

# Oculocutaneous albinism: an African perspective

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## Abstract

**Aim:** To describe the genetics behind oculocutaneous albinism (OCA), and explore OCA in an African context in terms of the effects on the health and education of individuals with OCA.

**Methods:** A literature-based review was conducted using Pubmed. Searches were restricted to English-based publications, focusing on OCA in Africa.

**Results:** The genetics behind OCA and the effects of OCA in terms of visual impairment and skin cancer are explained along with a description of what low vision services and low vision aids are available to those with OCA in Africa.

**Conclusions:** The review concludes with useful advice to those with OCA and those working with or teaching students with OCA.

**Key words:** Africa, Low vision, Oculocutaneous albinism

## Introduction

Derived from the Latin word *albus* meaning 'white', oculocutaneous albinism (OCA) is a relatively rare genetic disorder in which the body does not biosynthesise melanin normally. Melanin is a photo-protective protein whose role in the skin is to absorb ultraviolet (UV) light and prevent damage; without melanin the skin is more prone to sunburn and skin cancer. Lack of melanin results in a triad of signs – pale skin, light hair and pale iris colour – but the consequence of OCA is impaired visual acuity (VA). Visual impairment is caused by the irregular development of the nerve pathways and abnormal retinal development.<sup>1</sup> Children present with varying degrees of congenital nystagmus, hypopigmentation of the iris causing iris translumination, reduced pigmentation in the retinal pigment epithelium, foveal hypoplasia, large refractive errors and sometimes reduced colour vision.<sup>2,3</sup> The foveal hypoplasia causes the central cones to be spaced out, reducing central VA.<sup>4</sup> Misrouting of the optic nerves from excessive crossing of fibres at the optic chiasm can result in strabismus and reduced stereopsis.<sup>2</sup> Photophobia is a common complaint.

The purpose of this review is to describe the genetics

behind OCA, and explore OCA in an African context in terms of the effects on the health and education of individuals with OCA. The outcome of this review will be to provide useful advice to those with OCA and those working with or teaching students with OCA.

## Genetics

There are four different types of OCA, which result from a mutation in one of several genes (Table 1).<sup>1,2,4</sup> Each of these genes is chemically coded to produce proteins which are involved in the production of melanin. The mutation can result in the reduction of melanin production, or no melanin production.<sup>1</sup> All four types of OCA are autosomal recessive. Therefore if both parents are carriers of the mutated gene there is a 25% chance of them having a child affected with OCA and a 67% chance of their non-affected children being carriers.<sup>2</sup> A person with OCA will have non-affected children who all carry the mutated gene, provided the co-parent is not affected or carrying the mutation.<sup>2</sup>

It is possible to carry out genetic sequence analysis to determine whether a foetus has OCA; a sample for analysis would be obtained by amniocentesis at 16–18 weeks' gestation.<sup>5</sup> To genetically distinguish between OCA types 1B, 2 and 3, a sequence analysis of genes coding for tyrosinase, P protein and TRP-1 can be completed; however, this is not routinely available in the UK.<sup>5</sup> In Africa, genetics screening is widely unavailable; in fact there are only four human genetics screening and counselling departments in South Africa that provide services to patients from all over Southern and Central Africa.<sup>6</sup>

## OCA in the UK

In the UK, OCA is estimated to affect 3716 individuals (1 in 17 000 people).<sup>7</sup> Although the prevalence of skin cancer in the UK for individuals with OCA is unknown, the NHS advises that albinism does not alter life expectancy but does increase the risk of developing skin cancer.<sup>8</sup> Children with OCA present early in childhood to UK hospital eye clinics. Although OCA cannot be cured, in the UK children and adults will be provided with glasses or contact lenses if a refractive error is present and with low vision aids in the form of large-print books, magnifiers, telescopes and electronic vision enhancement systems (EVES) (CCTV, tablets and smart phones).<sup>8</sup> There has been some discussion on whether a tenotomy and re-attachment of the horizontal eye muscles can help reduce nystagmus amplitude and

**Table 1.** Summary of the classification of OCA types

Type	OCA type 1 (tyrosinase-negative)	OCA type 2 (tyrosinase-positive)	OCA type 3 (rufous)	OCA type 4
Gene mutation	Chromosome 11	Chromosome 15	Chromosome 9	Chromosome 5
Most frequently seen in	Caucasian descent	Sub-Saharan African descent	South African descent	East Asian descent
Signs	White hair; blue eyes; white skin	Hair may be yellow, auburn, ginger or red; eyes can be blue-grey or tan; skin is white at birth	Hair may be ginger or red; eyes are hazel or brown; skin is reddish brown	Hair may be yellow, auburn, ginger or red; eyes can be blue-grey or tan; skin is white at birth
Changes with age	<i>Type 1b</i> : Can start to produce melanin in childhood; hair changes to golden blonde; skin tans slightly; irises change colour and become less translucent <i>Type 1a</i> : remains unchanged and does not produce melanin with ageing	With sun exposure skin may develop freckles, moles or lentigines		With sun exposure skin may develop freckles, moles or lentigines
Vision	6/60 (logMAR 1.0) or less in type 1a and 6/36 (logMAR 0.8) in type 1b	May reach 6/18 (logMAR 0.5)	May be normal as the hypopigmentation is not sufficient to alter retinal development	May reach 6/18 (logMAR 0.5)
Symptoms	Photophobia is common			

thus improve VA, but the NICE guidelines advise that there is currently insufficient evidence on its efficacy.<sup>8</sup> Those with OCA remain under the care of the ophthalmologist/orthoptist during their childhood so that their vision can be monitored; and then under the care of their optometrist for refractive correction and sunglasses/tinted lenses to help with photophobia for the rest of their life. Individuals in the UK diagnosed with OCA have access to genetic counselling with a geneticist and can be referred to a dermatologist for assessment of their skin and advice regarding skin protection creams available on prescription.<sup>8</sup> There are a number of UK charities which provide information and peer support to those affected by OCA or the parents of children diagnosed with OCA.<sup>8</sup> These charities include The Albinism Fellowship ([www.albinism.org.uk](http://www.albinism.org.uk)), National Blind Children's Society ([www.nbcs.org.uk](http://www.nbcs.org.uk)) and Nystagmus Network ([www.nystagmusnet.org](http://www.nystagmusnet.org)).<sup>8</sup>

### OCA in Africa

The total population of individuals with OCA in Africa is unknown and the incidence of OCA will vary both between countries and within regions. In Tanzania, the OCA population is estimated at 31 345 (1 in 1429 people), which is nearly 8.5 times the UK OCA population.<sup>9</sup> Zimbabwe has an OCA population similar to the UK, estimated at 3050 (1 in 4000–5000 people), but its total population is a quarter of the UK's.<sup>9</sup> In South Africa's Venda region the incidence is 1 in 1970; two clans with the same number of individuals with OCA have an incidence of 1 in 825 people in the Vhatavhatsindi clan but 1 in 3107 people in the Vhalaudzi clan.<sup>9</sup> The higher incidence in the Vhatavhatsindi clan is

attributed to them living in a relatively isolated and inaccessible area with a restricted gene pool, which leads to a high level of intra-community breeding.<sup>9</sup>

In comparison with Western countries, OCA in Africa has significant health consequences. In Tanzania half the albino population will develop advanced skin cancer between 20 and 30 years of age, with less than 2% of albino children in Tanzania living to be 40 years of age.<sup>7</sup> The head and the neck are the most affected sites, with squamous cell carcinoma the most common type of skin malignancy seen with OCA; in contrast in a white population basal cell carcinoma was the most common variant.<sup>10–12</sup> Late presentation with advanced skin cancer and lack of therapeutic facilities such as radiotherapy and chemotherapy result in early mortality.<sup>10</sup> In one study men were affected with skin cancer more than women, which the authors suggested was because men tended to work outdoors,<sup>10</sup> but in other studies an equal proportion of affected men and women were reported.<sup>11,12</sup> In a retrospective study of 64 patients with OCA who had skin cancer treated in a hospital in Tanzania over a 9-year period, 83% of those presenting were 21–40 years old and 81% presented with tumours more than 5 cm in diameter; the duration of the illness was a median 24 months.<sup>10</sup> In this hospital, where surgery was the only treatment option available, surgical site infection was the most common complication (55% of the 20 cases with complications), and there was a mortality rate of 6%.<sup>10</sup> The authors found a re-occurrence rate of 30% and reported that this was as a consequence of delayed presentation and failure to complete treatment.<sup>10</sup> A 2-year retrospective review of hospital charts in a Nigerian hospital reported that 20 albinos presented with on average 1.9 lesions but that 70% of the patients did not complete their treatment or were lost to follow-up, despite half of these patients

needing radiotherapy.<sup>11</sup> The Nigerian authors concluded that skin cancer was the major cause of morbidity among individuals with OCA in Africa and that poverty and under-education resulted in late presentation.<sup>11</sup>

In a special school for the visually impaired in South Africa, attended by 112 children with OCA, focus group interviews found that although the children knew 'creams' should be used on their skin, they did not understand the term SPF and as a result the application of sunscreen lotions was only 37.8%.<sup>3</sup> There was an insufficient supply of sunscreen lotions and the cost and access to the lotions made it prohibitive for them to use sunscreen.<sup>3</sup> Death from skin cancer is the most serious and likely consequence of OCA in Africa; poor supply and high costs of skin protection creams and under-education of those affected by OCA and their communities is resulting in advanced multiple skin lesions. Better education regarding skin protection is the primary preventative measure.

In a secondary school survey of 38 albino children living in South Africa, 74% did not know what caused albinism, with 79% reporting that their parents had never explained albinism to them.<sup>13</sup> In a Zimbabwe study of 138 children with OCA, only 10.9% of the students understood that OCA was an inherited condition; in fact 50.7% pupils stated they had no idea why their skin was pale.<sup>14</sup> This shows a high level of under-education regarding this condition. This high level of under-education regarding OCA was also reported following a WHO albinism pilot survey of 12 African countries, which found that people with OCA did not fully understand their condition.<sup>15</sup>

Black, yet white, people in parts of Africa have been shunned because of having albinism. OCA is coupled with stigma and superstitions; recently there have been 100 albinos murdered in Tanzania and their body parts sold on the black market.<sup>16</sup> OCA can cause extreme light sensitivity, which contributes to the physical and social exclusion of individuals with OCA.<sup>13</sup> In fact the South African school survey reported that 86% of the children with OCA felt they had fewer friends.<sup>13</sup> In conclusion, it appears that individuals with OCA in Africa must deal with more than just their visual impairment and potential health concerns from skin cancer. Not only must they cope with stigma and discrimination from within their communities due to the under-education of what causes OCA, but also the possibility of harm because of mythology that wealth can be generated from selling albino body parts and the poor legal structures in dealing with the resultant murders.

### Education of children with OCA in Africa

A questionnaire study of 38 albino children (aged 11–16 years) attending a special school for the visually impaired in South Africa found that 95% had received an eye test and 18% were wearing spectacles.<sup>13</sup> The school adapted for the children by allowing them to walk up to the blackboard to read and by using a mobile blackboard; the children also had access to CCTV, magnifiers and large-print books.<sup>13</sup>

In a second school survey by the same author in Zimbabwe, where the subjects were recruited from all

the primary and secondary mainstream schools in 5 of the 9 districts in Zimbabwe, 138 pupils completed a questionnaire and interview.<sup>14</sup> In this survey, 95.6% of the children reported the inability to see the blackboard or their school books. This resulted in delay in completing their school work and making mistakes.<sup>14</sup> Access to health care was different for the Zimbabwe children; only 45.7% had received an eye test with 24.6% wearing spectacles. Classroom adaptations for the children in the Zimbabwe group showed that 94.2% sat at the front of the classroom, they used large-print books, were allowed additional time to complete their work, they had their own books rather than sharing with classmates and they were given extra tuition.<sup>14</sup> However, 70% felt that no special provision was made for them in their schools.<sup>14</sup> None of the children reported using CCTV or magnifiers.

Interestingly, 84% of the South African children felt they had equal or greater intelligence than normally pigmented people and yet 87% of the children felt that children with OCA should be educated in a special school for their primary education and 74% for their secondary education.<sup>13</sup> The children would appear to be unaware that education through a special school rather than the local mainstream schools would contribute further to their social exclusion in the community.

### Use of low vision aids for OCA

Although the impact on VA is variable, the majority of albinos living in the UK will eventually be registered as either visually impaired or severely visually impaired. Low vision aids are prescribed based on the quality of VA and the goals of the individual. There are advantages and limitations to all low vision aids; however, in Africa the concerning limitations are availability and cost. At the Kwale District Eye Centre in Kenya, funding towards the costs of treatment is obtained from a number of charities including Eyes for East Africa, Sightsavers International, CBM International (formerly known as Christian Blind Mission) and individual donations.

OCA is associated with hypermetropia, myopia and astigmatism; correction of refractive errors with spectacles will optimise VA although not correct it to normal levels. Photophobia is helped by dark-tinted glasses; however, there is a further reduction in VA with dark lenses. A hat with a brim is helpful for reducing glare and photophobia, and has the advantage of skin protection for the face and head.

EVES provide maximum magnification through real image magnification; they allow the user to change the size, contrast and brightness of the material according to their needs. The devices allow a better working distance for near and intermediate tasks, an increased field of view, and in the UK would be the mainstay for visually impaired individuals in education and workplace environments. However, these devices are expensive to purchase, install and maintain and they require battery or electrical power sources, something still beyond most individuals with OCA living in Africa.

Telescopes can be used for near, intermediate and distance viewing. Telescopes can be hand-held or

spectacle-mounted to be hands free, and come in various sizes and shapes. Telescopes are ideally suited for short-term viewing, such as reading signs or a blackboard, and for locating a distant object. Telescopes can be used as binocular or monocular devices, and this is particularly useful for individuals with nystagmus, as the amplitude of the nystagmus increases with monocular viewing. Unfortunately, telescopes are still costly, have a reduced field of view and have limited magnification available; they can also be cosmetically unacceptable to many people. However, in my own personal experience with school children in Kenya, telescopes are welcomed; once the child and school teacher is given adequate training, they maximise the child's ability to view the blackboard, allowing them to keep up with their classmates.

For near magnification, individuals with OCA in Africa have two options available to them: hand and stand magnifiers. Hand magnifiers are the most common type of optical aid, as they are easily available and inexpensive. They have a wide range of magnification powers available up to  $\times 12$  (<http://www.eschenbach.com/products-hand-held-magnifiers.htm>). The aids are portable, allowing for use at home as well as at school. They are discreet and therefore more cosmetically acceptable to most people. They are available with illumination systems but this requires batteries or electrical power sources and increases the cost of purchasing and using the appliance. Unfortunately, hand magnifiers have disadvantages. Because they are hand-held only one hand is free, which limits manual tasks. They can be tiring to hold, and the correct working distance must be found and maintained; incorrect positioning affects the power, and accommodation will also alter the power. Also, as the magnification power of the device increases, there is a decrease in the field of view.

Stand magnifiers have a magnifying lens attached to some form of stand so that the magnifier sits on the page being viewed. The major advantages of the stand magnifier over the hand magnifier are that the correct working distance is always maintained and the hands are left free; in fact some have a high stand to allow the child to write in the books underneath the stand or the teacher to point to a word. The lens has good light-gathering properties, although illuminated stand magnifiers require batteries or electrical power sources. As with hand magnifiers they come in a wide range of powers up to  $\times 12.5$  (<http://www.eschenbach.com/products-stand-magnifiers.htm>). For children, stand magnifiers are useful for stabilising the effects of accommodation. However, stand magnifiers are more expensive than hand magnifiers, they are bulky and less portable, they are cosmetically more noticeable than a small hand magnifier and have a smaller field of view compared with a hand magnifier.

A unique study which documented the use of low vision appliances with blind East African school children found that after an accurate refraction 9.1% of students had no actual visual impairment, 21.7% were visually impaired and 69.2% were severely visually impaired or blind.<sup>17</sup> Of the 50 students who required low vision aids, 21 students found stand magnifiers easiest to use (range +20D to +76D), with only 2 students preferring a hand

magnifier of  $\times 6$  power; 19 students required only high reading adds (+6D to +20D) and 8 students preferred hyperoculars (+24D to +48D).<sup>17</sup> The researchers visited 5 blind schools in Kenya and Uganda in the mid-1990s and discovered that no low vision services or low vision aids were available in any of these schools for the blind. However, with more voluntary ophthalmic charities now working in Africa this situation may have improved. The authors reported that many of the teachers in these schools were visually impaired or blind themselves and as a result 57% of students with low vision were being taught through braille only despite 79% being capable of reading N5–N8 print.<sup>17</sup> This study highlighted the need for accurate refraction and access to glasses among the low vision population, as more than 50% of the group had a spherical or astigmatic refractive error greater than 2D and, as highlighted earlier, 9.1% did not have any visual impairment when glasses were prescribed and did not need to be educated in a school for the blind.<sup>17</sup> The authors concluded that in Africa it was easy to purchase or grind glass with powers up to +28D to make inexpensive lower powered magnifiers, and that these magnifiers along with accurate refractions and dispensed glasses would provide a cost-effective treatment to help the visually impaired read. They advised that when low vision aids were dispensed support, training and follow-up would be required, especially for children, and that higher powered aids would require additional supervision.<sup>17</sup>

#### **Low vision assessment of a school-aged child with OCA in Africa**

At the Kwale District Eye Centre, a teacher is employed who has been trained in children's eye health, visual development and low vision by the resident ophthalmologist and the visiting eye professionals who visit the centre for short periods. From his continual active learning about low vision and its management the teacher now works as a low vision therapist at the eye centre and as a visiting teacher for visually impaired children. This an invaluable role linking health and education to children, where in many situations it may be impossible for the children and their families to travel the long distances to the nearest eye centre.

When assessing a child with OCA it is important to identify the problems encountered by the child at home and in school; the child and parents' understanding of OCA, particularly regarding the cause of the albinism; and the effect on both their VA and skin. It is important to meet with the school principal and teacher to establish their attitudes towards OCA, and when necessary educate them diplomatically on the causes and consequences of OCA and how they can maximise the child's ability to see and study in their school. Once the child is assessed and the low vision appliance obtained which matches his or her needs, the child and teacher should be trained on how to use the appliance. The child should then be reviewed at a later date in the school to ensure that the appliance is being correctly used to maximise the child's VA.

## Conclusion

Sub-Saharan Africa is a socio-economically disadvantaged region that has the highest mortality and disability rates in the world, and the higher incidence of blindness in Africa compared with Europe is attributed to the difference in socio-economic status.<sup>18</sup> OCA has significant consequences on the health and well-being of individuals living in Africa, skin cancer being the leading cause for premature death in individuals with OCA. However, with the relevant education and proper skin protection, skin cancer could be prevented, delayed, or detected earlier and so treated effectively. Social exclusion and lack of acceptance of those with OCA by the community still occurs because of under-education about the condition. Programmes delivered in communities, preferably by those with OCA, detailing the cause and effects of OCA, will educate communities, remove superstitions and allow individuals with OCA to integrate and contribute to their communities. Individuals with OCA can perform well educationally when they are provided with accurate refractions and low vision aids to assist with school work and they have the ability to achieve equal status in their community; in 2010 the first individual with OCA was elected to the Tanzanian parliament.<sup>19</sup> Finally, the outcome of this review is to provide useful advice, and this can be found in Appendixes 1–3.

## Appendix 1. Recommendations for managing OCA in Africa

1. Engage with school teachers, principals and community leaders and involve them in the education of individuals with OCA about OCA.
2. Run community educational events, ideally with individuals with OCA who can offer peer support and guidance.
3. Ensure the individual with OCA has regular eye examinations to check for changes in their eyes, such as a change in glasses or cataracts developing.
4. Ensure that individuals with OCA are given skin protection creams, advice on skin protection, and wide brimmed hats. If possible the community nurses should examine the skin for early signs of damage and refer when necessary, and most importantly educate those with OCA about skin protection.
5. Try to raise funds for low vision appliances and skin protection creams.
6. When low vision appliances are provided, ensure that they meet the needs of the individual and are used correctly.

## Appendix 2. Advice for individuals with OCA in Africa

1. Always wear a wide brimmed hat to protect your head, face and eyes from the sun.
2. Wear sunglasses with 100% UVA and UVB protection; the darkness of the lens is not an indication of the protection. Darker tints will help reduce glare further.
3. Wear long-sleeved heavy cotton shirts, trousers and skirts to protect your skin from the sun.

4. Try to avoid intense sunlight, especially in the middle of the day.
5. If possible use skin protection cream with SPF 30+, applying it 15 minutes before entering sunlight. Skin protection creams are necessary for parts of your body not protected by clothes, such as your hands, face and feet.
6. Try to find out about OCA, what causes it and how it affects you, then educate others about OCA. This will reduce the stigma and superstitions that you may face.
7. As a student, talk to your teacher about what you can and cannot see; your teacher will want you to maximise on your education.

## Appendix 3. Advice for teachers with an OCA student in Africa

1. Always sit the student at the front of the class, in the middle of the room, away from windows.
2. Allow the student to wear a hat indoors if the room is very bright.
3. Allow the student to use an abnormal head posture or hold the books/print close to their eyes, to allow them to maximise on their remaining vision; because of this the student will need their own class books.
4. If the student cannot see the writing on the blackboard from the front of the classroom, you can enlarge your writing or allow the student to come up to the board.
5. If possible enlarge the size of the writing on their tests. If the student uses a magnifier, allow them additional time to complete the test: reading with a magnifier is slow as the student can only see a few words at a time. If neither is possible you might consider verbally testing the student by asking them the questions.
6. Try to avoid the student sitting in the sun. If classes or breaks are given outside, try to find a shaded location for the student to sit in.
7. Educate the other students about OCA, by sharing your knowledge of the causes and effects of OCA. This will result in the community having a greater understanding of the condition.

The author declares there are no competing interests.

The author visits the Kwale District Eye Centre in Kenya as a volunteer. The main role is the continual education of the centre's staff, and the reviewing of children with low vision and/or strabismus for an orthoptic assessment. On recent trips the author travelled with the low vision therapist to the schools and observed his assessments and management plans while liaising with the principals, teachers, parents and the extended community involved with the children. The majority of the children seen on these field trips had OCA.

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