Original Article

Prevalence of haemoglobin variants among the Ika ethnic nationality of Delta state

Adu E.M*, Isibor C.N, Ezie E

Department of Medical Laboratory Services, Antiretroviral Therapy Centre, Central Hospital, Agbor, Delta State.

*Corresponding author: adumatthew10@yahoo.com

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ABSTRACT

Background: Haemoglobin genotype is an important blood component that determines haemoglobinopathies. Distribution of haemoglobin variants was investigated among the Ika ethnic nationality of Delta State, Nigeria. Aim: The present study was conducted to determine the prevalence of haemoglobin variants and also to provide information for instituting genetic counseling services to reduce haemoglobinopathies between couples. Methods: A total of 600 subjects were screened for haemoglobin genotypes using paper electrophoresis. Results: The prevalence of HbAA, HbAS, HbAC and HbSS were 78%, 19.5%, 0.5% and 2% respectively. There was no relationship (P>0.05) between sex and haemoglobin variants. There was a low prevalence of abnormal haemoglobin variants observed which is consistent with previous studies. Conclusion: The sickle cell trait (HbAS) is the largest variant in this population; we therefore advocate mandatory haemoglobin genotype screening for all intending couples in order to reduce the sickling gene pool.

Keywords: Haemoglobin variant, haemoglobinopathies, prevalence, genotype, deoxyribonucleic acid, electrophoresis

INTRODUCTION

Haemoglobin genotype is an important blood component that determines haemoglobinopathies.[1] Haemoglobin is the oxygen carrying pigment of red blood cells, defects in its genes can produce abnormal haemoglobin which leads to condition known as haemoglobinopathies.[2]

Haemoglobinopathies are conditions caused by qualitative structural abnormalities of the globin polypeptide chains that result from alteration of the deoxyribonucleic acid (DNA)
genetic code for the chains.\(^2\) Haemoglobin genotype includes normal haemoglobin (HbAA) and other abnormal ones like haemoglobin S (HbS) which is a variant form of haemoglobin.\(^1\) It differs from normal haemoglobin (HbA) by the substitution of valine, a neutral amino acid for glutamic acid at position 6 in the β-chain which causes a change in the properties of haemoglobin that result in sickling of red blood cells.\(^1\) Another variant is haemoglobin C (HbC) which results from the substitution of lysine a basic amino acid for glutamic acid at position 6 in the β-chain.\(^1\) Various studies have been carried out on haemoglobin variant in the Niger Delta region\(^3,4\) but there are few published data in Delta state. Due to the paucity of data in Delta state, we investigated the distribution and frequency of haemoglobin variants of the Ika ethnic nationality of Delta state, Nigeria.

**METHODOLOGY**

**Study population**
A total of 600 (132 males and 468 females) out-patients attendee in Central Hospital, Agbor, were recruited for this study. This Hospital serves the two Ika Local Government area (Ika South and Ika North East) and even beyond. Participants were duly informed about the investigation and their consent was given. Ethical clearance was obtained from the institution ethics committee.

**Sample collection and preparation**
2mls of venous blood was collected into EDTA container and inverted severally for proper mixing. About 1ml of the blood sample was washed three times using normal saline (0.85%NaCl) to remove plasma proteins. The washed cells were re-suspended with equal volume of normal saline. The red cell suspension was mixed with equal volume of distilled water to lyse the blood cell. The resulting lysate was used for haemoglobin genotype determination.\(^5\)

**Haemoglobin electrophorese**
The standard method described by Evans\(^6\) was used for haemoglobin electrophoresis. A small quantity of haemolyste of venous blood from each of the subjects was placed on a cellulose acetate membrane and carefully introduced into the electrophoretic tank containing Tris-EDTA-borate buffer at pH 8.6.\(^6\) Electrophoretic separation was then allowed to operate for 15-20minutes at an electromotive force (e.m.f) of 160v. The results were read immediately against known haemoglobin (AA, AS, AC, SC and SS) which serves as controls.

**Statistical analysis**
Statistical Package for Social Sciences (SPSS Version 10, Chicago IL) was used for data analyses. Frequency distribution and percentage prevalence scores of the various parameters were generated.

**RESULTS**
The overall frequencies of the various haemoglobin genotypes in this study are shown in table 1. Out of the 600 subjects screened, 468 (78%) were HbAA, 117 (19.5%) were HbAS, 12(2%) were HbSS and 3(0.5%) were HbAC. Of the 600 people tested, 132 (22%) were males while 468(78%) were females.

<table>
<thead>
<tr>
<th></th>
<th>HbAA (%)</th>
<th>HbAS (%)</th>
<th>HbAC (%)</th>
<th>HbSS (%)</th>
<th>TOTAL (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>MALES</td>
<td>102 (21.8)</td>
<td>30 (25.6)</td>
<td>0 (0)</td>
<td>0 (0)</td>
<td>132 (22%)</td>
</tr>
<tr>
<td>FEMALES</td>
<td>366 (78.2)</td>
<td>87 (74.4)</td>
<td>3 (100)</td>
<td>12 (100)</td>
<td>468 (78%)</td>
</tr>
<tr>
<td>TOTAL</td>
<td>468 (100)</td>
<td>117 (100)</td>
<td>3 (100)</td>
<td>12 (100)</td>
<td>600 (100%)</td>
</tr>
</tbody>
</table>

Figure 1 shows the general distribution of the various haemoglobin genotypes among males and females subjects

**DISCUSSION**
Haemoglobin genotypes and blood groups are all inherited blood characters. The
Inherited disorders of haemoglobin are the most common gene disorders with 7% of the world population being carriers.\(^\text{[7]}\) The prevalence of HbSS among black population in the United States was reported to be 9% and 30%-40% generally for Africans.\(^\text{[6,9]}\) In another report, the geographical distribution of Sickle cell anemia (SS) was given as 3-9% for USA black Americans, 1-8% for US whites, 3-7% for Europe (UK, Pakistanis-blacks), 2-8% for other European countries (Mediterranean), 1-3% for Caribbean, 1-3% for Middle East, 1-10% for Africa.\(^\text{[10]}\) The frequency of the sickle cell trait (AS) was also reported as 8-16% for USA blacks, 8-10% for USA whites, 6-15% for Europe (UK, Pakistanis-blacks), 1-15% for Europe (Mediterranean), 3-85 for Caribbean, 7-85 for Middle East, 15-30.5% for Africa and 40.5% for West Africa and Nigeria.\(^\text{[10]}\)

![Haemoglobin Variants](Image)

**Figure 1:** A Bar chart showing the distribution of the various haemoglobin genotypes among males and females

In our study, the frequency of HbAA was 78% while HbAS was found to be 19.5%. This is in line with the findings of Jeremiah that revealed a frequency of 80.2% for HbAA and 19.68% for HbAS in the Niger Delta area.\(^\text{[8]}\) Esan *et al.*\(^\text{[1]}\) who reported 72.8% for HbAA and 23.5% for HbAS in Ekiti State. Akhigbe and co-workers\(^\text{[11]}\) reported a prevalence of 71.03% for HbAA, 22.19% for HbAS, in Ogbomoso South West Nigeria. Also, the present study reveals a frequency of 2.0% for HbSS and 0.5% for HbAC. This is consistent with the reports of Egesie *et al.*\(^\text{[12]}\) and Nwafor and Banigo,\(^\text{[13]}\) who reported a frequency of 2% and 3% for HbSS respectively. However, this is in contrast with the report of Jeremiah\(^\text{[3]}\) that reported 0% for HbSS and HbAC. Esan *et al.*\(^\text{[11]}\) in their work reported 1.86% for HbAC and 0.69% for HbSS. The low prevalence of HbSS in this study can be attributed to increased awareness of the disease, improved socio-economic conditions and other environmental and genetic factor which have an overall effect on the sickling gene pool.\(^\text{[3,4]}\) Erhabor and his colleagues\(^\text{[4]}\) attributed this low prevalence to abnormal haemoglobin carrier screening as well as genetic counseling program for prevention of haemoglobin disorders.

The frequency of HbAS detected in this study (19.5%) is in accordance with previous studies in Nigeria and other African setting which observed a prevalence of 20-40% in Nigeria and Africa in general.\(^\text{[10,14]}\) HbAS has been thought to offer some protective role against Plasmodium falciparum malaria.\(^\text{[15]}\) Adu *et al.*\(^\text{[16]}\) attributed this to the presence of large amount of acute phase reactants and 𝛽-globulin in their plasma which confers immunoregulatory mechanism on the HbAS individuals. Akhigbe and his co-workers in their work attributed this to the substitution of hydrophilic amino acid glutamate with hydrophobic amino acid valine (as in HbSS) or lysine (as in HbC) causing increasing binding affinity between haemoglobin molecules with polymerization of haemoglobin deforming red blood cells which rapidly cleared from the circulation.\(^\text{[17]}\)

The frequency of HbAC in this study is 0.5% which is in agreement with previous work that reported a prevalence of 2% for HbAC and 4% for HbSC in the Niger Delta area.\(^\text{[12]}\) Also a study of 204 individuals of black ethnic background in Spain reported 3.4% prevalence for HbAC and 0.5% for HbCC.\(^\text{[18]}\)
We observed a gender-associated risk for haemoglobin S trait (HbAS) and HbSS of 74.4% and 100% for females as against 25.6% and 0% for males. Also the females have 100% for HbAC gene as against 0% for males. Though the reason for this female gender susceptibility for abnormal haemoglobin gene is unknown but calls for increased effort on the part of both parent and health workers to sensitize the public especially the female folk on the need to undergo genetic counseling before marriage.

CONCLUSION

The knowledge of the various abnormal haemoglobin variants is vital in the safe and effective transfusion services in this locality as well as in civic registration and forensic medicine. It will also be useful in genetic counseling of prospective couples to make informed decisions aimed at reducing the sickling gene pool of this ethnic nationality and in Nigeria. We therefore recommend a mandatory genetic counseling and screening of all intending couples in order to avert the sickling gene pool in our population.

REFERENCES


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Conflict of Interest: None declared