Bilateral Coloboma of iris with posterior subluxation of lens in right eye – A case report and review of literature

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Abstract

A 45yr male, presented with defective vision of both eyes since one and half year, which has progressed gradually. Patient developed pain, redness, watering in right eye since one month. On examination of right eye, cornea was hazy, there was a coloboma of iris inferiorly and cataractous lens was subluxated posteriorly, fundus view is not possible.

Keywords: Coloboma of iris, subluxation of lens into posterior chamber

Introduction

Here we are presenting a case of Coloboma of iris of both the eyes, and subluxation of cataractous lens into posterior chamber, in right eye, and immature cataract in left eye.

Case report: A 45yr old male presented with gradual painless progressive fall of vision in both eyes since one and half year. History of redness, pain, watering, of right eye present. Patient takes mixed diet, known smoker and alcoholic since 20yrs. Family history: nil significant. General examination: patient is moderately built and moderately nourished. There is no pallor, cyanosis, icterus, clubbing, pedal edema, lymphadenopathy. Bp: 130/90mmhg, PR: 82/min, CVS: S1S2+, RS: clear. On ocular examination: there is coloboma at 6 ‘o clock position in both eyes. Eccentric pupil in both the eyes, right eye pupil...
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mid dilated, not reacting to light. Left eye pupil reacting to light. Lens: posteriorly subluxated lens with greyish brown opacity in right eye and in left eye the lens is greyish white. Vision: right eye: PL+, left eye: CF 2meters. Fundal examination: right eye: no glow, left eye: CLO, OD and Vessels: hazily seen. Intra ocular pressure examination: LE: 12.2mmHg, RE: 20.5mmhg.

**Discussion: Coloboma**

A coloboma-the absence of part or all of an ocular tissue-may affect the iris, ciliary body, choroid, or all 3 structures. Histologically, colobomas appear as an area nearly or entirely devoid of tissue. A coloboma (from the Greek *koloboma*, meaning defect) [1] is a hole in one of the structures of the eye, such as iris, retina, choroid, or optic disc. The hole is present from birth and can be caused when a gap called the choroid fissure, which is present during the early stages of prenatal development, fails to close up completely before a child is born. The classical description in a medical literature is a key whole shaped defect. A coloboma can occur in one eye (unilateral) or both eyes (bilateral). [2] Most cases of colobomata affect only the iris. People with colobomata may have no vision problems or may be blind, depending on the severity. It effects less than one in every 10,000births. Colobomas can be associated with a mutation in the *PAX2* gene. Eye abnormalities have been shown to occur in over 90% of children with Fetal Alcohol Syndrome. Colobomata form one of the commonest congenital malformations of the eye, in which the tissues of the uvea and the retina or their prolongation on to back of the iris are badly developed or deficient. They are due to defective closure of the embryonic cleft, in which case they occur in the lower part of the eye (typical colobomata). Colobomata of the iris found in other directions are called atypical. A coloboma of the iris may involve this tissue only, when it is usually pear shaped, the deficiency extending from the pupil towards, but not always as far as, the ciliary body, usually running downwards and slightly inwards [3]. A coloboma of the iris may also be associated with a similar coloboma of the lens, choroid and retina, or the later condition may occur alone. An inferiorly tapered cornea, giving it a pear like shape is a strong indicator of a defect in closure of the embryonic cleft giving rise to an iris and choroidal coloboma.

Other ocular malformations that include coloboma or are related to it:

Charge syndrome, a term that came into use as an acronym for set of unusual congenital features seen in a number of new born children. The term stands for coloboma of the eye, heart defects, atresia of nasal choana, retardation of growth and/or development, genital and/or urinary abnormalities, and ear abnormalities and deafness. Although these features are no longer used in making a diagnosis, the name has remained [4].

**Cat eye syndrome:** The term “cat eye” was termed because of the presence of vertical colobomas in the eyes of some patients.

**Patau syndrome** (trisomy13) including microphthamia, peters anamoly, cataract, iris and or fundal colobama, retinal dysplasia or retinal detachment, sensory nystagmus, cortical visual loss, and optic nerve hypoplasia.

**Treacher Collins syndrome**: Coloboma is part of a set of characteristic facies that features craniofacial malformations, such as down slanting eyes, ear anamolies, or hypoplasia of zygomatic bone and jaw (micrognathia).

**Treatment:** No treatment is available for visual impairment cause by coloboma at present. Specialized contact lenses can be used later in life for colobomos of iris, and glasses can be used to help with vision problems.

**Ectopia lentis**

Ectopia lentis refers to a displacement of the lens from its normal position. The lens may be completely dislocated, rendering the pupil aphakic (luxated), or partially displaced, still remaining in the pupillary area (subluxated). Ectopia lentis may be hereditary or acquired. Acquired causes include trauma, a large eye (e.g. high myopia, buphthalmos), anterior uveal tumours and hypermature cataract [5].

Management of subluxated lens in this case was by surgical extraction.

**References**