

Artigo original

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Taxa elevada de albinismo oculocutâneo no estado da Bahia, região nordeste do Brasil

High rate of oculocutaneous albinism in the state of Bahia, northeastern of Brazil

ABSTRACT

Oculocutaneous albinism (OCA) is a detrimental genetic trait that affects the production of melanin, resulting in hypopigmentation of the skin, eyes and hair, with a worldwide distribution but higher prevalence in the African continent. In this article we aim to analyze the OCA distribution in cities of state of Bahia, northeastern Brazil, with high rate of African descent. This study had a descriptive and exploratory approach and used a convenience sample consisted of 481 subjects of both sexes, aged 1-65 years registered in Albinism Association of the State of Bahia (APALBA). It was found high rates of albinism in 34 of these cities, in individuals of different age groups, suggesting a founder effect and also the role of random processes as internal migration, family size, fertility, as well as economic, natural and cultural factors. The occurrence of *quilombola* communities in some of these cities was also associated with high rates of black population and the increased frequency of albinism. Although the lack of melanin provides greater susceptibility to skin cancer for people with albinism, natural selection does not act against the albinism gene to a severe level. Advances in public health, allied to greater attention to albinism, are factors which may contribute to reduce the deleterious consequences of the disorder.

RESUMO

O albinismo oculocutâneo (ACO) é uma característica genética prejudicial que afeta a produção de melanina, resultando na hipopigmentação da pele, olhos e cabelos, com distribuição mundial, tendo maior prevalência no continente africano. Este artigo objetiva analisar a distribuição de OCA nos municípios da Bahia, nordeste do Brasil, estado com alta taxa de descendência africana. O estudo teve abordagem descritiva e exploratória, utilizando uma amostra de conveniência constituída por 481 sujeitos de ambos os sexos, com idade entre 1 a 65 anos, cadastrados na Associação de Albinismo do Estado da Bahia (APALBA). Foram encontrados altos índices de albinismo em 34 desses municípios, em indivíduos de diferentes faixas etárias, sugerindo um efeito fundador e o papel de processos aleatórios como migração interna, tamanho da família, fertilidade, além de fatores econômicos, naturais e culturais. A ocorrência de comunidades quilombolas, em algumas dessas cidades, também foi associada a altas taxas de população negra e ao aumento da frequência de albinismo. Embora a falta de melanina forneça maior suscetibilidade ao câncer de pele para pessoas com albinismo, a seleção natural não age contra o gene do albinismo a um nível severo. Avanços na saúde pública, aliados à maior atenção ao albinismo, são fatores que podem contribuir para reduzir as consequências deletérias deste distúrbio.

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KEYWORDS

Albinism Oculocutaneous; Founder Effect; African Continental Ancestry Group.

PALAVRAS-CHAVE

Albinismo Oculocutâneo; Efeito Fundador; Grupo de Ancestralidade Continental Africano.

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INTRODUCTION

The Brazilian population is the outcome of many different ethnicities, initially resulting from the merge of the original indigenous population with Portuguese immigrants and enslaved Africans during the colonization period. In the last centuries there was a new migratory flow of different foreign peoples that were settled in different regions of the country, which made up peculiar ethno geographical features of Brazil (SALZANO & FREIRE-MAIA, 1970). There was a predominance of African miscegenation in the North-eastern states of Bahia, important centre of trading and distribution of black slaves, with cultural and genetic implications in the composition of the current population. Studies on the genetic characteristics in this region show frequent diseases in the African population, such as oculocutaneous albinism on the island of Lençóis in the state of Maranhão, attributed to genetic drift (FREIRE-MAIA et al., 1978). Monteiro and others (2017) assert that geographic location is a preponderant factor in the occurrence of genetic characteristics in populations and the analysis of different regions shows a strong correlation between genetic and environmental factors and also socioeconomic factors including the social exclusion, which takes place nowadays (BRASIL, 2001).

Albinism is a heterogeneous group of genetic disorders that lead to a reduction or congenital absence of melanin. Gronskov e outros. (2007) estimate that 1 in 70 people are carriers of a gene related with albinism. Among their different forms, the Oculocutaneous Albinism (OCA), of autosomal recessive inheritance, is the most important category, reaching about 1:20,000 people worldwide. This condition is sub-classified into two main forms: OCA1 (MIM 203100), mapped in 11q14.3, which produces the enzyme tyrosinase, crucial for the synthesis of melanin, and OCA2 (MIM 203200) mapped to 15q11.2-q12, in which tyrosinase is present and functional but is impaired by mutations in OCA2, leading to disturbance in the synthesis of melanin. The TYRP1 protein, associated with OCA2 gene, is higher in black people and Indians melanocytes compared to Mexican, Chinese and Europeans, suggesting that this protein may play a significant role in ethnic differences arising from melanogenesis and consequent skin pigmentation (NEWTON et al., 2001). This finding supports studies on the worldwide distribution of diseases, which states that OCA1 albinism displays the same frequency in all populations, while OCA2 is about twice more common among black people, with an estimated incidence of 1:10,000 in Africans and African-Americans, and about 1:36,000 in Caucasians (ALALUF et al., 2003).

This study analyses the distribution of albinism in Bahia state's, where black and mulatto population is predominant in relation to other states of Brazil.

MATERIAL AND METHODS

BAHIA STATE DEMOGRAPHIC CHARACTERISTICS

According to the IBGE (2008), Bahia is the fourth most populous state in Brazil, with an estimated population of 15,126,371 inhabitants with high rate of black and mulatto population, living in 417 cities grouped into 7 mesoregions: Metropolitan of Salvador; South-Central Bahia; North-

Central Bahia; South Bahia; Northeast Bahia; São Francisco Valley; Far West Bahia.

DATA COLLECT

This is a descriptive and analytical study. Frequency of albinism in the state of Bahia was estimated by consulting the database of APALBA (People with Albinism Association in the state of Bahia) in the years 2013-2015.

The methodological study procedures are in accordance with the ethical principles defined by the Declaration of Helsinki and it was approved by the Ethics Committee of Nursing Faculty of Federal University of Bahia under number 082461/2014.

STATISTICAL ANALYSIS

The descriptive statistical analysis and graphics analysis were made through R version 3.4.4 (R Core Team, 2018) using R packages as TidyR version 0.8.1, Ggplot2 version 3.0.0 and Dplyr version 0.7.7.

RESULTS AND DISCUSSION

This study has recorded 481 cases of albinism in 90 cities in the state of Bahia, categorized in mesoregions. Table 1 shows the population rates of albinism (albino rate) in 34 cities of different mesoregions, in which the frequency of albinism was higher than 1:10.000 inhabitants, which is the expected value for African descendants. Albino rate concerns the number of people with albinism per 10.000 habitants and was calculated for each city. Figure 1 shows the topographic distribution of these cities. Figure 2 shows the Sapinhos Island, small isolated inserted in mangrove areas in Marau city, inhabited for fishermen and their families including five siblings with albinism.

In order to generate non-parametric box-and-whisker plot (figure 3), a graphical tool that represents the variation of observed data, all cities were arranged based in percentage of black and mulatto populations in order to each sub-group presents 11 (± 1) cities, resulting in 3 groups with 88-78%, 78-72% and 71-54%.

Albinism is a group of genetic disorders that leads to hypopigmentation. It is considered a rare genetic disease and it is included in the National Register for the study of genetically isolated populations, identified by National Isolated Census (CENISO) (CASTILLA & SCHULLER-FACCINI, 2014).

In the population taken for study, cases of albinism were recorded in different mesoregions state, with rates 4~6/10.000 in seven of these cities (Inhambupe, Miguel Calmon, Macajuba, Morro do Chapéu, Acajutiba, Gavião, Capim Grosso). Individuals with albinism in the municipality of Marau, were resident in Sapinhos Island, with 101 inhabitants and only accessible by waterways (OLIVEIRA, 2005). Here the founder effect can be contributing to the high rate of this recessive trait, despite the low adaptive value, with increased risk of skin cancer, in a sunny environment where fishing is the main source of livelihood. Beiguelman (1994) observed that the distribution of certain frequencies gene alleles in human populations has high probability of being a result of genetic drift.

Cities	Mesoregions	Number of albinos	Population size (IBGE, 2010)	Rate of Albinism (n/10.000 habitants)	Percentage of black and mulattos people (IBGE, 2010)
Lamarão	Northeast Bahia	1	9027	1,1	88,85
Tanquinho	Central-North Bahia	2	8008	2,49	88,64
Maraú	South Bahia	5	19097	2,61	88,38
Castro Alves	Metropolitan of Salvador	10	25419	3,93	85,8
Santa Bárbara	Central-North Bahia	4	19064	2,09	84,12
Ibirataia	South Bahia	2	18946	1,05	83,89
Rafael Jambeiro	Central-North Bahia	3	25555	1,17	82,12
Quijingue	Northeast Bahia	6	27243	2,2	81,55
Laje	South-Central Bahia	3	22206	1,35	81,39
Inhambupe	Northeast Bahia	22	36290	6,06	80,7
Caldeirão Grande	Central-North Bahia	3	12485	2,4	78,64
Miguel Calmon	Central-North Bahia	13	26466	4,91	78,64
Teofilândia	Northeast Bahia	7	21484	3,25	78,61
Macajuba	Central-North Bahia	5	11229	4,45	78,36
Morro do Chapéu	Central-North Bahia	15	35207	4,26	76,96
Ibicoara	South-Central Bahia	2	17301	1,15	76,67
Itaberaba	Central-North Bahia	13	61623	2,1	76,4
Acajutiba	Northeast Bahia	6	14 830	4,04	76,24
Quixabeira	Central-North Bahia	1	9548	1,04	75,38
Cansanção	Northeast Bahia	6	32923	1,82	75,08
Nova Fátima	Northeast Bahia	3	7602	3,94	75,01
Aporá	Northeast Bahia	2	17720	1,12	73,58
Jaguarari	Central-North Bahia	5	30342	1,64	72,82
Riachão do Jacuípe	Northeast Bahia	7	33081	2,11	71,47
Planaltino	South-Central Bahia	1	8822	1,13	69,41
Gavião	Northeast Bahia	3	4561	6,57	69,06
Maracás	South-Central Bahia	3	24615	1,21	68,75
Remanso	São Francisco Valley	5	39149	1,27	68,32
Capim Grosso	Central-North Bahia	12	26529	4,52	67,91
Coronel João Sá	Northeast Bahia	3	17066	1,75	67,45
Brotas de Macaúbas	South-Central Bahia	4	10718	3,73	66,75
Ribeira do Pombal	Northeast Bahia	9	48271	1,86	66,03
Macaúbas	South-Central Bahia	16	47067	3,39	61,27
Abaíra	South-Central Bahia	1	8324	1,2	54,31

Table 01. Cities of Bahia state with albinism rate greater than 1:10,000.

The rate of albino people reflects the amount of people with albinism inside a 10.000 group people per city. Through box-and-whisker plot (figure 3) it's possible to notice a higher individual rate of each city per group when comparing the 3 groups, which reflects in the average value.

Although group 3 counts on the city with the highest rate of albinism, Gavião, with 3 albino cases within a population of 4561 peoples (6,57 albino rate), the city reports only 69% of citizens as black and mulattos, which lines up the city within group 3, specific to the 11 cities with the lowest black and

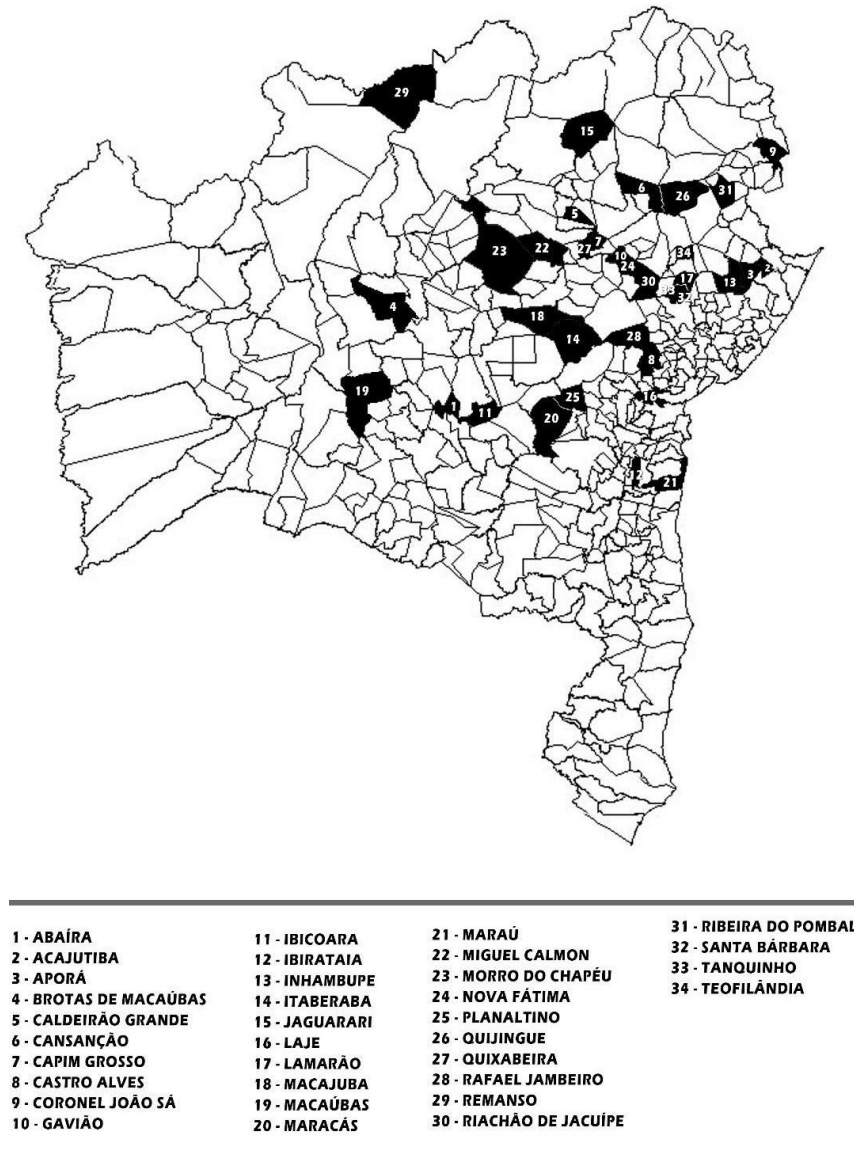


Figure 01. Topographic map of the cities in the state of Bahia (Brazil). Cities with albinism ratio of $>1:10.000$ are highlighted in black.

mulattos rate that presents average value for albino rate as 1.86. The increased prevalence for Gavião seems to be explained in another way. In general, the mean values of albino rate floats related to percentage of black population. This confirms that among the cities with higher black and mulatto rate, albinism is indeed more prevalent.

The existence of remnants of *quilombo* in some of these communities is an important factor associated with higher frequency of African descent and possible increase in albinism rate in the investigated populations, in view of the relative isolation in which they remained until the second half of last century.

In urban areas, more densely populated, with high rates of individuals with albinism, it should be considered, beyond the influence of the historical patterns of colonization, African Diaspora and internal migration, and the occurrence of other factors, such as family size and fertility. The analysis of the spatial dispersion of the population draws

attention not only to the importance of migration leading to decentralization of cities, but also to morphoclimatic conditions, economical, natural and cultural factors.

Studies associating sun exposure and the occurrence of cancer in albino skin lesions treated in a public centre of cancer treatment in the state of Bahia shows that the average age of albinos with cancer was 42.8 years, significantly increased when compared to the average of 65, one year for individuals without albinism (MOREIRA et al., 2013).

However, it should be taken into consideration that between the albinos with skin cancer 56% had basal cell carcinoma, the most common type of skin cancer in Brazil and greater chance of cure, which helps to minimize its deleterious effect. Moreover, it is observed that despite the increased susceptibility to skin cancer presented by albino, this condition, when detected early, has high rates of cure (KLIGERMAN et al., 2002).



Figure 02. Sapinhos Island (Environmental Protection Area, Marau-BA)

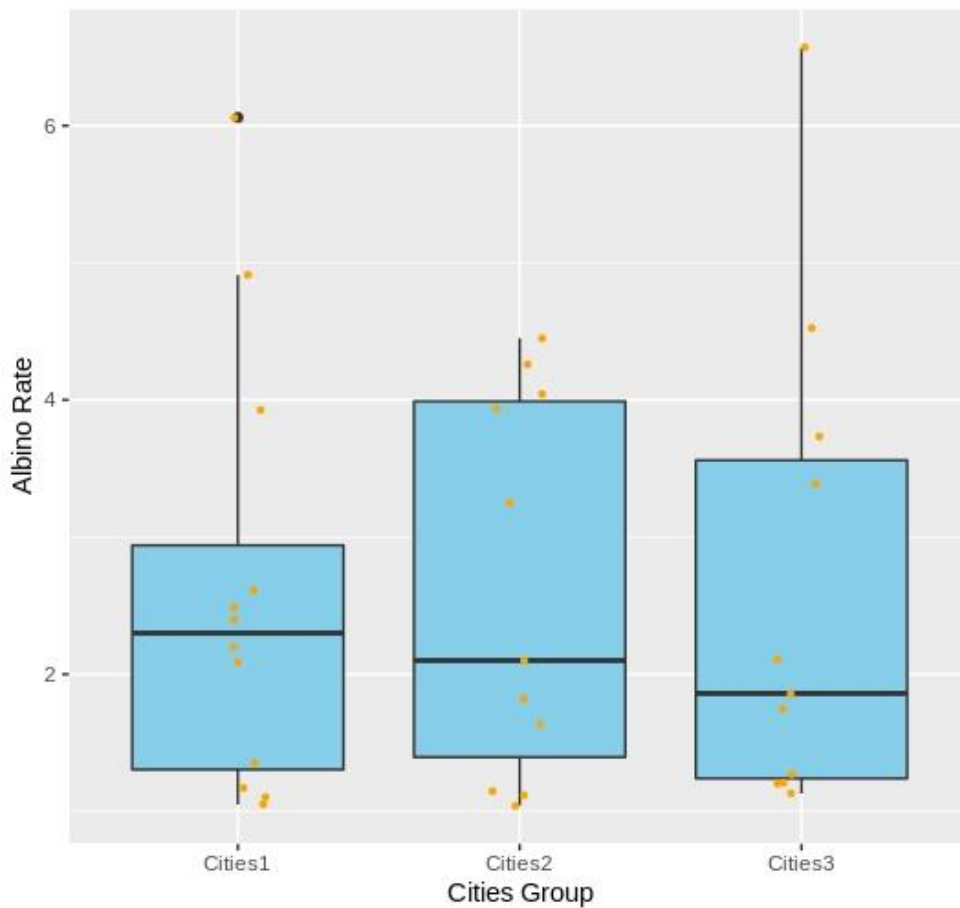


Figure 03. Box-and-whisker plot correlating albino rate with all 34 cities arranged in 3 groups due percentage of black and mulattos populations: “Cities 1” account cities with 88-78%, “Cities 2” account cities with 78-72% and “Cities 3” account cities 71-54%.

There was also no record of early mortality that could be attributed to hypopigmentation or decrease of the rate of marriages associated to natural selection or adaptive disadvantage. Notwithstanding the improvement of health conditions and medical advances, the albinism is

stigmatizing condition and the patients face health difficulties arising from low vision and are more prone to skin cancer.

CONCLUSION

This study presents original data on the distribution of Oculocutaneous Albinism in the state of Bahia, allowing associating the higher frequency of albinism with African descent. The results suggest the occurrence of founding effects and random processes, such as internal migration, family size, fertility, as well as economic, cultural and natural factors associated with the studied region. The correlation between genetic and environmental factors, and also social exclusion, makes it relevant to maintain and increase public policies to prevent the deleterious effects of this condition, such as the occurrence of skin cancer associated with greater susceptibility to sun exposure.

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