CONDITION

# Oculocutaneous albinism

Oculocutaneous albinism is an inherited condition that reduces pigment in the eyes, skin, and hair.

**WHAT IT IS**
Inherited reduction of pigment in eyes, skin, and hair

**BODY SYSTEM**
Eyes, skin, and hair

**MAIN SYMPTOMS**
Reduced vision, light sensitivity, involuntary eye movements, pale colouring

**WHAT CAUSES IT**
Inherited gene changes affecting pigment production

**HOW IT IS DIAGNOSED**
Eye and skin examination, confirmed by genetic testing

**MAIN TREATMENT**
Vision support, sun protection, and skin monitoring

**CAN IT BE CURED?**
No, inherited pigment changes cannot be reversed

**IS IT AN EMERGENCY?**
No, but sudden vision loss needs emergency care

**LONG-TERM OUTLOOK**
Lifelong; health outlook usually good with protective care

## Overview

Oculocutaneous albinism, often shortened to OCA, is a group of inherited conditions that affect the production of melanin. Melanin is the pigment that gives colour to your skin, hair, and eyes. It also helps protect the skin from ultraviolet, or UV, radiation and has an important role in eye development before birth.

People with OCA make little melanin or make a form that does not work as expected. The amount of pigment differs between genetic types and between individuals. OCA does not always mean completely white hair or very pale skin.

The condition is present from birth. It affects both the eyes and the skin, which is why it is called "oculocutaneous": "oculo" refers to the eyes, while "cutaneous" refers to the skin.

Low pigment can make the eyes sensitive to light and affect how the retina and visual pathways develop. The retina is the light-sensitive layer at the back of the eye. These changes often cause reduced vision that glasses cannot fully correct.

Skin with less melanin burns more easily and has less natural protection from long-term UV damage. Sun protection and skin monitoring are therefore an important part of lifelong care.

OCA does not usually affect intelligence. Most people can study, work, have relationships, and take part in everyday activities, although vision support and practical adjustments may be needed.

## Symptoms

Features are usually noticed in infancy or early childhood. Their severity can differ even among relatives with the same genetic type.

Common eye and vision features include:

Nystagmus often begins during the first months of life. Some people turn or tilt their head because a certain position reduces the eye movement and helps them see more clearly.

Eye colour may be blue, grey, hazel, or brown. The coloured part of the eye, called the iris, may let more light through than usual. Eyes do not generally appear red, although certain lighting or photographs can make reflected blood vessels visible.

Skin and hair may be lighter than those of other family members. Some people have very pale skin and white or light-blond hair. Others have more colour, and their hair or skin may darken with age. Freckles and moles can also develop.

The visual effects are usually present in both eyes. Vision may range from a mild reduction to severe visual impairment.

- Reduced sharpness of vision
- Sensitivity or discomfort in bright light, called photophobia
- Rapid, involuntary eye movements, called nystagmus
- Eyes that do not point in the same direction, called strabismus
- Short-sightedness, long-sightedness, or astigmatism
- Reduced depth perception

## Causes and risk factors

OCA is caused by changes in genes involved in making or handling melanin. Several genetic types are known. The type can influence how much pigment develops, but it does not predict every person's experience.

Most forms follow an autosomal recessive inheritance pattern. This means a person usually inherits one altered copy of the same gene from each parent. The parents are often healthy carriers and may not know they carry the change.

When both parents carry a change in the same OCA-related gene, each pregnancy has its own chance of resulting in a child with OCA. A genetic counsellor can explain the chances for a particular family after reviewing test results and family history.

OCA can occur in any ethnic group. Having a relative with OCA or having parents who carry changes in the same gene increases the chance of inheriting it.

Rarely, albinism is part of a broader genetic syndrome. For example, Hermansky-Pudlak syndrome can involve unusual bleeding and, in some types, lung or bowel problems. These associated conditions are not features of most nonsyndromic OCA.

## Diagnosis

OCA may be suspected when a baby or child has reduced pigmentation together with nystagmus or other vision differences. Diagnosis involves looking at the eyes, skin, hair, medical history, and family history.

An eye specialist can check visual development, eye alignment, focusing problems, and the structures inside the eye. Testing may show underdevelopment of the central retina, known as foveal hypoplasia. Optical coherence tomography, or OCT, can create detailed images of the retina.

A test called a visual evoked potential may be used in some cases. It measures how signals travel from the eyes to the brain. People with albinism often have a characteristic difference in how these nerve pathways are arranged.

Genetic testing can confirm a genetic cause and may identify the OCA type. It can also help distinguish nonsyndromic OCA from forms linked with other health problems. However, testing does not identify the cause in every person.

Conditions such as isolated nystagmus, other inherited retinal conditions, and pigment changes from different causes may need to be considered. A skin biopsy is not usually required.

## Treatment and management

There is no treatment that restores typical melanin production or reverses the way the eyes developed before birth. Management focuses on making the best use of vision, protecting the skin, and identifying complications early.

Glasses or contact lenses can correct short-sightedness, long-sightedness, and astigmatism. They may improve vision but do not correct all visual effects of OCA. Low-vision services can suggest magnifiers, telescopic devices, enlarged text, screen readers, and other tools.

Tinted lenses, sunglasses, hats, and changes to indoor lighting may reduce glare. Sunglasses should block UV radiation, but lenses do not need to be very dark to provide UV protection.

Children may benefit from school adjustments, such as:

Strabismus surgery may improve eye alignment. Surgery can also help some people who use a marked head position because of nystagmus. It does not usually remove nystagmus or restore typical vision.

Skin protection includes using broad-spectrum sunscreen, wearing protective clothing and a brimmed hat, seeking shade, and avoiding tanning beds. Sunscreen should be reapplied according to its label, especially after swimming or sweating.

Regular skin reviews can help find sun damage or skin cancer early. The timing of reviews should reflect your skin findings, UV exposure, and local specialist advice.

If testing suggests a syndromic form of albinism, management may also include checks for bleeding, lung, immune, bowel, or neurological problems.

- Sitting where glare is limited and teaching materials are visible
- Receiving large-print or digital materials
- Using magnification or text-to-speech technology
- Having extra time for tasks that rely heavily on vision

## Complications

Reduced vision can affect reading, recognising faces at a distance, driving, sport, and travel in unfamiliar places. Glare may make outdoor vision harder. Falls or injuries are possible when depth perception is limited, although practical adaptations can reduce these risks.

Repeated UV exposure can cause sunburn, early skin ageing, rough precancerous patches called actinic keratoses, and skin cancer. A skin cancer in someone with OCA may contain little

pigment, so it may look pink, red, or skin-coloured rather than dark.

Social misunderstanding, unwanted attention, bullying, and discrimination can affect emotional wellbeing. Support from family, schools, low-vision services, counselling, or albinism organisations can be helpful.

Complications involving bleeding, the immune system, lungs, or bowel suggest a syndromic form rather than uncomplicated OCA and need separate assessment.

## Prevention

OCA cannot be prevented through diet, skincare, or choices made during pregnancy. It is inherited, and nothing a parent did caused the genetic change.

Genetic counselling can help people understand inheritance, testing options, and reproductive choices. This provides information rather than preventing the condition.

Some complications can be reduced. Consistent UV protection lowers the risk of sunburn and long-term skin damage. Eye care, suitable glasses, and low-vision support can improve function and help children develop skills confidently.

## Living with it and what to expect

OCA is lifelong. The eye development changes do not go away, and vision does not usually become typical. In nonsyndromic OCA, vision is often broadly stable after childhood. Any new or rapid loss of vision should not be assumed to be part of albinism.

Nystagmus may become less noticeable over time, although it often remains present. Hair and skin pigmentation may increase with age in some genetic types. Increased colour does not necessarily mean that the eyes or vision have changed.

Children may need updated support as schoolwork becomes more visually demanding. Adults may use different aids for work, transport, parenting, or independent living. Occupational therapy, orientation and mobility training, and assistive technology can help with specific tasks.

Whether you can drive is based on your measured vision and local licensing rules, not the diagnosis alone. Some people meet the required standard, while others use public transport or other travel options.

Nonsyndromic OCA does not usually shorten life. Long-term health is strongly influenced by UV protection, access to eye and skin care, and whether a broader genetic syndrome is present.

## When to get medical help

Arrange a routine appointment if a baby develops persistent involuntary eye movements, does not appear to track objects, or has unusually strong light sensitivity. Early eye assessment can identify focusing problems and support visual development.

Seek a prompt, non-emergency review for:

Get urgent eye care for sudden flashes, many new floaters, or a shadow or curtain across your vision. These can be signs of a retinal problem and are not expected features of OCA.

Seek emergency care for sudden loss of vision, severe eye pain, a serious eye injury, uncontrolled bleeding, or major difficulty breathing. These symptoms need immediate assessment whether or not you have OCA.

- A new or changing skin growth, especially one that bleeds or does not heal
- Increasing difficulty with vision, glare, reading, or daily activities
- New easy bruising, frequent nosebleeds, or unusually heavy bleeding
- New breathing or persistent bowel symptoms in someone with possible syndromic albinism

## Common questions

Is oculocutaneous albinism contagious?

No. OCA is inherited through genes. It cannot spread through touch, shared objects, blood, food, sex, or close contact.

Is OCA the same as ocular albinism?

No. OCA affects pigmentation in the eyes, skin, and hair. Ocular albinism mainly affects the eyes, with skin and hair colour often close to that of other family members. The inheritance patterns can also differ.

Will my vision get worse over time?

The vision changes caused directly by nonsyndromic OCA are usually stable after visual development is complete. Normal age-related eye conditions can still occur. Sudden or continuing deterioration needs assessment because it may have another cause.

Can someone with OCA drive?

Some people can, while others do not meet the legal vision standard. This is based on visual sharpness, field of vision, and local licensing rules. An eye or low-vision assessment can

measure these abilities.

Can two parents without albinism have a child with OCA?

Yes. Most forms are recessive, so two unaffected parents can each carry one altered gene copy. If both carry changes in the same OCA-related gene, they can have a child who inherits both copies.

Can skin or hair become darker with age?

Yes, in some genetic types. Pigment may gradually increase during childhood or adulthood. Even if the skin darkens or tans slightly, it may still have less natural UV protection and require consistent sun protection.

## Sources

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