Ocular Coloboma Associated with Iris Heterochromia: A Case Report

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Coloboma of the eyelids, lens, iris, ciliary body, choroid, retina and/or optic nerve arise from failed or incomplete closure of the embryonic fissure during development. A ocular colobomas may extend from the pupillary frill of the iris anteriorly to the optic disc posteriorly. The ocular colobomata may be associated with many systemic and ocular congenital anomalies. A coloboma can occur in one eye (unilateral) or both eyes (bilateral). People with coloboma may have no vision problems or may be blind, depending on severity. We report a case of unilateral typical coloboma of the iris, retina, and choroid associated with heterochromia of the iris.

Keywords: Coloboma, Embryonic fissure, Heterochromia of iris

INTRODUCTION

Coloboma is a term derived from a Greek word koloboma meaning mutilated or curtailed.¹ Coloboma of the eyelid, lens, iris, ciliary body, choroid, retina and/or optic nerve arise from failed or incomplete closure of the embryonic fissure of the neuroectodermal optic cup around 5-8 weeks of gestation.² This closure starts at the equator of the eye and continues anteriorly and posteriorly. An intrauterine insult during this period can result in the development of ocular coloboma of varying size and location. A coloboma may extend from the iris margin to the optic disc and involve one or more defects along the fusional lines.

The prevalence of ocular coloboma has been reported to be between 0.5 and 2.2 cases per 10,000 live births.³,⁴ The ocular coloboma may be sporadic or inherited and maybe associated with systemic and ocular disorders.⁵ A coloboma affecting the posterior segment of the eye can be unilateral or bilateral.

Anteriorly located coloboma often appears as a defect in the iris tissue. The coloboma may also involve the ciliary body and zonules. They are classified as “typical” if found in the inferonasal quadrant of the affected structure and “atypical” if found elsewhere. Posteriorly located coloboma can involve the optic nerve, retina, and choroid. If the retina is involved, it is reduced to glial tissue with the absence of retinal pigment epithelium (RPE) or choroid. This appears as a white area often with pigment deposition at the junction of the coloboma and normal retina.⁶ Chorioretinal coloboma is usually asymptomatic but may produce significant visual field defects in the superior quadrant.

People with coloboma may have no vision problems or may be blind; depending on severity of the size coloboma has effect on the vision can be mild or more severe depending on the size and location of the gap. If, for example, only a small part of the iris is missing, vision may be normal, whereas if a large part of the retina or optic nerve is missing, vision may be poor and a large part of the visual field may be missing. This is more likely to cause problems with mobility if the lower visual field is absent. Other conditions can be associated with a coloboma. The colobomas involving the eyeball should be distinguished from gaps that occur in the eyelids. While these eyelid gaps are also called colobomas, they arise from abnormalities in different structures during early development.

The colobomas can be associated with a mutation in the PAX2 gene.

Most often, isolated coloboma is not inherited, and there is only one affected individual in a family. However, the
affected individual is still at risk of passing the coloboma on to his or her own children.

In cases when it is passed down in families, coloboma can have different inheritance patterns. Isolated coloboma is sometimes inherited in an autosomal dominant pattern, which means one copy of an altered gene in each cell is sufficient to cause the disorder. Isolated coloboma can also be inherited in an autosomal recessive pattern, which means both copies of a gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of a mutated gene, but they typically do not show signs and symptoms of the condition.

Less commonly, isolated coloboma may have X-linked dominant or X-linked recessive patterns of inheritance. X-linked means that a gene associated with this condition is located on the X-chromosome, which is one of the two sex chromosomes. A characteristic of X-linked inheritance is that fathers cannot pass X-linked traits to their sons.

**CASE REPORT**

A 40-year-old female patient (Figure 1) presented to us with complaints of defective vision for near work since 2 months. She gave a history of defective vision for distance the left eye since childhood. The patient had normal intelligence and normal general features.

**On Examination**

Unaided visual acuity for distance was 6/6 (right eye) and 6/24 (left eye) not improving any further with pin hole or refraction correction. Her visual acuity for near was N/8 (both eyes), getting corrected to N/6 with +1.0 D sphere.

The ocular motility was full in both eyes with no tropia or phoria. Intraocular pressure measured by rebound tonometer 12 mmHg (right eye) and 18 mmHg (left eye). Anterior segment evaluation by slit lamp revealed normal eyelids, conjunctiva, cornea, and anterior chamber in both the eyes. The patient had light irides in the right eye with heterochromia of the iris (Figure 2), left eye had a typical keyhole iris coloboma in the inferonasal quadrant extending to the ciliary body (Figure 3). The anterior chamber angle and lens in both eyes were normal.

The posterior segment evaluation revealed a normal fundus picture in the right eye (Figure 4). The left eye showed a normal optic disc and a large retinochoroidal coloboma (Type 3 according to IDMAN classification) in the inferonasal quadrant seen 1 disc diameter inferior to the optic disc (Figure 5). No evidence of retinal detachment or choroidal neovascularization adjacent to the coloboma. The macula showed a dull foveal reflex.

Confrontational visual fields were normal in the right eye and with a superotemporal quadrant.

Figure 1: 40-year-old female patient

Figure 2: Right eye showing heterochromia of iris

Figure 3: Left eye showing iris coloboma in the inferonasal quadrant
Systemic examination was normal, and no associated congenital anomalies could be detected.

**DISCUSSION**

The coloboma is usually caused by the failure of the closure of the embryonic fissure. The coloboma can involve multiple layers of the globe, including the iris, ciliary body, lens, optic nerve, choroid, retina, and RPE.

The ocular coloboma may have associated congenital anomalies involving the cardiovascular, central nervous, musculoskeletal, gastrointestinal, genitourinary, and nasopharyngeal systems. The ocular coloboma may be associated with many systemic syndromes such as:

- Coloboma, heart defects, choanal atresia, retardation of growth, and ear anomalies syndrome
- Basal cell nevus syndrome
- Oro-facial-digital syndrome
- Aicardi syndrome
- Trisomy 13 (Patau syndrome)
- Trisomy 18 (Edwards syndrome)

Hence, management of a patient detected to have ocular coloboma should involve a thorough systemic evaluation to rule out these associated anomalies.

The ocular associations of ocular coloboma include:

- Heterochromia of iris (difference in the color of the iris between the two eyes)
- Microphthalmos
- Cataract
- Glaucoma
- Retinal dysplasia
- Nystagmus

The most common complications that are known to occur in patients of ocular coloboma are:

- Retinal detachment: This may occur in 23-42% of patients with retinochoroidal coloboma
- Cataracts of multiple varieties are associated with coloboma including pigment deposits, subcapsular, cortical, anterior and posterior polar, and total opacification
- Choroidal neovascularization

The patients with ocular coloboma should undergo a detailed ocular examination to detect the above mentioned complications. Appropriate correction of any refractive error should be done. If detected at an early age, occlusion therapy for amblyopia management may be necessary. These patients have to be kept on a lifelong follow-up to monitor the development of the complications.

**CONCLUSION**

We have reported a patient with unilateral ocular coloboma associated with heterochromia of the iris. The ocular coloboma may be unilateral or bilateral and may occur in isolation or in association with other congenital ocular and systemic anomalies. The diagnosis of ocular coloboma should therefore be followed up by a detailed systemic work-up and family screening and the patient should be monitored 6-12 monthly to detect any complications.

**REFERENCES**