Bilateral optic disc pit with maculopathy in a patient with cleft lip and cleft palate

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Optic disc pit (ODP) is small, gray-white, oval depression found at the optic nerve head. It is a congenital defect that occurs due to imperfect closure of superior edge of the embryonic fissure. Cleft lip and palate are also congenital midline abnormalities occurring due to defect in the fusion of frontonasal prominence, maxillary prominence and mandibular prominence. There is only one case report describing the occurrence of ODP in a young patient with cleft lip and palate who also had basal encephalocele. We describe a 52-year-old patient with congenital cleft lip and palate with bilateral ODP with maculopathy but without any other midline abnormality.

Key words: Basal encephalocele, cleft lip and palate, optic disc pit with maculopathy

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Optic disc pit (ODP) is small, gray-white, oval depression found at the optic nerve head. It was first described by Wiethe in 1882.[1] It is considered as a type of optic nerve head coloboma.[2] Histologically, it is described as a small congenital defect in the cribriform plate where a herniation of the dysplastic retina extends to the subarachnoid space in the optic nerve, which is surrounded by a layer of collagen-rich tissue.[3] Cleft lip and palate are also congenital midline abnormalities occurring due to defect in the fusion of frontonasal prominence, maxillary prominence and mandibular prominence.[4] There is only one case report describing the occurrence of ODP in a young patient with cleft lip and palate who also had basal encephalocele.[5] We describe a 52-year-old patient with congenital cleft lip and palate without any other midline abnormality with bilateral ODP with maculopathy.

Case Report

A 52-year-old man presented to our hospital with complaint of painless, progressive decrease in vision in both eyes for 6 months. There was no history of trauma, recurrent redness or any ocular surgery. The patient was born with a bilateral cleft lip and cleft palate, for which surgical repair was done in childhood and he was prescribed an obturator for the small residual cleft in the palate [Fig. 1]. On examination, the best corrected visual acuity was 20/100 in both eyes. The anterior segment was unremarkable in both eyes. The patient was orthophoric, all ocular movements were full and free, and pupillary reactions were also normal. The applanation tonometry by Golmann applanation tonometer revealed an intraocular pressure of 18 mm in both eyes. The fundus examination revealed bilateral ODP inferotemporally with maculopathy with hyperpigmentation at the macula [Fig. 2]. Refraction under cycloplegia did not reveal any significant refractive error. Telecanthus and hypertelorism were ruled out by measuring inner canthal distance, outer canthal distance and interpupillary distance.

The fundus flourescein angiography showed a normal arterio-venous filling with window defects at the macula but no leakage [Fig. 3]. The Spectral domain optical coherence

tomography of the right eye revealed an ODP with maculoschisis with thinning of the retinal layers, with a neurosensory detachment not communicating with the pit in the right eye while the left eye showed an ODP with maculoschisis with thinning of the retinal layers. There was also hyper-reflectivity of retinal pigment epithelium-choriocapillaries complex in both eyes [Fig. 4]. The general physical examination of the patient was unremarkable. The magnetic resonance imaging of the brain and spine done to rule out any other midline or neurological anomalies were within normal limits. Ultrasonography of the abdomen and pelvis was also done to rule out any renal abnormality and was unremarkable.

The patient was not advised surgery in view of poor visual prognosis due to retinal thinning and pigmentary changes at macula. He was also referred to the Department of Plastic Surgery, where he was advised to continue using the obturator.

Discussion

The optic disc pit is small, gray-white, oval depression found at the optic nerve head. It is a congenital defect that occurs due to imperfect closure of superior edge of the embryonic fissure.[2] It appears as a crater like indentation on the surface of optic nerve head usually with a steep temporal wall.[6] It usually occurs as an isolated anomaly. However, variants of optic disc colobomas especially morning glory syndrome have been associated with the papillorenal syndrome and basal encephaloceles.[5,7,8]

Papillorenal syndrome, also known as Renal-coloboma syndrome is an autosomal dominant disorder consisting of bilateral optic disc anomalies associated with hypoplastic kidneys, implicating PAX2 and PAX6 at chromosome 10 as the causative genes. These genes play an important role in the normal development of kidney, mid-brain, hind-brain, ears and eyes.[7]

Developmental anomalies of the optic disc have also been seen in patients with basal encephalocele associated with cleft lip and palate.[5,8] These include the morning glory syndrome, pale discs, coloboma of the optic nerve head, megalopapilla, optic nerve dysplasia, peripapillary staphyloma and ODP. The occurrence of basal encephalocele with mid-facial clefting is known as frontonasal dysplasia. It has a wide clinical spectrum consisting of hypertelorism, primary telecanthus, a broad

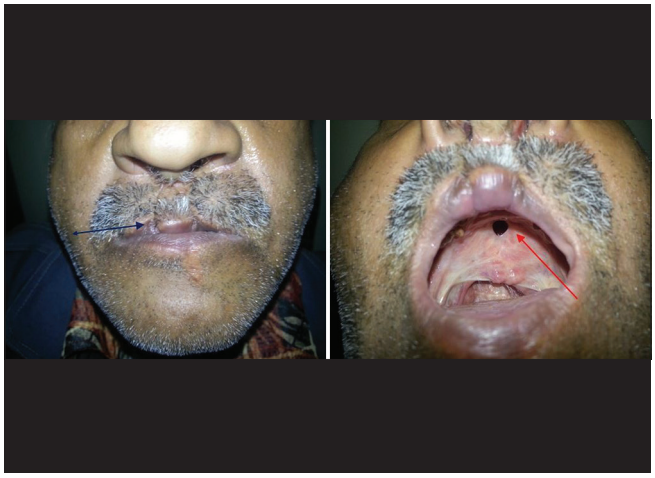

Figure 1: Showing repaired bilateral cleft lip (blue arrow) and palate with residual fistula (red arrow)

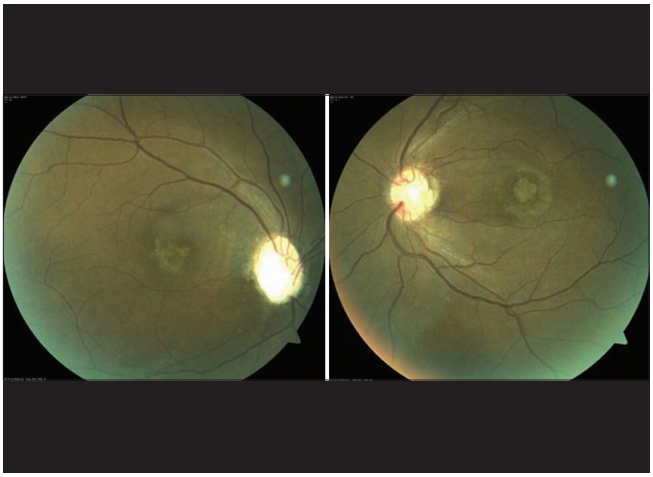

Figure 2: Fundus of both eyes showing optic disc pit inferotemporally with maculopathy

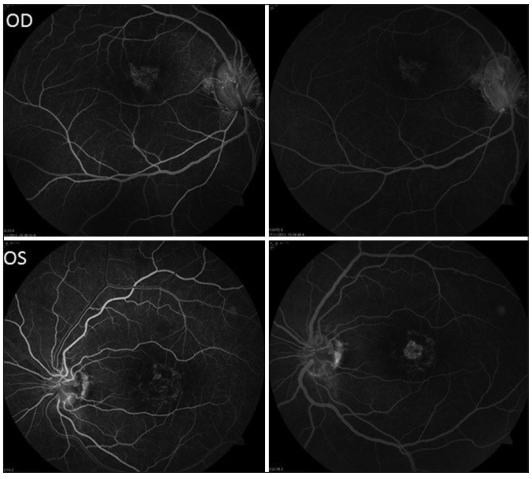

Figure 3: Fluorescein angiography of both eyes showing window defects at macula, but no leakage

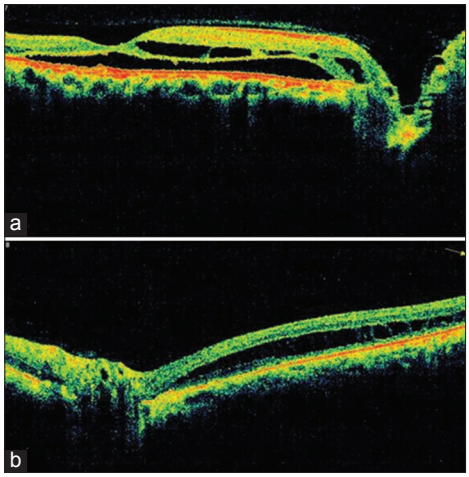

Figure 4: Spectral domain optical coherence tomography of right eye showing schisis and neurosensory detachment (a) and of left eye showing only maculoschisis (b)

nasal tip which is frequently cleft, median cleft upper lip and premaxilla, median cleft palate, anterior or basal encephalocele, and agenesis of the corpus callosum. The pathogenesis is unknown, but a nonseparation theory, which assumes that during closure of the neural tube, the ectodermal elements fail to separate, has been implicated.[9] The cleft lip and palate occurs due to nonfusion of frontonasal, maxillary and mandibular prominences. However, the presence of optic nerve anomalies is not explained by these theories. Itakura *et al.*, have suggested that if the sphenoid encephalocele prevents fusion of the palate, which precedes formation of the optic nerve, then the later development of the optic nerve could be abnormal.[9]

The co-existence of cleft lip and palate with ODP in the absence of basal encephalocele cannot be explained by the above-mentioned theory, and the only common pathway in the pathogenesis of these two independent disorders is the gestational age of 6–8 weeks, at which the frontonasal, maxillary and mandibular processes fuse and the embryonic fissure in the primitive eye closes. Hence, it may be hypothesized that an insult at this time may have caused these two anomalies to co-occur.

To the best of our knowledge, there is only one case of ODP with maculopathy, with cleft lip-palate with basal encephalocele reported in the literature.[5] This is the first case report of ODP with maculopathy with cleft lip and palate without a basal encephalocele found incidentally in the same patient or there might be a possible unexplained association. Thus, a possibility of ODP should be kept in mind, in a patient of cleft lip and palate with visual complaints, irrespective of the presence or absence of basal encephalocele.

The various options for management of maculopathy associated with ODP include gas tamponade with or without laser barrage and surgically with pars plana vitrectomy with or without internal limiting membrane peeling with or without gas tamponade.[3] It has been seen that multilayer schisis pattern of maculopathy has poorer visual prognosis and may require multiple surgeries as compared with serous detachment pattern.[10] Our patient also had multilayer schisis at the macula along with retinal layers thinning. Hence, no surgery was undertaken for this patient.

References

1. Wiethe T. A case of optic nerve deformity. Arch Augenheilkd 1882;11:14-9.
2. Halbertsma KT. Crater-like hole and coloboma of the disc associated with changes at the macula. Br J Ophthalmol 1927;11:11-7.
3. Georgalas I, Ladas I, Georgopoulos G, Petrou P. Optic disc pit: A review. Graefes Arch Clin Exp Ophthalmol 2011;249:1113-22.
4. Sadler TW. Langman's Medical Embryology. 11th ed. Philadelphia: Wolters Kluwer, Lippincott Williams and Wilkins; 2010.
5. Goldhammer Y, Smith JL. Optic nerve anomalies in basal encephalocele. Arch Ophthalmol 1975;93:115-8.
6. Kranenburg EW. Crater-like holes in the optic disc and central serous retinopathy. Arch Ophthalmol 1960;64:912-24.
7. Sanyanusin P, Schimmenti LA, McNoe LA, Ward TA, Pierpont ME, Sullivan MJ, *et al.* Mutation of the PAX2 gene in a family with optic nerve colobomas, renal anomalies and vesicoureteral reflux. Nat Genet 1995;9:358-64.
8. Hodgkins P, Lees M, Lawson J, Reardon W, Leitch J, Thorogood P, *et al.* Optic disc anomalies and frontonasal dysplasia. Br J Ophthalmol 1998;82:290-3.
9. Itakura T, Miyamoto K, Uematsu Y, Hayashi S, Komai N. Bilateral morning glory syndrome associated with sphenoid encephalocele. Case report. J Neurosurg 1992;77:949-51.
10. Skaat A, Moroz I, Moisseiev J. Macular detachment associated with an optic pit: Optical coherence tomography patterns and surgical outcomes. Eur J Ophthalmol 2013;23:385-93.

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