

A Unique Albino Village of Bhatti Tribe in Rural Sindh, Pakistan, with Oculocutaneous albinism Manifestations: An Epidemiological Study

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Abstract

Background: Oculocutaneous albinism is a disease with an autosomal recessive inheritance pattern in most cases. People with Oculocutaneous albinism face many health, psychological and financial issues. In this study, we report a unique village of Bhatti tribe in Jacobabad District, Pakistan, in which 40 children and adults with albinism live. The aim of this study was to observe the pattern of inheritance, complications and socioeconomic impacts of this condition on the community.

Methods: Detailed clinical history and relevant data were recorded on a specially designed performa followed by clinical examination by a consultant dermatologist together with his team to observe the extent of the disease and associated complications in two seasons (winter and summer).

Result: This village, with a total population of 810, had 40 cases, 17 male and 23 female, ranging in age from 6 months to 35 years. About 65% of the affected individuals were younger than 16 years of age. There were 22 involved families in this village and 2 of them had 3 or 4 affected members. In this area, which is geographically close to the equator, all 40 cases (100%) had photodermatosis and 36/40 cases (90%) had bacterial skin infections during summer. In addition, they had limited outdoor exposure during this season. These complications also caused occupational, social and educational limitations 9 months a year. Eye problems were present in all 40 cases; however, all 40 cases were malignancy free.

Conclusion: Oculocutaneous albinism is challenging not only because of its management but also because of its social and financial impacts. The current trend of consanguineous marriages, prevalent in this village, puts further emphasis on the role and the importance of marriage counseling in such situations. (*Iran J Dermatol* 2009;12:42-46)

Keywords: oculocutaneous albinism, epidemiology, health education, psychosocial impact

Introduction

Oculocutaneous albinism is a congenital heterogeneous group of autosomal recessive disorders manifested by hypopigmentation of the skin, hair, and eyes. The basic defect lies either in the decreased or absent biosynthesis of melanin pigment. It has different forms with Oculocutaneous albinism type I and II being the most common varieties. Type I Oculocutaneous albinism (tyrosinase deficient), which is more severe, results

from reduced or absent activity of tyrosinase, the key enzyme in the biosynthesis of melanin. The individuals with albinism have a unique physical appearance which causes many problems in the society in the form of social discrimination and integration^{1,2}. Associated ocular problems (Photophobia, Nystagmus, and Poor vision) along with extreme photosensitivity of skin further complicate this condition. Therefore, the ability to acquire education and social skills are affected in such cases.

In different parts of the world, the ratio of population affected with this disorder varies. It is 1:37000 in the USA whereas the reported ratio was about 1:4182 in Zimbabwe³. However, very few isolated pockets of populations having high frequency of albinism have been identified and reported from different continents such as Tonga, Pedi, Venda from South Africa^{4,5,6} and an ethnic group (Tili) in West Bengal⁷. This phenomenon could be due to the prevalence of unique family practices and social customs prevalent in these communities such as family intermarriages or having several wives from the same family or tribe (polygamy) with increasing breeding tendencies or due to geographic isolation with limited mobility^{4,8,9,10}. The clustering of a large number of cases of any disease in a particular community is helpful in conducting researches as a large number of cases can be observed and examined at the same time and place. The clinical presentations, complications and the socioeconomic impacts of that particular disease can be observed at the same

time. Also, it occasionally helps to identify the pattern of the inheritance and to localize involved genes.

The published clinical data regarding albinism is sparse in Indo-Pak subcontinent. A few cases of albinism were initially reported to us in the outpatient department and revealed the presence of the similar disease affecting a large number of children/adults in the village. This village was inhabited mostly by people from a single Bhatti tribe. In view of this interesting and unique features mentioned to us, it was decided to organize and conduct an epidemiological, clinical and psychosocial study on albinism in this unique village of Bhatti tribe located in a remote area in Jacobabad District, Sindh, Pakistan.

Patients and Methods

Study Design: It was a cross-sectional study which included all patients with albinism irrespective of age, sex, ethnic and tribal background living in the



Figure 1: Photograph showing large number of Albino children

village of Bhatti tribe located 20 km southeast of Jacobabad city, Jacobabad District of Upper Sindh, Pakistan. Upon visiting this particular village, a large number of cases with OCA were identified. All affected individuals had white hairs, hazel eyes and milky white skin. An informed written consent was taken after explaining the nature and purpose of this study to the adult albinos or parents of the children with albinism. They were interviewed and a performa, specially designed in relation to the disease and aim of the study, was filled for each patient. Skin swabs were sent from all these patients for culture and sensitivity. For psychological assessment of the adults, a few questions were included in the performa after consultation with a psychiatrist. The infants and children were assessed through their parents' statements regarding their mental well-being. The filled performa and these cases were finally assessed by the psychiatrist. Detailed physical examination was performed to observe the extent of the disease and associated complications. Skin swabs were sent from all these patients for culture and microorganism sensitivity. This process was conducted twice, once in the summer and the other in the winter by a consultant dermatologist and his team and the purpose was to observe and compare clinical findings and complications in two extreme climates prevalent in that region. This area is one of the hottest places in Asia in summers and very cold in winters due to being close to the mountains of Balouchistan receiving waves of cold winds from central Asia. All gathered data was tabulated for further analysis.

Result

Total population of the village was 810 individuals in two tribes, Bhatti (745) and Brohi (65). There were 40 (4.38% of the total population) Albinos, 17 (42.5%) males and 23 (57.5%) females in the age range of 6 months to

Table 1. Out door activities measured in hours of the Albinos during summer and winters

	Summer		Winter	
	Male	Female	Male	Female
Limited 0-4 hrs	15/17 (88%)	21/23 (91.30%)	1/17 (5.88%)	0
Average 4-6 hrs	2/17 (11.76%)	2/23 (8.69%)	2/17 (11.76%)	12/23 (52.17%)
unlimited >6hrs	0	0	14/17 (82.35%)	11/23 (47.82%)

35 years. The number of affected individuals in each age group is as follows: 0-10 yrs (22, 55%), 11-20 years (10, 25%), 21-30 years (7, 17.5%), 31-40 years (1, 2.5%). 65% were below 16 years. A group of these children are shown in figure 1.

The affected individuals belonged to 22 families (total families: 94) in the village. There was one Brohi family (out of the total 7 Brohi families) with a single case of albinism. In this case, the male had married a normal female from an affected family of the Bhatti tribe. All the other 21 affected families belonged to the Bhatti tribe (out of 87 Bhatti families). In these 22 families affected, 2 families had 3 affected members, whereas the remaining families had either one or two affected members. All families were closely related and their elders recalled having heard of albinism in previous generations. All adult males with albinism were unmarried whereas four adult female albinos were married and had normal children and none had history of any albinos born to them.

All cases (40 cases, 100 %) had a history of severe sunburn during the summer and most of them had complaints of associated blister formation. Of them, 90 % (36 cases) had severe bacterial infections such as folliculitis, furunculosis and impetigo during the summer. About 85% had staphylococcal infections and the rest had streptococcal or E coli while one case had Pseudomonas infection. Of them, 20 % (8 cases) had fungal infection (confirmed as candidiasis culture); most of them were females suffering from chronic paronychia. None of the Albinos had skin malignancy. All 40 cases (100%) had eye complications such as poor vision, nystagmus and photophobia. Of 40 Albinos, 10 % (4 cases) either were going or had gone to school.

Of them, 85% (34 cases) had limited exposure to outdoor activities during the summer applying to both males and females. In the winter, 76.47% (13/17) of males and 47.82% (11/23) of females had unlimited outdoor activity (Table1). All adult albinos were jobless. Two albinos were performing religious services voluntarily. Females were only performing household activities. All affected families were living below the poverty line with an income less than 1000 rupees (20 dollars) per month. All cases had psychological disturbances and behavioral changes.

Discussion

This study was unique in the sense that a large number of albinos were studied more specifically regarding their health and psychosocial problems

for the first time in Pakistan and most probably in South Asia. Various studies have been conducted in different regions, mostly in South Africa, where different prevalence rates have been reported.^{4, 5, 6} There were many interesting findings/observations in this study. This was a large group of albinos (4% of the total population) living in a single village belonging to one particular tribe (Bhatti tribe). Jacobabad District is one of the hottest spots in Asia because of its geographical location. The temperature is 50° C or higher most of the days during the summer months. Therefore, the delicate/sensitive skin of this large group of albinos is continuously exposed to high ultraviolet radiation resulting in severe sun burns in almost all cases along with photodermatitis/photoageing which was an expected finding^{1,11,12,13,14}. In addition to these facts, most of them are economically poor and cannot afford to buy sun protective medicines and clothing required for this condition which further contributes to their problems^{1,9}.

The high rate of infections and lack of health facilities has made this population more vulnerable to complications such as cellulites/generalized sepsis. Infections are more common in these affected individuals due to the sensitive nature of their skin which is susceptible to solar damage resulting in lesions such as bullae /ulcers which may get secondarily infected. This area is the hottest in the country which results in a high rate of skin infections even in normal individuals. We could not observe a single case with sun related skin malignancy in this population of albinos which was a surprising finding in individuals with such skin types. There could be a number of reasons for this low occurrence. Firstly, the majority of the cases were children and might not have as yet received enough radiation for lesions to develop. Secondly, most of the albinos did not expose themselves enough to the sun so as to have received a cumulative dose of radiation enough for the malignancy to occur. A low malignancy rate has also been reported in one study in Indonesia¹⁵.

The presence of severe visual abnormalities hampered them from performing their normal daily activities. The affected families had a low socioeconomic status and could not afford photo protective and corrective measures for their eye problems as reported earlier⁴. These visual problems could be one of the reasons for low school attendance. The cosmetic problems of the albinos and their different facial appearances due to ocular defects and sarcastic remarks from fellow children were additional major factors which

resulted in the lack of acceptance leading to their low school attendance. Similar attitudes and behavioral problems have been reported in the past^{1,11}. Moreover, in other parts of the world there are schools with special visual aids and methods that enable such children to have reasonable education^{1,11,16}. The most interesting observation was the discouragement from school authorities and pressure on school authorities from the parents of normal children not to admit these albino children because they feared it was contagious or that their condition was God's curse. Similar attitudes also have been reported from other communities and countries in the past^{8,9,17}.

This disease leads to many social issues as observed in this study. Some of albinos felt that they were different and abnormal and had a feeling of being "let down". Social discrimination was seen as an obstacle in their relationships and maintaining an occupation. There was also a feeling of helplessness among albinos as they were not getting any health care/financial support from health personal/social organizations. There were complains that many NGOs and local influential people visited them for their own personal interests/gains without providing any relief or support which resulted in personality changes observed in them. Some studies done in the developing world have cited similar reasons in affected families^{1,2,11,17,18}.

Many albinos referred to their cosmetic disability as one of the reasons for their joblessness. However, some were doing religious services but either without financial gains or with minimal salaries not enough for survival. This area was rural and under developed and there were either not many job opportunities available or they were outdoor such as farming or labor work which could not be performed by them⁴. This unemployment and the fact that many members in a single family were affected with the disease in some cases were factors further contributing to the financial/social pressures in the affected families. Similar economical and occupational limitations have been reported in developing countries in the past¹⁷.

Health education and counseling regarding the disease, especially solar related complications, did not exist in this population. None of the albinos or their close relatives was given genetic counseling which was important because of the trend of intermarriages within their tribe. Preventive measures, such as barrier creams or wearing caps or long-sleeved light colored clothing to protect their skin, were not taken as they were unaware of

the importance of those measures. Even general practitioners and nursing/paramedics staff were not fully aware regarding the nature and the management of this condition.

Cultural beliefs stating that " children are born with a different color if their mother is unfaithful" are present in Africa ⁴ but were fortunately not present in this population. Maximum age of the albinos in this study was 35 years and no older albino was seen; however, three albinos younger than 45 years of age had expired due to the reasons not known to the relatives. They might have died due to the some complications of this disease.

This study would enable us to have a good knowledge of this disease and would help in making them self-reliant, more active participants and useful to the society. There are plans with the help of some NGOs (Non Governmental Organizations) to develop a model village as the Albino Model Village which would have a health centre and specialized education regarding this condition. It would have a vocational training centre for teaching/training different indoor skills so that they can earn their living. Genetic counseling could be provided regarding this condition and other genetically transmissible conditions to improve the general health of the population.

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