ALBINISM ALLELIC FREQUENCY IN HUMAN SUBJECTS LIVING IN THE NORTHERN GUINEA-SAVANNAH OF NIGERIA

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Abstract: Albinism an autosomal recessive allele is a disease characterized by lack of pigmentation in the eye, skin and hair. Albinism allelic frequency was estimated by counting the number of albinos per 10,000 individuals in the Cities (Kano, Kaduna, Sokoto, Gusau and Minna) of Northern guinea savannah. The allelic frequency was low, with range 0.013 to 0.011. Kano population has a frequency of 0.013, Kaduna and Minna 0.012, Sokoto and Gusau 0.011, respectively. For the genotypic frequencies, homozygous recessive genotype was found to 0.016%, the heterozygous genotype was 2.5% and the homozygous dominant genotype was 97.2% in Kano population. In Kaduna and Minna populations, homozygous recessive has 0.014%, heterozygous has 2.3% and homozygous dominant genotype has 97.4%, while homozygous recessive has 0.012%, the heterozygous genotype has 2.1% and homozygous dominant has 97.6% in Sokoto and Gusau populations. In conclusion the allele has a low frequency within the sub region, which could be as a result of purifying selection and mate preference in the populations.

Keywords: Albinism, heterozygote, homozygote, genotype, allele

Introduction

Albinism is a heritable congenital skin condition characterized by lack of pigmentation in the eye, skin and hair. There are conflicting reports about its prevalence. A report by U.S. institute of health cited 1 in every 20,000 as the frequency of albinism worldwide (US Institute of health 2007). Another report, by a national group serving persons with albinism "vision for tomorrow" cited 1 in every 17,000 (Vision for tomorrow 2013), the U.N. high commissioner for human right reported 1 in every 15,000 people (U.N. 2013). Although OCA2 (Occulocutaneous albinism 2) occurs worldwide, it has a relatively high frequency in several African populations. In West Africa estimate range from 1 in 1100 in the Igbo of Nigeria to 1 in 3800 among the Bamalike of the Cameroon (Witkop *et al.*, 2013).

In Souh Africa the frequency varies between different ethnic groups, being highest among the Swazi-sotho at about 1 in 1500 to 2000 (Kromberg *et al.*, 1999) and lowet among the Ngumi ethnic groups at 1 in 4500 (Lund 2016). Albinism an autosomal recessive loss of functional mutation is a trait passed from parents that are either afflicted or carriers to their offspring. A mutation in human TRP-1 gene may result in the deregulation of melanocytes, tyrosinase enzymes, a change that promotes brown versus black melanin synthesis, resulting in an oculocutaneous albinism genotype (Boissy *et al.*, 1996). All races are affected by albinism, but prevalence are more in some part of the world than other parts, for example in Africa 20 out of 100,000 people are albinos, while in USA and Europe only 5 out of 100,000 people are albinos (Kruijt, 2011)

Associated with a number of defects such as photopobia, amblyopia, nystagmus and lack of skin pigmentation, albinism makes skin susceptible to sunburn and skin cancers. This also affects essential granules present in immune cells, leading to increased susceptibility to infection (Caplan *et al.*, 2014). Albinism result from inheriting recessive gene alleles, it is known to affect all vertebrates, including humans. It is due to absence or defect of tyrosinase, a copper-containing enzyme involved in production of melanin. It is the opposite of melanism (Joel *et al.*, 2014).

Albinism affected individuals, faces discrimination, violence, ridicule and other social and cultural challenges. In many African societies it is socially stigmatized. In a study conducted in Nigeria on albino children it was stated that they experienced alienation, avoided social interaction and were less emotionally stable. Furthermore, affected individuals were less likely to complete schooling and employed, and find partners in life. Many cultures around the world have developed beliefs regarding people with albinisms (Sasasha and Jacquelyn, 2013).

Albinism can reduce the survivability of an animal, for example, it has been suggested that albino alligators have an average survival span of only 24 hours due to the lack of protection from, ultraviolet and their lack of camouflage to avoid predators (McCardle, 2015)

Albinos have characteristic pink or red eyes because the pigment they lack in the iris allows the blood vessels of the retina to be visible.

The P gene for OCA2 (occulocutaneous albinism 2), which is located on chromosome 15, codes for an integral membrane protein with trans-membrane domains (Lee *et al.*, 1994).

Although the function of this P polypeptide remains uncertain, it may be active in transporting tyrosine into melanosomes. It has been suggested that there is locus homogeneity among southern African negroids with occulocutaneous albinism. The most common mutation so far identified in people with this type of albinism from Africa is a 2.7 kb deletion that removes an entire exon of the P gene (Kedda *et al.*, 1994). Albinos may experience sight problems, which may be near sighted or far sighted. They may have astigmatism (defect in the curvature of the lens) resulting in blurred vision. Due to lack of pigmentation their irises do not prevent high light intensity from entering the eye, thereby making it very sensitive to the sun light (Hong *et al.*, 2006).

Oculocutaneous is the most common type of albinism; it affects the eyes, hair and skin. Hair and skin remain completely white throughout life, in its most severe form. However people with less form of affliction are born with white hair and skin that turn slightly darker as they age (Magna 2014). Most individuals with oculocutaneous albinism experience nystagmus (abnormal flicking of the eyes) and sensitivity to bright light. The second most common type of the condition is known as ocular albinism, in which only the eyes lack pigments, the hair and skin are normal.

It is premised that the early hominine evolved in east Africa around 3 million years ago. The striking phenotypic change from primate to early hominine is thought to have stem from reduction in body hair, with the exception of the head which is covered with hair for insulation to facilitate thermoregulation against ultraviolet radiation, in early hominines there is a

drastic reduction in hair in other parts of the body that are less exposed to ultraviolet radiation. The initially exposed skin can be assumed to be non-pigmented, resembling the light skin of our relatives the primates. A positive selection advantage must have been inferred to hair loss phenotypes in general, in these early ancestors dwelling the tropical environment that are bestowed with melanin producing genes that probably gave them survival advantage over their counterparts that don`t have. They survive and therefore left more offspring and became the most dominant groups in the environment (Greaves, 2015).

There are other genetic mutations which are proven to be associated with albinism, however all changes lead to alterations in melanin production in the body. Albinism usually occurs with equal frequencies in both sexes, except ocular albinism, which is passed onto offspring through x-linked inheritance. Thus ocular albinism occurs more frequently in males as they have a single x and y chromosomes, unlike females, whose genetics are characterized by two x chromosomes (Occulocutaneous Albinism, 2017).

Variations in living organisms can be broadly classified into discrete and cryptic variations. Discrete variations are visible variants with large effect on phenotypes, such as flower color in plants, shell patterns in snails, mutations such as dwarfism as well as a complete absence of pigment in the skin that is referred to as albinism or albinoid. Cryptic variation isthose variations that do not give rise to obvious differences in phenotypes. Cryptic variations can only be revealed by techniques that study differences in proteins and the DNA itself. The MN blood groups, ABO blood groups and the sickle cell traits are examples of cryptic variations (Raymond *et al.,* 2017)

Albinism which is a double recessive autosomal trait falls under discrete type of variation since it is tangible; it is therefore possible to diagnose it by physical appraisal.

Materials and Methods

This study was conducted in five major Cities within the northern guinea-savanna of Nigeria. The Cities due to their high populations were choosing to form the study areas. The cities are Kano, Longitude 8º31`00``E, Latiude12º00`00``N, Kaduna, Longitude 7º25`46.214E, Latitude 10º36`33.5484``N, Sokoto, Longitude 5º14`51.1872``E, Latitude 13º021`.1428``N, Gusau Longitude 6º39`50``E, Latitude 9º39`50``N and Minna Longitude 6º32`46.7376``E, Latitude 9º35`0.7980``E, respectively (GPS, 2016).

Sampling was done by visiting ward heads in the Cities and enquiring about the presence of albinos residing in their domains. The population of each city was obtained from 2006 census data. Random sampling was adopted in the sampling, using equal sampling size techniques of Kogi (Kogi, 2015).

$$n = \frac{z^2(1-p)}{2d^2}$$

Where: z = 1.96 at 95% confidence interval, p = proportion of infected individuals, p = proportion of individuals uninfected, d = level of precision (level for test of significance).

Genotyping

Genotypes were group into three columns; homozygote dominant (AA), heterozygote (Aa) and homozygote recessive (aa). Genotypes were obtained by counting the numbers of individuals that falls into each phenotypic class, that is for the two homozygous (dominant and recessive). There was no any method available as to the best of my knowledge used in determining the heterozygous genotype at the time of the study, we were rendered handicapped, identifying heterozygous was not possible by physical examination, no laboratory techniques was available for us. We then opted to use a proportion of 1/7 available in the literature, (Vision for Tomorrow 2013). Albinism allelic frequency is the fractional representation of the allele in the population. It was calculated by multiplying the number of individuals counted in each population by two, to get the total number of alleles (remember allelic frequency is twice the number of genotype). The total number of alleles was then used to divide the number of albinism allele copies in the populations.

The genotypic frequencies are the number of individuals that fall under each genotypic groups divided by the total number of individuals in the population; AA/n, Aa/n, aa/n

Hardy-Weinberg principle (Hardy, 1908; Weinberg, 1908) was used to estimate expected genotypic frequencies; according to the Hardy-Weinberg equilibrium principle, the frequencies of two alleles are p and q, the frequencies of the genotypes are: $p^2 + 2pq + q^2$, this is represented as;

(AA)	(Aa)	(aa)
p^2	2pq	q^2

The expected frequencies of each genotype, multiply by the number of individuals in the population is the expected number of individual genotype in each population under Hardy-Weinberg equilibrium.

Chi-square was used to test level of significance among the populations;

$$\chi^2 = \frac{\sum(observe - \exp ected)^2}{\exp ected}$$

Results and Discussions

Information on allelic and genotypic frequencies are provided in Table 1. The albinism allelic frequency ranges from 0.013 to 0.011 in the studied populations, with Kano population having 0.013, Kaduna and Minna 0.012, Sokoto and Gusau have 0.0011 each. The average albinism allelic frequency across the whole population is low 0.003. This low frequency among the population could be due to the effect of natural selection against the allele. The lack of pigmentation in the skin coded by the allele makes it very sensitive to ultraviolet radiation and thereby more prone to cancer, which is lethal. Mate preference and negative selection may be responsible for this low frequency.

Table 1: Allelic and genotypic frequencies of albinism trait in the studied populations

Population		No. Counted	Genotypic	Frequency	Allelic	Frequency
		AA	Aa	aa	A	a
Kano	10000	0.972	0.025	0.00016	0.986	0.013
Kaduna	10000	0.974	0.023	0.00014	0.987	0.012
Sokoto	10000	0.976	0.021	0.00012	0.988	0.011
Gusau	10000	0.978	0.021	0.00012	0.989	0.011
Minna	10000	0.974	0.023	0.00014	0.987	0.012

A=dominant allele a= recessive allele AA=homozygous dominant, Aa=heterozygous, aa=homozygous recessive

The homozygous recessive frequencies are 0.00016 in Kano population, 0.00014 in Kaduna and Minna populations, 0.00012 in Sokoto and Gusau populations. This low frequency, despite the fact that natural selection is weeding out the genotype, it is still persisting and did not wipe-out the allele completely. Perhaps the fitness cost suffered by albinism alleles when in homozygous is balanced by a fitness advantage they enjoy when in heterozygotes (heterozygous advantage). The heterozygous frequencies are 0.025 in Kano population, 0.021 in Sokoto and Gusau populations, 0.023 in Kaduna and Minna populations, respectively.

Table 2: Number of individuals (observed and expected) under the three genotypic groups in each populations

Populations		Genotypes		
		AA	**AS**	**SS**
Kano	Observed	972	25.63	016
	Expected	973	26.20	0.17
Kaduna	Observed	974	23.90	0.14
	Expected	981	23.70	0.15
Sokoto	Observed	976	21.73	0.12
	Expected	978	22.04	0.12
Gusau	Observed	978	21.75	0.12
	Expected	984	22.51	0.12
Minna	Observed	974	23.01	0.14
	Expected	982	23.68	0.15

Table 3: Observe and expected numbers of individuals studied along with their χ^2 values

Population	Genotypes				χ^2	LOS
		AA	**Aa**	**aa**		
Kano	observed	972	25.63	0.16	0.014	ns
	expected	973	26.20	0.17		
Kaduna	observed	974	23.90	0.14	0.052	ns
	expected	981	23.70	0.15		
Sokoto	observed	976	21.73	0.12	0.008	ns
	expected	978	22.04	0.12		
Gusau	observed	978	21.75	0.12	0.062	ns
	expected	984	22.51	0.12		
Minna	observed	974	23.01	0.14	0.084	ns
	expected	982	23.68	0.15		
$\chi^2_{0.05} = 5.991$						

A=dominant allele a= recessive allele AA=homozygous dominant, Aa=heterozygous, aa=homozygous recessive

Deviation of the observed number of individuals from the expected in the populations are not statistically significant (Table 3), suggesting that the populations conform to Hardy-Weinberg principles.

Conclusions

Albinism allelic frequencies has been found to be low (average allelic frequency 0.03) in the studied populations. This could be attributed to purifying selection (negative selection) against the allele and mate preference or choice among the individuals. Although the allelic frequencies were low, the trait stillpersist in the populations due to heterozygous advantage (over dominance) they enjoy in the populations.

Conflict of Interest

Authors have declared that there is no conflict of interest reported in this work.

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