

What You Should Know About Colobomas

By David J. Browning, MD, PhD

The human eye is a multilayered structure that requires exquisite developmental orchestration during embryonic development. If the movement of precursor tissues does not proceed in a normal sequence from weeks 5-7 in utero, then congenital ocular abnormalities are the result. The most common of these is actually a family of related anomalies, all falling under the term coloboma. The embryonic eye begins as a plate of multilayered tissue. This plate begins to curve and deform into a sphere and the edges of the curving plate of tissue must meet at the bottom of the eye-to-be. When this goes awry, there will be absence of some ocular tissue in the bottom region of the eye. This absence of some normal ocular tissue is called a coloboma. Somewhere between 2 and 14 persons per 100,000 births will have a coloboma.¹

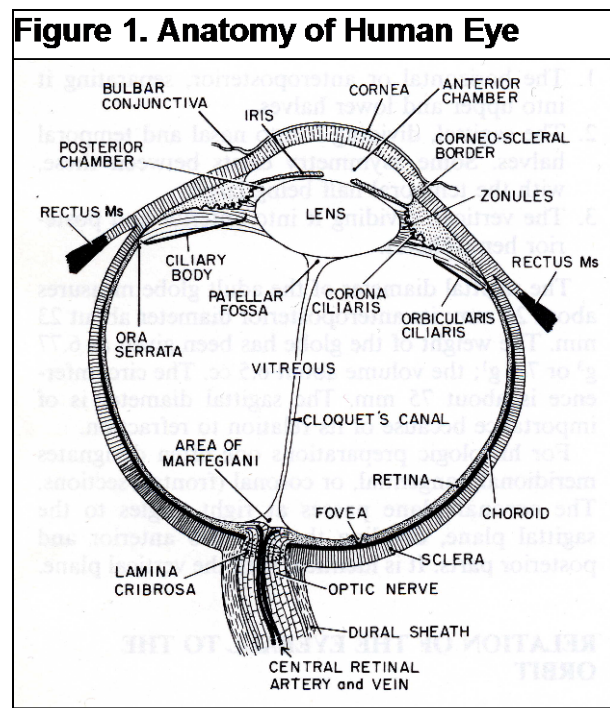
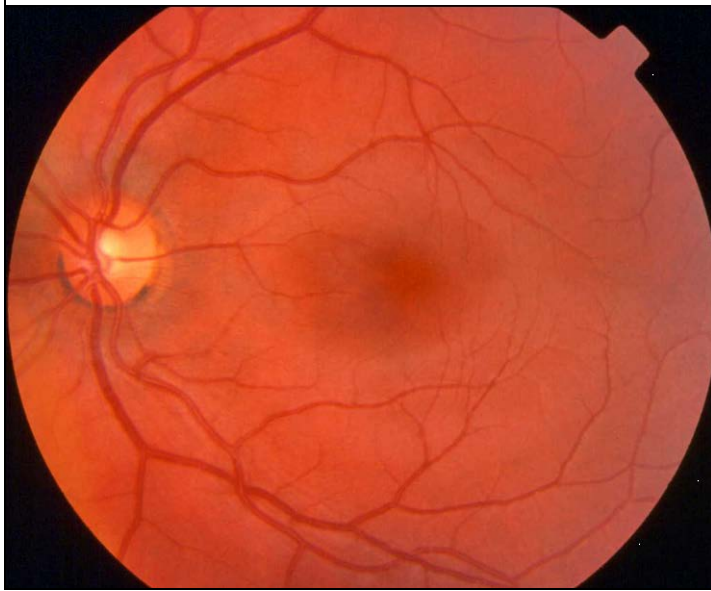


Figure 2. View of Normal Eye Interior

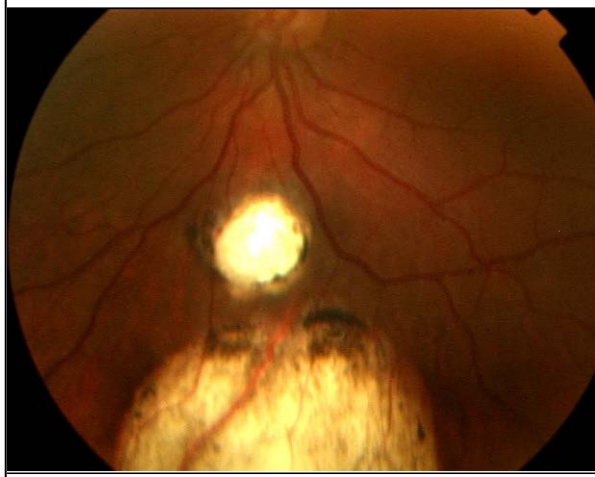


Types of Colobomas

The most common coloboma involves the iris. This produces a keyhole shaped pupil. Iris colobomas often do not affect vision. Less common, but potentially more visually handicapping colobomas involve the retina, choroid, optic nerve, or all three.

The retina is the neural lining of the back of the eye. The choroid is a layer of nourishing

Figure 3. View of Eye with Coloboma



blood vessels beneath the retina. The optic nerve connects the back of the eye to the brain and carries the neural signals from the eye to the brain. These normal structures are identified in figure 1. A normal appearance to the fundus, or back of the eye, as seen by the doctor looking through the eye's pupil, is shown in figure

2. A retinal-choroidal coloboma is shown in figure 3.

The white area with sharp borders has only a rudimentary retina, and no choroidal layer. The white sclera is seen through the clear remnant of undifferentiated retina. The level of visual handicap depends on the size and location of the absent retina and choroid. In the case shown, there was a lost area of visual field superiorly (since the upper field is perceived by the lower retina), but excellent central vision, because the central retina was uninvolved by the coloboma.

Colobomas can also affect the optic nerve. In one particular type, called a Morning Glory Anomaly, the optic disk has a large excavated funnel shape with an elevated rim of peripapillary tissue. The vessels emerge in a peculiar radiating pattern reminiscent of a flower.

Genetic Implications of Colobomas

Most colobomas occur sporadically and have no genetic implications. That is, in these cases, the affected patient will not have an increased risk of having a child with a coloboma. In some cases, colobomas are inherited, usually with an autosomal dominant pattern. In such cases, each child of an affected patient has up to a 50% chance of having a coloboma. In some cases a new genetic mutation is responsible for the coloboma. Colobomas have been described in association with trisomy 13, CHD7 mutation, 4p deletion, 14q deletion, and PAX2 mutation.²

Colobomas as Part of a Multisystem Condition

Sometimes colobomas are part of several congenital abnormalities. Occasionally ocular colobomas are associated with congenital forebrain malformations, basal encephaloceles, cleft lips, agenesis of the corpus collosum, and endocrine dysfunction.

One syndrome, called CHARGE, standing for Coloboma, Hear defects, Choanal Atresia, Retarded growth, Genital abnormalities, and Ear anomalies, is well recognized in newborns. These patients have a normal karyotype (the appearance of the 46 human chromosomes). Aicardi Syndrome is another multisystem syndrome involving females only. It reflects an X-linked dominant genetic mutation that is lethal to males. Affected females have microphthalmia (small eyes), orbital cysts, colobomas, retinal detachment, cataract, and mental retardation.

Consequences of Colobomas

Fifty-five percent of patients with a coloboma will have both eyes affected.² Twenty-two percent of patients with a coloboma will have some other ocular abnormality such as cataract or glaucoma.¹ Approximately 8% of eyes with retinal-choroidal colobomas eventually develop retinal detachment. Surgical repair is needed for these eyes. Smaller percentages develop abnormal blood vessels growing under the retina adjacent to the coloboma called choroidal neovascular membranes. Laser treatment is used to stop growth of such abnormal blood vessels. Children with colobomas can develop superimposed amblyopia, commonly called “lazy eye”, in which the brain involved in visual processing does not develop full potential. Patch therapy may be needed in such cases.

Colobomas represent a range of congenital ocular abnormalities. All patients with colobomas need periodic ophthalmic examination to detect the possible associated problems and treat them as necessary.

Once you have read this brochure, if you would like to find more in-depth information about colobomas, an excellent resource is the National Library of Medicine

website section called PubMed. It can be accessed via any search engine, or directly at this link, <http://www.ncbi.nlm.nih.gov/entrez/query.fcgi>. It includes an extensive database of articles published in peer-reviewed medical journals from all over the world.

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References

1. Skalicky SE, White AJR, Grigg JR, Martin F, Smith J, Jones M, Donaldson C, Smith JEH, Flaherty M, Jamieson RV. Microphthalmia, anophthalmia, and coloboma and associated ocular and systemic features. Understanding the spectrum. *JAMA Ophthalmology* 2013;131:1517-1524.
2. Shah SP, Taylor AE, Sowden JC, et al. Anophthalmos, microphthalmos, and coloboma in the United Kingdom: clinical features, results of investigations, and early management. *Ophthalmology* 2012;119:362-368.