

Retinal detachment in a child with severe early childhood onset retinal dystrophy

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DESCRIPTION

A 10-year-old boy was seen with the complaints of blurring of vision in his left eye (LE) since 3 months. The child had a history of poor vision in both the eyes since early childhood. There was no history of trauma. Birth and family history were insignificant. On presentation, the best corrected visual acuity in right eye (RE) was 2/60 and hand motions in LE. The spherical equivalent of refractive error was +2.5D in RE and could not be determined in LE. Fat atrophy of bilateral orbits with deep-set eyeballs were noted. A left dominant alternate divergent exotropia of 90 prism dioptres was also seen. Slit lamp examination of LE revealed few pigments on corneal endothelium, posterior synechiae at 5 o'clock position, small (mention size 1 or 2 mm) posterior subcapsular cataract and retrolental pigments. A relative afferent pupillary defect was present in LE. The intraocular pressure as recorded with non-contact tonometry was 14 mm Hg and 10 mm Hg in RE and LE, respectively. Anterior segment of the RE was normal. Dilated fundus evaluation of RE revealed an isolated macular coloboma measuring three disc diameters in its longest dimension (figure 1), while LE had a total retinal detachment with inferior subretinal bands and membranes, along with a macular coloboma as in the fellow eye. No retinal break could be localised, both eye (BE) optic disc was pale with slightly thin arteries. No pigmentary were apparent in either eye. Optical coherence tomography of BE macula revealed an excavation of macula with altered retinal and choroidal architecture (figure 2).

The patient was diagnosed as severe early childhood onset retinal dystrophy (SECORD) with

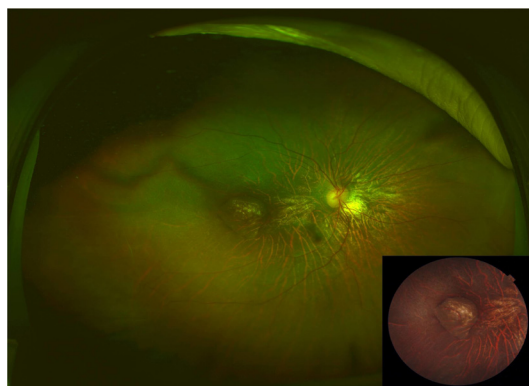


Figure 1 Ultrawide fundus image of right eye showing a pale optic disc, attenuated arteries and a macular coloboma. Inset fundus photograph shows a magnified image of the macular coloboma.

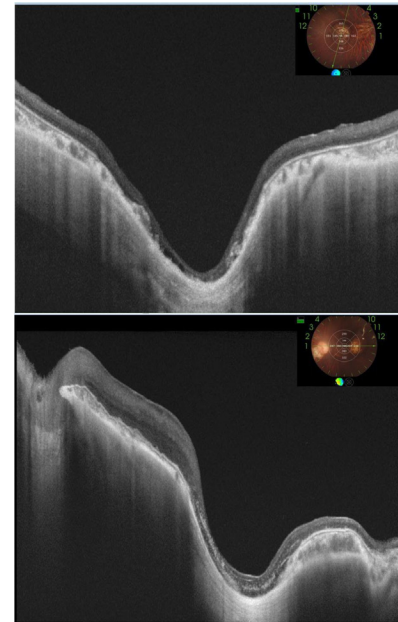


Figure 2 Swept source optical coherence tomography through macular region of right (up) and left (below, after surgery with silicone oil in situ) eyes with the inset picture showing axis of the B scan. Fovea of both eyes show poorly differentiable inner retinal layers, thinning of an excavated choroid and scleral ectasia. While right eye fovea is thin (98 u), left eye fovea is thickened (388 u). Right eye additionally shows focal areas of outer retinal cavitation, while left eye shows multiple scattered intraretinal hyper-reflective areas.

macular coloboma in BE, with a rhegmatogenous retinal detachment (RRD) in LE (in lieu of subretinal proliferative vitreoretinopathy (PVR) and pigments behind the lens}. Detailed systemic examination of the child and ocular examination of family members revealed no abnormalities. Twenty-five-gauge vitreoretinal surgery was done under general anaesthesia for LE along with 360° encircling band. No retinal break could be discerned during the surgery despite scleral depression. Two rows of laser photocoagulation was performed near the equator in 360°. Visual acuity in LE was 1/60 at third week of follow-up, with attached retina (figure 3). Patient and family counselling was done and low vision aids suggested with a future plan of elective silicone oil removal. Gene mapping was denied by the guardians of the child due to expenditure.

This child was diagnosed to have SECORD in preference to Leber's congenital amaurosis (LCA) due to preservation of some form vision in the second decade of life and as historically the vision



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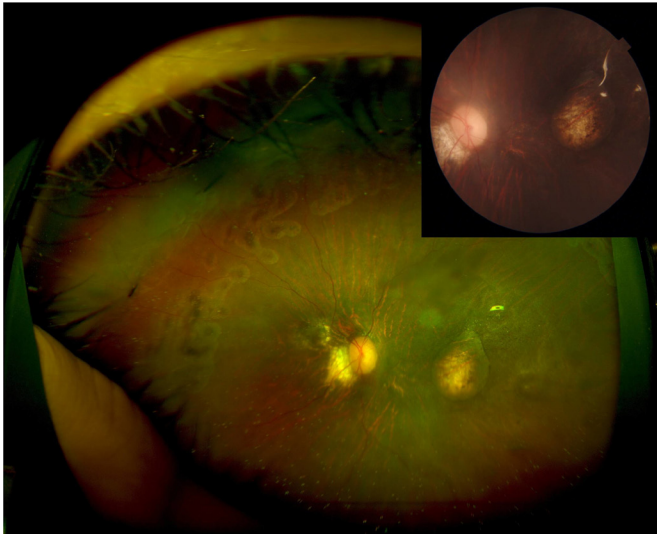


Figure 3 Ultrawide fundus image of the left eye after surgery with silicone oil in situ. Retina is attached, while site of posterior retinotomy, endolaser and indentation due to the scleral buckle can be seen. Inset fundus photograph shows a magnified view of the macular coloboma.

loss had been deciphered at around 5 years of age.¹ Pseudo-macular coloboma, though classically described with LCA, may be present across different phenotypes of retinitis pigmentosa (RP). It represents extensive tissue loss at fovea.¹ To our knowledge, this is the first report of an RRD with LCA/SECORD. While localised RD related to coats-like reaction may occur, a case of tractional RD has been described previously in a case of CEP290-associated LCA.^{1,2} Our case and the case described by Cunningham *et al*² indicates the need to observe for complications and manage them appropriately despite such a setting of progressive retinal degeneration.

RD is a rare accompaniment of RP, perhaps due to intraretinal migration of pigment that obliterates the potential subretinal space and makes RD unlikely. For this reason, RRD rarely occurs in individuals who have the typical tapetoretinal pigmentation.³

Rishi *et al*³ have described a series of 31 patients of RP who developed RRD. Authors noted precocious vitreous degeneration with detachment of posterior vitreal cortex in a male child to have a high OR of developing RRD. Such degeneration was also noted in our case during surgery. While the least age of presentation in that series was 4 years, a diagnosis of LCA/SECORD was not specified by the authors in their series, and the authors had also included traumatic retinal dialysis and syndromic RP in their study sample. Interestingly, they found myopia to be a risk factor for RD in RP, while our patient was hypermetropic. Other considerations in such patients is a possibly higher risk of PVR.

Learning points

- ▶ Severe early childhood onset retinal dystrophy (SECORD) or Leber's congenital amaurosis (LCA) may rarely be associated with rhegmatogenous retinal detachment. This occurrence could be because of vitreous degeneration in absence of the pigmentary retinopathy that is typical of retinitis pigmentosa.
- ▶ Although SECORD or LCA can have progressive retinal degeneration, complications can occur during follow-up and may need urgent management. Hence, periodic surveillance should be done.

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