# Children with oculocutaneous albinism in Africa: Characteristics, challenges and medical care

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Oculocutaneous albinism (OCA) is an inherited condition characterised by significantly reduced pigment in skin, hair and eyes, visual defects and an increased risk of skin cancer. In the South African black population, 1 in 4 000 people is affected. Quality of life in children with albinism is influenced not only by health problems, but also by stigmatisation, rejection and cultural issues. This review aims to explore the latest literature available on the epidemiology, genetics, clinical characteristics, psychosocial issues and possible management strategies, focusing on affected children. The knowledge provided here is required of health professionals if a more fully informed service is to be offered to these children and their families.

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Oculocutaneous albinism (OCA), an inherited single-gene condition which occurs in all ethnic groups worldwide, includes a number of autosomal recessive disorders affecting pigmentation. OCA is rare globally, but the prevalence is much higher in Africa than elsewhere. While there are several types of albinism, the most common in Africa is OCA2.[1] The gene that when mutated causes OCA2, used to be called the *P* gene, but is now referred to as the *OCA2* gene. This gene is on chromosome 15q11-12. A 2.7kb deletion in the gene accounts for most (~80%) *OCA2* mutations in southern Africa.[2] Owing to lack of understanding and acceptance of the condition in many African communities, as well as the stigmatisation of people with albinism and lack of adequate healthcare for them, the quality of life of those affected is often poor.

For the present article, relevant reports (particularly those from sub-Saharan Africa) on the epidemiology, genetics, types and characteristics of OCA in black African populations were sourced from national and international publications and from the recently published book *Albinism in Africa*,[3] and reviewed. In addition, the published literature on the psychosocial issues associated with the condition, on community attitudes and cultural background, as well as the health challenges for affected children and their families, were explored and articles were selected for inclusion and discussion. Lastly, information on general management strategies focusing on affected children in southern Africa was sought and the findings outlined. The aim of this review was to gather information on children with OCA in the black population, with regard to epidemiology, genetics, clinical characteristics, and psychosocial and management issues.

## Epidemiology

One of the first South African surveys on albinism was performed by Dr G Oettle[4] in the Transkei and Ciskei regions, and it showed a prevalence of ~1 in 3 759. A similar rate of 1 in 3 900 was found in Soweto, the large, urban 'township' south-west of Johannesburg which comprises an almost exclusively black population.[5] The latter study showed that the prevalence of OCA varied in different local ethnic groups, and it appeared to be more common in those of Sotho extraction than in those of Nguni origin. One exception was the Swazi, who, although included in the Nguni group, had a higher rate of albinism, partly due to a higher rate of consanguineous marriage.

Countries neighbouring South Africa (SA) also have high rates of OCA (Table 1). In the SA population of 56 million people,[6] ~14 000 have albinism. The World Health Organization estimated, although actual data were limited, that prevalence rates across Africa varied from 1 in 5 000 to 1 in 15 000 and tens of thousands of people in sub-Saharan Africa probably have albinism. Their clinical care consequently poses challenges to health departments across the continent.[7]

## Types of albinism

Seven types of albinism have been reported from various countries around the world, and these have been described in detail by Montoliu *et al*.[8] The OCA1-3 types occur in Africa, but the OCA4-7 types have not been reported in African populations to date. Only parents who both carry a mutation for the same type of albinism are at risk of having affected children.

OCA1, caused by mutations in the *tyrosinase (TYR)* gene, is the most severe albinism type. Affected individuals have no pigment in

**Table 1. Rates of albinism in southern African countries**

| Country | Rate | Size of population subgroup studied | Reference |
|---|---|---|---|
| Botswana | 1 in 1 307 | 18 300 (Isolated village) | Kromberg, 1985[9] |
| Namibia | 1 in 1 755 | 2 113 077 (Census population) | Lund and Roberts, 2018[10] |
| South Africa | 1 in 3 900 | 803 511 (Soweto) | Kromberg and Jenkins, 1982[5] |
| Swaziland | 1 in 1 951 | 160 000 (Hhohho district) | Kromberg, 1985[9] |
| Zimbabwe | 1 in 4 720 | 1 312 032 (School children) | Lund and Roberts, 2018[10] |

hair, skin and eyes, skin which is most susceptible to sun damage and vision which is severely compromised. However, this type is very rare in Africa. It has never been found in SA and, so far, only one African case, in Cameroon, has been reported and confirmed with molecular testing.[11]

OCA2, caused by mutations in the *OCA2* gene, is the most common type found in Africa; there are 3 sub-types.[1] OCA2A shows the least pigmentation, although some can accumulate with age, the hair might change from white at birth to yellowish later and eye colour from blue to light brown. Visual deficits are not as severe as those in individuals with OCA1, but the skin is still highly susceptible to solar damage and skin cancer. OCA2AE is similar to OCA2A, but slightly more general pigment might accumulate and large (1 - 2 cm) pigmented ephelides, or dendritic freckles, develop with age on sun-exposed areas, particularly the face. Infants with these 2 types look the same clinically and have a similar phenotype at birth.

OCA2B, or brown albinism, is rarer than OCA2A and OCA2AE. Individuals with this type have more skin pigment than the first 2 types, usually have light brown eyes, fewer visual defects, can tan more easily and have a lower risk of developing skin cancer.[1] They may be more difficult to diagnose at birth, as neonates in the black population in general can be quite pale in colour.

OCA3, or rufous albinism, is rarer in Africa than OCA2, and it seldom occurs elsewhere in the world. It is caused by mutations in the tyrosinase-related protein 1 (*TYRP1*) gene on chromosome 9.[12] Two point mutations together account for 95% of the mutations found in OCA3 in southern Africa. Prenatal diagnosis is theoretically possible, but the phenotype is not severe and uptake is not expected. Affected individuals have a very distinctive colouring with brick reddish skin; dark yellow, gold or reddish hair; brown eyes and a lower risk of skin cancer than occurs in OCA1 and 2.[12,13] The phenotype may not be recognised in infants at birth, but becomes obvious in childhood.

Where a family wants to find out what type of albinism a child has, the diagnosis can usually be made clinically in the first decade of life (with the aid of the comprehensive table provided by Kromberg *et al.*[1]). If doubt exists, the diagnosis can often, but not always, be confirmed by molecular testing, where such laboratory services are available.

## Genetics of OCA2

OCA is inherited as an autosomal recessive condition. Therefore, where a couple has a child with albinism, each parent carries one recessive mutation for the condition. The recurrence risk is then 25%, or 1 in 4, that they will have an affected child, for each subsequent pregnancy. Genetic testing, using molecular techniques, can be offered to most at-risk family members (such as unaffected siblings of an affected person, who have a 67% chance of being carriers, and unaffected half-siblings who have a 50% chance of being carriers) and new partners (who, if they have no family history and are unrelated to the affected person, have a 3 - 4% chance of being carriers) of a parent with an affected child, who want to know their carrier status. Also, prenatal diagnosis is available, provided a family work-up has been performed to identify the familial mutations.[14]

Genetic counselling can be offered to those who wish to better understand the genetics of OCA, clarify their risks of recurrence or occurrence, and learn how to manage the psychosocial and health issues associated with the condition.

In SA, 1 in 30 black individuals carries a mutated copy of the *OCA2* gene, so ~1 in 4 000 infants is born with the condition. While 78% of OCA2 mutations are accounted for by a common 2.7kb deletion mutation at the *OCA2* locus,[2] no other common mutation/s have been described in this population group.

## Characteristics

The most obvious signs of albinism at birth are the pale skin and hair colour which is very striking in a black community. As the neonates open their eyes, the nystagmus is usually noticeable, and the red reflex, pale eye colouring, photophobia and lack of focus become apparent. About one-third of those affected have strabismus.[15]

### Skin cancer and sun damage

Both children and adults with OCA are highly susceptible to sun-induced solar and ultraviolet radiation damage. Skin cancer has been investigated in affected South Africans by several experts. Oettle[4] found that individuals with albinism represented 9.6% of all cutaneous cancers in black patients in the Transvaal, while Rippey and Schmaman[16] observed that 11% of their squamous and 41% of basal cell carcinoma cases, in the black population of Johannesburg, occurred in people with OCA.

In the 1980s, a cohort of 111 people with albinism was investigated in Johannesburg and 23% were found to have sun damage, skin lesions and solar keratoses.[17] The age range was 1 - 72 years and the rate of solar damage increased with age (Table 2). Altogether, 20% of children in the age group 1 - 19 years had precancerous skin lesions and/or solar keratoses. Also, those with ephelides (OCA2AE) had significantly ($p<0.01$) fewer skin lesions (33%, $n=8/24$) than those without ephelides (OCA2A) (67%, $n=16/24$). A second series of patients had skin biopsies and the head (e.g. cheek, eye and face) was found to be the most common site affected. Squamous cell carcinomas were about 5 times more common than basal cell carcinomas.[17]

### Visual defects

OCA is associated with the absence or significant reduction of melanin synthesis and generation of the melanin intermediate L-3,4-dihydroxyphenylalanine (L-DOPA), which disrupts normal embryonic development of the visual system. This disruption causes changes in the architecture of the eye, deficits in the retinas and optic tract misrouting.[18,19] The result is poor binocular and stereoscopic vision, reduced visual acuity, refractive errors and often myopia.[20] The anatomical changes to the visual system cause hypopigmentation of the irides, retinal pigment epithelium and choroid, as well as foveal hypoplasia and abnormal decussation of the optic tract. The lack of pigment in the choroid of a child with albinism can be observed using high-definition photography[21] and it is readily noticeable clinically.

Almost all children with albinism have nystagmus, which is usually horizontal, but occasionally vertical or rotational. It becomes evident soon after birth, but generally improves with age. OCA is also associated with hypermetropia, myopia and astigmatism,[22] as well as photophobia.[23] Correction of refractive errors with spectacles can optimise visual acuity, but not restore it to normal levels.[22] Most affected people benefit from prescription spectacles and testing

**Table 2. Presence of sun damage and/or skin cancer in people with albinism by age (Kromberg *et al.*[17])**

| Age group, years | Total | Sun damage present, *n* (%) |
|---|---|---|
| 1 - 9 | 32 | 2 (6.0) |
| 10 - 19 | 44 | 6 (14.0) |
| 20 - 29 | 14 | 3 (21.9) |
| 30 - 49 | 16 | 10 (61.5) |
| ≥50 | 5 | 5 (100) |
| Total | 111 | 26 (23.4) |

should be undertaken regularly for this purpose. Photophobia is mitigated by using dark glasses, although these further reduce visual acuity. Binocular devices are preferable for individuals with nystagmus, as monocular viewing can increase the amplitude of the nystagmus.

### Development in childhood

Children with congenital blindness and other visual deficits often show delayed motor development and slow milestones,[24] which is not a reflection on their intelligence. In the case of children with OCA, intelligence is within the normal range.[25] However, some milestones (such as sitting and crawling) were found to be delayed, probably related to reduced visual acuity, in a sample of local affected infants.[26] Where stimulation programmes are available, possibly at a paediatric neurology clinic, these children should be enrolled because, with a little extra stimulation, they can reach these milestones more timeously.

## Psychosocial issues

Parents with children with albinism may have to deal with various psychosocial difficulties from the birth of the affected infant onwards, owing to cultural beliefs and general ignorance which surround the condition. Further, they themselves might have very limited knowledge of the causes, genetics and clinical management of the condition (as was found to be the situation in local families with boys with haemophilia[27]) which makes it difficult for them to cope with the psychological and health issues that arise.

Children with albinism have several psychosocial problems which they have to face as they develop. Initially, there may be some maternal and paternal rejection which might have a long-term impact on their psychological well-being. Then there might be slightly delayed milestones owing to poor visual acuity. As the child grows and starts socialising with other children, there is likely to be some discrimination, distancing and rejection in the community. Stigmatisation often occurs at school but may change to partial acceptance as the other children become aware that the affected child is no different from themselves (apart from skin colour and visual problems). Teachers need to be aware of these issues and should adopt ways of making life easier for affected children, both in the classroom and in their social interactions. The children themselves need to be assisted in understanding their condition, becoming more assertive and requesting the help they require.

### Maternal-infant bonding

When an infant with albinism is born, the parents may experience feelings of shock and distress. In the case of the mother, delayed maternal-infant bonding may occur.[26] Nurses in the delivery ward may be uncertain how to react and sometimes reluctant to touch and handle the infant, suspecting that the condition might be contagious.[28] They may also convey inaccurate information about the condition to the parents.

At first, the mother may be reluctant to hold and breastfeed the infant and may show signs of debilitating depression and sleeplessness.[26] However, by 3 months after delivery, the mother generally starts to accept and bond with the child and is more willing to cope with feeding and holding them close. Bonding is difficult in this situation, as it depends on direct visual contact between mother and child, which is hindered by the presence of the nystagmus, visual deficits and reduced ability to focus in the child. Also, the partially rejecting mother may not be willing to look closely at the child owing to her anxiety and reluctance to accept that she has produced an affected infant. However, by 9 months after delivery, the mother's unhappiness has generally abated (although there is evidence that some mothers experience chronic sorrow) and maternal-infant interaction is comparable to that in other mothers with unaffected infants.[26]

While there is little research on fathers of a child with albinism, mothers have reported that fathers were shocked and upset by the birth, sometimes questioning the mother's faithfulness, denying paternity and occasionally deserting the mother and child.[26] However, where the father has a family history of albinism, he is more likely to accept the child. In other cases, the father may progress through various emotional stages and reach acceptance of the child either around a similar time to the mother or later.

### Intellectual maturity and body image

Studies have shown that children with albinism have intelligence well within the normal range. Beckham[29] compared intelligence in a group of children with OCA with that found in their unaffected siblings, and there was no significant difference. Similarly, Stewart and Keeler[30] found no evidence to support the idea that the condition was associated with diminished intelligence. Manganyi *et al.*[25] investigated intellectual maturity and body image differentiation in black SA children with OCA and a matched control group of unaffected children. The results showed some significant differences ($p<0.02$) between the groups: the former group showing slightly higher levels of intellectual maturity than the latter. The better performance of the affected children may have been related to compensatory mechanisms, such as those mentioned by Keeler[31] who suggested that their 'inability to compete physically often leads them to sedentary and intellectual pursuits'.

The findings of the study[25] on body image indicated that, although both groups showed body concepts intermediate between relatively well differentiated and diffuse, children with OCA generally expressed a more differentiated body image than the controls. The authors suggested that body image may be an artifact of intellectual maturity and subject to the same dynamic mechanisms. However, the affected children had more problems creating a self-image than undertaking the other drawings (two human figures, a male and a female). This finding suggests that they faced a negative self-evaluation (identity), and could not significantly influence their projections.

### Attitudes to albinism

Although attitudes to albinism appear to be perceived as generally negative in black communities, they tend to vary from fear to adoration and from infanticide to reverence. Reports of affected people being protected at a king's court in Central Africa contrast with those that they were seen as monsters by the local communities.[32] Livingstone[33] described the attitudes he observed during his travels in Africa, from infanticide in Botswana to the favouring, elsewhere, of affected people as doctors by certain chiefs. They were sometimes ascribed special physical or spiritual characteristics, which also set them apart and prevented their full integration in society.

More recently, attitudes were investigated in a group of teenagers with OCA ($N=35$) and a group of matched unaffected controls in Soweto, Johannesburg.[34] A significant difference ($p<0.05$) was detected in the male group, in which control subjects were more negative towards albinism than were affected males. Regarding the social distance items, responses were generally quite positive but, as social distance was reduced, the difference between the groups grew wider. The most significant finding was that associated with the death myth: nearly half the controls (43%, $n=15$) believed that people with OCA vanished at the end of their lives and did not die natural deaths; a further 8 (23%) were unsure what happened,

while a third of those affected (31%, *n*=11) reflected this strong community belief and were not sure what would happen at the end of life. The development of more positive community attitudes is probably hampered by the nature of this widely prevailing and invasive myth. The origin of the myth lies in antiquity, but may be associated with the special significance of the colour white for many local ethnic groups.[34,35] White beads, skins and cow tails are used by traditional healers when communicating with the spirits. Further, as people with albinism are white (the colour of the spirits) and all other people are black, and as spirits cannot die, it may be that this belief became associated (possibly to explain why these individuals were born the colour they were) with affected people at some time in the past.

One way to change these negative attitudes would be to make sure that affected people are better integrated into their community so that others get to see, become more familiar with, understand and accept them. Linn[36] observed that discrepancies between verbal attitudes and overt behaviour are seen to be 'a function of the level of involvement with the attitude objects, as well as the amount of prior experience with it.'

### Discrimination and stigmatisation

Children with albinism face social discrimination from birth. Mothers observe such discrimination when taking their infant out in public for the first time.[37] Stigmatisation reflects not only on the child, but also on the mother, father and sometimes the family as well. The parents have to find answers to questions about the child that will not only allay the suspicions of relatives, but will also negate their beliefs in prevalent community myths. When affected children start attending school, they might find that no-one wants to sit next to them or be their friend.[38] Stigmatisation can take many forms from total and aggressive rejection (e.g. throwing stones) to more subtle avoidance and name-calling. With the help of a supportive family, children have to learn to cope so that they do not resort to withdrawal from interaction with others, and their self-concept and self-respect are not damaged. To assist them, families with an affected child often have to become advocates for albinism.

### Cultural beliefs: the impact on people with albinism

Owing to the recent spate of attacks on people with OCA which, although much more common in Central Africa have begun to spread to SA, families with an affected child, particularly in rural areas, have to be aware of the dangers of the child moving around in public without protection. The attacks are motivated by the desire of traditional healers for what they believe to be powerful medicines, made from the tissue or bones of people with albinism. Attacks on defenceless affected children have predominated, as their tissue is believed to make the most powerful medicine and they are easier to abduct.[39] For this reason, parents have to be aware of the risks and take necessary precautions. However, both the public and traditional healers are in need of enlightening education, so that they recognise the absurdity of this mistaken belief and do not participate in perpetuating the myth and in making or purchasing such 'medicine'.

Similarly, the death myth must be shown to be a superstition. Appropriate community education programmes should expose these issues as erroneous and reiterate that people with albinism are like other unaffected people, except for skin colour and visual problems, and subject to life and death processes as any other person.

## Medical care strategies and recommendations

Children with albinism should have their needs recognised and addressed early, so that the intervention opportunities of childhood are not missed and their quality of life is not negatively affected.

Ophthalmologists recommend that the child has regular visual assessments, preferably starting at 6 months of age, but definitely before starting school. Parents should be encouraged to have the child assessed and should then help the child to learn to use prescription spectacles, as well as dark glasses (with polarised lenses and UVA and UVB protection[22]), where appropriate. The eyes can also be protected by the daily use of wide-brimmed hats, even in the classroom, if necessary.

Dermatologists strongly recommend that sun-screen cream of sun protection factor (SPF) 30+ use starts in infancy, as soon as children are exposed to the sun. Children and parents need to accept that the use of sun-barrier cream on exposed areas of the body is a life-long necessity and that sun exposure during the midday hours should be avoided wherever possible. Anti-actinic cream should be applied 15 minutes before entering sunny areas and the skin should be protected by heavy cotton clothing, long-sleeved shirts, trousers and hats. The lips of children with albinism are also subject to sun damage and need protection by means of moisturising barrier lip balm with SPF >30. If signs of sun damage occur, the child should be taken for treatment as soon as possible.

Some children with OCA are enrolled in schools for the partially sighted (usually with boarding facilities); however, this should not be necessary. If teachers in community schools are informed on how to assist affected children to benefit from schooling, they should be able to attend their local schools. When they start school, the children should have access to small telescopes and magnifiers. Teachers should be advised that the child needs to sit in the front of the classroom, be allowed to walk up to the board if necessary, be encouraged to use visual aids and be seated away from glare. Children should be allowed to hold books near their eyes and use an abnormal head posture, if necessary. They should also have large-font text in tests and examinations, and should be given more time to complete tests. In addition, teachers should educate fellow students to ensure a greater understanding of the condition and the child who has it.[22] With appropriate teacher-pupil education, affected children should be able to keep up with their classmates at school. Useful guidelines for optimising vision in children have been drawn up by Kammer[20] and McBride.[22]

Affected children, once old enough to understand, should be informed about the genetics and complications of their condition and how to explain it to others. They also need to accept that there are cultural beliefs about albinism in most communities, which can make life difficult for them, and they need to learn how to counter these beliefs and manage ensuing problematic situations.

Genetic counselling should be provided, so that parents and their children understand the nature and cause of albinism, the mode of inheritance and prognosis of the condition, and can explain these facts to all who ask them. Social workers and clinical psychologists suggest that parents have counselling so that they can help their children face the stigmatisation and discrimination they are bound to encounter. Children could benefit from appropriate supportive therapy. Further, joining up with an albinism support group (run by the Albinism Society of South Africa) is often found to be very beneficial.

## Conclusion

OCA is a relatively common genetic condition locally. Affected children need regular ophthalmology and dermatology follow-up so that medical issues can be identified in the early stages when appropriate treatment can provide the most benefit.

Because it is so obvious, but often unexpected and little understood, albinism is surrounded by myths and superstitions, particularly in an African population. Children with the condition – if they are well

accepted by their family and community, if they are encouraged to take the necessary preventive measures to avoid succumbing to the visual and skin complications, if they learn to cope with the discrimination they experience, and with a little help from informed health professionals and educators – should be able to enjoy a good quality of life.

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