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Research Article

Gene Frequencies of Haemoglobin Variants in Amorji Community of Enugu State, South East, Nigeria

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Abstract: Nine hundred and seventy three randomly selected volunteer residents of Amorji Nike community, in Enugu East Local Government area of Enugu State, Nigeria were screened for their haemoglobin genotypes. Six haemoglobin genotypes exist in the community. Persons homozygous for normal adult haemoglobin, *HbAA* were highest in haemoglobin frequency (64.3%) followed by the heterozygotes *HbAs* (24.8%), and *HbAc* (6.8%). Sicklers gave percentage frequencies of 0.7%, 1.1%, and 2.3%. For Hbcc, and Hbss respectively.

Key Words: normal and abnormal haemoglobins, sicklers.

INTRODUCTION

There are normal and abnormal haemoglobins. The normal haemoglobin, HbA is the predominant form in adults and consists of two alpha protein chains and two beta protein chains (Steinberg, 2001) Foetal haemoglobin, HbF, is the primary haemoglobin produced by the foetus during pregnancy. It also has two alpha and two gamma protein chains. It usually disappears after about six to nine months of delivery to be replaced by the adult type, HbA. (Steinberg, 2001).

The inheritance of abnormal haemoglobins (Hbs, Hbc, Hbd, Hbi, Hbm, Hbe, etc) which may occur due to gene mutation is the cause of sickle cell disease. (Okafor, 2000) For instance, Glutamic acid of HbA is replaced by valine in the beta chain of the globin molecule of Hbs (Ingram, 1957). Other abnormal haemoglobins are formed in the same manner; an amino acid being inserted at the wrong spot (point mutation) within the globin chain. Over 400 different kinds of mutant haemoglobins have been discovered in humans. (Alimba *et al*, 2010) An abnormal haemoglobin occurs in about one out of every 10,000 persons and is usually detected by electrophoretic methods (Weatherall and Clegg, 2001). A sickler inherits one copy of each abnormal haemoglobin gene from each parent. Thus, the sickle cell disease can manifest in the form of Hbss, Hbcc, Hbsc, Hbsd, Hbse, Hbee, Hbdd, Hbsi, Hbsm, Hbsg, Hbmm, Hbdd, Hbgg, Hbic etc and such similar combinations; these are all sicklers. When one inherits a normal haemoglobin, HbA from one parent and the abnormal type from the other parent, the person is a sickler cell carrier. (HbAs, HbAc or HbAi etc).

The sickler (e.g Hbss) is often anaemic, at any slightest stressful situation. This is because the life span of his sickled erythrocytes is usually less than 10 to 20 days, unlike the life span of a normal erythrocyte which is about 120 days. (Guyton and Hall, 2011). This means that when the sickler is exposed to a stressful situation like prolonged exercise, cold weather, pregnancy, malaria, malnutrition, injury, infections, dehydration, air travel etc, his abnormal, haemoglobin molecules polymerise to form 'tactoids' thereby making the erythrocytes to assume a peculiar sickle shape. The sickled cells block the blood vessels, which reduces the supply of oxygen to other erythrocytes, so that more erythrocytes become sickled, thereby causing pain, impair circulation, and decrease the oxygen–carrying capacity of the erythrocytes, making the latter's life span to be shortened (Platt *et al*, 1991; Ekeke, 2001). In essence, the sickler is often anaemic. His haemoglobin level may be reduced from the normal 15g/dl to about 6 to 8mg/dl or even less. The 'crisis' situation in a sickler is a combination of ***serious anaemia***, ***much pain*** (especially at the joints of the hands, abdomen, legs, back) and ***acute ill health*** (Steinberg, 2001).

The heterozygote of sickle cell disease (or the sickle cell carrier) HbAs or HbAc or HbAi etc might on certain occasions like air travel be slightly anaemic but has the health advantage of being resistant to malaria caused by *Plasmodium vivax* or *P. falciparum* (Maina *et al*, 2010*)* The homozygous normal haemoglobin HbAA has no resistance to *P. vivax* or *P. falciparum. (*Maina *et al*, 2010).

The gene frequencies of sickle cell genotypes vary within the black race. In Western Nigeria (a predominantly Yoruba ethnic group) about 50% of sicklers belong to the Hbss category (Lesi, 1991). In another survey carried out in Western Nigeria by Adeyemo and Soboyejo (2006), the percentage frequencies were: HbAA (70%), HbAs (26%), Hbss (1.3%), HbAc (1.3%), Hbsc (0.7%), and Hbcc (0.7%).

Since there is not much information on gene frequencies of sickle cell genotypes in many communities of Eastern Nigeria (a predominantly Igbo ethnic group) this study was undertaken. The present report seeks to determine more precisely the gene frequencies of sickle cell genotypes in Amorji-Nike community of Enugu State in South Eastern

Nigeria.

MATERIALS AND METHODS

Nine hundred and seventy three randomly selected volunteers who reside at Amorji –Nike in Enugu East Local Government area of Enugu State, Nigeria, were used for the survey. After ethical clearance from relevant authorities, 1.0ml of blood was collected from each subject and 0.5ml of it was mixed with 0.5ml 2% sodium metabisulphite solution on a clean glass slide and left for 25 mins. Sickling of the erythrocytes was indication of either a sickler or a sickle cell carrier. If there was no sickling of the erythrocytes, it was an indication of HbAA. The remaining 0.5ml blood sample was subjected to electrophoresis in Tris buffer solution for 25 mins at emf of 240 volts, using blood samples of known genotypes as control. This was a confirmatory test for HbAA, and various types of heterozygotes eg HbAs, HbAc etc as well as various types of sicklers (Hbss, Hbcc, Hbsc, Hbsi, Hbci, Hbsm etc.

RESULTS AND DISCUSSION

Normal adult haemoglobin, HbAA gave a percentage frequency of 64.3%; followed by the heterozygotes, HbAs and HbAc that gave 24.8 and 6.8% respectively. Sicklers were 2.3% (Hbss), 1.1% (Hbsc) and 0.7% (Hbcc) Fig.1.

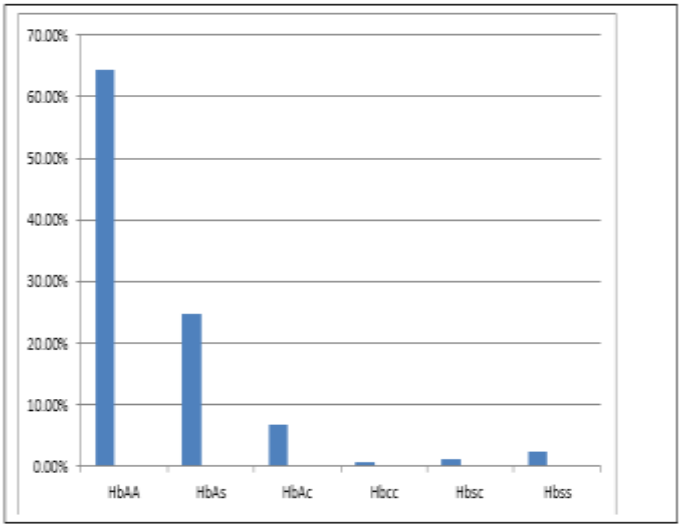

Fig. 1 Gene frequencies of Haemoglobin variants in Amorji-Nike Community of South-East – Nigeria.

However, Alimba et al (2010) carried out their own survey in a Western Nigeria population and got percentage frequencies of 76.6%, 20.4%, 2.0%, 0%, 0.4% and 0.7% for HbAA, HbAs, HbAc, Hbcc, Hbsc and Hbss respectively. Adeyemo and Soboyejo (2006) carried out their own studies at the University of Lagos, Lagos-Nigeria and reported percentage frequencies of 70%, 26%, 1.3%,1.3%, 0.7% and 0.7% for HbAA, HbAs, HbAc, Hbss, Hbsc and Hbcc respectively. All these reports, as well as those of others (Bakare et al, 2006, Egesie *et al*, 2008) seem to be in accordance with the estimate made by Fleming and Lehman, (1982) as well as by Nwafor and Banigo (2001) that in an average black population, HbAA ought to be between 55 and 75% of the population. In many communities within the black race, most available informations point out that HbAA has always been of highest occurrence when compared to other haemoglobin variants (Bakare, *et al*, 2006; Okafor *et al*, 2009). In any case, the report of Alimba *et al*, (2010) was very slightly higher than the 55 to 75% estimate.

In Kenya, East Africa, HbAA frequency ranged from 74 to 97%, also much higher than the estimated range (Moormann *et al*, 2003).

Sick cell carriers in this study was found to be 31.6%. Alimba *et al* (2010) got 22.4% while Adeyemo and Soboyejo reported 27.3%. Based on these reports as well as those of others (Zaccheaus, 2006) it could thus be estimated that the percentage frequency for sickle cell carriers in a black African population ought to be within the range of 20 to 35%.

It is easy to understand why HbAA is commonest in most black populations. Genetic inheritance of both normal and abnormal haemoglobins is dependent on the sexual union of parents. If both parents were free of abnormal haemogobin, offspring would equally be free. Prospective couples are hereby advised to go for genetic screening before marriage. This would help to produce a sickle cell disease free, generation. It is estimated that 7% of the World's population are sickle cell carriers (Weatheral, 2001).

It is on record also that about 300,000 children are born with sickle cell disease annually worldwide. (Okpala et al, 2002).

Sicklers should take time to find out the chemicals used in the preservation of some of the foods and drinks they consume. For instance, wines preserved with sodium metabisulphite, a generally used food/drink preservative, causes almost instant sickling of erythrocytes of sicklers. Sicklers are often faced with the risk of alloimmunization from allogenic blood transfusion. Despite the complications associated with blood transfusion, it is still used as a life-saving and prophylactic approach in handling sickle cell anaemia. However, it should be sparingly applied and limited to severe sickle cell anaemia cases of about <4.0gm/dl or during surgery. The sickler should avoid taking acidic types of soft drinks, or those that have been heavily gassed. Excessive alcohol consumption is not appropriate for sicklers due to the excess heat energy it would generate on an already entropic solution.

Since HbAA has a high frequency in Amorji –Nike community of South Eastern Nigeria and this gene resists neither *P vivax* nor *P falciparum* malaria, it is expected that malaria should be prevalent in Eastern Nigeria where so many species of *Anopheles* mosquitoes thrive (Conner *et al*, 1976).

The low frequencies of sicklers observed in this study(4.1%) and similar other studies (Lesi, 1991; Adeyemo and Soboyejo, 2006; Okafor *et al*, 2009) implies that the sickling gene pool is gradually reducing in the black population. It could be attributed to increased awareness of the disease through genetic counselling, socio-economic conditions and other environmental and genetic factors, which have an overall effects on the sickling gene pool. (Zaccheaus, 2006)

It is equally possible that the Hardy Weinberg equilibrium must have been disturbed, which led to more people acquiring normal haemoglobin (HbAA) gene and sickle cell trait (HbAs, HbAc, HbAo etc) while the homozygous sickle cell gene is gradually tending to zero (Zaccheaus, 2006).

The importance of knowledge of haemoglobin variants in a given population is a useful tool in marriage counselling, paternity disputes, medical diagnosis, medical statistics and genetic information.

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