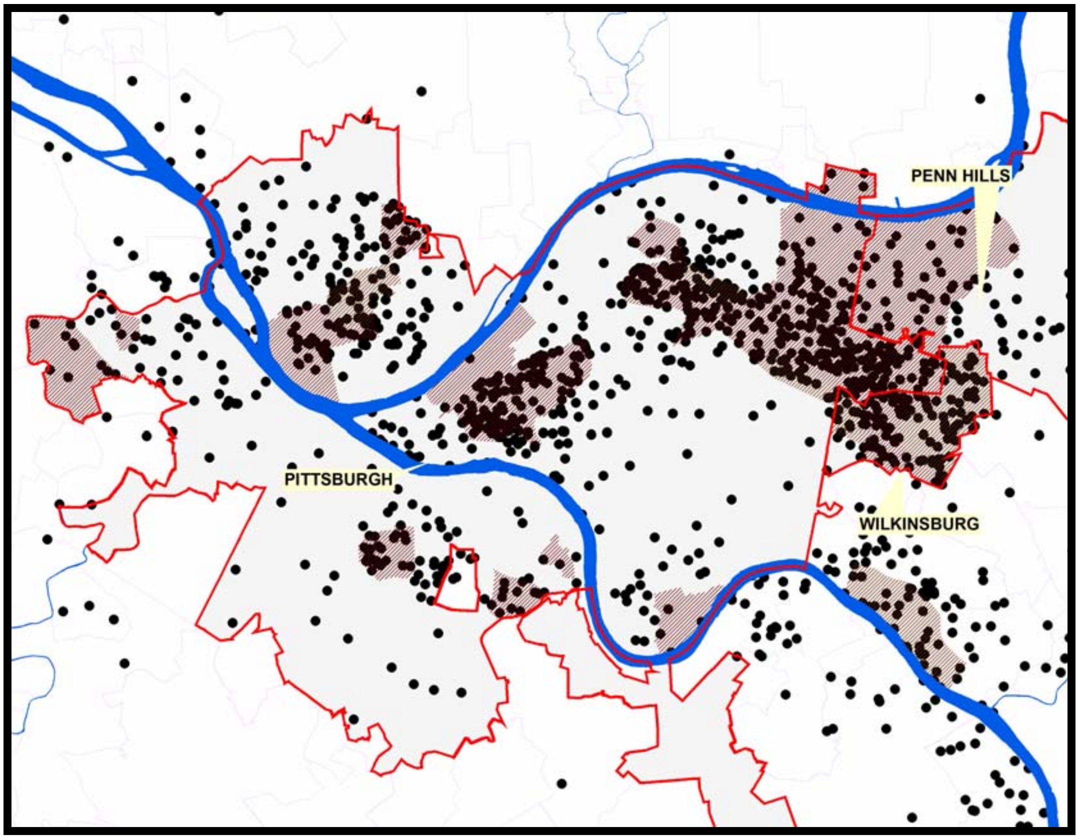

Figure 3. Health Empowerment Zone Map (1 dot = 100 African-Americans)

Subjects recruited can be divided into two categories: individuals who are members of the Healthy Black Family Project but not the Minority Research Recruitment Database and individuals who are members of both the HBFP and the Minority Research Recruitment Database. The Minority Research Recruitment Database is a feature of the HBFP which is offered to individuals who complete their family health history with a genetic counseling student. It was developed as an effort to increase African-American and minority participation in research. The database is composed of names, addresses, telephone numbers, and personal and family health information of those individuals who would like to be contacted in the future for research studies. Participants provide written informed consent to be included in the database. For those individuals who are members of the HBFP but not the Minority Research Recruitment Database, contact was made for this study through flyers posted at the Kingsley Association and advertisements in the HBFP monthly mailings (See Appendix B). The flyer stated the topic of genetics but did not specifically mention sickle cell or newborn screening. Individuals who saw a flyer then contacted the researchers to sign up for a focus group. For those individuals who are members of both the HBFP and the Minority Research Recruitment Database, contact was made by telephone and the study was presented according to a telephone script (See Appendix B). Telephone calls were made at several times of day on both weekends and weekdays to be as inclusive as possible. At the time of recruitment, participants were also notified that focus groups would require 2 hours of time, and they would receive refreshments and a $25 gift card to Giant Eagle, a grocery store located in Pittsburgh, for their commitment. After signing-up for a focus group, each participant received a reminder letter in the mail (See Appendix B) and a follow-up phone call the day before his/her scheduled focus group.

4.1.2 FOCUS GROUPS

Four focus groups were conducted over a period of two months in December and January of 2006-2007 at the Kingsley Association. The Kingsley Association was selected as the location for the focus groups because it is a familiar building for HBFP participants and is closely located to where a majority of HBFP participants reside. For each focus group between 10 and 15 individuals were recruited with an expectation that due to cancellation or no show approximately 10 individuals would attend the group. The four focus groups were conducted at a variety of times and days of the week in order to gather as many participants as possible. The groups were held on two Saturdays at 2 PM, Wednesday at 6 PM, and Thursday at 6 PM. The groups consisted of 11, 6, 10 and 8 participants for a total of 35. Each group was held in a community room of the Kingsley Association, and participants sat together at a common table for discussion.

Audio recordings of the focus groups were collected for later transcription and notes of the discussion were taken by an assistant to the moderator. Each focus group began with participants choosing an alias to use during discussion and then completing a pre-discussion survey. Focus groups were conducted according to a moderator's guide found in Appendix C.

4.1.3 SURVEY

Each participant was asked to complete a survey designed to characterize demographics and experience with sickle cell trait and disease (See Appendix D). The survey was designed to take approximately 10 minutes to complete and consisted of 19 questions. The survey is separated into two sections. The first section focuses on demographic questions and information about genetics knowledge, personal health, and health insurance status. The second section is directed at the participant's familiarity with sickle cell disease and trait and knowledge of personal trait status and trait status within his/her own family. Information gathered from the administration of this survey was analyzed using Microsoft Excel.

4.1.4 TRANSCRIPTION

The audio tapes for each focus group were transcribed word-for-word by a transcriptionist employed by the Center for Minority Health. Microsoft Word was used to manage the four transcripts. Transcripts adhered to participants' grammar, pauses, place holders, and unfinished sentences but have been edited in some cases to be more understandable by the reader.

4.1.5 QSR NUDIST VIVO® AND CODING

The QSR NUDIST VIVO® software is a program designed to manage and facilitate analysis of qualitative data. NUDIST is an acronym for Non-numerical Unstructured Data Indexing, Searching, and Theorizing. The NVIVO program allows transcripts or other qualitative data to be uploaded into the software where the researcher can then store and analyze the data. The program is designed to permit the researcher to develop his/her own codes and apply those throughout multiple transcripts in one project. These codes or nodes can be further broken down and linked by node trees in order to generate a thematic map of the data. The software allows the researcher to capture all direct quotations labeled by a particular node from across all transcripts. This feature is helpful for comparison of themes during analysis.

Before applying the NVIVO program to the transcripts of this study, each transcript was read twice by the researcher to gain familiarity with the data and the audio tapes were listened to once while reading each transcript to ensure reliability. As the transcripts were read, the researcher wrote down notes of potential themes and other features of the data. After performing the background research of this study and reading the transcripts, it was determined that the inductive approach to thematic analysis would best capture the responses of this population and allow for the unanticipated. The use of interpretative themes is most fitting for the research questions of this project in the attempt to understand the underlying ideas, attitudes, and assumptions which shape African-Americans' views of sickle cell. However, explicit themes are necessary for some topics.

The transcripts were uploaded to NVIVO and codes were generated to reflect the participants' views and opinions according to thematic analysis. Codes were revised throughout the process in order to better refine and capture the beliefs of participants. Some passages of the transcript required only a single code while other passages required multiple codes due to the richness of the information. The NVIVO software then allowed comparison of codes across transcripts to create a theme which summarizes the views of multiple individuals.

5.0 RESULTS

5.1 SURVEY RESULTS

5.1.1 DEMOGRAPHICS

A total of 35 individuals participated in the focus groups and completed the pre-discussion surveys. The average age of participants was 53 years with a median age of 57 years and range of 26-77 years. Of these individuals, 91% are female and 91% are African-American. The remaining 9% of individuals categorize their race as “Other”. Approximately 82% of participants have children. The majority of participants has an income between $20-35,000 (32%) and has an educational background of 1-3 years of college or technical school (40%). When asked about personal health, 57% indicated they are in good health, 54% believe they are overweight, and 17% are smokers. Approximately 97% have a form of health insurance, 57% have one personal physician, and 17% could not go to the doctor within the last year because of cost. (See Figures 4-13).

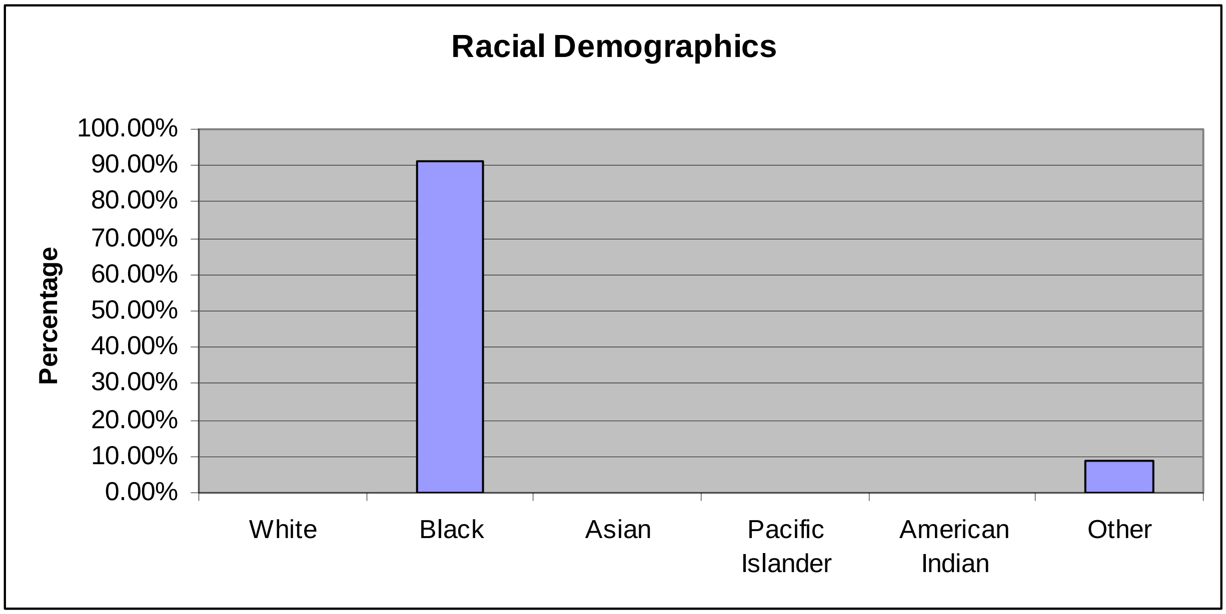

Figure 4. Racial Demographics of Focus Group Participants (N=35)

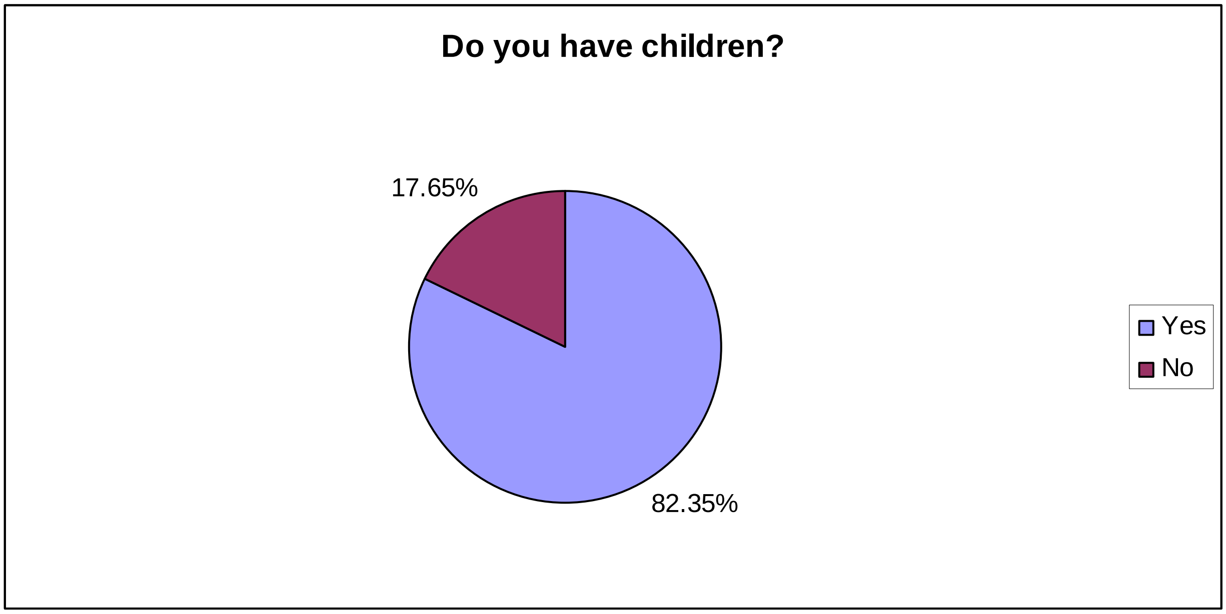

Figure 5. Percentage of Focus Group Participants with Children (N=34)

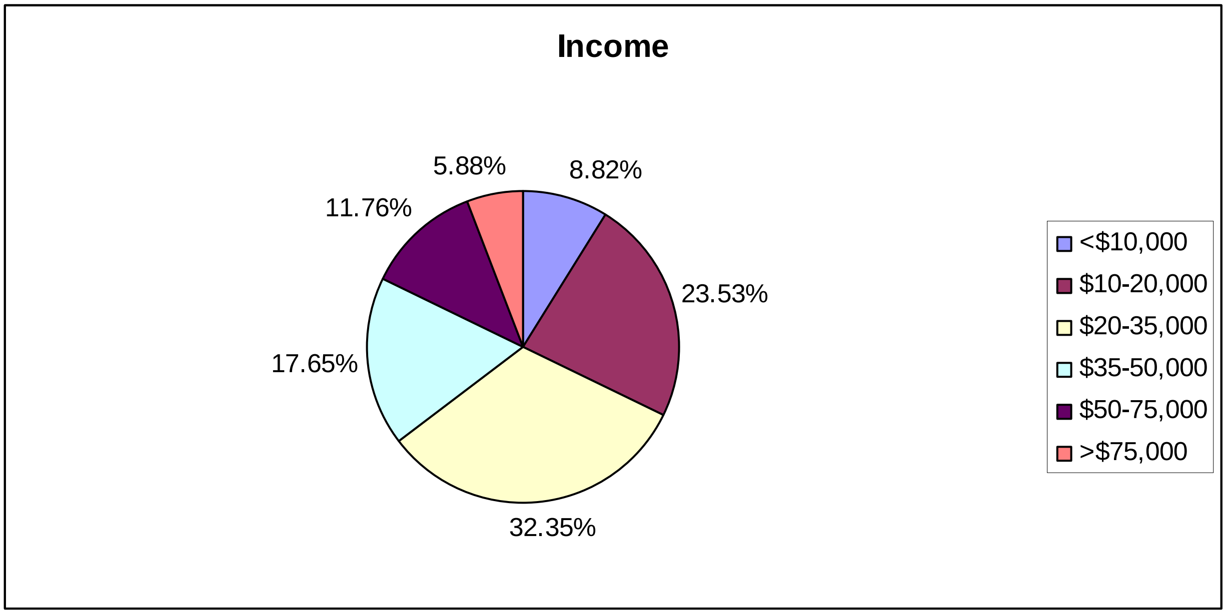

Figure 6. Income Levels of Focus Group Participants (N=34)

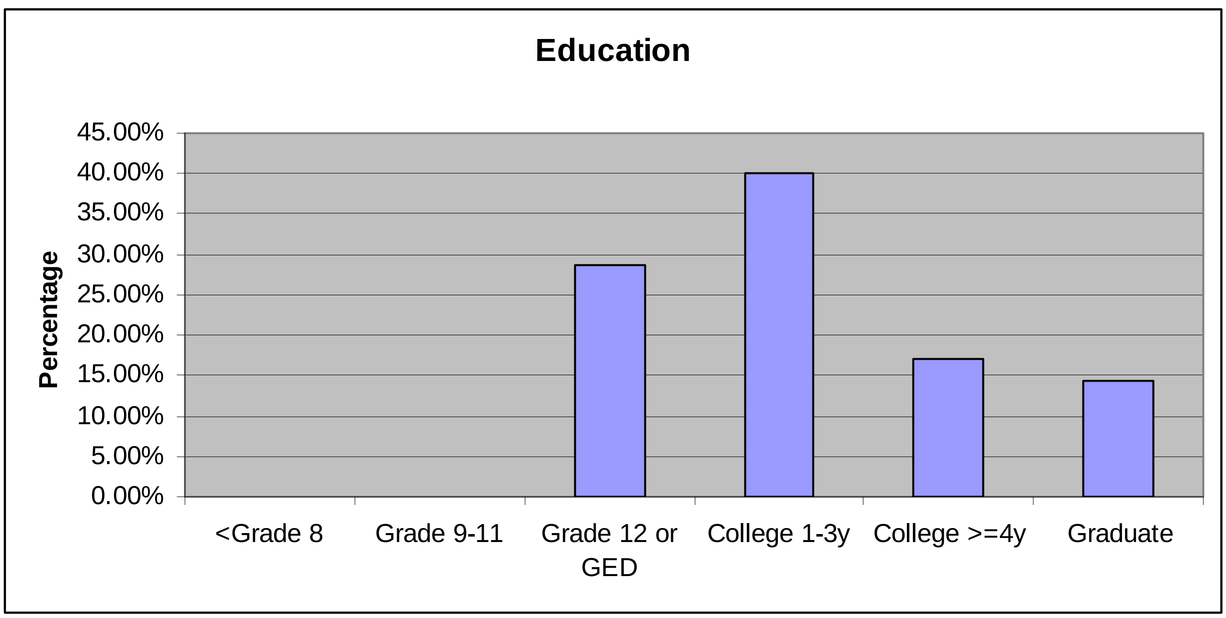

Figure 7. Education Level of Focus Group Participants (N=35)

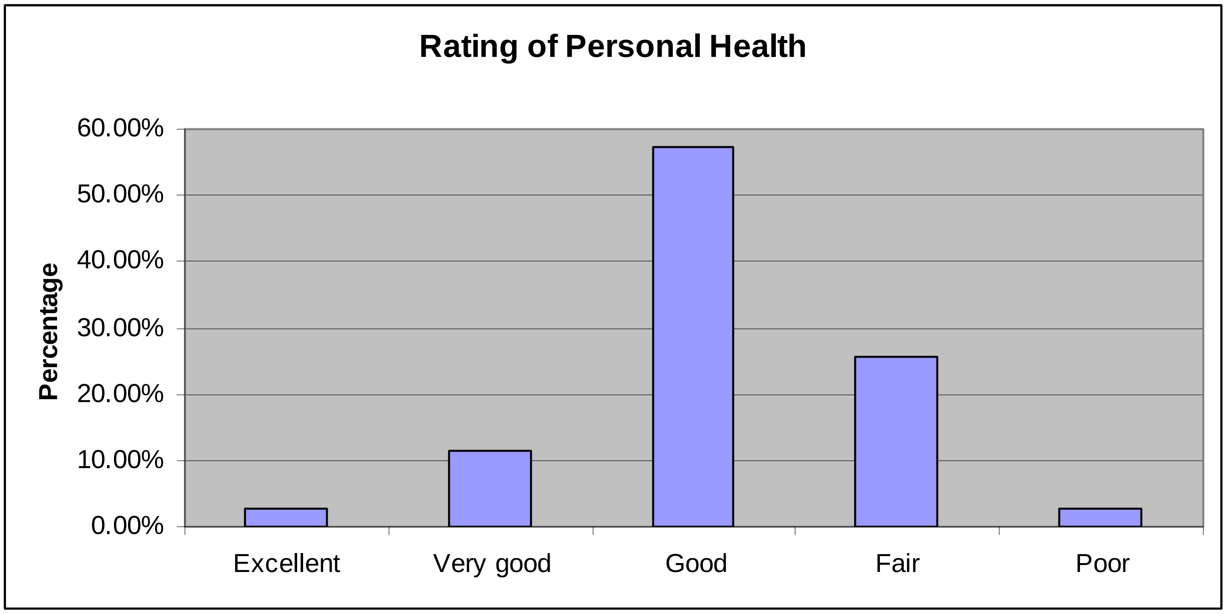

Figure 8. Rating of Personal Health among Focus Group Participants (N=35)

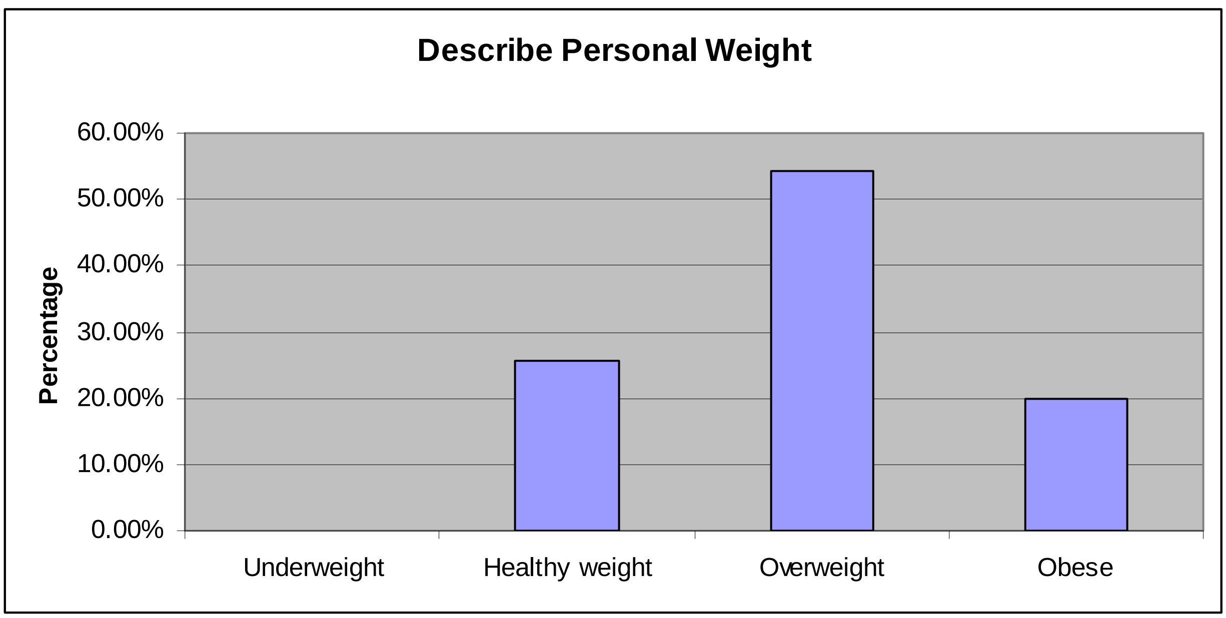

Figure 9. Description of Personal Weight among Focus Group Participants (N=35)

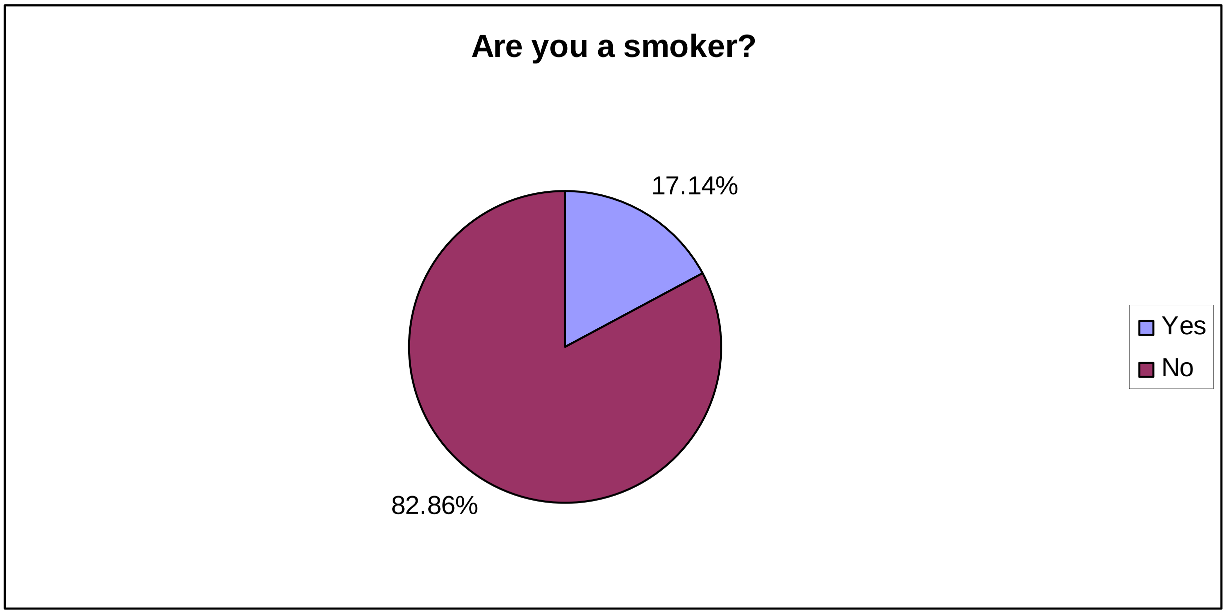

Figure 10. Smoking Status among Focus Group Participants (N=35)

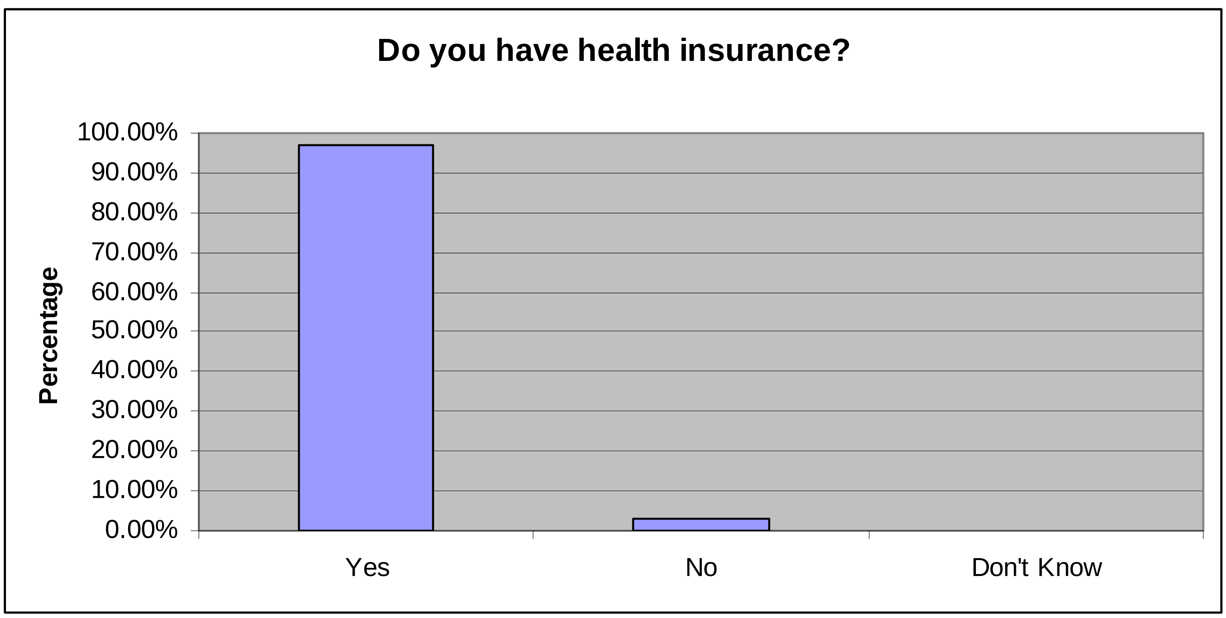

Figure 11. Health Insurance Status among Focus Group Participants (N=35)

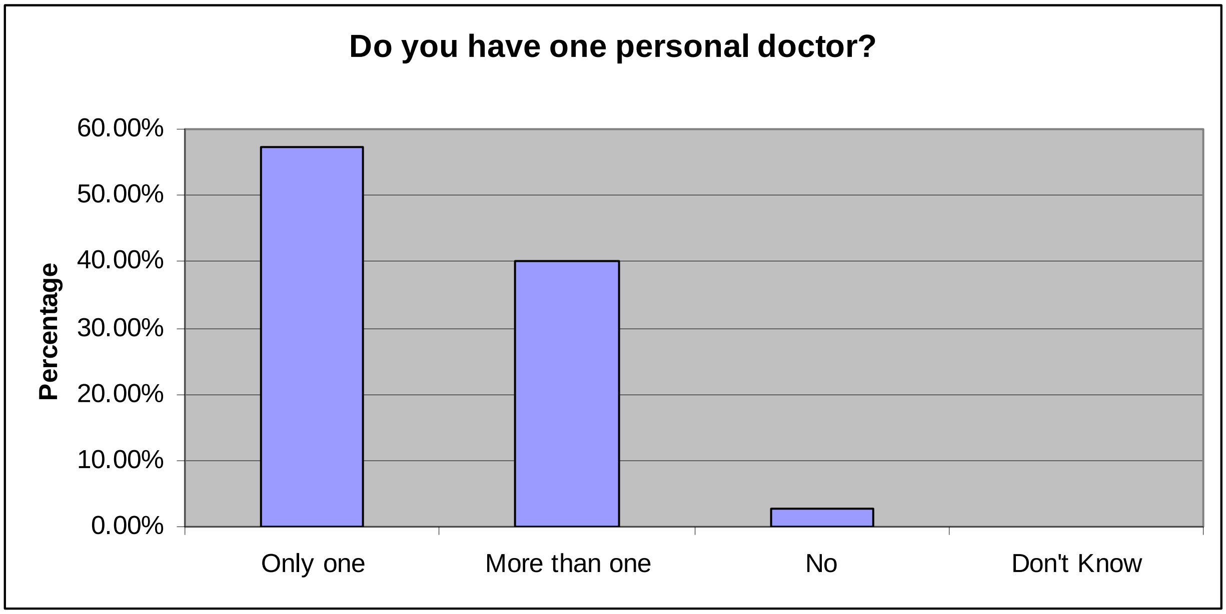

Figure 12. Number of Personal Physicians among Focus Group Participants (N=35)

Figure 13. Ability to Attend Physician Appointment Based on Cost among Focus Group Participants (N=35)

5.1.2 FAMILIARITY WITH GENETICS AND SICKLE CELL

A pre-discussion survey assessing personal rating of genetics knowledge and familiarity with sickle cell trait and sickle cell disease found that a majority of individuals believe they have a fair knowledge of genetics (47%) and a majority knows someone with sickle cell trait (64%) and someone with sickle cell disease (64%). Of those individuals who know someone with sickle cell trait or disease, 20% know an individual who was found to have trait or disease by newborn screening. (See Figures 14-17).

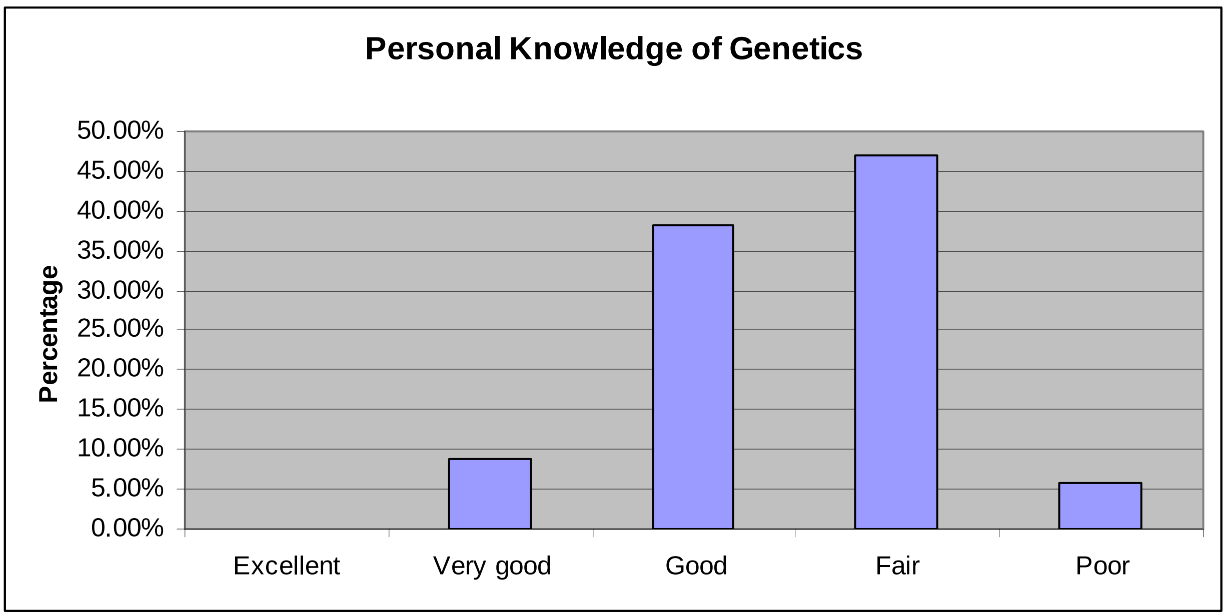

Figure 14. Rating of Personal Knowledge of Genetics among Focus Group Participants (N=34)

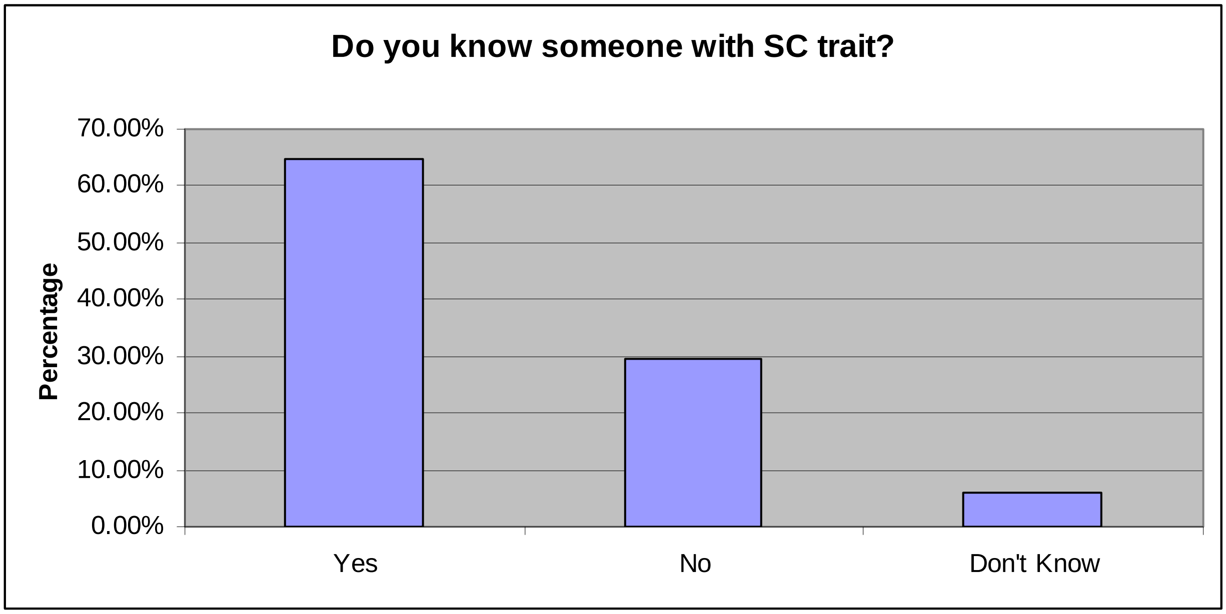

Figure 15. Knowledge of Someone with Sickle Cell Trait among Focus Group Participants (N=34)

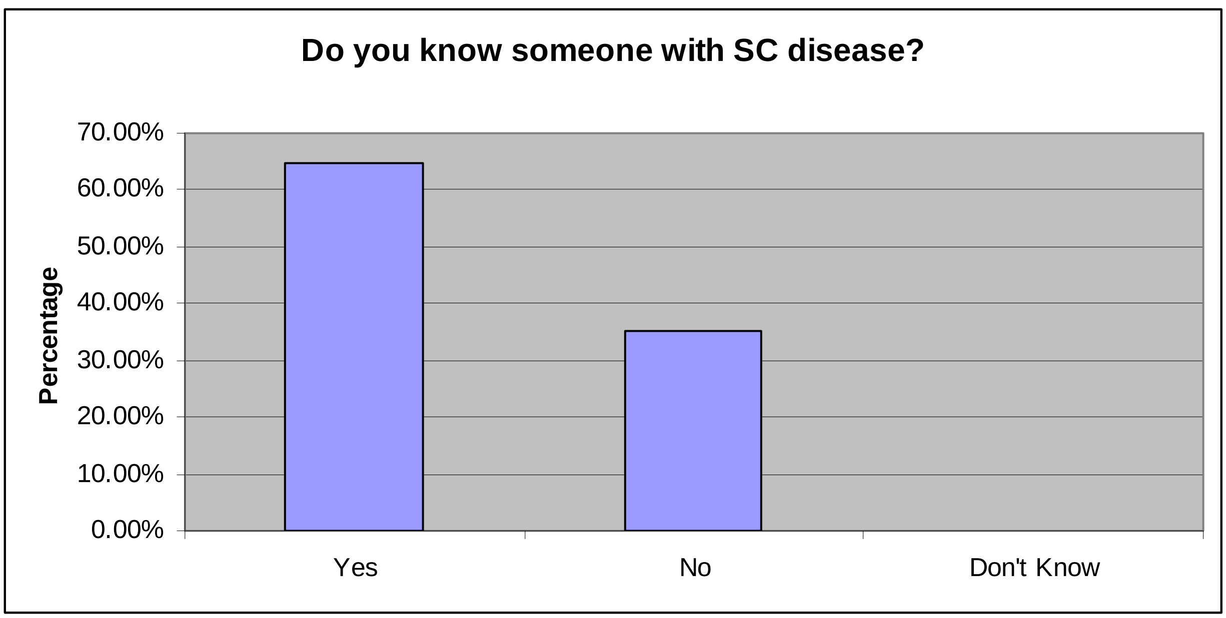

Figure 16. Knowledge of Someone with Sickle Cell Disease among Focus Group Participants (N=34)

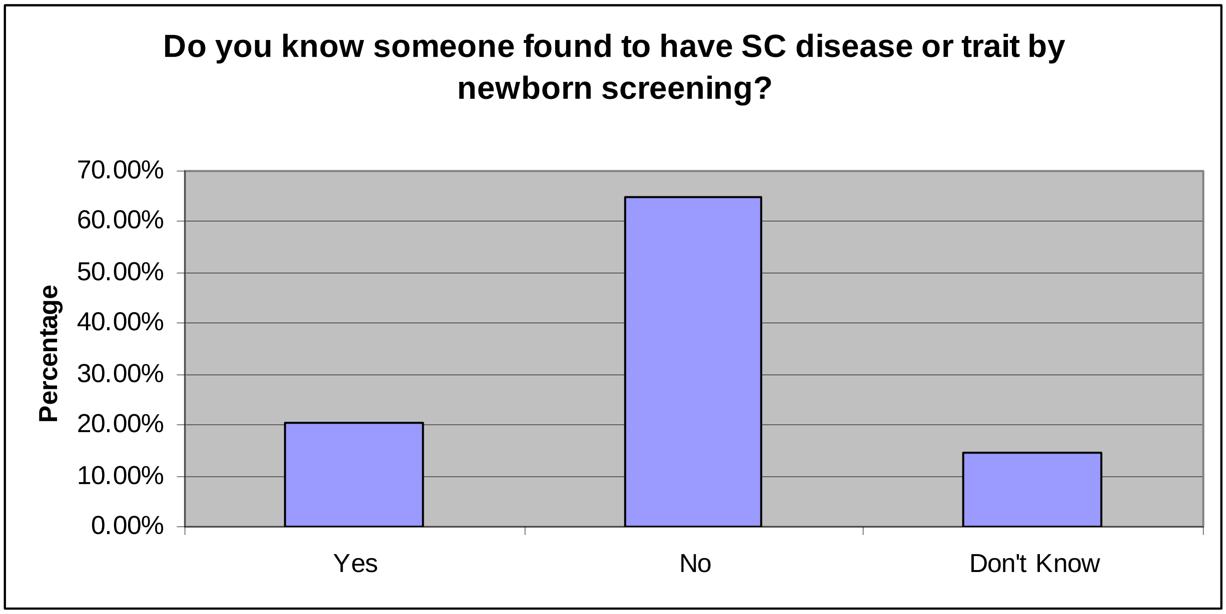

Figure 17. Knowledge of Someone with Sickle Cell Disease/Trait Detected by Newborn Screening (N=34)

5.1.3 KNOWLEDGE OF PERSONAL AND FAMILIAL SICKLE CELL TRAIT STATUS

The majority of participants do not know their own sickle cell trait status (51%). For those individuals with a spouse or partner, only 26% know that individual's sickle cell trait status. Among participants with children, only 40% know their child or children's sickle cell trait status. (See Figures 18-20).

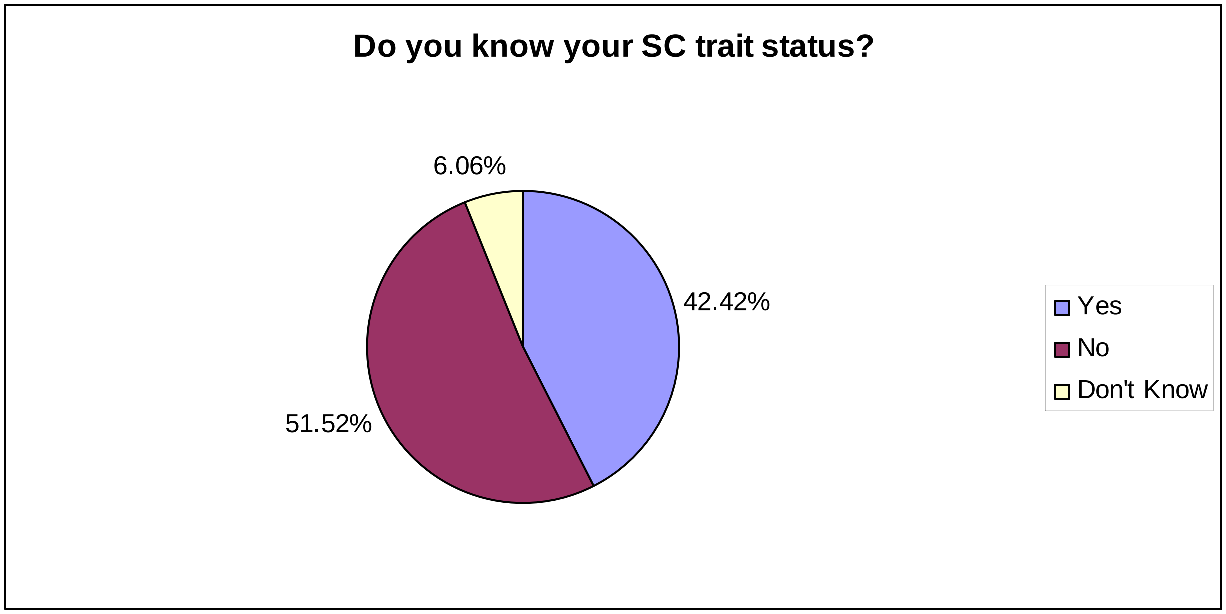

Figure 18. Knowledge of Personal Sickle Cell Trait Status among Focus Group Participants (N=33)

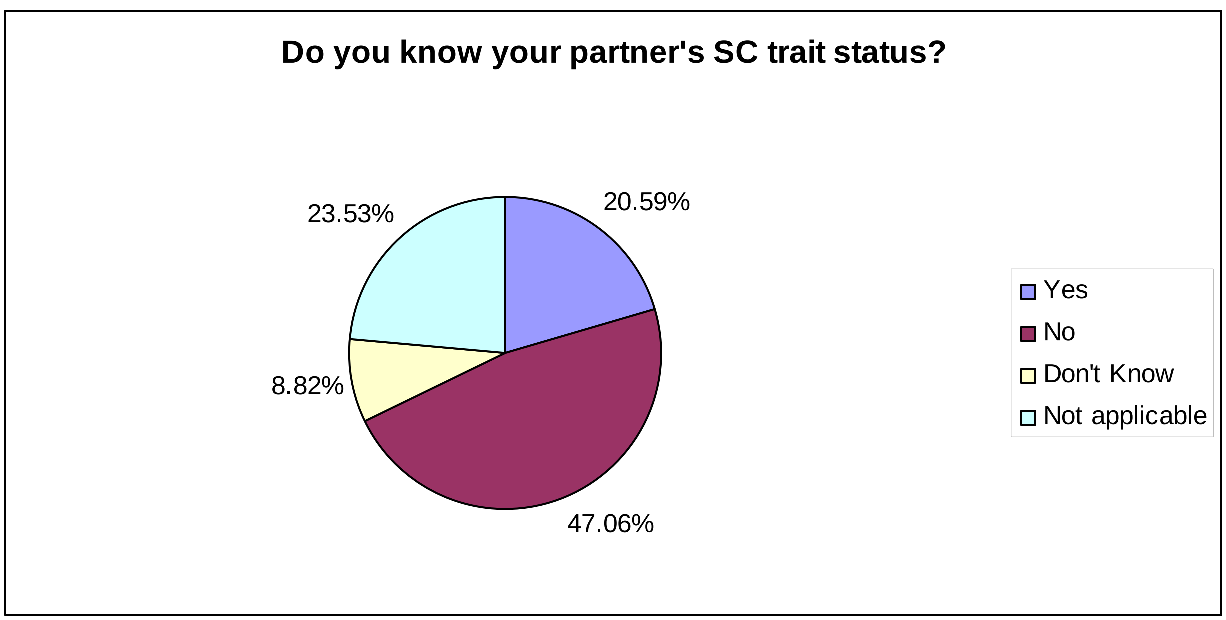

Figure 19. Knowledge of Spouse/Partner’s Sickle Cell Trait Status (N=34)

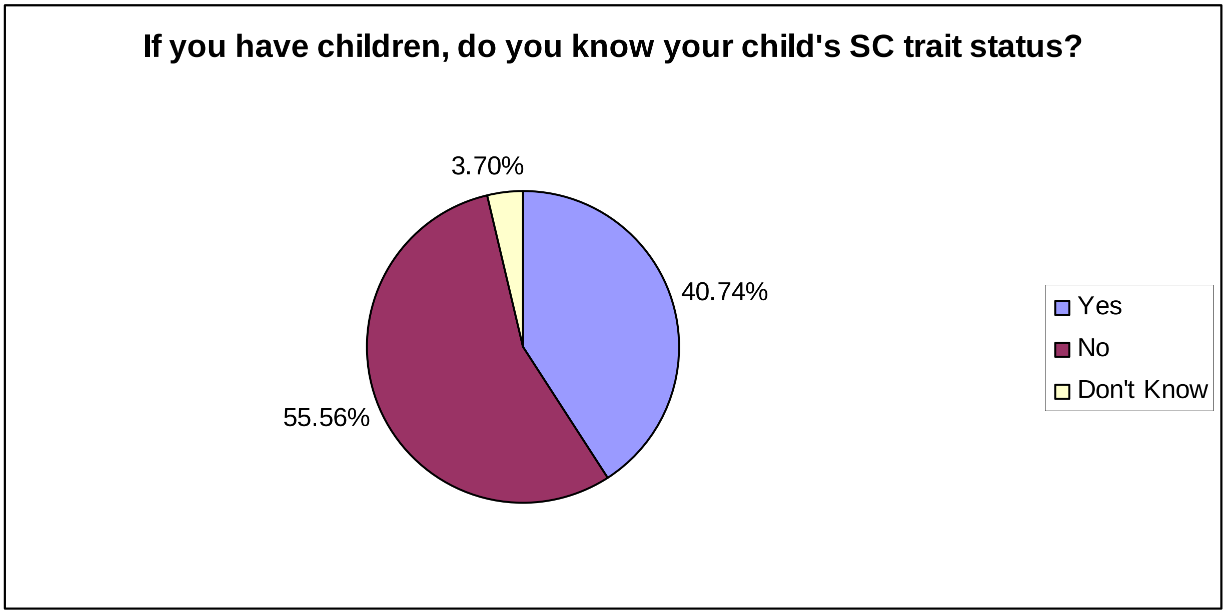

Figure 20. Knowledge of Child/Children's Sickle Cell Trait Status (N=27)

5.2 THEMATIC ANALYSIS

5.2.1 ASSESSING AFRICAN-AMERICANS' ATTITUDES AND BELIEFS ON GENETICS

Initial focus group discussion began with conversation about participants' impressions of genetics and what the word means to them. Through this conversation, participants identified their primary resources for genetics information as high school and college coursework, experiences on the job or as a participant in research studies, physicians, the media, and family and friends (Table 4). The educational background of focus group participants ranged from completion of high school to graduate school, and therefore, the exposure to biology and genetics course work varied for each individual. The majority of individuals who stated their educational background is a primary resource for genetics education also explained that they had advanced degrees or multiple courses in biology. One woman stated, “I have a degree in biology so I had a lot of courses in different areas.” Other individuals discussed on the job experience with genetics such as “I worked in a research lab…and we get lots of medical information.” Two individuals mentioned their participation in genetics research which provided them with first-hand experience and knowledge of genetics. For many individuals, the primary care physician provides genetics information and often the first conversation about the importance of family history. As a participant describes here, “A lot of times you go see doctors, and if you are a first time patient, they ask a lot of questions about the medical family history.” Another individual stated, “Especially when something goes wrong with you and they’ll come in and introduce themselves and bring up your background and it will come up that way.” The greatest number of participants discussed relying on information gathered from the media to understand genetics. Media influences included the internet, newspaper, magazines, and television programs including those for entertainment, health and science information, and news.

The theme of using family and friends as a primary resource for health/genetics information was highlighted over the course of focus group conversations. In general, participants valued a particular individual as a resource because of his/her association with the medical profession and a feeling of familiarity and trust. One woman describes her relationships with friends and family:

> I think that is true that the black community will have someone that works at a hospital or has been at a hospital for a long period of time and by that I mean my sister she’s 72 and we consider her the physician of the family. She has the PDR’s. She’s had a long time experience with different illnesses and various things. My girlfriend she’s worked at Allegheny General for 25 years when I want a specialist but that is the way I think it happens. I think in the black community too is that word of mouth.

Another participant describes how she relies on friendship to help understand health information:

> I have a friend who is a physician's assistant so every now and then we'll have like a woman session. If she's there but if that's not what we're talking about we just get together every now and then for fun but if there is anything we need to discuss we ask her.

The theme of the importance of familiarity with the health professional as a component of the information sharing process appears again during discussion of the dissemination of newborn screening information.

Throughout each focus group discussion, individuals were able to characterize what genetics means to them. During these moments of conversation, distinct themes emerged including genetics is inheritance, genetics is defined by technical terms, genetics is defined by the media, and genetics is influenced by the environment (Table 4). The majority of individuals situated their understanding of genetics within the context of inheritance or "traveling down the line from generation to generation." There were some individuals who expressed a feeling that genetics is inescapable and that the health of past relatives has an effect on the current individual. Participants stated that "Inheritance is something that you are born with." and "Your chromosomes, the X and Ys and then that determines certain elements (pause) if you have pre diagnosis history." In contrast, there were participants who discussed how genetics can be shaped by environmental influences such as nutrition or how a mother's health can impact the outcome of a pregnancy. For example, a woman described the connection between mother and baby, "Sometimes if the blood pressure is up they will put you on bed rest because it definitely will have an affect on you as well as the child." Many individuals used technical language such as "genes" and "DNA" to describe what genetics meant to them and made statements such as "When a child is born you get DNA from your parents, mother and father, and of course, your DNA is made up of all the past DNA and so you get half and half and you are who you are." Other individuals described genetics by using popular media stories such as actor Michael J. Fox and stem cell research or media icon Oprah Winfrey and tracing African heritage. A participant discussed how media shapes her view of genetics in the following passage:

> I've heard the word genetics used in trying to trace back our ancestors. Oprah was talking about it where she was trying to find as far back as she could reach with the testing of genetics seeing where her racial boundaries were and I found that to be quite fascinating.

The news and entertainment stories which participants chose to describe genetics had a great deal of attention in the media during the time period these focus groups were conducted.

Table 4. Attitudes and Beliefs about Genetics

Primary Sources of Genetics Information	Definition of Genetics
• Education • Employment • Research participation • Physician • Media • Family/Friends	• Inheritance • Defined by technical terms • Defined by the media • Influenced by the environment

5.2.2 ASSESSING AFRICAN-AMERICANS' ATTITUDES AND BELIEFS ON PRENATAL TESTING

Over the course of focus group discussions, participants were able to correctly identify methods of and indications for prenatal testing and converse as a group about the benefits and risks/limitations of prenatal testing. Within each focus group, there were several individuals who had recently undergone prenatal testing or knew a friend or family member who had experienced prenatal testing. Participants identified ultrasound, amniocentesis, and viral studies as prenatal tests but did not use these words explicitly (Table 5). A young woman referred to amniocentesis as "Being tested while you are pregnant with the long needle thing in your stomach." When identifying the types of prenatal tests available focus group participants fell into either one of two categories: chromosome testing and DNA testing (Table 5). Most frequently individuals mentioned chromosome testing for Down syndrome. “I always think of chromosomal problems like Down syndrome and different problems that happen when you are pregnant and they can tell through prenatal testing.” Sickle cell testing was the only DNA test mentioned during discussion. “A lot of black kids have sickle cell they might be able to detect that.”

Table 5. Attitudes and Beliefs about Prenatal Testing

Prenatal Testing Methods	Prenatal Testing Indications
• Ultrasound • Amniocentesis • Viral studies	• Chromosome testing: Down syndrome • DNA testing: Sickle Cell

Overwhelmingly, participants voiced the opinion that prenatal testing has multiple benefits. Themes which emerged during conversation about the benefits of prenatal testing included the value of awareness and the value of choice (Table 6). Participants within each focus group were able to identify the benefit of having information about the pregnancy. However, some believed this awareness would ultimately benefit the pregnancy and the baby while others discussed how this awareness would benefit the mother or the family. Several participants brought up the potential to correct a health problem that is identified prenatally: “Can’t they do surgery while the baby is inside if they find something in order to help the baby before they come out.” Another participant stated prenatal testing was performed “to see what they can try and fix.” Some individuals felt a health problem identified prenatally would allow the child to receive the appropriate care in the newborn period:

> When a child through prenatal you find out something is wrong with that child and that child is born, if you know before you leave the hospital you can treat things so much differently-even different formulas and stuff. If you have a child with different types of diseases or something, you have to be very careful with. So if you know before hand, then you don’t make the mistake of hurting your own child from not knowing.

Awareness of a genetic condition during the prenatal period also benefits the mother and the family. For some participants, awareness of a genetic condition allows the mother to be knowledgeable about possible recurrence and in select cases, make changes to lower the chance of recurrence. One woman described how parental awareness could help prevent spina bifida:

> You could probably take that information to your next pregnancy. When you get pregnant again, if your body was too low with a certain vitamin or some type of nutrition you didn't have while you were pregnant, you could try to boost up on that before you get pregnant the next time so you won't have to face any difficulties or anything like that.

Additional focus group discussion of the value of parental awareness highlighted the importance of how awareness leads to personal choice.

The choice described by participants was often between termination or continuation of a pregnancy in light of prenatal test results indicating the child would have a genetic or other health condition. However, there was another faction of focus group participants who described the choice as whether or not to undergo prenatal testing. The importance of choice in situations which affect the family is described by a participant who stated:

> People have a right to make individual choices. They have a right to make family choices. You can look into things and you can take part in genetic testing or the procedure you were talking about as far as amniotic fluid, but I don't know it still has a lot to do with human choice.

Several individuals stressed the value of choice when deciding whether or not to continue a pregnancy if a genetic condition has been detected by prenatal testing. One woman felt that prenatal testing is performed "to see if there is any deformity in the child and then it gives the mother the right to say whether or not she wants to go full term." Another participant pointed out that the choice to continue a pregnancy is often presented by the physician:

> There also are certain diseases that run in certain races and so this kind of testing while baby is still in utero can help identify them. For instance, Tay-Sachs is a disease that runs in certain groups of people and retardation and certain things like that. So if you know in the first term, then the doctor will say to you, you can make a decision at this time to go forth to go full term or you can terminate therapeutically at this point in time and so that's one very good reason why genetic testing in terms of prenatal is so important.

The value of choice becomes important for each woman or couple when assessing whether or not he/she will be able to care for a child with a genetic health condition. During conversation a participant brought up this point when she said "…Sometimes too they can detect different birth defects before the person is full term to see if that person is able to take that child because the child is going to come deformed or something or maybe its better medically to abort that child." However, no participant mentioned specific social situations or financial situations which would cause a person to choose to terminate a pregnancy.

Community members also valued the opportunity to choose whether or not to have prenatal testing. For some individuals, the detection of a health problem in a pregnancy would cause overwhelming stress and he/she would prefer to be ignorant of a problem throughout the course of the pregnancy. These individuals demonstrated that they are aware of their personal limitations in the ability to handle stress. This feeling is captured by the sentiments of a female participant:

> I think if you know that you are not going to terminate the child and then you know there is something wrong with it, it could be real stressful for you which would also be a problem for the baby. So if you are going to do it, I think you should decide before that if you do want to terminate it, but if you don't, I don't think you really should know ahead of time.

Another participant discussed her personal experience deciding whether or not to undergo prenatal testing:

> When I was trying to get pregnant, my husband and I, we had a very difficult time getting pregnant so then they say once over 40 if you get pregnant you may be more at risk. So then they were going to test and I said if I am not going to terminate why would I want to test because it will just make me more anxious to have another miscarriage.

For these and other individuals, it is important to think through the decision to have prenatal testing and to choose what will be of benefit to their life and situation.

Focus group participants identified fewer risks than benefits of prenatal testing, but participants placed significant emphasis on the risk to lose the pregnancy, the possibility test results may cause stress on the marriage or family relationship, and chance that prenatal test results are inaccurate (Table 6). When asked to identify potential risks of prenatal testing, many made statements such as “You could have a miscarriage.” There was also a subset of individuals who believed the real risk came not from the prenatal test but from the treatment for the condition identified by the prenatal test. One woman described her fear of electronic monitors used during labor and delivery:

> A lot of these doctors want to go up and they are fooling around and do damage to these unborn. You know just like they want to stick these monitors on these kids. I didn’t want it. They did it, and I told them if anything happens I’m suing you because I don’t think you should be putting things on babies’ heads.

Another woman feared the medication which may need to be administered to the mother to treat the baby and stated, “I think a disadvantage comes when you have to give medication or whatever and that medication has an adverse reaction.” Not only did participants fear prenatal testing could harm the fetus but they also expressed concern about the impact of prenatal test results on family relationships. One participant characterized a possible interaction between a couple experiencing the process of prenatal testing:

> (Prenatal testing is performed)…To see if there is any deformity in the child and then it gives the mother the right to say whether or not she wants to go full term because it may be traumatic to raise a child with special needs. So they give the parents cause they tell them to have a conference to decide what they want to do and sometimes doing that it will split up a family because mom says ‘I’m having my child I don’t care what is wrong.’ and dad is like ‘I’m not putting up with a child that has some special needs.’ and so now you don’t have a dad in the picture because he just felt he can’t handle that kind of situation and so a lot of women won’t take it. They’ll just say we’ll find out what happens when the child gets here.

Stress on a relationship is an important consideration in the decision whether or not to pursue prenatal testing despite the benefits named by participants. Participants also voiced concern about the possibility of inaccurate test results and the stress which may emerge when considering this possibility. This theme of distrust of prenatal test results is captured in the following passage:

> I had prenatal testing and the whole time we waited for the test results I think I stressed myself. Like I worried about it so much it was like I don't know just cause more aggravation. That is the only thing I didn't like about the prenatal testing was like you had to wait and after the baby came out more testing and none of the results were true cause like the sonograms weren't really efficient. They could see some degree of the child, but they are not really that great so it was like I am more worrying.

In some cases, the perception of the inaccuracy of prenatal testing is shaped by frustration with the test's limitations. In the previous statement, the participant understood that ultrasound cannot detect or characterize all health conditions but still expressed anxiety that further testing would be required for her child.

Table 6. Risks and Benefits of Prenatal Testing

Benefits	Risks/Limitations
• Value of awareness • Value of choice	• Risk of miscarriage • Relationship stress • Possible inaccurate results

5.2.3 ASSESSING AFRICAN-AMERICANS' ATTITUDES AND BELIEFS ON NEWBORN SCREENING

Focus group discussion revealed three distinct themes characterizing participants' knowledge of newborn screening: those who are unaware, those with accurate but incomplete knowledge, and those with misinformation (Table 7). The greatest majority of focus group participants did not respond or made movements such as shoulder shrugging to indicate uncertainty when asked to describe newborn screening. Common responses indicated a lack of awareness such as "Never heard the phrase (newborn screening)" and "So is this (newborn screening) something new? I mean, how recent?" Each group contained at least one individual who was able to identify a genetic condition which can be detected by newborn screening. Specifically, individuals correctly believed sickle cell, cystic fibrosis, and phenylketonuria are screened for in the newborn period. However, none of these individuals identified that newborn screening is a heel-stick blood test performed in the first days of life. Interestingly, one participant accurately identified that the number and type of condition tested for by newborn screening varies from state to state. "There is a lot (of tests) out there and Pennsylvania probably only does 9 or so, but there are other states who do. So I don't know if our state should do more or if that is a requirement of the parent." This participant is accurate in her understanding of the differences between state programs but indicates that she still has incomplete information as to who decides what is included on a screening panel. In contrast to participants with accurate but incomplete information, there were a number of individuals who are misinformed about the use of newborn screening and who were unable to provide an accurate description. These participants cited blood type, heart murmur, diabetes, prenatal drug exposure, jaundice, and asthma as conditions tested for by newborn screening.

Table 7. Knowledge of Newborn Screening

Knowledge of Newborn Screening
• Unaware • Accurate but incomplete knowledge • Misinformation

Participants only identified benefits of newborn screening after a common definition of the process was shared by the focus group moderator. The benefits of screening included themes of preparation and treatment (Table 8). With each of theses themes, the benefit indicated by the participant was for the child and not for the parent or family. Individuals expressed a desire to have the information gathered from newborn screening to prepare to care for that child in the appropriate way. One mother discussed the importance of knowing her child had a genetic condition called glucose-6-phosphate deficiency or G6PD in order to protect him from substances which could aggravate his condition.

> My son, I adopted him, he was an infant and he had G6PD - something I knew nothing about. So it was good to know that - to have that information from the hospital. I know what not to have around him, what not to give him especially like aspirin, no mothballs around him. Cause my mom is from the old school. She used to have mothballs so I was real glad that I knew.

Another participant focused on the value of knowledge and the sense of security from being aware of a child's genetic condition. "You need to know what to look for. If it's there, if something happens, you need to know what you're dealing with." There were also participants who valued advance knowledge to prepare the appropriate services for a child in order to maximize his/her potential. For example, "I think of to get as much early intervention as possible to help the child in ways that the child could be stimulated." The possibility for early treatment when a condition is identified by newborn screening was also explored by participant discussion. "I know you can test for a lot of conditions that can be treated within the first 3 months you can give medication to the baby to be okay." This participant recognized the potential to provide treatment to prevent the effects of an untreated genetic condition. The concept of using awareness and treatment to improve the outcome for a child with a serious condition is also elucidated in these comments:

> …And with cystic fibrosis at least they can start a plan of treatment so the child can have a productive or as much of a productive life as possible as opposed to finding out when they're older and now, you're starting from scratch when you could have prevented something. It's the same thing like a child that is diagnosed with autism. If it's further down the road, then you're behind the eight ball. Where as if you find out early enough, you can start doing things to help get the child in the right direction and get them in mainstream.

This participant and others articulated the benefit of knowing a genetic condition is present in order to provide a child the best care and opportunities for a healthy life. Conversation demonstrated that when individuals are provided with accurate information about newborn screening they find it to be a valuable service which improves the ability to care for their child/children.

Table 8. Benefits of Newborn Screening

Benefits of Newborn Screening
• Preparation • Treatment

5.2.4 KNOWLEDGE OF SICKLE CELL DISEASE AND TRAIT

Analysis of participants' conversation about sickle cell disease and trait identified the primary theme of reliance on experience (Table 9). Each participant referred to witnessing an acquaintance, family member, or friend living with sickle cell disease or sickle cell trait when describing a feature of the condition or its inheritance. These experiences allowed participants to speak confidently as they described the symptoms of the disease such as pain, weakness, and shortened lifespan. One woman describes what she has learned about pain crises from talking with a co-worker:

> I have a friend she is a co-worker and she has a daughter that has it. She has a son he doesn't but her daughter has had it since birth and what I've noticed is it seems like the older she is getting, because she just turned 13, the older she is getting the more episodes or crisis. It's painful. Her mother tells me she can't even get out to walk and she's 13 and she only stand this tall. She has had bouts in the hospital. They have to put her on morphine so you can imagine a little body like this it affects her psychologically.

Another speaks about the pain he witnessed when a family member went into a crisis stating, "I do know it's very painful very, very painful because he used to ball up just fall on the floor and he'd be in a ball crying." One participant who was a junior high school teacher articulated her interaction with a student diagnosed with sickle cell disease.

> If you've ever watched a child suffer from sickle cell- I had students that had sickle cell…I was her teacher. She broke my heart to watch her sit in my classroom and the pain that she suffered with and to know she suffered all her life and even sadder, was she knew she was never going to see 50 the way her health was.

This quotation also highlights the belief that sickle cell disease shortens the lifespan. When describing the symptoms of sickle cell disease, participants referred to individuals they had seen pass away at young ages and made statements such as "When I was younger, there was a girl she lived to the age of 12, and she had sickle cell" and "I had a friend who had sickle cell and she would go into a crisis quite a bit. She didn't survive. She passed away in her early 30s." When discussing additional symptoms like weakness/tiredness, individuals continued to voice personal experience observing sickle cell disease in the family or community. "My husband's niece has it and the only thing I know she gets very weak and has to have a lot of bed rest." Another participant based her knowledge of sickle cell disease symptoms on children she has observed in her church.

> …If they get over exerted like there are some young girls at church that are involved in everything and they are involved in their school activities so what happens with them is when they are just over extended for some reason that brings about an episode.

In addition to articulating an understanding of sickle cell disease symptoms, several focus group participants were able to identify the cause of symptoms and the differences between disease and trait. Some were able to connect the name of the condition to the abnormal shape of the cell. Statements included "…From what I understand it's like your cells are an abnormal shape" and "The shape of the cell is like a moon or crescent or something and it stops the flow of the blood." Participants distinguished sickle cell disease from sickle cell trait by the presence or absence of symptoms and again relied on personal experience to make this distinction.

> I had a friend who had sickle cell and she would go into a crisis quite a bit. She didn't survive. She passed away in her early 30s and in her family there were a number of people who had the trait and when you asked the question, the main thing I thought of was the trait don't go into crisis situation.

Participants' reliance on personal observation of individuals with sickle cell disease and trait in order to understand the condition also led to significant misconceptions about the inheritance pattern. Individuals understood that sickle cell disease and trait runs in families. "Right so it has to be genetically in that family - older sister had it (sickle cell disease) and one of her brothers had it and one died from it." However, there was significant uncertainty and misconceptions that emerged as participants attempted to make sense of the pattern of disease and trait seen within a family. Some individuals questioned why only some children have disease when each parent has trait.

> You would think, at least I do, if the mother or the father or both that all the siblings would have it. Why is it maybe just one out of three or four may have it and the other ones no?

Other participants were uncertain as to why sickle cell disease would appear for the first time in a family.

> My grandson's cousin he was the first in our family or someone that I knew that had sickle cell. I mean it was a surprise to me that his first cousin was born with sickle cell and then later on, my grandson, my daughter's son, has a trace of sickle cell. So I don't know where that came from. It was surprising to me that he has the trait. I am clueless as to whether he received that from his father because I know of no one on my side of the family that ever had that that I am aware of.

One woman observed a pattern where only females were carriers of sickle cell trait in her family. "I have the sickle cell trait and when you were talking about different family members that have it, it seems like the females in my family have it. I had the trait, my daughter, my sister." Participants did not articulate what they believe the chances are to have a child with sickle cell disease when each parent has trait nor did they discuss the chance for a child to inherit sickle cell trait when one or both parents have trait.

Apart from discussion about the inheritance and clinical features of sickle cell disease, participants were keenly aware of the social issues that emerge from a sickle cell disease diagnosis. Again participants focused on their observations of family dynamics when a child has sickle cell disease. This participant described the stress in a family as they try to manage caring for a sick child.

> I watch my friend. I ache for her because unfortunately we both work for a major corporation - I won't mention - but the area she works in they don't show sympathy for the time she needs to be off when her daughter is in the hospital and she'll spend the night at the hospital and she'll go to work. Her husband the type of job he has he can adjust his schedule a little bit so he'll be there during the day while she goes to work and she goes home washes up goes to the hospital, spends the night at the hospital, gets up, and goes to work.

Another participant focuses on the helplessness a family feels when a child with sickle cell disease is in pain.

> And the drain it takes on family members as parents to know something is wrong with your child and you're helpless and what I see my girlfriend go through at times there is nothing she can do to ease the pain and discomfort.

Childhood disease can cause tension and stress within a family as they try to manage caring for a child and maintaining jobs and other relationships. Participants gained intimate knowledge of these strains from discussion with individuals who have a child with sickle cell disease. Other social issues that were addressed by participants' conversations focused on the stress an individual with sickle cell disease may experience as he/she interacts with the health care profession. A woman describes the difficulty an adult may have acquiring pain medication from a skeptical physician in order to treat his/her sickle cell disease.

> The other thing sometimes the medical profession is not very sympathetic and they think the young people want the drug and there is really no pain. So there are a whole lot of things that are going on. They think you just want some morphine drip. Come on! You want to be out doing things you want to do and being active just like your other friends but because you are in the hospital and they can't pin point the pain its almost like back pain you can't really pin point it.

Additional participants spoke of social/personal strains on an individual with sickle cell disease when they miss school or work due to time spent in the hospital.

Table 9. Knowledge of Sickle Cell

Knowledge of Sickle Cell
• Reliance on experience - Symptoms - Distinguish disease and trait - Misconceptions/Uncertainty about inheritance - Social issues

5.2.5 BARRIERS TO EDUCATION AND AWARENESS OF SICKLE CELL DISEASE AND NEWBORN SCREENING

Participants' discussion revealed that there are a significant number of individuals who have inaccurate or incomplete information about sickle cell disease and newborn screening. However, it was important to analyze conversation in order to determine why this is so. Each focus group provided insight into the barriers that prevent an individual from acknowledging the presence of sickle cell disease in the community and the chance that a child could be born with the condition within his/her family, as well as, the barriers to accessing information. The researcher categorized these barriers as personal, familial, and societal/cultural (Table 10).

Themes identified as personal barriers include: fear, denial, and overwhelming responsibilities. For some individuals, fear was used to describe inability to trust the medical establishment due to past abuses directed against African-Americans. Participants discussed the Tuskegee syphilis study and eugenic attempts to eliminate African-Americans. As one participant said, "Like the Tuskegee experiments, our parents pass that on to their children and grandchildren. That's don't trust the police, don't trust the medical that is passed on." Participants did not feel that newborn screening for sickle cell is an attempt to discriminate or harm African-Americans, but the general fear of testing and medical professionals prevent individuals from going to the doctor and seeking information. This distrust and fear was also evidenced in young focus group participants. One young woman stated:

> I am praying on it because it is really crazy the way they use black people for experimentation and then they don't teach you a dog gone thing in the schools about what is going on and it probably has to occur with the people…start speaking up and saying something.

Fear also played a central role for some individuals who believed that if you think about disease it will happen to you or your family members. A male participant stated:

> I believe in not worrying about something. If you start thinking about these different diseases coming your way sooner or later it's going to smack you right in the face. I don't deal with it. When it happens, it happens.

Another woman expressed a sentiment that was heard repeatedly during each focus group. "Sometimes people they don't want to know. They like to stay ignorant to the fact. If I don't know it, then it can't hurt me." This fear certainly prevents individuals from acknowledging the risk for sickle cell disease in their families and communities and inhibits them from seeking information from health professionals. The fear of disease is not specific only to sickle cell. One focus group attendee spoke of her experience with relatives who did not want to acknowledge the risk for polycystic kidney disease in the family.

> I have to be real strong even with the rest of the family. This is something I have to do. I have brothers and a sister. 'I don't want to know. I don't want to know.' But like I said,

> I want to save my grandchildren. I am trying to protect my grandchildren. I was trying to protect my children. It was not something that I went into. I was forced into it.

This fear of knowing about the increased risk for and even, at times, trying to avoid thinking about the risk appears different from the denial of facts that other participants spoke of. These participants described having an accurate knowledge of the risk for a particular disease in the family but openly denied its impact on their personal health. A woman summarizes this barrier, "Even those things that the family has experienced, I don't worry just because they experienced them. Doesn't mean me and mine are going to experience it." Another participant expresses this sentiment as a rejection of family history.

> Sometimes we take on different things that we hear and we accept and sometimes the doctors will impart things and make you believe that you are going to have this. No I am not going to have this and I am not accepting what you are saying and I am not going to receive it. My grandmother had that and yes my mother had that but that does not mean I am going to have that. Now you may want me to have this but I am giving this back to you. I am not having cancer. I am not accepting diabetes, and I am not claiming it from this point on so I am giving it back to you.

For this participant, it appears that accepting the risk for disease in the family would be too overwhelming and it is a self-preserving to deny the increased risk.

A personal barrier for participants of all ages was the feeling of too many responsibilities and no time to be concerned about the risk for sickle cell disease. In some cases, this could be characterized as avoidance behavior while for others it is an uncertainty about what questions to ask. For young mothers, there was a sense that other daily challenges take precedence over thinking about newborn screening for sickle cell disease/trait. This participant describes the overwhelming responsibilities and concerns of young mothers and the inability to focus on non-pressing issues.

> I believe as a lot of our people have other things to worry about beside whether my kid has sickle cell. There are other issues whether they are going to have food on the table,

> whether their mother is a drug addict. A lot of these kids are out there. They are raising themselves because their parents are drug addicts. They have other issues that are more important to them than this.

When asked whether or not they are concerned about the risk of sickle cell disease in the family, several focus group participants also believed sickle cell disease is not a personal priority. Individuals made statements such as "I have too many other things. I don't even want to go there." As demonstrated by this participant, a feeling of no time to think about sickle cell may be avoidance behavior.

The absence of open communication about risk for disease is the primary family barrier to sickle cell disease awareness and education. In general, participants believed that a feeling of shame prevents older relatives from sharing a history of sickle cell or any other disease. In the following passage, a participant describes the breakdown in communication about health issues in her family.

> I'll give you a for instance. My dad is 82. I just found out about a year ago that my dad is the oldest brother out of 6 siblings. My cousin is doing a family history so he called. He is a radio announcer in Baltimore. He said I'm going to do a family history and ask everybody what school they went to, who they married, and so forth. So I asked my dad he said well you know one of my brothers has diabetes and my other brother has this and my sister has this but don't tell them I told you so. What I am saying is if his brothers and sisters have these illnesses and he doesn't want me to know how are their children supposed to know. How is this going to go down from generation to generation?

Participants were aware that a lack of family discussion about risk for disease prevents future generations from making informed decisions about their health. A participant shared the following, "I think a lot of times most families we don't sit together to talk about family history and those things pass down from generation from ancestors and so once you learn your family history then you can have a better picture." There is also a positive belief among participants that families are improving communication. A participant expressed this opinion saying, "I think more families are talking more about it. It's not seen as you have to be ashamed about it." The shame and stigma of disease is decreasing as individuals come to understand the importance of knowledge and prevention.

Focus group discussion illuminated the primary societal barrier to sickle cell education and awareness. The central theme identified was a lack of awareness due to little media attention, minimal physician focus on the topic, and ignorance among the increasing number of teenage mothers. Comments made by participants were striking for this lack of awareness in the community.

> Because you didn't hear about it a lot anymore. I hadn't heard about sickle cell since what the 60's or 70's and knew people that I went to elementary school with and part of high school and middle school maybe that had it and they passed on many years ago in their 20's or 30's and you just didn't hear about it anymore. I thought maybe no one was getting it anymore.

Other participants also believed that due to the lack of attention and their own lack of awareness on the topic of sickle cell that the disease prevalence had decreased or that a curative treatment had been developed.

> I think of people not being made aware of what sickle cell really is. Before I became employed with the school, I hadn't heard about sickle cell for many, many years and I thought wow it had passed on but I'm like wow it's still alive and it's still untreatable, incurable.

This lack of awareness also led to sense of frustration and even anger for individuals who had a first encounter with the condition when a child was born with disease or trait. The following comments are from a female participant who recently became the mother of a child with sickle cell trait.

> I didn't know about it either until I had my son and they told me he had sickle cell, the trait, but it's the fact that I never - I mean my husband is here - but I never heard of it either. I could have had it. He could have had it and we had kids and they could have had the actual sickle cell.

Focus group conversation brought attention to the primary reasons why participants feel there is a lack of sickle cell awareness in the community. First, participants explained the belief that there is not enough sickle cell disease activism and health education within the community and that leads to minimal media attention on the condition. A male participant spoke of the absence of public health advertising needed to increase awareness in the community.

> I remember in the 80's it was on billboards that is the first time I had heard about it. I came back here from some place else and it was on billboards they had some celebrities on TV talking about it then after the 80's you don't hear about it anymore.

Participants believe that other public health issues such as HIV/AIDS are given more media attention within the community.

> It's (sickle cell) a disease that affects mostly African Americans and then I'll bring this up. AIDS affects everyone. Every day, every time you can see radio, print, TV about AIDS and HIV 'Get tested.' How come nobody is out in the communities saying to African Americans put it (sickle cell) on TV. It doesn't matter whether it affects Caucasians or Mexicans or whoever. Why isn't it out there? Like I said I didn't know about it 'til I was 30 something years old.

Participants acknowledged the significance of preventing HIV/AIDS in the African-American community but feel that sickle cell disease is also a health condition which can be impacted by media attention and campaigns to increase awareness and decrease prevalence.

> What I'm saying is that they are paying millions of dollars for commercials like herpes is the greatest thing to have. If you have it they are spending money on things like that that people can prevent. Of course you have to put the awareness out there about HIV but how are you getting it? I am saying for the imperfections that we all inherited put that out there too cause you don't know it's out there.

Individuals recognize the lack of awareness in the community as a whole and would like to have more attention given to sickle cell in the media.

Participants believe that a lack of awareness also stems from physicians not providing the necessary information about sickle cell. Throughout the conversations, participants expressed their frustration with their lack of knowledge and felt that their physician should have supplied the information regardless of whether or not the patient asked about carrier testing. In many cases, participants believed discussion about sickle cell should occur at the same time as other conversations with their physician regarding family history and risk for multifactorial conditions such as hypertension and diabetes.

> You go in the doctor's office waiting for him. You see 'If you have diabetes, take your socks off and shoes.' You see mammograms. You see all these different things on the wall. There are flu shots. I don't see anything about sickle cell. I don't see anything about lupus. I see more about stem cell research which I have no clue really what it is but I see a lot of it and bone marrow transplants for African Americans cause that is so rare. If it only affects a certain group of individuals be it minorities, then it seems like there is not a lot of information out there and they (physicians) don't care.

There was a strong feeling among the focus group participants that when a patient does not have awareness of sickle cell disease risk it is important for the physician to ask questions and provide them with the appropriate information.

The sentiment of the need for physicians to educate patients was also expressed when participants discussed the lack of sickle cell awareness among young mothers. One woman stated:

> Mothers are not physicians and so you can't expect them to be informed and know what questions to ask and so I think if you are a professional concerned physician then you will supply that kind of information to them and particularly young mothers. Girls are having babies at 11 and 12, 15, 18 years old they don't have any questions to ask they don't know and so it's the responsibility of the physician.

Participants felt that teenage mothers are ignorant of the risk for sickle cell disease and therefore, healthcare providers must guide these young women, ask questions about family history, and take the time to explain sickle cell disease and trait.

> I think also with mothers being so young today they are not aware. What do they know about sickle cell? Adults don't know about it. With the age of the mothers being so

> young, they don't know and a lot of times the doctors they take advantage of that and they may just mention it but it will blow over. This is a young mother. She is probably concerned about how am I going to take care of this child - fearful and frightened. So doctor probably knows that but that parent doesn't really focus on that.

The young mothers are inexperienced with what questions need to be asked when speaking with the doctor and participants strongly believed that physicians need to take the time to have a conversation about sickle cell and carrier status.

Table 10. Barriers to Awareness and Education of Sickle Cell and Newborn Screening

Personal	Family	Societal
• Fear • Denial • Overwhelming responsibilities	• Absence of open communication	• Too little media attention • Minimal physician discussion • Ignorance among increasing number of teenage mothers

5.2.6 IMPROVEMENTS TO COMMUNICATION ABOUT SICKLE CELL AND NEWBORN SCREENING

Participant discussion stimulated suggestions of how to improve awareness about sickle cell and improve communication about newborn screening. Participants were enthusiastic when making suggestions of how to expand access to information for members of their community. Their suggestions included holding community discussion groups, involving the church in information dissemination, increasing public health messages using the media, developing school programs focused on inheritance and sickle cell, and adding a sickle cell trait status question to the family history questionnaire provided by physicians (Table 11). At the conclusion of each focus group, participants were excited to continue the conversation about inheritance and sickle cell with other community members. Many mentioned holding further discussion groups at the Kingsley Association where they could feel comfortable asking an expert.

> That is what is missing in our community all discussions about vital issues critical issues so this is a real important thing is because we got to talk to each other we have to discuss things so this is very good.

Participants also pointed out that conversations about prenatal testing, newborn screening, and family history need to occur at churches as well. Individuals felt that these topics involve moral issues which impact the family and are best discussed in the church. Several participants wanted to see increased use of the media in the sickle cell public health campaign. Suggestions included commercials and billboards. None of the focus group participants mentioned knowledge of the current project developed by the Sickle Cell Program at Children's Hospital of Pittsburgh that is targeted at sickle cell education in public schools or the Murray-Irvis Sickle Cell Society of Pittsburgh. However, when making suggestions participants described the need for such a program aimed at young people and educators.

> I think someone needs to come out to the school district to let the people know sickle cell is still alive, it is still incurable disease, and we are still working. I don't know how many agencies are involved in working on sickle cell but you don't even hear about that anymore with foundations. People need to be made aware that sickle cell is in children. You have it here in your school. People need to know. They need to be made aware.

Focus group participants also wanted to add a question about personal sickle cell trait status and family history of sickle cell to current medical questionnaires completed at the physician's office. They believed this would encourage physicians to discuss the topic with their patient and/or stimulate the patient to ask the physician questions about sickle cell.

Participants were adamant that awareness of sickle cell would improve if a physician communicated the information with compassion, empathy, and patience. One woman spoke of her frustration with doctors who do not give of their time to listen to patient concerns. "No one has the time I mean these doctors come in and look at you. They are not in there a good five or ten minutes while you are thinking of something to ask they are on their way out and you're like wait a minute." Another participant responded to her comment by stating, "That's why I stayed with the doctor I'm with. She pulls up a chair and sits down." For these individuals it was important to receive information about sickle cell and newborn screening from a physician they have a relationship with.

> It's that piece of mind just knowing that all you got to do is get on the phone or go and say this, this, and this and you know you are going to be taken care of. You know you are going to be talked to like you are a person. They got your interest at heart with them that means so much to a person especially when you are hurting.

Individuals appreciated communicating with their obstetrician about newborn screening due to the familiarity and the established relationship. "Obstetrician - cause during the pregnancy because you are relating to them and they know a lot about it (newborn screening)." For these participants, it was more important to hear the information in the prenatal period by the obstetrician because of this relationship and not because of the hectic time period after the birth. Participants also recognized that it is important to ask questions and independently seek information. "My PCP, I'll call her. We'll talk. We have a very close relationship and then from there I'll start my research I'll go out to the internet." When a patient is informed, it improves communication and the relationship with the health professional.

> I am my most prized advocate I am very, very informed and very knowledgeable and I know that I need to be informed and knowledgeable so that whatever is wrong with me I am an informed partner with my PCP and together we discuss my treatment and my treatment options.

Participants acknowledged that communication cannot be a one-sided process.

Table 11. Improvements to Communication about Sickle Cell and Newborn Screening

Increase Awareness
• Community discussions • Church involvement • Increase media messages • Adding sickle cell question to physician questionnaires • Acknowledging importance of physician-patient relationship • Developing school sickle cell programs

5.2.7 APPROACHES TO DECREASING SICKLE CELL DISEASE PREVALENCE

Focus group participants had the belief that increased awareness of sickle cell disease in the community will lead to greater family communication and ultimately, lower rates of sickle cell disease. Participants believed that family discussion will lead to more informed choices for future generations. In the following passage, a woman discusses her hope that by telling her son he has sickle cell trait he will think through his selection of a spouse/partner.

> I first found out that my oldest son was a sickle cell trait and I was told at that time and by them talking let me know that they explained to me at that particular time that he had to be very careful who he had kids by. He had to make sure that person was not a trait too and I'll tell you something that is something else that I had to, you know, like when they talked about getting married going with the traits and stuff because I told them you don't want to have it if you can prevent it. You don't want to have a sick child like that. I mean if you are going to be a trait and your mate is a trait and you both know that then at least you can say well we choose not to have children and choose not to let that to have a sick child.

Intertwined with this desire to open communication in families and to allow informed decisions is a belief that the chance to have a child with sickle cell disease will lead to great fear. Participants believe that this fear will cause a couple who both have trait to choose not to have children. A participant mentions a friend who is unwilling to continue taking the risk of having a child with sickle cell disease, "I had a girlfriend who said both her and her husband have the trait. They have two kids and they are stopping. They don't want to risk it." Another participant discusses how she would not have had children if she was aware of a genetic condition in her family.

> If somebody talked to me about genetic stuff when I was young, you wouldn't have to worry about my legs being closed because they would have stayed closed. I mean seriously some things you don't want to be responsible for passing on to your children.

Participants also believed that exposing young people to families with a child who has sickle cell disease or allowing young adults to speak with a person who has sickle cell disease would cause them to think about their own trait status and reproductive decisions.

> It's like you have to see it. This is real. Look at these children that are in the hospital. Look at these parents getting up at 4 in the morning. They are missing work. It's the grace of god that the father has a job where they are understanding and the mother essentially works part time because her time is going back and forth to the hospital that has to be a part of the reality when you do a round table discussion. Bring a child in with sickle cell and let them hear what they go through because it brings more meaning. 'I don't know what I am going to eat tonight.' Well you know what? How would you like to not know what you are going to eat tonight, not know if you are going to have a job tomorrow, and not know if you are going to get up at 2am to take your child to the hospital because they are sick again. That is a reality. So I would rather deal with 'I don't know what we're going to eat tonight' but 'I know I took the time out to make sure genetically you didn't have the trait and even though I have the trait, we are going to have a healthier child.'

6.0 DISCUSSION AND CONCLUSIONS

The specific aims of this study were to assess African-American community members' attitudes and beliefs regarding sickle cell disease and trait, prenatal testing, and newborn screening. However, the overarching goal of the project was to characterize the barriers to education and awareness about sickle cell and newborn screening in order to determine if a community based intervention project could be developed to improve knowledge of these topics. Qualitative methodology was pursued to explore these subjects in the African-American community of Pittsburgh and to understand the perspective of the people the intervention project would serve. It is the belief of the researcher that the contribution of this study is not only in the analysis of what African-American community members do and do not know about genetics, newborn screening, and sickle cell but to understand why the majority do not have a familiarity with these topics and to determine if an intervention could be developed to improve education and awareness.

This study included the opinions of 35 individuals of African-American and/or mixed ancestry who reside in the Pittsburgh communities surrounding the Kingsley Association of East Liberty. The individuals who participated in this research project were self-selected and many spoke of the importance of giving back to the Healthy Black Family Project by contributing their time to research. Of the 35 participants, the majority (74%) are members of the Center for

Minority Health's Minority Research Recruitment Database and therefore, already inclined to partake in research.

Pre-discussion survey results characterized the demographics of focus group participants. Approximately 91% of participants are female. This percentage is comparable to the statistics collected on gender by the Family Health History Initiative of the Healthy Black Family Project. Of the 420 individuals who have completed their family health history, 83% are female. Unfortunately, the uptake of HBFP services by African-American men has been low, and the recruitment of the current study is reflective of the gender gap seen in the larger project. However, the Family Health History Initiative is currently developing methods to reach out to the male population. Studies examining the barriers to African-Americans' participation in research have demonstrated that men and women differ in the types of barriers described and the impact those barriers have on participation (BeLue, 2006; Hoyo, 2003). Specifically, African-American men in qualitative studies have stated that they are concerned about the costs and time commitment of research participation (BeLue, 2006; Hoyo, 2003). Additionally, men have been found to have a low interest level and participation rate in research because of a lack of knowledge and awareness about the process of research (Hoyo, 2003). In contrast, African-American females have described barriers such as the researcher-participant relationship and the need to have the researcher make the participant feel comfortable (BeLue, 2006). These barriers could be contributing to the current study where the majority of participants are female. Women may have felt more comfortable than men attending the focus groups because they had previously met with the researchers and are familiar with the Healthy Black Family Project and Kingsley Association. Male participation may have been negatively impacted by the two hour time commitment advertised for the focus groups.

The median age of focus group participants is 57 years with a range of 26-77 years of age. Though discussion of topics such as newborn screening and prenatal testing may be more appropriate for individuals of childbearing age, the overall goals of the project were attained by capturing the opinions of diverse members of the community. It is also important to point out that as evidenced by group conversation sickle cell disease pervades community life regardless of age. Many of the older focus group participants are primary caretakers for grandchildren and had intimate knowledge of prenatal testing, newborn screening, and sickle cell.

The 35 participants who comprised this research study are representative of the household income level of the Health Empowerment Zone but have a higher level of education. The majority or 32% of focus group participants have a household income between $20-35,000 and 56% have a household income between $10-35,000. Data collected by the US Census in conjunction with information compiled by the Center for Minority Health (CMH) and the Allegheny Health Department has shown that the median household income within the Health Empowerment Zone is $26,167 with 27% falling below the poverty level (Robins, 2005). The 2005 US poverty threshold for a family of four with two children under age 18 is $19,806 and the poverty level for a family of two with no children is approximately $13,000 (US Census Bureau, 2005).

Data collected by CMH and the Allegheny Health Department has also shown that approximately 81% of individuals living in the Health Empowerment Zone are high school graduates (Robins, 2005). In the current study, 100% of participants completed high school with 72% completing post-high school education. The difference in educational background between focus group participants and those living in the Health Empowerment Zone indicates that this sample of individuals is not representative of the larger African-American population.

Importantly however, this study was able to demonstrate that a sample of middle class and working poor African-Americans with a high level of education has both inaccurate and incomplete information about newborn screening and sickle cell. Therefore, one could infer that a sample of African-Americans who more closely resemble the population living in the Health Empowerment Zone would have even less accurate knowledge of these topics.

The current study found that a majority of focus group participants do not know their personal sickle cell trait status or that of their spouse/partner and children. A previous study surveying 147 African-Americans demonstrated that 31% know their personal trait status while a survey of 282 African-Americans in a large metropolitan area of California identified that only 15.9% know if they are a carrier for sickle cell (Midence, 1994; Wright, 1994). This research project has shown that 42% of the 35 participants know their own sickle cell trait status. This percentage is higher than expected, but given that 97% of participants have health insurance and only 17% have difficulty seeing a physician due to cost, individuals may have greater interaction with the medical profession and access to carrier testing for sickle cell. The large percentage of participants with knowledge of their personal sickle cell trait status may also be due to their age. The majority of focus group participants experienced their childbearing years during the early 1970s which is the time period when federal funding and state sickle cell screening programs were offered (Markel, 2006).

Interestingly, of the 27 focus group participants with a spouse or partner only 26% know that individual's sickle cell trait status. This figure emphasizes the theme which emerged from focus group discussion regarding the lack of family communication about health conditions. Of the 82% with children, only 40% of participants know their child's sickle cell trait status. This may be reflective of the fact that many participants are past their childbearing years, and newborn screening for sickle cell did not become mandatory in Pennsylvania until 1990 (Olney, 2000). As participants indicated throughout focus group discussion, parents of older children may not have known to ask their physician to perform carrier screening due to a lack of awareness of the risk for the condition. Additionally, physicians may not have explained the importance of sickle cell carrier screening to their African-American patients.

Analysis of focus group transcripts demonstrates that participants fall into one of three knowledge categories when discussing genetics, prenatal testing, newborn screening, and sickle cell. These categories are those who are unaware, those who have accurate but incomplete knowledge, and those who are misinformed. Unfortunately, all of these categories indicate that the current work to educate individuals about newborn screening and sickle cell is not sufficient. There were many individuals who had pieces of the larger issue when discussing each of these topics. However, amidst each statement indicating accurate knowledge were statements where participants held an inaccurate view or had no information to contribute to the discussion. For example, participants had an accurate understanding of the symptoms and clinical course of sickle cell disease but in general were misinformed about the autosomal recessive inheritance of the condition. Individuals accurately identified that sickle cell trait and disease travel through families but did not understand the risk to children conceived by parents where one or both are sickle cell carriers. These findings support the work of Treadwell et al. who also found significant misconceptions among African-Americans about the inheritance pattern of sickle cell in various family scenarios (2006). When discussing newborn screening, there were a significant number of participants who indicated they are unaware that such a program exists. Focus group work in the community setting by Catz et al. identified a similar lack of awareness about newborn screening (2005). However, in the present study there were also many participants who inaccurately believed newborn screening was testing performed to determine if a baby had jaundice, prenatal drug exposure, or other non-genetic health conditions.

During analysis of focus group transcripts, it was interesting to witness the impact of providing brief genetics education on the flow and topics of conversation. For those individuals who did not have an understanding of newborn screening or prenatal testing, it was important for the moderator to share a definition so that conversation could proceed. This small amount of education stimulated focus group discussion and allowed individuals to debate the risks and benefits of prenatal testing and newborn screening and to articulate the issues which impact the community. After individuals determined the differences between prenatal testing and newborn screening, they were able to deliberate the benefits of having knowledge in the prenatal period versus after birth. In the prenatal period, participants described the benefit of having genetic information so that they were aware of the risks and could make choices about continuing or terminating a pregnancy. For newborn screening, benefits of knowledge were for treatment and preparation. The theme of preparation as a benefit of newborn screening was also identified in focus groups conducted by Catz et al. 2005 and Laskey et al 2003. Interestingly, individuals in the current study could articulate not only the procedural risks of prenatal testing such as miscarriage, but the risks to the family relationship and individual mental health when genetic information is learned. However, newborn screening was viewed as only beneficial with no risks or downfalls to having this type of genetic knowledge. This may be due to participants' less sophisticated knowledge of newborn screening when compared to prenatal testing.

As African-American participants attempted to make sense of complicated issues such as genetic disease and genetic testing in the prenatal or newborn period, the theme of reliance on experience was expressed. When discussing the resources participants access for genetics information, very few individuals spoke of seeking out information from the library, books, or internet resources. Similarly, a quantitative study by Nicholson et al examining the relationship between race and women's use of health information resources found that African-American women had 50-70% less odds of using print news media, computer resources, or health organizations for information when compared to Caucasian women (2003). Instead of these traditional sources, focus group participants relied on what they knew from on the job experience, participation in research studies, stories they had witnessed on television, and discussions with physicians, family, and friends. Similarly, when participants described the symptoms and inheritance of sickle cell disease they alluded to observations of children, adults, and families with sickle cell disease and how the condition impacts daily life. Research on the information seeking behaviors of African-Americans has demonstrated the theme of the importance of personal sources such as family, friends, and the church (Nicholson, 2003; Talosig-Garcia, 2005). Nicholson et al demonstrated that African-American women are more likely than Caucasian women to report family/friends and the church as useful health information resources (2003). Focus group participants in the current study relied on familiar sources and past experiences to explain their attitudes and beliefs on a topic and spoke confidently regardless of whether or not the information they shared was accurate.

Focus group participants also expressed the theme that familiarity increases the ability to trust. For example, participants spoke of a reliance on friends and family who had an affiliation with the healthcare profession as a resource for genetic and other health information. This familiarity allowed them to converse with great ease and trust and therefore, rely on that particular individual as a primary resource for health education. Participants also valued the familiarity and relationship with a personal physician. Similar to the findings of Treadwell et al.

2006, participants believed that in order to be receptive to what the physician is communicating there must be a mutual respect and established relationship between physician and patient.

In addition to the importance of the physician-patient relationship in the communication/education process, focus group participants identified specific personal, familial, and societal barriers to awareness that are at work in their family and community lives. The personal barrier described as fear and distrust of the medical profession due to past abuses is an issue deeply rooted in the African-American culture. The theme of distrust of the medical profession emerged in focus groups conducted by Treadwell et al and Zimmerman et al as well. Additionally, a qualitative study of African-American community attitudes toward medical research found that the Tuskegee syphilis study was an important negative factor in the decision to participate in research for older individuals while distrust of physicians was a personal barrier for both younger and older participants (Hamilton, 2006).

Participants also feared acknowledging the risk for a health condition like sickle cell to occur. Many individuals believed that simply by thinking about a risk it would cause the disease to develop in themselves or a family member. This magical thinking was an interesting disconnect among participants determined to have a high level of education in comparison to the general population of the Health Empowerment Zone. This fear led to denial or rejection of family history for some participants. African-Americans' denial of multifactorial health conditions has been documented in the literature. Specifically, denial has been associated with diseases which have a significant racial health disparity such as prostate cancer, heart disease, breast cancer, and stroke (Gullatte, 2006; King, 2001; Richardson, 2004; Rucker-Whitaker, 2006). These studies have demonstrated themes of anxiety and repression of fear which leads African-Americans to deny their risk for disease (Gullatte, 2006; King, 2001; Richardson, 2004;

Rucker-Whitaker, 2006). A study by Gullatte et al documented this denial as "'If you name it, you claim it'" (2006). This sentiment is similar to the feeling expressed in the focus groups of the current study that if you think about a disease it will happen. It is difficult to determine the cause for such a belief in this sample of African-Americans. However, the presence of multiple individuals in each of four focus groups who expressed this opinion indicates that it may be part of a larger cultural belief system.

It is important to examine the connection between the barriers of fear, denial, and a societal/cultural lack of awareness with the absence of open communication within African-American families. Participants stated they are limited in their knowledge of family disease risk because older generations do not want to discuss a personal or family history of health conditions. Issues of personal and family shame prevent relatives from discussing their health and the risk to develop disease. Participants also brought attention to the lack of family conversation about sickle cell trait status and history of sickle cell disease. They believe the absence of discussion causes future generations to have misconceptions about sickle cell that lead them to fear and deny their family history. The absence of family communication about sickle cell is also a factor in the ignorance among teenage parents. Participants stated that if parents never talk with their children about sickle cell trait status and the risk for disease then a lack of awareness is perpetuated into the next generation.

6.1.1 RECOMMENDATIONS

To address these barriers to education and awareness of sickle cell and newborn screening, it is essential to design an intervention which is nested within the larger Healthy Black Family Project. The Healthy Black Family Project has become enmeshed in Pittsburgh's African-

American community as it allows participants the opportunity to learn about health and make lifestyle changes with the support of the community. Within this framework, an intervention targeted at improving awareness about sickle cell disease would build upon the trust and rapport which has already been established with the community. Additionally, a sickle cell intervention could be implemented alongside other health promotion programs to allow participants the opportunity to learn and interact in a familiar community environment.

Based on the information gathered from this study, it is essential to continue the health education mission of the Healthy Black Family Project and to design interactive programs where participants can learn from experience. Guest speakers from the local community such as families who have a child with sickle cell disease should be invited to the Kingsley for community conversation. Therefore, adults and children can freely ask questions which they would not otherwise ask a speaker from outside the community. The personal story of the family and the individual with sickle cell would provide participants with the experience and observation to draw on as they attempt to understand the condition and explain it to other family members and friends.

The Family Health History Initiative should continue and expand to reach more families within the African-American community in order to provide individuals with an accurate assessment of disease risk and tools to open discussion about health within the family. During meetings with the genetic counseling student, participants should be specifically asked about their personal sickle cell trait status and family history of sickle cell trait/disease. If an individual has a family history of sickle cell trait/disease, the genetic counseling student can then provide education about the inheritance of the condition, the differences between disease and trait, and specific questions to ask a physician about carrier screening and sickle cell disease.

The Healthy Black Family Project also has the potential to extend the reach of its media work to include public health messages about sickle cell disease and create literature to be disseminated at health fairs, churches, and community clinics. Participants' concerns about the lack of sickle cell awareness among young parents and single teenage mothers are also valid. Therefore, interactive programs targeted at youth and young mothers should be developed to provide education on genetics, sickle cell, and other health topics. Currently, the Hemoglobinopathy Program of Children's Hospital of Pittsburgh is developing sickle cell education programs for young people in the Pittsburgh public school system.

In summary, a community approach to prevention needs to be implemented to address this community health problem. The Center for Minority Health and the Healthy Black Family Project in conjunction with the Hemoglobinopathy Program of Children's Hospital of Pittsburgh are in a unique situation to provide such an intervention and improve awareness and education about sickle cell disease. The Center for Minority Health has the framework in place to address sickle cell at the community level in the same way as they have so tirelessly worked to alleviate the burden of multifactorial disease in Pittsburgh's African-American population.

APPENDIX A: INSTITUTIONAL REVIEW BOARD APPROVAL LETTERS

University of Pittsburgh
Institutional Review Board

Exempt and Expedited Reviews

3500 Fifth Avenue
Suite 100
Pittsburgh, PA 15213
Phone: 412.383.1480
Fax: 412.383.1508

University of Pittsburgh FWA: 00006790
University of Pittsburgh Medical Center: FWA 00006735
Children's Hospital of Pittsburgh: FWA 00000600

TO: Stephen Thomas, Ph.D.

FROM: Sue R. Beers, Ph.D., Vice Chair S R Beers

DATE: November 16, 2006

PROTOCOL: Assessing the Attitudes and Beliefs of African-Americans Toward Newborn Screening for Sickle Cell Disease

IRB Number: 0610018

The above-referenced protocol has been reviewed by the University of Pittsburgh Institutional Review Board. Based on the information provided in the IRB protocol, this project meets all the necessary criteria for an exemption, and is hereby designated as "exempt" under section 45 CFR 46.101(b)(2).

- If any modifications are made to this project, please submit an 'exempt modification' form to the IRB.
- Please advise the IRB when your project has been completed so that it may be officially terminated in the IRB database.
- This research study may be audited by the University of Pittsburgh Research Conduct and Compliance Office.

Approval Date: November 16, 2006

SRB:kh

APPENDIX B: SUBJECT RECRUITMENT

B.1 FLYER ADVERTISEMENT

A HEALTHY BLACK FAMILY PROJECT RESEARCH STUDY

Participate in a Focus Group on Genetic Testing and Screening

We value your opinion.

Brought to you by:
Center for Minority Health of the Graduate School of Public Health
University of Pittsburgh

Share your opinions on topics related to genetic testing and screening in small focus groups at the Kingsley.

Participate in the focus group and receive a $25 gift card to Giant Eagle and refreshments.

Call Katie or Leah at 412-383-9822 for more information or to schedule a focus group.

Must be 18 & over and willing to commit approximately 2 hours of your time.

Principal Investigator: Stephen B. Thomas, PhD

B.2 TELEPHONE RECRUITMENT SCRIPT

Telephone Contact (Individuals who have completed family history)

Hello, my name is Katie Hoffman from the Healthy Black Family Project. I met with you at the Kingsley in (month) to draw out your family history. I was calling to invite you to participate in a research study that I will be conducting. This research study will be in the form of a focus group. A focus group is where several people come together to discuss a specific topic and share their opinions. I am interested in hearing the opinions of African-American community members on the topic of newborn screening for sickle cell disease and I would like to hear your thoughts about this topic. I hope to collect these opinions to help improve how information is transmitted to new parents about their child's sickle cell status. The groups will take place at the Kingsley and will take about 2 hours of your time. During this group, you will be asked to share your thoughts and opinions on specific topics and questions posed to the group. This research study will only be open to members of the Healthy Black Family Project. There are some possible risks with this research such as familiarity with other participants in the group which could cause discomfort in sharing your opinion. Your name will not be used to identify your responses, but there is a small risk of breach of confidentiality due to the audio recording of the session. These recordings will be kept in a locked file drawer at the Center for Minority Health and then destroyed upon the completion of this study. To thank you for your participation, we would like to offer some refreshments during the group and a $25 gift card to Giant Eagle. Your participation is voluntary and you can withdraw your participation at any time. Would you be interested in attending the group and offering your opinions? If you have any questions about this research, please contact me at 412-383-9822.

B.3 PARTICIPANT LETTER

Date

Dear Name,

The Center for Minority Health at the University of Pittsburgh would like to thank you for agreeing to participate in the focus group on Genetic Testing, Screening, and Research. We value your opinion and we are very excited you will be joining us for our small group discussion about topics related to research, genetic testing, and genetic screening. This letter serves as a confirmation for your scheduled focus group. Please contact Katie Hoffman or Leah Slattery if you will not be able to attend your meeting or have any questions.

Date:

Time:

Location: The Kingsley Association
6435 Frankstown Avenue
Pittsburgh, PA 15206

Contacts: Katie Hoffman and Leah Slattery
412-383-9822

Sincerely,

Katie Hoffman, BS
Graduate Student
Center for Minority Health
412-383-9822

Leah Slattery, BS
Graduate Student
Center for Minority Health
412-383-9822

APPENDIX C: MODERATOR'S GUIDE

I. **Introduction** (10 minutes)

A. Good afternoon/evening and welcome to our session. Thank you for taking the time to join our discussion on sickle cell and newborn screening. My name is Katie Hoffman and I am with the Healthy Black Family Project from the Center for Minority Health at the University of Pittsburgh. Assisting me is Leah Slattery who will be taking notes on the conversation during the focus group. Please begin by filling out the survey in front of you by circling your answers. We will begin when everyone has completed this form.

You have been invited because you are a member of the Healthy Black Family Project and your opinions on sickle cell and newborn screening are important to us. Our research team is trying to understand the attitudes and beliefs of African-Americans toward newborn screening for sickle cell disease. We would like to hear your opinions in order to determine how information can be better provided to new parents in your community about sickle cell and newborn screening. We need your input to make future newborn screening for sickle cell in Pittsburgh successful. We will ask you a number of questions and we need your honest thoughts and ideas. Please feel free to share your point of view especially if it is different from what others have said. There are no wrong answers, only different points of view.

Now, let me share some ground rules. We want to make sure that everyone feels comfortable expressing his or her own opinion. We ask that you are respectful of everyone by listening to one another and waiting until the person speaking has finished before you begin. Please do not have side conversations with your neighbor during the discussion. All of your comments are helpful and would provide us with useful information. We have placed cards on the table in front of you to help us remember your names during the focus group.

As you have probably noticed, we have a microphone on the table. We will be tape recording our discussion so that we don't miss any of your comments. To assure you of your confidentiality, we will only be using the names you selected for today and will not use any names in our final reports.

The group discussion will last about 2 hours, so feel free to enjoy your meal as we begin. We will have a 10-minute break and restrooms are located near the gym on the first floor. You should have already completed the brief survey and we will collect it before we begin.

II. **Assessing Knowledge of Genetics**

Question 1

Where do you receive trusted medical information?

Probe: From a Physician, Nurse, Pharmacist, Friend/Family member, TV, radio, newspaper, internet, seminar, etc.?

Probe: Why is this your most trusted source of medical information?

Question 2

What comes to mind when you hear the word "genetic" or "genetics"?

Probe: Where have you heard this word? Who has used the word?

Probe: How do you think "things" are passed down to or inherited from one generation to another? Why?

III. **Assessing Knowledge of Prenatal Testing and Newborn Screening for Genetic Conditions**

Question 3

When you hear the words "prenatal testing" what does it mean to you?

[Definition shared if no one answers: Prenatal genetic testing is testing performed on the developing baby while a woman is pregnant. It can be used to gather information about genetic health conditions in the baby.]

Probe: Are there benefits of prenatal testing? Why?

Probe: Are there risks of prenatal testing? Why?

Question 4

What are some reasons it would be helpful to know a child has a genetic condition before leaving the hospital?

Question 5

When you hear the phrase "newborn screening" what do you think of?

Probe: Where have you heard the phrase used?

Probe: Who has used it?

Probe: Under what conditions or circumstances has it been used?

IV. **Assessing Importance of Newborn Screening**

Question 6

Sometimes newborn babies are stuck in their heel to draw a small amount of blood for testing. Please tell me what you know about this type of test?

Probe: Why do you think it is necessary?

Question 7

What kind of information would you like to know before a child's blood is taken?

Probe: Who would you want to tell you?

Probe: When would you like to be told that information?

Probe: How would you want to receive that information (in print, face-to-face, video)? OR Would you like to receive it from some other source?

V. **Assessing Understanding of Sickle Cell Disease**

Question 8

Please tell me what you know about the condition called sickle cell.

Probe: (If they mention sickle cell disease and trait) What is the difference between the two?

Probe: Do you think one is more serious than the other? How?

Probe: How does someone get sickle cell?

VI. **Assessing Perceived Risk of Sickle Cell**

Question 9

Earlier we talked about how things are inherited or genetics. Do you think there is a risk in your family for a future child with sickle cell trait? Do you think there is a risk in your family of a future child with sickle cell disease?

Probe: Why or why not?

VII. **Assessing Opinions on Transmission of Sickle Cell and Newborn Screening Information**

Question 10

Scenario: It is three days after the birth of your child and you just found out your child has sickle cell disease. How helpful would this information be to you?

What if your child had sickle cell trait? How helpful would this information be to you?

Question 11

What is the best way for new parents to receive information about newborn screening for sickle cell?

Probe: When should this information be provided?

Probe: Who should provide this information?

Probe: Under what conditions should this information be provided?

Question 12

What information do you think is important for parents to know about newborn screening?

Question 13

How can the community help new parents understand information about sickle cell?

VIII. Closing

A. Thank you for your time and energy given to this discussion! It was a pleasure listening to your thoughts and opinions. We will need your signature to indicate you have received your $25 gift certificate to Giant Eagle before you leave.

APPENDIX D: SURVEY

We thank you for participating in this focus group. This survey is to gather information about the community's interactions with sickle cell. If there is a question that you do not feel comfortable answering, you can skip it and continue on. Please answer the following questions to the best of your ability. The survey should take approximately 10 minutes. We would like to thank you in advance for your willingness to participate in this study.

Section 1: General Information

1) What is your age?
__ __ age in years

2) What is your gender?
1 Male
2 Female

3) Are you Hispanic or Latino?
1 Yes
2 No
3 Don't know

3a) Which one or more of the following would you say is your race? **(Check all that apply)**
1 White
2 Black or African American
3 Asian
4 Native Hawaiian or Other Pacific Islander
5 American Indian, Alaska Native
6 Other [specify] ___________________________

4) What was the total household income from all sources last year?
1 Less than $10,000
2 Between $10,000 and $20,000
3 Between $20,001 and $35,000
4 Between $35,001 and $50,000
5 Between $50,001 and $75,000
6 Greater than $75,000

5) What is the highest grade or year of school you completed?
1 Grades 8 or less (Elementary)
2 Grades 9 through 11 (Some high school)
3 Grade 12 or GED (High school graduate)
4 College 1 year to 3 years (Some college or technical school)
5 College 4 years or more (College graduate or post-graduate)
6 Graduate level (Masters or PhD)

6) How would you rate your knowledge on genetics?
1 Excellent
2 Very good
3 Good
4 Fair
5 Poor

7) How would you describe your general health?
1 Excellent
2 Very good
3 Good
4 Fair
5 Poor

8) Do you smoke?
1 Yes
2 No

9) How would you describe your weight?
1 Underweight
2 Healthy weight
3 Overweight
4 Obese

10) Do you have one person you think of as your personal doctor or health care provider?
1 Yes, only one
2 More than one
3 No
4 Don't know / Not sure

11) Was there a time in the past 12 months when you needed to see a doctor but could not because of the cost?
1 Yes
2 No
3 Don't know / Not sure

12) Do you have any kind of health care coverage, including health insurance, prepaid plans such as HMOs, or government plans such as Medicare?
1 Yes
2 No
3 Don't know / Not sure

Section 2: Sickle Cell and Newborn Screening

13) Do you know your sickle cell trait status?
1 Yes
2 No
3 Don’t know

14) Do you know your partner/spouse’s sickle cell trait status?
1 Yes
2 No
3 Don’t know
4 Not applicable

15) Do you have children?
1 Yes
2 No

16) If you answered “Yes” to the previous question, do you know your child’s sickle cell trait status?
1 Yes
2 No
3 Don’t know

17) Do you know someone with sickle cell disease?
1 Yes
2 No
3 Don’t know

18) Do you know someone with sickle cell trait?
1 Yes
2 No
3 Don’t know

19) Do you know someone that was found to have sickle cell disease or trait by newborn screening?
1 Yes
2 No
3 Don’t know

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