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Clinical-Medical Image

Rare Presentation of Unilateral Chorio-Retinal Coloboma in a Patient with Bilateral Retinitis Pigmentosa

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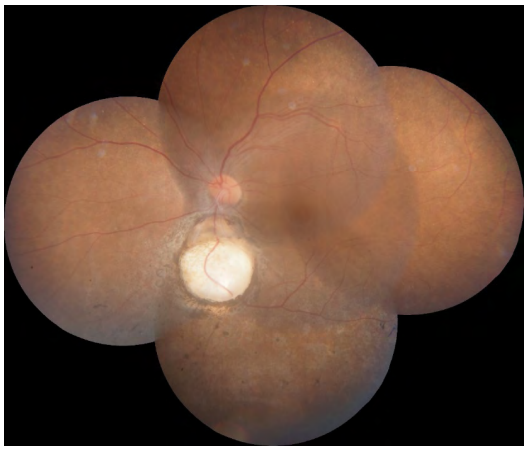

Figure 1: Colobomas which occur due to defective closure of the embryonic fissure.

Clinical Image

Inherited Retinal diseases are an incurable cause of visual morbidity occurring in children and young adults. Retinitis pigmentosa is one of the commonest examples of inherited retinal diseases with a prevalence of 1 per 4000 individuals. Colobomas are known to occur in approximately 1 per 10,000 people. Colobomas occur due to defective closure of the embryonic fissure between 6^{th}–7^{th} weeks of development fetal life, and are under diagnosed as they don't cause symptoms until they involve the disc or macula or both. Retinitis pigmentosa and choroidal coloboma are known to usually exist as two separate entities. The fundus photo shows features of Retinitis pigmentosa, with an isolated chorio-retinal coloboma inferior to disc, in a 30 years old lady, who, presented with decreased peripheral vision and nyctalopia. There was no history of consanguinity. On evaluation she was found to have 6/6 vision bilaterally. Anterior segment was unremarkable in both eyes. Right eye fundus showed features of Retinitis pigmentosa. Humphrey Visual Fields 24-2 tests showed bilateral visual field defects, explaining the loss in peripheral vision. After extensive literature review, such association of unilateral coloboma in a patient with bilateral Retinitis pigmentosa, with no other systemic anomalies is extremely rare, and has been reported only in a few cases (Figure 1).

Keywords: Coloboma; Retinal diseases; Eye

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