

Combined cavitory anomalies of the optic disc with maculopathy

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DESCRIPTION

Cavitory optic disc anomalies can cause maculopathy. The most common of these associated with maculopathy is the optic disc pit. Very rarely, maculopathy can accompany optic disc coloboma. We report a case of combined cavitory disc anomalies (coloboma and disc pit) with maculopathy.

A 22-year-old young woman presented with decreased vision in the left eye (LE) for 6 months. The right eye (RE) had poor vision since childhood. Best corrected visual acuity was 2/60 in the RE and 6/24 in the LE. Fundus examination of the RE showed a retinochoroidal coloboma (RCC) involving the disc and macula (**figure 1A**) and the LE showed a cavitory defect involving inferior half of the disc with adjoining macular thickening and retinal pigment epithelium changes (**figure 1B**). Optical coherence tomography (OCT) scans through the left macula showed retinal schisis with foveal detachment (**figure 1C**).

Closer observation of the left optic disc revealed a circular greyish focal dip on the temporal margin suggestive of a pit (**figure 2A**, arrow). OCT scans through the optic disc in this region showed a narrow slit-like tract extending from the schitic retina to the depths of the lamina cribrosa (**figure 2B,C**, arrows). Vertical line scan through the larger cavitory defect of the left optic disc showed a widened scleral canal, posteriorly displaced lamina cribrosa with dysplastic retina lining the wall (**figure 2D**, arrow) consistent with a colobomatous defect.

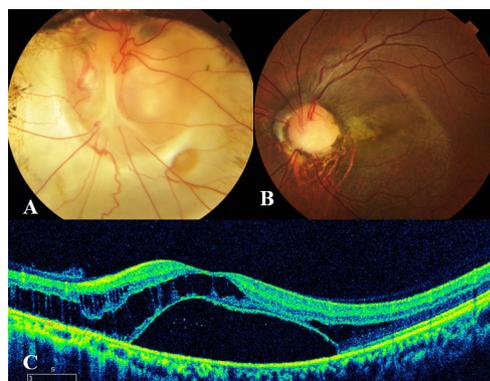


Figure 1 (A) Fundus photograph of the right eye showing a coloboma involving the disc and inferior retinochoroidal layer. (B) Left eye shows a cavitory anomaly of the inferior half of the optic disc with adjoining retinal pigment epithelium changes and macular thickening. (C) Optical coherence tomography scan through the left macula showing retinal schisis in outer layers and subretinal fluid.

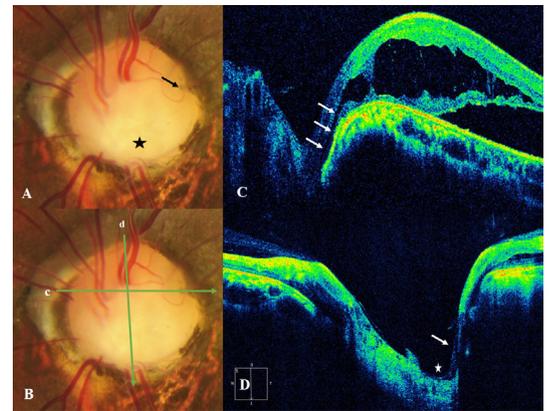


Figure 2 (A) Colour photograph of the left optic disc showing a sharply delineated, colobomatous excavation of the inferior half of the disc (asterisk) and a pit which is seen as a greyish focal depression on the temporal edge of disc (arrow). (B) The positions of the horizontal and vertical line scans corresponding to (C) and (D) are shown. (C) Horizontal line scan through the pit showing a narrow tract (arrows) extending deep into the lamina cribrosa. (D) Vertical scan through the disc showing an excavation in the inferior portion of the disc (asterisk) with herniation of the dysplastic retina (lacy appearance) (arrow).

Hence, a diagnosis of LE optic disc pit maculopathy with disc coloboma was made.

Optic disc pits are congenital cavitory anomalies of the disc which can present in isolation or in combination with an RCC.^{1,2} The coexistence of a disc coloboma with a pit is a rare entity. Maculopathy in such cases may develop either from disc pit or rarely from the coloboma.^{2,3} In a study on cavitory disc anomalies using swept source OCT, the author observed that the anatomical and physiological barrier between the subarachnoid space and the vitreous cavity is compromised by the presence of pits and colobomatous defects in the disc. In the present case, the presence of both these defects in close proximity could have increased the risk of developing maculopathy.

Optic disc colobomas need to be differentiated from acquired causes like glaucomatous cupping. A characteristic bowl-shaped white glistening defect with well-delineated edges in an abnormally large disc should raise suspicion of a disc coloboma. Dysplastic retina herniates into the excavation and takes on a lacy appearance on OCT.¹ The macular thickening in cavitory disc anomalies presents with retinal schitic changes which may be misdiagnosed as macular oedema of retinal vascular origin. Careful evaluation of the disc for pit lesions and



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demonstration of anatomical defects using OCT helps in arriving at the correct diagnosis. Vitrectomy with or without tamponade and laser have been shown to be successful in resolving maculopathy changes secondary to optic disc pits.^{4,6}

This case emphasises the need for a meticulous imaging of the optic nerve head in cases with cavitory disc anomalies and maculopathy to arrive at the correct diagnosis and plan appropriate management.

Learning points

- ▶ Cavitory disc anomalies can present with varied combinations including coloboma and pit.
- ▶ Meticulous imaging of the disc and macula is needed to demonstrate morphological changes and hence correct diagnosis.
- ▶ The management of such cases changes as many of the conditions are treatable.

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Disclaimer Case reports provide a valuable learning resource for the scientific community and can indicate areas of interest for future research. They should not be used in isolation to guide treatment choices or public health policy.

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