Isolated unilateral temporal coloboma of the lens

Abstract:

Lens coloboma is a rare congenital anomaly that occurs due to failure of the foetal fissure to close completely causing an incomplete formation of thelens, so usually inferonasal. Temporal lens coloboma is extremely rare; reported cases were associated with ocular or systemic anomalies.We report a rare caseof unilateral atypical temporal lens coloboma without any iris abnormality and posterior segment coloboma or any other ocular or systemic anomaly.

An 18-year-old female presented with a gradually progressive painless diminution of vision in her left eye for two years. There was no history of trauma, and family history was non-contributory. The systemic examination was normal. Ocular examination revealed a vision of 6/6 in the right eye and 6/60 in the left eye improving to 6/24 with pinhole and unilateral atypical temporalcoloboma of the crystalline lens in the left eye after dilatation. The rest of the anterior segment and fundus examination was normal. No abnormality was detected in the right eye. The best corrected visual acuity of 6/6 OD and 6/9 OS was achieved with spectacle correction at 6 months of follow-up.This case is a rare presentation of unilateral atypical coloboma located temporally without any iris abnormality and posterior segment coloboma or any other ocular or systemic anomaly.Early correction of refractive error and astigmatism results in good prognosis.

Keywords: Lens coloboma, atypical coloboma, temporal coloboma, embryonic fissure, zonules.

1. INTRODUCTION

"Lens coloboma is a rare congenital anomaly, characterized by notching at the equator associated with a deficiency or absence of the zonules at that site"[1]."Coloboma of the lens may betypical coloboma (occurring at the site of the embryonic fissure) or atypical coloboma (not occurring at the site of the embryonic fissure)"[1,2]. Typical lens colobomas are common, present infero-nasally and usually associated with coloboma of the uveal tract or other ocular developmental anomalies or systemic conditions [1-5].

Several cases of unilateral and bilateral lens coloboma with and without ocular or systemic abnormalities have been described in the literature [3-11]. Few case reports described the atypical lens coloboma located supero-temporally and infero-temporally associated with ocular or systemic abnormalities [3, 12]. Temporal lens coloboma is extremely rare; reported cases were associated with ocular or systemic anomalies [13]. Here, we present an unusual case of unilateral lens coloboma located temporally without any iris abnormality and retinochoroidalcoloboma and not associated with any other ocular or systemic anomaly.

2. CASE PRESENTATION

An 18-year-old female presented with decreased vision in the left eye,which she had noticed for two years. However, she did not consult for decreased vision or use spectacles.There was no history of trauma, and family history was non-contributory.

The systemic examination including the cardiovascular examination was normal. On ocular examination, visual acuity of 6/6 in the right eye and 6/60 improving to 6/24 with a pinhole in the left eye was noted. Undilated anterior segment examination was within normal limits in both eyes. Examination under dilatation revealed a coloboma located temporally in the left eye with a flattened and concave lens equator and absence of zonules (Figure 1). The iris was normal and the gonioscopy showed normal architecture of the angle structures. The fundus was normal; no coloboma was seen in the posterior segment with scleral indentation and indirect ophthalmoscopy with a 20D lens.Keratometry reading was 44.00 @ 180^0 / 44.25 @ 90^0OD and 43.50 @ 180^0 /43.75 @ 90^0OS.

The best corrected visual acuity of 6/6 was achieved without addition OD and 6/18 with -1.25 DS, -2.0DC at 180° OS. The patient was advised to wear a full-time spectacle and regular follow-up. At the 6-month follow-up, her BCVA was 6/6 OD and 6/9 OS.Patient refused tests for genetic analysis due to lack of affordability.

3. DISCUSSION

"Lens colobomais an incomplete formation of the lens due to failure of the foetal fissure to close completely, so usually inferonasal, and associated with a deficiency or absence of the zonules at that site"[1,2]."Segmentally defective or absent development of the zonules results in a coloboma of the lens secondary to the flattening of the equator in the region of the zonular defect. There is no actual loss of lens substance. The lens assumes a more spherical shape in that region, which is thought to be the cause of high astigmatism"[6]."Lens coloboma is considered to be a misnomer and might better be called coloboma of the zonules or of the ciliary body, as the process actually involves the segmental notching and contraction of the lens due to an absence of the zonules in that location"[1]."The zonules fail to form in the anterior secondary vitreous or the marginal bundle of Druault during the 3rd-4^{th} month of

gestation, due either to the failure of condensation and differentiation of the vitreous substance or to the failure of the lens to induce such changes"[14].

The embryonic origin of atypical coloboma is undetermined. Several theories have been proposed to explain the occurrence of atypical colobomasare the rotation of the foetal fissure, presence of accessory clefts or it may be because of intrauterine inflammation [15,16]. "Atypical ciliary body coloboma may be caused by persistence of mesodermal tissue from the embryonic vascular system" [17]."This tissue may block the forward growth of the neuroectoderm, producing a defect in the iris and ciliary body, that result inabsence of the zonules and lens coloboma". [17]

The occurrence of multiple colobomas, and atypical coloboma in the same eye with typical ones disproved the Vossius theory of foetal fissure rotation. Manz, in 1888, had assumed an atypical accessory fetal cleft to explain atypical coloboma, but no theory could be proposed due to the lack of evidence at that time. However, the presence of accessory fetal cleftsin chick embryos was reported long before in 1958 by Ammon, and later on it was reported in amphibian and mammal's embryos as well [15].

Szily (1907) described"atypical clefts in the edges of both optic cups in a 4-week-oldhuman embryo as due to the mechanical pressure of blood vessels passing over the optic-cup margin and hindering growth at those points. In his opinion, these accessory clefts are always anterior and thus could not be the cause of choroidal defects, though they do account for coloboma of the iris and ciliary body". Wolfrum (1908) described "a 5-week-old embryo in which the optic-cup margin showed three depressions other than the fetal cleft with blood vessels, but he differed with V. Szily in that he thought these vessels to be so delicate and fragile that they could not possibly offer any resistance to the growth of the cup margin". "He believed the fissure to be due to a variation in the growth energy of various portions of the cup margins, so that some regions grew at a faster rate than those immediately adjacent and thus caused such irregularities"[15].

Lindahl (1912) was "the first to study a series of human embryos in order to determine the frequency of accessory notches in the optic cup and relate them to the atypical colobomata"."Lindahl disagreed with the previous contributors in that the small accessory notches were an exceptional occurrence and could occur at any point. He found that besides the optic-cup fissure, there were four accessory notches, arranged symmetrically in such a fashion as to give the pupillary opening a five-sided shape, and these notches close in the same manner as does the fetal fissure. If, however, anything occurs to prevent their closure, atypical colobomata result. Lindahl considered it a transient phase in normal development and said that only when their closure is prevented do abnormal changes occur"[15].

Ronnes (1934) studied "a series of human embryos and concluded that notches occur in the margin of the embryonic optic cup due to variations in the growth energy of the margin". "The persistence of these notches is due to a loss in growth energy of the tissue at this point and the forward growth of the adjacent tissue. The presence of connective tissue in these regions can be an attempt to close the defect" [18].

Jennifer C. Hocking et al. proposed that "the embryonic origin of superior coloboma could not be explained by conventional models of eye development, reanalysed the morphogenesis of the dorsal eye, and revealed the presence of the superior ocular sulcus (SOS), a transient division of the dorsal eye conserved across fish, chicks, and mise"[19]. But this hypothesiscould not explain the occurrence of isolated temporal coloboma.

“Recent molecular studies have demonstrated that the earliest developmental processes are controlled by a complex network of eye field transcriptional factors (Pax6, Six3,and Six6), cell cycle regulators, and diffusible signalling molecules” [20-22].“These act in harmony to form different ocular compartments, regulate cell proliferation, migration, and apoptosis, and specify cell identities. There are two key genes sonic hedgehog (SHH) and PAX6. Sonic hedgehog regulates embryonic morphogenesis through an intracellular signalling network” (20, 23).“The PAX6 gene influences early ocular morphogenesis. Both these genes act as transcriptional regulators of many other genes that are also associated with coloboma. The timing of expression and site of expression is also important. Coloboma-related genes (SHH and SIX3) acting prior to eye development (before 20 days’ post conception) are associated with severe neurological deficits and systemic anomalies. Other coloboma genes acting later in eye development (after 20 days’ post conception) are usually associated with either milder CNS and systemic anomalies or isolated coloboma (PAX2, MAF1, CHX10, RBP4)”[20].

Sporadic, unilateral colobomas are most likely non-genetic. Environmental factors like maternal infection, drug intake, and vitamin A deficiency have also been reported to be associated with coloboma [24-27].

Isolated atypical temporal lens coloboma without any associated ocular or systemic anomaly is a rare condition and can be easily missed if not examined after mydriasis. Lens coloboma causes high astigmatic refractive error and if left uncorrected result in refractive, meridional or anisometropic amblyopia. So early diagnosis and treatment are essential. If severe astigmatic refractive error cannot be corrected by refraction, lens extraction with IOL placement is considered to prevent amblyopia.This patient presented late; even when she noticed decreased vision two years ago, she did not consult as she had no problems with reading or routine work and lacked medical facilities near her vicinity. She consulted us only when her family moved to the city. School and preschool screening are recommended for early detection and management of eye disease, especially congenital eye anomalies.

4. CONCLUSION

Temporal lens coloboma is extremely rare; previously reported cases were associated with ocular or systemic anomalies. This case report is noteworthy as a rare presentation of isolated unilateral temporal coloboma without any iris abnormality, and posterior segment coloboma or any other ocular or systemic anomaly. Isolated lens coloboma can be easily missed if not examined after mydriasis. Early correction of refractive error and astigmatism prevents amblyopia and results in a good prognosis. The embryonic origin of atypical coloboma remains unclear. Several theories have been proposed to explain the occurrence of atypical colobomas however, further research in molecular science is needed to determine the origin of the isolated atypical lens coloboma.

COMPETING INTEREST

Authors have declared no competing interest.

CONSENT

Written informed consent was obtained from the patient for publication of the case.

ETHICAL APPROVAL

As per international standard or university standards written ethical approval has been collected and preserved by the author(s).

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FIGURE CAPTIONS

Figure 1: Slit lampretro illumination view of the left eye under mydriasis showing temporal lens coloboma and absence of zonules (white arrow); normal zonular attachment visible inferiorly (black arrow).

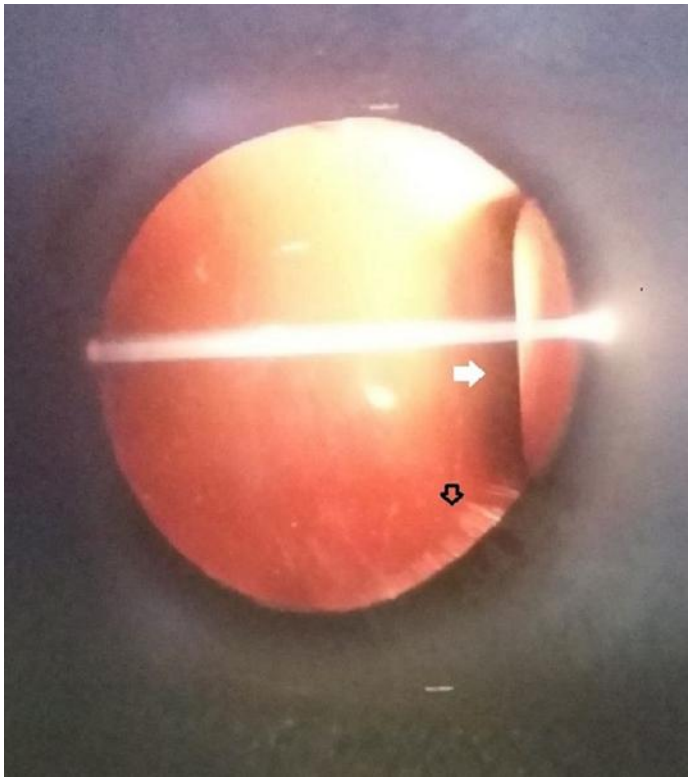

Figure: 1