Disc within Macular Coloboma

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Figure 1: Fundus examination revealed punched out defect at posterior pole with disc inside the defect.
Figure 2: Fundus examination revealed punched out defect in macular area with normal disc.

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Introduction

A 25 year old male presented to us with history of diminution of vision in Right eye noticed 1 month ago, when he went to ophthalmologist for regular examination.

On examination best corrected visual acuity was 20/120 in Right eye & 20/20 in Left eye. Anterior segment examination was essentially normal in both eyes. Fundus examination revealed punched out defect at posterior pole with disc inside the defect (Figure 1). After careful examination it was noticed to be punched out defect in macular area with normal disc (Figure 2). Left eye fundus was normal. Pre-natal history was not significant. Investigations did not support any inflammatory cause.

Absence of ocular tissue resulting from fetal fissure to close is called Ocular coloboma. It’s defined atypical, when it involves the areas not originating from embryonic cleft.

Macular coloboma is an atypical coloboma, which should be differentiated from inflammatory causes like ocular toxoplasmosis. Histological evidence of lack of choriocapillaries & RPE in coloboma area is evaluated in a case.