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Review Article on Albinism

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The term albinism typically refers to oculocutaneous (ok-u-low-ku-TAY-nee-us) albinism (OCA) — a group of inherited disorders where there is little or no production of the pigment melanin. The type and amount of melanin your body produces determines the color of your skin, hair and eyes. Albinism is associated with a number of vision defects, such as photophobia, nystagmus, and amblyopia. Lack of skin pigmentation makes for more susceptibility to sunburn and skin cancers. In rare cases such as Chédiak–Higashi syndrome, albinism may be associated with deficiencies in the transportation of melanin granules. This also affects essential granules present in immune cells leading to increased susceptibility to infection [1].

Different types of albinism can have different patterns of inheritance, depending on the genetic cause of the condition. Oculocutaneous albinism (OCA) involves the eyes, hair and skin. Ocular albinism (OA), which is much less common, involves primarily the eyes, while skin and hair may appear similar or slightly lighter than that of other family members. Mutations in several different genes, on different chromosomes, can cause different types of albinism. According to the National Organization for Albinism and Hypopigmentation, "With ocular albinism, the color of the iris of the eye may vary from blue to green or even brown, and sometimes darkens with age. However when an optometrist or ophthalmologist examines the eye by shining a light from the side of the eye, the light shines back through the iris since very little pigment is present. Researchers have also identified several other genes in which mutations can result in albinism with other features [2].

One group of these includes at least nine genes (on different chromosomes) leading to 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. Like OCA, HPS is inherited in an autosomal recessive manner. Because individuals with albinism have skin that entirely lacks the dark pigment melanin, which helps protect the skin from the sun's ultraviolet radiation, their skin can burn more easily from overexposure. Individuals with vision problems may need corrective lenses. They should also have regular follow-up exams with an ophthalmologist. In rare cases, surgery may be needed. Individuals with albinism should also have regular skin assessments to screen for skin cancer or lesions that can lead to cancer. Research helps us better understand diseases and can lead to advances in diagnosis and treatment.

This section provides resources to help you learn about medical research and ways to get involved. The human eye normally produces enough pigment to color the iris blue, green or brown and lend opacity to the eye. In photographs, those with albinism are more likely to demonstrate "red eye", due to the red of the retina being visible through the iris. Lack of pigment in the eyes also results in problems with vision, both related and unrelated to photosensitivity. People with albinism are at risk of isolation because the condition is often misunderstood. Social stigmatization can occur, especially within communities of color, where the race or paternity of a person with albinism may be questioned. Families and schools must make an effort to include children with albinism in group activities. Contact with others with albinism or who have albinism in their families or communities is most helpful. NOAH can provide the names of contacts in many regions [3].

References

1. Manga, Prashiela, Robyn Kerr, Michèle Ramsay, and Jennifer GR Kromberg. “Biology and genetics of oculocutaneous albinism and vitiligo–common pigmentation disorders in southern Africa.” South African medical journal 103, no. 12 (2013): 984-988.
2. King, Richard A., C. G. Summers, J. W. Haefemeyer, and B. Leroy. “Facts about albinism.” International Albinism Center, University of (2001).
3. Kruijt, Bastiaan, Luuk Franssen, Liesbeth JJM Prick, Johannes MJ van Vliet, and Thomas JTP van den Berg. “Ocular straylight in albinism.” Optometry and vision science 88, no. 5 (2011): E585-E592.

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