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The Impact of Premarital Genotype Screening on Sickle Cell Disease, Among Undergraduate Students in Nigeria.

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ABSTRACT

Genetic compatibility needs to be carefully considered in an effort to lessen the frequency of sickle cell disease (SCD), especially in Nigeria and other African where the disease has a high burden with morbidity and mortality. The population for the study are undergraduate of Al-hikmah University Ilorin, the multi-stage sampling technique was used in sample selection of the 341 respondents, and data was collected using a self-administered questionnaire. Findings indicate 88.3% fell within age range of 18 – 25 years, 70.1% were female and single representing 93.5% and 76% have heard understand SCD and are generally familiar with the complications of SCD. Only 56% have good knowledge of PMS as the cheapest prevention, while 63% don't know about premarital sickle cell counselling. Only 56% knew their Hemoglobin genotype, however 12% knew Hemoglobin genotype of their fiancés and 69% are of the opinion of terminate relationship if the genotype result is incompatible. 68% support the establishment of laws prohibiting marriage in cases of PMS incompatibility. The statistical analysis given that P-value is 0.000, which is < alpha level of 0.05, therefore null hypothesis rejected, it was concluded that knowledge of genetic composition and the potential threat of SCD was robust, mandatory PMS with better counselling strategies and willingness in advocating PMS to intending couples was confirmed. Recommendations, carrier identification and genetic counselling enlightenment on SCD and PMS testing through mass media and screening centers should be made by the government and religious institutions

KEYWORDS: *Premarital, Genotype, Screening, Sickle cell, Disease.*

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1. Introduction

One of the most serious medical conditions in the world is sickle cell disease World Health Organization (WHO, 2017). Sickle cell disease (SCD) is a genetic inheritance that can be prevented, which affects a large number of people worldwide and is brought on by aberrant hemoglobin. Because of a single gene mutation. Babalola et al., (2019) Explain that an individual with sickle cell disease (SCD) has inherited two abnormal hemoglobin (Hb) genes, at least one of which is accountable for the creation of sickle hemoglobin (HbS). In contrast, the inheritance of two faulty HbS genes results in HbSS. The homozygous, or HbSS, clinical phenotype also referred to as sickle cell anemia is the most prevalent. In addition to SS, compound heterozygous HbSC is sometimes referred to as SCD, or sickle cell disease. Both hemoglobin SC disease (HbSC) and sickle cell anemia (SCA) are inherited disorders with an autosomal recessive pattern of inheritance.

Knowledge of premarital genotype screening presents an opportunity for individuals to become informed about their genetic predisposition to diseases (sickle cell) and for intending couples to be aware of the

possible genetic characteristics of their unborn children (Babalola et al., 2019). Preventing genetic inheritance that could have serious consequences if one believes that having children is one of the reasons for marriage. The cheapest methods of preventing genetic disease include premarital genotype screening and genetic counselling, prenatal diagnosis, preconception diagnosis and implantation of normal embryos after in vitro fertilization and in vitro therapy using stem cell transplantation (Al-qattan et al., 2019). Prevention of the disease through carrier identification and genetic counselling remains the only realistic approach to reduce the impact of the disease and allow better use of available resources in the low income countries where the condition is most prevalent (Al-qattan et al., 2019)

According to the WHO approximately 240 million people are carriers of genetic disease and at least 500,000 affected individuals are born annually (Colombatti et al. 2022). The prevalence of genetic disease is becoming higher in society, creating more stress despite difficulties people encounter in life. Sickle cell disease contributes to the equivalent of 15% of under five deaths on the African continent, more than 9% of such deaths in West Africa (Thomson et al., 2023). The literature analysis noted that mothers have low knowledge and awareness of their genotype, despite the fact that sickle cell trait (AS & AC) is a risk factor for having children with SCD. It has been noted that some individuals are unaware of sickle cell illness prior to marriage, and some go on to become mothers to children with the illness. In the modern world, couples simply move forward with marriage decisions regardless of their genetic status, which presents a significant difficulty when it comes to incompatibility (Colombatti et al., 2022). Most youths today are either unmarried or intending to get married and will procreate in the future. This group of persons is the target population who will benefit from appropriate interventions aimed at preventing and controlling genetic disease, especially sickle cell disease. Thus, there is a need to ascertain the impact of premarital genotype screening on sickle cell disease among young undergraduates University students of Al-Hikmah Ilorin. Nigeria, examine the effect of their demographic data and potential respondents' engagement in addressing the issue of PMS and SCD, so as to direct interventions in reducing the reproductive risk of genetic diseases, thereby contributing to a decrease in the prevalence of genetic diseases in Nigeria.

2. Methodology

The study was conducted at Al-Hikmah University, Ilorin Kwara State. Nigeria. Ilorin is one of the biggest cities in North Central Nigeria. Geographically it is situated in the guinea savannah belt at latitude 8" North and longitude 4" East (8°30′N 4'33'E), with three local Government The country's capital, Abuja, is 500 kilometers from the ancient city of Ilorin, and 300 kilometers from Lagos. Al-Hikmah University is one of the Universities in Ilorin, established in 2005 The University granted a license to function as a Conventional Private University (License No. 010), seven faculties and postgraduate School, comprise Al-Hikmah University's current structure. Descriptive design was adopted for the study and a questionnaire was used to elicit necessary information from the respondents. The population unit of study are undergraduates of the University who are between the ages of 18-30 years. The survey was administered to 357 undergraduate respondents. Out of the 357 copies of questionnaires, 346 was retrieved, equalling 96.9% of response rate. Also, out of the 346 retrieved, only 341 (98.5%) were useful, as there are incomplete questionnaires and missing responses. They were selected across levels, disciplines and faculties. The Multi-stage sampling technique was used in sample selection of the 341 who participated voluntarily. The process via which the components of the study sample are determined is known as the sampling technique. It is thought of as a sampling tool as well. For the study, a survey-style descriptive research design was used. The study's population consisted of all enrolled regular undergraduate students at Al-Hikmah University in Ilorin, Kwara State; a total of 6,200 respondents were used. stage one, stratified, sample random sampling technique were chosen, since they already exist in a stratum, two (2) departments from each of the seven (7) faculties on the Al-Hikmah University Ilorin campuses the Faculty of Natural Sciences, the Faculty of Applied Sciences, the Faculty of Humanities, the Faculty of Social Sciences, the Faculty of Health Sciences, and the Faculty of Agriculture. In the second stage, The Sample size was determined by Fisher's formula of; $n = Z^2pq/ d^2$ the calculation resulted 357 respondents the targeted population at the department level were chosen using the proportionate sample technique. While stage three involved recruiting participants directly through departmental student organizations, a simple random sampling technique using the class rolls of each department as the sampling frame was used to select participants with a variety of backgrounds, ages, academic disciplines, and levels; informed consent was obtained from each participant prior to their involvement in the study. The questionnaire was pilot-tested prior to data collection. Data obtained was analyzed and presented in the frequency table, percentages, and chi-squared analysis. Anonymity and confidentiality of information were maintained. The study is a cross sectional survey to assess the

understanding of premarital genotype screening on sickle cell diseases. Quantitative research is chosen for its ability to explore the subjective knowledge, attitude, and perception of individuals in their natural context.

3. Results

Table 1: Demographic characteristics of the respondents

Variable	Frequency	Percentage
Age 18 – 25	301	88.3
26 – 33	40	11.7
Total	341	100%
Gender: Male	102	29.9
Female	239	70.1
Total	341	100%
Marital Status		
Single	319	93.5
Married	22	6.5
Total	341	100%
Religion		
Islam	305	89.4
Christianity	36	10.6
Total	341	100%
Year of study		
100	44	12.9
200	75	22.0
300	103	30.2
400	91	26.7
500	28	8.2
Total	341	100%

Data from table 1 above presents the respondents demographic characteristics, age fell within the 18–25 (88.3%) of the total. The gender distribution slightly skewed towards females (70%), the respondents had diverse academic years of study with 300 level accounting for (30.2%), 400 level (26.7%) 200 level (22.0%) 100 and 500 level smallest group. Based on religion, (89.4%) practice Islam and (10.6%) practice Christianity, respectively. (93.5%) are single representing. While the tribe distribution Yoruba (68.0%) Hausa (21.7%) as shown in table 1.

3.3.1 Knowledge of premarital genotype screening on sickle cell disease

Table 2: Respondents' Knowledge of premarital Genotype screening on Sickle cell

Knowledge of premarital genotype screening and sickle cell disease was assessed using eight (8) knowledge items**.** Each correct response was awarded one (1) point**,** while each incorrect response was awarded zero (0) points**.**

Statement	**Respondents Opinion (frequency)**		**Percentage %**	**Score**
What do you understand by sickle cell disease SCD	Sexually transmitted	38	11	0
	Inherited blood disorder	208	60.9	1
	An Infectious disease	82	24	0
	Punishment from God	13	4	0
	Total	**341**	**100%**	**1**
What is the mode of sickle cell disease SCD transmission and sickle cell carrier/trait	Contact with infected person	65	19.1	0
	Infection from the mother	82	24	0
	Hereditary from the parents	191	56	1
	Contacted after childbirth	3	0.9	0
	Total	**341**	**100%**	**1**
Have you ever heard the term "sickle cell Anemia"	Yes 215		Yes 63	1
	No 126		No 37	0
	Total	**341**	**100%**	**1**
Which of the following genotype is called sickle cell	AA	7	2	0
	AS	62	18	0

Anemia	SS 177 SC 95 **Total 341**	52 28 **100%**	1 0 **1**
Which of the following are Medical complications are caused by sickle cell disease	Repeated Pain episode 167 Avascular Necrosis 119 Stroke & Anemia 17 Cough & stool 38 **Total 341**	49 35 4.9 11.1 **100%**	1 0 0 0 **1**
Do you know if there is a cure for sickle cell disease	Yes 133 No 208 **Total 341**	Yes 39 No 61 **100%**	0 1 **1**
What is premarital genotype screening	Medical screening for intending couple 201 Genotype test 72 Medical checkup 17 All of the above 51 **Total 341**	59 21 5 15 **100%**	1 0 0 0 **1**
PMS program is available in Nigeria	Yes 242 No 99 **Total 341**	Yes71 No 29 **100%**	1 0 **1**

Result in table 2 indicate that a significant proportion of the respondents (76%) have heard about sickle cell disease SCD, the majority of respondents (61%) understand sickle cell disease as inherited blood disease, (56.1%) understand sickle cell disease as hereditary from the parents, and the majority of respondents (63%) have heard the term Sickle cell anemia, similarly significant proportion of the respondents (59%) and (28%) understand that an individual with genotypes SS and SC has sickle cell disease, respectively. The respondents were generally familiar with the complications associated with sickle cell disease; (49%) and (35%) understood repeated pain episodes, and avascular necrosis, anaemia, and stroke as common complications of SCD. However, (61%) believe that SCD has no cure yet, and (39%) of the respondents are of the opinion that SCD has a cure as indicated in table 2. Regarding the knowledge of premarital genotype screening, (59%) of the respondents defined it as the medical screening carried out for intending couples, and (71%) of the respondents knew that PMS test is available in Nigeria. The (59%) of respondents understand that PMS comprises Genotype test, HIV/AIDs test and blood group screening. A significant number (58%) understand PMS as prevention against hereditary diseases.

3.3.2 Attitude towards premarital genotype screening on sickle cell diseases

Table 3 Attitude towards premarital genotype screening on sickle cell diseases. Attitude questionnaire were scored using a dichotomous system. A positive response ("Yes") was assigned a score of 1**,** while a negative response ("No") was assigned a score of 0

Statement	Frequency	Percentage %	Sore
Are you aware of premarital sickle cell counseling	Yes 126 No 215 **Total 341**	Yes 37 No 63 **100%**	0 1 **1**
Will you be part of advocate/recommend on PMS for all couple	Yes 259 No 82 **Total 341**	Yes 76 No 24 **100%**	1 0 **1**
Do you know your genotype composition	Yes 191 No 150 **Total 341**	Yes 56 No 44 **100%**	1 0 **1**
Do you know the genotype of your fiancés /fiancée	Yes 41 No 300 **Total 341**	Yes 12 No 88 **100%**	0 1 **1**

Result in Table 3 Shows that 63% of the respondents don't know about premarital sickle cell counselling, only 37% aware of premarital sickle cell counselling. While 76% of the respondents are ready to recommend and advocate PMS for all intending couples and young unmarried. (56%) of the respondents knew their Hemoglobin genotype composition, (44%) don't know (out of which (82%) are ready to undergo PMS and (18%) are not interested in PMS). However only 12% of respondents knew the Hemoglobin genotype of their fiancés /fiancée.

3.3.3 Perception of premarital genotype screening on sickle cell diseases

Table 4. Perception of premarital genotype screening on sickle cell diseases

The perception of respondents towards premarital genotype screening was assessed using four dichotomous (Yes/No) items. Each response reflecting a positive perception was assigned a score of 1, while a negative perception was assigned a score of 0. For negatively worded items, reverse scoring was applied

Statement	Respondents Opinion	Percentage %	Score
Would you still go on to marry your partner even if the genotype result shows (incompatibility) possibility of having a child with sickle cell disease	Yes 106 No 235 **Total 341**	Yes 31 No 69 **100%**	0 1 **1**
Is premarital genotype screening against your religious belief	Yes 27 No 314 **Total 341**	Yes 8 No 92 **100%**	0 1 **1**
Will you support mandatory of PMS in Nigeria	Yes 232 No 109 **Total 341**	Yes 68 No 32 **100%**	1 0 **1**
Will you support the establishment of regulations and laws prohibiting marriage in case of a positive premarital screening results	Yes 198 No 143 **Total 341**	Yes 42 No 58 **100%**	0 1 **1**

Findings in table 4 indicate that (69%) are of the opinion of terminate their relationship with their partner if the genotype result shows (incompatibility) possibility of having a child with SCD. However, 92% claimed that PMS has nothing to do with their religious belief. (68%) and (58%) of the respondents support mandatory PMS for all intending couples in Nigeria and the establishment of regulations and laws prohibiting marriage in cases of incompatible PMS results, respectively.

4. Hypothesis

Relationship between socio-demographic characteristic and knowledge regarding premarital genotype screening on sickle cell diseases.

Table 5 Age * Knowledge cross tabulation

Count				
		Knowledge		Total
		Good	Poor	
Age	18 – 25	161	140	301
	26 – 33	37	3	40
	34 – 41	0	0	0
	42 – Above	0	0	0
Total		198	143	341

Calculated chi-square p-value is 0.000, which is less than the common alpha level of 0.05, therefore reject the null hypothesis and conclude that there is a significant relationship between age and knowledge of SCD. Age influences a person's knowledge about sickle cell disease and premarital genotype screening.

Table 6 Gender * Knowledge cross tabulation

Count				
		Knowledge		Total
		Good	Poor	
Gender	Male	34	68	102
	Female	101	138	239
Total		135	206	341

Given that p-value of the Chi-Square Tests is 0.123, which is greater than the common alpha level of 0.05, the null hypothesis is thereby accepted. This means there is no significant relationship between gender and knowledge Therefore, being male or female does not significantly influence a person's knowledge about

sickle cell disease and premarital genotype screening.

Table 7 Academic level knowledge cross tabulation

Count				
		Knowledge		Total
		Good	Poor	
Academic Level	100L	15	29	44
	200L	35	40	75
	300L	73	30	103
	400L	56	35	91
	500L	21	7	28
Total		200	141	341

Pearson Chi-Square is 0.000 which is the p-value indicates that there is very strong evidence against the null hypothesis. Given that the p-value is less than the common alpha level of 0.05, therefore reject the null hypothesis. This means there is a significant relationship between academic level and knowledge. This result suggests that the academic level of a student influences their knowledge about sickle cell diseases and premarital genotype screening.

5. Discussion of findings

This cross-sectional study assessed respondents' knowledge, attitudes, and perceptions regarding premarital genotype screening (PMS) for sickle cell disease (SCD). The socio-demographic characteristics showed that most respondents were aged 18–25 years and were single. This finding is not unexpected because the study population comprised undergraduate students, who are within the age group most likely to make future marital decisions. The predominance of unmarried respondents provides an appropriate context for assessing knowledge and perceptions of premarital genotype screening before marriage. The study found that a majority of respondents correctly identified sickle cell disease as an inherited blood disorder and recognized that it is transmitted genetically from parents to their offspring. However, a considerable proportion of respondents still held incorrect beliefs regarding the cause and mode of transmission of SCD, suggesting persistent knowledge gaps that may influence informed reproductive decision-making. These findings are consistent with those reported by Adigwe (2022), who observed relatively good knowledge of the hereditary nature of SCD among Nigerian youths but also identified misconceptions regarding the disease. The findings underscore the need for sustained public health education on the genetic basis of sickle cell disease.

More than half of the respondents correctly identified the SS genotype as sickle cell anaemia and reported being aware of their own haemoglobin genotype. This level of knowledge is comparable to the findings of Hussaini et al. (2019) among undergraduate students in Kano State, who reported relatively high awareness of genotype composition. In contrast, Isah et al. (2016) reported lower levels of genotype knowledge among nursing students in Sokoto. Similarly, the proportion of respondents in the present study who knew their genotype was higher than that reported among adolescents in Benin City and the Federal Capital Territory by Faremi and Olatubi (2018). These differences may reflect variations in educational exposure, health promotion activities, and access to genotype screening services across study settings. The findings further indicate that respondents generally demonstrated a positive understanding of premarital genotype screening. Most participants recognized PMS as an important preventive strategy for reducing the risk of sickle cell disease among offspring and were aware that screening services are available in Nigeria. This finding agrees with Boardman et al. (2020), who reported positive perceptions of premarital screening among young adults. Nevertheless, a small proportion of respondents lacked adequate knowledge of the components and purpose of premarital genotype screening, suggesting that misconceptions still exist despite the overall satisfactory level of awareness.

The study also demonstrated generally favourable perceptions towards premarital genotype screening. Most respondents indicated that they would not proceed with marriage if genotype results revealed incompatibility associated with a high risk of having children with sickle cell disease. Similar findings have been reported by Al-Aama (2010) and Al-Kindi et al., (2019), who found high acceptance of premarital genotype screening and increased willingness to reconsider marriage decisions based on genotype compatibility. Furthermore,

the majority of respondents reported that premarital genotype screening does not conflict with their religious beliefs, suggesting that religious barriers to screening may be less pronounced within the study population.

A substantial proportion of respondents supported making premarital genotype screening a prerequisite for marriage and also supported legislation aimed at reducing marriages between genetically incompatible couples. This level of support was higher than that reported by Al-Kindi et al., (2019), indicating increasing acceptance of preventive strategies against sickle cell disease. However, despite this favourable perception, previous studies have shown that social stigma, emotional attachment, cultural expectations, and concerns about interference with personal choice remain important barriers to premarital genotype screening and adherence to genotype compatibility recommendations (Boardman et al., 2020).

The inferential analysis revealed statistically significant associations between selected socio-demographic characteristics and respondents' knowledge of premarital genotype screening and sickle cell disease. Specifically, age and academic level were significantly associated with knowledge scores, suggesting that older students and those at higher academic levels were more likely to possess better knowledge. Similar associations have been reported in previous studies, where higher educational attainment was associated with improved awareness and understanding of sickle cell disease prevention (Emmanuel, 2015). However, because this study employed a cross-sectional design, the observed associations should not be interpreted as evidence of causal relationships. Rather, they indicate that these characteristics are associated with differences in knowledge within the study population. Overall, the findings suggest that although respondents demonstrated generally good knowledge and favourable perceptions of premarital genotype screening, important misconceptions remain regarding the genetic basis of sickle cell disease and aspects of premarital screening. These findings highlight the need for continuous health education, strengthened genotype counselling services, and sustained awareness campaigns within tertiary institutions to promote informed reproductive health decisions and reduce the burden of sickle cell disease in Nigeria.

6. Conclusions

One of the key conclusions drawn from the study is that the respondents have relatively robust knowledge about the genetic composition. However, the study revealed respondents support for mandatory PMS to all intending couples in Nigeria, and also express their support towards the establishment of regulations and laws in Nigeria prohibiting marriage in case PMS results incompatibility. Correspondingly, the respondents' willingness to advocate and recommend PMS to young unmarried. However, the majority of the respondents don't know and have never heard about premarital genetic counselling. This indicates the need for enlightenment and promoting informed decision-making. This finding underscores the statistically significant relationship between other socio-demographic characteristics at the 5% significant level.

7. Recommendation

For education and prospective carrier identification, religious institutions and NGOs should prioritize premarital counselling on genetic diseases in their various domains, with minimal emotional pressure at minimal or no cost to be feasible. Public enlightenment and health education on sickle cell disease and premarital genotype through mass media programs, and screening centres across primary health should be made available, affordable and accessible to all by government.

List of abbreviations

Hb: Hemoglobin gene
HbS: Sickle Hemoglobin
HbSS: homozygous Sickle cell anemia Hemoglobin
HbSC: compound heterozygous Sickle cell Hemoglobin
PMS: Premarital genotype Screening
RBCs: Red blood cells
SCA: Sickle cell anemia
SCD: Sickle cell disease

Declaration

Ethics approval and consent to participate

Ethical approval was obtained from ethical review committee of Al-Hikmah University through the Department of Public Health and Postgraduate School of the University respectively. Prior to participating in

the study, all potential participants were provided with a clear and comprehensive explanation of the research purpose, procedures and benefits. Informed consent form was given. Participants was assured that their participation is voluntary, and they have the right to withdraw at any stage without facing any consequences.

Consent for publication

Not applicable

Availability of data and material

The datasets used or analyzed during this study are generated from primary source and available with the corresponding author on reasonable request, included in this manuscript submitted to Bulletin of the National research center and its supplementary information file.

Competing interests

The author declares that no competing interests

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Authors' contributions

The corresponding author confirms sole responsibility for the study, include conception and design, data collection, analysis and interpretation of result, and manuscript preparation,

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