Optic Albinism

Albinism is a condition in which there is a deficiency of melanin (produces pigment) in the body. The lack of melanin in the body causes certain parts of the eye to form abnormally. Typically in the development of the iris, melanin is used to block the light, and in the retina, it is used to absorb the light. Since the light is not blocked or absorbed in albinism, a condition known as photophobia occurs. Photophobia is when light scatters in the eye causing bright light to be painful and uncomfortable.

Figure 1. Fundus picture of a patient with albinism (a) and fundus picture of a normal eye (b).

http://www.ojrd.com/content/2/1/43

Another area of the eye affected in development is the fovea. The fovea depends on melanin to develop properly. Due to the lack of melanin in the development of the fovea (located in the center of the retina, it is a pit filled with millions of cones used to provide color sensitivity), pit may not form, and there will be a decreased number of cones present.

Our optic nerves are a series of nerves that travel from the eyes to the brain. Half of the optic nerves for the right eye connect to the right side of the brain, while the other half connect to the left side of the brain, and the same occurs in the left eye. The images that are projected into the two sides of the brain are then combined by the brain to form a single imagine. When there is a lack of melanin such as in Albinism, the optic nerves cross, and the imagines are never combined. People with Albinism will see a single image, with a lack of depth. The misaligned optic nerves can lead to strabismus, which is where one eye moves independently of the other.


Individuals will also experience involuntary movement of the eyes, most commonly from side to side but may occur up and down as well. This involuntary movement or Nystagmus will be compensated by the brain over time, and the image produced in the brain will not move.

Another portion of the eye which is effected by the lack of Melanin in development is the cornea. The cornea may be curved in an irregular shape leading to the following: nearsightedness, farsightedness or distorted images.
Forms of Optic Albinism

Oculocutaneous Albinism involves eyes, hair and skin and is inherited in an autosomal recessive pattern (one mutation from each parent).

Ocular Albinism primarily involves the eyes, while skin and hair may appear lighter than that of other family members. Ocular Albinism is inherited on the X chromosome, due to this, more men are affected than women.

Detection of Albinism

Babies can be diagnosed with Albinism prior to birth with DNA testing. Methods commonly used are chorionic villi sampling and amniocentesis. These tests are primarily conducted if the unborn child has siblings with Albinism or other family members.

Most people are diagnosed with Albinism when born though. If the child is born with hair and skin that is determined to be lighter than other family members, doctors will perform eye exams looking specifically at the retina, and the iris for the transparency discussed earlier.

Treatments for Albinism

Albinism itself is not life threatening, and while there are no ways stimulate the production of melanin in the body, there are many treatments available. Sunglasses can help to comfort the light sensitivity experienced due to photophobia. Glasses or contact lenses can be used to assist in correcting the nearsightedness, farsightedness and astigmatism. Surgery to the eye-muscles may be an option for correcting twitches in the eye as well.

There are many easy ways to assist with vision such as large-print books, computer displays, video magnifiers, telescopic lenses, and speech synthesizers.

Great Websites for support and or fellowship:

http://www.albinism.org
The National Organization for Albinism and Hypopigmentation (NOAH) is a great resource for parents find support and ideas on raising children with Albinism.

http://www.albinism.org.uk/
Albinism Fellowship in the United Kingdom and Ireland.

http://www.positiveexposure.org/
An arts organization which uses visual arts to impact genetics, mental health and human rights.

References:


