

Albinism

Albinism is a non-contagious, genetically inherited condition that affects people worldwide regardless of race or ethnicity. It results in a lack of pigmentation (melanin) in any or all of the hair, skin and eyes, causing vulnerability to the sun and bright light. As a result, most persons with albinism have a visual impairment of varying degrees and are prone to developing skin cancer. In almost all types of albinism, **both parents** must carry the gene for it to be passed on, even if they do not have albinism themselves.

When both parents carry the gene, then there is a 25 percent chance at each pregnancy that the child will have albinism. When both parents carry the gene, other children without albinism have a 50% chance of carrying the gene. Frequency of albinism in Africa is 1 in 5,000 to 1 in 15,000 with certain tribes exhibiting higher rates of 1 in 1,000. In Europe and in North America, frequency is 1 in 17,000 to 1 in 20,000. Among indigenous groups in Fiji and Panama, rates can be as high as 1 in 700 and 1 in 125 respectively. Higher frequencies can sometimes be explained by consanguineous relationships e.g. intermarriage among cousins such that the gene pool becomes tighter. "Person with albinism" is now preferred to "albino" because the former puts the person ahead of his condition while the latter equates him to it. This is the global trend as we see in the common phrases: persons with disabilities and persons with leprosy for example

- **Types of Albinism**

- **Albinism only affecting the eyes.** This is called Ocular albinism (OA), and is much less common. Most cases are recorded in males. Children with OA may have skin and hair colour that are normal but slightly lighter than those of other family members.
- **Albinism affecting the skin, hair and eyes.** This is known as OCA or Oculocutaneous (pronounced ock-you-low-kew-TAIN-ee-us) albinism. This is the most visible form of albinism in Tanzania.
- Using DNA research, 4 forms of OCA are now recognized – OCA1, OCA2, OCA3 and OCA4; some of these are further divided into subtypes. In each category and sub-category of OCA, the degree of colour/melanin apparent in the individual varies.
- **Albinism plus:** Researchers have also identified a type of albinism that comes with additional characteristics. One of these is known as Hermansky-Pudlak Syndrome (HPS). In addition to albinism, HPS is associated with bleeding problems and bruising. Some forms are also associated with lung and bowel disease. HPS is a less common form of albinism but should be suspected if a person with albinism shows unusual bruising or bleeding.

Visual problems associated with Albinism

Development of the optical system is highly dependent on the presence of melanin, and the reduction or absence of this pigment in sufferers of albinism may lead to:

- Misrouting of the retinogeniculate projections, resulting in abnormal decussation (crossing) of optic nerve fibres.
- Photophobia and decreased visual acuity due to light scattering within the eye (ocular straylight). Photophobia is specifically when light enters the eye, unrestricted - with full force. It is painful and causes extreme sensitivity to light.
- Reduced visual acuity due to foveal hypoplasia and possibly light-induced retinal damage.

Eye conditions common in albinism include:

- Nystagmus, irregular rapid movement of the eyes back and forth, or in circular motion.
- Amblyopia, decrease in acuity of one or both eyes due to poor transmission to the brain, often due to other conditions such as strabismus.
- Optic nerve hypoplasia, underdevelopment of the optic nerve.

The improper development of the retinal pigment epithelium (RPE), which in normal eyes absorbs most of the reflected sunlight, further increases glare due to light scattering within the eye. The resulting sensitivity (photophobia) generally leads to discomfort in bright light, but this can be reduced by the use of sunglasses and/or brimmed hats.

Genetics

Oculocutaneous albinism is generally the result of the biological inheritance of genetically recessive alleles (genes) passed from both parents of an individual for example OCA1 and OCA2. A mutation in the human TRP-1 gene may result in the deregulation of melanocyte tyrosinase enzymes, a change that is hypothesized to promote brown versus black melanin synthesis, resulting in a third oculocutaneous albinism (OCA) genotype, "OCA3". Some rare forms are inherited from only one parent. There are other genetic mutations which are proven to be associated with albinism. All alterations, however, lead to changes in melanin production in the body. Some of these are associated with increased risk of skin cancer

The chance of offspring with albinism resulting from the pairing of an organism with albinism and one without albinism is low. However, because organisms (including humans) can be carriers of genes for albinism without exhibiting any traits, albinistic offspring can be produced by two non-albinistic parents. Albinism usually occurs with equal frequency in both sexes. An exception to this is ocular albinism, which it is passed on to offspring through X-linked inheritance. Thus, ocular albinism occurs more frequently in males as they have a single X and Y chromosome, unlike females, whose genetics are characterized by two X chromosomes.

There are two different forms of albinism: a partial lack of the melanin is known as hypomelanism, or hypomelanosis, and the total absence of melanin is known as amelanism or amelanosis.

Diagnosis

Genetic testing can confirm albinism and what variety it is, but offers no medical benefits except in the cases of non-OCA disorders that cause albinism along with other medical problems which may be treatable. There is no 'cure' for Albinism. The symptoms of albinism can be assisted by various methods.

Treatment

- PWAs often show a combination of any of the following:

NAME OF CONDITION	CONDITION	TREATMENT or REHABILITATION (where available)
Nystagmus	regular movement of the eyes, side to side or in circular motion	Surgical intervention today minimizes but cannot completely cure nystagmus
Strabismus includes “crossed eyes” (esotropia), “lazy eye” or an eye that deviates out (exotropia)	Muscle imbalance in eyes	For strabismus, surgery may improve the appearance of the eyes In the case of esotropia or “crossed eyes,” surgery may help vision by expanding the visual field (the area that the eyes can see while looking at one point).
Photophobia	Sensitivity to bright light and glare	PWAs need light to see just like anyone else. Sunglasses or tinted contact lenses help outdoors. Indoors, it is important to place lights for reading over a shoulder rather than in front.
Near or far sightedness Usually with astigmatism or some blurriness	far-sightedness or nearsightedness	Various optical aids are helpful to people with albinism including bifocals, strong prescription reading glasses and bioptics* (glasses which have small telescopes mounted on)
Foveal hypoplasia	No normal development of the inside surface of the eye that receives light- known as the retina	See photophobia
Optic nerve misrouting	nerve signals from the retina to the brain do not follow the usual nerve routes	

*Some countries permit driving with biotic lenses for some PWAs

2. Skin Problems and how to prevent them

- People with many types of albinism need to take precautions to avoid damage to their skin caused by the sun.

- Damage to the skin can be avoided by regularly wearing sunscreen lotions, hats and sun-protective clothing which is clothing that covers skin from exposure to the sun.

3. Medical Challenges & how to avoid them

- In many countries, most people with albinism live normal life spans and have the same types of general medical problems as the rest of the population.
- Those with additional conditions such as Hermansky-Pudlak Syndrome may have a shortened life span by lung disease or other specific challenges.
- In countries with sunny & hot or tropical climates like Ghana, persons with albinism who do not use skin protection may develop life-threatening skin cancers.
- Persons with albinism in such countries must use appropriate skin protection, such as sunscreen lotions rated 20 SPF or higher and proper clothing to enjoy outdoor activities and a normal life span.

4. Social Challenges & how to remove them

- People with albinism are at risk of isolation because the condition is often misunderstood.
- Social stigmatization can occur, especially within communities where other family members have dark skin and appearance.
- Families and schools must make an effort to include children with albinism in group activities.
- Contact with other persons with albinism, or others who have persons with albinism in their families can be most helpful.

5. Educational Challenges & how to overcome them

- Due to significant vision impairment most persons with albinism cannot read from the blackboard in a normal classroom set-up.
- Teachers and educators must make extra effort to re-organize the classroom so that persons with albinism are positioned in front of the class and moved closer to the blackboard as needed.
- This re-positioning must be done so that the person with albinism is **not** staring into sunlight to see the blackboard.
- To enhance re-positioning, the blackboard must be kept “black” at all times so that the contrast of the white chalk may enhance read-ability.
- If re-positioning fails, teachers and educators must be prepared to photocopy their notes and give copies to each person with albinism in their class.
- Exams, teachers’ notes and other printed/photo-copied hand-outs must be clear and with large enough print.

- If needed, extra time must be granted for exam-writing for the persons with albinism. (Actual time plus half of actual time is suggested). This is especially important if the exam questions are provided in small or regular print, and if the exams are re-used/faded copies.
- For day-to-day support, teachers and educators may find it useful to assign a capable student to each person with albinism for peer support in the learning process.

Epidemiology

Albinism affects people of all ethnic backgrounds; its frequency worldwide is estimated to be approximately one in 17,000. Prevalence of the different forms of albinism varies considerably by population, and is highest overall in people of sub-Saharan African descent.

International Albinism Awareness Day

International Albinism Awareness Day was established after a motion was accepted on 18 December 2014 by the United Nations General Assembly, proclaiming that as of 2015 the 13th of June would be known as International Albinism Awareness Day. This was followed by a mandate created by the United Nations Human Rights Council that appointed Ms. Ikponwosa Ero, who is from Nigeria, as the very first Independent Expert on the enjoyment of human rights by persons with albinism.

RESOURCES AND EXTERNAL LINK

1. albinism@ohchr.org
2. International Albinism Awareness Day
3. Under The Same Sun: www.underthesamesun.com sun
4. **NOAH**
The National Organization for Albinism and Hypopigmentation
<http://www.albinism.org>
5. www.positiveexposure.org
6. **The Vision for Tomorrow Foundation**
www.visionfortomorrow.org
7. **HEALTH AND SKIN CARE FOR PEOPLE WITH ALBINISM / SEASIDESKINCARE.COM**
8. <http://seasideskincare.com/health-and-skin-care-for-people-withalbinism>.
9. **EXPERT OPINION Dr. Murray Brilliant, Geneticist.**
<https://ictr.wisc.edu/Brilliant>