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Full Length Research Paper

Level of awareness of genetic counselling in Lagos, Nigeria: its advocacy on the inheritance of sickle cell disease

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A descriptive analysis of reported cases of sickle cell disease and the level of awareness about genetic counseling in 30 hospitals were carried out. Additionally, 150 individuals between ages 16 - 45 were randomly selected for evaluation of genetic counselling awareness. The main tools for this study were questionnaires, which were taken to hospitals, and individuals completed the others. The numbers of reported cases of sickle cell disease recorded in private, public and teaching hospitals were 14 and 57; 143 and 89; 272 and 57 for the periods of 1995 - 2000 and 2001 – 2005, respectively. A general informal genetic counselling took place mostly occasionally in the hospitals visited. 122 (86%) individuals have had the knowledge of genetic disease and only 43 (30.3%) individuals have been exposed to genetic counselling. 64% of individuals agreed that genetic counselling would help in the prevention of genetic disease.

Keywords: Sickle disease, genetic counseling, genetic testing, advocacy.

INTRODUCTION

Genetic disorder is a disease condition that occurs as a result of mutations, which could be fatal and cause varying degrees of harm (Taylor et al., 1997). Gene mutation is changes in gene structure and it causes sudden and spontaneous changes in the phenotype (Hartwell et al., 2000). It occurs as a result of a change in the nucleotide sequence in the DNA molecules in a particular region of chromosomes. The genes that have been altered are referred to as mutant genes. Gene mutation can cause loss, addition or rearrangement of bases in the gene. The mutation takes different forms and these include duplication, insertion, deletion, inversion or substitution of bases (Odunlade, 2005). Examples of diseases that occur as a result of substitution mutation are sickle cell anemia, cystic fibrosis, phenylketonuria and hemophilia.

Sickle cell disease is a common genetic condition that affects hemoglobin – inheritance of mutant hemoglobin genes from both parents resulting in HbSS. It occurs at a frequency of 1 out of 1600 among black people. Another sickling variant is HbSC disease, which is milder sickling disorder. It is present in 1 of 1100 African Americans. Symptoms are similar to sickle cell disease but less frequent and severe. In S-beta thalassemia, the individuals inherit one sickle cell gene and one gene for beta thalassemia, another inherited anemia. Sickle cell trait is the heterozygous carrier state of HbAS. These individuals are generally healthy as non-carriers. Prevalence varies from one country to another. Sickle cell trait occurs in about 8% African Americans and 20 - 30% in Nigeria (Reid and Famodu, 1988) and 20 - 40% in Africa (Fleming and Lehman, 1982).

Sickle cell anemia contributes the to equivalent of 5% of under-five deaths on the African continent, more than

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9% of such deaths in West Africa, and up to 16% of under-five deaths in individual West Africa countries (WHO, 2005). Many also die before their reproductive age. Sickle cell anemia poses serious health concern, especially in developing countries. Awareness on genetic understanding and screening is not a common practice and the diagnosis is usually made when it is presented with a severe complication. Even when tragedies such as two or more miscarriages, still births, or children die in infancy, many at times doctors do not order a blood test to take a closer look at genetic make up of parents or refer them to a genetic counselor. Therefore, the most important challenge is to raise the awareness on its causes and prevention through health education. In Nigeria, no genetic counseling clinic is known as at the time of this study. In many developed countries, genetic testing and genetic counseling have been commonly done to identify carriers.

Increasingly, genetics seems to provide the answer to the basis of disease and to offer insight into the status of the health of populations. Medical genetics offers genetic testing as a tool for diagnosis and through genetic counseling, individuals will be provided with an accurate understanding of genetic inheritance and what it means to be 'at risk'. The usefulness of genetic testing relates to the efficacy of disease prevention and the right of a person to know his or her own genetic heredity. This will bring about a substantial impact on health improvement. Genetic screening for personal health will provide information about the health and well being (not necessarily reproductive health) while in other cases it may be used for health related reproductive risks. Testing for genetic disorder will also provide a person with relieve of the uncertainty of not knowing, especially when such a person has a previous record of any suspecting disease. For example, a family member is currently suffering from the diseases-infertility, miscarriage, still birth or early child mortality. It puts people concerned in a better position to make major life decisions and it will also benefit family members either now or in the future. Additionally, genetic testing has become a tool for parents to decide whether to have only affected and/ non-affected offsprings.

When genetic testing has been carried out to establish a diagnosis of hereditary or congenial diseases in affected patients with high accuracy, it is usually accompanied with genetic counseling. Genetic counseling will help establish a diagnosis of hereditary diseases in affected patients, take measures to alleviate the clinical manifestations of such disease, predict the probability of development of a disease in families/individuals not yet affected and possibly prevent it. Furthermore, in the prediction of giving birth to an offspring with a genetic disease and proffering options to take decisions, this will help to distort the general confusion about the potential benefits and risks of genetic testing. Information about genetic testing and counseling should be part of regular medical practice to achieve desired level of knowledge and a change in attitude.

Interestingly, genetic counseling now has a very broad scope with the availability of several new DNA tests. It is simply a communication process, which involves diagnosis, explanation and options. However, different countries have different ways of defining genetic counseling (UNESCO, 1995). In 2006, the Genetic Counselling Task Force of the National Society of Genetic Counsellors (NSGS) provided a new definition of genetic counseling as – the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease (Resta et al., 2006). This process integrates the following: interpretation of family and medical histories to assess the chance of disease occurrence or recurrence, education about inheritance, testing, management, prevention, resources, research and counseling to promote informed choices and adaptation to the risk or condition.

The notion that genetics defines one's identity and one's destiny is not deeply rooted in the Nigerian cultures. There is varying degree of public awareness and understanding of issues concerning human genetics in general and genetic testing in particular. It is expected that genetics could find applications in new strategies of prevention and risk minimization based on better understanding of it. Similarly, only few doctors in Nigeria today can be defined like those in the western world with regards to training in the field of genetic counseling. Furthermore, genetic practice has not been given the serious attention that is needed. Similarly, genetic services are absent. Access and availability to genetic services will help in referring patients to genetic counselors, which will improve the detection of genetic risk factors.

Urbanization in Nigeria is growing rapidly and the health services are increasingly under strain. At least 70% of the population live in towns and cites. The pattern of medical care in these cities is not intrinsically different from what is obtained in other countries. The only draw back is the non-availability of facilities like modern equipment and poor working conditions. Government provision includes General hospitals, primary health centers as well as University teaching hospitals. Owner-ship of health establishments is divided among federal, state and local governments. The Ministry of Health coordinates health services throughout the country including those provided by the government, by private institutions and missions.

There is a general acceptance of modern medical care. Western medical care was formally introduced into Nigeria until the 1860s, when the Sacred Heart Hospital was established by the Roman Catholic Missionaries in Abeokuta (Southwest Nigeria). In the past, the Federal government has launched various health programmes such as Primary Health Care Plan (PHC), Expanded Programme on Immunization (EPI), National Family Health programmes and many other ones. Implementation of these programs took place mainly through collaboration between the Ministry of Health and participating government councils, which receives direct subvention from the Federal government. The mission hospital still remains important components of the health care system medical services to the Nigeria people.

It is well known that not every ill person consults health care professionals; many do visit traditional homes. The social and cultural factors may influence the health of many individuals in Nigeria especially among the rural dwellers. Cultural and spiritual health beliefs vary among different ethnic groups. Additionally, people visit traditional healers because they lack financial capacity to visit modern hospitals. Nowadays, both western medicine and traditional medicine are being used among the urban and rural dwellers. Currently, there is a challenge to provide continuing health facilitates to the people by the government.

The health care system constitutes the precondition for the emergence of the genetic conception of health. There should also be a safe and supportive environment in the country for taking up genetic screening programs. To this end, Nigerians need to be exposed to genetic testing/ genetic counseling and incorporating genetic counseling programs into our health care system. Emphasis on genetic counseling has been low in Nigeria; this could be because its importance has not been fully realized by the Ministry of Health. Moreover, religious and cultural beliefs of people regarding the origin of sickle cell disease and its treatment are drawbacks. There is a general belief that genetic disorders are caused by witchcraft powers of the wicked. Even where diagnosis has been made they ignore advice. The need to promote awareness among health care professions and the general public about the great potentials embedded in genetic testing and genetic counseling to make substantial impact on health improvement is of great importance in Nigeria. Hallowell and Richards (1997), have reviewed studies of risk awareness of carriers of genetic disorders and individuals who attended genetic counseling.

Awareness of an identifiable genetic risk factor in a family makes it possible to discuss and offer genetic counseling. In one study, limited alertness and awareness about genetic and tetratogenic risk factors among GPs were noticed (Aalfs et al., 2003). GPs are supposed to play an important role in the referral for reproduction counseling, which preferably should take place before conception. It is important to increase public awareness, education and understanding of genetic concepts being organized for both the public and the media. Extensive education on health programmes such as raising awareness about the importance of the genetics testing and genetic counseling could be extended to the students at different levels of secondary and tertiary institutions. The implementation of population genetic screening programmes has been extensively reviewed by Garson et al., (2006). With adequate genetic counseling, the high frequency of sickle cell anemia disease could be prevented in Nigeria.

The objective of the present study was to estimate the frequency of reported cases of sickle cell anemia in some private and public hospitals in Lagos metropolis (Southwest area) for a given period. Furthermore, it was to evaluate the level of awareness about genetic counseling among medical practioners in these hospitals and among some selected individuals in the University of Lagos community and environs. Advocacy on genetic counseling in the area of communicating information about sickle disease among Nigerians is also emphasized in this paper.

MATERIALS AND METHOD

Survey /collection of data on the reported cases of sickle cell disease from hospitals

The criteria for selection of hospitals were based on those with standard care of medical services. Forty (40) hospitals were randomly selected across Lagos city, Nigeria, which is an urban commercial centre. Private (30), Public (8), and teaching hospitals (2) were visited. A survey on the number of reported cases of sickle cell disease in the hospitals from 1995 – 2005, was carried out. The findings of the frequency were recorded in a pre-designed form by the hospitals and the sickle cell disease data were collected from the medical records of the hospitals.

Survey on the level of awareness of genetic counseling in hospitals

The level of awareness of genetic counselling in these hospitals was equally evaluated. The main tools were questionnaires. These were taken to the hospitals with a letter of introduction. The questionnaires consist of 10 questions. It is aimed at knowing the name of the hospital, its location, medical personnel's designation, medical personnel's knowledge about genetic disorder, prenatal diagnosis, genetic counseling and if they possess genetic units. The structured questionnaires were self administered following informed consent. A total of 30 hospitals responded, giving a res-

ponse rate of 75%. Officers of medical records, pediatrics and obstetric departments jointly completed the questionnaires. In addition to collecting data concerning sickle cell anemia, they were urged to complete the level of awareness about genetic counseling, since; they were not used to providing such information.

Survey on the level of awareness on genetic counseling among the individual subjects

A randomized cross-sectional study of level of awareness of genetic counseling was evaluated among 150 individuals between the ages of 16 and 45 years. The individuals to complete the questionnaires were identified on a daily basis from the University of Lagos community. This community was used for this study because it is an enlightened society where they are likely to be interested in this kind of survey. This was designed to know the subjects' name, age, sex, academic status, what they think is the cause of genetic disorders, assess the level of awareness about the knowledge of genetic counselling where he/she had received the genetic counseling and possibility of the use of concept of genetic counseling in the prevention of genetic diseases. This was carried out within a period of five months. During the completion of the questionnaires, salient facts of heredity of sickle anemia cell were explained by the study team to help complete the form. For the purpose of this study, genetic counseling was seen as a communication process that entails diagnosis, explanation and options, thus providing information to people concerned about the risk of inheritance of a genetic disease. Respondents were asked to complete the questionnaires immediately after receiving it in the presence of the study team. Communication among individual subjects was not allowed during completion of the questionnaires. In most of the instances, individuals were encouraged to take part in this survey. The choice of media as a place where they received genetic counseling was based on the answer provided by respondents.

Measures

The frequency of sickle cell disease data from the hospitals was measured as incidence. Hospitals' awareness about genetic counseling was evaluated on number and response rate. Level of awareness on genetic counseling among the individual subjects was based on response rate (Yes/No options). Places where respondents have received genetic counseling were assessed by giving five places. The respondents indicated others.

Data analysis

Hospitals return both form/questionnaires and those returned by individual subjects were collated, evaluated and a descriptive statistics was used to describe the outcome measures. Numbers of reported cases were stated and percentages were calculated in other cases.

RESULTS

Frequency of reported cases of sickle cell disease

Table 1 shows the occurrence of reported cases of sickle cell disease. Most of all the private hospitals visited had cases of sickle cell anemia between the years 2001 - 2005 compared to 1995 - 2005, while public hospitals had more cases of sickle cell anemia between 1995 - 2001 than years 2001 - 2005, which is the same in the teaching hospitals The numbers of affected patients are relatively high in public hospitals.

Background information on genetic unit, prenatal diagnosis and genetic counselling in hospitals

Forty (40) hospitals were visited for this study and thirty (30) hospitals responded; private hospitals (22), public hospitals (6) and teaching hospitals (2). Five (5) private hospitals out of the hospitals visited have genetic unit, while all the public and teaching hospitals do not. Similarly, only three private hospitals are carrying out prenatal diagnosis, this also correlates with the fact that most of the hospitals do not have genetic unit. Hence, prenatal diagnosis revealing genetic fitness may not be suggested to the patients. Table 2 shows that private hospitals carry out genetic counselling occasionally and others do it frequently. Public and teaching hospitals carry out genetic counselling frequently although they do not have genetic units (Table 2).

Respondents, knowledge about genetic disorder

Table 3 shows the respondents, knowledge about genetic disease. A high percentage of each age bracket has the knowledge of genetic disease when compared to those who do not have the knowledge of genetic disorders.

Studies on genetic counselling awareness

Table 4 shows the respondents, level of awareness about genetic counselling. Only 20.4% of age group 16 - 24 have heard about genetic counselling, 9.9% in age group (25 - 35) and none in age group (36 - 45). Ninety one (91) individuals are of the opinion that genetic counselling could assist in the prevention of children with sickle cell disease, 38% of individuals in age bracket (16 - 24), 22.5% of (25 - 35) and 3.5% of (36 - 45). About 126 individuals are willing to be educated more on the causes and prevention of sickle sell disease. Table 5 shows the places where individuals had received genetic counselling. Sixty four (64%) of individuals that received genetic counselling were exposed to it in schools, 22% in hospitals, 8% from other sources such as the media and in churches (7%).

Table 1 . Number of reported cases of sickle cell disease.

Hospitals	Frequency of reported cases of sickle cell anemia	
	1995-2000	2001-2005
Private		
A	-	10
B	-	19
C	-	2
D	11	20
E	-	2
F	-	1
G	3	2
H	-	1
I-V	-	-
Public		
A	-	-
B	138	45
C	275	44
D-F	-	-
Teaching		
A	97	35
B	175	22

- No Records

Table 2. Hospital's awareness about genetic counseling.

Hospital	Number of hospitals carrying out genetic counselling	Rate of genetic counselling among hospitals	
		Occasionally	Frequently
Private	19 (22)	11	8
Public	5 (6)	1	4
Teaching	2 (2)	1	1

Table 3. Respondents' knowledge of genetic disorders.

Age bracket	Percentage of people with knowledge	Percentage of people without knowledge
16 – 24	81 (57%)	10 (7%)
25 – 35	37 (26%)	9 (6.3%)
36 – 45	4 (3%)	1 (0.7%)
Total	122 (86%)	20 (14%)

DISCUSSION

The high figure of reported cases of sickle cell disease over ten years obviously reflects that this genetic disorder is still a common disease among Nigerians. This is partly due to the lack of knowledge of couples at risk before marriage and coupled with lack of awareness about its causes and prevention. The total frequencies of sickle cell disease recorded in private hospitals for the period of 1995 - 2000 and 2001 - 2005 were 14 and 57 respectively. In the public hospitals, a total of 143 and 89 reported cases were observed for the period between 1995 - 2000 and 2001 -

Table 4. Respondents' level of awareness on genetic counselling among the individuals.

Age Bracket	Reponses to exposure to genetic counselling		Responses of GC in the prevention of genetic disease		Reponses of willingness to be educated on sickle cell	
	Yes	No	Yes	No	Yes	No
16-24	29 (20.4%)	62 (43.7%)	54 (38%)	37 (26.1%)	80	11
25-35	14 (9.9%)	32 (22.5)	32 (22.5%)	14 (9.9%)	41	5
36-45	-	5 (3.5%)	5 (3.5%)	-	5	-
Total	43 (30.3%)	99 (69.7%	91 (64%)	51 (36%)	126	16

Table 5. Places where individuals who have received genetic counseling.

Places where they received it	No of people who have received genetic counselling
Hospital	11 (26%)
School	25 (58%)
Church	3 (7%)
Mosque	- -
Others (e .g . friends, Media)	4 (9%)
Total	43

*Others as indicated by the respondents.

2005 respectively. These high reported cases are because people prefer to visit the public and teaching hospitals, whereas some cases might be referrals from the private hospitals especially when the condition of the patient is very bad. It was observed that some hospitals do not keep statistics of reported cases of sickle cell disease. We were also made to understand that some parents seek for treatment in herbal and religious centres due to poverty and sometimes religious beliefs.

The findings gathered from the questionnaires that were completed by medical personnel in the hospitals visited showed that only 5 hospitals out of the 30 hospitals have genetic units, and only 3 hospitals carry out prenatal genetic diagnosis mainly to ensure the position of fetus before birth and some times to check multiple pregnancies. We learnt that the only prenatal test that is commonly recommended for pregnant women is ultrasound scanning which cannot reveal or detect genetic disorder such as sickle cell anemia. The doctors agreed that a general informal genetic counselling is done during antenatal and postnatal clinics in the private, public and teaching hospitals. This indicates that the importance of genetic counseling is not given so much regard yet in the hospitals. Most Mendelian genetic disorders are rare here, which may explain lack of interest in genetic issues.

It has been observed that most primary care physicians have not seen genetics as important. On the other hand, we understood that sickle cell clinics are organized in teaching and public hospitals for sickle cell patients once in a week.

From the questionnaires distributed among individuals, a total of 122 (86%) have heard about genetic diseases. This study shows that only 43 (30.3%) individuals of the total sample size have been exposed to a form of genetic counselling awareness on sickle cell disease. This shows that the level of awareness is very low. Limited awareness of knowledge of genetic disorders may lead to limited awareness of the implications of genetic risks among people (Watson et al., 1999). 20.4% of those individuals exposed to genetic counselling are within the age bracket (16 - 24) which are the young adults. This explains why 64% individuals were exposed to counseling awareness at schools. Others indicated that they have been informed about genetic counselling through the media (Health programmes provided by T.V stations), friends and very few through interactions with medical personnel. The respondents that provided the media may not have received genetic counselling through the media but might have been aware through health programmes.

The government, religious institutions and hospitals should be more involved in raising awareness about genetic counselling to help in the prevention of giving birth to offspring with sickle cell diseases. Establishment of proper guidelines as regards genetic counselling process is highly necessary for the implementation of genetic testing

/genetic counselling in Nigeria. It was observed that 58% agreed that genetic counselling would help in the prevention of sickle cell disease and about 89% are willing to know more about the causes and prevention of sickle cell disease. A high percentage is in the age bracket (16 – 24). Their interest in learning about this genetic disorder could be due to the fact that they are closest to marriage, however, some were still pessimistic (11%). Many people of the age bracket 36 – 45 were not willing to complete the forms for some ethical reasons, thus, few subjects were drawn from this age bracket.

A recent blood genotype study among 150 randomly selected individuals revealed 26% as sickle cell carriers (Adeyemo and Soboyejo, 2006). Therefore, there is an urgent need to expedite action on public health education to enlighten people about the inheritance of sickle cell disease and placing an emphasis on advocacy for genetic counselling at the national level for its prevention. Moreover, given the fact that individuals who are at risk (sickle cell trait carriers) could easily be identified by inexpensive blood tests, access to genetic testing/ counselling would help to explain genetic risks and this will lead to a reduction in births of affected offspring. More research is needed in the area of public perception of genetic testing/counseling in health care systems in Nigeria.

Advocacy for genetic counselling

Genetic counselling could help identify individuals who are suspected of a heritable disease such as sickle cell disease, at risk because of their family history and concerned about the possibility of having an affected child based upon personal or family history or ethnicity. In addition, they help to consider options. Individuals who have been trained at the master's level in this field usually provide formal genetic counseling. It is also provided by physicians and geneticists who have trained in residency programs accredited and certified by the right board. Personnel involved in medical care with task-oriented basic training can also counsel.

The counsellors make diagnosis (this may involve physical examination, laboratory tests e.t.c), calculate risk and explain the problem. Also, he/she discusses the result of genetic screening with the person who has been tested. In carrying out genetic counselling, provisions for prenatal diagnosis should be made available as well as screening of HbSS disorders at birth.

Prenatal testing such as the use of DNA analysis where DNA is prepared from a biopsy of chorionic villi (it can be obtained at 8 – 12 wks gestation) and from amniotic fluid cell (it can also be examined at 16 weeks's gestation). Investigational attempts are ongoing to isolate fetal cells from maternal blood for DNA assay. There is the need for trained genetic counsellors in our country to deliver clinical genetic services. Scott et al. (1988) have carefully reviewed twelve programs that currently grant master's-level degree in genetic counselling in the United States and Canada. They also reviewed the development and current status of training opportunities of genetic counsellors. In view of the above, the Federal Government of Nigeria should make funds available for the training of counsellors.

Nevertheless, raising public education awareness of the hereditary nature of sickle cell disease and genetic testing/ counselling should be made regularly through teaching in schools, religious centres etc. This will aid both parents and prospective couple's access to information about child bearing risks and in the diagnosis of sickle cell disease. Genetic services offer a range of diagnostic and genetic counseling services for people with, or at risk of genetic conditions or susceptibilities (of family histories of sickle cell disease) The establishment of genetic units in primary health centres all over the country in order to ensure adequate services for genetic counselling is of great importance. It will create opportunity for counselling to be done in a psychologically balanced environment. Also, effort should be put in continued research on the frequency of blood genotypes randomly to identify sickle cell traits (risk couples) in the population. This is quite necessary to monitor its onward transmission in future generations. Hindrances to genetic counseling may arise from illiteracy, improper history record and polygamy. It is possible to take detailed family histories and provide genetic counseling advice in primary care with minimal training of clinical staff (Rose, 1999).

The correct dissemination of knowledge is an important step towards the eradication of genetic disorders in the Nigerian populations. We advocate the implementation of genetic counseling with the primary goal of counseling, which is to inform and educate counselees about risk of sickle cell disease and risk management.

Limitations of study

Some of the hospitals included in this study could not provide data on the reported cases of sickle cell anemia because of improper record keeping. Therefore, the frequency provided here might not be an exact representative number for this period of investigation. Most individual subjects were mainly within ages 16 - 24. The results should therefore only be generalized with caution to this age group. The level of awareness about genetic

counseling was carried out using a small sample size and cannot be generalized for the large population.

Conclusion

Genetic counseling is one of the fundamental means of eliminating sickle cell disease in Nigeria and Africa at large if only a national program is implemented for the prevention of sickle cell anemia. Such program will ensure that every individual knows his/her hemoglobin genotype before getting to the child bearing age, and that hospitals carry out compulsory prenatal diagnosis and diagnosis at birth properly. Its effective advocacy must be emphasized.

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