

Albinism Terminology

Oculocutaneous Albinism (OCA): Oculocutaneous (pronounced ock-you-low-kew-TAIN-ee-us) Albinism is an inherited genetic condition characterized by the lack of or diminished pigment in the hair, skin, and eyes. Implications of this condition include eye and skin sensitivities to light and visual impairment.

Ocular Albinism (OA): Ocular Albinism is an inherited genetic condition, diagnosed predominantly in males, characterized by the lack of pigment in the eyes. Implications of this condition include eye sensitivities to light and visual impairment.

Hermansky Pudlak Syndrome (HPS): Hermansky-Pudlak Syndrome is a type of albinism which includes a bleeding tendency and lung disease. HPS may also include inflammatory bowel disease or kidney disease. The severity of these problems varies much from person to person, and the condition can be difficult to diagnose with traditional blood tests

Chediak Higashi Syndrome: Chediak Higashi Syndrome is a type of albinism in which the immune system is affected. Illnesses and infections are common from infancy and can be severe. Issues also arise with blood clotting and severe bleeding.

Melanin: Melanin is pigment found in a group of cells called melanocytes in most organisms. In albinism, the production of melanin is impaired or completely lacking.

Nystagmus: Nystagmus is an involuntary movement of the eyes in either a vertical, horizontal, pendular, or circular pattern caused by a problem with the visual pathway from the eye to the brain. As a result, both eyes are unable to hold steady on objects being viewed. Nystagmus may be accompanied by unusual head positions and head nodding in an attempt to compensate for the condition. Nystagmus appears more prominent as students focus visually on a target.

Null Point: A null point refers to the point in which a student has positioned his/her head to greatly reduce or eliminate nystagmus.

Strabismus: Strabismus is a misalignment of the eyes caused by a muscle imbalance. The eyes deviate upward, downward, inward, or outward simultaneously or independently of each other.

Photophobia: Photophobia is an abnormal discomfort or sensitivity to light and glare. Individuals with albinism experience photophobia because the lack of pigment in the iris does not allow the eyes to filter out light.

Refractive Error: Refractive errors are vision problems that happen when the shape of the eye keeps you from focusing well. The cause could be the length of the eyeball (longer or shorter), changes in the shape of the cornea, or aging of the lens. This could

manifest in a student being nearsighted or farsighted. Students with albinism sometimes experience refractive errors in addition to the impaired vision associated with albinism, but this is not always the case, thus, not all students with albinism will be prescribed or benefit from glasses.

Foveal Hypoplasia: The retina, the surface inside the eye that receives light, does not develop normally before birth and in infancy.

Optic Nerve Misrouting: The nerve signals from the retina to the brain do not follow the usual nerve routes.

Optical Devices: An optical device is a tool used by a student with low vision to improve access to a given near or distance tasks. These devices are typically prescribed by a Low Vision Specialist, and some examples include hand-held magnifiers, monoculars (telescopes), and/or video magnifiers.

Scotoma: A scotoma refers to a partial loss of vision in an area of the eye where otherwise normal vision exists. It is also known as a blindspot.