Oculocutaneous Albinism

Albinism is an inherited condition that occurs at birth and is essentially characterized by an absence or decrease in the pigment that gives color to the skin, hair, and eyes. There are different types of albinism, including oculocutaneous albinism (OCA). Oculocutaneous albinism is the most common and causes decreased pigment in the eyes, hair, and skin. It also causes vision problems such as blurred vision, rapid eye movements, strabismus (squinting) and increased sensitivity to light.

Oculocutaneous Albinism (OCA) Variants

OCA 1	It comes from a genetic defect in an enzyme called tyrosinase. This enzyme helps the body change the amino acid tyrosine into pigment.
OCA 2	People with OCA2 produce a minimal amount of melanin pigment and may have a hair color ranging from very light blonde to brown.
OCA 3	It results from a genetic defect in the TYRP1 protein, a tyrosinase-related protein, and can have a substantial amount of pigment.
OCA 4	It is caused by a defect in the SLC45A2 protein that helps the enzyme tyrosinase to function.
OCA 5, 6 y 7	These variants are rare and have reported mutations in three additional genes. To identify these individuals, more genetic tests need to become available.

Not all people with oculocutaneous albinism have the same characteristics.

Risk Factors

Albinism is an inherited genetic disorder. There is a high chance that if both parents carry the oculocutaneous albinism gene, the child will acquire it. However, if the child receives only one oculocutaneous albinism gene from the parent, the child will not show symptoms.

Signs and Symptoms

Skin: color may be white to brown.
Hair: color can range from white to brown; eyebrows and eyelashes are characteristically lighter.
Eye color: eyes can be very light blue to brown and may change with age.
Vision: a common sign of oculocutaneous albinism is impaired vision.

Some of these are:

- **Nystagmus:** rapid, involuntary lateral movement of the eyes.
- Head movement, to reduce involuntary eye movements.
- **Strabismus:** inability of both eyes to focus on the same point or move at the same time.
- **Myopia:** blurred vision of distant objects.
- **Photophobia**: sensitivity to light.
- **Astigmatism:** atypical curvature of the cornea causing blurred vision.
- Atypical development of the retina causes reduced vision.
- **Abnormal optic nerve decussation:** nerve signals from the retina to the brain that do not follow normal nerve pathways.
- **Legal blindness**: vision less than 20/200, or total vision.

Diagnosis

The physician should perform the following tests: physical examination, specifically skin and hair pigmentation; retinal examination; and genetic testing to confirm and determine the type of variant.

Treatment

There is no cure for oculocutaneous albinism. There are methods for eye and skin care.

These include:

- **Eye Care**: Visiting an ophthalmologist regularly, wearing glasses or contact lenses that are prescribed for sun protection, and being treated for nystagmus or other vision problems.
- **Skin care:** Apply broad-spectrum sunscreen, stay in shaded areas, wear a hat or cap, visit a dermatologist every 6 to 12 months, and avoid using medications that make you more sensitive to the sun.

Social and Emotional Aspects

For recommendations, tools, and resources on how you and your family can address the emotional and physical concerns that arise during your treatment, visit your primary care physician or contact:

Medical Advice Line
1-844-347-7801
TTY/TDD 1-844-347-7804
APS Health
787-641-9133

References:

https://www.mayoclinic.org/es/diseases-conditions/albinism/symptoms-causes/syc-20369184 2023
https://www.mayoclinic.org/es/diseases-conditions/albinism/diagnosis-treatment/drc-20369189#:~:text=El%20diagn%C3%B3stico%20del%20albinismo%20se,con%20la%20de%20otros%20familiares. 2023
https://albinism.org/boletin-informativo-que-es-el-albinismo/ 2021
https://www.aao.org/salud-ocular/enfermedades/albinismo 2022

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