

Prevalence of albinism in the South African Negro

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Summary

Two hundred and six individuals (113 males and 93 females) affected with oculocutaneous albinism have been ascertained in 126 Black (South African Negro) families living in Soweto, Johannesburg. The prevalence is approximately 1/3900 and the carrier rate about 1/32. Autosomal recessive inheritance has been demonstrated for the condition and consanguinity has been shown to be an important factor in the different prevalences found in the different ethnic groups represented in the study. The sex ratio of 1,21 is not significantly different from unity for a sample of this size.

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Oculocutaneous albinism (OCA) is 'a hereditary defect in the metabolism of melanin resulting in the decrease or absence of this pigment in the skin, mucosa, hair or eyes'.¹ Among Negro peoples affected individuals have a chalky-white skin, blue or more often light brown eyes, and white or sand-coloured hair; they usually suffer from photophobia and nystagmus because of a lack of pigment in the eyes, and the condition is inherited as an autosomal recessive.

There are two distinct types of albinism, one in which tyrosinase is deficient (the so-called tyrosinase-negative (ty. neg.) type) and the other in which tyrosinase is present (the tyrosinase positive (ty. pos.) type). The two conditions are complementary, so that if a ty. pos. albino marries a ty. neg. albino the offspring will be normally pigmented. The two types can be differentiated clinically: the ty. pos. albino has darker straw-coloured hair, often many small spidery pigmented patches (ephilides) chiefly on the exposed parts of the body, and brown eyes; the ty. neg. albino has lighter hair, blue or light brown irides and no ephilides. The majority of Negro albinos seem to have brown eyes rather than the pale blue eyes found in the Caucasoid albino.

The intellectual maturity of the albino has been studied, and albinos were found to have average intellectual maturity with a similar range to that of a matched Black control group.² In the same study it was shown that the albino's body image tends to be more clearly defined than that of the normally pigmented individual.

Albinos have a tendency to develop cancer of the skin, since they have no natural protection, in the form of skin pigment, against the ultraviolet rays of the sun. They also suffer from

impaired visual acuity. Socially, however, they appear to be well accepted, although this acceptance stops short of marriage and few albinos in Negro populations are married (according to data obtained in Barnicot's³ study in Nigeria, Vallois's⁴ study in the Cameroons and our experience in South Africa).

The incidence of albinism differs greatly among populations. The rate in Holland is 1/20 000,⁵ in Northern Ireland about 1/10 000,⁶ in Norway about 1/9 600,⁷ and in Nigeria about 1/5 000.³

The aim of the present study was to determine the prevalence of albinism in the population of Soweto, Johannesburg, as well as to investigate the prevalence among the various ethnic groups in Soweto; to establish its mode of inheritance; to investigate the consanguinity rate among parents of albinos; and to determine the sex ratio of the affected individuals.

Subjects and methods

The field work was carried out in Soweto, the Black satellite city close to Johannesburg. The city of Johannesburg, which includes Soweto, has a Black population of approximately 803 511 (National Census, 1970), made up of representatives of many of the rural groups from all over South Africa; the vast majority of these people live in Soweto. For the present study we shall use this census figure for Soweto, although it was estimated that census underenumeration in Soweto was approximately 17%,⁸ since this is the only total for which an ethnic group breakdown is available.

The subjects were ascertained by means of seven different methods. One of us (J.G.R.K.) visited every one of the 120 schools in Soweto, discussed the condition with principals and teachers, and asked for information concerning affected children. Similarly the 6 health clinics in Soweto, St John's Eye Hospital and Baragwanath Hospital were visited and information was requested there. Blank lists were left at these places with a request that the names and addresses of any affected patients be recorded. Welfare organizations and municipal social workers were also approached for information. The last method of ascertainment was through families with albinism, and as the survey progressed they came forward with information concerning other albinos they knew.

After the subjects had been ascertained each was visited personally and interviewed, usually with the help of an interpreter. A predesigned facesheet was completed indicating name, address, age, sex, ethnic group, occupation, whether or not the parents were related, number, names, and ages of the siblings, and whether any of them were albinos and whether or not there were other albinos in the family. Frequently not all of this information could be obtained in the first interview, in which case follow-up interviews or interviews with other members of the family were arranged. The subjects were also requested to present themselves for a medical examination at our laboratory. They were then examined, photographed and given a special anti-actinic cream for use on the exposed parts of the body to prevent skin cancer. Blood samples were collected from the albinos and as many family members as possible; these have been typed for a wide range of genetic polymorphisms as part of a study designed to establish linkage between them and the locus (or loci) for OCA.

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The present study does not take into account the two different types of OCA, and the data in this paper refer to all those individuals who could be clinically defined as having oculocutaneous albinism. Studies are, however, underway to provide information which will permit a more refined analysis on the basis of the ty. neg. and ty. pos. types.

Segregation analysis was carried out assuming complete ascertainment by the *a priori* method of Hogben as outlined by Emery.⁹ The data were also subjected to analysis by the much simpler 'singles' method of Li and Mantel.¹⁰

Results and discussion

As a result of the different methods of ascertainment 213 individual albinos were found in 126 different families, and 206 of these were personally seen by one of us (J.G.R.K.). The remaining 7 were either in the rural areas or away from home at the time of the interviews.

For the majority of the subjects, answers were obtained to all the questions set out on the facesheet. Information on consanguinity, however, was more difficult to obtain and occasionally where parents were dead or unknown the children and relatives could not respond to this item. Data on consanguinity were therefore obtained for only 90 of the 126 families.

Prevalence

The prevalence of the condition was found to be 1/3 900, using the data for those personally seen by the authors and using the population figure as reported in the census. The carrier rate is therefore 1/32 and the gene frequency is 0,0160 for this population, as calculated using the Hardy-Weinberg Law.

This prevalence is substantially higher than that reported for all Caucasoid population groups, but it is similar to that reported for Nigeria (1/5 000)³ and the Transkei (1/4 000).¹¹ It is, however, much higher than the rate of 1/15 430 reported by Hitzeroth and Hofmeyer¹² for albino females in the Black population of South Africa.

Four pairs of twins of whom both were albinos were found. This gives a figure of 1/53 births, which is not significantly different from the rate of 1/37¹³ for twin births in the general Black population ($\chi^2 = 0,45$; $P > 0,50$).

Prevalence among the various ethnic groups

Prevalence was calculated for each ethnic group in comparison with the total number of each group living in Johannesburg as reported by the National Census (1970) (Table I). It was found that the highest prevalence occurred in the Southern Sotho

group, for which the rate was 1/2 254. The Swazi group showed the second-highest rate, 1/2 716, but this was a small group of only 29 000 individuals as compared with the Zulu, Tswana and Southern Sotho groups, which are each represented by over 100 000 members living in Soweto. The third-highest rate occurred among the Tswana people, i.e. 1/3 481 people. The lowest rates were encountered in the Shangaan, Pedi, Venda, Xhosa, Ndebele and Zulu peoples.

Chi-square tests were performed to find out whether there were any significant differences between the rates in the various ethnic groups. There were no significant differences between any of the 3 Nguni groups (Zulu, Xhosa and Swazi) but the differences between the Xhosa/Zulu and the Southern Sotho were significant at the 1% level. This information showed a pattern very similar to that observed by Hitzeroth;¹² he found that the Tswana, Swazi and Southern Sotho groups showed the highest rates of albinism and the Venda, Xhosa and Zulu the lowest rates. Hitzeroth also found highly significant differences between the Nguni and Sotho-speaking groups.

Hitzeroth obtained his information from the files of those females who reported to the Bantu Reference Bureau of the former Department of Bantu Administration and Development, and his prevalence figures were on the average three times lower than those found in the present study. For example, the Tswana group was found to have a rate of 1/900 and the Zulu a rate of 1/15 705 compared with rates for the same groups of 1/3 480 and 1/4 459 respectively found in the present study. It seems likely that there were many albino women who did not present themselves for registration at the Bureau and who were therefore omitted from Hitzeroth's compilation.

Although the present study is based upon the Soweto population, which is about second generation in the city, the prevalence estimated here may still be somewhat lower than the actual prevalence, either because albinos may prefer to remain in the rural areas where they can maintain a relatively sheltered existence or because they may have been sent to the rural areas as children to live with relatives. On the other hand it may be that albinos are over-represented in the urban areas, since they may come to the industrial areas to seek employment. They may consider themselves unsuited for work in the rural areas because of the detrimental effect of exposure to the sun, which is probably greater when people perform manual labour in these areas. It has been found that albinos in the Cuna Indian society fitted much better into a community where a variety of indoor work opportunities was available.¹⁴ If this is the case in South Africa, the estimated prevalence figures for Soweto may be too high.

Mode of inheritance

The number of sibships included in the present study was 126, and there were 213 affected and 467 normal individuals. The

TABLE I. PREVALENCE OF ALBINISM BY ETHNIC GROUP

Ethnic group	No. of albinos	Johannesburg population	Prevalence	Gene frequency
Zulu	55	245 258	1/4 459	0,0149
Xhosa	17	81 511	1/4 794	0,0144
Swazi	11	29 872	1/2 716	0,0192
Ndebele	3	13 593	1/4 531	0,0148
Tswana	42	146 184	1/3 481	0,0169
S. Sotho	53	108 190	1/2 041	0,0211
Pedi	8	77 604	1/9 700	0,0101
Venda	6	31 936	1/5 323	0,0137
Shangaan	2	57 228	1/28 614	0,0059
Other	0	12 135		
No information	9			
Total	206	803 511	1/3 900	0,0160

data subjected to analysis by the method of Hogben⁹ are presented in Table II, and the expected numbers of affected individuals together with the variances are also shown. The difference between the observed and expected is 6,5, which is 0,70 times the standard error (9,36). There is therefore close agreement between the observed and expected numbers of affected siblings, indicating autosomal recessive inheritance.

TABLE II. OBSERVED AND EXPECTED NUMBERS OF ALBINOS ACCORDING TO SIBSHIP SIZE IN 126 SOWETO FAMILIES

Size of sibships	No. of sibships	Total No. individuals	No. of affected individuals	
			Obs.	Exp. (variance)*
1	11	11	11	11,0 (0)
2	12	24	13	13,7 (1,5)
3	17	51	22	22,1 (4,5)
4	8	32	10	11,7 (3,4)
5	18	90	19	29,5 (9,0)
6	18	108	33	32,9 (13,0)
7	16	112	34	32,3 (15,5)
8	8	64	20	17,8 (9,4)
9	7	63	20	17,0 (9,7)
10	3	30	7	7,9 (4,8)
11	3	33	8	8,6 (5,4)
12	3	36	8	9,3 (6,1)
13	2	26	8	6,7 (4,5)
Total	126	680	213	220,5 (87,6)

* Expected numbers calculated assuming complete ascertainment. Obs. = observed; Exp. = expected.

Using the 'singles' method of Li and Mantel¹⁰ of testing for recessive inheritance when there is complete ascertainment, the data were shown to be equally compatible with the recessive hypothesis. The number of singles was 77 and the proportion of affected individuals in the 126 sibships was 0,23 with a standard error of 0,02. This method is, in the opinion of its originators, just as reliable as more involved methods.

Consanguinity rate among parents of albino children

The consanguinity rate, calculated from the data obtained on the 90 families, was 24% (see Table III). This figure appears to be high, but the incidence of parental consanguinity for the parents of albinos in 21 sibships in Japan was nearer 50%.¹⁵

When the figures for the different ethnic groups were analysed, however, it was found that among the Tswana almost 42% and among the Southern Sotho 27% of the matings of parents were consanguineous, whereas only 4% of the Zulu matings were between relatives. The only significant difference between the major groups was that shown between the Zulu and the Tswana, which was significant at the 2% level. These data offer some indication as to why the Tswana and Southern Sotho show a high prevalence of albinism and the Zulu a low prevalence. It is probable that the gene frequency is actually similar in the different ethnic groups, but the different prevalences are due to the number of consanguineous matings in some populations as opposed to others.

Among the Southern Sotho and the Tswana people consanguineous matings are preferred. Schapera¹⁶ quotes Tswana proverbs which indicate two of the reasons for consanguineous marriages: 'Child of my paternal uncle, marry me, so that the cattle should return to our kraal', and 'side by side with his cousin a man is always happy'. The marriage partner is therefore generally chosen from among those close relatives who would cause the cattle given as *bogadi* (similar to the *lobola* of the Nguni groups) to remain in the family, or from among those relatives through whom the family can increase its political influence. The latter type of marriage occurs particularly among the 'nobles' of the Tswana and Southern Sotho people. According to Kuper¹⁷ the percentage of consanguineous marriages is about 20,5% in Southern Sotho people and probably more than this among Tswana groups. When, as is already happening, the local political systems and traditional beliefs break down, the need to marry a relative will be diminished and it may be anticipated that in the future, in the urban areas at least, this consideration will no longer operate and a decrease in the prevalence of albinism among Sotho-speaking peoples might be expected.

Among the Zulu and other Nguni groups marriage between relatives is virtually taboo, and no marriages or sexual relations of any kind are allowed with blood relatives at least four generations removed on the father's or mother's side.¹⁸

The degrees of relationship between the parents are also shown in Table III. Of those parents who were reported as being related almost half (9 out of 22) were first cousins. Of the 9 families where the parents were first cousins 6 belonged to the Tswana group.

Sex ratio

It has been claimed by some writers^{3,5,19} that albinism occurs more frequently among males than among females. Froggatt⁶ found this to be the case in his Northern Ireland study (81 males

TABLE III. DEGREE OF CONSANGUINITY AMONG PARENTS OF ALBINO CHILDREN, ANALYSED ACCORDING TO ETHNIC GROUP

Tribe	No. of families	Consanguinity			Total	Consanguinity rate (%)
		Cousins		More distant*		
		1st	2nd			
Zulu	23	1			1	4,3
Xhosa	10			2	2	20,0
Swazi	6	1		1	2	33,3
Tswana	24	6		4	10	41,7
S. Sotho	22	1	1	4	6	27,3
Pedi	2				0	—
Venda	2		1		1	—
Shangaan	1				0	—
Total	90	9	2	11	22	24,4

* Exact relationship unknown to respondent.

and 57 females), with the difference significant at the 5% level. He felt that the reason for the sex differences was social and that albino males tend to come forward to be counted more readily than females.

The present study ascertained more males than females (Table IV), but the sex ratio of 1,21 does not significantly deviate from unity. Oettlé's¹¹ study in the Transkei showed a tendency in the opposite direction; he found more females than males, with a sex ratio of 0,08. These tendencies may be explained by the nature of the populations in which these studies were carried out. The normal Johannesburg Black population consists of more males than females (421 170 males to 382 341 females; National Census, 1970). South African rural populations, on the other hand, consist of more females than males because the latter migrate to the towns in search of employment. Barnicot³ also found more male than female albinos in his Nigerian study, but this was possibly due to his methods of ascertainment and to the fact that males tend to volunteer for such studies more frequently than females.

TABLE IV. SEX RATIO OF ALBINO SUBJECTS

Population	Males	Females	Ratio	Reference
Soweto, RSA	113	93	1,21	Present study
Transkei	92	104	0,89	Oettlé¹¹
Nigeria	24	17	1,42	Barnicot³
N. Ireland	81	57	1,42	Froggatt⁶

Conclusion

A study of the prevalence of oculocutaneous albinism among the South African Negro inhabitants of Soweto has been carried out. Through seven different methods of ascertainment 206 affected individuals, 113 males and 93 females, were found in 126 families. The Black population of the area is approximately 803 511. The prevalence figure for albinism is therefore 1/3 900 and the carrier rate approximately 1/32. The number of albinos ranged from 1/2 254 in the Southern Sotho to 1/4 700 in the Xhosa, 1/9 700 in the Pedi and 1/28 614 in the Shangaan people. Consanguinity was found to play an important role in the prevalence in the various ethnic groups. The rate of consanguineous matings was 41% in the Tswana and only 4% in the Zulu group. The Tswana were noted to have a social system in which marriages between cousins are the preferred marriages

in the community. In the total sample, among the parents of albinos for whom information was available, there were 24% of consanguineous matings of various types.

Segregation analysis confirms that the data are compatible with a recessive mode of inheritance.

The study highlights several areas in which further research is required. Whether the high prevalence of the albino gene in this community is caused by some heterozygote advantage of the carrier or is due to random genetic drift in the form of founder effect is uncertain. Oettlé¹¹ has suggested that the carrier may have a paler skin colour than the normal individual and so may have a social advantage. These and other questions are puzzling, and studies are already underway in order to try to provide answers to them.

It can be concluded that the rate of albinism in the local population is higher than that reported for any Caucasoid population, marginally higher than the rates reported in other African countries, and shows a great variation among the various ethnic groups which go to make up the people of Soweto.

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