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Fact Sheet

Coloboma of the Eye

By Alycia Nurton

What is a coloboma of the eye? Basically, a coloboma is used to describe a missing piece, or tear in any part of the eye. More precisely, a coloboma is a gap or defect in part of the structure of the eye. This gap can occur in a range of areas and be of varying sizes. The most common for of gap is caused by an imperfect closure of a cleft, present in the fetus, but usually closed by birth date. This gap can occur in various parts of the eye including the eyelid, iris, lens, choroid or optic disc. Colobomas of the iris and/or retina are the most common types (Hartshorne & Hartshorne, 1998).

What are the symptoms? Coloboma of the eye can have the physical manifestation of a keyhole-like shaped pupil or a notch or cleft on the pupil, but diagnosis is often delayed because the child is unable to verbalize their visual abilities.

About 80% of individuals with CHARGE syndrome are affected with one or more colobomas types (Hartshorne & Hartshorne, 1998)

Joubert syndrome and Lenz microphthalmia syndrome are two other syndromes that can be associated with colobomas. “Joubert syndrome is a very rare autosomal-recessive form of agenesis of the cerebellar vermis. Joubert syndrome is a neurological disorder that results in abnormal formation of the brain and affects balance and coordination” (Acosta & Pearl, 2003, p. 542). “Lenz microphthalmia syndrome is an extremely rare, genetic, X-linked syndrome of multiple congenital anomalies” Opitz, 2003, p. 654).

What causes a coloboma of the eye? There is evidence that colobomas are genetic and can be passed down through the family, although some cases have appeared in individuals with no prior family history. They eyes of a fetus develop quickly in the first three months of the pregnancy, during which the intricate parts of the eyes are formed and nourished through a gap known as the fetal cleft. Once the eyes are completely developed the gap should close, but when it does not close completely or not at all, the gap is known as a coloboma (Royal National Institute of the Blind, 2006).

What are the vision implications? Central vision may be affected depending on the location and size of the coloboma. A coloboma of the eye can range from mild to serious. The seriousness of the coloboma depends upon the extent and location of the gap. If the coloboma is located in the lens and is large, it may also affect the iris and choroids and increase the risk of retinal damage (Royal National Institute of the Blind, 2006).

When a coloboma is of the iris, vision is usually not affected. A coloboma of the retina may cause blind spots in the visual area of one or both eyes; the severity of the impairment depends on the placement of the coloboma. Optic nerve colobomas can cause the greatest impairment because the optic nerve is what transmits visual signals to the brain. Detailed and precise vision may be affected by a macula coloboma (Hartshorne & Hartshorne, 1998).

Is there treatment? A coloboma cannot be medically treated. If your child has a coloboma, he or she should be closely monitored according to the seriousness of the eye involvement. The risk of detached retina or glaucoma does increase slightly with a coloboma. Hospital visits might be more frequent during the early years but will depend on the individual. If the eye is small or severely distorted, then artificial eyes or lenses can be fitted for cosmetic and/or for promoting socket growth (Micro and Anophthalmic Childrens [sic] Society, n.d.). Children with colobomas may have an aversion to strong light because the pupil cannot react to light in the normal way. Some doctors may recommend tinted glasses to help deal with bright lights (Micro and Anophthalmic Childrens [sic] Society, n.d.).

References

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Fact Sheets from the Colorado Services to Children and Youth with Combined Vision and Hearing Loss are to be used by both families and professionals serving individuals with vision and hearing loss. The information applies to students ages birth through 21 years of age. The purpose of a Fact Sheet is to give general information on a specific topic. More specific information for an individual student can be provided through personalized technical assistance available from the Colorado Services for Children and Youth with Combined Vision and Hearing Loss Project. For more information call (303) 866-6681 or (303) 866-6605.