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Introduction
As clinical genetics has continued to play important roles in the field of medicine today, prenatal genetic testing has been used as a successful tool in combatting chromosomal abnormalities in countries where it is available and accessible.[1] Prenatal diagnosis in general employs a range of diagnostic technologies to gain information about embryo or foetal wellbeing,[2] including the detection of susceptibility to and presence of genetic diseases.[1]

It is reported that genetic disorders and congenital abnormalities occur in 2%-5% of all live births.[4] Sickle cell disease is one of the commonest genetic disorders in Nigeria with carriers of the mutant gene accounting for about 24% of the population and about 150,000 affected children born annually with this disease in Nigeria alone.[5] Some other genetic diseases found in Nigeria include Down syndrome and Turner’s syndrome. Research shows that Down syndrome in Nigeria has an incidence of 1 in 865 live births with a high incidence of cases among young mothers.[6] It is possible that the low prevalence of genetic diseases maybe the result of underreporting as well as the poor...
standard of healthcare facilities which means that many children born with genetic diseases are less likely to survive.\[1\]

Prenatal genetic testing is divided into screening and diagnostic testing. A screening test determines whether a pregnancy is at increased risk for a specific disorder such as Down syndrome. Screening tests include first trimester ultrasound scan and blood tests before 3 months and second trimester maternal serum screening. Diagnostic testing involves first trimester chorionic villous sampling (CVS), mid-trimester amniocentesis, and biopsy of the foetal skin or percutaneous umbilical blood sampling (PUBS).\[7\]

Developing countries like Nigeria have been lagging behind in their implementation of public policies for the care and prevention of genetic diseases.\[3\] Prenatal diagnosis and treatment in Nigeria is still not routinely offered to pregnant women in most antenatal clinics.\[8\] Studies have shown that the increasing incidence of genetic diseases and mortality rates attributed to genetic diseases in Nigeria and Africa is still as a result of ignorance and negligence of parents.\[9\] In addition, a World Health Organization report lists poor education, low literacy rates, little or no knowledge about genetics and misconceptions at various levels among the barriers to the availability of genetic services in developing countries.\[1\] Ideally, pregnant women should be aware that they could find out the genetic status of their children in order to be well aware of possible genetic or congenital defects as they are the ones who are the most affected by decisions concerning prenatal diagnosis and they will be the most affected in making decisions concerning the quality of life of their unborn children as it would be their obligation to nurse these children if they turn out to have a genetic disease or congenital anomaly.\[8\]

This study set out to assess the knowledge of pregnant women about genetic diseases and prenatal genetic testing, willingness to test, attitudes towards testing, use of common tests available as well as willingness to terminate affected pregnancies. To the best of our knowledge, no published study has examined this in Lagos, Nigeria.

**Materials and Methods**

**Study background**

Lagos State is located in the South Western part of Nigeria. The study was carried out in Amuwo-Odofin a Local Government Area in the Badagry Division of Lagos State. It includes two local council development areas (LCDA): Amuwo-Odofin LCDA and Oriade LCDA.

**Study design**

This was a descriptive cross-sectional study among 327 pregnant women who attended antenatal clinics in a secondary health facility (Amuwo-Odofin General hospital) and the antenatal clinics in two primary health care centres (Festac and Agboju primary healthcare centres) all within Amuwo-Odofin LGA.

**Study participants**

The study participants were women in any of the three trimesters of pregnancy, registered and found at the facility for antenatal care and who gave written informed consent. They were recruited consecutively as they arrived in the clinic. Based on the proportion of weekly antenatal clinic attendees, 213 respondents were recruited from the Amuwo-Odofin General Hospital, 89 respondents were recruited from the Agboju primary healthcare centre, and 53 respondents were recruited from Festac primary healthcare centre. Data was collected using structured interviewer-administered questionnaires containing both closed and open ended questions which took approximately 15 minutes to complete.

**Inclusion and exclusion criteria**

All pregnant women in any of the three trimesters of pregnancy, registered and found at the facility for antenatal care and who gave written informed consent were included in this study. Pregnant women who were not previously registered and found at the healthcare facilities for antenatal clinic services were excluded from the study.

**Study tools**

The questionnaire was adapted from a similar study done in Western Australia\[10\] and from a review of available literature.\[2,9\] Information was obtained on their sociodemographic characteristics in terms of age at last birthday, educational status, religion, marital status, occupation, spouses’ occupation and household monthly income. Characteristics of pregnancies which were elicited were respondents’ gravidity and number of previous pregnancies with a genetic condition. The number of respondents’ relatives with a genetic disease was also elicited.

The following were the items which assessed respondents’ knowledge of genetic diseases: Genes come from both parents, causes of genetic diseases, ability to affect a developing foetus, inheritability and transmission of sickle cell disease Down syndrome as a genetic disease and risk of having an affected child as well as respondents’ major source of knowledge about genetic diseases.

Respondents’ knowledge of prenatal genetic testing including first trimester ultrasound and blood tests, second trimester maternal serum screening, amniocentesis and chorionic villous sampling were assessed using the following items: possibility of prenatal diagnosis of genetic diseases, ability of an ultrasound to detect every kind of birth defect, what constitutes first trimester screening, possibility of additional tests if screening determines there is increased risk.

Knowledge was categorized as ‘good’ or ‘poor’ based on whether they scored above or below 70% of correct responses to the items of knowledge asked.

Willingness to test was assessed with a single item on the questionnaire with respondents indicating ‘Yes’, ‘No’ or ‘Don’t know’ in response to the question, ‘Are you willing to undertake...
Attitudes to testing were assessed with eight items which evaluated the respondent’s perception of the need for, use and personal disposition towards prenatal genetic tests. Respondents were also asked if they would proceed to undergo diagnostic testing if screening tests revealed that pregnancies were at increased risk. The respondents were also required to indicate ‘Yes’, ‘No’, ‘I don’t know’. ‘Good’ attitude was determined by the respondent giving 70% or more of positive responses.

Uptake of the various screening and diagnostic options were assessed using six items to determine whether the respondent had used any of the prenatal genetic tests in the past. Also, items were provided for possible reasons why the respondent had or had not made use of testing as the case may be.

An item was finally included asking if the respondent would opt to terminate the pregnancy if diagnostic tests came out positive for a genetic disease

**Data analysis**

Data was obtained and analysed electronically using Epi-Info 7.2 statistical software. Data was presented using frequency distribution tables. Independent variables were presented as frequencies and percentages with means and standard deviations where applicable Chi-square and Fishers exact were then used to test whether there was any significant association between the outcome and independent variables. Level of significance was set at 0.05.

**Ethical approval**

Approval for this study was obtained from the health research and ethics committee of the Lagos University Teaching Hospital. The participants were informed about the significance of the study and how honest and fair answers were important when answering the questionnaires. Consent was sought before administration of the questionnaires. No names were printed on the questionnaires and the respondents were assured of the confidential nature of the study. They were also given the choice to participate or not in the study.

**Results**

Respondents were mostly in the age range of 29-38 years (56.9%). The mean age was 30.0 ± 4.2. Respondents were predominantly Christians (91.7%), were married (99.4%) and 41.9% had post-secondary education. More than half (52.6%) were unemployed and majority (41.9%) earned between N18,001-N50,000 naira (50-139 dollars) monthly [Table 1] Majority (43.7%) of the respondents were experiencing their first pregnancy and majority (94.8%) reported that they had no previous pregnancies with a genetic condition and no relatives with a genetic disease (97.3%) [Table 1].

More than half (69.4%) of the respondents had poor knowledge of genetic diseases, that is, scored less than 70% on the knowledge questions. Mean total knowledge score was 4.45 ± 2.79 standard deviation. Majority of the respondents (75.5%) knew that marriage of two genotype AS partners could lead to a child with sickle cell disease, that sickle cell disease was inherited (50.2%) and that genetic diseases could affect a baby developing in the womb (57.5%), however more than half (55.1%) did not know that Down syndrome was a genetic disease and could not answer most of the other knowledge questions. Just about half (49.5%) of the respondents stated their major source of knowledge to be radio, television, books, magazines and newspapers as seen in Table 2.

Although 50.8% of the respondents stated that they had heard about one prenatal genetic test or another, with majority of the respondents (66.5%) stating that they had heard about first trimester ultrasound scan and blood tests before 3 months of pregnancy, almost all the respondents (97.6%) had poor knowledge of prenatal genetic testing. Mean total knowledge score was 2.12 ± 2.12. Majority of the respondents did not know the answers to the knowledge questions although half of the respondents (50.2%) answered correctly that it was possible to find out if their unborn children had a genetic disease. Majority of the respondents (48.9%) stated their major source of knowledge of prenatal genetic testing to be the health personnel as seen in Table 3.

There was no significant association between the sociodemographic variables and knowledge of prenatal genetic testing as seen in Table 4. Knowledge of prenatal genetic testing was not also significantly associated with willingness to test (p=0.16).
When assessed for willingness to undergo prenatal testing, majority (61.8%) of the respondents stated that they would opt for a prenatal genetic test if made available. Respondents’ level of education was significantly associated with willingness to test (p<0.001) as seen in Table 4. 49.5%, 29.2%, 12.4%, 3.9% of women who were willing to test had post-secondary, secondary school, vocational education and no formal education respectively.

Only 23.9% of the respondents had good attitude scores. Knowledge of genetic diseases (p=0.02), respondents’ employment status (p=0.03) and previous use of tests (p<0.01) significantly correlated with attitudes to testing. Majority (63.6%) thought the screening tests were valuable and that the information gained from the tests would help them plan for the future (62.9%). However, only less than half (48%) of the respondents thought that the cost of the screening tests should not influence whether they are done or not. Only 22.7% expressed confidence in the accuracy of test results. More than half of the respondents (58.7%) stated that they would worry about how such tests would affect the health of their baby. Also, only 39.4% indicated that they would opt for a diagnostic test if screening test came out positive. There was a significant association between respondents’ willingness to proceed with diagnostic tests and attitude scores. (p<0.001)

<table>
<thead>
<tr>
<th>Table 2: Knowledge of genetic diseases.</th>
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<tbody>
<tr>
<td>Knowledge item</td>
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<tr>
<td>If genes come in pairs, one copy from each parent</td>
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<tr>
<td>If genetic diseases are caused by inheriting two abnormal genes from the parent</td>
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<tr>
<td>If genetic diseases are caused by an infection</td>
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<tr>
<td>If genetic diseases can affect a baby developing in the womb</td>
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<tr>
<td>If one family member has a genetic disease, will all family members develop the disease</td>
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<tr>
<td>If Sickle cell disease can be inherited</td>
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<tr>
<td>If marriage of two AS partners could lead to a child with sickle cell disease.</td>
</tr>
<tr>
<td>If Down syndrome is a genetic disease.</td>
</tr>
<tr>
<td>If no family member has Down syndrome, there is no risk in my children.</td>
</tr>
<tr>
<td>If it is possible to have a gene for a genetic disease but no symptoms</td>
</tr>
</tbody>
</table>

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In the study sample, only 27.2% stated that they had made use of a prenatal genetic screening or diagnostic test. Knowledge of genetic diseases was significantly associated with use of tests (p<0.018). Majority (26.9%) of the participants who had made use of a prenatal screening or diagnostic test had made use of ultrasound and blood test before three months of pregnancy [Table 5]. When asked for possible reasons why they made use of a prenatal screening or diagnostic test majority (19.6%) stated that they wanted to see the baby, others (18%) stated that they wanted to make sure the baby was healthy and 11.3% stated that it was just a routine thing that is done when they are pregnant. When asked for possible reasons why they had not made use of a prenatal genetic test, majority (35.5%) of the respondents who had not made use of a prenatal genetic test stated that they didn’t know about them, 10.7% of them stated that they were not necessary and 5.5% stated that they were not aged 35-37 years or older [Table 6].

Only 10.1% of the population stated that they would opt to terminate their pregnancy if a prenatal diagnostic test came out positive for a genetic disease such as Down syndrome. Majority of the respondents (73.4%) were undecided about this and 16.5% stated that they would not opt to terminate the pregnancy. Knowledge of genetic diseases significantly correlated with decision to terminate affected pregnancies (p=0.02).

**Discussion**

The modal age group of participants in this study was 29 to
38 years which is older than the modal age group of 25 to 34 years observed in a similar study done in Ibadan, another city in southwest Nigeria. The observation is that this group of respondents fall into an age group at higher risk of genetic disease particularly down syndrome among their children. The knowledge of genetic diseases found in this study was poor; however, women seemed to know more about sickle cell disease than about diseases such as Down syndrome. This can be attributed to the greater prevalence of sickle cell disease in this environment. It also underscores the effectiveness of previous and on-going efforts at educating the masses about sickle cell disease and its risk factors.

Our findings from this study showed that pregnant women in the study population had poor knowledge of prenatal genetic testing. Majority of the respondents who had heard about prenatal genetic testing (48.9%) stated their major source of knowledge to be Health personnel while more than a quarter (28.2%) stated their major source of knowledge to be Radios, Television, books, magazines and newspapers. This was similar to the study done among western Australian women where Twenty-nine per cent (29%) stated their source of information to be pamphlets while 60% first found out about the tests through their General practitioners or specialist.

Although about half of the respondents (50.2%) knew that prenatal testing was possible, respondents had deficiencies in the nature of these tests, the modalities involved and the associated risks, especially of miscarriage associated with invasive tests like amniocentesis and chorionic villous sampling. This information is necessary if pregnant women are to make informed decisions concerning prenatal testing, are important in eliciting informed consent and should be incorporated in pre-test counselling for prenatal testing.

The knowledge evaluation of the population was 21% which was lower than the overall knowledge evaluation of women in the study done in Ibadan (55%), much lower than the knowledge evaluation of the participants in the study done in Western Australia (62%), and also in Denmark (82%). The fact that the study in Ibadan was carried out in a tertiary hospital as opposed to a primary and/or secondary hospital where this study was carried out may account for the difference in knowledge scores. Tertiary health facility attendees have increased access to specialist care and improved medical technology available in these facilities than at the primary healthcare levels. This would underscore the paucity of information and services of prenatal genetic testing that is available to women who do not access care at tertiary centres. Results of our study however, was in line with a study done in Pakistan where only 26% of women had any knowledge about Down syndrome or its screening.

In our study, none of the independent variables were statistically significantly associated with knowledge of prenatal genetic testing and knowledge of testing was not significantly associated with willingness to test. The studies in Ibadan and Western Australia both found associations with marital status, level of education and age. However, there was no difference in the mean knowledge score of women who accepted prenatal diagnosis and those who declined in Ibadan showing that knowledge was a weak determinant of willingness to test. Another study carried out in Sweden found that knowledge of Down syndrome and its consequences was not a major factor which determined willingness to undergo testing. Also, the study done in Denmark showed that there were no significant differences found in knowledge of Down syndrome between women declining or accepting prenatal diagnosis for Down syndrome. Both groups had varying and in several respects, low knowledge about Down syndrome and its consequences. It was therefore concluded that knowledge of Down syndrome at these levels is not a major factor when women decide to accept or decline prenatal diagnosis for Down syndrome.

Furthermore, 61.8% of the respondents in our study were willing to opt for prenatal genetic testing. Willingness to test was significantly associated with respondents’ level of education. Majority of women who were willing to undergo testing had post-secondary education. This was similar to that reported in Ibadan with overall acceptance at 68% with educational attainment being the only predictor.

Though majority of the respondents felt that tests were valuable, general attitude scores were negatively influenced by concerns about cost of testing as only less than half of the respondents felt cost of testing should not determine decision to test, majority of the respondents expressed a lack of confidence in the accuracy of test results and were worried about possible negative impacts of testing modalities on the health of their babies and the outcome of their pregnancies. All these may explain why majority of the respondents were generally not willing to undergo further testing once a negative result was gotten from screening tests. This study varied with results from Ibadan, Denmark and Croatia where majority of the participants had favourable attitudes. Our study agrees with a study done in the United States which analysed how a group of women who refused the offer of maternal serum screening accounted for their decisions and compared their explanations with those who took the test. This study found that refusal did not signify rejection of and/ or resistance to the offerings of science and technology. Rather, women who refused often employed biomedical categories, particularly the concept of ‘risk’, to reject its very offerings.

Our findings however agreed with a similar study carried out among Nigerian women to ascertain their attitude to prenatal screening tests where education and good socioeconomic status had a positive correlation with women’s attitude to prenatal screening services. Results of this study also differed with the study done in Ibadan where age range 30 to 34 years and tertiary education were some of the demographic predictors of attitude towards acceptance of prenatal diagnosis. However, it agrees with this study in that employment status was a predictor of attitude.

The fact that only a minority of respondents had undergone any testing at all including first trimester ultrasound scans
raises concern about the quality of antenatal care received in those areas and suggests increased rates of missed diagnosis and by consequence, missed opportunities for counselling and subsequent supportive care. Unlike in a similar study carried out to evaluate predictive factors towards uptake,[20] only knowledge of genetic diseases was associated with use of tests in our study. Ignorance still remained the major reason for non-uptake, which agrees with similar studies on the subject.

Finally, in our study, only a minority of the respondents were in favour of terminating affected pregnancies. To possibly understand this decision, a look at the study done to elucidate reasons for accepting or declining prenatal diagnosis for Down syndrome, corresponding statements declining prenatal diagnosis were ‘termination of pregnancy is not an option’.[21] It would seem therefore that pregnant women are concerned about having to make the decision to terminate their pregnancy and many are not comfortable with making it. In our study, religion was not significantly associated with decision to terminate affected pregnancies.

The fact that our study did not take into account the number of antenatal visits the respondents had made, considering that majority of them were experiencing their first pregnancy, constitutes a limitation to this study.

Conclusion
More attention needs to be paid to the overall education of adolescents in order to bridge the gaps in knowledge about genetic diseases and prenatal genetic testing. Pregnant women should also be educated about the genetic diseases especially down syndrome including the burden, risk factors, consequences and various modalities of risk assessment and diagnosis as well. There is room for improvement in subsidising healthcare costs in the study area in order to facilitate use of tests. Finally, attention should also be paid to providing supportive care and genetic counselling for parents who are willing to keep affected pregnancies.

Authors’ Contributions
Chibuzor Franklin Ogamba was responsible for the concept and design of the study, definition of intellectual content, literature search, and the acquisition of data, analysis and interpretation of the data as well as preparation of the manuscript.

Alero Ann Roberts contributed to the design of the study, definition of intellectual content, the interpretation of the data as well as the manuscript editing and manuscript review.

Mobolanele Rasheedat Balogun contributed to the design of the study, interpretation of data, manuscript editing and manuscript review.

Chibuikem Anthony Ikwuegbuenyi contributed to the data inputting and analysis, interpretation of data, manuscript editing and manuscript review.

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Conflict of Interest
The authors disclose that they have no conflicts of interest or competing interests. The authors state that the manuscript has been read and approved by all the authors, that the requirements for authorship as stated in the instructions to authors have been met, and that each author believes that the manuscript represents honest work.

Availability of Data and Materials
The datasets used and/or analysed during the current study are available from the corresponding author on reasonable request.

References


