Retinopathy of prematurity in oculocutaneous albinism

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We report a case of retinopathy of prematurity (ROP) in an infant with oculocutaneous albinism (OCA), with the challenges faced in diagnosis, and subsequent management. Poor fundus contrast and blanching of retinal vessels on indentation caused significant visualization problems in detection of ridge and extraretinal vessel proliferation. Careful examination revealed zone 2 Stage 3 ROP with preplus disease in both eyes. Laser photocoagulation was attempted, but laser uptake was poor. The disease regressed over 3-week close follow-up. ROP along with OCA is a rare finding. There is a need for high index of suspicion and caution while screening and managing such babies.

Key words: Hypopigmented fundus, oculocutaneous albinism, retinopathy of prematurity

Access this article online

Quick Response Code:

Website: www.ijo.in

DOI: 10.4103/ijo.IJO_931_17

Oculocutaneous albinism (OCA) is a rare inherited genetic disorder of melanin pigment synthesis characterized by a significant reduction of the pigment in various structures of the body. The disorder exists with a broad spectrum of presentation ranging from only a partial depigmentation to complete lack of pigment from all structures of the body.[1]

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Manuscript received: 30.09.17; Revision accepted: 29.11.17

Cite this article as: Chandra P, Salunkhe N, Kumar V, Kumawat D, Tewari R. Retinopathy of prematurity in oculocutaneous albinism. Indian J Ophthalmol 2019;67:960-2.

The presence of retinopathy of prematurity (ROP) in a child with OCA is a rare scenario, probably the first of its kind to be reported to the best of our knowledge. The diagnosis of ROP is mainly clinical. OCA presents with nonpigmented retina with easily visible large choroidal blood vessels that hamper the visualization of small immature vessels near the ridge in ROP. The described case highlights the importance of careful screening and diagnostic-therapeutic challenges in the management of ROP in the presence of OCA.

Case Report

An infant born at 29 weeks gestation with birth weight of 900 g was brought by parents at 32-week postmenstrual age for ROP screening. The infant had a history of respiratory distress requiring mechanical ventilation in the neonatal period. The infant had generalized uniform skin and hair depigmentation. Pediatric evaluation had ruled out any possible syndromic association. Parents did not exhibit any feature of ocular or cutaneous albinism.

The eyelashes and iris were depigmented in both the eyes. The pupils were dilating well without any evidence of tunica vasculosa lentis. Fundus screening with indirect ophthalmoscope with 28D noncontact lens and RetCam 3 wide-field imaging (Clarity Medical Systems, Inc.,) revealed light colored fundi due to the lack of pigment in the retinal pigment epithelium (RPE) and easily visible large choroidal vessels [Fig. 1]. A diagnosis of OCA was made.

Both the eyes had mild retinal vascular tortuosity and dilatation in posterior pole suggestive of preplus disease. On initial examination, a demarcation ridge could be noted in zone 2 in right eye easily but barely in the left eye suggestive of Stage 2 disease. Minimal pressure from the scleral depressor/RetCam probe resulted in blanching of retinal vessels and the vascular ridge. However, careful examination with a 20D lens revealed brushfire like fibrovascular proliferation into vitreous cavity at the area of ridge in both eyes suggestive of Stage 3 disease [Fig. 1].

The ridge neovascularization progressively worsened over the next week, though plus disease was still not obvious. RetCam-assisted fluorescein angiography was performed, but the imaging was unclear due to poor dye circulation, and vessel blanching by RetCam hand-piece pressure. Therefore, after obtaining written informed consent from the parents, laser photocoagulation was planned in both eyes. During the laser procedure, great difficulty was encountered in visualizing the avascular area under scleral indentation because of poor contrast, underlying choroidal vessels, and blanching of retinal vessels. The entire avascular area was lasered, but laser uptake was poor due to lack of pigment in RPE. In spite of increasing the laser power and duration, laser reaction could not be clearly seen.

The ridge regressed over the next 3 weeks, and vessel caliber became normal in both eyes [Fig. 2]. The laser scars were sparsely seen in the temporal periphery. Cycloplegic refraction and regular follow-up were advised to the parents. Genetic testing for OCA could not be performed due to unavailability of facility at our center. Based on uniform generalized depigmentation, we believe it to be OCA Type 1.

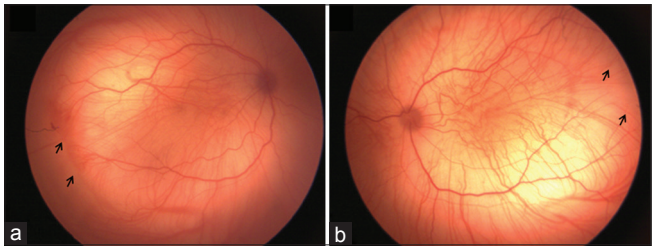

Figure 1: (a) Fundus photograph of the right eye showing hypopigmented retina along with easily visible large choroidal blood vessels, zone 2 retinopathy of prematurity with the presence of brushfire like neovascularization extending into the vitreous depicting Stage 3 disease (black arrows); (b) fundus photograph of the left eye showing faint ridge (black arrows) in zone 2 with avascular retina anterior to the ridge

Discussion

ROP is a preventable disease of preterm infants. If undetected, it may lead to irreversible blindness. Timely screening and management help in reducing the risk of complications. Identifying ROP is crucial and depends on good indirect ophthalmoscopy skills using scleral depression.

OCA is characterized by the presence of pale fundus due to lack of melanin pigment in the RPE and easily visible large choroidal vessels.[1] ROP with OCA is a chance occurrence and coexistence does not exhibit causality. However, the screening for ROP in such conditions is particularly difficult due to poor contrast for detection of ridge and fine neovascular fronds. Moreover, scleral indentation for peripheral retinal examination leads to blanching of the small fine vessels, and can also obscure "plus" disease in such patients. RetCam imaging is also challenging, and gentle handling of RetCam probe is essential as even slight pressure leads to blanching of the fine blood vessels thereby causing an error in staging of disease. It is easy to mistake choroidal vasculature as complete retinal vascularization in these eyes. The retinal and choroidal vessels can be differentiated clinically on the basis of color, caliber and vascular branching pattern. Retinal arteries and veins run parallel, are dark red, thin, well delineated, and divide in a dichotomous fashion towards the periphery while the choroidal vessels are light pink, ribbon-like, randomly branching vessels arranged in a segmental/lobular manner with peripheral anastomosis.

Laser photocoagulation is the preferred treatment in eyes with Type 1 prethreshold ROP.[2] Patients with OCA lack the vital RPE pigment necessary for laser uptake. Performing laser photocoagulation in such patients becomes difficult owing to poorly visible laser reaction and increased propensity of

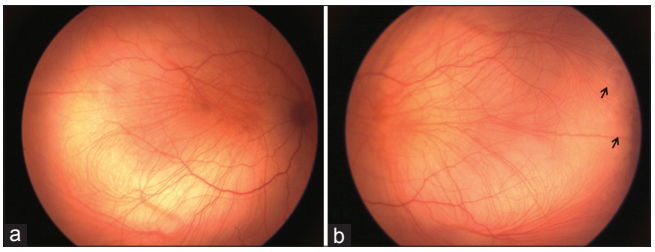

Figure 2: (a) Post-laser fundus photograph of the right eye revealing the disappearance of the ridge along with regressed retinopathy of prematurity; (b) Post-laser fundus photograph of left eye showing faintly visible laser scars in the temporal avascular retina with regressed disease (black arrows)

complications on increasing laser power. Poor visibility of laser spots may lead to inadvertent skip avascular areas and progression of disease.[3] Laser at high power and increased duration has been attempted previously in such eyes with rhegmatogenous retinal detachments to coagulate hemoglobin within the choriocapillaris and indirectly lead to retinal adhesion effect.[4,5] Owing to the failure in detection of laser reaction, cryotherapy can be tried as a second step in such eyes as retinal blanching/whitening can be better picked up.[4] However, the efficacy of chorioretinal scar from both of these procedures is poor in such eyes as both need RPE for absorption of energy and scar formation. Intravitreal anti-vascular endothelial growth factor therapy may be advantageous over laser/cryotherapy in these cases as it overcomes the limitations of visualization, has less risk of overtreatment, allows for faster regression and better preservation of visual field. However, the long-term safety remains a concern for these agents.

We faced similar challenges while performing laser photocoagulation in our patient. There was difficulty in visualizing the laser spots as well as keeping a track of the ridge which would blanch on scleral indentation. Follow-up examinations, however, showed faint laser scars and disease regression. Although we did not require repeat laser procedure, laser augmentation may be needed if skip areas are visible. Although the disease activity was apparently less in the left eye and disease may have regressed spontaneously on follow-up, we did notice a progressive worsening of ridge neovascularization and with persisting visibility issues, laser treatment was a safer choice to obtain good predictable outcomes.

Conclusion

This case highlights unique challenges in diagnosis and management of ROP with OCA. It emphasizes the importance of careful screening and gentle scleral indentation for accurate diagnosis and management of disease.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

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