Pre-marital screening for sickle cell haemoglobin and genetic counseling: awareness and acceptability among undergraduate students of a Nigerian University

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Background: Sickle cell disease (SCD) is a genetic disease which is more prevalent in developing countries. Pre-marital screening for sickle cell disorder is helpful in the prevention and control of the condition. Aim: The aim of this study was to assess the level of awareness and acceptability of pre-marital genetic counseling and screening for sickle cell haemoglobin among undergraduate students of Ebonyi State University Abakaliki, South eastern, Nigeria. Methods: A cross-sectional descriptive study was carried out, and participants were selected using multistage sampling technique. Data was collected using a pre-tested, self-administered questionnaire and analyzed using SPSS software, version 20. Results: A total of 329 participants were studied; 158 (48%) were males and 171 (52%) were females, with mean age of 22.3± 2.7 years. Three hundred (91.2%) participants were aware of pre-marital genetic counseling and screening for sickle cell haemoglobin and 319 (97%) accepted to go for pre-marital genetic counseling and screening for sickle cell haemoglobin while 316 (96%) accepted that it is important for people to go for pre-marital genetic counseling and be screened for sickle cell haemoglobin. Conclusion: This study found high level of awareness and acceptance of pre-marital genetic counseling and screening for sickle cell haemoglobin among the students. However, more need to be done to increase public awareness. The present study will emphasizes the need to institute formal pre-marital counseling and screening for sickle haemoglobin among the youth to help them take informed decision about their marriage to prevent procreation of children affected by sickle cell disease.

Key words: Sickle cell disease, sickle cell traits, premarital screening, genetic counseling, haemoglobin, Nigeria

INTRODUCTION

Sickle cell anemia is a genetic disorder in which an abnormal haemoglobin leads to chronic haemolytic anaemia with numerous clinical consequences. The sickle haemoglobin gene was reported to develop in response to severe malaria endemcity and is protective against malaria as heterozygous individuals with sickle cell gene (AS individual) have been shown to have a decreased risk of severe malaria. On the other hand, homozygous individuals (SS individuals) are at increased risk of severe malaria attacks.

Sickle cell disease is a public health problem and afflicts over 100 million people worldwide, predominantly in black people (in Africa,
Europe and the Americans), Arabs and those of Asian ancestry.\[3\] It is estimated that sickle cell anaemia contributes to 5% 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 African countries, including Nigeria.\[4\] About 24% of Nigerian have sickle cell gene and 2% of all newborns in Nigeria are homozygous for sickle cell gene, and are therefore likely to suffer from sickle cell disease.\[5-7\]

Premarital genotype screening creates an opportunity for people to take informed decision on the genetic predisposition of their unborn children. It is therefore important that people are screened and informed of their genetic make-up in order to help them take informed decision about their marriage so as to avoid procreation of children with genetic inheritance of grave consequences. Genetic diseases can impact greatly on a person’s life, causing physical and emotional pain, developmental problems and even death.\[8\]

Different methods of preventing genetic diseases include genetic counseling and premarital screening, preconception diagnosis and implantation of normal embryos after in vitro fertilization, prenatal diagnosis and abortion of the affected fetus and in-utero therapy using stem cell transplantation.\[8\] However, the only realistic approach to prevent and reduce the impact of the disease is through genetic counseling and carrier identification.

Nigeria has the highest burden of sickle cell disease in the world with about 150,000 children born annually with sickle cell disease.\[5\] Considering the burden of genetic diseases especially sickle cell disease in Nigeria, there is need to evaluate the level of awareness and acceptability of premarital genotype screening and genetic counseling among the populace. The youths are particularly the right target population for interventions aimed at preventing and controlling genetic diseases such as premarital genetic counseling and screening because most of them are either unmarried or preparing to get married and will procreate in the future. Several studies have given different reports on the level of awareness and acceptability of premarital genetic screening among the youth,\[9-12\] but there is paucity of such studies in our locality hence the objective of this study is to determine the level of awareness and acceptability of pre-marital screening for sickle cell haemoglobin and genetic counseling among undergraduate students of Ebonyi State University, Abakaliki, South eastern, Nigeria.

**METHODOLOGY**

**Study design**
Descriptive cross-sectional study was carried out between September and November 2015 at Ebonyi State University, which is located in Abakaliki, Ebonyi State, South eastern Nigeria.

**Sample size**
The sample size for this study was determined using the formula\[13\] for estimation of population prevalence and was based on a 95% confidence level, and a prevalence of awareness about premarital screening for sickle cell gene among students, which is 78.9%, as reported by a previous study,\[14\] and a desirable degree of accuracy set at 0.05 level. Minimum sample size of 293 was calculated. However, 350 questionnaires were given out to account for refusals. Out of this number, 329 were correctly and completely filled. These questionnaires were considered valid and were used for data analysis.

**Sampling method**
Multistage sampling technique was used for the study. Simple random sampling by ballot method was used to select four out of the ten faculties existing in the school. Simple random sampling technique by ballot method was also employed to select four departments, one department each out of the four chosen faculties, as well as to select students from the chosen departments.

**Study instrument and data collection**
The tool for data collection was a pre-tested, semi-structured self-administered questionnaire. Information sought in the questionnaire included socio-demographic characteristics, awareness and acceptance of genetic counseling and premarital genetic screening for SCD. During data collection, three research assistants were employed to administer the questionnaires. They were adequately trained and mobilized for the exercise and they assisted in the administration and retrieval of the questionnaires.
Ethical issues
Approval for this study was gotten from Research and Ethics Committee of the institution. Questionnaires were administered only to students who gave their consent.

Statistical analysis
The questionnaire was used to elicit necessary information from the participants. Data was cleaned for inconsistencies in the responses and was entered into a computer using statistical package for social sciences (SPSS) software, version 20, which was also used for the analysis. Descriptive statistics were used to compute percentages and averages. Chi square test was used to assess the relationship between variables. Results were presented in tables and charts, and expressed as percentages/proportions, means and standard deviation.

RESULTS
A total of 329 students participated in the study and were made up of 158 (48%) males and 171 (52%) females, with male to female ratio of 1:1.1. The ages of the participants ranged between 16 and 36years with mean age of 22.3±2.7years. Most of the participants, 323 (98.2%) were christians and 305 (92.7%) were single (table 1).

Three hundred participants (91.2%) were aware of premarital genotype screening and genetic counseling (figure 1). Lectures were the most frequent source of information, 115 (35%). Other sources of information include health workers 63 (19.1%), friends and colleagues 55 (16.7%), family members 37 (11.2%), radio and television 34 (10.3%), library 11 (3.3%), internet 5 (1.5%) and posters 1 (0.3%).

One hundred and ninety one (58.1%) of the participants had adequate knowledge about genetic counseling and premarital genotype screening. Two hundred and seventy (82.1%) of the participants have heard about haemoglobin genotype and all (100%) the participants have done their haemoglobin genotype test. Table 2 showed the result of haemoglobin genotype test result of the participants.

Table 3 showed the responses of participants to questions on acceptance of premarital genotype screening and genetic counseling. Three hundred and nineteen (97%) participants accepted to go for premarital genetic counseling and 316 (96%) accepted that it is important for people to go for premarital genetic counseling and be screened for haemoglobin genotype. Three hundred and twenty (97.1%) accepted that it is good to know partner’s genotype before marriage while 246 (74.8%) did not accept to continue with the marriage if there is risk of sickle cell disease in the offspring. Other information is as shown in table 3.

<table>
<thead>
<tr>
<th>Characteristics</th>
<th>Frequency</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sex</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Male</td>
<td>158</td>
<td>48</td>
</tr>
<tr>
<td>Female</td>
<td>171</td>
<td>52</td>
</tr>
<tr>
<td>Total</td>
<td>329</td>
<td>100</td>
</tr>
<tr>
<td>Age</td>
<td></td>
<td></td>
</tr>
<tr>
<td>&lt;20</td>
<td>31</td>
<td>9.4</td>
</tr>
<tr>
<td>20 – 25</td>
<td>266</td>
<td>80.9</td>
</tr>
<tr>
<td>&gt;25</td>
<td>32</td>
<td>9.7</td>
</tr>
<tr>
<td>Total</td>
<td>329</td>
<td>100</td>
</tr>
<tr>
<td>Religion</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Christianity</td>
<td>323</td>
<td>98.2</td>
</tr>
<tr>
<td>Islam</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>Traditional</td>
<td>1</td>
<td>0.3</td>
</tr>
<tr>
<td>Others</td>
<td>5</td>
<td>1.5</td>
</tr>
<tr>
<td>Total</td>
<td>329</td>
<td>100</td>
</tr>
<tr>
<td>Marital status</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Married</td>
<td>22</td>
<td>6.7</td>
</tr>
<tr>
<td>Single</td>
<td>305</td>
<td>92.7</td>
</tr>
<tr>
<td>Divorced</td>
<td>2</td>
<td>0.6</td>
</tr>
<tr>
<td>Total</td>
<td>329</td>
<td>100</td>
</tr>
</tbody>
</table>
Ugwu: Pre-marital screening for sickle cell haemoglobin and genetic counseling

**Figure 1:** Awareness about premarital genotype screening among the participants

**Table 2:** Participants’ haemoglobin genotype

<table>
<thead>
<tr>
<th>Haemoglobin Genotype</th>
<th>Frequency</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>AA</td>
<td>208</td>
<td>63.2</td>
</tr>
<tr>
<td>AS</td>
<td>98</td>
<td>29.8</td>
</tr>
<tr>
<td>Not sure</td>
<td>23</td>
<td>7.0</td>
</tr>
<tr>
<td>Total</td>
<td>329</td>
<td>100</td>
</tr>
</tbody>
</table>

**Table 3:** Responses to questions on acceptance of pre-marital genotype screening and genetic counseling

<table>
<thead>
<tr>
<th>Variable</th>
<th>Acceptance</th>
<th>Response</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>N (%)</td>
<td>N (%)</td>
</tr>
<tr>
<td>I agree to go for premarital genotype screening and genetic counseling</td>
<td>319 (97)</td>
<td>6 (1.8)</td>
</tr>
<tr>
<td>People should go for premarital genetic counseling</td>
<td>323 (98.2)</td>
<td>1 (0.3)</td>
</tr>
<tr>
<td>It is important for people to be screened for haemoglobin genotype</td>
<td>316 (96.0)</td>
<td>5 (1.5)</td>
</tr>
<tr>
<td>It is good to know partner’s genotype before marriage</td>
<td>320 (97.1)</td>
<td>6 (1.8)</td>
</tr>
<tr>
<td>Partner’s genotype should influence decision for getting married</td>
<td>313 (95.1)</td>
<td>7 (2.1)</td>
</tr>
<tr>
<td>One can continue with marriage even if there is risk of SCD in the offspring</td>
<td>23 (7.0)</td>
<td>246 (74.8)</td>
</tr>
</tbody>
</table>
Table 4: Relationship between acceptance and knowledge about premarital genotype screening and genetic counseling

<table>
<thead>
<tr>
<th>Variables</th>
<th>Knowledge genotype of premarital screening</th>
<th>Total</th>
<th>$\chi^2$ ($P$-value)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Adequate</td>
<td>Inadequate</td>
<td></td>
</tr>
<tr>
<td>Acceptance of premarital genotype screening and genetic counseling</td>
<td>178 (60.8)</td>
<td>115 (39.2)</td>
<td>293</td>
</tr>
<tr>
<td>Not accepted</td>
<td>13 (36.1)</td>
<td>23 (60.8)</td>
<td>36</td>
</tr>
</tbody>
</table>

This study also showed that a higher percentage of students from the Faculty of Medicine, 74.9%, had better knowledge about premarital genotype screening and genetic counseling compared to students from other faculties: Faculty of Health Science and Technology (57.9), Faculty of Agriculture (42.0%) and faculty of law (33.9%). Association between knowledge and faculty was statistically significant ($\chi^2 = 48.5$, $P = 0.0001$).

This study also showed statistically significant relationship between acceptance and knowledge about premarital genotype screening and genetic counseling ($\chi^2 = 8.0$; $P = 0.005$) (table 4).

DISCUSSION

This study found that the ages of the participants ranged between 16 and 36 years with equal sex distributions. The participants were all young people most of whom were unmarried making them ideal for the study on awareness and acceptability of premarital genotype screening and genetic counseling, as also implicated by previous studies.\[15,16\] Awareness and acceptability of premarital genotype screening and genetic counseling will help them to understand their genetic make-up and the possibility of transmitting genetic disease to their unborn children. This will thus help them take informed decision about their marriage so as to avoid procreation of children affected with genetic disease such as sickle cell disease.

This study also found high level of awareness of premarital genotype screening and genetic counseling among the participants. This is similar to the report of Omuemo et al.\[16\] who also reported high level of awareness and acceptability of premarital genetic screening among their study population. High level of awareness among the participants may be because the students are usually mandated to screen for their haemoglobin genotype as part of medical screening on admission into the University. Major source of information about premarital genetic screening include Lectures, health workers, friends and colleagues. This corroborates with the findings of the study conducted in Jos, Nigeria,\[17\] which also reported health workers, family members and friends as the major source of information. These findings showed that the media has to do more in terms of creating awareness about sickle cell disease.

Though most of the participants were aware of premarital genotype screening and genetic counseling, some of them lack adequate comprehensive knowledge about it. Some of the participants did not know the importance and usefulness of premarital genotype screening and counseling and did not border to know their genotype off-head. Part of the reason being little or no importance attached to the result of the genotype test as some participants merely did the test as part of mandatory medical screening tests required for admission into the school. This is similar to the findings of the study conducted by Sani et al.\[18\] in Zaria, Nigeria who reported that many of their participants did not know their genotype and there were no obstacles preventing them from genetic counseling and testing. Among the participant who screened and know their genotype, all of them claimed not to have sickle cell disease. This may be because of fear of stigmatization and discrimination exhibited against people living with sickle cell disease as reported by previous studies.\[19,20\]

This study also found that students from faculty of medicine had significantly better
knowledge about premarital genotype screening and genetic counseling compared to students from other faculties. This is not surprising considering the fact that medical students are better exposed to enlightenment on issues concerning the prevention and control of sickle cell disease and other health issues compared to students from other faculties. This buttresses the fact that proper education about sickle cell disease including method of prevention and control such as premarital genotype screening and genetic counseling will help people to take informed decisions and appropriate actions to control sickle cell disease. Similar study conducted among health care workers and medical students showed better knowledge about issues concerning sickle cell disease since they interact more with clients suffering from the disease.\[21\]

Majority of the participants exhibited high level of acceptability for premarital genotype screening and genetic counseling. This corroborates with findings of previous studies,\[22\] which reported acceptance of premarital genotype screening among the participants following counseling. The partners of the participants who were not subjected to genetic counseling were reluctant to go for genotype screening. This still buttresses the fact that education and enlightenment enhances peoples acceptability for premarital genotype screening and genetic counseling, a realistic method of prevention and control of sickle cell disease.

**CONCLUSION**

This study showed high level of awareness of premarital genotype screening and genetic counseling among undergraduate students of Ebonyi State University. Majority of the participants accepted to go for pre-marital genetic counseling and screening for sickle haemoglobin. Majority of the participants also accepted that it is important for people to go for pre-marital genetic counseling and screening for haemoglobin genotype. However, some of them exhibited non acceptability due to ignorance about the methods of prevention and control of sickle cell disease, Therefore education and awareness on the genetics of sickle cell disease is paramount to the acceptability of premarital genotype screening. This will equip people to take informed decisions and actions about their marriage, to prevent procreation of children affected with sickle cell disease.

**LIMITATIONS OF THE STUDY**

The study population was made up university undergraduates who are usually screened for their haemoglobin genotype as part of mandatory medical screening on admission into the University. This may have contributed to the high level of awareness and acceptability of pre-marital genetic counseling and screening for sickle cell haemoglobin among them. Applying the same findings to the general population should therefore be done with caution.

**ACKNOWLEDGEMENT**

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**CONFLICT OF INTEREST**

The author declares no conflict of interest.

**REFERENCES**


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Conflict of Interest: None declared
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