

Cancer Association of South Africa (CANSA)



Fact Sheet on Albinism and its Implications

Introduction

Albinism, is a group of inherited disorders (usually an autosomal recessive inherited condition). It results in little or no production of the pigment melanin in the body. Albinism is also known as achromia, achromasia or achromatosis. This hereditary disease can be found in humans (affecting all races), mammals, birds, fish, reptiles and amphibians. Even though it is a hereditary condition, in most cases, there is not necessarily a family history of albinism.



[Picture Credit: Albinism]

Both parents must carry a defective gene to have a child with albinism. When neither parent has albinism but both carry the defective gene, there is a one in four chance that their baby will be born with albinism.

The type and amount of melanin one's body produces determines the colour (or tone) of the skin, hair and eyes. Most people with albinism are sensitive to sun exposure and are at increased risk of developing skin cancer.

Melanin also plays a role in the development of the optical nerves before birth. All forms of albinism cause problems with the development and function of the eyes.

[Picture Credit: Albinism 2]

The photo on the right shows a picture of the world's largest family with albinism – it includes the father, mother, and five children.



Although there is no cure for albinism, people with the disorder can take steps to improve their vision and avoid too much sun exposure. Albinism does not limit intellectual development, although people with albinism often feel socially isolated and may experience discrimination.

According to The National Organization for Albinism and Hypopigmentation, it is estimated that one in every 17 000 people worldwide has some type of albinism. In South Africa the incidence among Black people is about 1 in 3 900 people with oculocutaneous albinism (eye and skin involvement) while in the White population it is less common with an incidence of 1 in 15 000 people. According to available statistics there are about 11 500 affected individuals in South Africa.

(Mayo Clinic; Medical News Today; Albinism Society of South Africa; National Organization for Albinism and Hypopigmentation; Hong, Zeeb & Repacholi).

Prenatal Testing for Albinism

For couples who have not had a child with albinism, there is no straightforward test to determine whether a person carries a defective gene for albinism. Large genetic studies on albinism have been inconclusive, making it look less likely that, at least for the medium-term, effective genetic tests are possible.

In the case of parents who already have a child with albinism. It is possible to test using either amniocentesis (introducing a needle into the uterus to draw off fluid) or chorionic villous sampling (CVS). Cells in the fluid are examined to see if they have an albinism gene from each parent.

(Medical News Today).

Incidence of Albinism in South Africa

Type OCA2 albinism is the most prevalent autosomal recessive disorder among southern African Blacks, affecting 1:3 900 individuals; while albinism type OCA3, although rare, is most prevalent in southern Africa. Another common pigmentation disorder in southern Africa is vitiligo, which affects 1 - 2% of people worldwide. Vitiligo is a complex, acquired disorder in which melanocytes are destroyed due to an autoimmune response.

(Manga, *et al.*).

A study determined the frequency and distribution of albinism among the Vhavenda ethnic group living in the relatively low-income north of South Africa in a clan-oriented society. A retrospective study of birth records from regional hospitals gave an incidence of OCA of 1 in 1970, whereas a survey of mainstream schools gave a frequency of only one pupil with albinism in 13 319 as most affected children attended the regional special school. A community-based field study of 35 rural villages gave a prevalence of 1 in 2239 for OCA. One clan, the Vhatavhatsindi, had a significantly higher frequency of 1 in 832. This epidemiological study provides the necessary data for developing health care and welfare system for families affected by albinism in this region.

(Lund, *et al.*, 2009).

According to Mswela (2016) in South Africa, albinos make up about 1 in every 4 000 people. The genetic condition of albinism in South Africa has a high frequency among the Sotho people of Northern South Africa. One study, carried out in 1982, of the incidence of oculocutaneous albinism amongst the South African black population determined seven diverse ways of establishing who was affected by the disorder. Among the 126 families that had members who were affected by albinism, were males and 93 females. At the time, the Black population of Soweto was more or less 803 511. Based on these statistics, the incidence of albinism was found to be 1 per 3 900. The carrier rate of the albinism gene is around 1 in every 32 persons. The number of persons living with albinism was estimated to be 1 per 2 254 amongst the Southern Sotho, 1 per 4 700 amongst the Xhosa, 1 per 9,700 amongst the Pedi and 1 per 28 614 among the Shangaan inhabitants of South Africa.

Causes of Albinism

The cause of albinism is a mutation in one of several genes. Each of these genes provides the chemically coded instructions for making one of several proteins involved in the production of melanin. Melanin is produced by cells called melanocytes, which are found in the skin and eyes. A mutation may result in no melanin production at all or a significant decline in the amount of melanin.

In most types of albinism, a person must inherit two copies of a mutated gene — one from each parent — in order to have albinism (recessive inheritance). If a person has only one copy, then he or she will not have the disorder.

Different genes are responsible for the different types of albinism.

Oculocutaneous albinism (OCA) is the most common type of albinism. Several different genes have been identified that may cause OCA.

Mutations (changes) in different genes cause different types of OCA. For example:

- OCA1A and OCA1B are caused by mutations in the tyrosinase gene
- OCA2 is caused by mutations in the P gene
- OCA3 is caused by mutations in the TRP-1 gene
- OCA4 is caused by mutations in the SLC45A2 gene

How OCA is inherited - OCA is an autosomal recessive condition. This means one needs to inherit two copies of the faulty gene (one from the mother and one from the father) to have the condition. It is estimated that about one in 70 people carry the gene that causes OCA. Carriers of the gene are not affected by the condition and have a normal amount of melanin. If both parents carry the gene, there is a one in four chance their child will have albinism.

Ocular albinism (OA) - There are two types of ocular albinism. These are caused by different genes and are also inherited in different ways:

- OA1 is caused by a mutation in the GPR143 gene
- autosomal recessive ocular albinism (AROA) is caused by mutations in either the tyrosinase gene or the P gene

(Mayo Clinic; NHS UK).

OCA2 is the most common form of albinism worldwide due to its high prevalence in southern Africa, where it occurs in 1/3 900 Blacks.

(Manga, 2013).

Signs and Symptoms of Albinism

Since birth, people with albinism have little or no pigmentation in their eyes, skin and hair (*oculocutaneous albinism*) or sometimes in the eyes alone (*ocular albinism*). The degree of pigmentation varies. Some people gain a little pigmentation in their hair or eyes with age. Some individuals develop pigmented freckles on their skin. Someone with complete absence of melanin is often referred to as an 'albino'. One with only a small amount of melanin is described as 'albinoid'.

People with albinism are very pale with fair hair and very light eyes. In some people, the eyes appear red or purple, depending on the amount of pigment. This can happen because the iris actually has very little colour. The eyes appear pink or red because the blood vessels inside of the eye show through the iris.

A person with albinism is generally as healthy as the rest of the population. However, problems with vision and skin are particularly common.

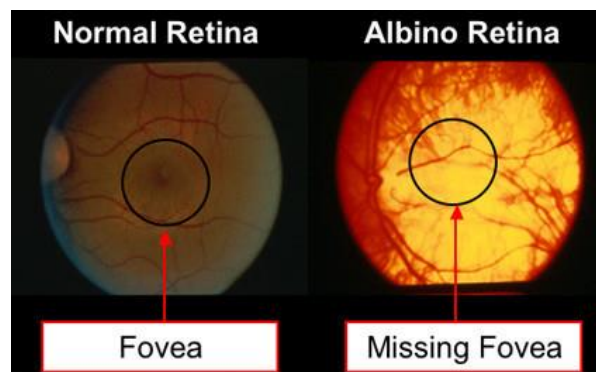
Eye Problems in Individuals with Albinism

Individuals with albinism lack pigmentation in the eye. In a 'normal-sighted' eye, pigment is found in different parts of the eye and performs a function in each part. In addition, albinism alters the structure of the eye and the optic nerve. It is important to note that because the eye develops differently in someone with albinism, conventional treatments, such as surgery or eyeglasses, do not correct the problem. Although people with albinism always have problems with vision, the degree varies greatly among individuals. Some are legally blind, while others have vision that is good enough to drive a car. Most are able to read without using Braille.

[Picture Credit: Albino Retina]

The following parts of the eye are affected in an individual with albinism:

The Retina - The retinal pigment epithelium contains the cells that convert visual input into signals to send to the brain. A lack of pigment in these cells leads to a decreased ability to process visual input. It also leads to a decreased ability to absorb stray light, leading to light sensitivity and the red reflex that is sometimes noticeable as a violet hued iris in people with albinism.



The fovea is the centre of the macula – the area of the retina responsible for one's central, sharpest vision. In a normal eye, the macula is the area of a person's best vision that focuses on an image such as a person's face. The fovea zeros in on detail such as seeing a freckle on someone's face. In people with albinism, the fovea is underdeveloped and lacking in pigment. This is known as *foveal hypoplasia*. Foveal hypoplasia is perhaps the greatest cause of vision problems in an individual with albinism.

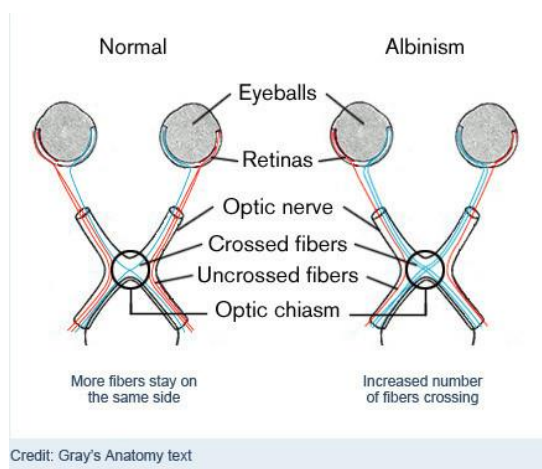
The Optic Nerve - the optic nerve is the connection from the eye to the brain. Between the eye and the brain, there is an area known as the optic chiasm. This area is where the optic nerve splits into separate fibres which are routed to the brain.

In normal-sighted people, most of the fibres stay on the same side of the brain while some cross to the opposite side. This helps with 'binocular vision' (using both eyes) and, in turn, depth perception.

In people with albinism, too many fibres cross to the opposite side, giving the brain an abnormal signal. This leads to the *difficulty with depth perception* that often occurs in albinism.

The Iris - the iris is the coloured part of the eye. It helps control the amount of light entering the eye.

An insufficient amount of pigment in the iris of individuals with albinism results in a decreased ability to filter light entering the eye. The decreased pigment also leads to trans-illumination defects of the iris, which is light reflecting back through the poorly pigmented iris. The detection of trans-illumination defects during an eye examination by an ophthalmologist is an indication of albinism.



People with albinism often are *sensitive to bright lights and glare*, which is usually helped by wearing sunglasses.

The differences in the structure of the eye of individual with albinism all lead to a wide range of visual acuity. One the first things a parent often wonders is 'what will my child see? What will his/her acuity be?' People with albinism are generally considered to have 'low vision' or be 'visually impaired' which means that their vision reduction cannot be corrected with standard glasses or contact lenses. It is not possible to predict what a child's vision will be in infancy because it depends on many factors. However, the most common vision problems associated with albinism are:

- Reduced visual acuity. Primarily due to the differences in the retina described above, individuals with albinism have a reduced visual acuity. Visual acuity refers to the ability to see fine detail. An individual's visual acuity is an indication of the clarity or clearness of what they are seeing. Visual acuity is typically measured by reading letters on an eye chart. The measurement given is based on what a 'normal' human being should be able to see when standing 6 metres away from the eye chart. If an individual has 6/6 vision, it means that when he stands 6 metres away from the chart, he can see what a 'normally-sighted' individual can see at 6 metres away. If an individual has 6/30 vision, it means that when he stands 6 metres away from the chart he can only see what a 'normally-sighted' person can see when standing 30 metres from the chart. In general, an individual with albinism, even after their vision is 'corrected' with glasses or lenses, must be much closer to the chart to see the same lines than a 'normally-sighted' person. Best-corrected visual acuity is the measure of best acuity while wearing corrective lenses like glasses or contact lenses.
- Light Sensitivity. The lack of pigment in the retina and iris generally makes people with albinism sensitive to bright light and glare. One may notice a child squint or cover their eyes in environments with bright light. Tinted glasses and sunglasses outside can help a child most effectively use their vision.

- Nystagmus. This disorder is characterised by an irregular, side-to-side involuntary eye movement that may be side-to-side, up and down or rotary. Although people with nystagmus do not sense that their eyes are moving, this disorder does make it more difficult to focus on visual details. Nystagmus does not go away or disappear but it usually becomes less noticeable with age. Conversely, the eye movements usually become more noticeable if a child is tired, nervous, or stressed. There is a procedure, known as a *tenotomy*, that, in early stages, has shown some promise in improving nystagmus.
- Strabismus is a muscle imbalance of the eye which leads to crossing of the eyes or a 'lazy eye'. In esotropia, the eye turns inward. In exotropia, the eye turns outward. If uncorrected, this can lead to amblyopia, or the loss of some vision function in the weaker eye. Strabismus is correctable with glasses, eye-patches and/or surgery.
- Delayed Visual Maturation. A small percentage of children with albinism show no signs of usable vision for the first few months of their lives. They do not seem to track objects or make eye contact. It may 'seem' like they don't see anything. While this can be very frightening for a parent, *it is temporary and is not thought to indicate less vision overall*. At about six months of age, some parents report that their child acted as though a 'switch' was turned on and it seems that from this point, their vision developed at the same rate as that of other children with albinism.

(The Vision for Tomorrow Foundation; The National Organization for Albinism and Hypopigmentation).

Albinism and Light Sensitivity

Children with albinism can have profound light sensitivity. In a normal eye, the iris helps to shield the retina from bright light. When a child has albinism, their iris is sometimes so light in colour that it cannot properly control the amount of light that hits the retina. Also, because the back of the eye also lacks pigment, light is not absorbed properly and scatters, creating more light sensitivity.

[Picture Credit: Quality Sunglasses]



These children require sun protection, including quality sunglasses or tinted contact lenses.

Some children with albinism may benefit from a permanent tint in their prescription eyeglasses that is light enough to function indoors. Children with albinism may also benefit from photochromic lenses. Photochromic lenses darken to a grey or brown shade when in sunlight and automatically lighten back to clear indoors. Many different types of photochromic lenses are available today and individuals with albinism may benefit from photochromic lenses that turn darker outside but do not necessarily lighten up completely when indoors. They remain slightly tinted indoors (About health).

Albinism and Glare

Glare is light that is reflected off surfaces such as water, waxed floors and white sand. Glare can make even the cloudiest day uncomfortable for children with albinism. Because glare can be debilitating to these children, polarised sunglass lenses are highly recommended. Polarised sunglasses reduce not only the amount of light that enters the eye but they also virtually eliminate associated glare. Polarised lenses can make children with albinism much more comfortable and deliver a much better visual experience for them. Polarised lenses are available in many different colours and are available in both constant tints and photochromic options.

To further enhance comfort for children with albinism, many doctors and opticians recommend adding a mirror coating to their sunglass lenses. A mirror coating further reduces the amount of light that reaches the eyes and deflect the light that bounces up and enters the eye from below (About health).

Albinism and Skin Cancer

Kromberg, *et al*, (1989) investigated the presence of skin cancer in 111 individuals with albinism belonging to the Black population of Johannesburg. The overall rate was 23.4%, the risk increasing with age. Identifiable risk factors included: environmental exposure to ultraviolet radiation; inability to produce ephelides ('freckles'); and possibly ethnicity. The head was the site most commonly affected, and squamous cell carcinoma was far more common than basal cell carcinoma. No melanomas were detected.

Besides giving skin, eyes, and hair their colour, melanin helps protect the skin from the sun. It does this by causing skin to tan instead of burn — which is why people with darker skin (more melanin) are less likely to burn than people with lighter skin. So people with albinism can sunburn very easily.

People with light skin are also particularly at risk for skin cancer. So it is important for people with albinism to use a sunscreen at all times and to wear clothing that offers protection from the sun, such as broad rim hats, dark-coloured clothing, or long pants and long-sleeved shirts.

A review of 775 normally pigmented Africans and 18 African albinos with malignant skin tumours showed that squamous cell carcinoma was the most common tumour type, in contrast to Caucasians, in whom basal cell carcinoma is most frequent. In African albinos squamous cell carcinoma of the head and neck region was most frequent. However, the proportion of basal cell carcinomas was low also among albinos but higher than among normally pigmented patients. In contrast to the normally pigmented patients, there were no squamous cell carcinomas on the limbs in albino patients. We suggest that this difference was due to environmental factors, such as chronic leg ulcers, which might have been less influential in the albinos, who seldom lived more than 30 years. No cases of cutaneous melanoma or Kaposi sarcoma were found in the albino group. (Yakubu & Mabogunje, 2009).

Skin cancers are the most common cancers among albinos in our environment. Skin cancers are a major risk associated with albinism and are thought to be a major cause of death in African albinos. Albinism and exposure to ultraviolet light appears to be the most important risk factor in the development of these cancers. Late presentation and failure to complete treatment due to financial difficulties and lack of radiotherapy services are major

challenges in the care of these patients. Early institution of preventive measures, early presentation and treatment, and follow-up should be encouraged in this population for better outcome.
(BioMed Central Dermatology).

Prejudice and Albinism

People with albinism are just like anyone else, with the same lifespan and ability to reach their goals. But living with any medical condition during the teen years can be difficult, and albinism is no exception.

Because the social scene can be more about fitting in than standing out, teens with albinism may face bullying or prejudice. Voicing any frustration or sadness to a family member or friend who understands can help. So can talking to a counsellor or therapist to get ideas on coping with the challenges that come up during the teen years.
(KidsHealth.Org).

There is growing evidence of social discrimination and stigmatisation directed towards this population. Along with their differences in appearance, a lack of knowledge about albinism in the community leads to such stigma. For example, the aetiological beliefs about albinism continue to be heavily influenced by culture and superstition, rather than genetics.
(Hong, *et al.*).

During September, 2015, the National Prosecuting Authority in South Africa said that Siyabonga Gwala, 18, Lindokuhle Khumalo, 19, Mandla Mabuza, 19, and Bhekukufa Gumede, 65, a traditional healer and pastor had appeared at the Manguzi Magistrate's Court for the alleged murder of Thandazile Mpunza, 20, a young woman with albinism. It is suspected that the murder was linked to witchcraft because of her albinism.

Mpunza was reported missing by her family. Her remains were found in a shallow grave after the suspects confessed and led the police to the shocking discovery.
(Amanda Khoza, News24).

Best Sunscreen for People with Albinism

People with albinism should use sunscreens labelled SPF 20 to 30. Using sunscreens with SPF higher than 30 offers little benefit, and more concentrated chemicals might be more likely to irritate or cause an allergic rash in individuals with albinism. The US Food and Drug Administration (FDA) also proposes limiting the SPF factor to 30. Titanium and zinc oxide screens provide very broad spectrum coverage and are ideal for people with albinism.
(National Organization for Albinism and Hypopigmentation).

South African Support Groups

There is still a certain amount of stigmatisation of people with albinism in the South African community. Both public education about the condition and counselling for affected individuals and their families are required.

A small parent support group in Johannesburg has been functioning under the auspices of The South African Inherited Disorders Association (SAIDA) with more than 30 members all over the country. The objectives of this group are to educate the public about albinism, to provide support for affected families and to support research into the condition. A second large group (with more than 200 members) has also been established in Soweto. (Albinism Society of South Africa).

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Where to Obtain Additional Information and Support



The Albinism Society of South Africa (ASSA)

Address

Physical Address: Lara's Place, 187 Bree Street, Jhb, 2000.

Postal Address:

P O Box 9881 Johannesburg 2000

Contact Person: Nomasonto Mazibuko (Executive Director)

Contact

Tel: 011 838 6529

Non Profit Organisation: 009-389 NPO

Support is also available from The South African Inherited Disorders Association (SAIDA)



SAIDA - *The South African Inherited Disorders Association*

Contact Person:	Helen Malherbe - National Chairperson
Contact Number:	083 399 4353
Email:	info@saida.org.za
Postal Address:	SAIDA c/o Department of Human Genetics P.O Box 1038 Johannesburg 2000

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Albinism

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Albinism 2

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Albinism Society of South Africa

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<http://www.medicalnewstoday.com/articles/245861.php>

National Organization for Albinism and Hypopigmentation

<http://www.albinism.org/publications/ocular.html>

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Quality Sunglasses

http://www.mmf.umn.edu/initiatives/insight/2007/spring/Family_gives_back.cfm

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