

# Cancer Association of South Africa (CANSA)



## Fact Sheet on Albinism and its Implications

### Introduction

Albinism, is a group of inherited disorders. It is 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



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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. (Hong, Zeeb & Repacholi)).

**Alvarez, I., Smittenaar, R., Handley, S.E., Liasis, A., Sereno, M.I., Schwatzkopf, D.S. & Clark, C.A. 2020.**

“Albinism is a congenital disorder where misrouting of the optic nerves at the chiasm gives rise to abnormal visual field representations in occipital cortex. In typical human development, the left occipital cortex receives retinal input predominantly from the right visual field, and vice-versa. In albinism, there is a more complete decussation of optic nerve fibers at the chiasm, resulting in partial representation of the temporal hemiretina (ipsilateral visual field) in the contralateral hemisphere. In this study, we characterize the receptive field properties for these abnormal representations by conducting detailed fMRI population receptive field mapping in a rare subset of participants with albinism and no ocular nystagmus. We find a nasal bias for receptive field positions in the abnormal temporal hemiretina representation. In addition, by modelling responses to bilateral visual field stimulation in the overlap zone, we found evidence in favor of discrete unilateral receptive fields, suggesting a conservative pattern of spatial selectivity in the presence of abnormal retinal input.”

**Nakkazi, E. 2019.**

“People with albinism have been kidnapped and targeted because of myths and superstition around the condition. In some African societies, it is believed that the body parts of people with albinism bring wealth. Although people with albinism are still being killed for their limbs, hair, and nails, civil society organisations say the attacks against people with this condition have substantially decreased in most African countries.”

**Federico, J.R., Krishnamurthy, K. 2019.**

“Albinism, from the Latin *albus*, meaning "white," is a group of heritable conditions associated with decreased or absent melanin in ectoderm-derived tissues (most notably the skin, hair, and eyes), yielding a characteristic pallor. The most commonly thought of presentation is that of oculocutaneous albinism (OCA). OCA is a group of phenotypically similar genetic disorders derived from errors in melanin synthesis. As the name implies, the most dramatic effects are in the eyes and skin. The skin manifestations are more heterogeneous and appear along with a spectrum of severity depending upon the subtype of OCA. The ocular structures rely upon melanin for signaling as they develop, in utero; thus, misrouted optic nerve fibers yield more uniform ocular manifestations of the disorder. To date, seven types of nonsyndromic albinism (OCA1 to OCA7) have been described. These are all due to isolated genetic mutations whose constellation of signs and symptoms do not manifest so broadly that they can be classified as syndromic. A discussion on albinism, however, would be incomplete without the mention of isolated ocular albinism (OA1) and the syndromic albinisms: Hermansky-Pudlak syndrome (HPS) and Chediak-Higashi syndrome (CHS). The syndromic albinisms have the same hallmark lack of dermal and ocular pigment as OCA. They, however, involve genes that encode for proteins that have more extensive applications to cellular function. Loss-of-function mutations in these genes, therefore, yield predictable systemic consequences associated

with the syndromes mentioned. Examples include inactivation of genes involved in lysosomal synthesis (and not simply melanin synthesis) that lead to bleeding diathesis in HPS and propinquity to infection in CHS. Other conditions may present like albinism with congenital nystagmus and/or generalized hypopigmentation. Most of these are included in the Differential Diagnosis section. Of special mention is a pair of syndromes that derive their albino-like features because of deletions in the same genes that are mutated in OCA type 2: Angelman (AS) and Prader-Willi (PWS) syndromes.”

**Franklin, A., Lund. P., Bradbury-Jones, C. & Taylor, J. 2018.**

**BACKGROUND:** Albinism is an inherited condition with a relatively high prevalence in populations throughout sub-Saharan Africa. People with oculocutaneous albinism have little or no pigment in their hair, skin and eyes; thus they are visually impaired and extremely sensitive to the damaging effect of the sun on their skin. Aside from the health implications of oculocutaneous albinism, there are also significant sociocultural risks. The impacts of albinism are particularly serious in areas that associate albinism with legend and folklore, leading to stigmatisation and discrimination. In regions of Africa those with albinism may be assaulted and sometimes killed for their body parts for use in witchcraft-related rites or to make 'lucky' charms. There is a dearth of research on the psychosocial aspects of albinism and particularly on how albinism impacts on the everyday lives of people with albinism.

**DISCUSSION:** There is a growing recognition and acceptance in Africa that people with albinism should be considered disabled. Thomas's social-relational model of disability proposes it is essential to understand both the socio-structural barriers and restrictions that exclude disabled people (barriers to doing); and the social processes and practices which can negatively affect their psycho-emotional wellbeing (barriers to being). In this article, we combine a social model of disability with discussion on human rights to address the lacuna surrounding the psychosocial and daily experiences of people with albinism.

**CONCLUSION:** Through using this combined framework we conclude that the rights of people with albinism in some regions of Africa are not being enacted. Our debate highlights the need to develop a holistic concept of rights for children and young people with albinism which sees human rights as indivisible. We illuminate some of the specific ways in which the lives of children with albinism could be improved by addressing 'barriers to being' and 'barriers to doing', at the heart of which requires a shift in attitude and action to address discrimination.

### **Prenatal Testing for 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.

### **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

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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.

### **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.

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.

The most common vision problems associated with albinism are:

- Reduced visual acuity. Visual acuity refers to the ability to see fine detail.
- Light Sensitivity. The lack of pigment in the retina and iris generally makes people with albinism sensitive to bright light and glare.
- 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.
- Strabismus is a muscle imbalance of the eye which leads to crossing of the eyes or a 'lazy eye'.
- 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.

**Karlén, E., Milestad, L. & Pansell, T. 2019.**

**PURPOSE:** Albinism degrades visual function due to developmental disorders of the eye and visual pathways, larger refractive errors, absent binocularity and poor fixation control. Reading spectacles is commonly prescribed in our clinic and well tolerated. The purpose was to evaluate whether the accommodative response is typical or affected in comparison to a reference group.

**METHODS:** Twenty-two children with albinism (median: 13.5 years) and 12 controls (median: 13 years) underwent a full optometric examination and an objective accommodation measurement (WAM-5500 @ 6 Hz; Grand Seiko) in response to minus-lens-blur (-1, -2 and -3 D) and to a prolonged near viewing task (20 cm) for 5 min.

**RESULTS:** Children with albinism displayed less accommodation to minus lens-blur and during sustained near viewing ( $p < 0.001$ ) compared to the reference group. Higher visual acuity correlates

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with a better accommodative response ( $r \geq 0.5$ ;  $p \leq 0.04$ ). The subjective and objective measures of accommodation did not correlate. The habitual reading distance was always closer than the point towards which the subjects with albinism seemed to accommodate according to the measurements at 20 cm.

**CONCLUSION:** Children with albinism benefits from reading spectacles due to a combination of close habitual reading distance and a poor accommodation. Objective recording of accommodation is not critical for a correct judgement of near visual function. Children already wearing reading spectacles were those with least accommodative response.

### **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.

**Ramos, A.N., Fraga-Braghiroli, N., Ramos, J.G.R., Scope, A. & Fernandes, J.D. 2019.**

“Oculocutaneous albinism (OCA) increases predisposition to skin malignancies. Nevertheless, the differential diagnosis between melanoma and naevi in patients with OCA is still challenging, because pigmentary lesions have rarely been described in this population. We aimed to describe the

dermoscopic patterns of naevi in patients with OCA. We prospectively evaluated 83 naevi from 37 patients with OCA in a single centre in Brazil. Lesions were analysed by eye and by dermoscopy and were grouped by dermoscopic pattern. Eight main patterns were identified: homogeneous structureless pattern (n = 28; 33.7%), globular pattern (n = 27; 32.5%), reticular pattern (n = 8; 9.6%), peripheral reticular pattern with central hypopigmentation (n = 8; 9.6%), peripheral globules (n = 8; 9.6%), irregular brown globules with pink background (n = 2; 2.4%), reticular globular disorganized pattern (n = 1; 1.2%) and peripheral reticular globular with central hypopigmentation (n = 1; 1.2%). We found previously undescribed dermoscopic patterns in patients with OCA, in addition to confirming previously described patterns. These descriptions may help the understanding of pigmented naevi in patients with OCA.”

### **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.

### **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).

### **Medical Disclaimer**

This Fact Sheet is intended to provide general information only and, as such, should not be considered as a substitute for advice, medically or otherwise, covering any specific situation. Users should seek appropriate advice before taking or refraining from taking any action in reliance on any information contained in this Fact Sheet. So far as permissible by law, the Cancer Association of South Africa (CANSA) does not accept any liability to any person (or his/her dependants/estate/heirs) relating to the use of any information contained in this Fact Sheet.

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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

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### Albinism 2

<http://twentytwoords.com/worlds-largest-albino-family/>

### Albinism Society of South Africa

<http://doctors-hospitals-medical-cape-town-south-africa.blaauwberg.net/details.php?id=580>

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**National Organization for Albinism and Hypopigmentation**

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**NHS UK**

<http://www.nhs.uk/Conditions/albinism/Pages/causes.aspx>

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