

Vitreoretinal Intervention in Trivial Trauma Related Ocular Complications in Three Cases of Ehlers-Danlos Syndrome

V. H. Karambelkar¹, G. Gadre², Ankit Sharma³, Viraj Pradhan⁴

¹Professor, ²Assistant Professor, ^{3,4}Resident, Department of Ophthalmology

Krishna Institute of Medical Sciences and Research Centre, Karad, Satara, Maharashtra, INDIA.

Corresponding Addresses:

¹virajharikarambelkar@gmail.com, ²anks.sharma166@gmail.com, ³virajpradhan@gmail.com

Case Report

Abstract: Vitreoretinal surgical intervention was done in three separate cases of serious ocular complications in Ehlers-Danlos syndrome. In the first case with endophthalmitis with perforated ciliary-staphyloma - pars plana vitrectomy with intravitreal antibiotics was done, followed by scleral patch graft in second stage. The second case, a one-eyed patient with equatorial staphyloma with 360 degrees supra-choroidal haemorrhage and vitreous haemorrhage was taken for suprachoroidal drainage with vitrectomy and silicon oil insertion. Third case with globe rupture with anterior dislocation with massive suprachoroidal haemorrhage had no PL. After 2 weeks on steroids patient was developed perception of light with faulty projection of rays and he was taken for vitreoretinal intervention twice. At 6 months follow-up the cases had encouraging vision with the first two maintaining stable globe with corrected visual acuity of 6/60 and the third case had finger counting close to face. EDS type VI is associated with ocular problems and to the best of our knowledge this is the first report (*pubmed central and google search*) with vitreo-retinal intervention.

Keywords: Ehlers-Danlos syndrome, hyperextensibility, kyphoscoliosis, vitreoretinal.

Introduction

EDS is a group of inherited connective tissue disorders, caused by a defect in the synthesis of type 3 collagen resulting in fragility of joints, blood vessels, bowel and viscera, with symptoms such as loose, overly flexible joints, stretchy easily bruised skin, abnormal wound healing and scar formation. Kyphoscoliotic (type VI as per older classification) type of EDS is very rare autosomal dominant defect due to deficiency of an enzyme called Lysyl hydroxylase. Four major diagnostic criteria for type VI EDS are loose hyperflexible joints, low muscle tone, scoliosis at birth (worsens with age), and a fragility of eyes which may give blue sclera or globe rupture. Because of undue fragility of the corneal-scleral tissue and abnormal wound healing, the surgical prognosis is usually unfavourable. We report two cases of EDS kyphoscoliotic type who presented with spontaneous perforation of anterior staphyloma with

endophthalmitis and posterior globe rupture with supra-choroidals with retinal detachment in one-eyed patient.

Case 1

A 21-year-old male, was referred with gradual progressive bulging of the left eye since two months and pain, redness and diminution of vision since 7 days. Visual acuity in RE was perception of light with defective projection and in the LE 6/24 and 6/18 (with -0.75D spherical). Patient had bilateral ptosis. Slit lamp examination showed conjunctival and ciliary congestion and mucopurulent discharge. In the inferotemporal quadrant there was anterior scleral staphyloma (13mm x 5mm) with two areas of extreme scleral thinning with bluish uveal tissue visible through it. There was 1.5mm thick hypopyon with grade 2 flare and 4+ cells. Pupil was eccentric towards staphyloma and non-reacting. Lens showed early cataractous change. No glow was visible on indirect ophthalmoscopy. RE examination was unremarkable. Ultrasonography of the RE showed plenty of dot echoes and moderate reflective membranous echoes suggestive of endophthalmitis. Patient had thoracolumbar kyphoscoliosis, multiple areas of scarring over legs, elbow region and back. There was cutaneous hyperextensibility and joint laxity. There were no obvious septic foci, was afebrile and blood culture and routine lab investigations were unremarkable. In view of endophthalmitis urgent lensectomy and vitrectomy was done with intravitreal vancomycin, cefotaxim and dexamethasone. Iris hooks were placed for non-dilating pupil, PVD induction was done and near total vitrectomy was done along with removal of cyclitic membrane. Immediate postoperative period the size of staphyloma decreased, hypopyon had resolved. Hyphema, diffuse vitreous haemorrhage and significant hypotony was noted. Culture from vitreous tap revealed staphylococcus aureus sensitive to vancomycin, hence intra-vitreal vancomycin and dexamethasone were repeated. Severe hypotony

persisted till 8thPOD; hence 0.3cc of 100 percent C3F8 was injected through pars plana rout. Systemic steroids of 1mg/kg body weight were started with tapering dosages with intravenous cefotaxim 1gm twice daily for 5 days. Progressive thinning of staphyloma with exposure of uveal tissue was noted with persistent hypotony. staphylectomy with homologous processed scleral patch graft was done to save the globe on 9th post operative day. Sclera patch graft was processed form a donor globe after removing the corneal button and complete stripping of the uveal tissue. The sclera thus harvested was processed in 100 percent alcohol. After repositioning the uveal tissue, graft of 13x 6mm was then sutured with 7-o-vicryl on the scleralside and 10-o nylon monofilament interrupted sutures on the corneal side. The graft was then covered with undermined conjunctiva. Intraocular pressure gradually builds up and fundus glow became healthy. Graft exposure was noted after 17 days and revision of scleral corneal and conjunctival sutures was done. At the time of discharge the visual acuity was perception of hand movements close to face and IOP had build up. After a follow up of 2 months the graft was well accepted, globe stable (FIGURE) and central fundus could be visualized. The patient had best corrected vision of 6 meters snellen.

Case 2

A 37 year old male was referred to our OPD with the complaint of sudden painless diminution of vision in LE immediately following alleged assault with fist 5 days back. The patient had lost vision in the other eye 1 year back following suspected trivial trauma with finger. At presentation the visual acuity was no perception flight in RE and perception of light with inaccurate projection of rays in left eye. On ophthalmic examination RE was an enophthalmic, prethysical eye with a mature presenile cataract and LE had lid edema with ptosis, slit lamp evaluation revealed superior subconjunctival haemorrhage in fornix, clear cornea with 3mm hyphema with dispersed blood and cells, traumatic mydriasis and clear lens. On indirect ophthalmoscopy there was no glow in LE. Applanation pressures were 12mm in LE. B-scan USG of LE showed dense dot echoes suggestive of vitreous haemorrhage with 360 degree bullous suprachoroidals with attached retina. He had gross skin laxity (figure) with easily bruisability and multiple scars (figure) on the extensor aspect of hand, arms, elbow, knee and legs. Significant kyphoscoliosis (figure) was present. No obvious hearing abnormality was present. Parents had no similar abnormalities. One of the 6 siblings died in early childhood had similar deformities but rest had no such abnormalities. The patient was started on oral prednisolone 1.5 mg/ kg/ day tapering weekly, locally prednisolone acetate eye drop 10 times, homatropine eye drop thrice, timolol 0.5% twice and propped up position.

On 10 days review, visual acuity was perception of light with intact projection of rays and on slit lamp evaluation hyphema was present (altered blood) with grade 3 cells, clear lens. Red glow was present on retroillumination but no fundus details were visualized on indirect ophthalmoscopy. Repeat B-scan USG revealed multiple dot echoes (vitreous haemorrhage) reduced suprachoroidals with clot lysis and shallow RD. Surgical intervention was planned as clot lysis had occurred, the suprachoroidals had reduced and shallow retinal detachment had appeared. Radial sclerotomy around 11 mm from limbus in the inferotemporal quadrant was fashioned and the supra choroidal blood was drained followed by A/C wash. Phacoaspiration with vitrectomy, drainage retinotomy, fluid air exchange and endolaser barrage (around the site of incarceration and suspected break in the area of posterior scleral rupture in superonasal quadrant). inferior Iridectomy was done with 1200cs silicon oil infusion. Retinal folds were present in the macular area due to incarceration. Shallow residual suprachoroidals were present intraoperatively and immediate intraoperative period. At three months followup patient had attached retina with complete disappearance of suprachoroidals and best corrected visual acuity of 6/60. Silicon oil removal was done at 105 days. At one month post silicon oil removal patient was maintaining the vision with +9D spherical correction and retina was attached.

Discussion

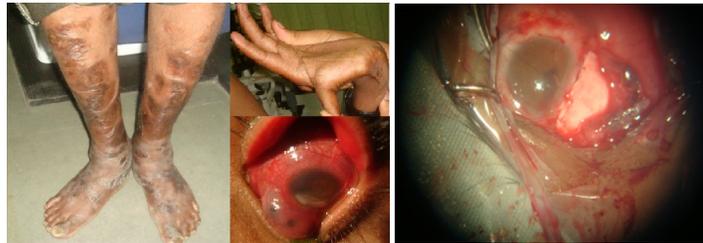
EDS is a rare disorder with subtype VI having ocular involvement. Although minor ophthalmological abnormalities are common, major ocular complications are infrequent. [1]. It is believed that EDS is usually transmitted as an autosomal dominant trait. Various researchers have reported other modes of inheritance like X-linked and recessive. Pedigree charting in the present case series was suggestive that the cases were sporadic. All the cases in our series had typical features of EDS, satisfying the four major criteria: thoracolumbar kyphoscoliosis, cutaneous hyperextensibility, articular laxity, and easy bruisability, hyperpigmented scars over elbows, shin and legs. Based on the typical clinical features these cases could be easily differentiated from Marfan syndrome and osteogenesis imperfecta. Reports of corneal translocation [2] and sclera transplant [3] Serious ocular complications were present in all the three cases with the 1st case having suprachoroidals with sclera rupture and retinal detachment and phthisical other eye; 2nd having ciliary staphyloma with perforation and endophthalmitis both following trivial trauma. Third case had blue sclera, high myopia and had been operated for retinal detachment surgery following which he developed secondary glaucoma due to emulsification of silicone oil.

one eye and sclera rupture with partial subconjunctival dislocation of lens in the other eye secondary to trauma. In all the cases the trauma was trivial which in normal subjects may not have resulted in such serious outcome. Vitreoretinal surgical intervention was done successfully in all the three cases and ambulatory vision could be restored. Hence ophthalmologist should be aware of these complications so that the patients could be warned to take adequate precautions as well as be prepared with the difficult management options. EDS patients may be predisposed to serious life threatening vascular

abnormalities and complications which should be kept in mind during anaesthesia.

References

1. Beighton P (1968) "The Characteristics of the Ehlers-Danlos Syndrome", p. 93-98. M.D. Thesis, University of London
2. Judish G F et al (1976) Ocular Ehlers Danlos Syndrome with normal LysylHydroxylase activity. Arch Ophthalmol 94:1489-1491
3. Nakazawa M et al (1986) Homograft of Preserved Sclera for Posttraumatic Scleral Staphyloma in Ehlers Danlos Graefe's Arch Clin Exp Ophthalmol, 24; 247-250.



Case 1 Photographs



Case 2 Photographs