CASE REPORT

Aborted choroidal coloboma: fundus imaging and optical coherence tomography

Brijesh Takkar, Pradeep Venkatesh, Sudarshan Khokhar, Meghal Gagrani

Dr Rajendra Prasad Centre for Ophthalmic Sciences, All India Institute of Medical Sciences, New Delhi, India

Correspondence to
Dr Brijesh Takkar, britak.aiims@gmail.com

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SUMMARY

Choroidal coloboma is characterised by poor embryonic development of chorioretinal structures and is of different types. We present a case of choroidal coloboma where the retinal pigment epithelium was present, clinically and on imaging, but rest of the structures were poorly formed. This observation suggests that in some cases, fusion of the fetal cleft may occur aberrantly, resulting in an aborted choroidal coloboma.

BACKGROUND

Choroidal coloboma (CC) is an embryonic maldevelopment of the eye with a large spectrum of clinical presentation. The variations seen in cases of CC are essentially linked to the pattern in which the fetal cleft closes.^{1–3}

In this report, we have discussed clinical and imaging features of a case of CC which showed partial closure of few layers of the cleft. This case is also marked by clinical presence of the aberrantly closed fetal cleft. We discuss these findings as an aborted CC and compare them with similar documented cases.

CASE PRESENTATION

A 54-year-old man presented to our clinic with complaints of loss of near vision. He had low vision in the right eye (RE) since childhood but had not taken any optical/surgical treatment for the same. Best corrected visual acuity was 6/36 in the RE with a spherical error of −5.5 D and cylinder of −2 D at 90°, while left eye was 6/6. He was found to have a typical incomplete iris coloboma in the RE with early senile cataract. Fundus examination of the RE revealed a flat pear-shaped vertically oval lesion near the third branch of the inferior retinal vasculature, slightly nasal to the vertical meridian (figure 1A). The lesion was subretinal and had variegated appearance due to heterogenous pigmentation. The inferior part of the lesion had denser melanin and continued further towards peripheral retina as a vertical tract. This tract ended in another densely pigmented flat postequatorial lesion which had depigmented lacunae within it. Rest of the RE fundus was normal. The left eye was within normal limits. Intraocular pressure was normal in both the eyes.

The patient was diagnosed to have RE aborted choroidal coloboma (ACC) with senile cataract and anisometropic amblyopia.

INVESTIGATIONS

Spectral domain optical coherence tomography (OCT) of the RE macula revealed grossly normal macular architecture with slightly shallow foveal dip (figure 1B). An inferiorly imaged OCT scan through the lesion revealed grossly thin neurosensory retinal layers in the region of the ACC, with prominent scleral–choroidal interface. The retinal pigment epithelium (RPE)–Bruch’s membrane complex appeared hyper-reflective due to melanin. Interestingly, the outer retinal layers suddenly terminated at the border of the ACC and could not be seen within it. Also the choroidal vasculature was seen as poorly developed within the lesion, evidenced by a few islands of large choroidal vessels (figure 1C). Neither OCT or ultrasound could detect scleral ectasia.

TREATMENT

Surgery was deferred because of minimal cataract and poor visual prognoses, and the patient was advised regular follow-up.

DISCUSSION

CC results from failure of fusion of the fetal cleft in the developing optic cup.^{1–3} Commonly advocated theories include the epiblastic theory, mesodermal theory and the lenticular theory.³ However iris, ciliary body and CC can occur independently of each other and in different proportions, which has led to different suggestions regarding the manner and direction in which the cleft closes.⁴ Of note, Duke-Elder mentions ‘an aberration in fusion of the fetal cleft does not necessarily entail a failure of fusion’.³ The findings of this case seem to be a manifestation of this hypothesis. To the best of our knowledge, we could not find a prior report of this nature in literature.

In the earliest stages of normal fusion of the fetal cleft during the second month of gestation, the outer layer of the optic cup invaginate into the cleft. These cells are the future RPE cells and they are said to develop melanosomes only after the fusion has occurred.⁵ This is the reason why CC do not have pigment. The sensory retina develops in the cleft region from the neuroblastic layers and the choroidal vasculature develops later in the second month of gestation. Histopathology of CC and OCT-based studies have consistently reported poorly developed outer sensory layers in the region of the coloboma while the inner retinal layers

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Figure 1 (A) Montage of fundus photographs depicting the inferiorly placed flat and vertically oval pear-shaped lesion (white arrow) which has variegated pigmentation. A tract of altered subretinal pigmentation emanates from the inferior part of the lesion (arrowheads) and continues to fuse into a densely pigmented postequatorial black lesion (black arrow). These findings are suggestive of an aborted choroidal coloboma (ACC). (B) Optical coherence tomography (OCT) scan of the macular region shows nearly normal architecture with a shallow foveal dip. Inset photograph shows the iris coloboma in the anterior segment infrared OCT projection. (C) Inferiorly placed horizontal OCT scan through the ACC. The blue vertical arrows delimit the area of the ACC. The overlying vitreous is healthy but the retina is grossly thin and the outer retinal layers suddenly terminate at the border of the ACC. The retinal pigment epithelium is thick and densely hyper-reflective due to excessive melanin. The choroidal vasculature is poorly formed with scattered large vessels (star). The sclera has been imaged as a thick and densely hyper-reflective structure (arrowheads). The inset shows the OCT raster scan positioned over the ACC.

continue as the intercalary membrane which bridges the open cleft.^{4 6 7}

The presence of dense pigment in our case is an indirect evidence of fusion of the cleft, which corroborates with the intact RPE layer seen in the OCT scans through the lesion (figure 1C). Therefore, in this case the CC had been aborted and the outer layer of the optic cup had actually fused. However due to the ‘aberration’ mentioned earlier, the outer neuroblastic layer failed to develop normally into outer retina and the choroidal vasculature was very poorly developed (figure 1C). As seen in the OCT scans, in some areas, the RPE appeared to directly overlie the sclera. These features have been reported on OCT before in established cases of typical CC by Gopal *et al.*^{6 7}

Prior to this case, similar cases of ‘partially pigmented flat lesions’ or ‘posterior dipping of the ora serrata’ with minimal scleral ectasia have been labelled as forme fruste colobomatous lesions.^{6 7} As these lesions cannot develop into a typical CC later in life, the term ACC is more befitting. The vertical tract seen in our case (marked as arrowheads) actually represents the line of aberrant fusion of the cleft, and such subretinal tracts in this location should alert the ophthalmologist towards possibility of an ACC. In her original description of the types of CC, Ida Mann has described Type 5 CC as a well-circumscribed circular white ectatic defect which is seen in the peripheral retina and

Type 6 CC as a vertically oval area of pigmentary disturbance with choroidal thinning but no white area.⁸ The CC in our case is most probably an aborted form a combination of Types 5 and 6 coloboma as per that classification system. The inferior most dark pigmented lesion having depigmented lacunae within it could have been a peripheral lesion of a type 6 coloboma as per Ida Mann’s classification system,^{8–10} or even a patch of solitary congenitally hypertrophic RPE (CHRPE). In fact, the pear-shaped ACC in our case shares common features like pigment hypertrophy and defective outer retinal layers with CHRPE,¹¹ though its location along with presence of iris coloboma and thinning of choroidal vasculature indicate otherwise.

Learning points

- ▶ An aborted choroidal coloboma (ACC) may manifest as a solitary lesion in the inferior fundus.
- ▶ Presence of retinal pigment epithelium should not deter a surgeon from diagnosing CC.
- ▶ Aberrantly closed fetal cleft may manifest as a clinically apparent vertical tract.
- ▶ Such lesions are likely to be innocuous by their own virtue, though other associations of ocular coloboma may impact the visual acuity and should be ruled out at the earliest.

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Competing interests None declared.

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