

Albinism in life

By Allen Little for Albinism Trust © 2015

The symptoms of albinism depend on the specific type of albinism and are mainly related to the eyes, skin and hair. Generally people tend to want an understanding about what those of us with Sight Impairments can and can't see. They ask questions based on their own capabilities to see things. It is important to understand 6/6 does not translate into perfect vision and does not indicate other important aspects of sight such as peripheral vision, the ability to identify colours or depth perception. Having 6/12 vision means you can see at six metres what a person with normal vision can see at 12 metres away. A visual acuity of 6/24 satisfies requirement's for **Registration with the Blind Foundation.**



Ophthalmoscope use

The ophthalmoscope is a tool used in medicine to examine the interior of the eye. Observing the inner parts of the eye, such as the optic disk, retinal blood vessels, retina, choroid and macula, can help diagnose diseases of the eye. An Ophthalmoscope works by redirecting bright light through a mirror into the eye. That light reflects off the retina and returns to the instrument in the form of a magnified image assisting the clinicians examination. An issue for people with Albinism is the experience of having ones eyes examined by Ophthalmoscopy. This examination is important for the Clinician who is seeking to see a good view of the ocular pathology. Most eye care professionals are aware of the need to explain and prepare patients with Albinism about the brightness of the light emitted from this instrument. If the bright light is intolerable and uncomfortable it's important to mention this fact to the examining clinician at the time. It should be noted that ophthalmoscopy often necessitates dilation of the pupils; this can last for several hours and exacerbates the photophobia both during the investigation and for some time afterwards.

Symptoms of The Eye -

With Albinism the colour of the iris is usually blue/grey or light brown with some people due to lack of pigment in the iris, having a reddish or violet hue reflected through the iris. Vision problems in albinism result from abnormal development of the retina, lack of pigment in the retina and abnormal patterns of nerve connections between the eye and the brain. People with albinism can have unusual pathways for sending nerve signals from the eye to the brain.

One major abnormality of the eye in albinism involves lack of development of the fovea. The fovea is a small but most important area of the retina inside the eye. The fovea is the central area of the retina that enables sharp vision, such as for reading and recognising faces; this area of the retina does not develop in albinism. This results in lack of sharp vision and blurring of objects.

Other symptoms of the eyes include -

- Decreased sharpness in vision (visual acuity)
- Functional blindness / Low Vision
- Involuntary, irregular and rapid movement of the eye (Nystagmus)
- Lazy eye (Amblyopia)
- Sensitivity to bright lights or glare (Photophobia)
- Deviation of the eyes (Strabismus)
- Distortion of a viewed image (Astigmatism)

Symptoms Affecting The Skin -

- Patchy or total absence of the melanin pigment leaving the areas pale
- Susceptibility to rapid sun burning

Symptoms of The Hair -

- Forelock (portion of the hair near the forehead) turns white (seen in Waardenberg syndrome)
- Complete absence of pigment in the hair resulting in white hair

Other Symptoms include -

- Bleeding tendency (seen is Hermansky-Pudlak Syndrome)
- Problem with bowels (seen is Hermansky-Pudlak Syndrome)
- Breathing problems due to lung fibrosis (seen is Hermansky-Pudlak Syndrome)
- Deafness
- Increased susceptibility to infections nervous system disorders

Albinism is -

- **a congenital disorder** characterized by the complete or partial absence of pigment called melanin in the skin, hair and eyes. It is also known as hypopigmentation, oculocutaneous albinism or ocular albinism.
- **an autosomal recessive type** of inherited disorder, which means that the chance of the disorder being passed from one generation to the next is low. Statistics show that about 1 in 17,000 people suffer from some form of albinism.
- **an inherited defect** affecting melanin production and metabolism. Melanin is a naturally occurring pigment, which is responsible for the colour of our skin, hair and eyes. It protects the skin from the harmful effects of ultraviolet light.
- People with Albinism in the tropics are darker as their sun exposure is more, in comparison to people who live in temperate climates. If the gene encrypting this pigment is defective, the body becomes pale and white.

People who are affected by albinism are called albinos. They have absence of colour in the hair, skin, or iris of the eye; or lighter than normal skin and hair. Lack of skin pigmentation makes albinos more susceptible to sunburn and skin cancers. Vision problems like extreme far-sightedness or near-sightedness, nystagmus, strabismus, photophobia and astigmatism are common in people with albinism.

Albinism can be diagnosed merely by observation of major or total absence of pigmentation of the skin, hair and eyes. Genetic testing, electroretinogram, chemical testing of hair and blood tests aid in diagnosing albinism.

There is no cure for albinism. Treatment is aimed to ease the symptoms. Treatment of the eye conditions consists of visual rehabilitation, or the prescription of tinted lenses. It is important that albinos use sunscreen before sun exposure to prevent premature skin aging or skin cancer.

Albinos can live a normal life span, however, some forms of albinism can be life threatening. The lives of people with Hermansky-Pudlak syndrome can be shortened by lung disease. People in tropical countries who do not use skin protection may develop life-threatening skin cancers.

Albinism may cause social problems, because people with albinism look different from their families, peers, and other members of their ethnic group. There is no known way to prevent albinism. Genetic counselling should be considered for individuals with a family history of albinism or hypopigmentation.

What are the Types of Albinism?

Oculocutaneous Albinism (OCA) - It is the most common and severe form of albinism. It affects the eyes, skin and hair. People with OCA also suffer from vision defects, usually myopia or short sightedness and require glasses.

OCA can be categorized into different types based on the specific gene involved.

- In Type 1 albinism there is a defect in the metabolism of an amino acid called tyrosine that leads to failure in converting tyrosine to melanin. The conversion normally happens due to an enzyme called tyrosinase, however, if there is a defect in the enzyme it fails to happen.
- In Type 2 albinism there is a defect in the 'P' gene. In this type there is only minor defect in the pigmentation at birth.
- In Type 3 albinism there is a genetic defect in TYRP1, a protein related to tyrosinase.
- In Type 4 albinism there is a genetic defect in the SLC45A2 protein that helps the tyrosinase enzyme to function.

Ocular Albinism (OA) - In this condition the skin colour is usually normal and eye colour may be in the normal range. But close examination of the eye reveals that there is little or no pigment in the retina. It accounts for about 10 to 15% of the albinism cases. It can be inherited either by an X-linked or an autosomal recessive process.

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Since OA is not easily recognised or diagnosed, it is important that children who may be exhibiting visual difficulties say at school, are examined as soon as possible. The visual effects of OA can be profound and are often mistaken for learning difficulties.

Although the visual defects of albinism can not be treated, some symptoms may be ameliorated with appropriately tinted glasses. There are now tints available that have been developed specifically for albinism which can be incorporated into prescription glasses or some contact lenses.

There are some conditions in which there is only a specific area of the body that is affected by the condition. These include-

Waardenberg Syndrome - In this type of syndrome there is only a small lock of hair on the forehead that is white and not pigmented or there maybe just one or both eyes (the iris) that has absence of pigment.

Chediak-Higashi Syndrome - Lack of melanin pigment all over the body with few areas being spared.

Tuberous sclerosis (white leaf macule) - Tuberous sclerosis are small localized areas of de-pigmentation.

Hermansky-Pudlak Syndrome (HPS) - This condition is associated with a great range in degrees of pigmentation, from no pigmentation to almost normal colouring.

It is a single-gene disorder inherited in an autosomal recessive manner. It is a form of albinism associated with a bleeding tendency (bleeding disorder) along with problems of lungs (lung fibrosis) and bowel diseases.

Diagnosis of albinism will require an examination of the eyes which involves a comparison family pigmentation.

A medical doctor (ophthalmologist) or optometrist should conduct your child's eye examination. The exam will include an assessment of potential nystagmus, strabismus and photophobia. The doctor will use a device to visually inspect the retina and determine if there are signs of abnormal development. A simple test can measure the brain waves produced when light or a reversing pattern is flashed into each eye. This can indicate the presence of misrouted optical nerves. Disorders other than albinism can affect skin pigmentation, but these don't cause all of the visual problems associated with albinism.

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Other tests to diagnose albinism include -

- ✓ **Genetic Testing** - Genetic testing is the most accurate way to diagnose albinism and its specific type. This is helpful in families with albinism and is useful for specific, isolated populations who carry the trait in them.

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- ✓ **Eye Examination** - An ophthalmologist or optometrist should perform a complete examination of the eye of an affected individual. An electroretinogram test may be done to determine vision problems in albinism.
- ✓ **Chemical Testing of Hair** - Chemical testing of hair also provides an easy confirmation of the diagnosis of albinism.
- ✓ **Hairbulb pigmentation test** - It can be used to identify carriers. It is done by incubating a piece of the person's hair in a solution of tyrosine, an amino acid the body uses to make melanin. If the hair turns dark, it means the hair is making melanin. Light hair means there is no melanin synthesis.
- ✓ **Tyrosinase test** - It is more precise than the hairbulb pigmentation test. It measures the rate at which hair converts tyrosine into another chemical (DOPA), which is then made into pigment.
- ✓ **Blood Test** - Recently, a blood test has been developed that can identify carriers of the gene for some types of albinism.
- ✓ **Prenatal Diagnosis Of Albinism** - Amniocentesis and chorionic villus sampling can also diagnose some types of albinism in pregnancy.

Several types of hypopigmentation in humans are called albinism. The phenotype for different types of albinism varies according to the amount of pigment in the hair, skin and iris, the reduction in visual acuity and the degree of nystagmus and strabismus.

Cutaneous and ocular melanin pigment can range from complete absence throughout the lifetime of the individual to the development of nearly normal levels, including the ability to tan. Visual acuity ranges from 20/40 to 20/400, and visual development in an affected infant is slower than normal. Foveal hypoplasia and altered routing of the optic nerves are found in all types of albinism and are the most constant feature of this condition.

The demonstration of optic track misrouting by visual evoked potential studies provides the critical diagnostic procedure for questionable cases of albinism, and this is the single definitive diagnostic test to confirm a diagnosis of albinism.

Visual perception is the ability to interpret the surrounding environment by processing information that is contained in visible light. The resulting perception is also known as eyesight, sight, or vision (adjectival form: visual, optical, or ocular). The various physiological components involved in vision are referred to collectively as the visual system, and are the focus of much research in psychology, cognitive science, neuroscience, and molecular biology, collectively referred to as vision science.

<p>These notes are published by Albinism Trust as a Public Education Resource and have been checked by Mr Gordon Sanderson, Associate Professor, Ophthalmology Section. Otago Medical School, Dunedin.</p>
