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Prenatal Diagnosis in Sickle Cell Disease: In the Eyes of the Couple at Risk

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Author's contribution

The sole author designed, analysed, interpreted and prepared the manuscript.

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ABSTRACT

Background: Prenatal diagnosis of sickle cell disorders provides a couple at risk the opportunity to make informed decisions whether or not to birth a child with Sickle Cell Disease (SCD).
Aim: To explore the knowledge of prenatal diagnosis and its acceptability among parents at risk of having another child with SCD
Methods: A self-administered structured questionnaire was used to obtain information on socio-demographics, knowledge and attitude towards prenatal diagnosis. The respondents were parents of SCD patients seen at the haematology consultant paediatric clinic of the University of Port Harcourt Teaching hospital. Data were analysed using SPSS version 20.0.
Results: Forty-six parents were interviewed and they were all Christians. Thirty-two (69.6%) were females and fourteen (30.4%) were males. All the respondents were from the south-south region of Nigeria. Four (8.7%) had lost children from complications of SCD. Twenty-four (52.2%), had heard about prenatal diagnosis of sickle cell disease while twelve (50%) of them heard about it from health care professionals. Four (8.7%) had done prenatal diagnosis in previous pregnancies while 32 (69.6%) were willing to do it in their next pregnancy. The most common reasons given for not doing prenatal diagnosis were religious beliefs, personal beliefs and fear of the procedure.
Conclusion: There is still a gap in knowledge and utilization of prenatal diagnosis by at risk couples. Appropriate information and regular counselling should be given to at risk parents as a key method of preventing SCD.

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Keywords: Prenatal diagnosis; sickle cell disease; pregnancy; knowledge and utilization.

1. INTRODUCTION

Sickle cell disease (SCD) is one of the most common genetic diseases worldwide as reported by the World Health Organisation (WHO) [1]. Nearly 300,000 infants are born in Africa each year with major haemoglobin disorders [1]. The high burden in this region is attributed to the survival advantage conferred by sickle cell trait against Plasmodium falciparum [1]. Despite this high burden, prenatal diagnosis and newborn screening rates in Nigeria are low. Prevention of SCD through carrier identification, genetic counselling, and prenatal diagnosis (PD) plays a significant role in reducing the impact of the disease and allows better use of available resources for the existing patient populations [2].

Prenatal diagnosis refers to the use of techniques to detect the presence or absence of foetal abnormalities [3,4,5]. In the context of sickle cell disease, prenatal diagnosis will detect the haemoglobin phenotype of the foetus, hence giving the couple room to prepare themselves for the birth of the child or terminate the pregnancy. [3] A couple at risk refers to a couple who are both healthy carriers of sickle cell trait (HbAS), one has the sickle cell trait (HbAS) and the other Haemoglobin C trait (HbAC) or one has sickle cell anaemia (HbSS) and the other sickle cell trait (HbAS) [6]. Prenatal testing should be preceded by genetic and psychological counselling done by a medical expert [7]. The at-risk couple should be given accurate and comprehensive information for informed decision making. This information should include indications for testing and the risks, benefits, and limitations of the prenatal test in detail and in a language understandable by the patient [8]. Prenatal diagnosis for sickle cell disease can be done through two major techniques, chorionic villus sampling (CVS) and amniocentesis [2]. Though CVS is the method of choice, sometimes, at-risk couples may be identified late in the 2nd trimester and so, can still be offered amniocentesis [2].

Chorionic villus sampling is a relatively safe procedure that involves obtaining foetal tissue between 8 and 12 weeks of gestation under ultrasound guidance, to access foetal DNA [7]. The sample is analysed by mutation detection methods [7]. A transabdominal or transcervical/ vaginal route can be used depending on individual patient peculiarities, the location of the placenta and the choice of the obstetrician [8,9]. Amniocentesis is done at 16 weeks gestation by harvesting amniotic fluid cells from 20-30 mls of amniotic fluid aspirated from the amniotic cavity trans abdominally using a 20-22 gauge spinal needle under anaesthesia [8]. Both CVS and amniocentesis are minimally invasive and may cause some risk to the mother and foetus. There is a risk of a foetal miscarriage of 0.5-1.0% [2]. The costs for amniocentesis and CVS vary by geographic location, laboratory, and the specific tests done [8].

Newer methods of prenatal diagnosis are done by isolating foetal cells from maternal blood for DNA assay (non-invasive prenatal diagnosis) [10]. There is also a technique called cordocentesis for fetal blood sampling and DNA analysis at 18 to 19 weeks gestation [10]. Celocentesis is another technique where the celomic fluid is aspirated at 7-9 weeks gestation [10]. Pre-implantation diagnosis is another option especially for couples who would want to have a healthy baby but do not wish to terminate an affected pregnancy after prenatal diagnosis. Pre-implantation diagnosis involves the selection and use of unaffected embryos following *In vitro* fertilization [11]. The study aimed to explore the knowledge of prenatal diagnosis and its acceptability among parents at risk of having another child with SCD. Information from this research will aid the counselling of at-risk couples, in terms of addressing misconceptions and dispelling their fears with the overall purpose of improving the prenatal diagnosis rates among this population group.

2. METHODOLOGY

This was a hospital-based cross-sectional study done over three months (October 2018 to December 2018). The study population consisted of parents of children with sickle cell disease seen at the Haematology Clinic of the Department of Paediatrics, University of Port harcourt Teaching Hospital. The Paediatric Haematology Clinic caters for patients up to 18 years of age and runs every Thursday from 8 a.m. to 4 p.m. in the Department of Paediatrics. The patients are usually brought in by their parents or guardian. The clinic is run by two consultants and two senior registrars with the assistance of nurses.

All patient information was kept confidential and participants' identity during data collation was

completely anonymous. All individuals in the study population were given an equal chance to participate. Counselling on prenatal diagnosis was done for each participant after the filled questionnaire had been retrieved. The children of the few parents who declined participating were still given the full care at the outpatient clinic.

A self-administered structured questionnaire was used to obtain information on socio-demographics, knowledge and attitude towards prenatal diagnosis. The questionnaire was pretested among 5 parents at risk of having a child with SCD attending the paediatric haematology clinic with their children on an earlier date before the commencement of the study. Ambiguous words were replaced and typographical errors corrected. Data from the pretest were not included in the study. The data were analysed using SPSS version 20.0.

2.1 Study Instrument Reliability

The medical records of the children of 10 randomly selected study participants at the Haematology Clinic were reviewed to check the accuracy of their responses for state of origin and the number of children with SCD. These two questions were chosen because they represent significant medical detail that is likely to be accurately documented in the records. This reliability check found 100% concordance between the questionnaire responses and medical records.

Test-Retest reliability was assessed by administering the questionnaire twice to 10 randomly selected study participants (at least two weeks apart) who had returned with their children for their normal clinic follow up appointment during the study period. This reliability check also found 100% concordance between the two responses given.

3. RESULTS

A total of 50 questionnaires were distributed. 46 questionnaires were retrieved that were completely and correctly filled giving a response rate of 92%. Of these 46 respondents, there were 14 males and 32 females with an M: F ratio of 1:2. The largest age group represented was the 40-49 years age group (60.9%). All respondents were Christians. (Table 1).

The majority (69.3%) of respondents had just one child with sickle cell disease while 4 (8.7%) respondents had experienced the death of a child from complications of sickle cell disease.

Table 1. Socio-demographic characteristics of the respondents

Variables	Frequency(n=46)	Percent
Age group of parents		
20-29	4	8.7
30-39	12	26.1
40-49	28	60.9
50-59	2	4.3
60-69	0	0.0
Gender of parents		
Male	14	30.4
Female	32	69.6
Religion		
Christian	46	100.0
Level of education		
Primary	1	2.2
Secondary	7	15.2
Tertiary	38	82.6
State of origin		
Rivers	20	43.5
Akwa-Ibom	2	4.3
Delta	12	26.1
Edo	4	8.7
Cross Rivers	4	8.7
NR*	4	8.7

**NR- No Response*

About a half (52.2%) of respondents affirmed that they had heard about prenatal diagnosis and most of these persons got the information from health workers (50%), (Table 2).

Only four (8.7%) respondents had done prenatal diagnosis while 32 (69.6%) respondents would like to do prenatal diagnosis in pregnancy in future. The reasons given for not wanting to do prenatal diagnosis include personal beliefs (50%), religious reasons (33.3%) and fear of the procedure, (Table 3).

There was a statistically significant relationship between the number of children with sickle cell disease the parents have and their desire to do prenatal diagnosis, (Table 4).

Table 2. Knowledge of prenatal diagnosis of sickle cell anaemia of the respondents

Variable	Frequency (n=46)	Percent
How many children do you have/have you had with SCD		
One	32	69.6
Two	8	17.4
Three	6	13.0
Have you lost any child with sickle cell anaemia?		
Yes	4	8.7
No	42	91.3
Have you heard about prenatal diagnosis of sickle cell anaemia?		
Yes	24	52.2
No	22	47.8
If yes, from whom?		
Friends	8	33.4
Doctors/Nurses	12	50.0
Internet	2	8.3
NR*	2	8.3

NR-No response*

Table 3. Attitude towards prenatal diagnosis and practice of prenatal diagnosis of sickle cell anaemia of the respondents

Variable	Frequency (n=46)	Percent
Have you ever done prenatal diagnosis?		
Yes	4	8.7
No	42	91.3
Would you like to do prenatal diagnosis in your next pregnancy?		
Yes	32	69.6
No	10	21.7
Unsure	4	8.7
Reasons given for not wanting prenatal diagnosis(n=12)		
Religious reasons	4	33.3
Personal beliefs	6	50.0
Fear of the procedure	2	16.6

Table 4. Relationship between the number of children with sickle cell anaemia and the attitude towards prenatal diagnosis of sickle cell anaemia

	Number of children with sickle cell anaemia			Fishers exact test
	One(n%)	Two(n%)	Three(n%)	
Would you like to do prenatal diagnosis?				0.116
Yes	22(68.8)	6(18.8)	4(12.5)	
No	10(100.0)	0(0.0)	0(0.0)	
Unsure	0(0.0)	2(50.0)	2(50.0)	

4. DISCUSSION

Prenatal diagnosis is a very useful innovation and plays a key role in the preventive measures for sickle cell disease. Its benefit, however, is yet to be fully explored by many couples at risk of birthing a child with SCD. This study set out to explore the knowledge of prenatal diagnosis and how acceptable it is to at-risk couples.

In this present study, the sample population used consisted of parents who already have an affected child who accompanied their child/children to attend the sickle cell well-clinic. The respondents in this study were all Christians. This may be due to Christianity being the predominant religion in the South-south region of Nigeria, where this study took place. There were two times more females than males in this study. This can be explained by the fact that in general, mothers take up the role of child rearing while the fathers focus on providing for the family. Hence mothers more commonly accompany their children to the hospital.

In this study, about half of the respondents had heard about prenatal diagnosis and amongst these informed parents, half of them got their information from health workers. Since this study was carried out within the hospital, it is understandable that all of the respondents have come in contact with health care workers and may have had some counselling for SCD. While up to half of the study participants had heard about prenatal diagnosis in this present study, only 4 out of 46 respondents (8.7%) had ever done prenatal diagnosis. Also, 69.7% of respondents in this present study would like to do prenatal diagnosis in subsequent pregnancies and this was significantly associated with the number of affected children the respondent had. This is similar to the findings in a study done among adults in Saudi Arabia [12] where 68% of adults studied would consider prenatal diagnosis in future and this practise was significantly associated with their level of education, previous pregnancy with an affected baby and having prior knowledge of prenatal diagnosis.

The acceptance of prenatal diagnosis in this present study is, however, lower than what was reported in another study done among health care professionals and medical students in Saudi Arabia, [13] where 82.1% of the participants showed a consistent trend of accepting and encouraging PND when appropriate. This better acceptance of PND in this group maybe because they were health care professionals and medical students with better knowledge and understanding of the usefulness of PND as compared with the parents in this study [13,14].

The reasons given for deciding not to do prenatal diagnosis in this present study were religious beliefs, fear of the procedure and personal beliefs. Regarding religious beliefs, some people believe that 'fate' or 'God's will' will determine whether or not they have children with sickle cell anaemia [15]. Besides, because of religious beliefs, some at-risk parents may be unwilling to interfere with or terminate a pregnancy following a prenatal diagnosis of sickle cell disease as they may equate it with the crime of 'killing' an individual. All the participants in this present study were Christians and this religion preaches the sanctity of life as one of the ten commandments written in the bible are 'Thou shalt not kill'. Religion plays a significant role in the health choices of individuals especially in our country Nigeria [16]. Religion reflects a people's belief in God and the spiritual explanations of the human place in the world [16]. Religion is particularly important to families affected by SCD as they tend to use their faith as a coping mechanism for the psychosocial burden that living with SCD poses [17]. Many religions suggest that children are a gift from God and shouldn't be a matter of choice. Parents should accept the health condition of children just the way they are and depend on God for the sustenance of life. In a study by Alysse et al, [18] cultural and religious beliefs, as well as the high cost of the procedure, were reported as reasons for declining prenatal diagnosis.

In this present study, 16.6% of respondents who are not willing to do PND expressed a fear of the procedure as a reason for not opting for PND. The procedure of PND (via chorionic villus sampling or amniocentesis) is invasive and carries a risk of pregnancy loss. The risk of miscarriage is worse with CVS than with amniocentesis and CVS is the procedure more commonly done for PND in Nigeria as it can be done earlier in pregnancy than amniocentesis. Furthermore, most parents will still not want to proceed to terminate a pregnancy following the detection of SCD, hence PND is seen as being of no particular use. The decision to end a pregnancy following a prenatal diagnosis of SCD has an ethical twist to it, is often difficult and involves religious, psychosocial and cultural considerations [19]. Another influence on the decision for or against PND may be that if one

should weigh the benefit of PND and possible avoidance of the birth of a child with SCD and coping with the life challenges of having a child with SCA, in the face of SCA presently becoming a more 'manageable' disease, the latter may not be as terrible an option [19].

Personal beliefs were also given as reasons for not wanting to do prenatal diagnosis in this present study. Personal beliefs include all the many reasons why an individual may decide not to do the procedure. They range from no particular reason at all to negative perception resulting from knowledge of the negative experiences of others, etc. This is similar to the findings in a study in India where it was reported that the couples in the study were reluctant to undergo prenatal diagnosis and were ready to accept a child born with sickle cell disease instead [9]. Also, only 25 % of at-risk parents in another Indian study presented for prenatal diagnosis [3].

Other factors which may influence the decision of at-risk couples to do prenatal diagnosis include the high cost of the procedure and access to a facility. Presently, in Nigeria, the facility for amniocentesis and chorionic villus sampling are available only in very few centres in major cities like Lagos, Port-Harcourt and perhaps Abuja. The cost of the procedure is also in hundreds of thousands, which may be above the budget of many at-risk couples. Surprisingly, high cost and limited access to a facility were not listed by the respondents in this present study.

Though this study provides key insight into the awareness of prenatal diagnosis and reasons for not doing prenatal diagnosis among at-risk couples, it did not explore the attitude of the participants towards abortion following an unfavourable prenatal diagnosis. Another drawback of this study is that about a third of the participants were fathers. Some fathers may not be able to give factual details of procedures done in pregnancy.

5. CONCLUSION

There is still a gap in knowledge and utilization of prenatal diagnosis by at risk couples. Health workers handling patients with SCD or at-risk couples, therefore, should regularly update their knowledge of current trends in the prevention and management of SCD to give their clients adequate information. Furthermore, more effort needs to be put into information dissemination with enlightenment campaigns done within the community to compliment the spread of information done within the hospital setting by health workers. These will go a long way to prevent sickle cell disease.

CONSENT

Informed and written Consent was obtained from the parents before the study tool was administered.

ETHICAL APPROVAL

It is not applicable.

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COMPETING INTERESTS

Author has declared that no competing interests exist.

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