Case Report

Retinal Detachment Surgery in Oculocutaneous Albinos Patient

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This is an interventional case report of a 30 years old male patient of oculocutaneous albinism with high myopia associated with retinal detachment in both eyes. He had best corrected visual acuity of hand movement in right eye due to fresh rhegmatogenous retinal detachment and perception of light in fellow eye because of long standing retinal detachment. Micro incision vitrectomy surgery was done in right eye after flattening of retina with PFCL, endolaser photocoagulation attempted but failed to get adequate burns over tear and detached retina. Then cryopexy was performed around the retinal tear and 360° of peripheral retina.

Albinism is a genetically determined, heterogeneous group of melanin synthesis disorder which involves either hypopigmentation of eyes only (ocular albinism) usually inherited as X – linked or occasionally AR autosomal recessive (AR)[1] or hypopigmentation involves eyes along with skin and hairs known as Oculocutaneous albinism (OCA) is inherited as autosomal recessive.

Oculocutaneous albinism may be: a) Tyrosinase-positive albinos synthesize varying degree of melanin (pigment). The hairs may be white, yellow or red and darken with age; skin color is very pale at birth but usually darkens by age of 2 years, b) Tyrosinase-intermediate albinos has no melanin pigment at birth but varying degree of pigmentation with age, c) Tyrosinase – negative albinos are incapable to synthesize melanin and have no pigments in skin, hairs and ocular structures throughout life.

The Albinos usually have impaired visual acuity (VA) 20/40 to 20/200 or <20/200 due to foveal hypoplasia[1-3], may have high refractive error, strabismus or nystagmus at 2 – 3 month age and hypopigmented fundus with iris transillumination and amblyopia secondary to strabismus or anisometropia.

Albinos are at increased risk of cutaneous basal and squmous cell carcinomas before 4th decade of life[1]. The failure of endolasers in reattachment of retina due to melanin deficiency therefore Cryopexy is method of choice for attachment of sensory retina to retinal pigment epithelium and choroid by with freezing with cryo-probe to surface of sclera.

CASE REPORT

A 30 years old male patient of Oculocutaneous albinism (figure 1), with high myopia using 7.50 D and 8.00 D concave (minus) spherical lenses in right & left eyes respectively, visited our tertiary care eye hospital for sudden deterioration of vision in his right eye. His best corrected visual acuity (BCVA) was hand movement (HM) due to rhegmatogenous retinal detachment with macula off associated with giant retinal tear superiorly. BCVA in fellow eye was perception of light only, B-scan ultrasound showed total retinal detachment (figure 2). Vitreoretinal surgery is planned with guarded prognosis in fellow eye.

For retinal detachment in right eye micro incision vitrectomy surgery (MIVS) was performed with 23-G system and PFCL was used per operatively to flatten the retina. Argon endolaser photocoagulation was tried at edges of giant retinal tear but it failed to get adequate reaction due to insufficient amount of melanin pigment in retina; ultimately cryopexy around giant retinal tear and 360° to peripheral retina was carried out in order to reattach the retina and

finally silicone oil was injected into vitreous cavity as internal tamponade. At succeeding follow ups and after 3 months of surgery, retina was found attached and BCVA was improved from HM to 6/60 (decreased visual acuity was due to foveal hypoplasia).

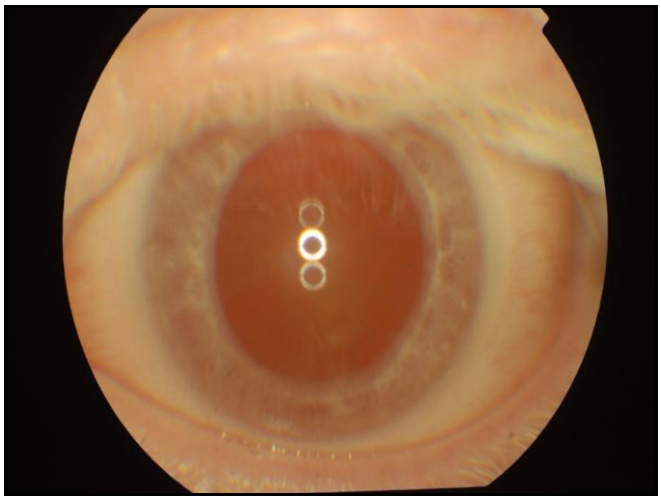

Fig. 1: Color Photograph – right eye of Albinos patient

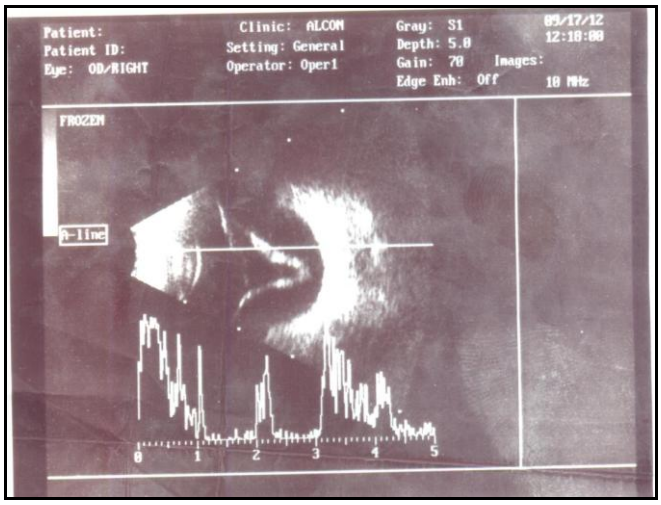

Fig. B: Scan of Right Eye showing total detachment of retina

DISCUSSION

Albinism resulting from mutation from genes coding for Tyrosinase, the enzyme used in synthesis of melanin; at least 14 genes are distinguished[4]. The melanin is synthesized from an amino acid tyrosine, which is first converted to dihydroxyphenylalanine through copper-containing enzyme Tyrosinase[5]. The Tyrosinase activity is assessed by using the hair bulb incubation test, which is reliable after five years of age.

Oculocutaneous albinism is most frequent type; whereas ocular albinism is caused by mutation in GPR 143 gene[6]. To diagnose the albinos patients the denaturing high performance liquid chromatography (DHPLC) couple with direct sequencing is an effective and exact test[7].

Rarely albinism is associated with inherited systemic disorders like Chediak-Higashi syndrome involving the leucocytes abnormality resulting in recurrent pyogenic infection and another is Hermansky-Pudlak syndrome which causes bleeding disorder due to platelet dysfunction that may be responsible for intra-operative bleeding in albinos patients, therefore pre-operative evaluation has significance.

With extensive literature search it was found that only two cases of retinal detachment in the albinos have been reported. One case of RRD in Oculocutaneous reported by Yang JW et al in Korea, shown the failure of endolasers in reattachment of retina due to melanin deficiency but successful result of cryopexy in case if albinism[8]. While the second case ocular albinism with RRD reported by M. Hiroshi et al in Japan shown reattachment of retinal hole & lattice degeneration with photo coagulation by Krypton laser[9].

R.J Hanson et al in their study on therapeutic challenge in proliferative diabetic retinopathy in OCA also suggest that no visible endo laser burns were seen in cases of retinopathy and ultimately cryopexy was done[10].

We are reporting as first case of RRD in Oculocutaneous albinism in our country and it observed that Argon endo laser photocoagulation is not successful to reattach the retina but cryopexy is more useful and effective.

In conclusion, the albinos patient should be rehabilitated promptly and timely; it is suggested that cryopexy should be attempted for retinal detachment in albinos patients and further work up to be needed.

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