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Evolutionary Relationship of Four Major Ethnic Populations in Nigeria Based on *Alu PV92* Insertion Polymorphism

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ABSTRACT Nigeria is a country located within the sub-Sahara region of Africa with four main geographical regions of diverse human population and ethnicity yet little is known about the evolutionary trait of these populations. As such, the *Alu* PV92 insertion polymorphism was used to depict the evolutionary and ancestral trait among the four main populations in Nigeria. Samples were obtained from 149 individuals from the four populations and DNA was extracted from their cheek cells. *Alu* PV92 DNA sequence was amplified by PCR and visualized on a 1.5 percent agarose gel for *Alu* insertion polymorphism. Among the 149 individuals, the frequency of *Alu* insertion (+) allele was 21 (7.05%) in the entire population and was predominant in the Ijaw-Ibibio population (4.36%). The Hardy-Weinberg equilibrium was not violated for the entire study population ($p>0.05$) suggesting that *Alu* polymorphism was responsible for the evolution of the population. The average heterozygosity (0.1191) and the Gst (0.0846) were relatively low compared to other populations predicting a low degree of interpopulation differentiation or diversity. Phylogenetic analysis showed the Ijaw-Ibibio population to exhibit the highest genetic distance from other populations suggesting Ijaw-Ibibio as the ancestral population. In conclusion, the four main populations of Nigeria were found to be closely related with a low level of genetic diversity except for the Ijaw-Ibibio population which showed the highest interpopulation differentiation and thus considered to be the ancestral population.

INTRODUCTION

Nigeria is a country located in the African continent within the sub-Sahara region at the Gulf of Guinea with a diverse human population and ethnicity. The earliest cultural population in Nigeria is believed to be the Nok people identifiable by their distinctive artifacts (Falola 1999). These skilled artisans were iron workers who habited a large area above the confluence of the Niger and Benue rivers between the fourth century B.C. and the second century A.D (Douglas 2004). Following the disappearance of the Nok people for over thousands of years, powerful kingdoms emerged within the region which formed the roots of some of the cultural groups existing in Nigeria today (Falola 1999). These early states induced the Yoruba kingdoms in Yoruba and the Edo Kingdom of Benin in South-West Nigeria, the Hausa Fulani cities and Nape. The earliest of the Nigerian Kingdoms, Kanem and Borno were located near Lake Chad in North-East Nigeria (Falola 1999). Today these kingdoms have segregated into four major geographical-ethnic populations principally the South-West (SW) region made up of the Yorubas, the South-East (SE) region dominated by the Igbos, the Northern (NO) region inhabited by individuals of Hausa-Fulani origins and the South-South (SS) region dominated by the Ijaw-Ibibios (Fig. 1). With this diversity, the evolution of Nigeria population still remains unclear.

Human genetic variations among individuals may be useful to give an insight and understanding of the evolution and migration patterns of populations. Generally, several genetic markers have been used to study population diversity and evolution. Genetic variants or polymorphisms that are found at loci or genes that code for expressible products are usually not very

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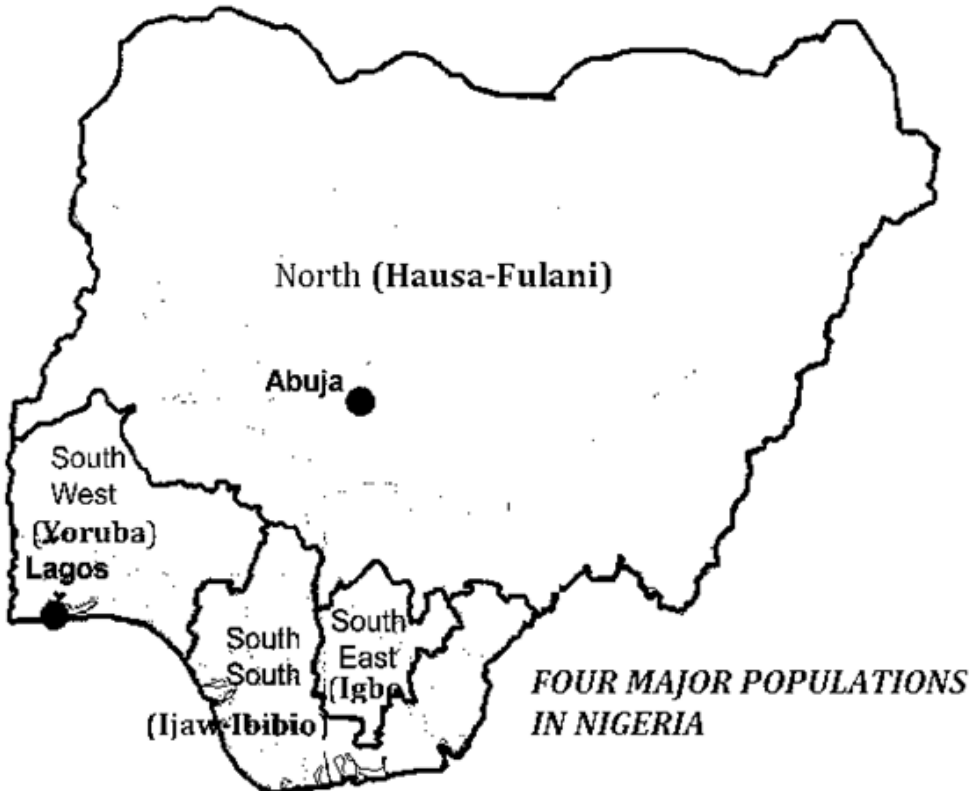

Fig. 1. Four major populations in Nigeria

suitable for evolutionary studies since they are usually internally regulated. On the other hand, allelic polymorphisms, especially those found in non-coding regions of the human genome are usually not internally regulated and thus, more likely to be evolutionarily neutral and suitable for genetic evolutionary studies. Several genetic insertion/deletion polymorphic markers have been identified in the non-coding region of the human genome among which is the *Alu* sequences.

The *Alu* family is one of the most successful mobile genetic elements with a copy number of over 50,000 within the human genome which is believed to have accumulated in approximately 65 million years of primate evolution (Ray et al. 2005; Schmid and Maraia 1992; Deininger and Batzer 1993). These *Alu* elements located in non-coding regions (intergenic spacers and introns) were originally identified about 45 years ago as a component in the human DNA renaturation curve which possessed the *Alu* I restriction sites (Tripathi et al. 2008). *Alu* elements are approximately 300 bp long followed by a tail of 20-30 adenosine bases and found to be ancestrally derived from 7SL RNA gene, which forms a part of the ribosome complex and mobilize through an RNA polymerase III–derived transcript in a process termed "retroposition" (Deininger 2011). Based on the mutation pattern of *Alu* sequences within the human genome, several sub-groups of related elements have been identified. One of such groups of *Alu* elements within the human genome is the human- specific (*HS*) group also known as the predicted variant (*PV*) group (Carrol et al. 2001; Batzer et al. 1990; Batzer and Deininger 1991).

The level of *Alu* transposition has changed over time, ranging from a single novel jump in every live birth, earlier in primate evolution, to about one in every 200 new-borns in present

days (Comas et al. 2001; Rowold and Herrera 2000). One or several of the *Alu* "chiefs" or "masters" are capable of jumping or transposing periodically (Micklos et al. 2013). Therefore, all primates that show an *Alu* insertion at a particular locus have inherited it from a common/shared ancestor. This is referred to as 'identity by descent' (Micklos et al. 2013). Because *Alu* elements can insert at a locus, copy itself for transposition and remain stable through evolutionary time from one generation to another, it has been considered suitable for population genetics and phylogenetic studies (Xing et al. 2005; Salem et al. 2003; Batzer and Deininger 2002). More so, since the distribution of these elements vary in geographically distinct human populations, it can serve as a useful genetic DNA marker to study human populations and their evolutionary relationship.

Objectives

This study was aimed to assess the evolutionary relationship of four major populations in Nigeria using *Alu PV92* insertion polymorphism.

MATERIAL AND METHODS

Sample Collection and DNA Isolation

Individuals from four major geographical regions of Nigeria were randomly recruited for the study after their consent was obtained. The cheek cells of the mouth were extracted with 0.9 percent saline solution whereby each participant gaggled saline solution without swallowing for 30 to 60 seconds. After the mouth wash, the resultant solution mixed with saliva was expelled into a 5 ml Agary non-vacuum plain sterile tube. DNA was extracted from the cheek cells using Bio-Rad *PV92* DNA extraction kit according to manufacturer's protocol.

PCR Amplification

PCR was performed with forward primer: 52 -AACTGGGAAAATTTGAAGAGAAAGT-32 and reverse primer: 52 -ATGGATGTAGTTGGTGTCATGGTCA-32 . The PCR cocktail contained 25 µl of one taq quick-load 2x master mix with standard buffer (New England Biolab (NEB), USA), 1 µl (10 µM) each of the forward and reverse primers (Inqaba-Biotech, South Africa), 3 µl of nuclease free water and 20 µl of genomic DNA to a final volume of 50 µL. The PCR amplification was done using a MiniJet thermal cycler (Biorad, USA) and started with a pre-denaturation step at 94°C for 2 min, followed by 40 cycles of denaturing step at 94°C for 30 sec, annealing at 60°C for 30 sec, and elongation at 72°C for 1 min with a final elongation step at 72°C for 5 min. The PCR amplicons were separated on 1.5% agarose gel electrophoresis at 100 volts for 30 min. The *PV92* homozygous (+/+), *PV92* homozygous (–/–) and *PV92* heterozygous (+/–) controls samples were amplified along with the participants' samples.

Data Analysis and Interpretation

The Hardy-Weinberg equation ($p^2 + 2pq + q^2 = 1$) was used to determine the expected genotypic frequencies (EF) and was compared with the observed genotypic frequencies (OF) for the various regional populations to test for Hardy-Weinberg equilibrium (HWE) using the chi-square (χ^2) test. Chi-square value > 5.991 from the critical table of χ^2 distribution with a degree of freedom of 2 was considered to be statistically significant ($p < 0.05$) and thus HWE was violated. Heterozygosity (*Het*) was calculated using 2pq from the Hardy-Weinberg equation and the average heterozygosity (*avg.Het*) determined, where p is the frequency of the *Alu* insertion (+) allele and q the frequency of the non-*Alu* insertion allele (-).

The G_{st} values, a measure of the relative magnitude of genetic differentiation among populations was calculated according to the equation of Nei (1987) as $G_{st} = \frac{nHet\text{-}avg.Het}{nHet}$, where $_nHet$ is the heterozygosity of the total population and *avg.Het* is the average heterozygosity.

The genetic distance was calculated as the differences of *Alu* insertion allelic frequencies between the various populations and was used to draw the phylogenic tree using the Minimum-evolution tree program of the Molecular Evolution Genetic Analysis (MEGA) software version 7.026.

To assess the relative amount of gene flow observed in the population, a plot of heterozygosity against distance from the centroid of each

population was done as described by Harpending and Ward (1982). Heterozygosity was the usual expected heterozygosity under HWE, and the distance from the centroid *ri* for a population *i* was determined using the equation $ri = \frac{(pi-p)^2}{(p)\ 91-p)}$ where *pi* and *P* are the frequencies of the *Alu* insertion in population *i* and total population respectively. Under an island model of population structure according to Harpending and Ward (1982), there should be a linear relationship between the theoretical heterozygosity and distance from the centroid. Theoretical heterozygosity *(hi)* was thus calculated as *hi* = *nHet* (1 - *ri*), where *hi* is the theoretical heterozygosity of population *i* and *nHet* is the expected heterozygosity of the total population. The outliers of this plot are of particular interest in this analysis such that plots above the theoretical heterozygosity are populations that have experienced more gene flow than average and those that fall below the theoretical heterozygosity are populations that have experienced less gene flow than average.

RESULTS

Genetic Variation within the Population

Samples were obtained from 149 individuals of the four ethnic populations (Hausa-Fulani, Yoruba, Ijaw-Ibibio and Igbo) in Nigeria and successfully amplified for *Alu PV92* gene (Fig. 2). The homozygous *Alu* insertion genotype (+/+) was present in 7 (4.70%) of the study participants, the heterozygous *Alu* genotype (+/-) was present in 7 (4.70%) of the individuals while 135 (90.60%) were homozygous with no *Alu* insertion genotype (-/-). Thus, the frequency of the *Alu* insertion allele (+) was 21 (7.05%) in the entire population and was predominant in the Ijaw-Ibibio population 13 (4.36%) (See Table 1).

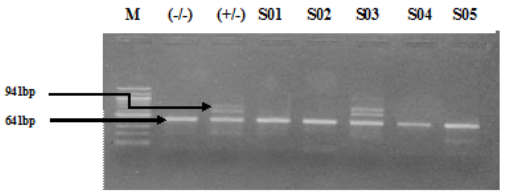

Fig. 2. Electrophoregram of the *Alu PV92* sequence amplification.
Legend: (-/-) indicates the control for the homozygous *Alu* non- insertion genotype while (+/-) indicates the control for heterozygous *Alu* genotype. S01, S02, S04 and S05 indicate samples which are homozygous for *Alu* non insertion genotype (-/-) while S03 is a heterozygous *Alu* genotype (+/-). **M** indicates 100bp molecular weight marker and **S** indicates sample

Evaluating the population for the HWE showed no significant differences ($p>0.05$) between the observed and expected genotype frequencies for the Hausa-Fulani, Yoruba and Ijaw-Ibibio populations indicating that Hardy-Weinberg equilibrium was not violated in any of these three populations. However, the HWE was violated ($p<0.025$) for the Igbo population. Moreover, the HWE was not violated ($p>0.05$) in the entire population (See Table 2). The heterozygosity was fairly different among the four populations ranging from a low of 0.0525 in the Igbo population to a high of 0.2465 in the Ijaw-Ibibio population and the average heterozygosity (*avg.Het*) was 0.1191 (see Table 3).

Genetic Differentiation among Populations

To examine the amount of genetic differentiation among the four populations, Gst value (a

Table 1: Genotype and allele frequencies of the various populations in Nigeria

Populations	*n*	*(%)*	*+/+ (%)*	*-/- (%)*	*+/- (%)*	*+ (%)*	*-(%)*	*Total (%)*
Hausa-Fulani	47	(31.54)	1 (0.67)	44 (29.53)	2 (1.34)	4 (1.34)	90 (30.20)	94 (31.58)
Yoruba	20	(13.42)	1 (0.67)	19 (12.75)	0 (0.00)	2 (0.67)	38 (12.75)	40 (13.42)
IIjaw-Ibibio	45	(30.20)	4 (2.68)	36 (24.16)	5 (3.36)	13 (4.36)	77 (25.84)	90 (30.20)
Igbo	37	(24.80)	1 (0.67)	36 (24.16)	0 (0.00)	2 (0.67)	72 (24.16)	74 (24.83)
Total	149	(100.00)	7 (4.7)	135 (90.6)	7 (4.70)	21 (7.05)	277 (92.95)	298 (100.00)

Legend: n: population number, +/+: homozygous *Alu* insertion genotype, -/-: homozygous *Alu* non-insertion genotype, -/-: heterozygous *Alu* genotype

Table 2: Observed and expected genotype frequencies to test Hardy-Weinberg Equilibrium

Populations	*Freq.*	*+/+ (n)*	*-/- (n)*	*+/- (n)*	*Total*	χ^2	*p-value*
Hausa-Fulani	OF	0.021 (1)	0.936 (44)	0.042 (2)	47	4.6301	<0.1
	EF	0.0019	0.9158	0.0842			
Yoruba	OF	0.050 (1)	0.950 (19)	0.000 (0)	20	1	<0.5
	EF	0.0025	0.9025	0.095			
Ijaw-Ibibio	OF	0.089 (4)	0.80 (36)	0.111 (5)	45	0.2495	<0.9
	EF	0.0207	0.7327	0.2465			
Igbo	OF	0.027 (1)	0.973 (36)	0.000 (0)	37	8.2916	<0.025
	EF	0.0007	0.9467	0.00525			
Nt	OFt	0.047 (7)	0.906 (135)	0.047 (7)	149	0.0793	<0.9
	EFt	0.0949	0.8639	0.1311			

Legend: Nt: Total population, OF: Observed Frequency, EF: Expected Frequency, OFt: total Observed Frequency, EFt: total Expected Frequency, Freq.: frequency, Ç²: Chi-square test

measure of the interpopulation variability) for *Alu* insertion was determined as shown in Table 3. The Gst was 0.0846 and differed significantly from zero, as judged by contingency chi-square analysis of the allele frequencies. Hence, the four populations are different or diverse with respect to the frequencies of the *Alu* insertion polymorphism.

To investigate the evolutionary relationship of these populations, a maximum-evolution tree was constructed directly from the genetic distance of *Alu* insertion allelic frequencies. The Ijaw-Ibibio population was shown to have the highest genetic distance (Table 4) and farthest away from the other regional populations while the Hausa-Fulani and Igbo populations clustered as shown in the phylogenetic tree (Fig. 3).

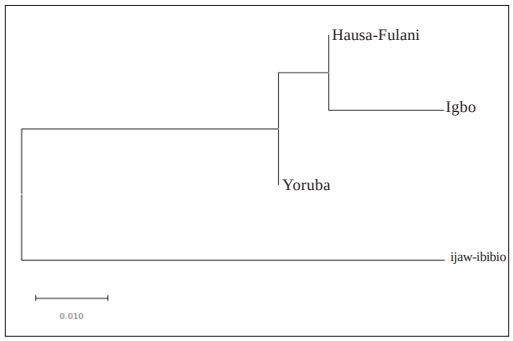

Fig. 3. Phylogenetic tree of the four ethnic populations of Nigeria

Gene Flow within Populations

To determine the relative amount of gene flow experienced by each population, we calculated the expected and theoretical heterozygosity of each population and the distance from the centroid as shown in Table 3 and evaluated their relationship as previously described by Harpending and Ward (1982). A plot of heterozy-

Table 3: Heterozygosity of the study population and distance from the centroid

Populations	*No. of alleles*	*Alu (+) allele*	*of Alu (+) allele freq.*	*Alu (-) allele*	*Alu (-) allele freq.*	*Het*	r_i	h_i
Hausa-Fulani	94	4	0.043	90	0.957	0.0823	0.0112	0.1286
Yoruba	40	2	0.05	38	0.95	0.095	0.0061	0.1293
Ijaw-Ibibio	90	13	0.144	77	0.856	0.2465	0.0841	0.1192
Igbo	74	2	0.027	72	0.973	0.0525	0.0284	0.1264
Nt	298	21	0.07	277	0.929	0.1301		
nHet						0.1301		
avg. Het						0.1191		
G_{st}						0.0846		

Legend: Nt: Total population, *avg.Het:* average heterozygosity, $_n$*Het*: heterozygosity of the total population. *Gst:* measure of interpopulation differentiation, *ri*: distance from the centroid, *hi*: theoretical heterozygosities of population *i* , freq: frequency.

Table 4: Genetic distance between regional populations of Nigeria

	Hausa-Fulani	*Yoruba*	*Ijaw-Ibibio*	*Igbo*
Hausa-Fulani	0			
Yoruba	0.007	0		
Ijaw-Ibibio	0.101	0.094	0	
Igbo	0.016	0.023	0.117	0

gosity versus distance from the centroid for the four populations in Nigeria showed only Ijaw-Ibibio population to be above the line of the predicted value (theoretical heterozygosity) while the Hausa-Fulani, Yoruba and Igbo populations were below the line of theoretical heterozygosity (See Fig. 4).

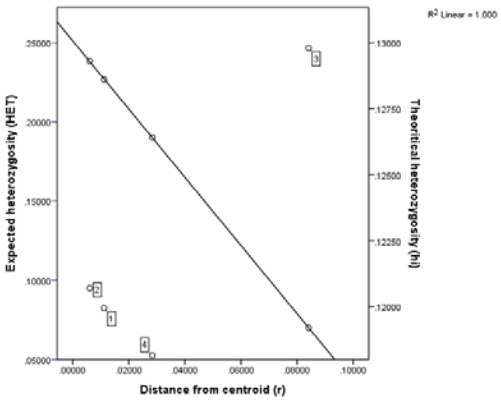

Fig. 4. Gene flow of populations in Nigeria
Legend: The numbers in the figure indicates the four different populations in Nigeria. 1: Hausa-Fulani, 2: Yoruba, 3: Ijaw-Ibibio, 4: Igbo

DISCUSSION

Alu insertion polymorphic sequences are known to have originated from common ancestral master genes since the primordial time of human existence and have been transmitted from one generation to another (Terreros et al. 2009). Because of their ability to transpose within the human genome, *Alu* insertion polymorphic sequences can serve as a unique source of nuclear genetic variability to study human population (Roy-Engel et al. 2001; Salem et al. 2003). More so, *Alu* polymorphism which is due to insertion of the element in the genome is considered as a forward mutation thereby facilitating an accurate estimation of the root in phyologenetic tree of population relationships (Batzer et al. 1994). In this study we assessed the *Alu PV92* polymorphism in four Nigerian populations; the Hausa-Fulani, Yoruba, Igbo and Ijaw-Ibibio populations. *Alu* insertion polymorphism was present in all four populations and greatest in the Ijaw-Ibibio population. Hardy-Weinberg equilibrium (HWE) was not violated in the Hausa-Fulani, Yoruba and Ijaw-Ibibio populations suggesting that this population may have evolved through natural selection process. Though, HWE was violated in the Igbo population probably suggesting that this population might have evolved through non-random mating or by mutations other than the *Alu* polymorphism, the fact that the HWE was not violated in the entire population suggests that the *Alu* polymorphism may be responsible for the evolution and diversity of the entire populations in Nigeria.

Heterozygoisity was highest in the Ijaw-Ibibio population (0.2465) but the average heterozygosity of the Nigeria population was lower (0.1191). Though low, this heterozygosity was high when compared to that of a study by Batzer et al. (1996) which showed Nigeria to have a heterozygosity of 0.091 for *Alu PV92*. This suggests that interpopulation breeding in Nigeria may have increased in recent time to exhibit such increase in genetic diversity. Also, the G_{st} for *Alu* insertion was 0.0846 and different from zero thus depicting some level of interpopulation differentiation or diversity. However, this value is comparably lower than that of other regions across the world (Panjaliya et al. 2010; Batzer et al. 1996; Stoneking et al. 1997) suggesting that these four populations in Nigeria are not very diverse compared to other populations across the globe (Chinniah et al. 2016). Moreover, this G_{st} is higher than that of other populations across the globe (Chinniah et al. 2016; Panjaliya et al. 2012; Litvinov et al. 2008) suggesting that Nigeria shows some degree of diversity comparatively higher than that of some populations in the world. The minimum- evolution tree which showed the genetic distance between populations was used to evaluate the evolutionary relationship between the four populations. The phylogenetic tree showed Hausa-Fulani and Igbo populations to be closely related while the Ijaw-

Ibibio and Igbo populations showed the largest genetic distance between them. The Ijaw-Ibibio population showed the largest genetic distance thus was farthest apart from the other populations, indicating the highest level of variability compared to other populations. This suggests the Ijaw-Ibibio population to be the possible ancestral origin of human population in Nigeria. The gene flow of the populations was investigated to further confirm this possibility by evaluating the relationship between the heterozygosity and the distance from the centroid. A plot of heterozygosity against distance from the centroid showed the Igbo, Yoruba and Hausa-Fulani populations to have a heterozygosity below the predicted value (theoretical heterozygosity) while only the Ijaw-Ibibio population was found to be above the predicted value indicating that the Ijaw-Ibibio population had the greatest level of gene flow in the population and thus affirm the Ijaw-Ibibio population as the ancestral population of Nigeria which other populations may have evolved.

CONCLUSION

The four main populations of Nigeria were found to be closely related with low level of genetic diversity except for Ijaw-Ibibio population which showed the highest interpopulation differentiation. As such, the Ijaw-Ibibio population may be considered to be the ancestral population of Nigeria. This study further supports the use of polymorphic *Alu* insertions to assess the direction of the evolutionary history of population groups and provides information about their diversity and relatedness. Though a few studies have used *Alu* polymorphism to assess the evolutionary relationship between Nigeria and other populations across the world, this is the first study to evaluate the interpopulation relationship within Nigeria. However, being a pilot study, the small sample size may be limiting to generalization. Thus, this finding may serve as a baseline for further studies within the Nigerian population.

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