KNOWLEDGE AND PRACTICE OF PREMARITAL GENETIC COUNSELLING AS A PREVENTIVE MEASURE AGAINST HAVING CHILDREN WITH SICKLE CELL DISEASE AMONG WOMEN IN LAGOS NIGERIA

BY

OLANIYAN, OLADELE HAKEEM

B.Sc CELL BIOLOGY AND GENETICS (UNILAG)

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CERTIFICATION

We certify that this work was carried out by Olaniyan H. Oladele, in the Department of Epidemiology and Medical Statistics, Faculty of Public Health, College of Medicine, University of Ibadan, Ibadan, Nigeria.

SUPERVISOR

Dr. Olufunmilayo I. Fawole, Reader

M.B.B.S. (Ib.), M.Sc (S. Africa), F.M.C.P.H. (Nig), F.W.A.C.P, Cert, Clin. Epid. (S.Africa)

Ag. H.O.D.

.....

CO –SUPERVISOR

Dr. Oyindamola B. Yusuf, Senior Lecturer

B.Sc. (Ib.), M.Sc. (Ib.), PhD (Ib.), Cstat (Uk).

DEDICATION

This project work is dedicated to all the past and present lecturers that committed themselves to making the department of Epidemiology and Medical Statistics a model among other departments in the faculty of Public Health and University of Ibadan in general.

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LISTS OF ACRONYMS

ANOVA	Analysis of Variance
FDA	Food Drug Administration
G6PD	Glucose-6-Phosphate Dehydrogenase deficiency
GF	Genotype Foundation
LASUTH	Lagos State University Teaching Hospital
LCDAs	Local Council Development Areas
LGAs	Local Government Areas
MST	Multistage Sampling Technique
NAFDAC	National Agency for Food and Drug Administration and Control
NGOs	Non-Governmental Organisations
NSGS	National Society of Genetic Counsellors
PGC	Premarital Genetic Counselling
PHC	Primary Health Centre
SAMI	Sickle Cell Advocacy and Management Initiative
SCA	Sickle Cell Anaemia
SCD	Sickle Cell Disease
TAHF	Temitope Awosika Help Foundation
TIF	Thalassaemia International Foundation
UK	United Kingdom
UNILAG	University of Lagos
USA	United States of America
WHO	World Health Organisation

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ABSTRACT

Nigeria currently has the highest incidence and prevalence of sickle cell disease (SCD) among all countries of the world and this continues to cause high morbidity and early death. SCD and other Haemoglobinopathies are a major public health concern among the black race. The aim of this study was to determine if women would still go ahead to have children with SCD after knowing they and their partners have sickle cell trait through blood genotype test.

This study was a descriptive hospital based study carried out in eight primary health care facilities across six Local Government Areas in Lagos state, Nigeria on 407 women. The main tools for this study were self and interviewed administered questionnaires containing both open-ended and closed-ended questions. The numbers of completed questionnaires were 407 out of the 420 administered, giving a response rate of 96.9%. Data were analysed by frequency counts, percentages, chi-square test and bivariate analysis at 5% level of significance.

The mean age of the women was 38.6±7.2, while for the children with SCD, it was 9.3±5.1

The study showed that 92.6% of the study participants accepted that it was important for unmarried individuals to meet a Counsellor before they plan a marriage. About 81.0% accepted that having a higher level of formal education increased the knowledge of SCD. The chi square test analysis showed a significant association between study participants' highest level of education and knowledge and practices related to genetic counselling (p=0.02), and between ethnic group and practices related to SCD genetic counselling (p=0.01). Study participants who had secondary school education as the highest level of education had an odds ratio of 0.90 and 95% confidence interval of 0.48-1.71 which was not significant for the bivariate analysis. While tertiary and above had an odds ratio of 0.66 and a 95% confidence interval of 0.33-1.30, which also was not significant.

About ninety percent of the study participants accepted that their life would have been different, better and stable if they had known about PGC and practiced it. About 83.0% of the women accepted that SCD is a serious disease and expressed that having children with SCD gave them a lot of psychological trauma. Forty two percent rejected that there was no reincarnation of a child that dies from SCD, 39.6% rejected that saying prayers daily could cure a child from SCD; while 81.3% rejected that it was the destiny of a child with SCD to have the disease.

This study showed good knowledge across employment status and marital status as 89.7% and 87.5% of the study participants responded correctly to questions asked on PGC. These results showed that the women would not go ahead with pregnancy or marriage if they had known that they and their partners were carriers of the sickle cell gene.

Public health education programmes on PGC should start right from the secondary schools as this will go a long way in making vital information available to the youths especially the females who are more vulnerable, in the future to make informed choices about pregnancy and marriage if they have sickle cell trait.

KEYWORDS: Premarital Genetic Counselling, Sickle Cell Disease, Reincarnation, Blood genotype test and Primary Health Care Facilities.

CHAPTER ONE

1.0 INTRODUCTION

1.1 Background

The history of non-communicable disease like sickle cell disease is as old as man himself. There is a global concern for the impact of haemoglobin disorders, mainly Sickle Cell Disease (SCD) on morbidity and mortality rates particularly in developing countries (WHO, 2008).

Since the 1980s the World Health Organisation (WHO) and the Thalassaemia International Federation (TIF) have recognised the need to support and facilitate the development and implementation of national policies for the management of thalassaemia and other haemoglobinopathies, mainly sickle cell disease. Since the late 1980s, the WHO, TIF and several sickle cell centres and organisations independently made an effort to promote the hereditary haemoglobin disorders as priorities on the health agenda of Member States, mainly as a consequence of the improvements achieved in the health infrastructure, public health services and the reduction in the annual infant mortality in affected countries of the developing world (WHO, 2008).

Sickle cell disease characteristic features make it most prevalent and suited among the populations of people in the sub-Saharan region of Africa. It mainly affects African Americans, with the condition occurring in about 1 in every 400 African American births (Kaye, 2006). The endemicity of malaria in this region is also one of the factors aiding this. SCD originates in tropical regions as a result of its advantage against malaria. It is predominant among people from African, Asian, Arabian and Mediterranean countries; nonetheless it is a global health problem because of population migration. SCD results in early childhood death if left untreated, and its

effect on the burden of health care is being recognised as a global issue in terms of chronic disease (WHO, 2006).

Inheritance of a single sickle haemoglobin (HbS) gene results in a healthy sickle cell carrier state, while the inheritance of the HbS gene from both parents, or HbS with another variant haemoglobin gene (e.g. HbC, Hbβ-thalassaemia) results in symptomatic SCD (Anie et al. 2010).

The prevalence of sickle-cell trait ranges between 10% and 40% across equatorial Africa and decreases to between 1% and 2% on the North African coast and less than 1% in South Africa (WHO, 2006). Although a single abnormal gene may protect against malaria, inheritance of two abnormal genes leads to SCD and confers no such protection, and malaria is one of the major causes of morbidity and death in children with SCD in Africa (WHO, 2006; McAuley et. al. 2010). This is because people who are carriers of the sickle cell trait are more resistant to malaria attack. Thus, mosquitoes and malaria fever thrive more here. Malaria is the evolutionary driving force behind sickle-cell disease, thalassaemia, glucose-6-phos-phatase deficiency, and other erythrocyte defects that together comprise the most common Mendelian diseases of humankind (Kwiatkowski, 2005).

Sickle cell disease (SCD) is one of the most important single gene disorders of human beings. In the United States, SCD affects about 72 000 people and 2 million are carriers (Creary et al. 2007). In Africa, more than 200 000 infants are born yearly with SCD (Makani et al. 2007). In the United States, mortality has decreased dramatically with newborn screening and better comprehensive care. The median age of death in patients with SCD in the United States is now 53 years for men and 58 years for women (Powars et al. 2005). However, SCD patients are still hospitalised frequently and by the fifth decade of life, 48% of surviving patients have documented

irreversible organ damage (Powars et al. 2005). Sickle cell anaemia (SCA) and Haemoglobin SC disease (HbSC) are the two most frequent types of sickle cell disease (SCD) in Cuba and as both are hereditary diseases with an autosomal recessive pattern of inheritance (Afolayan & Jolayemi, 2011). In Africa, where comprehensive medical care is less available, death in early childhood is usual (Makani et al. 2007).

Sickle cell anaemia (SCA) is a type of sickle cell disease in which there is a single point mutation on the β-globin gene (Afolayan & Jolayemi, 2011). SCA is caused by a structural variant of the major adult haemoglobin called S or Sickle haemoglobin (HbS), while HbSC is caused by the presence of two variants, one of them is sickle haemoglobin and another is haemoglobin C (HbC). These variants result from HbS and HbC allelic genes in beta globin locus in chromosome 11p 15.5. HbS allele differs from the normal allele A, in a single amino acid; at position 6 a valine replace a glutamic acid residue. In HbC allele, at the same locus lysine replace glutamic acid residue also at position 6.

Affected individuals with SCA are homozygous SS, because they inherit one HbS allele from each parent; while affected individuals with HbSC are heterozygous SC, because they inherit one HbS allele from a parent and one HbC allele from another one. These genes are quite common among African ancestry (Ruiz et al. 2007).

Occasional episodic acute complications characterise sickle cell disease (SCD) in childhood (Dampier et al. 2010). These acute complications often become more frequent in adolescents, who additionally begin to display variable degrees of chronic multi-organ dysfunction typical of adults with this disorder (Dampier et al. 2010). Pain resulting from sickle erythrocyte vaso-occlusion is the most common acute complication in children, with both pain and priapism potentially becoming frequently

recurrent complications. Avascular necrosis of hips or shoulder joints, and cardiopulmonary or renal dysfunction are examples of chronic complications that become increasingly prevalent in adolescents and young adults (Chiang & Frenette, 2005; Castro & Gladwin, 2005).

According to (Lagos health centre fights sickle cell disorder. *DAILY SUN.* Thursday, July 24, 2014: Page 40.) during the 2014 Annual sickle cell information and awareness programme in Lagos tagged: "Sickle Cell Disorder: The Survival Option", it was stated there that Nigeria presently has 40 million carriers of the sickle cell gene out of the country's one hundred and sixty million inhabitants. This figure was a result of statistics gotten on SCD and confirms the 25% prevalence of carriers among the population as a whole.

The chronic nature of SCD requires a life-long medical attention, expensive supportive symptomatic therapy, its specialised care, the associated high morbidity, reduction in life expectancy of the affected, poor school attendance, the potential risk of the development of drug addiction, especially to opiates, and its burden on the affected families all indicate that the condition is a major public health problem where ever its risk prevalence is high (WHO, 2006).

SCD is tackled using different preventive and treatment measures in recent times. One measure is premarital genetic counselling (Abioye-Kuteyi, 2009). Premarital genetic counselling (PGC) is also known as premarital counselling or simply genetic counselling. It is the expert advice given especially to intending couples in order for them to know if they have any inherited disorder that could be passed on to their offspring before their matrimonial union. It is like a co-joined twin to premarital screening. The result of premarital screening is what is used by genetic Counsellors to

advise would be couples or any other person about their inherited genetic disorder if they have any.

It is also very important that there should be an appreciation for the sensitivity of genetic information and the need for privacy and confidentiality while delivering genetic education and counselling fairly, accurately and without coercion or personal bias and with sensitivity to the patients' and families' culture, knowledge and language level (http://www.aafp.org/cg). PGC plays a major role in the health maintenance of an individual and possible prevention of different types of haemoglobinopathies.

Abdel-Meguid et al. (2000) stated that premarital genctic investigation plays very important role in the detection of many genetic disorders. They further said that premarital counselling is useful in order to explain the underlying nature of genetic diseases because when there is a lack of understanding, a variety of genetic diseases like SCD tend to surface.

Sickle haemoglobin (HbS) is the commonest and clinically significant haemoglobin structural variant (Piel et al. 2013). When a gene for a recessive disorder is present in a family, the diagnosis of the disease in a child serves as a marker of the extended family that is at increased genetic risk (Al Gazali, 2006).

Carrier screening and mutation identification are the comerstones prevention programme for the haemoglobin disorders in the developed world. The frequencies of these inherited characters have been extensively reported in various populations and ethnic groups in Nigeria (Erhabor et al. 2010).

However, prevention of the disease through carrier identification and genetic counselling has been advocated for in many of the different countries of the world and

remains the only realistic approach to reducing the impact of the disease and allow better use of available resources in the low income countries where the condition is most prevalent (WHO, 2006), like Nigeria. Since sickle cell disease can be controlled cost-effectively by programmes that integrate treatment with carrier detection and genetic counselling, WHO has recommended global development of these services (WHA, 2006a; WHA, 2006b). However, service development can be unexpectedly challenging, because it requires inclusion of genetic approaches in health systems (Modella & Darlison, 2008).

Therapies aim to minimise the effects of symptoms of SCD. Painful episodes (crises) are managed primarily with analgesia and hydration (De Montalembert, 2008). Analgesic pain control is usually in progressive stages and requires a variety of medications ranging from paracetamol for mild pain to morphine for severe pain. Blood transfusions may be required for stroke and other complications, and Hydroxyurea has also been found to be very effective in reducing the 'sickling' process and consequently the frequency of pain and hospitalisations experienced by patients (De Montalembert, 2008).

Some traditional health practitioners do claim that they have a cure for SCD. However, there has been no scientific proof to the veracity of their claim, or certification by the National Agency for Food and Drug Administration and Control (NAFDAC) in Nigeria to the efficacy and safety of their treatment. An example of one of these is the RUFUS NATURE GIFT HERBS NIGERIA LIMITED located in Ondo state, southwest, Nigeria. Likewise in the United States of America, there is a claim of a tea called RedCEL that sells at a dollar plus (www.inspire.com/...cell.../natural-remedies). It is believed that it tends to reduce the episodes of SCD. Also, the FDA (Food Drug Administration) there has not certified

this either. On the other hand, it was reported by Lagos health centre fights sickle cell disorder. *DAILY SUN.* Thursday, July 24, 2014: Page 40; during the annual sickle cell disease awareness programme in Onikan Health Centre, Lagos that a holistic approach to the treatment of SCD is very important especially in infants.

Bone marrow transplantation is a possible cure for SCD; however among other criteria, this requires a matched donor, and unfortunately is not feasible for all affected children Bhatia & Walters (2008) and also is very expensive.

1.2 Problem Statement

There is a high prevalence of children with sickle cell disease in Nigeria despite the awareness on checking of blood genotype before marriage. The relationship between religious/cultural beliefs and knowledge of sickle cell disease has not reduced the number of carriers of the disease in Nigeria. The annual increase of SCD due to neglect of premarital genetic counselling by intending couples because of excuse of respect for certain cultural values is worrisome. The lack of updated and reliable epidemiological data including incidence, carrier and prevalence rates as well as the clinical spectrum of SCD in Nigeria. The low level of recognition of SCD by government and also by some Non-governmental health agencies and funding bodies. Religious and cultural beliefs in Nigeria coupled with literacy problems are seriously hindering promotion of significant aspects of the control programme including spreading of community awareness and clear understanding of the nature of SCD, its prevention and management (WHO, 2006).

These disorders occur widely across the world, and both their natural history and prevalence vary considerably between the different regions of the world. Their prevalence may also vary (i) between countries of the same region; (ii) between different areas within a country and (iii) even between different medical centres within the same area (WHO, 2006). Despite the laboratory and clinical advancements towards their effective prevention and clinical management, increased numbers of annual affected births and high rates of mortality and morbidity are still observed in the majority of affected countries of the developing world (WHO, 2006). The statistics on SCD include: Around 7% of the global population carries an abnormal haemoglobin gene; 300,000-500,000 children are born with clinically significant haemoglobin disorders annually; about 80% of affected children are born in developing countries; about 70% are born with Sickle Cell Disease (SCD) and the rest with Thalassaemia Syndromes;50-80% of children with SCD die each year in low and middle income countries; 50,000-100,000 children with thalassaemia die each year in low and middle income countries (WHO, 2006).

On this premise, this study will assess knowledge and practice of premarital genetic counselling as a preventive measure against having children with sickle cell disease, among women in Lagos, Nigeria.

1.3 Justification

Of the sickle cell control strategies, premarital genetic counselling is increasingly practised in many countries of the world (Alao et al. 2009). Methods of preventing new haemoglobinopathy births include premarital screening and genetic counselling, prenatal diagnosis, preconceptional diagnosis and implantation of normal embryos

after in vitro fertilisation, and in utero therapy using stem cell transplantation (WHO, 2006).

Prevention of the disease through carrier identification and genetic counselling remains the only realistic approach to reduce the impact of the disease and allows better use of available resources in the low-income countries where the condition is most prevalent (Abioye-Kuteyi, 2009).

Programmes of population screening and genetic counselling can have a major impact on the birth rate of children with SCD and other genetic diseases (Al Arrayed, 2005). The prospective control of SCD by heterozygote detection through premarital screening, which is vital to the identification of the couples at risk, is of utmost importance. The success of such a programme to a large extent depends on the SCD knowledge in the community, the understanding of the full consequences of having a child with SCD, and people's attitude towards genetic screening and counselling (Abioye-Kuteyi, 2009). In 2004, the Bahrain Government passed a law requiring all couples planning to get married to undergo free premarital counselling (Al Arrayed & Al Hajeri, 2005).

Currently, there is no known cure for Sickle Cell Disease (SCD). Genetic testing and counselling have become standard of practice in many developed countries to confirm the diagnosis of hereditary diseases. Certain genetic conditions, such as sickle cell disease and glucose-6-phosphate dehydrogenase deficiency (G6PD), are checked regularly on all newborns in developed countries so that treatment can be initiated promptly (Bodamer et al. 2007).

It has been reported that the rise in the number of old and new cases between 2011 and 2013 of SCD as recorded by the sickle cell clinics in Onikan Health Centre, Lagos raises the need why there must be a way in which births from SCD should be

prevented (Lagos health centre fights sickle cell disorder. *DAILY SUN*. Thursday, July 24, 2014: Page 40). There has been a percentage rise of 150% of children with SCD as reported by Onikan Health Centre between 2011 and 2013 which is quite worrisome despite the increase in claims of different cures for the disease. Also, there is dearth of sickle cell centres/clubs and education in Nigeria.

Ibekwe, (2012)reported that a clinic in one of the rural areas of Edo state regularly has 50 new cases of sickle cell disorder patients weekly. This means that many of the undiscovered patients might have died. This gave a strong indication that sickle cell disease needs the urgent and utmost approach to finding the appropriate public health solutions to its alarming incidence and prevalence in Nigeria.

Ahmed, 2010 stated that genetic disorders and congenital abnormalities occur in about 2%-5% of all live births, account for up to 30% of paediatric hospital admissions and cause about 50% of childhood deaths in industrialised countries. These figures are even higher in developing countries like Nigeria, where the medical record is poor. Similarly, Memish & Saeedi(2011)noted that genetic disease affects as much as 5% of the world's population constituting a major public health problem in many parts of the world.

Access to beneficial drugs like Hydroxyurea, Neonatal Screening, Penicillin Prophylaxis and Prophylactic Immunisation even when available are almost out of reach of an Average man. With adequate genetic counselling, the high frequency of sickle cell anaemia disease could be prevented in Nigeria (Adeyemo et al. 2007). Therefore for Africans and in particular Nigerians, an important approach for controlling the disease is preventive, and this depends upon education, the detection of carriers, genetic counselling, prenatal screening for foetal genotype done in couples

who are both carriers and newborn screening for sickle cell genotype (Olatona et al. 2012).

Knowledge of the citizenry of a nation about SCD constitutes an important variable that influences the acceptability, practice and success of premarital genetic counselling (Alao et al. 2009). Understanding the culture of the participants is important to the appropriate delivery of genetic testing and counselling (Futter et al. 2009; Penn et al. 2010).

Sickle cell anaemia contributes the equivalence of 5% of under-five deaths on the African continent, more than 9% of such deaths in West Africa, and up to 16% of under-five deaths in individual West Africa countries (WHO, 2005). Among the different and many resulting complications of sickle cell disease is stroke. SCD could cause damage of blood vessel in the brain of children with the disease, whereby they in turn suffer speech impairment, difficulty with speaking and understanding or even trouble with reading and writing (Ibekwe, 2012). These in summary are the hallmark and features of stroke. Therefore, this further reveals that SCD is a disease of serious public health concern.

Genetic education, which is premarital genetic counselling, provides individuals and society with the autonomy to make informed decisions. Informed decisions are important for any genetic test including carrier screening so that individuals can make the best decision about testing for themselves. Genetic conditions such as sickle cell disease, cystic fibrosis, and Tay-Sachs disease are some of the commonest conditions where genetic education has had public health significance. Educating at risk populations about carrier status, providing them with the information to make informed decisions, and educating them on the benefits and limitations of testing can

inform these individuals. The result creates autonomous individuals who can make informed decisions (Gilani et al. 2007).

1.4 General and Specific Objectives

1.4.1 General Objective

To determine knowledge and practice of premarital genetic counselling as a preventive measure against having children with sickle cell disease among women.

1.4.2 Specific Objectives

1. To determine the proportion of women who had ever had knowledge of genetic counselling for SCD.
2. To assess the association between respondent's socio-demographic characteristics and knowledge of SCD.
3. To examine the association between respondent's socio-demographic characteristics and knowledge of PGC
4. To determine the association between respondent's socio-demographic characteristics and practice of PGC
5. To determine the proportion of women who have religious/cultural beliefs about SCD.

CHAPTER TWO

2.0 LITERATURE REVIEW

2.1 Historical Background

2.1.1 Research and Studies

The different scientific works and researches in the field of public health in the last decade have been progressively showing the importance and relationship between knowledge and practice of premarital genetic counselling and sickle cell disease. Jastaniah (2011) reported that early childhood death usually in Africa is due to SCD and also stated that the first case of SCD in Saudi Arabia was reported in the Eastern province of the country in the 1960s. These likewise have attracted a lot of attention in present time regarding the study of SCD. In a way to go merely beyond the reason for this study, past and recent works will be looked into. Deaths from Sickle Cell Disease complications occur mostly in children under five years, adolescents and pregnant women (WHO, 2008).

Adeyemo et al.(2007)reported that Western medical care was formally introduced into Nigeria in the 1860s. It was when the Sacred Heart Hospital was established by the Roman Catholic Missionaries in Abeokuta, Southwest Nigeria. They stated that presently, there is a general acceptance of modern medical care among Nigerians.

Anie et al. 2010 stated that the current or historic exposure to Plasmodium malaria infection in West Africa will lead to continued transmission of the HbS gene as carriers of sickle cell trait appear to be protected from malaria associated deaths and have improved survival. On the other hand, Jastaniah (2011)reported that SCD gene

initially were thought to spread by migration, but later indication of sickle gene mutation developed independently and spontaneously at least five times with four major halotypes ('Senegal', 'Benin', 'Bantu' and 'Cameroun') and the fifth, the 'Arab-Indian'. Memish & Saeedi, 2011; Jastaniah, 2011; Al Gazali et al.(2005);Al Arrayed & Al Hajeri, (2010);Al Arrayed & Al Hajeri, (2012) are of the opinion that more than 55% of SCD prevalence is as a result of the common consanguineous marriages practiced in the Arab world.

Whitehead et al. (2010) opined that most of the crises faced by the parents of children with SCD could have been prevented or better managed if their parents had knowledge of newborn screening. Methods of preventing genetic diseases include pre-marital screening and genetic counselling, prenatal diagnosis, preconception diagnosis, implantation of normal embryos after in-vitro-fertilisation and in-utero therapy using stem cell transplantation (WHO, 2006). Studies on SCD have focused on the causes, prevention, treatments, incidence and prevalence in the recent past. Abdulrahaman et al. 2013; Modella & Darlinson, 2008; Olatona et al. 2012 all agreed that there are over 300,000 global annual births of children with sickle cell disease. Jastaniah, 2011 found out that 200,000 of such births occur in Africa alone. Abdulrahaman et al. 2013 reported that 7% of the world's populations are carriers of the sickle cell traits (that is 269 million global carriers). Memish & Saeedi, 2011; Olatona et al. 2012 reported that 5% of the global populations have haemoglobinopathy genes. A work on genetic counselling has been studied by (Al Arrayed & Al Hajeri, 2012). Abioye-Kuteyi, 2009 found out that across the equatorial regions of Africa, healthy carriers of sickle cell trait constitute 10-40% of the population there. While in Northern Africa, it is 1-2% and less than 1% in Southern

Africa. He further stated that the carrier frequency for West African countries like Ghana and Nigeria is 15-30% and it is 45% in some areas in East African countries such as Uganda and Tanzania. Haemoglobin comprises four globin chains: foetal haemoglobin (Hb F) has two α and two gamma chains (α2γ2) and adult haemoglobin (Hb A) has two α and two β chains (α2β2). The α-globin and β-globin cluster (on chromosomes 16 and 11) and control globin-chain production. Due to spontaneous mutation, haemoglobin gene variants are present at low prevalence (carriers 1–1.5/1000) in all sizeable populations (Modell et al. 2007). Most haemoglobin gene variants are rare and many are harmless, but some are common because carriers are less likely than others to die from falciparum malaria. The most common of such variant, α plus (α+) thalassaemia is usually harmless (Odunlade, 2005).

However, people who inherit combinations of haemoglobins S, C, E, D Punjab, β thalassaemia, or α zero (α0) thalassaemia may have a serious haemoglobin disorder (Odunlade, 2005). The resemblance between thalassaemia and iron deficiency can confuse the diagnosis of either disorder (Wonke et al. 2007).

While the HbC (mild sickle cell disease) trait is largely confined to the Yoruba people of south-western Nigeria, the prevalence of sickle cell anaemia is about 20 per 1,000 births meaning that Nigeria alone, accounts for about 150 000 children with sickle cell anaemia annually (Abioye-Kuteyi et al. 2009). Sickle cell disease (SCD) is an autosomal recessive genetic blood disorder characterised by red blood cells that assume an abnormal, rigid and sickle shape (Oludare & Ogili, 2013). Afolayan & Jolayemi, 2011 stated that sickle cell disease (SCD) is one of the commonest but preventable inherited diseases. It is a disease that affects the red blood cells and is a lifelong ailment. Sickle Cell Disease (SCD) is the commonest genetic disease

worldwide and includes disorders affecting the structure, function or production of haemoglobin (Ohls & Christensen, 2004). There is a substitution of glutamate with valine in position 6 of the beta globin (β 6Glu→Val) (Serjeant & Serjeant, 2001).

The disease is expressed when Haemoglobin S (HbSS) is inherited from both parents, the homozygous child or haemoglobin SS (HbSS) suffers from sickle cell anaemia (SCA), while the heterozygous child or Haemoglobin AS (HbAS) is a carrier of a sickle cell trait (SCT). HbSS is the commonest pathological haemoglobin variant worldwide and majority of children born with SCA die before reaching five years of age (Weatherall et al. 2006). Sickle cell affects all races of the world; it affects the people of tropical Africa, Mediterranean Sea, Middle East and South India. It has contributed significantly to the high childhood mortality rate. Nigeria accounts for 50% of sickle cell disease (SCD) births worldwide and about 2.3% of her population suffers from SCD with 25% of Nigerians being healthy carriers (Afolayan & Jolayemi, 2011).

The unanimous verdict is that SCA or simply put SCD has no cure. However findings show that extracts of a local Nigerian plant *Fagara zanthoxyloides* called "Orin Ata" in Yoruba, a type of chewing stick and some nutritional supplements have shown alleviating effects on the painful symptoms of this genetic disorder (How herbal, nutritional cocktail boost health in sickle cell patients. The Guardian, Lagos, Nigeria.10.06.2008). Imaga (2013) further stated that research into the anti-sickling properties of medicinal plants has been rewarding.

Abdulrahaman et al. (2013); Alao et al. (2009); Anie et al. (2010); Abioye-Kuteyi,

(2009) reported a prevalence of 20 per 1,000 births, resulting in 150,000 children born annually in Nigeria with SCD genotypes. Adeyemo et al. (2007) reported that SCD contributes equivalent of 5% under-five deaths in Africa, 9% of this in West Africa and up to 16% of under-five mortality in individual West African countries. While many of the reviewed works used self- administered questionnaire; interviewed-based questionnaire was used only by (Al Arrayed & Al Hajeri, 2012; Nnaji et al. 2013).

2.1.2 Research Statistics

According to Modella & Darlinson (2008), 13% of SCD is as a result of migration; 85% of those with the disease and 70% of all affected birth occurs in Africa; haemoglobin disorders are significant health problems in 71% of 229 countries; 3.4% of annual under five deaths are as a result of the disease; haemoglobin disorders are endemic in 60% of 229 countries and that HbS accounts for 40% of carriers and causes over 80% of disorders worldwide because of its high localised prevalence. They both further stated that there are 948,000 new carrier couples with 1.7 million pregnancies to carrier couples and that over 9 million carriers become pregnant annually with risk of partner carriers from 0.1-40% and a global average risk of partner carriers of 14%.Abioye-Kuteyi et al. (2009) reported a response rate of 91%. Omuemu et al. (2013) reported a 95.2% response rate, while 93.3% was reported by (Oludare & Ogili, 2013).

Only Afolayan & Jolayemi (2011) reported a response rate of 100%. The response rate for this work was also reported. Al Arrayed & Al Hajeri (2010) reported that more than 90% of their study participants had heard of SCD, with 89% knew that it could be diagnosed by a simple blood test and

84% have the knowledge that SCD is a hereditary disease. Seventy two percent said that it could skip generation.

Animasahun et al. 2009 reported that 98.7% of 403 health professionals in a study carried out in Lagos had heard of Sickle Cell Anaemia, while (Omuemu et al. 2013) reported 95.8% in similar study carried out among 400 undergraduate students of the University of Benin. This work also found out the frequency knowledge of SCD of study participants

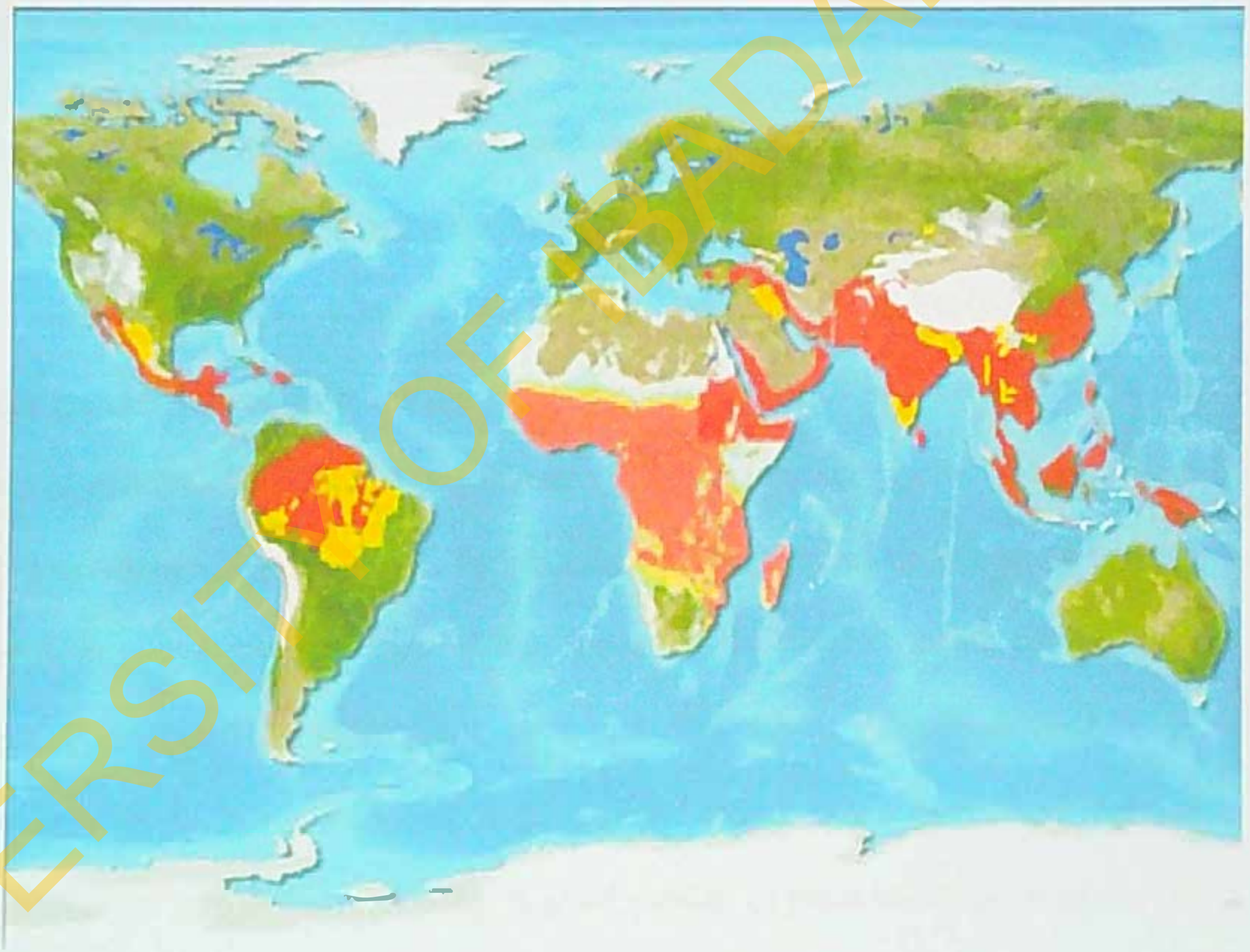

Figure 1: Areas with Sickle cell disease and other Haemoglobinopathies (Gustafon, 2006).

NOTE: The Areas in red have increased incidence and prevalence of SCD

In Africa, the highest prevalence of sickle-cell trait occurs between latitudes 15° north and 20° south, ranging between 10% and 40% of the population in some areas (Oludare & Ogili, 2013).

The term 'Ahotutuo' is used by the 'Twi' people in Ghana to describe SCD; and the closest English translations would include 'body biting', 'body chewing', and 'beaten up' (Anie et al. 2007).

In Africa, cultural factors are particularly relevant to these problems because of beliefs and traditional practices (Anie et al. 2010). In Nigeria, beliefs are usually influenced by cultural and religious values, which influence health behaviour such as coping strategies.

In some years past among the Yorubas and the Igbos, SCD was perceived as an evil due to the high level of ignorance then and certain cultural beliefs like taboo. Children born with this disease are called 'Abiku' and 'Ogbanje' respectively in these two ethnic groups. For example, among the Igbo communities, SCD is believed to be the result of malevolent 'Ogbanje' (reincarnation) that is repeated cycles of birth, death and reincarnation (Nzewi, 2001). People of these two cultures do not know that the disease was as a result of each of the parents of the children being carriers of a defective haemoglobin gene. They believe it was a sort of reincarnation. Such children with SCD are subjected to all sorts of torture when there is an episode (crisis), which includes skin burning. A good number of these children die at the end of the day. Up to this present time, some people in these two tribes still believe in this fallacy. People with SCD and their families undergo psychological trauma frequently. This majorly is as a result of the impact of pain and symptoms on their daily lives, and society's attitudes to SCD and those affected (Nzewi, 2001).

Whitehead et al. 2010 opined that most of the crises faced by the parents of children with SCD could have been prevented or better managed if their parents had knowledge of newborn screening or genetic counselling and practiced it.

While in 2006, the Genetic Counselling Task Force of the National Society of Genetic Counsellors (NSGS) provided a new definition of genetic counselling as – the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease (Resta et al. 2006).

This process integrates the following: interpretation of family and medical histories to assess the chance of disease occurrence or recurrence, education about inheritance, testing, management, prevention, resources, research and counselling to promote informed choices and adaptation to the risk or condition (Adeyemo et al. 2007).

Sickling occurs because of a mutation in the haemoglobin gene that decreases the cells' flexibility, resulting in vascular-occlusive complications such as painful episodes at extremities and chest, stroke, priapism, liver disease, leg ulcers, spontaneous abortion and renal insufficiency (Oludare & Ogili, 2013). The genes that have been altered are referred to as mutant genes. Gene mutation can cause loss, addition or rearrangement of bases in the gene (Adeyemo et al. 2007). The mutation takes different forms and these include duplication, insertion, deletion, inversion or substitution of Bases (Odunlade, 2005).

The trauma experienced from sickle cell disease cannot be over emphasised (Afolayan & Jolayemi, 2011). Kabiti, 2008 stated that patients with sickle cell disease may have recurrent illness and be hospitalised due to various complications of the disease. The cost implication and mental agony of the parents in particular are of significant note. He further identified physical deformities of the patients such as

frontal bossing, protruding abdomen, thin extremities and gnathopathy. Other impacts are: absenteeism from school due to frequent illness, difficulty in getting marital partner, damaging effect of the stigma of being a child with SCD on his/her psyche, reduced chances of getting pregnant for females, fear of frequent illness during pregnancy (females), increased abortion rate, anxiety on the possible genotype of the baby in-utero, needs for ante-natal diagnosis and its attendant risks, need for therapeutic abortion in case of an unfavourable genotype attendant risk, persistent state of anxiety and tension because the individual can get ill at any time and psychosocial trauma of the knowledge of imminent death (Kabiti, 2008). More importantly, the victims may be unable to realise/actualise his dreams in life and there is denial of some rights and privileges such as work, freedoms, marriage and sex.

Ibekwe, 2012 found out that before early screening for sickle cell disease and the use of preventive antibiotics in children, 35% of infants with sickle cell disease die from various infections and that about 30% of patients with disease have pulmonary hypertension. Pulmonary hypertension is a serious and potentially deadly type of hypertension whereby the pressure in the arteries of the lungs increases.

Kabiti, 2008 further stated that the cost of daily maintenance of sickle cell patient is colossal in terms of drugs, nutrition, prevention of crisis, hospitalisation and that the affected individuals or families suffer a burden of anxiety, frequent illness, excess mortality rates, ignorance and lack of appropriate health services and research.

Optional management of painful crises requires a team that includes a Haematologist, a family Physician that probably has the history of the patient during previous crises, Nurses, Psychiatrist, Physiotherapist, Pain Specialist and Social workers and the presence of a cohesive family unit to prevent psychological instability and development of chronic pain syndrome (Ibekwe, 2012).

Alswaidi et al. 2012 reported that despite results from a screening program for sickle cell disease and β-thalassemia in Saudi Arabia, 90% of couples at risk of having affected children in Saudi Arabia still decide to marry. (Al Arrayed & Al Hajeri, 2012) found out in a study of newborn screening in Bahrain between 1985 and 2010 that incidence of affected newborns with sickle cell disease declined by 75%. In the study, cord blood of infants was collected at birth and electrophoretic technique was used to detect haemoglobin fractions present in the cord blood samples.

While some studies have focused on the different preventive measures against SCD, Others have focused on premarital screening and premarital genetic counselling. Accordingly, Alao et al. 2009 suggested that premarital genetic counselling is increasingly practiced in many countries of the world. Many of the reviewed works carried out studies on genetic screening, awareness, knowledge and practice of genetic counselling. All the studies used a mixed population of males and females. This study however used a homogenous population of women. This removed the bias that would have happened if the husbands of the women interviewed were present. This study is different and unique from previous works because it provided data and information collected from the independent woman as well as data on their children with the SCD. Also, information was gotten on children who died as a result of SCD from the study participants.

Alao et al. 2009; Abioye-Kuteyi et al. 2009; Al Arrayed & Al Hajeri, 2010 and Olatona et al. 2012 studied the level of knowledge of SCD among Government workers and youths in Nigeria and Bahrain. They found out that more than half of their study participants showed poor knowledge of SCD.

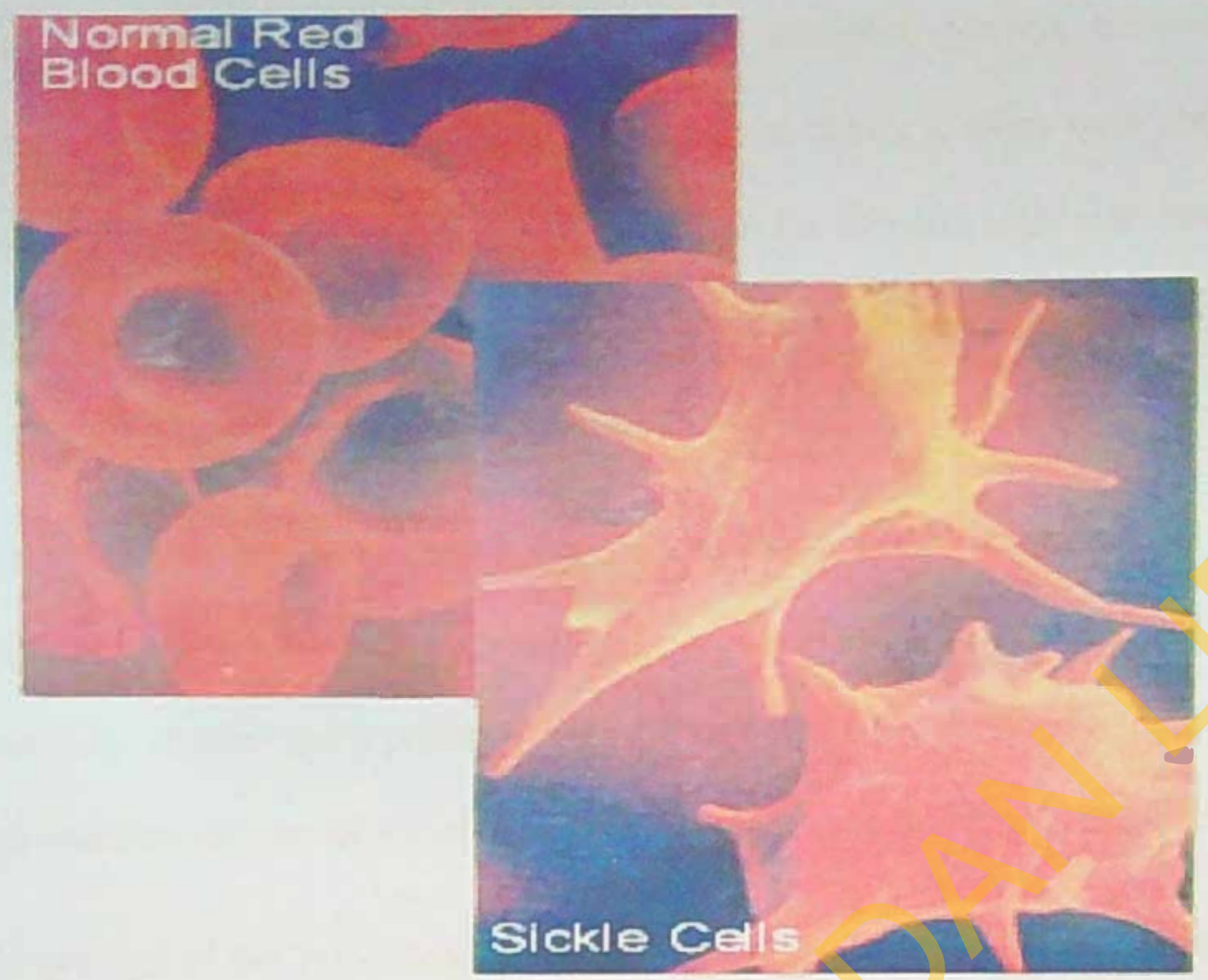

Figure 2: Normal Red blood Cells and Sickle Red Blood Cells (www.samc.com, 4/2/2006)

2.2 Knowledge of Sickle Cell Disease

2.2.1 Assessment of Knowledge

According to Olatona et al. 2012, knowledge of SCD was determined using a 22 point scale of less than or equals to eight (≤8) for poor; between nine and thirteen (9-13) for fair and fourteen to twenty two for good knowledge respectively. However in this study for the Likert scale questions; strongly agree and agree answers were coded as 1, while indifferent, strongly disagree and disagree answers were coded as 0. The respondents' knowledge of sickle disease was assessed on a 10 point scale similar to the assessment of (Gustafon, 2006) where answers to the knowledge of SCD and sickle cell trait (SCT) and acceptance of genetic testing and genetic counseling

questions were transformed into dichotomous variables. Correct answers to the knowledge questions were coded as 1, incorrect or unsure answers were coded as 0. A total point score of five points or less (≤ 5) on the 10 point scale represented poor knowledge of SCD, while a total point score of between four and ten (6-10) represented good knowledge on the 10 point scale. .

Animasahun et al. 2009 reported that Medical Doctors had a statistically significant knowledge of the best time of detecting the genotype responsible for SCD. They also stated that 85% of health professionals had been involved in the management of sickle cell anaemia and that 24.3% of them knew most of the complications of the disease.

Knowledge of the respondents on PGC was measured with a Likert scale of five points of: strongly disagree, disagree, indifferent, agree and strongly disagree. The level of knowledge of premarital genetic counselling was assessed on a seven point scale of poor and good knowledge similar to (Gustafon, 2006) where assessment was done on poor and good knowledge. This is different from that of (Olatona et al. 2012) where a scale was developed for poor, fair and good knowledge. Study participants who scored 3 points or less (≤ 3) were considered to have poor good knowledge and practice of premarital genetic counselling while those who scored between a total of four and seven points (4-7) of the 7 point scale were considered to have good knowledge and practice of PGC in this study.

Omuemu et al. 2013 reported a higher knowledge frequency of SCD in females than their male participants. This was not statistically significant. This differs from the study of Animasahun et al. 2009 where knowledge score of female health professionals was statistically significant. However this study further broadened the claims from both studies as participants for this study are a homogenous population of

women who were not health personnel as in the latter study. Whereas, Al Arrayed & Al Hajeri (2012) reported a higher knowledge in females and it was found to be statistically significant. A further limitation of previous reviewed works was that no data were collected from respondents on mortality of any of their children from SCD. This study got information on death of child/children from SCD from the participants if such had happened.

Abioye-Kuteyi et al (2009) reported that 95% of study participants had favourable attitudes towards premarital screening. Animasahun et al. (2009) stated that almost all study participants knew about screening. Eighty six percent of the respondents studied by Adeyemo et al. (2007) had knowledge of genetic disease like SCD, while 30.3% of the same participants had been exposed to genetic counselling. Omuemu et al. (2013) reported 78.9% awareness on genetic screening and a high percent in the level of acceptability of genetic counselling among participants. Oludare & Ogili (2013) reported that 86% of their study participants had positive attitude to premarital genetic counselling, while 65% had practised things related to premarital genetic counselling. Olatona et al. (2012) stated that almost half of their respondents were unaware of the availability of premarital screening for sickle cell disease in Nigeria.

Alao et al. (2009) stated that their respondents had 48% good knowledge of SCD in a study among 300 students of Benue State University, Nigeria. Omuemu et al. (2013) reported 78.9% in a similar study among 400 students of University of Benin, Nigeria. Eighty two percent of participants claimed to have SCD knowledge in a study done by Owolabi et al. (2011) among secondary school students in Abuja. This study also

found out the frequency of levels of SCD and PGC knowledge of participants from poor to good.

In a study in Bahrain, between 2006 and 2007, Al Arrayed & Al Hajeri (2010) reported that 51% of participants did not know the prevalence of sickle cell disease in Bahrain. However, there was a wide and large acceptance and good knowledge of SCD among the public there. Of the 2,000 respondents studied, 583 were professionals; 406 were students, while 618 were unemployed. Out of the overall respondents, 48.8% were school students; 45.5% were university graduates; 4.6% were postgraduates and illiterates were 1.1%. In addition, 53% were single, while 47% were married. Eighty percent of participants agreed that SCD had a negative impact on a child's school performance, while two-third of them said that certain food could trigger episodes (crises) of SCD.

Al Arrayed & Al Hajeri (2010) found out that respondents' level of knowledge and gender was significant and that professionals gave the most correct answers about SCD knowledge and it was statistically significant. Also, respondents with higher job status showed significant knowledge. This study also found out if women's level of education, monthly income and employment status were significant to PGC and SCD knowledge.

Odeiola et al. (2013) stated that difference in attitude towards premarital genetic screening was not statistically significant; difference in attitude due to religion was not statistically significant ($p = 0.689$) and that difference in attitude due to course of study was not statistically significant ($p = 0.585$) among polytechnic students in a study in Osun State, Nigeria.

2.2.2 Knowledge frequency based on socio-economic factors

All the respondents Abdulrahaman et al. 2013 interviewed were African descent resident in Sokoto and were mainly Hausas, while 75% of those interviewed by (Anie et al. 2010) were Yorubas. Schwartz et al. 2009 had 97.5% of their study participants to be of African-American origin. A sample of a hundred percent African-Americans was studied by (Palermo et al. 2008).

Odelola et al. 2013; Afolayan & Jolayemi, 2011; Oludare & Ogili, 2013 used a structured questionnaire to collect data from study participants on a four-point scale format of knowledge scores of strongly agree, agree, disagree and strongly disagree. A semi-structured questionnaire including questions on a five-point Likert scale of strongly disagree, Agree, Indifferent, Agree and strongly agree was used in this study.

Abioye-Kuteyi et al. 2009 reported that 69.7% of respondents had tertiary education as their highest level of education. They stated that 86.7% of respondents and 74% of their partners have had sickle cell screening before the study, while 25% of married and engaged study participants did not know their partners' sickle cell status. One third to two third of respondents stated that they will continue with their relationship even if they or their partners have sickle cell trait. In the work of (Animasahun et al. 2009), 33.7% of participants felt that genotype identification should be done during childhood.

2.3 Study type

This work was also hospital based study. Most all the studies were hospital based. Only Oludare & Ogili, 2013; Afolayan & Jolayemi 2011; Anie et al. 2010; Animasahun et al. 2009; Lattimer, 2010 used population based studies. This showed a

preference for hospital based study by many of the researchers. However, getting a large sample size could sometimes be a major challenge in hospital based studies especially for an uncommon disease like SCD. This is because some cases do not come to the hospital due to low self-esteem, ignorance, poverty and fear of disclosing information about themselves.

2.4 Socio-cultural Factors aiding Sickle Cell Disease

Barriers, limitations and challenges facing prevention of sickle cell disease were studied Vis a Vis premarital genetic counselling. Afolayan and Jolayemi, 2011 reported that of all the sickle cell carrier parents of children with SCD interviewed in a study in Irepodun Local Government in Kwara state, 87% of them stated that they regretted having children with SCD. They further said that the reasons they went ahead with their marital union were due to lack of enlightenment programme on sickle cell disease; little or no genetic counselling; ill disposition to premarital genotypic screening; inadequate medical facilities for adequate genotype test in rural areas; gross misrepresentation and wrong perception of SCD due to cultural beliefs; lack of knowledge of people on SCD and a nonchalant attitude to the results of screening due to love and interest in one's partner.

Oludare & Ogili, 2013 cited ignorance and poverty as the major reasons for the high prevalence of sickle cell disease. Adeyemo et al. 2007 opined that the practice of premarital genetic counselling is poor and that there is inadequate provision of genetic services in Nigeria. They stated further that most hospitals do not have genetic unit, while private hospitals carry out premarital genetic counselling occasionally. On the part of Memish & Saeedi, 2011, cultural beliefs were found to be the main obstacle in preventing SCD. Al- Gazali et al. 2006 said that there is lack of public health

measures directed at prevention of genetic disorders in low income countries. They also said that public health measures are affected by cultural, religious and legal limitations in some of such countries. Jastaniah, 2011 stated that environmental factors such as infections, malnutrition and unfavourable socioeconomic status negatively influence SCD and rate of survival of children with the disease. While 86% of SCD patients said that they were less involved in decisions about their medical care according to (Lattimer, 2010), a large percentage of them said that doctors and nurses were afraid to discuss about their health with them. 64% of participants said that hospital staff gave conflicting information about their status.

Furthermore, Amr et al. 2011 reported that there is stigma and fear of disclosure among females who have SCD. He stated that there are limited studies on rural/urban differences in the quality of life among patients with SCD in Saudi Arabia; limitations in studies on differential levels of support, coping styles and perceptions of illness in urban-rural among patients with SCD; low income among patients with SCD as well as other limitations which include low participation of females with SCD due to cultural factors restraining females not to disclose their sickle cell status, thereby making them seek treatment in private health facilities. This study found out if such restraints of females from knowing their sickle cell status is common or exist after the analysis of the collected data based on the knowledge of participants about SCD

Al Gazali, 2005 stated that the problems facing genetic counselling in the United Arab Emirates were due to absence of a good national database, deficiency of genetic services and absence of preventative alternatives to carrier couples. Anie et al. 2010 analysed various issues regarding sickle cell disease which included knowledge and

understanding; beliefs; pain experience; coping; health service utilisation; social issues including education and employment as well as quality of life.

While many of the studies did not report the effect of cultural factors on having a child with SCD, some of the studies reported the perception of SCD from the cultural and societal view. Olatona et al. 2012 reported that there is a myth, misinformation and stigmatisation of person with SCD. Anie et al. 2010 also stated that there is a negative perception of society towards SCD, sufferers of the disease as well as their close relations.

Oludare & Ogili, 2013 cited Ignorance and Poverty as the common factors for the high prevalence of SCD in Africa. This work found out if the socioeconomic status of the participants was related to the proportion of children with the disease as opined by (Oludare & Ogili, 2013)

Alswaidi et al. 2012 reported social and cultural factors as main reasons for couples proceeding with marriage despite advice on their sickle cell status. A hundred percent positive view on SCD from the religious angle was gotten from respondents in a study carried out in Mali after health education intervention (Meilleur et al. 2011). There was a bias towards religion in this study because all the participants were Muslims who had a difficulty in separating the origin of SCD from God or fate, instead of the immediate biological cause. This study was a classic example of a study where selection bias based on religion was the hallmark, as Christians, Traditionalists or other religious groups did not participate. Possible Confounders like level of Education and Employment status were also not considered. This result is different from the study of Alao et al. 2009; Oludare & Ogili, 2013 were religion was reported not to have any significant influence on SCD knowledge. On the other hand, Omuemu

et al. 2013 reported that the proportion of Christian participants who had knowledge of haemoglobinopathies were 95.8%.

2.5 Blood Genotypes

In a retrospective study of at-risk marriages between sickle cell trait carrier intending couples and effect of genetic counselling between 2004 and 2009 in Saudi Arabia by (Memish & Saeedi, 2011), they reported that there was issuance of marriage certificates to some couples despite the unfavourable test results after knowing their sickle cell trait status. They stated that the prevalence of SCD was 45.1 per 1,000 with a p-value of 0.803 which was not statistically significant. They found out that marriage cancellation of at-risk couples between 2004 and 2009 decreased from 10.1 to 4.0 per 1,000 persons-at-risk with p-value less than 0.001 after test and genetic counselling, making the result to be statistically significant. It showed that post genetic counselling reduced the number of persons-at-risk of at-risk marriages.

The percentage of haemoglobin variants was reported by Abdulrahaman et al. 2013 among study participants. They found out that the prevalence of SCD based on age groups indicated that the prevalence of HbAA was highest in the 11-20 years age group, while HbAS, HbAC, HbSC and HbSS (35%, 8.5%, 2.25%, 0.75%, and 4.25%) prevalence was highest among subjects<10 (less than 10) years old. The percentage distributions of different forms of haemoglobin (Hb) among the subjects were: HbAA 280(70%); HbAS 93(23.25%); HbAC 5(1.25%); HbSC 3(0.75%) and HbSS 19(4.75%). Among the male subjects, 93(67.9%) were HbAA, 18(14.9%) were HbAS; 1(0.83%) were HbAC; 1(0.83%) were HbSC and 8(6.61%) were HbSS. Among the 279 female subjects, 187(67.02%) were HbAA, 75(28.88%) were HbAS, 4(1.43%) were HbAC; 2(0.72%) were HbSC and 11(3.94%) were HbSS. They further

observed that all subjects with haemoglobin SS and SC were less than 20 years of age. Owolabi et al. 2011 also found out that HbAA had the highest prevalence among study participants. The percentage haemoglobin distributions reported were HbAA,70.5%; HbAS,17.1%; HbSS,4.8%; HbAC,3.8%; HbSC,1.7%; HbCC,1.4% and 0.7% for other genotypes. They also reported that 38% of respondents knew the cause of SCD; 48.7% knew their genotype and generally that the respondents' knowledge of premarital genetic counselling was poor. Only Lattimer, 2010 reported the highest percentage of HbSS of 68% among subjects. The haemoglobin types used in this study were HbAA, HbAS, HbAC, HbSS and other genotypes. This work also reported the percentage distribution of haemoglobin types among the study participants

2.6 Other reported distributions

2.6.1 Hospital distributions

According to Adeyemo et al. 2007, the reported cases of sickle cell disease between 1995 and 2000 for Private, Public and Teaching Hospitals were 14, 143 and 272 respectively. While between 2001 and 2005 for the three different hospitals in the same order, documented cases of SCD became 57, 89 and 57 respectively. For Public and Teaching Hospitals in the two different time periods, the count of reported cases dropped significantly from 143 to 89 for Public Hospitals and from 272 to 57 for Teaching Hospitals respectively. The reasons for the decrease of SCD cases in these two health institutions could possibly be due to cancellation of marriages of at-risk-persons after premarital screening and premarital genetic counselling as well as increase awareness in the knowledge of SCD among patients who come to these

hospitals. However for Private Hospitals, there was an increase of reported cases of SCD from 14 to 57 between the two time periods. This could probably be due to less emphasis on practice of premarital genetic, not encouraging the termination of at-risk-marriages and the possible claim of provision of treatments for children with SCD by the private health institutions.

Furthermore, Adeyemo et al. 2007 found out that out of the 30 study centres used; only five had genetic units and that 30.3% of the 150 respondents had been exposed to genetic counselling.

2.6.2 Age reported distribution

The age range of study participants had been studied. Anie et al. 2010 had 38% of participants in the age range of 14 and 18 years, while 62% were between 19 and 56 years. Alao et al. 2009 reported that 47% of study participants knew their haemoglobin phenotypes, while the study was not statistically significant with $p > 0.05$ (p greater than 0.05). However, they reported an overall mean score of knowledge (MSK) and standard deviation of 4.65 ± 1.66. The MSK reported for males was 4.58 ± 1.66 and 4.74 ± 1.64 for females. This study went a step further ahead of previous reviewed works by reporting both the age range of the study participants and their children who suffer from sickle cell disease.

About nine percent of respondents in one of the two groups (that is intervention) studied by Olatona et al. 2012 agreed to have children with SCD despite knowing that they have sickle cell trait. The mean age and standard deviation of the intervention group were: 25.0 ± 2.2. The second group, that is, the control group had 25.1 ± 2.4 for both mean age and standard deviation.

In a study by Abdulrahaman et al. 2013, all subjects less than 20 years were found to have HbSS and HbSC. The reported mean ages and standard deviations were 38.4 ± 12.8 for males and 38.4 ± 12.8 for female participants respectively. Dampier et al. 2010 reported a mean age and standard deviation of 9.6 ± 4.7.

Accordingly, Amr et al. 2011 matched 180 adolescent with SCD (cases) with 202 healthy adolescent (controls) with both cases and controls having an age range of 14 to 18 years. Personal interviews and a review of the medical records on health related quality of life of both groups were carried out. It was found out that there was significant health deterioration in adolescent with SCD. Also, health related quality of life scores were negatively associated with increasing age, female gender, rural residence, low family income, presence of disease-related complications and frequent hospital admissions. In addition to all these, sociodemographic correlates were significant determinants to worsen health related quality of life.

Amr et al. 2011 further stated that the health related quality of life of Saudi Arabian adolescent deteriorates upwards. That is, as they grow older. For the cases and controls, 16.8 ± 3.6 and 16.9 ± 1.7 were reported as their mean ages and standard deviations respectively. However, male subjects with SCD had higher mean score above 66th percentiles than females with SCD. More so, cases had lower sociodemographic profiles than controls, but this was found not to have statistical difference. Lattimer, 2010 in addition only reported a mean age of 31.2 and age range of 20 to 59 years for study participants. 65.7% of respondents interviewed by Abioye-Kuteyi et al. 2009 were between 21 and 30 years. Adeyemo et al. 2007 reported an age range of 16 to 45 years for study participants. Respondents interviewed by Afolayan & Jolayemi, 2011 were all married and were between 20 and 60 years.

Oludare & Ogili, 2013 reported 23.35 ± 0.25 for mean age and standard error of mean for all respondents. They both stated that there was a significant association between respondents' education and knowledge; attitude and practices to SCD and SCD premarital counselling. 61% of respondents here had secondary school education as their highest level of education. Age and education were found to be statistically significant with premarital genetic counselling and sickle cell disease. The age range of study participants and their children was also reported.

Palermo et al. 2008 identified neighbourhood socioeconomic distress using both publicly available census tract and African American respondents with a mean age and standard deviation of 12.14 ± 2.5. They reported that there were lesser studies on individual and family influences on children with SCD; that higher income was related to less child-reported disability from SCD and greater physical health related quality of life and that 25% of participants live in distressed neighbourhood. Memish & Saeedi, 2011 reported that the incidence of SCD was constant in a retrospective study carried out in Saudi Arabia between 2004 and 2009 despite reduction of at-risk marriages. They both stated that this could be as a result of the limitations and bias in aggregate data.

2.6.3 Religion reported distributions

While 95.3% of respondents interviewed by Omuemu et al. 2013 were Christians, 63.3% of them were between 20-24 years. Owolabi et al. 2011 interviewed respondents between the ages of 9-26 years, with mean age and standard deviation of 15.16 ± 2.13 and a modal age of 10-19 years in 97.8% of the overall participants. The percentage frequency of the different religions of respondents in this work was also reported.

In a study of 38 people in Mali by Meilleur et al. 2011, for a before and after genetic testing and counselling research, all the subjects were Muslims. This was so because majority of the people in Mali are Muslims. However in the work of Oludare & Ogili (2013), 70% and 30% of participants interviewed were Christians and Muslims respectively. There was no participant from either Traditional or any other religion. They further reported that out of these study participants, the percentage of those knowledgeable about premarital genetic counselling was 57% for Christians and 23% for Muslims. This work found out if there were participants from other religions apart from Islam and Christianity after collection of data and analysis.

2.6.4 HbSS Distributions

The most intensive prospective study on SCD based on a very large sample population was carried out by Memish et al. 2011. They used a total 1,572,140 men and women who were examined over a 6 year period. This figure represented 6% of the entire population of Saudi Arabia. They found out that the prevalence of couples who tested positive for sickle cell disease was 45.1 (42.4 for carriers and 2.7 for cases) per 1000 persons examined. The prevalence was highest in the Eastern region (134.1 per 1000), followed by Southern and Western regions (55.6 and 28.5 per 1000, respectively) and lowest in Central and Northern regions (13.7 and 13.5 per 1000, respectively). The prevalence of couples who tested positive for β-thalassemia was 18.5 (18.0 for carriers and 0.5 for cases) per 1000 persons examined. The prevalence was highest in the Eastern region (59.0), moderate in the Southern, and Western and Central regions (14.2, 10.2, and 10.1 per 1000, respectively) and lowest in the Northern region (3.9).

While Abdulrahaman et al. 2013 reported an observed prevalence of 4.75% for HbSS among 400 subjects of African resident in Sokoto; Owolabi et al. 2011 reported 3.8% for HbSS which was almost a similar prevalence in a study of 600 secondary school students in Abuja. However, Jeremiah, 2006 reported a zero percent prevalence of HbSS among 620 University students in a study in Port Harcourt Nigeria. The zero frequency observed in this Port Harcourt study, possibly implies that the sickling gene pool is gradually reducing in some African populations due to increased awareness and premarital counselling. The low prevalence of HbSS observed in this study could also be attributed to increased awareness of the disease, improved socio-economic conditions, improved premarital counselling, education, environmental and genetic factors which have an overall effect on the sickling gene pool. According to Abdulrahaman et al. 2013, the zero prevalence may also be attributed to an active programme of prenatal diagnosis among pregnant women in the southern part of Nigeria.

By comparison, the prevalence of HbSS among the black population in the United States is reported to be around 9% and 30%-40% generally for Africans (Gulbis et al. 2009). On the other hand, the number of people with homozygous SS in Sokoto, Nigeria is high. The reason for this high prevalence may be due to the absence of carrier testing programs and premarital counselling/testing for prospective couples prior to marriage in a bid to reduce the prevalence of haemoglobinopathies in the area (Abdulrahaman et al. 2013). Tshilolo et al. 2008 in a study reported that evidenced-based data from Belgium, a country with universal neonatal screening programme shows that neonatal screening is an excellent health education tool. Premarital genetic counselling likewise in Belgium is a strong preventive measure against SCD.

2.7 Study Methods

Anie et al, 2010; Abioye-Kuteyi, 2009; Animasahun et al. 2009; Omuemu et al. 2013; Meilleur et al. 2011 used minimum sample sizes of at least above 300 which is less than the 407 female participants used for this work. This number gave a good representation of the study population. However, the limitation in all these studies was that none of the participants was directly connected to the people with SCD, or asked questions about siblings with the disease. These limitations were addressed in this study. This is because almost all of the respondents in the reviewed works were healthy carriers or non-carriers.

Adeyemo et al. 2007 used randomisation to select subjects for their study. Multistage sampling technique was employed by Olatona et al. 2012; Alao et al. 2009. Likewise, Odelola et al. 2013 used multistage sampling technique in three stages namely: Purposive, Stratified and Proportionate. This study also used of multistage sampling technique which was broken into four stages of random sampling, systematic sampling, balloting to select study centres and finally random selection of study participants from the study centres.

While most studies used primary data, only Adeyemo et al. 2007; Dampier et al. 2010 used a combination of primary and secondary data for analysis. Primary data was used for this work.

A lot of the reviewed works used probability sampling procedure; only Palermo et al. 2008 used a non-probability sampling method with two groups; one with cases of SCD and the other, healthy individual control group.

A quasi experimental technique was used by Olatona et al. 2012 with two sets of respondents in intervention and control groups. Cross sectional studies were done in more than ninety percent of the studies. This study design reduces the problem of recall bias that often occurs in retrospective study and lost to follow up in prospective study which was done by Abdulrahaman et al. 2013. Alswaidi et al. 2012 used a case-control study design to determine the marriage status of 934 at-risk couples. Of the 934 couples, 824 were married (88.2%) and 110 (11.8%) were not. A case–controlled study was conducted on 104 couples who did not marry (cases) and 478 couples who did marry (controls) in order to assess relationships between various cultural and social factors and marriage decisions.

2.8 Care, Treatments and Management of SCD

2.8.1 Cultural and Religious beliefs

Studies showed that religious beliefs including prayer, faith in God and doctors, and a hopeful approach to health difficulties in Nigeria play a positive part in coping with SCD. Previous research by Anie et al. 2007 also revealed that compared with people with SCD living in the United Kingdom (UK), those in Nigeria commonly used praying and hoping as an affective coping strategy, which seems to be influenced by external factors such as religion, faith in God, culture superstitions, and stigma (Nzewi, 2001). This work likewise assessed the frequency of respondents on a Likert scale on their religious and cultural beliefs based on destiny/fate, prayers/belief in God, reincarnation and taboo/superstition respectively.

2.8.2 Treatments of SCD

Many of the previous work focused mainly on awareness on SCD and genetic screening. The earlier works advocated genetic counselling, health education and public awareness as preventive measures against SCD (Adeyemo et al. 2007). On the other hand, alternative therapy using phytomedicines has proven to not only reduce pain crisis, but also reverse sickling (*in vitro*) (Imaga, 2013). However in a study carried out in Saudi Arabia by Alswaidi et al. (2012), it was reported that marriage decisions for participants who did not receive genetic counselling was not statistically different from those that received genetic counselling.

Adeyemo et al. (2007) at the time of their study further argued that there was nonexistence of standard SCD clinics and agreed that genetic counselling or better still premarital genetic counselling would help prevent SCD. However, there was selection bias in the findings of Adeyemo et al. (2007). This was due to the use of study population with the highest level of education, which is the University, thus making respondents score high on knowledge assessment.

Adesina (2005) found out that extracts from Orin Ata are used by Traditional Healers in Nigeria and could compensate some inborn defects of SCA. He further suggested that this means healthier red blood cells, less anaemia/pain for individuals with SCD.

The recommendations and solutions to reducing and preventing sickle cell disease and various forms of haemoglobinopathies have been reported. Abioye-Kuteyi et al. (2009) recommended that carrier identification before marriage and genetic counselling were the only realistic reduction approach to SCD in low-income countries like Nigeria. They also emphasised the importance of increased local

government marriage registries, genetic counselling, health information awareness and health education. Adeyemo et al. 2007; Odelola et al. 2013 believe that genetic counselling will help establish a diagnosis of hereditary diseases, take measures to alleviate clinical manifestations of disease, predict probability of disease development in families/individuals and possibly prevent it, thereby providing options to take informed decisions. They further advocated the vigorous participation of Government at all tiers, hospitals and religious institutions, with their study reporting that 58% and 64% of participants agreed that genetic counselling would help prevent SCD and other genetic diseases respectively. Similarly, Memish & Saeedi, 2011 stated that counselling should be encouraged by engaging religious figures in the society, using programme information in high schools and encouraging singles to seek voluntary genetic testing.

According to Afolayan & Jolayemi, 2011, adequate mobilisation and educative programmes on genetic counselling are veritable tools to preventing SCD. They both strongly advocated for prohibition of marriages between non-compatible individuals. Education and information on SCD, according to Olatona et al. 2012 would help create autonomous individuals who could take informed decisions regarding marriage with other carrier individuals because they reported that educational intervention significantly increases the knowledge of SCD with p-value less than 0.001. Abdulrahaman et al. 2013 suggested that there should be formulation of genetic counselling policies to reduce the incidence of haemoglobinopathies in Nigeria. In addition, they said that there should be carrier screening and mutation identification as preventive measures. Jastaniah (2011) advocated the need for collaborations of Ministries of Health, Medical Institutions, Research Organisations and International

Donor Agencies. Screening of carrier couples and genetic counselling as an intrinsic part of healthcare in most countries were advocated by Modell & Darlinson (2008). Al Gazali (2006); Al Arrayed & Al Hajeri (2010) opined that genetic education through high school curriculums, genetic information and awareness via mass media educational campaigns are pivotal to preventing SCD. The various recommendations and limitations in this work were also reported.

2.9 Study Pre-test

Reliability test for questionnaires used in some of the previous works was reported. Odelola et al. (2013) reported a reliability of 0.76. Dampier et al. (2010) used Cronbach's coefficient alpha to assess internal reliability. They reported that Cronbach's alpha scores greater than 0.7 for all parent and child report scales except school functioning (child report) which was 0.69. The parent Cronbach's alpha scores were between 0.79 and 0.95, while child's report was between 0.69 and 0.88. Reported Pearson correlations for relationship between parent and child's report showed moderate range from 0.38 to 0.47. The test-retest coefficient reported by Afolayan & Jolayemi (2011) was 0.78. A pre-study focus group involving 6-8 participants with no validation of the questionnaire was carried out in Lagos, Nigeria (Anie et al. 2010). Omuemu et al. (2013) did a pre-test using self-administered questionnaire without reporting the number of study participants used in Edo, Nigeria. Only two participants were involved in the pilot study of Afolayan & Jolayemi (2011) in Kwara, Nigeria. Likewise, Meilleur et al. (2011) carried out a pilot study in Mali but the number of respondents involved was not reported. A pre-test involving 16 study participants in a was also carried out apart from the one for main study on study

participants in this study with both self-administered and interviewed-based questionnaires used.

Ethical approval was obtained for this study from the Ministry of Health, Lagos state. Informed consent of study participants was also obtained. In addition, permission to carry out this study was gotten from the various study centres. More so, ethical consideration was looked into. Palermo et al. 2008) obtained ethical approval and informed consent from study participants. In the study of Lattimer (2010) ethical approval was obtained from John Hopkins Medical Institution Review Board. Amr et al. 2011; Meilleur et al. 2011; Olatona et al. 2012 also obtained ethical approval for their study. However the Institutions the three works obtained it from were not reported.

2.10 Information sources

The different ways of getting information on PGC and SCD were studied. (Omuemu et al. 2013) reported that 79.5% of participants got information about SCD from electronic media, 63.3% from print media, 20.3% from lectures and 16.7% from health facilities. On the other hand, (Adeyemo et al. 2007) reported that 22% of participants got information about SCD from hospitals and just 8% from media and churches.

2.11 Statistical Techniques Application

Parametric and Non parametric statistical data analysis techniques like Chi square test, One way analysis of variance test (ANOVA), Kruskal Wallis test, Regression and Correlation analysis were used by Mcinish & Saeedi, 2011; Palermo et al. 2008; Lattimer, 2010. Al Arrayed & Al Hajeri, 2010 used Mann-Whitney U;

Kruskal-Wallis and ANOVA tests for statistical analysis. Jeremiah (2006) analysed data using T-test and ANOVA with level of significance set at 0.05% to test the formulated hypotheses. Descriptive and Inferential statistics including counts, frequencies, means and chi square tests were used in this study.

Descriptive statistics of frequencies, counts, percentages and means were used by Odelola et al. 2013; Palermo et al. 2008 to analyse demographic data. Palermo et al. 2008 further used multivariate regression analyses. The analyses revealed that individual/family socioeconomic distress was a significant predictor of children's functional disability and physical as well as psychosocial health related quality of life. The analyses also showed that distressed neighbourhood in addition to individual socioeconomic status is related to diminish physical health related quality of life in children. Memish & Saeedi (2011) tested for significance increasing/decreasing trends in pattern of their results by analysing with Extended Mantel Haenszel and Chi-square test. Dampier et al. 2010 used multiple regression models to control for haemoglobinopathies, gender and age, suggesting that reports of physical functioning, sleep/rest and fatigue declined in response to pain or avascular necrosis, while school functioning scales decline in response to pain or asthma.

While Dampier et al. 2010 used SAS/STAT statistical package for data analysis, Odelola et al. 2013 used STATA version 8.0. (Olatona et al. 2012) used Epi Info version 6.04. Memish & Saeedi (2011) used Open Epi version 2.2 and Microsoft Office Excel 2003 for all statistical analyses. SPSS version 15.0 was used by Al Arrayed & Al Hajeri, 2012; Lattimer, 2010. Palermo et al. 2008: Nnaji et al. 2013 used SPSS version 16.0. However, SPSS version 20.0 was used for data analysis in

this study. This version was designed to provide an easier, a better and a more precise analysis than the previous versions.

2.12 Male to Female Proportions

The proportion of male to female participants as well as the number of study participants has been reported. Al Arrayed & Al Hajeri (2010) studied 1106 females out of study participants of 2,000 which had 45% male and 55% female respondents. A similar proportion of 44% males and females 56% were reported by Lattimer (2010) but with a far different sample size of only 45 study participants. Palermo et al. 2008 studied 56 participants with a male to female proportion of 57% to 43%. A proportion of 53% males and 47% females made up the 1722 participants. Abdulrahaman et al. 2013 studied 400 participants having 30.25% male and 69.75% female proportion. A proportion of 50.2% and 49.8% males and females respectively were involved in a study of 600 secondary school students in Abuja, Nigeria (Owolabi et al. 2011). Omuemu et al. 2013 reported 52% males and 48% females in a study of 400 undergraduates in University of Benin, Nigeria. Four hundred and eight participants of a male to female proportion of 47.5% to 52.5% were interviewed in a study of psychosocial aspects of sickle cell disease in the UK and Nigeria in 2007 (Anie et al. 2010). Eighty participants were involved in the study of Afolayan & Jolayemi (2011) with proportion of male to female as 18.75% to 81.25%. Abioye-Kuteyi et al. 2009 reported a study participant of 300 individuals with male to female proportion of 49% to 51%. Schwartz et al. 2009; Al Arrayed & Al Hajeri, 2012; Odelola et al. 2013; Amr et al. 2011; Meilleur et al. 2011; Olatona et al. 2012; Oludare & Ogili, 2013; Animasahun et al. 2009; Adeyemo et al.2007 did not report the proportion of male to female in their study.

The lowest female proportion was reported by Palermo et al. 2008. This was 43%. Afolayan & Jolayemi (2011) reported the highest female proportion of 81.25%. This work did not have a report on male to female proportion because study participants were all females except for the proportion of male to female children with SCD as gotten from the collected data.

CHAPTER THREE

3.0 METHODOLOGY

3.1 Study Area

This study was carried out in Lagos, South West Nigeria. According to the 2006 census result, Lagos is the second most populous state in Nigeria after Kano state with over nine million inhabitants. However there is a controversy on the authenticity of this figure. The Lagos state government disproved this figure, stating that the state is the most populous in Nigeria with at least 18 million people as of 2013 (www.lagosstate.gov.ng). Lagos state is referred to as the economic nerve centre of Nigeria. This is because most of the indigenous and foreign financial establishments in Nigeria are located there.

The state is made of 20 Local Government Areas (LGAs) with additional 37 Local Council Development Areas (LCDAs). Ikeja is the capital of the state. The state is made up of people from almost all the different ethnic and religious groups in Nigeria. It shares boundary with Ogun state in the North and Atlantic Ocean in the South (www.nigeria.gov.ng/...states/lagos.state).

The highest numbers of sickle cell centres/clubs in Nigeria are in Lagos state. The study centres are the health facilities the study was carried out. Lagos alongside Edo is the only state with a standard sickle cell centre in Nigeria (Lagos health centre fights sickle cell disorder. *DAILY SUN*. Thursday, July 24, 2014: Page 40). There are also other non-governmental organisations (NGOs), clinics and hospitals that are involved in sickle cell disease prevention, management and treatment in the state. Nine primary health facilities and sickle cell clubs/centres were used.Eight of the

centres were used for the main study while the remaining one centre was initially used for the Pre-test.

The pre-test was done in Med-In Specialist Hospital, Ogudu. The eight Study centres used were from the following six local governments: Ikeja, Ikorodu, Kosofe, Lagos-Mainland, Oshodi-Isolo and Surulere. A minimum of one primary health care (PHC) facility or study centre was randomly selected form each of the six local governments for the study. The PHC facilities included: Genotype Foundation (GF) and Temitayo Awosika Help Foundation (TAHF) Sickle Cell Club Lagos State University Teaching Hospital. (LASUTH), both in Ikeja; Sickle Cell Club General Hospital Ikorodu; Dabma Sickle Cell Foundation and Sickle Cell Club General Hospital Gbagada, both in Kosofe; Medical Centre University of Lagos (UNILAG), Lagos Mainland; Sickle Cell Club Isolo General Hospital, Oshodi-Isolo and Sickle Cell Advocacy and Management Initiative (SAMI), Surulere.

3.2 Study Design

A cross sectional study was carried out.

3.3 Study Population

The study population was gotten by sampling from a population of women of reproductive age and caregivers who attend the eight PHC facilities with their children who have SCD.

3.3.1 Inclusion criteria

1. Women who come to the health facilities with children who have SCD.

2. Caregivers of the children with SCD. (They could be stepmothers, foster parents, grandmothers or nannies).
3. Mothers and caregivers of children with SCD attending the centres.
4. Mothers and caregivers who have a child or children who did not survive SCD.

3.3.1 Exclusion criteria

1. Women with children who have SCD and whose children are adults (that is above secondary school education).
2. Women who come to the centres with their fiancé or husbands to be for counselling.
3. Mothers and caregivers with more than one or more children living with SCD.

3.4 Sample Size Determination

The standard sample size formula for calculating single proportion was used.

This is given as:

$$N = \frac{Z\alpha^2 p(1-p)}{d^2}$$

A confidence interval of 95% at 5% level of significance gave Zα to be 1.96. The prevalence used was p equals 63.6% (WHO, 2008). This prevalence was used in a study on the management of haemoglobin disorder in Cyprus. p is the proportion of women (that is the respondents) who have children with sickle cell disease. p is also known as the estimated proportion of an attribute that is present in the population

(Nnaji et al. 2013). The sample size is represented by N. Thus; p=0.636, 1-p=0.364, d=0.05 and N=356. The calculation for N equals 356 is shown below:

$$N = \frac{[1.96^2 \times 0.636 \times (1 - 0.636)]}{0.05^2} = 356$$

A 10% additional attrition rate was considered. It also equals 0.1. This further brought the calculated sample size (n) to become 392, which is the minimum sample size. This was by adding 10% of 356 to 356 to give 392. The calculation for n is given below:

$$n = (0.1 \times 356) + 356 = 392$$

However, a total of 407 study participants were interviewed.

3.5 Sampling Technique

Respondents were selected based on the multistage sampling technique. First, a random sampling procedure was used to select 12 out of the 20 local government areas in Lagos state. Furthermore, systematic sampling method was used to select six LGAs out of the 12 LGAs. This was done by picking one LGA, skipping one LGA, then another one, skipping one, and continuously like that until all the six LGAs were selected. Balloting was used to select a minimum of one study centre from each of the six LGAs. Finally, women with children with SCD were selected and interviewed from each of the selected study centre.

3.6 Data Collection Methods and the Instrument

A pre- tested semi- structured questionnaire was used to collect data. It contained both open- ended and close- ended questions. Data and information collected focused on the objective of this study. These data included: sociodemographic characteristics, proportion of women with knowledge of premarital genetic counselling, knowledge and practice of premarital genetic counselling, as well as knowledge of sickle cell disease, religious and cultural beliefs regarding sickle cell disease.

Structured closed questions offering a dichotomous choice of 'yes' or 'no' as well as a Likert rating scale ranging from strongly disagree to strongly agree was used (Kaye et al. 2006). The Likert scale was later reduced to the nominal level by combining all agree and the disagree responses into accept and reject.

3.7 Study Variables (Exposure and Outcome)

These were the exposure and the outcome variables. The main exposure (independent variable) was knowledge and practice of premarital genetic counselling. The outcome (dependent variable) was children with sickle cell disease. Other independent variables include: Education, Cultural beliefs (Reincarnation & Taboos) and Religious beliefs (Destiny and Prayers). Other outcome variables were knowledge of sickle cell disease and knowledge of premarital genetic counselling.

3.8 Data Management and Analysis

Descriptive and inferential statistical techniques using SPSS Version 20 were used for data analysis. The level of significance was set at 0.05 (5%).

Open ended questions were collapsed to specific nominal scales by data coding before analysis.

Bar graphs were used to depict the percentage frequency of study participants on questions based on knowledge of SCD. Tables were drawn to show the relationship of sociodemographic characteristics, knowledge of premarital genetic counselling, sickle cell disease as well as frequency on the information about the children with sickle cell disease.

All Likert scale questions of strongly disagree, disagree and indifferent were collapsed to reject and represented with a code '0', while agree and strongly agree questions were collapsed to accept and represented with a code '1'. This was done to know the overall frequencies of study participants' response to questions asked on disagree and agree scales.

Chi square test and bivariate analysis were both also used for statistical analysis.

Chi square test was used to analyse categorical variables like marital statuses, monthly income, ethnic groups, education levels and employment statuses. This was done to test independence and association in any form of cross classification between the proportions of women who practice and do not practice PGC; those who have

knowledge and do not have knowledge of PGC and SCD across the categorical variables.

In this study correct answers were coded as 1, while indifferent and incorrect answers were coded as 0. The respondents' knowledge of sickle disease was assessed on a 10 point scale. A total point score of five points or less (≤ 5) on the 10 point scale represented poor knowledge of SCD, while a total point score of between four and ten (6-10) represented good knowledge on the 10 point scale.

The level of knowledge and practice of premarital genetic counselling was assessed on a seven point scale Study participants who scored 3 points or less (≤ 3) were considered to have poor knowledge and practice of premarital genetic counselling while those who scored between a total of four and seven points (4-7) of the 7 point scale were considered to have good knowledge and practice of PGC.

3.9 Ethical Consideration

1. Ethical approval was obtained from the Lagos state ministry of health ethics review committee.
2. Authorised permission was obtained from the selected study centres before commencement of study.
3. Study participants' written informed consent was obtained.
4. All collected data/information were treated with the highest degree of confidentiality.

3.10 Study Limitations

1. The difficulty of getting a large sample size of study participants in some of the study centres.

2. Some study participants not willing to neither sign study participant's informed consent after reading nor participate in the study.

3. The high cost of transportation in moving from one study centre to the other in the six local government areas.

3.11 Definition of Terms

1. **SCD**: It stands for sickle cell disease. It is also used as sickle cell disorder. It includes sickle cell anaemia (HbSS), mild sickle cell disease (HbSC) and thalassaemia.
2. **Haemoglobin**: The iron molecule in the red blood cell that delivers oxygen to the cells in the body after combining with oxygen. It is sometimes called 'the repanocytosis.'
3. **PGC**: It stands for Premarital Genetic counselling. It is a process of helping individuals or to be couples get information on the possibility of having children with inherited disease or not.
4. **Haemoglobinopathies**: They are inherited single gene diseases as a result of abnormal haemoglobins in the blood.
5. **Thalassaemia**: A name for a group of hereditary haemolytic diseases caused by faulty haemoglobin synthesis and it is widespread in the Mediterranean region, African and Asian countries.

CHAPTER FOUR

4.0 RESULTS AND ANALYSIS

The responses to proportion of women who had ever had knowledge of genetic counselling for SCD were transformed into dichotomous variables of 1 and 0. Code 1 was for correct answers, while code 0 was for incorrect answers. The codes 1 and 0 also stood for good and poor knowledge respectively. Mean was used to know the average age distribution. Frequency was used to determine the proportion of women with correct and incorrect knowledge. Chi-square test was used to analyse the significant association in any form of classification between the sociodemographic characteristics of study participants. Bivariate analysis was used to predict which factor (that sociodemographic characteristic) affected the outcome variable.

A total of 420 questionnaires were administered. Out of these, 407 were returned and collected giving a response rate of 96.9%.

The mean age of the women was 38.6±7.2. Most of the study participants were aged 35 to 44 years (56.0%), Christians (63.6%), Yorubas (75.4), married (81.6%), those with secondary education (50.9%), self-employed (66.1%) and those on a monthly income of #6,000- #20,000 (47.7%) (Table 1).

Table 1: Socio-demographic characteristics of respondents.

Characteristics	n=407 Frequency(Percent)
Age groups (years)	
< 25	6(1.5)
25-34	94(23.1)
35-44	228(56.0)
45-54	71(17.4)
≥ 55	8(2.0)
Mean Age: 38.6 ± 7.2	
Religion	
Islam	147(36.1)
Christianity	259(63.6
Traditional	1(0.3)
Marital status	
Cohabiting	4(0.99)
Single motherhood	32(7.99)
Separated/divorced	23(5.7)
Married	332(81.6)
Widowed	16(3.9)
Ethnic groups	
Yoruba	307(75.4)
Hausa/Fulani	8(2.0)
Igbo	65(16.0)
Others	27(6.6)
Educational level	
No formal education	8(2.0)
Primary	56(13.8)
Secondary	207(50.9)
Tertiary and above	136(33.4)
Employment status	
Employee	78(19.2)
Self-employed	269(66.1)
Unemployed	60(14.7)
Monthly income (naira)	
No income	45(11.1)
6,000-20,000	194(47.7)
21,000-35,000	108(26.5)
36,000-50,000	27(6.6)
51,000-75,000	19(4.7)
Above 75,000	14(3.4)

Footnote: Others were Other Ethnic Groups (Edo, Urhobos, Efik, Isoko, Tiv, Ebira, Ibibio, Ijaws, Birom and Kanuri).

There was no significant association between age group and knowledge of PGC (p=0.09) and for employment status (p=0.99). Ninety four percent of the study participants with tertiary and above level of education had good knowledge of PGC, 37.5% of those with no formal education had poor knowledge of PGC. Higher level of education was significantly associated with knowledge of PGC, (p=0.02) and also ethnic group (p=0.01) (Table 2).

Table 2: Association between Socio-demographic characteristics and Knowledge of Premarital Genetic Counselling.

Characteristics	Good n(%)	Poor n(%)	Total n	χ^2	[* means significance] p value (p<0.05)
Age groups(years)					
<25	13(100.0)	0(0.0)	6	8.02	0.09
25-34	76(87.4)	11(12.6)	94		
35-44	203(89.0)	25(11.0)	228		
45-54	65(91.5)	6(8.5)	71		
≥55	5(62.5)	3(37.5)	8		
			407		
Religion					
Islam	81(55.1)	66(44.9)	147	5.23	0.07
Christianity	115(44.4)	144(55.6)	259		
Traditional	0(0.0)	1(100.0)	1		
			407		
Marital status					
Cohabiting	4(100.0)	0(0.0)	4	4.13	0.39
Single motherhood	28(87.5)	4(12.5)	32		
Separated/divorced	23(100.0)	0(0.0)	23		
Married	292(88.0)	40(12.0)	332		
Widowed	15(93.8)	1(6.2)	16		
			407		
Ethnic groups					
Yoruba	276(89.9)	31(10.1)	307	27.40	0.01*
Hausa/Fulani	7(87.5)	1(12.5)	6		
Igbo	60(92.3)	5(7.7)	65		
Other Ethnic groups	19(70.3)	8(29.7)	27		
			407		
Educational level					
No formal education	5(62.5)	3(37.5)	8	10.34	0.01*
Primary	49(87.5)	7(12.5)	56		
Secondary	180(87.0)	27(13.0)	207		
Tertiary and above	128(94.1)	8(5.9)	136		
			407		
Employment status					
Employee	70(89.7)	8(10.3)	78	0.03	0.99
Self-employed	243(90.3)	26(9.7)	269		
Unemployed	54(90.0)	6(10.0)	60		
			407		

There was a significant individual – level predictor for knowledge of genetic counselling for SCD among the Igbos and other ethnic groups. Igbos and other ethnic groups were twice less likely to have knowledge of genetic counselling for SCD respectively and was found to be significant (OR=0.53; 95% CI= 0.42 – 0.92) and (OR=0.38; 95% CI= O.16 – 0.87) (Table 3).

Table 3: Bivariate analysis of knowledge of genetic counselling for sickle cell disease among Respondents' socio-demographic characteristics

variables	coefficients	Odds(OR) ratio	95% confidence interval (CI)	Significance (p value)
Educational level				
≤primary(reference)				0.00
Secondary	0.89	0.90	0.48 – 1.71	0.75
Tertiary and above	0.61	0.66	0.33 – 1.30	0.23
Ethnic groups				
Yoruba(reference)				0.00
Hausa/Fulani	0.35	1.42	0.77 – 2.61	0.26
Igbo*	0.86	0.53	0.42 – 0.92	0.01
Other ethnic groups*	0.98	0.38	0.16 – 0.87	0.02

Footnote: * indicates significance ($p < 0.05$)

The mean age of the children was 9.3±5.1. Children aged 5-9 years made up 31.5% of the total age groups; those currently at secondary level of education (37.6%), while females were 57.7% (Table 4).

Table 4: Socio-demographic characteristics of Respondents' Children with sickle cell disease.

Characteristics	n=407 Frequency(Percent)
Age groups (years)	
<5	88(21.6)
5-9	128(31.5)
10-14	119(29.2)
15-19	59(14.5)
≥20	13(3.2)
Mean Age: 9.3 ± 5.1	
Sex	
Male	172(42.3)
Female	235(57.7)
Current Education level	
None	12(2.9)
Kindergarten/playgroup	31(7.6)
Nursery	65(16.0)
Primary	146(35.9)
Secondary	153(37.6)

The proportions of the Study participants' responses in percent on knowledge and practice of genetic counselling related to SCD showed that ninety one percent of respondents answered that SCD is caused by inheriting genes from parents. About 84.0% knew the number of genes to be inherited to have SCD, some of the medical signs/complications of SCD (66.1%), appearance of Sickle Red Blood Cells (80.7%), possible cure for SCD (32.1%) and more than eighty percent knew how to check if someone carries gene for SCD (Figures 3-8).

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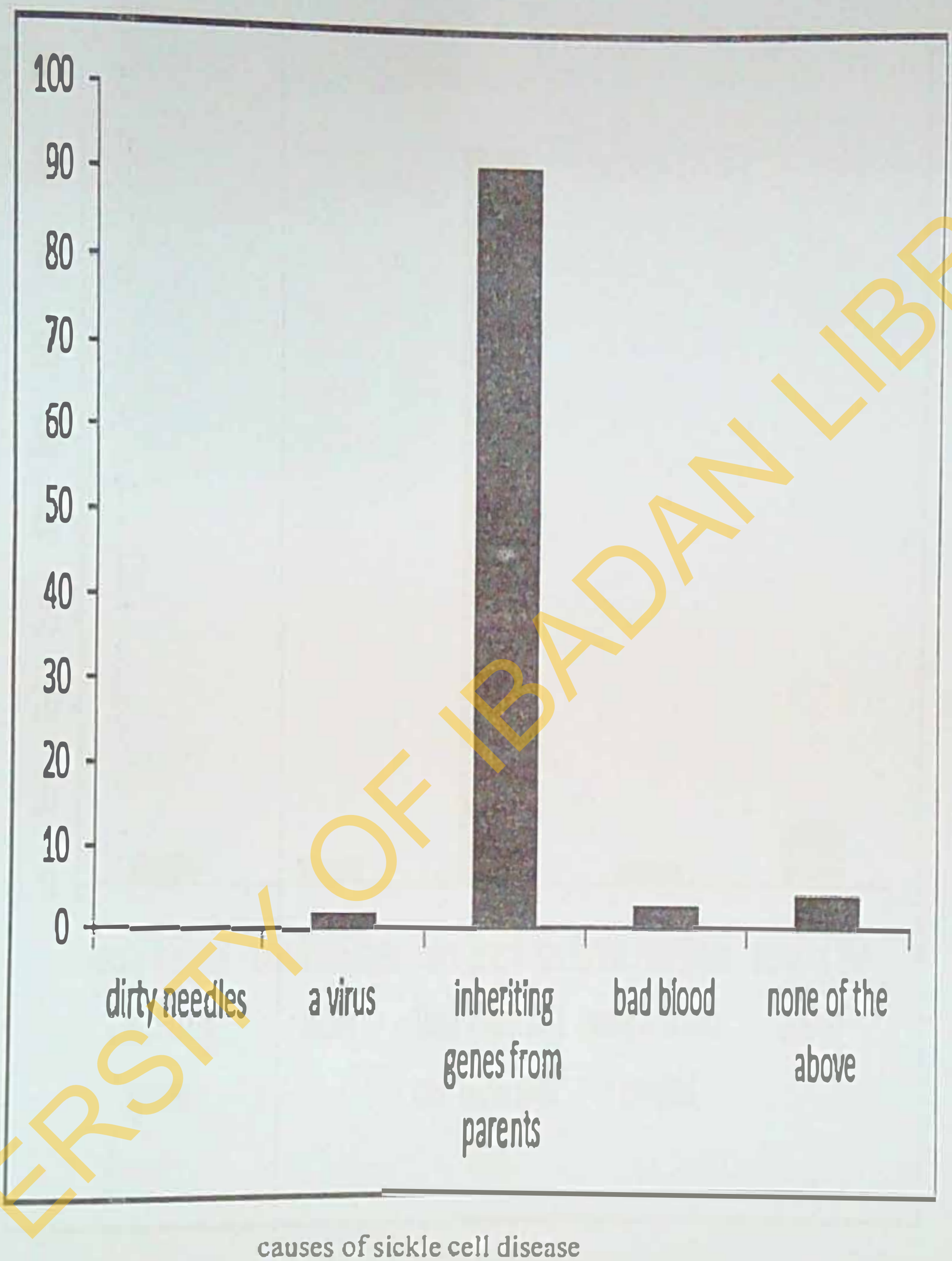

Figure 3: Study participants' responses in percent on causes of sickle cell disease

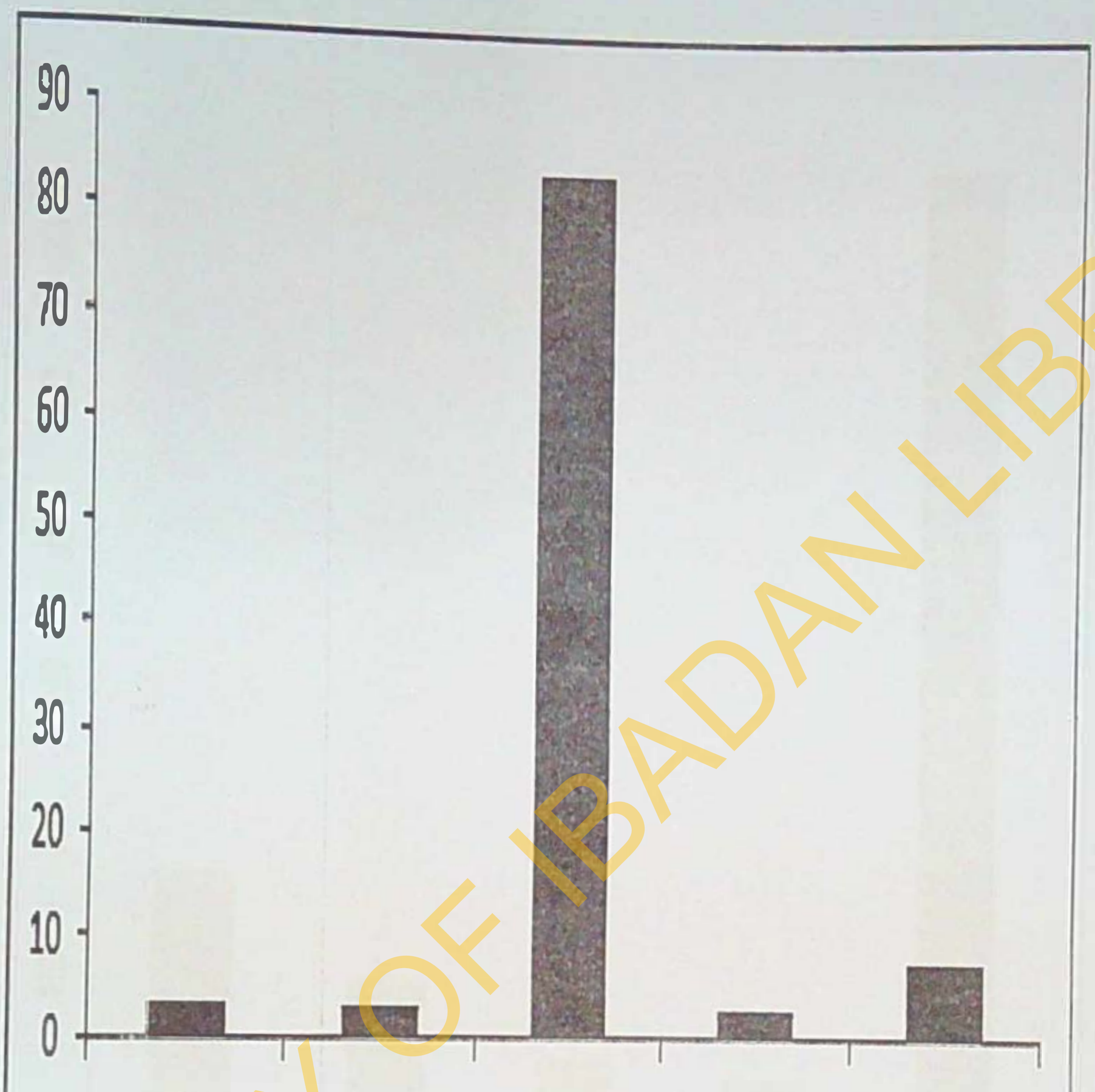

Figure 4: Study participants' responses in percent on number of genes to inherit to have sickle cell disease

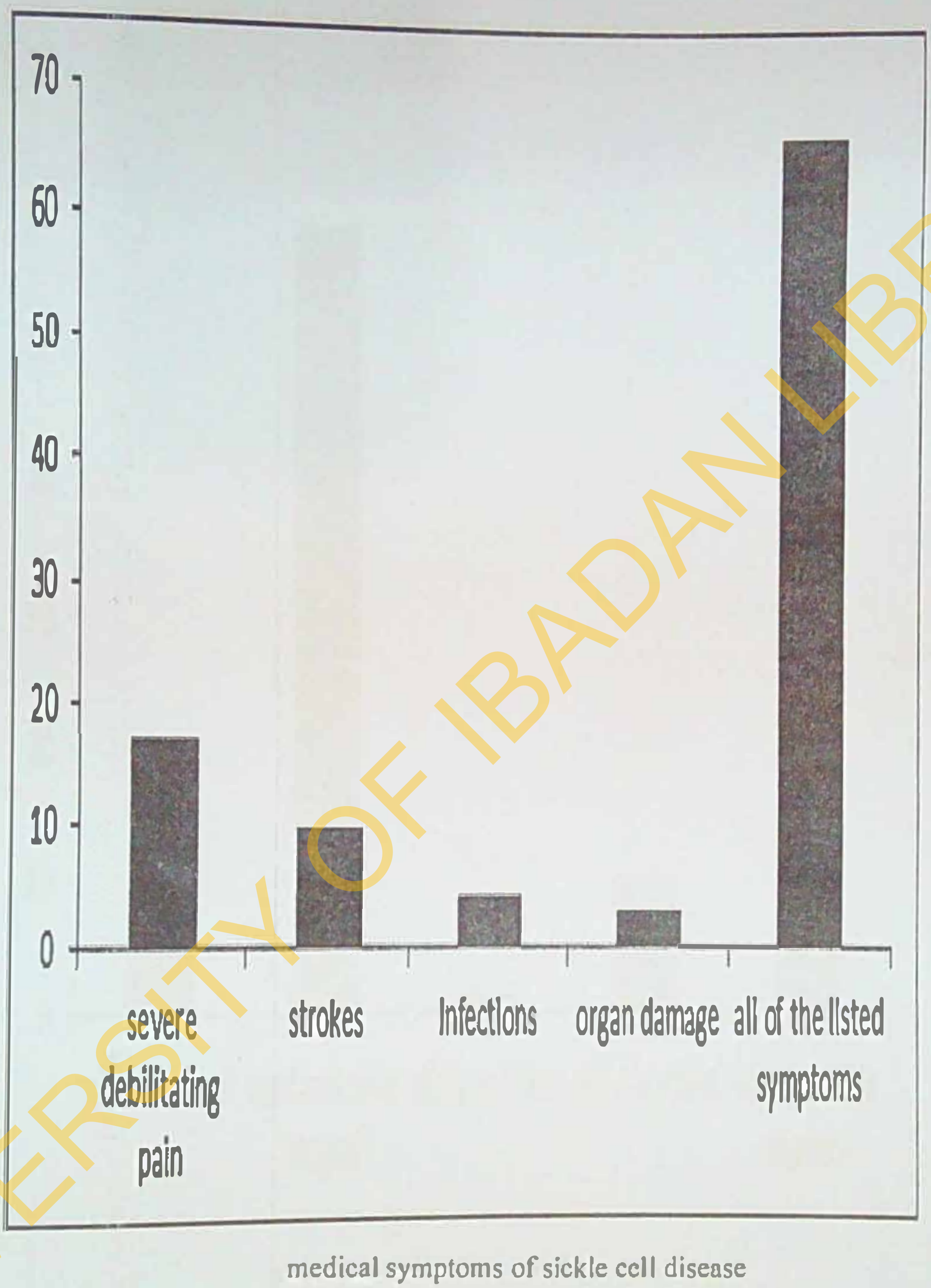

Figure 5: Study participants' responses in percent on signs/complications of sickle cell disease

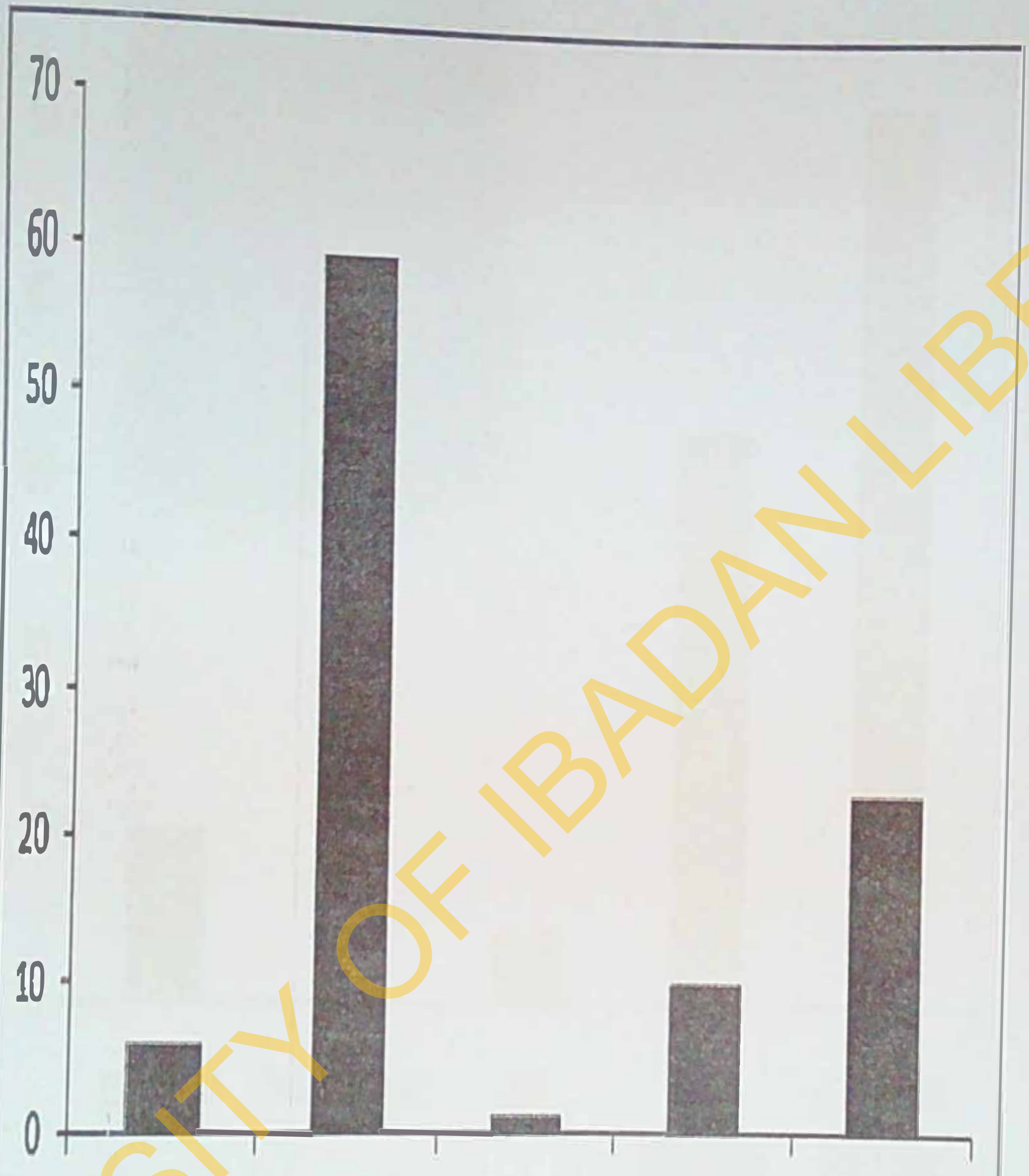

Figure 6: Study participants' responses in percent on appearance of sickle red blood cells

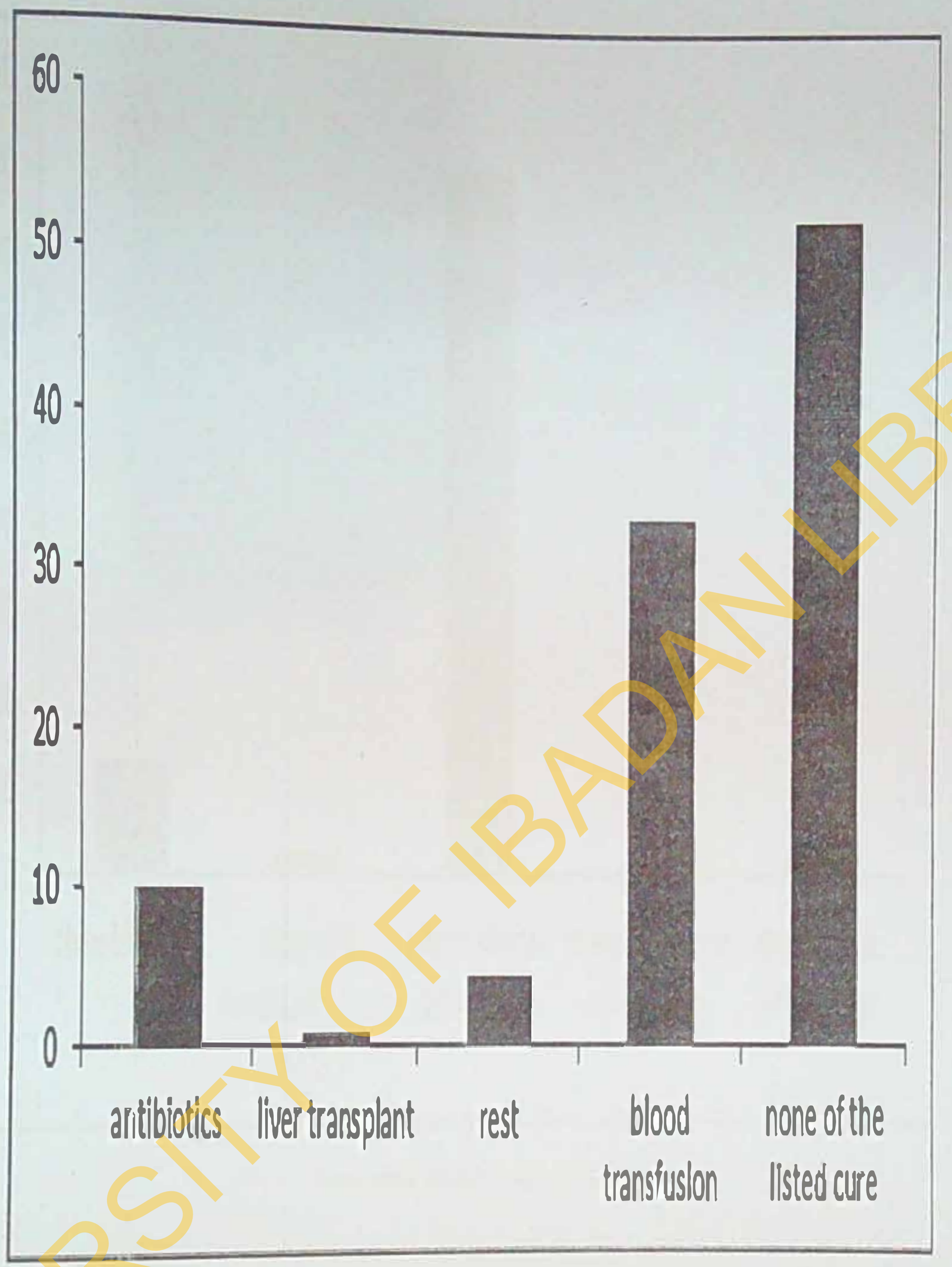

Figure 7: Study participants' responses in percent on cure for sickle cell disease

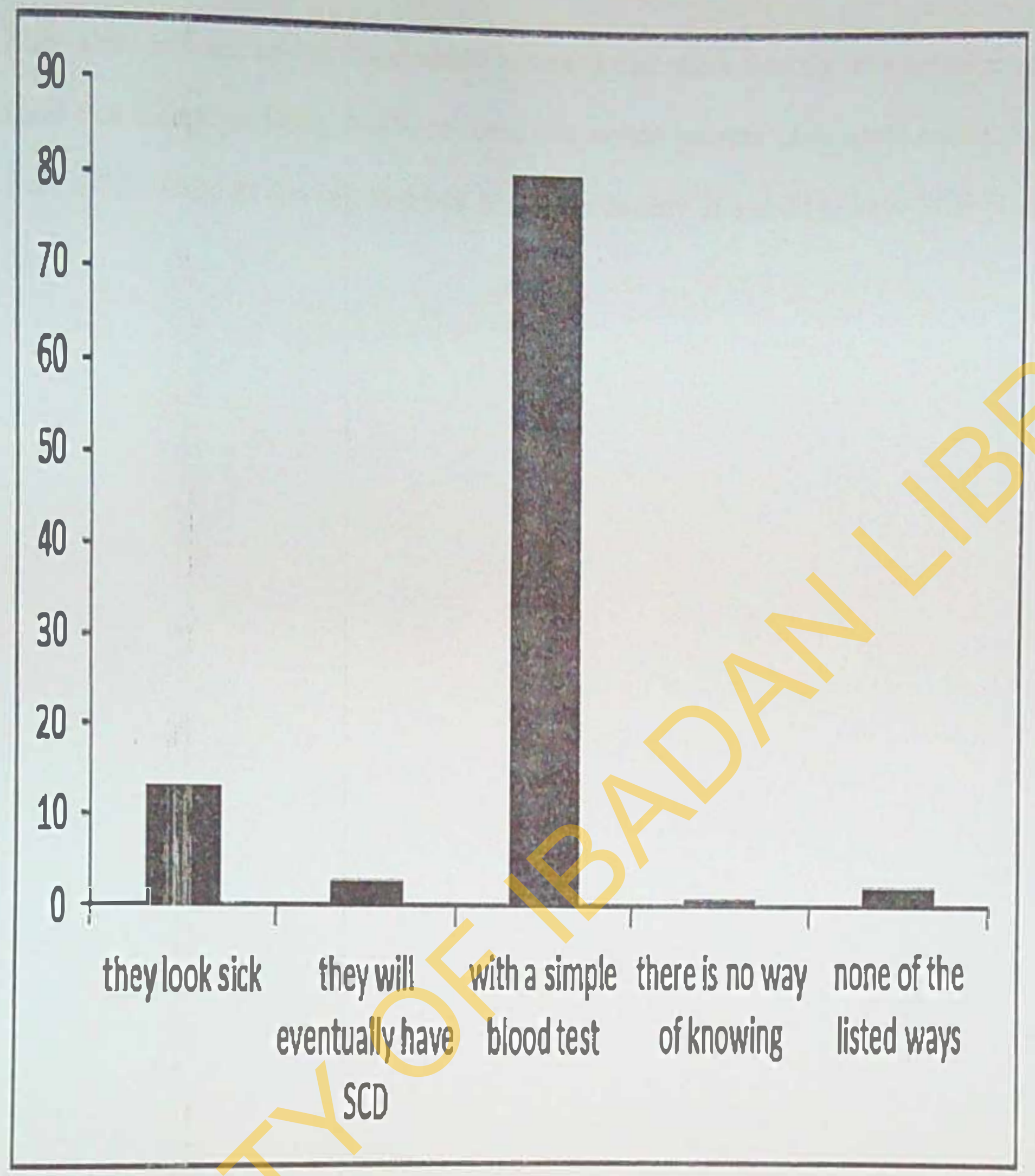

Figure 8: Study participants' responses in percent on how to detect someone carries genes for sickle cell disease

Fifty eight percent of the respondents accepted that there was no reincarnation of a child that dies from SCD, 39.6% rejected that saying prayers daily could cure a child from SCD; while 81.3% rejected that it was the destiny of a child to have SCD (Table 5).

Table 5: Respondents' religious/cultural beliefs for sickle cell disease.

Beliefs	n=407 Accept N (%)	Reject N (%)
Religious organisations play a role in preventing SCD	243(59.7)	164(40.3)
There is no reincarnation of a Child that dies from SCD	236(58.0)	171(42.0)
Saying Prayers daily could cure a child with SCD	243(60.4)	164(39.6)
It is the destiny of a child with SCD to have the disease	76(18.7)	331(81.3)
It is not a taboo to know if I have sickle cell trait	344(84.5)	63(15.5)
It is not a taboo to know if my partner has sickle cell trait	339(83.3)	68(16.7)
SCD could happen in any ethnic group	332(81.6)	75(18.4)

Chi square test analysis was used to examine the association between Socio-demographic characteristics and some outcome variables like: knowledge of PGC and SCD; practice of PGC and cultural/religious beliefs. A calculated chi-square test value that is more than a tabulated value shows significant relationship between the exposure and the outcome variables. Knowledge of PGC and SCD are both significantly associated with Educational level, Ethnic group and Employment status. Cultural/religious beliefs are significantly related to all listed Sociodemographic characteristics (Table 6).

Table 6: Association between Socio-demographic characteristics of Respondents and genetic counselling, sickle cell disease and religious/cultural beliefs

	Sociodemographic Characteristics	p-value * indicates significance (p<0.05)
proportion who had ever had knowledge of SCD	Age	0.37
	Religion	0.23
	Marital status	0.51
	Educational level	0.07
	Ethnic group	0.00*
	Employment status	0.39
knowledge of SCD	Age	0.00*
	Religion	0.56
	Marital status	0.15
	Educational level	0.03*
	Ethnic group	0.00*
	Employment status	0.01*
knowledge of PGC	Age	0.31
	Religion	0.16
	Marital status	0.00*
	Educational level	0.00*
	Ethnic group	0.05
	Employment status	0.00*
practice of PGC	Age	0.00*
	Religion	0.71
	Marital status	0.49
	Educational level	0.05
	Ethnic group	0.04*
	Employment status	0.08
religious/cultural beliefs	Age	0.01*
	Religion	0.00*
	Marital status	0.00*
	Educational level	0.02*
	Ethnic group	0.00*
	Employment status	0.00*

From Bivariate analysis done for practice of PGC, there was a significant individual – level predictor for practice of PGC among Igbos only. Igbos were three times less likely to practice PGC (OR=0.33; 95% CI= 0.16 – 0.58).

CHAPTER FIVE

5.0 DISCUSSION

The study was carried out to evaluate the knowledge, and practice of premarital genetic counselling among women in Lagos Nigeria related to knowledge of sickle cell disease. It has been discovered that the prevalence of SCD in this part of the world where this condition is common was largely due to ignorance of the people affected and poverty (Owolabi et al. 2011; Olatona et al. 2012). The study targeted population of women whose age range was 20-60 years with mean of 38.6±7.2, derived from the group data related to the age.

This study showed that 88% of the respondents that are married have good knowledge of premarital genetic counseling. This is quite expected as Lagos population is an urban area and not a rural area where the literacy rate is lower. This is in line with a study among students of the University of Lagos in 2007 which reported that 86% (Leadership Newspapers Nigeria. June 28, 2012. Four million Nigerians suffering from sickle cell disease Pg. 1. www.leadership.ng/nga/articles/28405/2012/06/28) of the respondents were knowledgeable of SCD and genetic counseling. Religious bodies, hospitals and non-governmental organizations in Nigeria such as Genotype Foundation (GF), Dabma Sickle cell foundation and Sickle Cell Advocacy and Management Initiative (SAMI) are some of the organisations helping to raise the level of awareness, cure and management of SCD in Nigeria. The religious bodies like Mosques and Churches through their counseling section seminars inform the public and most of them mandates genetic counseling before couples could be joined together.

The findings of this study for educational level was significant but not consistent with the work of Oludare & Ogili (2013) where 81% of the youths whose highest educational qualification was primary school had no knowledge of SCD and SCD premarital counselling. Only 12.5% of those with primary education in this study had little or no knowledge of PGC. This further buttresses the fact that education is vital in the campaign to reduce the prevalence of SCD and this implies that the government would need to develop innovative ways such as the teaching of SCD in the curriculum of secondary schools.

In addition in this study, knowledge and practice of genetic counselling towards SCD showed that age and educational qualification did not have a significant association, but employment status had a significant association. A study conducted on a literate population in Lagos in 2007 showed that public health education improved both knowledge and attitude of the study participants on SCD and screening uptake (Leadership Newspapers Nigeria. June 28, 2012.Four million Nigerians suffering from sickle cell disease Pg. 1. www.leadership.ng/nga/articles/28405/2012/06/28).

The general opinion of some women who usually want to go the way of love rather than having a broken relationship is mostly tended towards their religious beliefs on miracles and destiny. Some believe that, since it is a 1 in 4 chance of having a SCD child they will rather go for just one or two children. Selective termination of pregnancy (abortion) is another aspect that might influence some women as some religious denominations kick against all forms of abortion. Therefore, more public sensitisation is required to be carried out using all available resources such as the print media, social media, schools, mosques, churches and seminars. This will enhance the practice of the youths on SCD premarital counselling. Almost eighty nine percent of the study participants believed that it was important an Indivldual meets a Counsellor

before planning a pregnancy. This is an improvement compared to 64% reported in a study among students of the University of Lagos in 2007 (Leadership Newspapers Nigeria. June 28, 2012. Four million Nigerians suffering from sickle cell disease Pg. 1. www.leadership.ng/nga/articles/28405/2012/06/28). From the questionnaires administered among the women, a total of 379 (93.1%) believed that it was useful to know if they have sickle cell trait, while 367 (90.2%) believed that it was useful to know if their partner has sickle cell trait. This study showed that 329 (80.8%) of the respondents thought that having a higher level of education would increase the knowledge of sickle cell disease. This was close to the 86% reported by Adeyemo et al. (2007) for respondents with knowledge of sickle cell disease. Respondents who accepted that SCD was a serious disease and do give them psychological trauma whenever they remember that they have children with SCD were 337 (82.8%), while 312 (76.7%) accepted that it was a scary disease. Government, religious institutions, schools and hospitals should be more involved in raising awareness about genetic counselling to help in the prevention of giving birth to offspring with sickle cell diseases, which would in turn prevent the social and financial burdens of managing a child with sickle cell disease. Establishment of proper guidelines as regards genetic counselling process is highly necessary for the implementation of genetic testing /genetic counselling in Nigeria (Adeyemo et al. 2007). It was found after analysis that 59.7% accepted that religious organisations play a role in the prevention of sickle cell disease and about 58% believed that there was no reincarnation of a child that dies from sickle cell disease which is similar to the work of Nnaji et al. (2013).

While Adeyemo et al. (2007) reported that only 11% of their subjects were pessimistic about any cure for SCD from faith/prayers or otherwise, 39.6% of women in this study rejected that saying prayers daily could cure a child with SCD.

Some women of the age bracket 35 – 44 were not willing to complete the forms for some ethical reasons, thus, some subjects were drawn from this age bracket. Therefore, there is an urgent need to expedite action on public health education to enlighten people about the inheritance of sickle cell disease and placing an emphasis on advocacy for genetic counselling at the national level for its prevention. Moreover, given the fact that individuals who are at risk (sickle cell trait carriers) could easily be identified by inexpensive blood tests, access to genetic testing/ counselling would help to explain genetic risks and this will lead to a reduction in births of affected offspring. More research is needed in the area of public perception of genetic testing/counseling in health care systems in Nigeria (Adeyemo et al. 2007).

Omuemu et al. 2013 reported that 78.9% of study participants in a study among undergraduates in University of Benin knew that SCD could be detected by screening/simple blood test, while 80.8% of women in this study correctly said the same thing. This study revealed that there was a high level of knowledge of sickle cell disease among the respondents based on their blood genotypes. This is not surprising since the respondents are a population of women who are almost 100% mothers of children with sickle cell disease. Their major sources of information about sickle cell disease as revealed by the study were from the media (print and electronic), workshops/seminars, lectures and health facilities they bring their children to. Similar findings have been reported from studies among undergraduate students in different parts of the country (Animasahun et al. 2009).

In this study, over 90% percent of respondents with tertiary and above level of education had good knowledge of premarital genetic counselling In high-income countries that provide neonatal diagnosis and care for patients, most survive well into adult life (Modell et al. 2007). This is because there is limited use of prenatal

diagnosis; numbers of patients are rising steadily. Most affected children born in low-income countries still die undiagnosed, usually from malaria but things are changing (Model and Darlinson, 2008).

About 40% of Africa is now urbanised, and improved access to health care is leading to increased survival and rising demand for hospital services (Akinyanju et al. 2005). Community-based services including information, prophylactic antimalarial or antibiotics, and social support greatly improve survival and quality of life and reduce demand for acute hospital services – in short, it is less costly to make organised care available than not.

Over six million Africans will be living with a sickle cell disorder – clearly, care for these disorders must become part of primary care wherever they are common (Model and Darlinson, 2008).

There is a strong case for carrier screening in Africa. Cheap and simple methods for testing adults and newborns exist. Knowledge of risk allows a range of options, including limiting of family size, ensuring that at-risk infants are tested at birth, and requesting prenatal diagnosis. DNA-based early prenatal diagnosis is available at several African centres and is relatively inexpensive when only the sickle variant is sought. However, as few couples can afford even a subsidised fee there is insufficient information on likely uptake if the service were freely available (Adeyemo et al. 2007).

This study like many studies on SCD faced some challenges. As Abioye-Kuteyi et al. 2009 and Abdulrahaman et al. 2013 discussed challenges regarding studies on SCD. The challenges included: Refusal of a few study participants to participate in the study due to the psychological trauma they were passing through. There were excessive protocols and bureaucracy in allowing the study to be carried out with ease in some

Government health institutions. Challenge of funding whereby not all study participants who truly showed interest and participated in the study could get incentives. The number of research assistants recruited was not enough for a study of this nature. Questions were not provided on the family history of the study participants regarding past incidence of other genetic diseases.

More than 90% of the respondents in this study accepted that PGC is important for unmarried individuals. They believed that it was necessary to do PGC with their partner when either or both have haemoglobinopathy. This emphasised the urgent need for focused health education as the foundation of genetic counselling before marriage. As expected, knowledge scores for PGC and SCD were significantly higher in the group with tertiary and above education compared to the group with less education. With a generally favourable knowledge towards SCD and a significant association between some sociodemographic characteristics and outcome variables, the benefits are that morbidity and mortality from SCD would decline if PGC is embraced backed by legislation and implementation in Nigeria. This will further positively influence disposition of intending couples to genetic screening, partner discussion and marital decisions, the universal provision of and emphasis on sickle cell screening and genetic counseling especially long before marriage as recommended by WHO (Jeremiaj et al. 2007) Also, this will further facilitate informed rational marital decisions and reproductive behaviour that will lead to a sharp decline in new births of children with SCD among women (Rafi & Spicer, 2007).

Apart from direct assessment experience of socioeconomic characteristics of women who have children with SCD, recognition of important neighbourhood factors could

guide the clinician to identify specific subgroups of children with SCD who are at-risk (Palermo et al. 2008).

The finding from this study is commendable as it will help to make interventions such as health education more feasible. It will also contribute to a reduction in child mortality as desired in the Millennium Development Goal 4 (Omuemu et al. 2013). It is worth noting that majority of the respondents reported that their cultural and religious beliefs will not influence their decisions to accept genetic counselling for sickle cell disease. They said that they would not make the mistake they made earlier in life due to majorly their ignorance of not knowing their blood genotypes and that of their partners before pregnancy. This is at variance with the findings in a study conducted in North-Eastern part of Nigeria, a predominantly Islamic region, where religion was a factor militating against acceptability of prenatal diagnosis of sickle cell (Omuemu et al. 2013).

CONCLUSION

This study showed a high frequency for the proportion of women who had ever had genetic counselling for SCD. This was due to the fact that respondents have been exposed to series of genetic counselling since they discovered that their children have SCD. However, some of the women with low level of education found it difficult understanding the reasons why intending couples should go for genetic counselling before marriage. This affirms that more public awareness on the importance of having the knowledge of PGC and SCD should be intensified.

In addition, the relationship between the women's socio-demographic characteristics and knowledge and practice related to genetic counselling for SCD were significant. Information passed on to the mothers and caregivers when they come to the health facilities could be responsible for this. On the other hand, confounders like attending seminars on SCD and information through mass media could have also been responsible for the significant association.

Religious and cultural beliefs were found to be significant. However, they were less influential on the response of the women. This was because over eighty percent of the respondents rejected that it was the destiny of a child with SCD to have the disease, while almost half rejected that there was reincarnation of a child that dies from SCD. Although, this could be said to be encouraging; this is not the case in the rural and some parts of Nigeria where there are superstitious beliefs about SCD which negatively influenced by culture and religion.

It will be note-worthy to state here that all the necessary health authorities should be involved in reducing and preventing the public health challenges posed by SCD in order to drastically decrease the morbidity and mortality caused by SCD by making PGC compulsory for intending couples.

RECOMMENDATIONS

- Religious societies and organisations should create platforms to educate their members on the importance of PGC. They should make it a criterion especially before marriages are conducted.

- Sickle cell associations/clubs should be created at several locations in Nigeria as there are none in some states in Nigeria currently. This will serve as psychosocial support for parents and their affected children to freely talk about their conditions and be rehabilitated.

- Medical treatment of sickle cell disease should be provided and highly subsidised by the government if not entirely free to make it affordable and accessible to all.

- There should be advocacy for girl-child education because women with at least secondary school level of education were the ones that scored high for knowledge based questions in this study.

- Emphasis should be laid on PGC and SCD in health science and Biology syllabus right from the secondary school. This will give the students all the necessary information about PGC and SCD before adulthood.

- Government in collaboration with Non-Governmental Organisations (NGOs) should run rehabilitative centres for children or individuals with sickle cell disease where parents and their spouses are involved too.

- Future studies could focus on women who come for PGC and are about to get married.

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APPENDICES

APPENDIX 1: Letter of Introduction from Project Supervisor for Ethical Approval

DEPARTMENT OF EPIDEMIOLOGY AND MEDICAL STATISTICS

FACULTY OF PUBLIC HEALTH
COLLEGE OF MEDICINE
UNIVERSITY OF IBADAN, NIGERIA.

Telephone: +234-0702927692
(Direct Line)
+234-2-2410088 Ext: 2661
E-mail: emaph@comui.edu.ng

Ag Head: Dr. Olatunbosun Enwere

ACADEMIC STAFF
EPIDEMIOLOGY

Olatunbosun O. Enwere, Reader
M.Sc. (Epid. & Biost.)
FWACP (CPH)

Olatunbosun O. Ajayi, Senior Lecturer
M.Sc. (Epid.), FMCGP (Nig.)
MPH (USA) M.D.L.S.C. (Canada)

Senior Lecturer
M.Sc. (Epid. & Med. Stat.)

Senior Lecturer
M.Sc. (Epid. & Med. Stat.)

MEDICAL STATISTICS

Professor
M.Sc. (Med. Stat. London)

Professor
M.Sc. (Med. Stat. London)
M.Sc. (Biostatistics) (London)

Senior Lecturer
M.Sc. (Med. Stat.)

9th September, 2014

The Ethics Review Committee
Ministry of Health
Lagos State.

Dear Sir/Madam,

RE: LETTER OF INTRODUCTION – Olanlyan Olubukola Hakeem

I write to introduce to you the above-named MSc (Epidemiology and Medical Statistics) student with matric no. [illegible] in this department.

His work has been reviewed and approved by us and he requires ethical approval to study Master's project in your Ministry.

Olanlyan is to carry out a study on the topic: "Knowledge and Utilization of Premarital Genetic Counselling as a Preventive Measure Against Having Children with Sickle Cell Disease Among Women in Lagos, Nigeria".

Kindly give him all necessary assistance.

Yours sincerely,

COLLEGE OF MEDICINE
DATE
TIME
UNIVERSITY OF IBADAN

APPENDIX 2: Letter of Introduction for Ethical Approval by Project Student

Department of Epidemiology and Medical Statistics
Faculty of Public Health
College of Medicine
University of Ibadan
30th September, 2014.

The Permanent Secretary,
Ministry of Health,
Lagos State Secretariat,
Alausa-Ikeja,
Lagos State.

Sir/Madam,

A LETTER OF APPLICATION FOR ETHICAL APPROVAL

I, Olaniyan Olatunji Hakeem, a student of the above named institution of learning [illegible] letter to request for an ethical approval pertaining to my Master's Degree Project which [illegible] done in Lagos.

The topic of my study is "Knowledge and Practice of Premarital Genetic Counselling as a Preventive Measure against having Children with Sickle Cell Disease among [illegible] Nigeria". I have presented the topic as a proposal and it has been accepted and approved by my Project Supervisor.

Furthermore, I have attached other relevant materials to this letter.

I would be glad if my application is given a prompt attention and a favourable response in order for me to proceed with my project research as quickly as possible.

Thank you.

Yours faithfully,

Olaniyan Olatunji Hakeem
(MSc Epidemiology and Medical Statistics) 08062412774 olahakeemola@gmail.com

APPENDIX 3: Ethical Approval from Lagos State Ministry of Health

APPENDIX 4: Questionnaire used in the field study.

QUESTIONNAIRE NUMBER: .....

A QUESTIONNAIRE ON THE RESEARCH TOPIC:

"KNOWLEDGE AND PRACTICE OF PREMARITAL GENETIC COUNSELLING AS A PREVENTIVE MEASURE AGAINST HAVING CHILDREN WITH SICKLE CELL DISEASE AMONG WOMEN IN LAGOS, NIGERIA."

DEPARTMENT OF EPIDEMIOLOGY AND MEDICAL STATISTICS
FACULTY OF PUBLIC HEALTH
COLLEGE OF MEDICINE
UNIVERSITY OF IBADAN, NIGERIA.

My name is OLANIYAN Oladele Hakeem. I am a Master's student in the Department of Epidemiology and Medical Statistics of the University of Ibadan. I am conducting a study on the above topic in partial fulfillment of an MSc Epidemiology and Medical Statistics.

The study is to investigate the knowledge and practice of premarital genetic counselling as a preventive measure against sickle cell disease as well as to know the challenges of having children with sickle cell disease among women.

Your participation will go a long way in ensuring the success of the study. On the other hand, you have the right to or not to participate in the study.

All information gotten from you would be treated with full confidentiality.

It is hoped that the findings from this study will help improve the knowledge and practice of premarital genetic counselling and reduce the rise in sickle cell disease.

Thank you.

OLANIYAN Oladele Hakeem (08062412774; olahakeemola@gmail.com)

I have read the above introduction of the interviewer and have consented to participate in the study.

Thumbprint/Signature of Respondent ..... **Date .....**

SECTION A: SOCIODEMOGRAPIC AND SOCIOECONOMIC CHARACTERISTICS OF RESPONDENT.

1. Serial number .....
2. Age at last birthday .....
3. Religion [1] Islam [2] Christianity [3] Traditional [4] Others, please specify.....
4. Marital status [1] Cohabiting [2] Single motherhood [3] Separated/Divorced [4] Married [5] Widowed
5. If married, for how long? .....
6. Number of children .....
7. Ethnic group [1] Yoruba [2] Hausa [3] Igbo [4] Others, specify.....
8. Highest level of education attained [1] No formal education [2] Primary [3] Secondary [4] Tertiary and above
9. Employment status [1] Employee [2] Self-employed [3] Unemployed
10. Monthly income [1] #6,000-#20,000 [2] #21,000-#35,000 [3] #36,000-#50,000 [4] #51,000-#75,000 [5] Others, specify .....
11. Blood genotype [1] HbAA [2] HbAS [3] HbAC [4] HbSS [5] Others, specify .....

SECTION B: INFORMATION ON CHILD WITH SICKLE CELL DISEASE (SCD).

12. Age of child as at last birthday .....
13. Current level of child's education [1] None [2] Kindergarten/Playgroup [3] Nursery [4] Primary [5] Secondary
14. Sex of Child [1] Male [2] Female
15. Primary daily activities you give the child [1] Feeding [2] Bathing [3] Washing [4] Toilet cares [5] School homework [6] Managing drugs/treatment [7] Others, specify .....
16. Frequency of clinic visits in the last three months [1] 1-3 [2] 4-6 [3] 7-9 [4] 10-12 [5] 13-15
17. Frequency of pain crises in the last three months [1] 1-3 [2] 4-6 [3] 7-9 [4] 10-12 [5] 13-15
18. Length of hospital admission in the last three months [1] 1-5 hours [2] 6-10 hours [3] 24 hours [4] 2- 5 days [5] One week [6] Others, specify .....
19. Frequency of emergency hospitalisations of child in the past six months [1] 1-3 [2] 4-6 [3] 7-8 [4] Others, specify .....
20. Number of days child has been absent in school in the last six months [1] 1-5 [2] 6-10 [3] 11-15 [4] 16-20 [5] Others, specify .....
21. Reasons for child's absence in school in the last six months

    .....

    .....
22. Current treatment regimen of child[1] Oral fluid/intravenous saline [2] Paracetamol [3] Analgesic [4]Bed rest[5]Herbal medications[6]Bone marrow transplant [7]Others, specify .....
23. Which medical symptoms does the child show? [1] Jaundice [2] Stunting [3] Vaso-occlusion [4] Acute chest pain [5] Cholethiasis [6] Reduced abdominal circumference [7] Kidney failure [8] Others, specify .....
24. What are your feelings towards the child? [1] Positive [2] Negative [3] Indifferent
25. Please, briefly explain your choice of answer from above

    .....

    .....
26. Do you have any child who did not survive SCD? [1] Yes [2] No
27. If yes, please briefly explain about the child in details

    .....

    .....

SECTION C: KNOWLEDGE AND PRACTICE OF PREMARITAL GENETIC COUNSELLING (PGC).

28. Having premarital genetic counselling is important for unmarried individuals.
    Strongly Disagree 1 2 3 4 5 Strongly Agree
29. It is important I meet a Counsellor before I plan a pregnancy.
    Strongly Disagree 1 2 3 4 5 Strongly Agree
30. It is useful to know if I have sickle cell trait.
    Strongly Disagree 1 2 3 4 5 Strongly Agree
31. It is useful to know if my partner has sickle cell trait.
    Strongly Disagree 1 2 3 4 5 Strongly Agree
32. Knowing the risk of having a child with sickle cell disease would change how I plan a pregnancy.
    Strongly Disagree 1 2 3 4 5 Strongly Agree

33. My partner would be hard to convince to go for PGC.
Strongly Disagree 1 2 3 4 5 Strongly Agree
34. My life would be different if I had known about PGC and practiced it.
Strongly Disagree 1 2 3 4 5 Strongly Agree

SECTION D: RESPONDENT'S KNOWLEDGE OF SICKLE CELL DISEASE (SCD).

35. Sickle Cell Disease is a serious disease.
Strongly Disagree 1 2 3 4 5 Strongly Agree
36. Having a child with sickle cell disease would be very scary.
Strongly Disagree 1 2 3 4 5 Strongly Agree
37. My life would change if my child had sickle cell disease.
Strongly Disagree 1 2 3 4 5 Strongly Agree
38. Having a higher level of formal education can increase the knowledge of SCD.
Strongly Disagree 1 2 3 4 5 Strongly Agree

39. Sickle Cell Disease is caused by
[1] dirty needles
[2] a virus
[3] inheriting genes from parents
[4] bad blood
[5] none of the above

40. How many genes must someone inherit to have Sickle Cell Disease?
[1] zero, it is not caused by genes
[2] one from their mom
[3] two, one from their mom, and one from their dad
[4] three, one from mom, and two from dad
[5] none of the above

41. Sickle Cell Disease can cause
[1] severe debilitating pain
[2] strokes
[3] infections
[4] organ damage
[5] all of the above

42. Sickle Cell Disease makes red blood cells
[1] round and soft
[2] hard and sickle shaped
[3] sticky and blue
[4] stiff and round
[5] soft and sickle shaped

43. Sickle Cell Disease is easily cured by
[1] antibiotics
[2] liver transplant
[3] rest
[4] blood transfusions
[5] none of the above

44. How can you tell if someone carries the gene for sickle cell disease?
[1] They look sick
[2] They will eventually have Sickle Cell Disease
[3] With a simple blood test
[4] There is no way of knowing
[5] None of the above

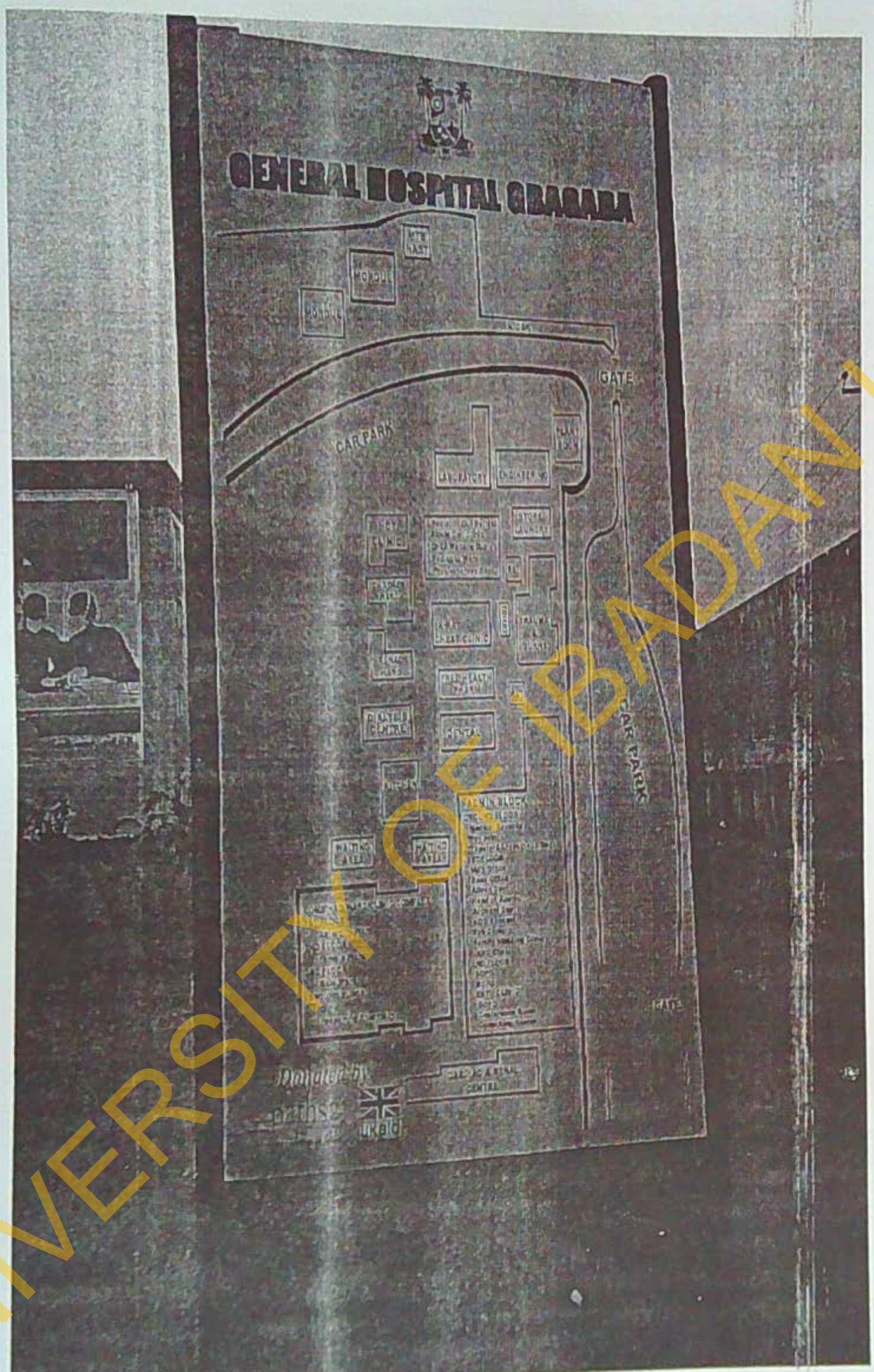
SECTION E: RELIGIOUS AND CULTURAL BELIEFS.

		STRONGLY DISAGREE(1)	DISAGREE(2)	INDIFFERENT(3)	AGREE (4)	STRONGLY AGREE(5)
45.	Religious organisations play a role in preventing SCD.					

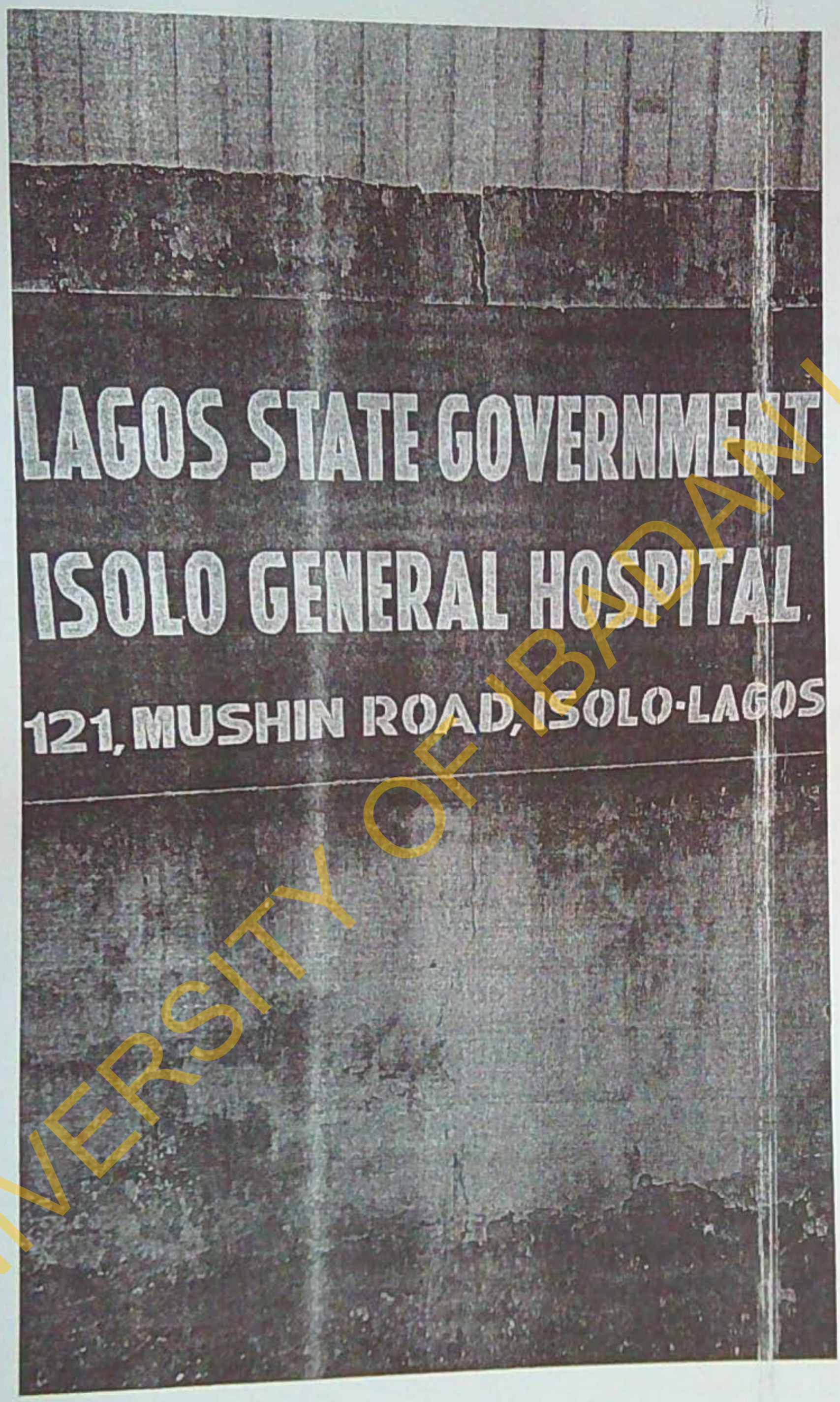
46.	There is no reincarnation of a child that dies from SCD.					
47.	Saying prayers daily could cure a child with SCD					
48.	It is the destiny of a child with SCD to have the disease.					
49.	It is not a taboo to know if I have sickle cell trait.					
50.	It is not a taboo to know if my partner has sickle cell trait.					
51.	SCD could happen in any ethnic group.					

APPENDIX 5: Project Student with SAMI Coordinator after interview with Study participants.

APPENDIX 6: Frontage of one of the study centres used for this Project work



APPENDIX 7: Entrance to one of the study centres used for this Project work



APPENDIX 8: Other terms used in this Project work and their definition.

1. **Priapism**: A persistent and usually painful erection of the penis that lasts for more than four hours in the absence of sexual arousal.
2. **Vaso-occlusion**: A common and painful complication of sickle cell anaemia in adolescents or adults leading to obstruction of blood circulation in the blood vessels due to sickled red blood cells.
3. **Avascular necrosis**: It is also called 'osteonecrosis.' It is a disease where there is cellular death of bone components due to interruption of the blood supply leading to destruction of joint articular surfaces.
4. **Cardiopulmonary disease**: A disease that affects the heart and the lungs at the same time.
5. **Mutation**: It is a change in the structure of the gene caused by alteration of a single base unit of the DNA or the deletion, insertion or rearrangement of larger sections of the genes or chromosomes resulting in a variant (different) type of gene and may be transmitted to subsequent generations.
6. **Gnathopathy**: A condition also called sickle cell jaw protrusion as a result of sickle cell disease.
7. **Taboo**: A statement made by a social custom or culture on the prohibition or ban of certain things.
8. **Reincarnation**: The rebirth or coming back to life of a particular soul after its death into another person's body.
9. **Destiny**: It also means fate. It is a power of thought in a Supreme Being to control all events of life.

10. **Opiates**: They are narcotic analgesic drugs used to directly depress the central nervous system. They are also usually reserved for the treatment of acute (short-term) back or leg pain. Examples are heroin, morphine and tramadol. They are also called opioids or narcotics.
11. **Frontal bossing**: It is also called Skull bossing. It is an unusually prominent forehead that is sometimes associated with a heavier than normal brow ridge.
12. **Thin extremities**: A person who suffers thin extremities has thin hands and legs with big and fat stomach. The body parts are disproportionate.
13. **Response rate**: The proportion or percentage of those invited to participate in a research and actually do so out of the total expected participants.
14. **Pre-test Questionnaire**: It is a type of questionnaire used before the main study to those of the individuals who will be used in the main study. It is used to know the pattern of response of items and also to know the amount of time it may take to respond to the questionnaire.
15. **Reliability test**: It is a statistical concept that is based on the association between two sets of scores representing the measurement obtained from the questionnaire when it was used with a group of individuals.

APPENDIX 9: Tables showing some other variables studied in this Project work.

Table I: Length of marriage in years of study participants

Number of years	n = 407 Frequency(Percent)
none	74(18.5)
less than 1 year	2(0.5)
1-3	9(2.2)
4-6	27(6.6)
7-9	32(7.9)
10-15	139(34.2)
16-20	64(15.7)
21 years or more	60(14.7)

Table II: Blood genotypes of study participants

Blood genotypes	n = 407 Frequency(Percent)
HbAA	10(2.5)
HbAC	300(73.7)
HbAS	42(10.3)
HbSS	7(11.5)
HbSC	8(2.0)

Table III: Study participants who had lost one or more of their children as a result of sickle cell disease

Response	n = 407 Frequency(Percent)
Yes (had child/children who died)	53(13.0)
No (does not have child who has died)	354(87.0)

Table IV: Number of days child had been absent from school in the last six months due to sickle cell disease.

Numbers of days	n = 407 Frequency(Percent)
None	119(29.2)
1-5	137(33.7)
6-10	85(20.9)
11-15	27(6.6)
16-20	22(5.4)
3 weeks or more	17(4.2)

Table V: Frequency of pain crises of study participants' children with sickle cell disease in the last three months.

Pain crises	n = 407 Frequency(Percent)
None	83(20.4)
1-3 times	234(57.5)
4-6	45(11.1)
7-9	23(5.7)
10-12	20(4.9)
13-15	2(0.5)

Table showing medical symptoms showed by Respondents' children with sickle cell disease.

Medical symptoms	n = 407 Yes Frequency	No (percent)
Jaundice	223 (54.8)	184 (45.2)
Stunting	102 (25.1)	305 (74.9)
Vaso occlusion/avascular necrosis	93 (22.9)	314 (77.1)
Acute chest pain	207 (50.9)	200 (49.1)
Cholelithiasis	8 (2.0)	399 (98.0)
Reduced abdominal circumference	35 (8.6)	372 (91.4)
Kidney failure	4 (1.0)	403 (99.0)
Priapism	5 (1.2)	402 (98.8)
Brain infarction/strokes	4 (1.0)	403 (99.0)
High temperature/fever	14 (3.4)	393 (96.6)
Cough and catarrh	3 (0.7)	404 (99.3)
Headache, stomach and body pains	70 (17.2)	337 (82.8)
Swollen abdomen/body parts	11 (2.7)	396 (97.3)
Liver infections	3 (0.7)	404 (99.3)