Rare association of juvenile retinoschisis with retinochoroidal coloboma

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DESCRIPTION

A 20-year-old man presented with history of diminution of vision since 8 years. On examination his best corrected visual acuity was 20/20 in the right eye and 20/200 in the left eye. Anterior segment examination was essentially normal in both eyes. Right eye fundus showed stellate maculopathy with schitic changes in the foveal region (figure 1A). Left eye fundus showed stellate maculopathy with schitic changes in the foveal region and an isolated wedge-shaped retinochoroidal coloboma in the inferior fundus (figure 1B). The retinochoroidal coloboma in the left eye was not involving the disc or macula. Optical coherence tomography of both eyes showed altered foveal contour, increased central macular thickness and schitic changes in the inner retinal layers with splitting of the retinal layers at the level of outer plexiform and

inner nuclear layer (figure 1C, D). Electroretinogram of both eyes showed reduced photopic and scotopic responses. Both eyes showed reduced oscillatory potential (figure 1E). A diagnosis of juvenile retinoschisis was made based on fundus features and ancillary tests. The patient and family members were counselled regarding the disease nature, prognosis and genetic association. Topical dorzolamide BD was prescribed for the macular schitic changes.

Congenital retinoschisis is a rare bilateral condition characterised by vitreous degeneration and splitting of the retina between the nerve fibre and ganglion cell layers. Patients will typically have a cystic-like stellate maculopathy or a foveal schisis with or without peripheral retinoschisis.^{1 2} It is an X-linked recessive trait with mutation in RS 1 (XLR1S1) gene, occurring mostly in males.² Optical coherence tomography and electroretinogram aids in confirmation of diagnosis. In our case, the macular schisis and electroretinogram findings were in favour of the juvenile retinoschisis. Reduction of a wave amplitudes in our case can be explained by affection of both photoreceptors and inner retinal layers.³ There have been few reports wherein acquired retinoschisis and retinochoroidal coloboma have been described in the same patient. Juvenile retinoschisis with concurrent presence of morning glory coloboma has been described in one report so far.⁴ The concurrent presence of retinoschisis and coloboma in our case can be explained in terms of mutations in the XLR1S1 gene, which may affect embryonic differentiation that can also lead to colobomatous change as described in earlier case report.⁴ However, in our case the genetic evaluation was not conducted as the patient and family members did not give consent for genetic testing. The presence of peripheral coloboma in our case can be due to same genetic origin of both disease and it can also be linked with the embryonic evolution of the eye as the coloboma in our case is inferior and peripheral in line with the closure of embryonic fissure.

Figure 1 (A) Right eye fundus showed stellate maculopathy with schitic changes in the foveal region. (B) Left eye fundus showed stellate maculopathy with schitic changes in the foveal region and an isolated wedge-shaped retinochoroidal coloboma in the inferior fundus. (C) (Right eye) and (D) (left eye) optical coherence tomography of showed altered foveal contour, increased central macular thickness and schitic changes in the inner retinal layers with splitting of the retinal layers at the level of outer plexiform and inner nuclear layer. (E) Electroretinogram of both eyes showed reduced photopic and scotopic responses with reduced oscillatory potential.

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Patient's perspective

Medical counselling helped me and my family members to have better understanding of the disease nature, its inheritance pattern and prognosis.

Learning points

- ▶ Association of juvenile retinoschisis with coloboma or any other embryonic defects should be kept in mind owing to common differentiation of structures embryonically due to mutations in XLR51 gene.
- ▶ Counselling of patients and the family members in any rare disorder regarding nature of disease is an important part of clinical practice for improvement of quality of clinical care.
- ▶ Regular follow up should be advised to the patient and parents and the patient should be monitored for progression.

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