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Albinism

Complete or partial lack of pigment
From Mary Kugler, R.N., former About.com Guide
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Congenital hypopigmentary disorders, known as albinism, result from a defect in the production of pigment (melanin) in the skin, eyes, and hair. This is due to the dysfunction of the melanin-producing cells (melanocytes).

This article looks at the types of albinism which occur most often These are oculocutaneous albinism (OCA) types 1, 2, and 3, and ocular albinism. Other disorders which have albinism as a symptom are Chediak- Higashi syndrome, Hermansky-Pudlak syndrome, and Waardenburg syndrome.

Albinism affects both males and females, and is apparent from birth. Research indicates that OCA type 1 occurs in 1 individual per 40,000 population, OCA type 2 occurs in 1 individual per 15,000, and ocular albinism in 1 individual per 50,000 population. Research has not yet found how often OCA type 3 occurs, although it has only been genetically confirmed in African and African American individuals.

Inheritance

OCA type 1 is caused by a gene on chromosome 11 and is inherited as an autosomal recessive trait, meaning the an individual must inherit two defective genes for the disorder to occur. OCA type 2 is caused by a gene on chromosome 15 and is also inherited as an autosomal recessive trait. OCA type 3 is caused by a gene on chromosome 9 and is also inherited in an autosomal recessive manner. Ocular albinism is caused by a gene on the X (female) chromosome. It is also a recessive disorder, meaning that a male inheriting one defective X chromosome will have ocular albinism, but a female would have to inherit two defective X chromosomes to have the disorder.

Symptoms

All types of albinism have some lack of pigment, but the amount varies from types to type.

- OCA type 1 usually causes complete absence of pigment in the skin, hair, and eyes, but some individuals may have some degree of pigmentation. OCA type 1 also causes sensitivity to light (photophobia), reduced visual acuity, and involuntary eye twitching (nystagmus).
- OCA type 2 causes a minimal to moderate degree of pigmentation in the skin, hair, and eyes. It also causes eye problems similar to OCA type 1.
- OCA type 3 has been difficult to identify based on appearance alone. It has been most noticeable when a very light-skinned child is born to dark-skinned parents. OCA type 3 also causes eye problems, but not as severe as those of OCA type 1.

- Ocular albinism affects only the eyes, causing minimal pigmentation in them. The iris may appear translucent. Reduced visual acuity, nystagmus, and difficulty controlling eye movements may occur.

Diagnosis

Albinism is present at birth, and it is usually diagnosed based on the infant's appearance. If necessary, genetic testing can be done to confirm a diagnosis, but this is not routinely done.

Treatment

There is no treatment or cure for albinism. Since individuals with albinism have little or no melanin in their skin, they need to use a broad-spectrum sunscreen and wear adequate clothing when outside to prevent ultraviolet-induced damage to the skin. The use of sunglasses will reduce the symptoms of light sensitivity as well as protecting the eyes. An ophthalmologist can treat other eye or vision symptoms. Individuals with albinism should see a dermatologist regularly to be screened for skin cancer. Albinism does not alter life expectancy or have other serious health effects.

Information for this article was taken from:

Boissy, R. E. (2003). Albinism. eMedicine, accessed at http://www. emedicine.com/derm/topic12.htm