Ghana Journal of Education and Teaching
2024 edition, Vol. 14 No. 2 ISSN: 0855-4064

Genetic Testing: Knowledge, Attitude and Genetic Risk Information of Women, Implications for Counselling among Girls in Oyo State, Nigeria

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Abstract

Genetic diseases or congenital abnormalities resulting in developmental disorders among children that has led to untold hardship in families could have been averted if women had gone through genetic testing before marriage or conception. This study examined the Knowledge, Attitude and Genetic Risk Information (GRI) of Women on Genetic Testing in Oyo State, Nigeria. The study adopted a descriptive survey design. Questionnaire was used to elicit information from 200 respondents. Three null hypotheses were raised and data collected were analysed using frequency counts, percentages, means, standard deviation, Chi-square, Pearson Product Moment Correlation (PPMC).Chi-square results socio-demographic characteristics (age, educational qualification, employment status, marital status, number of children, tribe and religion) and knowledge on Genetic Testing of respondents on genetic testing showed that educational qualification (X^2= 41.64, p=0.00) and tribe (X^2=12.60, p=0.00) influenced their knowledge. Secondly, religion X^2 = 7.97, p=0.02 was significant in influencing the attitude of the respondents towards genetic testing when compared with socio-demographic characteristics. However, genetic risk information and attitude towards genetic testing was significant using PPMC as (r= 0.17, p = 0.02). In conclusion, educational qualification and tribe influenced the knowledge of the respondents on genetic testing. Religion and the availability of genetic risk information positively affected individuals' attitude towards genetic testing. It was recommended that education on genetic testing and genetic risk information should be organised so as to educate women about genetic testing, thereby influencing their attitude positively towards their participation in genetic testing.

Keywords: Knowledge, Attitude, GRI, Women, Genetic Testing

1.

The condition caused by abnormalities in an individual's DNA is called genetic disease. It can be inherited from either or both parents and it can also happen due to spontaneous genetic mutation. While genetic testing is a process of analysing someone's Deoxyribonucleic Acid (DNA) in order to determine if there is a mutation that can cause disease, however, scientific advances play important roles in the field of medicine nowadays. For example, pre-natal genetic testing has been successfully utilised as a tool in ameliorating chromosomal abnormalities in countries where it is available and accessible for the inhabitants whether citizens or foreigners living in those countries. There have been commercially available pre-natal genetic screening tests due to the availability of clinical genetics give room for the circulation of cell-free fetal DNA in a maternal blood sample to identify fetal risk for specific congenital defect or defects as the case may be. (Webber, 2023).

Globally, prevalence of genetic disorders and congenital abnormalities is assumed to be 2%-5% of all live births because it is rare. For example, sickle cell disease is one of the most common genetic disorders in Nigeria with carriers of the mutant gene accounting for about 24% of the population and about 150,000 children born yearly are affected with this disease in Nigeria alone. Some other genetic diseases found in Nigeria are: Down syndrome and Turner's syndrome. According to the finding of Hanan et al, (2015), Down syndrome in Nigeria has an incidence of 1 in 865 live births with a high incidence of cases among young mothers. The scanty record of prevalence of genetic diseases especially in Nigeria, is largely attributed to substandard healthcare facilities available at health centres leading to the mortality of some children born with genetic diseases or congenital abnormalities. Which could have been averted if the parents had done genetic testing before marriage (Hanan, Saher & Fatma, 2015).

Moreover, congenital diseases or congenital abnormalities can be prevented, or reduced to the barest minimum by providing accessible and adequate pre-natal and post-natal genetic testing routines to the populace at no-cost or subsidised rate and performing non-invasive pre-natal testing (NIPT) especially between 9th and 10th week of pregnancy. The high-risk group is consanguineous marriages that can lead to a recessive syndrome due to marriage from the same ancestor but could be subjected to proper and adequate genetic counselling in order to have a safe and sound baby. The increasing availability of predictive genetic testing for late-onset diseases means that there is a growing need to understand the psychological consequences of such testing. According to Lin, Hasbullah, Sivam, Shanmugam, Augustine, Htay, Moe and Soe, 2022, when newly diagnosed patients with genetic disease or rare hereditary disorders are discovered, genetic testing can assist in early detection and receive disease-specific treatment as soon as they show up.

Genetic testing also known as DNA testing is a kind of test that has the capacity to identify defects in genes, chromosomes or proteins in an individual's body. This is done through the sample from the blood, hair, skin, tissue or amniotic fluid. The test can detect or confirm if there is any genetic disorder and can equally determine if there is any chance of developing or transferring a genetic disease (ACOG, 2022; AMA, 2022).

Although, the psychological consequence of genetic testing for adults is determined but that of children who are the most vulnerable to emotional and psychological trauma are not fully

established regarding the emotional distress and the effect on their self-esteem (Broadstock, Michie, Marteau, 2000). Various methods are available for smart and reliable testing of the DNA in humans. People at risk are duly referred to a qualified physician, who specialises in investigating, diagnosing and treating individuals who are suspected of having, or who do actually have, genetic disorders. In the process of examination, the background to the disorder is carefully considered, as well as any personal and family precedents and symptoms are fully ascertained. If a specific genetic disorder is suspected, a genetic test is proposed instantly and the diagnosis is carried out to establish the truth about its existence (Phadke & Gowda, 2013).

Some genetic tests check if there are deletions or insertions in the genetic materials. There are several methods to detect such changes but (Multiplex Ligation Dependent Probe Amplification (MLPA) analysis or Polymerase Chain Reaction (PCR) analysis are the most commonly used. Also, DNA changes can be visibly enormous: a missing or added piece of a chromosome, an entire chromosome, or swapping of chromosome fragments (called translocation) will have to be analysed by microscopically coding at the entire chromosome. Considering the studies on chromosomes or genes, genetic testing can also include biochemical tests to identify the presence or absence of Radiometric-microbiological assay (RMA), epigenetic changes (of DNA associated molecules) and biochemical tests, key proteins or their products. The proteins are made by genes; therefore, an abnormal protein implies that the gene has mutated or transfigured (Phadke, *et al.* 2013).

1.1 Statement of the Problem

Genetic diseases or congenital abnormalities are real and are promoting issues in the family due to the stress involved in taking care of children with genetic diseases. Moreso, genetic testing raises concern about privacy and confidentiality of sensitive genetic data. Hence, ensuring informed consent and autonomy in decision making regarding genetic testing is key in order to uphold ethical standards. Ethical consideration also includes the responsible use of genetic information to avoid stigmatization and discrimination based on revealed genetic dispositions. Therefore, promoting equity in access to genetic testing services and ensuring that individuals understand the implications of genetic testing results are essential ethical considerations for example, having witnessed or experienced directly or indirectly, the emotional and psychological consequences on parents of children with congenital abnormalities in the society, in order to ameliorate the future occurrence of these abnormalities and the accrued stress and financial burden on the significant others for instance, parents, siblings, the vulnerable children, government and the society at large this study set to assess the knowledge of women on genetic diseases, pre-natal genetic testing, willingness to do the test, attitudes of women towards genetic testing, use of common tests available as well as willingness to terminate affected pregnancies in case they had married before discovering their genetic statuses, the knowledge of mothers' opinions on newborn screening, their attitudes towards the need for consent and the impact of a diagnosis of a genetic disease or carrier status. Therefore, genetic education is necessary to promote informed decision-making, reduce genetic essentialism and improve genomics literacy. For this cause, this study was on Genetic Testing: Knowledge, Attitude and Genetic Risk Information among Women in Elere, Iddo Local Government Area, Oyo State, Nigeria.

1.2 Purpose and Objectives

The general purpose of the study was to: examine the awareness and attitude of women towards genetic testing in Elere, Iddo Local Government Area, Oyo State, Nigeria

The specific objectives are as follows:

(a.) determine the socio-demographic characteristics of the women in Oyo State regarding genetic testing;
(b.) evaluate the knowledge of women on genetic testing in Oyo State;
(c.) assess the genetic risk information available to respondents
(d.) identify the attitude of women in Oyo State towards genetic testing

1.3 Justification of the Study

Genetic testing is the most reliable way to make an accurate diagnosis of some specific disorders. It has numerous advantages irrespective of the final results positive or negative. A gene mutation test result provides a sense of relief from the fear of uncertainty and prepares people for adequate access to informed decisions about managing their health irrespective of their gender. For example, a negative result can eliminate the need for unnecessary checkups and screening tests in some cases in the future. A positive result guides a person towards available preventive procedures, monitoring and treatment options. Some test results equally allow people to make decisions about having children or not depending on the couple. Newborn screening is used to identify genetic disorders early in life in order to commence treatment as early identification is key in managing the disease or diseases identified. The study will serve as a guide to counselling girls on the importance of genetic testing; acquaint them with the knowledge on genetic testing and redirect them to having a right attitude towards genetic testing.

1.4 Hypothesis of the Study

$H0_1$: There is no Significant Association between the Socio-Demographic Characteristics of the Respondents and their knowledge on genetic testing

$H0_2$: There is no Significant Association between the Socio-Demographic Characteristics of the Respondents and their Attitude towards Genetic Testing

$H0_3$: There is no Significant Relationship between Genetic Risk Information and Attitude of the respondents towards Genetic Testing

Theoretical Framework

This research is hinged on Health Belief Model (HBM) which is the tool that scientists use to predict the behavior of human beings at any point in time. This model was developed in the 1950s and was formally proposed by a group of social psychologists who are Godfrey Hocbaum, Irwin Rosenstock, Rosenstock and Kirscht. The theorists propounded that an individual's willingness to change his or her behavior is related to their health perceptions (Boskey, 2023).

The model assumes that one's belief about health and health conditions largely determines one's health-related behavior. The salient factors that affect one's approach to health are:

a. How susceptible someone thinks he or she is to illness

b. One's thought about the consequences of becoming ill
c. Accessibility of information that prompts one to take action
d. Any barrier one thinks might pose a risk in one's way to success
e. One's confidence in one's ability to succeed in life
f. The thought of the benefits of engaging in healthy behavior (Boskey, 2023).

The Components of Health Belief Model include:

a. Perceived severity: Changing one's health behavior pends on one's feeling on how grave the consequences will be.

b. Perceived susceptibility: Some people will not change their health behavior unless they are at risk.

c. Perceived benefits: People may not agree to change their behavior unless there are benefits to gain from it.

d. Perceived barriers: Some will not change their health behavior because of the difficulty in doing so. E.g. it may involve time, money and deliberate effort to accomplish this.

e. Cues to action: These are external events that initiates a desire to make a change in health behavior.

f. Self-efficacy: This deals with someone's belief in his or her ability to make a health-related change (Boskey, 2023).

Health Belief Model can be used to create programs and interventions designed to assist in the prevention of health challenges, support behaviour change of girls and treat behaviour where necessary. It is also effective in producing behaviour change interventions. According to research, Health Belief Model (HBM) is used for designing strategies to promote healthy behaviour, improve the prevention and treatment of health conditions (Boskey, 2023).

Methodology

Method of Data Analysis

Research Design

In this study descriptive research design was employed for collection of data.

Population

The population of the study consisted of 200 females with age range of 18-60 years in the study area, Oyo State.

Sample and Sampling Technique

Simple random sampling technique was adopted in this research to determine the proportion of respondents from different locality in the study area, Ibadan, Oyo State.

Research Instrument

The questionnaire is made up of three sections, namely, Section A: Socio-demographic characteristics of the respondents {sex, age, religion, marital status etc.}. Section B: Basic information of the respondents. Section C: Genetic risk information available to respondents. Section D: Attitude of the respondents to genetic testing.

Validity and Reliability of Research Instrument

The validity and reliability test in this study was conducted to assess the knowledge, attitude and genetic risk information available to individuals in order to determine their attitude to genetic testing. Face validation was carried out by experts in the researcher's department and the reliability of the instrument was 0.60 based on the test-retest method using Cronbach's alpha. 200 copies of the instrument (questionnaire) were personally administered to the respondents by the researchers in Oyo State. The researchers gave the respondents ample time to fill the questionnaire after which they were collected and analysed using descriptive statistics e.g. means, percentages, frequency and standard deviation and inferential statistics such as Pearson's Product Moment Correlation coefficient (PPMC) and Chi-square was used to test the hypotheses of the study

Method of Data Analysis

Data collected was analysed using both descriptive and inferential statistics. Descriptive statistics such as means, percentages, frequency counts and standard deviation while inferential statistics such as Pearson's Product Moment Correlation coefficient (PPMC) and Chi-square were used to test the hypotheses of the study

Presentation of Findings

Table 5: Frequency Distribution of Socio–Demographic Characteristics of Respondents

Variables	Frequency	Percentage
Age		
18-30	51	25.5
31-40	87	43.5
41-50	24	12
51-60	35	17.5
Above 60	3	1.5
Educational qualification		
No formal education	1	0.5
Primary school	10	5
SSCE	26	13
OND	21	10.5
HND	54	27
B.Sc.	73	36.5
M.Sc.	15	7.5
Employment status		
Unemployed	32	16

Employed	110	55
Self employed	58	29
Marital status		
Single	37	18.5
Married	140	70
Divorced	9	4.5
Widow	14	7
Number of Children		
None	42	21
One	12	6
Two	60	30
Three	64	32
Five or more	22	11
Tribe		
Yoruba	162	81
Igbo	35	17.5
Hausa	3	1.5
Religion		
Christianity	158	79
Muslim	40	20
Others	2	1

From table 1, it was observed that the largest age group was 31-40 (87 persons or 43.5% of the sample). The distribution showed that females from all age groups were well represented. For educational qualifications, the largest group were those with B.Sc. degrees (73 individuals or 36.5% of the sample). This implied that the majority of the respondents were fairly educated. In terms of employment status, the largest group were those who were employed (110 individuals or 55% of the sample). In this study, the married formed the majority of the respondents, accounting for 70%. Considering employment status, the largest group were those who were employed (110 individuals or 55% of the sample). In terms of marital status, the largest group were those who were married (140 individuals or 70% of the sample). Regarding the number of children, the largest group were those with three children (64 individuals or 32% of the sample). In terms of tribe, the largest group was Yoruba (162 individuals or 81% of the sample). In terms of religion, the largest group was Christianity (158 individuals or 79% of the sample). These results provide a general overview of the distribution of individuals in the sample based on the six variables.

Result of Hypotheses

Hypothesis 1: There is no Significant Association between the Socio-Demographic Characteristics of the Respondents and knowledge on Genetic Testing

The socio-economic characteristics considered were age, educational qualification, employment status, marital status, number of children, tribe and religion. The significant relationship was determined at 0.05 level. The chi-square statistical analysis revealed that there was a significant association: between Educational qualification and knowledge on genetic testing (χ^2=41.64, p-value=0.00) and between Tribe and knowledge on genetic testing available to the respondents (χ^2=12.60, p-value=0.00).

Table 2: Test of Significant Association between the Socio-Demographic Characteristics of the Respondents and Knowledge on Genetic Testing using Chi-Square

Variables	**χ^2**	**df**	**p-Value**	**Decision**
Age	0.44	4	0.98	NS
Educational qualification	41.64	6	0.00	S
Employment status	2.73	2	0.26	NS
Marital status	2.49	3	0.48	NS
Number of children	3.38	4	0.50	NS
Tribe	12.60	2	0.00	S
Religion	0.05	2	0.97	NS

NS = Not Significant
S = Significant

Hypothesis 2: There is no Significant Association between the Socio-Demographic Characteristics of the Respondents and their Attitude towards Genetic Testing

The chi-square statistical analysis showed that there was a significant association between Religion and attitude of women towards genetic testing (χ^2=7.97, p-value=0.02) at 0.05 level of significance. There was no significant relationship between (Age, Educational qualification, Employment status, Marital status, Number of children, and Tribe) and attitude towards genetic testing.

Table 3: Chi-Square Test of Significant Association between the Socio-Demographic Characteristics of the Respondents and their Attitude towards Genetic Testing

Variables	**χ^2**	**df**	**P-Value**	**Decision**
Age	4.95	4	0.29	NS

Educational qualification	6.89	6	0.33	NS
Employment status	2.57	2	0.28	NS
Marital status	3.20	3	0.36	NS
Number of children	1.96	4	0.74	NS
Tribe	3.16	2	0.21	NS
Religion	7.97	2	0.02	S

NS = Not Significant
S = Significant

Hypothesis 3: There is no Significant Relationship between Genetic Risk Information and Attitude towards Genetic Testing

PPMC was used to test for significant relationship between genetic risk information and attitude of the respondents towards genetic testing. The significance of this association was determined at 0.05 using Cronbach's Alpha. The correlation test showed a significant relationship between genetic risk information and attitude towards genetic testing (r= 0.17, p-value = 0.02).

Table 4: PPMC Test of Significant Relationship between Genetic Risk Information and Attitude towards Genetic Testing

Variables	**r**	**p-Value**	**Decision**
Relationship between genetic risk information and attitude towards genetic testing	0.17	0.02	S

S = Significant

DISCUSSION

The results of the study revealed that the respondents had positive knowledge on genetic testing based on educational qualification and tribe. This suggests that age, employment status, marital status, number of children and religion did not have a significant impact on the knowledge individuals have on genetic testing. On the other hand, the study found a significant positive relationship between religion, genetic risk information and attitude toward genetic testing. This suggests that religion and exposure to genetic risk information play a role in shaping individuals' attitudes toward genetic testing. This indicates that the type of religion practiced by an individual can determine the attitude towards genetic testing and the more access to genetic risk information individuals have, the more positive their attitude toward genetic testing will be. In conclusion, these results suggest that while individual factors such as age, employment status, marital status, number of children, and tribe did not significantly impact the attitude of individuals towards genetic testing, educational qualification and religion play a role in restructuring individuals'

attitudes towards genetic testing. Additionally, the availability of genetic risk information positively influences individuals' attitudes toward genetic testing.

CONCLUSION

Based on the results of the study, it was concluded that educational qualification and tribe influence the knowledge of the women on genetic testing. But age, employment status, marital status, number of children and religion did not influence their knowledge on genetic testing. Moreover, religion has a strong relationship with the attitude of the respondents towards genetic testing. Also, there was a significant association between genetic risk information and the attitude of the respondents towards genetic testing. However, age, employment status, marital status, number of children and tribe do not have any significant association with genetic risk information and attitude toward genetic testing. This suggests that there may be other factors that influence an individual's attitude toward genetic testing, such as cultural beliefs, religious affiliation and personal experiences among others.

From the findings, it was recommended that more education and awareness campaigns be organised to educate individuals about genetic testing, particularly in areas where access to information and healthcare services may be limited or not accessible. This could involve partnerships with local organisation, community leaders, religious leaders, teachers, counsellors, policy makers, in addition to improved access to information, collaboration with Health Care Providers, Promoting Ethical and Legal Considerations, Encouraging More Research in other climes to reach the global population. Moreover, Reproductive Genetic Technology (RGT) is an implication for ethical and religious bodies like Judaism, Christianity and Islam all because prenatal genetic testing can influence the decision of parents to abort a pregnancy. Also, the three religions are of different opinions on the beginning of human life with different doctrines to back it up (Peter, Steven, Sohiel, Makenna, Ameen, 2024). It is noteworthy that genetic testing may prompt complex moral dilemma because patients may find it difficult to reconcile their religious values with the ambiguity of genetic risk and the terrible choices they may need to make sometimes. Even genetic professionals have their share of the issues because they may experience conflicts concerning how to respond when a client’s religious belief influences him or her to make decisions that are against their medical interest. For example, religious law prohibits abortion, but some religious individuals often opt to have their pregnancy terminated if it is deformed while others usually prefer to depend on God for a miracle.

RECOMMENDATION

Based on the results of the study, the following recommendations were deduced:

1. Enhanced Access to Information: The availability of information and resources on genetic testing can be improved through the development of educational materials, such as brochures, posters, handbills and websites, which can be easily accessible, with stimulating captions and understandable to the public irrespective of the age and status.
2. Sensitisation of women and girls on the importance of genetic testing: A widespread campaign should be conducted to educate individuals especially women and girls about the availability, importance, and potential benefits of genetic testing. This should be made up of

workshops, seminars and public meetings to make people to better understand the subject and ginger a positive attitude towards the genetic testing.

3. Targeted Campaigns: Targeted campaigns aimed at specific populations e.g. in religious centres, markets, schools, health care centres, palaces, village squares or town halls etc., where with low educational qualifications, could be reached more effectively to enhance their knowledge, to be equipped with genetic risk information and general understanding on genetic testing. This could involve partnerships with local organisations, community leaders, market leaders and health care providers to reach these populations.
4. Synergy among Health Care Providers: Strong collaboration between health care providers, genetic counselors, and other healthcare professionals could ensure that individuals have access to accurate and timely information on genetic testing, as well as access to genetic testing services.
5. Promoting Ethical and Legal Considerations: It is important to ensure that the public highly informed about the ethical and legal considerations surrounding genetic testing, such as genetic discrimination, privacy, and confidentiality. This can be done through the development of educational materials and adequate sensitisation on the subject matter.
6. Giving Room for Further Studies: Further researches in different parts of Nigeria are needed to better understand the attitude and beliefs of individuals towards genetic testing, as well as to assess the impact of different educational and sensitisation programmes on the subject matter. This will help to improve future campaigns and ensure that they are effective in promoting greater knowledge and understanding on genetic testing

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