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Original article

Haemoglobin Phenotype of Newborn at Asaba Specialist Hospital, Delta State Nigeria: The Need for Newborn Screening

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Abstract

Background: Sickle cell disease (SCD) is an inherited autosomal recessive haemoglobin disorder that contribute significantly to the morbidity and mortality of children especially in Nigeria. Despite the high morbidity and mortality associated with SCD in Nigeria, early detection through newborn screening is not readily available. This study is aimed at documenting the different pattern of haemoglobin phenotype among the neonate following institutionalization of newborn screening for SCD. ***Subjects and methods:*** A prospective study involving eligible newborn babies seen in Asaba Specialist Hospital between January 2021 and December 2023. Venous blood was collected from the babies into an ethylenediaminetetraacetic acid sample bottles. The samples were analyzed using high-performance liquid chromatography (HPLC) techniques, and the haemoglobin phenotypes obtained were documented. Data were analyzed using the Statistical Package for Social Sciences (SPSS) version 23. ***Results:*** A total of 5,103 neonates were recruited during the period under review. Majority (3297; 79.1%) of the subject had FA (normal Phenotype) while 801 (19.2%), 45 (1.0%), 19 (0.4%), 7 (0.2%), 1 (0.02%) had haemoglobin phenotype FAS, FS, FAD, FAC and FAE respectively. ***Conclusion:*** There is presence of wide spectrum of abnormal haemoglobin phenotype among the subjects. We recommend that newborn screening for sickle cell disease should routinely done for all neonate in Nigeria.

Keywords: ***Newborn, Haemoglobin, Genotype, Sickle-cell.***

Introduction

Sickle Cell Disease (SCD) is a group of inherited red blood cell disorders caused by the inheritance of two abnormal haemoglobin genes, one of which is haemoglobin S (Hb S). This abnormality results in the production of abnormal (sickle) shaped red blood cells and chronic haemolytic anaemia [1,2]. Sickle cell anaemia (Hb SS) is the most severe form of sickle cell disease [3]. It occurs following inheritance of two genes that code for haemoglobin S. Other SCD phenotype include Hb SC, Hb Thalassemia, Hb SD, Hb SE and Hb SO. The sickle cell trait (Hb AS) occurs with the inheritance of one copy of the sickle gene and one normal gene and it is often asymptomatic [1].

Globally, over 300,000 babies with SCD are born annually, with about 75%-85% of these children in Sub-Sahara Africa [4]. In Nigeria, about 150,000 children are born annually with haemoglobinopaty which amount to about 33% of global annual birth [5,6]. Furthermore, 2%-3% of the total newborn population in Nigeria are Hb SS while 24.7% are carriers [7,8]. It is estimated that 8%-16% of children Under-5 dies from haemoglobinopathies and its complications in Sub-Saharan Africa with death occurring before the fifth birthday in 50%-90% of undiagnosed children who suffer sickle cell anaemia [8-10]. Advances in the management of sickle cell disease in the United States of America (USA) have led to a decline in mortality from about 26% to approximately 1-2% in the first 18 years of life [11]. This achievement is possible due to institutionalization of universal newborn screening in all 50 states in the USA.

Newborn screening (NBS) program allow for early detection and intervention which help to prevent morbidity and mortality associated with the disease [12,13]. It has been estimated that widespread newborn screening and follow up care could save the lives of almost 10 million children by 2050 [14]. There is no national neonatal screening program for SCD in Nigeria, however few newborn screening programs have been established in some hospitals in the country in the last few years.

This study is aimed at identify the different haemoglobin phenotype patterns and ultimately to institutionalize NBS program for comprehensive SCD care among the populace.

Materials and Method

Study design

This is an observational, prospective study that screen newborn delivered-in or referred to Asaba Specialist Hospital, Asaba from January 2021 to December 2023.

Study site
The study was conducted at the Asaba Specialist Hospital (ASH). The hospital was established in 2019 as one of the state owned tertiary hospital. The hospital has a 20-bedded Sickle Cell Center (SCC) that is dedicated to the care of sickle cell patients. The sickle cell center treats an average of 300 patients per month and an annual admission rate of 200-250 patients.

Subjects and method
In a bid to offer a holistic and comprehensive care to the SCA patient in the Sickle Cell Center of the hospital, new born screening (NBS) for haemoglobin phenotype was established in 2021. All the newborn delivered in our center had their cord blood collected and sent to the sickle cell center for analysis/screening while heel prick samples were collected by trained health care workers on Guthrie cards between days 1 to 7 after birth for babies whose cord blood samples were not available and for those babies delivered outside our hospital.

Sampling technique
All newborns who were delivered in Asaba Specialist Hospital or referred to the center and whose parents consented were consecutively recruited into the study within the study period. Any baby who has been transfused was excluded.

Haematological analysis
Screening for haemoglobinopathies was done by HPLC. The HPLC was done using the HPLC BIO-RAD VARIANT 11 haemoglobin testing system V2 (LB0002570, USA, 2010), according to the manufacturers protocol. At the end of each analysis, the result were printed out and read as percentages (%) on the chromatographic form. The values in percentage of haemoglobin phenotypes in each sample (chromatogram) were reported in order of the highest to that with the least value and the allocation of the phenotype for each sample was based on the haemoglobin with the highest percentage after the fetal haemoglobin.

Follow-up
Babies with SCD were followed up at the Sickle Cell Centre (SCC) periodically where early and comprehensive care were instituted.

Data analysis
Data was analyzed with the Statistical Package for the Social Sciences (SPSS) version 23.0. Frequencies, proportions, and percentages were computed for categorical variables.

Ethical consideration
Ethical approval was gotten from Ethical Committee of Asaba Specialist Hospital, Asaba Delta State, Nigeria. Informed consent was gotten from parents of each baby.

Results

Table 1 shows the distribution of haemoglobin phenotype of the subjects. A total of 5,103 neonates were recruited for the study. Majority 3297(79.1%) of the subject had FA (normal Phenotype) while 801 (19.2%), 45 (1.0%), 19 (0.4%), 7 (0.2%), 1 (0.02%) were FAS, FS, FAD, FAC and FAE respectively.

Table 1: Distribution of haemoglobin phenotype

Haemoglobin phenotype (Hb)	Frequency (n =5,103), n (%)
FAA	3297 (79.0)
FAS	801 (19.2)
FAC	7 (0.2)
FAD	19 (0.4)
FAE	1 (0.02)
FSS	45 (1.0)

Figure I showed that 3297 (79.0%), had normal haemoglobin phenotype (FA), 828 (20%), had trait (FAS, FAC, FAD, FAE), while 45 (1.0%), had haemoglobin phenotype SS.

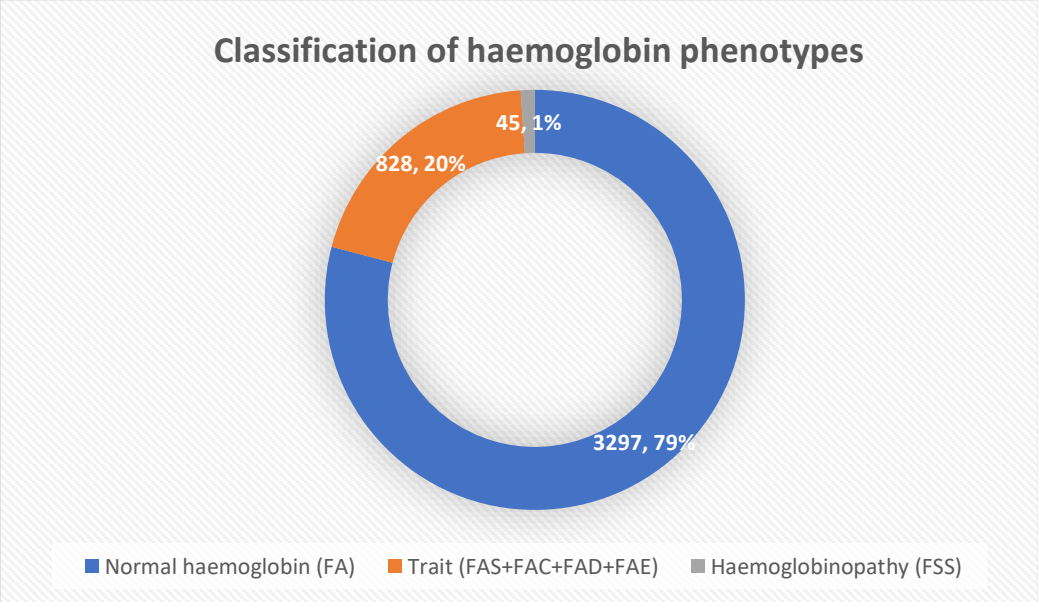

Figure 1: shows the distribution of normal hemoglobin, trait and haemoglobinopaty.

Discussions

This study of haemoglobin phenotype pattern of neonates seen at the Sickle cell Center of the Asaba Specialist Hospital, Asaba reveals the presence of normal as well as abnormal haemoglobin variants. The prevalence of the various phenotypes is as follows: FA (79.1%), FAS (19.2%), FS (1.0%), FAD (0.4%), FAC (0.2%) and FAE (0.02%).

The prevalence rate of haemoglobin phenotype FA 3297(79.0%) in this study is similar to what has been documented in other parts of Nigeria where newborn screening has been carried out [7,15-18]. This shows that the prevalence of Hb FA has not really changed over the years and a significant proportion of children in Asaba has normal haemoglobin despite the presence of abnormal variants. This finding from this study is in contrast to what was reported by Ohene-Frempong *et al.* [19] in Ghana who reported a prevalence rate of 96.2% for haemoglobin phenotype FA while Tshilolo also reported a prevalence of 81.6% in Democratic Republic of Congo [20]. The high abnormal haemoglobin phenotype

in this study could explain the low prevalence of Hb FA in our study compared to other studies by Ohene-Frempong *et al.* [19] and Tshilolo *et al.* [20].

The prevalence rate of haemoglobin FS in this study 1.0% is similar to 1.2% that was reported in Folayan *et al.* [21] in Bida, Nigeria and 1.9% in Abuja [18]. The similarity in the method adopted in screening of this children could explain this observation as HPLC was adopted in both studies. However, this finding is much lower than what has been reported by Abdulrahaman in Sokoto, Nigeria who reported a prevalence of 4.75% for haemoglobin FS among the populace and this he attributed to the high consanguinity rate in the area [22]. It has been reported that Africa has a sickle cell disease prevalence range of 1.2% to 1.5% in contrast to 0.01% to 0.62% prevalence in Europe and other part of America. This finding is possibly due to the high prevalence of malaria transmission in Sub-Saharan African.

The prevalence rate of FAS (19.2%) in this study is similar to what has been reported in other parts of Nigeria [18,21,22]. This finding is sharp contrast to what has been documented in Europe and America. The role of malaria in perpetuating the sickle cell carrier state could explain the high prevalence of FAS among the Nigerian subjects. The prevalence rate of FAD in this study is 0.5%, which is similar to 0.4% documented by Folayan *et al.* [21] Haemoglobin FAD appears to be rare phenotype in Nigeria. The prevalence of 0.2% for FAC in this study is low when compared to what has been documented earlier by other researchers. A prevalence of 1.3% was reported among neonates in Abuja, Nigeria [18] while 1.6% was also reported among neonates in Bida, Nigeria [21]. Tribe, ethnicity and other demographic variables might explain obvious difference observed.

Conclusion

The institutionalization of newborn screening for sickle cell has demonstrated a wide spectrum of haemoglobin phenotypes among the subject. This has afforded us opportunity for early initiation of comprehensive SCD care. It has also supported the call for routine neonatal screening for SCD in Nigeria.

Limitation

This is a hospital-based study and may not truly reflect the true prevalence of various haemoglobin phenotype in the general population.

Declarations

Financial support and sponsorship

None

Conflicts of Interest

None

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