

## Albinism (Achromasia) — Definition and Genetics

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**Albinism is a hereditary condition characterized by a low level or complete absence of melanin production, which can be confined to the eyes or widespread in the body. People with albinism present with white skin, white or yellow hair, and blue eyes. While the condition is rarely associated with any significant mortality, people with albinism tend to suffer from bleeding disorders, photosensitivity, and impaired vision. Treatment of visual impairment with low-vision aids and early-age eye patching might be needed.**



### Definition of Albinism

Albinism refers to a group of **melanin synthesis disorders** that are **inherited**. Patients develop a **reduction or a complete absence of melanin pigment**. The most common **mutation** associated with albinism involves the **metabolic pathway** that is responsible for the production of melanin from **tyrosine**. Albinism occurs due to genetic defects and can be passed on from one generation to another; this condition makes the body unable to produce the pigment melanin.

**The most severe form of albinism is called oculocutaneous albinism.**

**Oculocutaneous albinism** is defined as the reduction or absence of melanin in the skin, the hair, and the optic system. **Ocular albinism** is confined to the optic system without

cutaneous involvement.



Image: "Fundus picture of a patient with albinism (a) and fundus picture of a normal eye (b)." by Grønskov K, Ek J, Brøndum-Nielsen K . License: [CC BY 2.0](https://creativecommons.org/licenses/by/2.0/)

## Epidemiology of Albinism

The prevalence of albinism is approximately **1 in 17,000 people** in the United States. There are an estimated 18,000 people with albinism in the United States.

**Oculocutaneous albinism (OCA) types 1 and 2** are responsible for most cases of the condition. OCA1 has a prevalence of 1 in 40,000 people, while OCA2 has a prevalence of 1 per 15,000. OCA2 is more common in African Americans than in whites.

A special type of albinism known as **the Hermansky-Pudlak syndrome** is the most common cause of albinism in Puerto Rico with a prevalence of 1 in 2,700. This form of albinism is very rare in other populations.

Albinism is not associated with any significant mortality, and most affected individuals are of normal intellectual capacity. OCA and ocular albinism are associated with an **increased risk of visual impairment** in addition to **skin photosensitivity**. Individuals with albinism are also known to suffer from **social anxiety** due to their different appearance as compared to their peers.

OCA is common in both sexes, but ocular albinism is an X-linked recessive form of the disease that is only reported in males.

## Pathophysiology of Albinism

OCA types 1 and 2 are related to **mutations in genes responsible for tyrosinase and P protein**, respectively. These mutations are not X-linked. On the other hand, patients with ocular albinism are likely to have an X-linked form of the disease that involves the protein named **OA 1**.

**Melanin**, which is a pigment that is **photosensitive, protects the skin** by absorbing **ultraviolet light** from the sun. The absence of this photoprotective pigment is associated with a **higher risk of skin damage by sun exposure**. Individuals with albinism are therefore **at risk of developing sunburns**.

Melanin is also found in the **eyes**, especially in the **neuroectoderm**, which is responsible for the development of the **outer pigmented ciliary epithelium**. When melanin is absent, the **fovea** fails to develop normally. Additionally, the absence of melanin in the eyes was found to be associated with **abnormal connections between the retina and the brain**, hence **abnormal visual processing** can happen in those with albinism.

Individuals with albinism are thought to develop **decreased visual acuity** for several

reasons. Firstly, increased access of light to the retina might actually result in **retinal damage**. Additionally, as discussed above, foveal development is impaired during early life because of the absence of melanin.

## Clinical Presentation of Albinism

Individuals with albinism, in addition to the characteristic loss of melanin from the skin, also report **ocular symptoms**. **Photophobia** and **decreased central vision** due to **hypoplasia of the fovea** are common findings. Additionally, **easy bruising, frequent nosebleeds, and prolonged bleeding after surgery or dental work** are common in people with albinism. Individuals with **Hermansky-Pudlak syndrome** might have a **family history** of albinism.

Individuals with **OCA type 1** have **skin hypopigmentation at birth**, white hair, and blue irides. Individuals with **OCA type 2** have **normal skin pigmentation or absent skin pigmentation**. Some might have a complete absence of melanin at birth making the condition difficult to distinguish from OCA type 1.

The main difference between OCA type 2 and OCA type 1 is that the former is associated with **increased pigmentation as the child ages**. In fact, this increased pigmentation is very significant clinically because **visual performance is reported to improve** in those with OCA type 2.

African Americans affected by OCA type 2 usually have **yellow hair** at birth, which remains yellow throughout life. They also have **white skin that does not tan**.

Ocular albinism type 1 is an X-linked trait that presents with **eye involvement** without any significant skin changes. These individuals can develop **decreased visual acuity, fundus hypopigmentation, foveal hypoplasia, and nystagmus**. X-linked ocular albinism almost always affects males; females typically are carriers.

### Testing for albinism and risks involved in genetic testing

Testing for albinism is conducted on the patient's blood sample. There are some risks involved in blood testing, such as light-headedness, bleeding, and bruising.

## Diagnostic Workup of Albinism

Individuals with a complete **absence of melanin from their skin, hair, and eyes** are easily diagnosed clinically with albinism. Confirmation of the diagnosis requires specific and sophisticated **laboratory tests** to be performed.

The determination of **tyrosinase enzyme activity** from the hair bulb is useful. This test can provide quantifiable information by using radiolabeled tyrosine to measure the amount of melanin produced.

Individuals who have reduced enzyme activity for tyrosinase will show an inconclusive test result, and the diagnosis cannot be ruled out. Thus, **genetic testing** is the only diagnostic tool that can definitively confirm the diagnosis of albinism.

Genetic testing for albinism is usually provided for individuals with **familial forms** of albinism and not for those with sporadic cases of albinism. Individuals with albinism are at risk of **bleeding complications from surgeries**. **Bleeding time and platelet aggregation tests** are advisable for any person with albinism who is going to undergo major surgery.

People with albinism are at risk of developing **foveal hypoplasia**, among other foveal abnormalities. **Optical coherence tomography** is useful in the evaluation and assessment of the fovea and other parts of the retina. People with albinism are also at risk of developing **diminished visual acuity; visual evoked potentials** have been found to correlate with visual acuity in individuals with albinism.

Finally, individuals with albinism can have a **skin biopsy** performed **for research purposes**. Histological examination of a skin biopsy reveals the presence of **macromelanosomes**.

## Treatment of Albinism

Individuals with albinism might need **low-vision aids** and **tinted glasses** to correct for refractive errors and photophobia, respectively. Due to refractive errors, people with albinism might develop **strabismus**. **Eye-patching** should be used as early as possible, ideally by the age of 6 months, for the optimal correction of strabismus.

Individuals with albinism who wish to alter their hair and eye color might benefit from **nitisinone**, which increases plasma tyrosine levels and helps increase melanin levels in the skin, eyes, and hair of people with **hereditary tyrosinemia type 1**.

### Preventive measures for albino patients:

Patients with albinism should always take certain precautions. Shielding the eyes and skin from the sun is recommended. Patients should use a sunscreen with high sun protection factor when going out in the sun and cover themselves up with proper clothing when going out in daylight. To protect their eyes, they should wear sunglasses designed for UV protection.

**Retinal detachment** is a common complication in people with albinism. **Retinal detachment repair surgery** might be needed; unfortunately, the prognosis for restoration of normal vision is usually poor. It is postulated that vision cannot be improved with surgery in people with albinism for two main reasons: the **absence of retinal pigmented epithelium** and the **abnormal retina-brain neural connections and cortical processing of visual stimuli**.

## References

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