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Living with albinism

Dr Shari Parker talks about the challenges she faces every day.

IF you have seen *The Da Vinci Code*, *Matrix Reloaded*, *Lethal Weapon* or *The Princess Bride*, you could be forgiven for thinking that people with albinism are evil and villainous with violent tendencies or supernatural powers.

Nothing could be further from the truth. I know this from a personal perspective. I am a doctor and a person with albinism. I am not evil or villainous. I do not have violent tendencies or, sadly, any supernatural powers. I just have white hair, very fair skin and visual impairment.

What is albinism?

Put simply, albinism is an inherited condition where the body produces less melanin than normal. 'Albus' comes from Latin and means white.

Albinism is broadly classified as either ocular albinism (OA), which affects the eyes, or oculocutaneous albinism (OCA), which affects the eyes, skin and hair. Within this framework, there are several subtypes of albinism, depending on the genes affected. There are some rare forms of albinism that are associated with other systemic manifestations.

The inheritance of most common forms of albinism is autosomal recessive. It affects about one in 17,000 Australians. This means that there are about 1000 Australians living with albinism and about one in 75 people are carriers.

Albinism is diagnosed clinically. Genetic testing is possible, and albinism can be classified as tyrosinase positive or negative based on the analysis of hair root bulbs. The purpose of these tests is largely academic. There is no cure for albinism.

Skin and hair features

The most visually startling and obvious feature of albinism is pale or white hair and skin.

This means that people with albinism do not tan, sunburn extremely easily and are more susceptible to solar damage. Regular surveillance for solar damage is essential. Actinic keratoses and squamous and basal cell carcinomas are common. Amelanotic melanomas can also occur, a fact to which I can personally attest.

The cutaneous aspects of albinism are managed by sun avoidance, frequent liberal application of broad-spectrum 30+ sunscreen, and by wearing broad-rimmed hats and long-sleeved clothing. Essentially the Cancer Council's 'Slip Slop Slap' message taken to the extreme.

Ocular features

Due to the lack of melanin, the retina, particularly the fovea, fails to develop properly. The optic nerve fibres cross abnormally in the optic chiasm and there is transillumination of the iris (ie, the iris does not filter light out).

Visual impairments include pendular nystagmus, monocular vision with poor depth perception and photophobia. Many people with albinism also have strabismus and refractive errors. People with OA manifest the eye features.

People often think of red eyes when they think of albinism. Strangers have said to me, "Oh, you don't have albinism, you don't have red eyes". Most people with albinism have pale-blue or, occasionally, violet-coloured eyes. The red eye misconception is largely due to flash photography. The flash travels unfiltered through the iris and pupil, is bounced back off a non-pigmented retina with unobscured blood vessels and appears red on the photo. Thanks to digital photography, it is much rarer now for people with albinism to appear as aliens with glowing red eyes in family or group photos.

The average corrected vision of people with albinism is about 6/36 to 6/60. Functional vision, however, is generally much better than this.

As the visual impairment with albinism is congenital, most people with albinism have learned compensatory tricks, using experience and intelligence to interpret what can be seen. Vision is often better in practice than in theory. This is a situation where impairment does not necessarily equal disability or limit activity.

Having said this, in situations involving bright light or glare, such as at the beach or when sitting facing a window, functional vision can be much

poorer and at times people with albinism can literally be 'blinded by the light'.

I am often asked what I can see. Looking through the "wrong" end of a pair of binoculars is a fair approximation. I have a normal visual field and my vision is not blurred. Things just appear less detailed. The situation is, of course, much worse in an environment where glare is a factor.

Visual impairments in albinism are managed with visual rehabilitation measures. These include monocular telescopes (affectionately known as a 'spyglass'), magnifying glasses, large print text, closed circuit television, sunglasses and corrective eyewear.

Surgery to correct strabismus is possible, as is surgery to dampen nystagmus. This can improve acuity to a degree.

Other common effects

Most people with albinism are unable to drive. Functional mobility training to facilitate the use of public transport and to help people with albinism get around independently are useful. In the US, some states allow the use of bioptic lenses for driving. Bioptic lenses have a small telescope mounted on a pair of glasses. This is sadly currently not allowed in Australia.

Anecdotally, people with albinism often report musculoskeletal complaints of the neck and back. With the visual impairment, comes the need to bring items close to the eyes to see them in any detail. This can lead to poor posture, muscular tightness and headaches.

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Impaired depth perception can predispose people with albinism to poor balance and falls, especially with the accumulation of other co-morbidities that can occur with ageing.

Psychosocial aspects

The psychosocial aspects of albinism are significant and often more troublesome for people with albinism than the physical effects. Movies, myth and the stigmatisation of anyone appearing different are largely to blame.

Name-calling, teasing and bullying in childhood are often reported by people with albinism. Kids can be mean and so often pick on the odd one out. This can lead to social isolation, poor development of the sense of self and depression.

When the child with albinism starts school, teachers need to receive education about the condition. In my experience, it is crucial that the child with albinism is made to feel as normal as possible. Children with albinism already look different and any additional treatment that further separates them from their peers should be avoided.

Later in life, more subtle discrimination, particularly in the workforce by employers and co-workers, can occur. This is largely due to ignorance about albinism and its effects.

Vision Australia is an invaluable resource for people with albinism and parents of children with albinism. The Albinism Fellowship of Australia is a useful support group (www.albinismaustralia.org). This group has been set up to provide support, education and fellowship to those with albinism, parents of children with albinism, their friends and families, and relevant professionals. I am on the committee of this organisation and am their medical liaison officer.

Despite the potential negative psychosocial implications of albinism, most people with albinism I have encountered are confident, strong-willed, determined and successful people. Many people with albinism are parents and work in a diversity of roles ranging from actors and accountants to lawyers, waitresses and aerospace engineers. Some are in health professions including nursing, massage therapy, speech pathology, occupational therapy, social work, chiropractics and medicine.

Personal Insights into albinism

So how does all this relate to me as a person with albinism and as a medical practitioner?

Corrected, my vision is 6/60 in one eye and 6/36 in the other. You are probably asking yourself, "How on earth did she become a doctor with such poor vision?"

I have an older sister with albinism. At birth, my parents were told to "put her in the sun and she'll brown up". When the diagnosis of albinism was finally made at three months, my parents were instructed to bring her back at age five so she could be placed in a blind school. They did not follow this advice and we were educated in mainstream schools.

I received my fair share of name-calling during my childhood. These included being called "Casper", "Snow white", "Shari the short-sighted" and regularly being told that I was "as blind as a bat". Thanks to a fairly tenacious personality and a group of loyal friends, this schoolyard banter did not have any long-lasting effects.

I naturally gravitated away from activities in the glaring sun. I tended to enjoy indoor activities such as music, drama, reading and studying.

This was fortuitous as I decided to study medicine in my early teens. I had a fascination with all things medical, perhaps borne out of frequent childhood medical interactions. I studied hard and obtained entry into the medicine course at the University of NSW in Sydney. Thankfully, I always received support and encouragement from my teachers and family.

Like most medical students, I found identifying structures on cadavers, histology and histopathology quite difficult. Though unlike most, this was due largely to the fact that I could barely see the fine structures. The only way I scraped through these subjects was with my theoretical knowledge of what 'should' be there and on my pattern recognition skills.

My nystagmus proved to be a hindrance in clinical examinations, because the condition is much more severe with stress and anxiety. My nystagmus, and therefore my vision, in these situations is poorer. The nystagmus may even have been a barometer for the examiner for when I did not know the answers.

My nystagmus is also distracting for people in a psychotic state, as I learned when trying to interview people during psychiatry terms. They

would fixate on my 'dancing' eyes and any further interviewing would be nigh impossible.

I got through medicine, did an honours year for the bachelor of medical science on disability in the elderly and eventually graduated from medicine with honours in 1999.

I then set about obtaining registration with the NSW Medical Board. Having declared my albinism, I needed certification from an ophthalmologist that I was fit to be registered. The first one I saw told me quite blatantly that she would not sign such a form because I clearly could not be a competent medical practitioner with such poor vision.

Thankfully, I was referred to another ophthalmologist who understood that the measured visual acuity does not necessarily equate with disability. The form was signed and I received conditional registration.

The conditions on my registration are that I undertake procedures within my visual ability and that I inform my employer. This works well in practice and my vision is rarely an issue. I certainly never held aspirations to become a surgeon.

I chose to specialise in rehabilitation medicine. I am now in my final year of advanced training.

My choice of specialty undoubtedly was influenced by my albinism. Perhaps I have a greater empathy for the challenges my patients are facing due to the obstacles that I have had to overcome throughout my own life.

I can honestly say that my albinism has never been raised as an issue when caring for patients in my seven years since graduation. Perhaps the diminution of one sense has led to the heightening of the other senses, especially that 'sixth' sense.

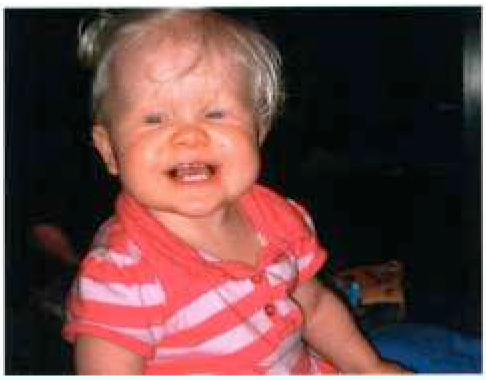
The main challenge that my albinism poses for me in my work life is not being able to drive. While I am very adept at using public transport and taxis, these routes of transport can be extremely frustrating when work demands travel between hospitals and home visits.

I have found that as I have grown older, I am much more comfortable with my albinism. In the past, I certainly never advertised my albinism and tended not to talk openly about it, unless it came up. I have perhaps mellowed with age and am now keen to inform others of this often misunderstood condition.

Now I find myself seven years post-graduation, a competent doctor in my final year of specialist training. I am married and a mother of two toddlers. Life is rich and the albinism is but one component of my personal makeup. ●

Dr Parker is an advanced trainee in rehabilitation medicine at Prince of Wales Hospital and Eastern Suburbs Private Hospital in Sydney. She is also on the committee of the Albinism Fellowship of Australia and is their medical liaison officer.

Attendants of the inaugural conference of the Albinism Fellowship of Australia in Sydney



Teachers need to receive education about albinism before a child with the condition starts school.

Five things I love about albinism

1. The lack of one special sense often leads to a heightened perception in the other senses. There are countless famous people with albinism who are musicians including Winston Foster ('King Yellowman' — a Jamaican Dancehall musician) and Brother Ali.
2. Not being able to drive means that people with albinism are kinder to the environment, we often walk more and thus have a higher level of fitness.
3. We tend to have younger-looking skin because we are usually so meticulous about sun protection.
4. We are always easy to spot in a crowd.
5. We generally have resilient personalities and are very determined individuals.

Five things I hate about albinism

1. Being called an "albino". We are people first and the albinism comes far down the list. Please call us people with albinism.
2. The widespread negative portrayal of people with albinism in movies and stories as being evil, villainous or having mystical powers.
3. Not being able to drive.
4. The inhumane prejudice people with albinism experience around the world. In Zimbabwe, there is a belief that having sex with a person with albinism will cure a man of AIDS. This has resulted in the widespread rape and infection with HIV of women with albinism.
5. Societal prejudice based on skin colour in general.

Five things you can do to help your patients with albinism

1. Be aware of and sympathetic to their significant visual impairment.
2. People with albinism may be eligible to apply for Centrelink benefits such as the Disability Support Pension (Blind) and Mobility Allowance, the Taxi Subsidy Scheme and travel passes for the visually impaired.
3. Be alert to the possibility of poor self-esteem, depression and social isolation that can affect people with albinism of all ages as a consequence of discrimination and bullying.
4. Perform regular skin checks for the presence of skin cancers, especially SCCs and BCCs. Look for amelanotic melanomas.
5. Refer them to the Albinism Fellowship of Australia. (www.albinismaustralia.org).