Photo Essay

Retinopathy in incontinentia pigmenti

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Key words: Incontinentia pigmenti, peripheral avascular retina, retinal detachment

An 8-month-old girl was brought in with wandering eye movements as noted by the parents. The child was delivered at term by cesarean section with birth weight of 1.9 kg with uneventful neonatal period. Skin examination revealed whorls and streaks of pigmentation on the abdomen, arms, and thighs [Fig. 1]. On ocular examination, child could not fixate and follow light in both eyes. Anterior segment examination was normal. Fundus examination by indirect ophthalmoscopy revealed total retinal detachment with disorganized retina in the right eye Fig. 2a]. Left eye revealed normal posterior pole, peripheral fibrovascular proliferation with localized tractional detachment in inferotemporal quadrant in the left eye with avascular retinal periphery [Fig. 2b, RetCam system, Clarity Medical System, Pleasanton, CA, USA]. An abnormal vitreoretinal interface was made out with OCT (optical coherence tomography) in the left eye [Fig. 3]. Fluorescein angiography confirmed peripheral retinal non-perfusion with abnormal arborization and leakage of the peripheral retinal vessels [Figs. 3-6]. Prophylactic laser photoablation was done to avascular retinal periphery [Fig. 7]. Both parents had normal skin pigmentation; fundus screening was advised. The patient followed-up 3 months later and peripheral retinal neovascularization was regressed [Fig. 8]. Skin pigmentation did not show any noticeable change.

Discussion

Incontinentia pigmenti is a multisystem disease with a variable expression. It is of an X-linked dominant inheritance and is lethal in the male fetus.[1] Wald *et al*. noted the similarity of the retinopathy in incontinentia pigmenti to that of retinopathy of prematurity (ROP) and suggested that peripheral retinal ablation, as proposed by the Cryotherapy for ROP Cooperative Group, may be beneficial.[2] Laser photocoagulation, intravitreal bevacizumab, and ranibizumab have also been used for management of retinal neovascularization.[3–5]

Incontinentia pigmenti should be considered in differential diagnosis of patients with peripheral retinal vascular non-perfusion, peripheral retinal neovascularization, or infantile retinal detachment. Differential diagnosis includes ROP, familial exudative vitreoretinopathy (FEVR), and Norrie's disease. Referral for skin involvement, dental disorders, speech therapy, and pediatric neurology might be necessary.

In conclusion, retinal vascular abnormalities are common in incontinentia pigmenti which can lead to exudative retinopathy and can be prevented with laser photocoagulation of peripheral avascular retina.

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

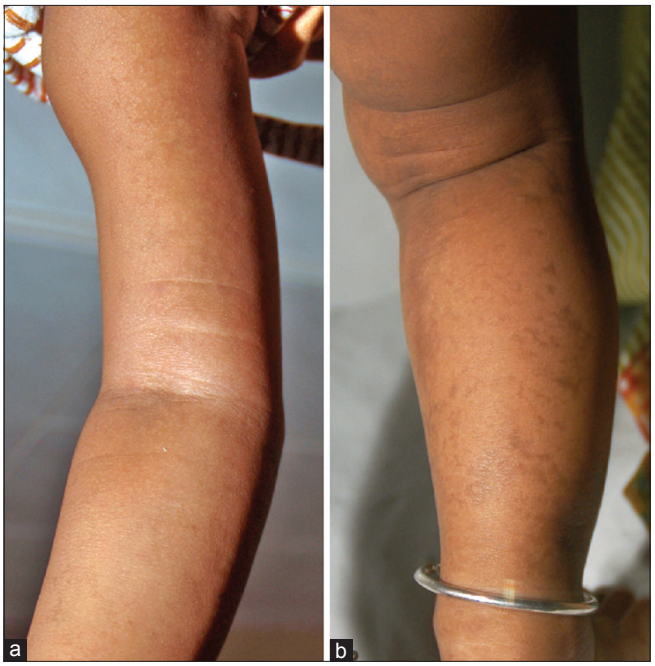

Figure 1: Streaks of skin hyperpigmentation are seen on the arms (a) and thighs (b)

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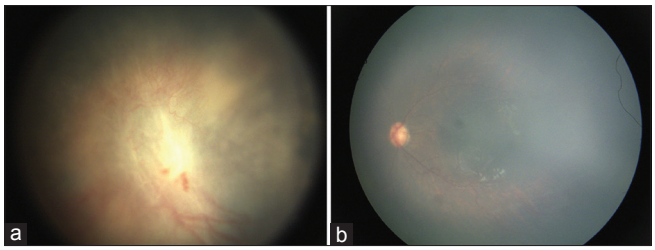

Figure 2: (a): Fundus photo of right eye shows total retinal detachment with disorganized retina. (b): Fundus examination of the left eye reveals a normal posterior pole

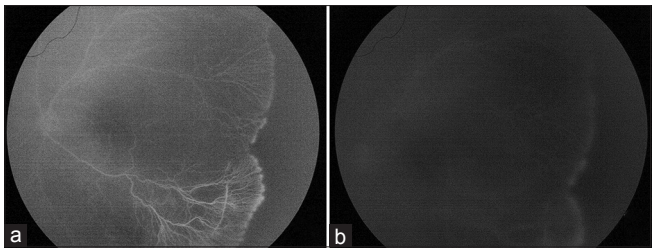

Figure 4: (a): Fundus fluorescein angiogram (FFA) of the left eye shows peripheral temporal retinal capillary non-perfusion (arteriovenous phase). (b). Late phase FFA shows diffuse hyperfluorescence due to leakage in the corresponding area

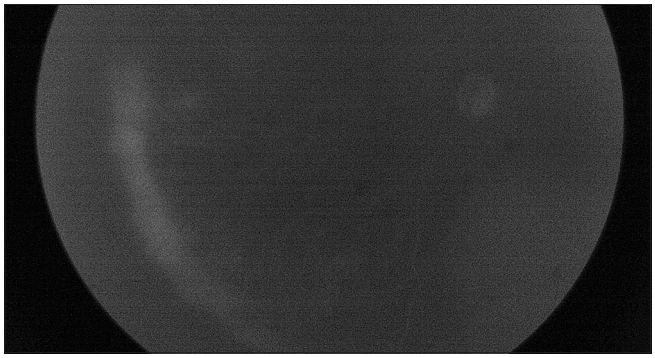

Figure 6: Late phase fundus fluorescein angiogram shows diffuse hyperfluorescence due to leakage in the corresponding area as Fig. 5

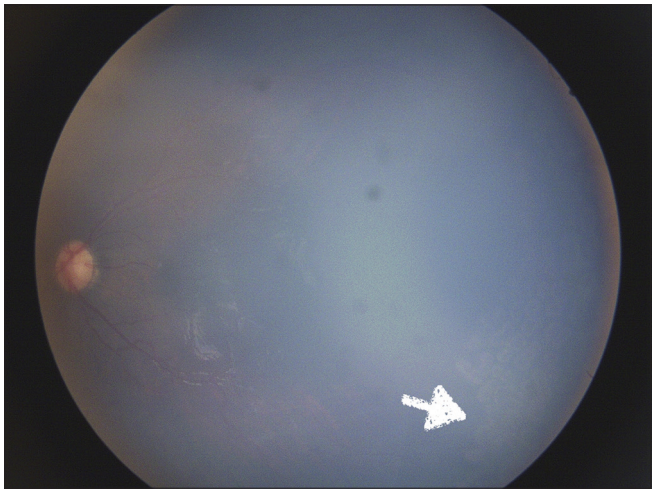

Figure 8: Color fundus photo shows old scatter laser marks in the retinal periphery

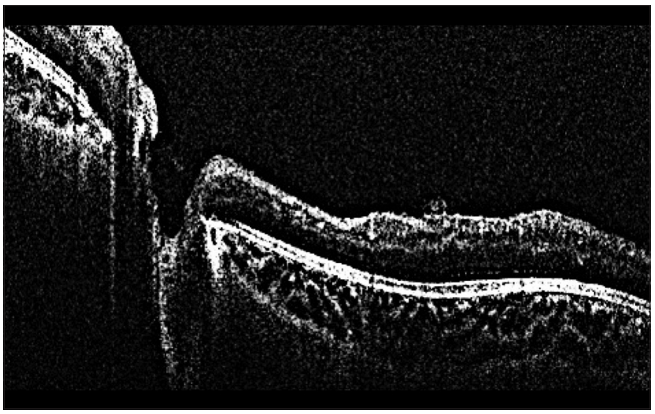

Figure 3: Optical coherence tomography of the left eye shows abnormal vitreoretinal interface at posterior pole

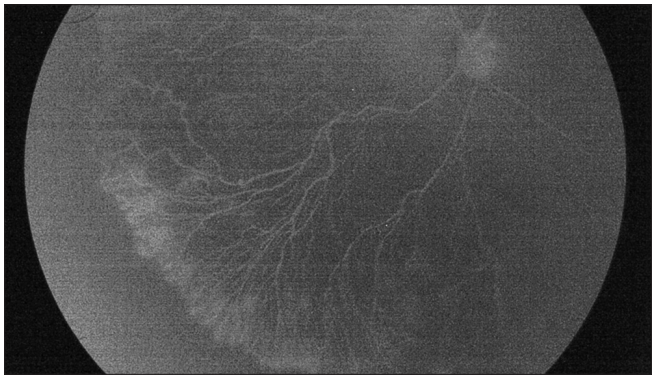

Figure 5: Fundus fluorescein angiogram shows peripheral nasal retinal capillary non-perfusion (arteriovenous phase)

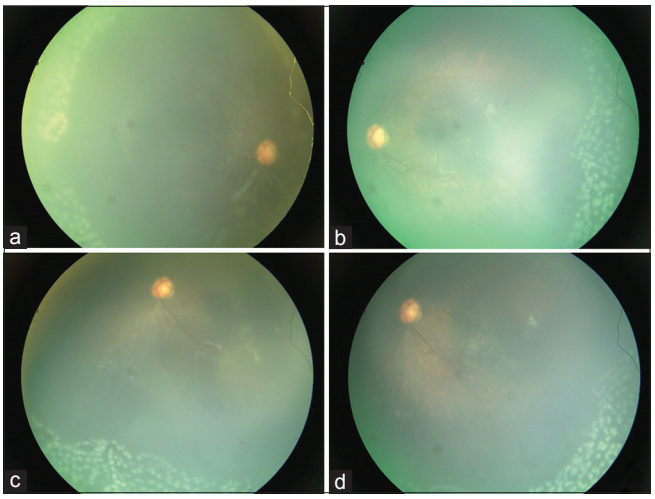

Figure 7: Color fundus photos (a-d) shows fresh scatter laser marks in the avascular retinal periphery

and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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