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Stickler Syndrome Type 1 with Cornea Perforation: A Case Report

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Abstract: Ophthalmologic anomalies are the most characteristic and most serious manifestations of the Stickler syndrome. In the eye, abnormalities of vitreous gel architecture associated with high myopia and retinal detachment is described. There is a substantial risk of retinal detachment. Here in we report a rare association of Stickler Syndrome with megalocornea and lagophthalmos which lead to cornea perforation.
Keywords: Stickler Syndrome, vitroretinopathies, megalophthalmos, cornea perforation.

INTRODUCTION

Stickler Syndrome is a group of genetic disorders that is thought to arise from a mutation of several collagen genes (collagen types II, IX and XI) during fetal development. It is estimated to be in 1 in 7500 new-borns [1]. It is a sex independent autosomal dominant full trait with complete penetrance but with variable expressivity (even interfamilial). It is characterized by distinctive facial abnormalities, ocular problems, hearing impairment and joint problems [2]. It is a progressive disorder and likely to become more severe with age.

CASE REPORT

A 4 months old Malay baby girl with Stickler Syndrome was referred by Paediatrics Unit for bilateral eye buphthalmic eyes (Figure 1). She was born borderline premature at 35 weeks with birth weight of 2.25kg. Mother antenatally has uneventful pregnancy and the child was born via emergency lower section caesarian section due to breech in labour.

Echocardiography, brain ultrasound, abdominal ultrasound and hearing assessment were done to the child which revealed normal findings. The child was initially had poor weight gain due to poor suckling but after training and interventions by occupational and speech therapist, the child was able to suck well and started to have a good weight gain.

There was strong family history of ocular problem (Figure 2) with her elder brother also has Stickler syndrome with buphthalmic eyes and secondary glaucoma, her father is blind due to bilateral eye advanced glaucoma and her mother also has pthysical right eye with low vision of the left eye.

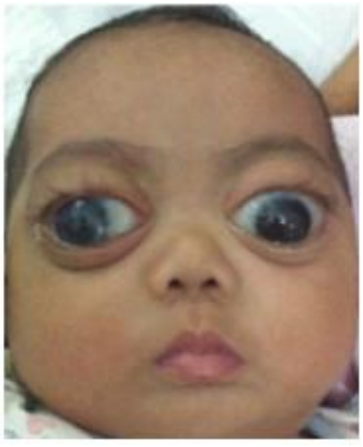

Fig- 1: Prominent bilateral buphthalmic eyes

Fig- 2 Strong family history of ocular problem

General examination shows that the child has flattened facial appearance with depressed nasal bridge, anteverted nares, micrognithia and low set ears. Oral examination shows that she has macroglossia, bifid uvula and cleft palate. The child also has hyper mobility and hyper flexibility of the joints and clubbed foot.

Ocular examination shows that the child has buphthalmic eyes with lagophthalmos bilaterally. Close examination shows that the child has right eye cornea perforation with iris plugging infernasally with cornea abscess and hypopyon. The left eye also has inferior cornea epithelial defect.

Examination under anesthesia (EUA) reveals the size of right eye cornea perforation was 8.0mm x 9.5mm. Right eye tectonic Penetrating Keratoplasty was done to the patient (Figure 3) on 19/2/2014, followed by bilateral eye tarsorraphy.

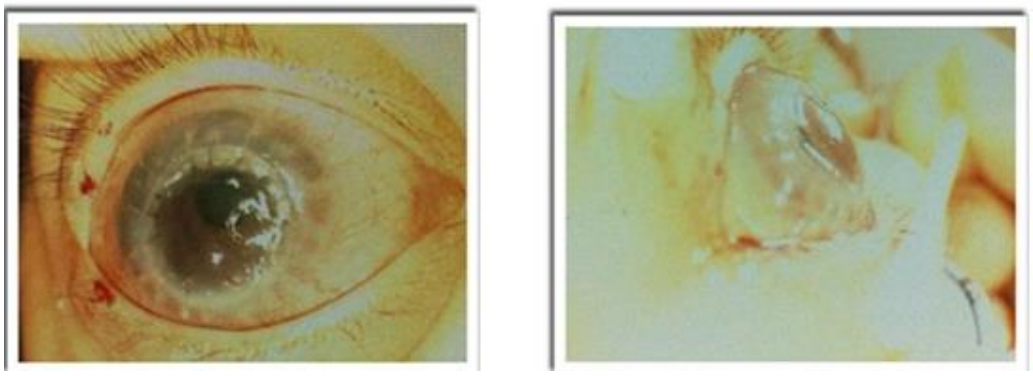

Fig-3: Penetrating Keratoplasty done for right eye cornea perforation

Subsequent EUA after 2 weeks showed the right eye cornea graft was clear with no evidence of infection and left eye epithelial defect was also completely healed. The cornea diameter was 15mm on the right with cornea thickness of 715um and intraocular pressure of 8mmHg. The left cornea diameter was 16mm on the left with central cornea thickness of 394um and intraocular pressure of

18mmHg. Left eye trabeculotomy was subsequently performed to the patient.

Latest EUA showed the right cornea graft remained clear with intraocular pressure of 11mmHg and the left eye cornea also clear with intraocular pressure of 8mmHg. Bilateral optic disc was pink with cup disc ratio of 0.2. Membranous vitrous was present in both eyes. Otherwise, retina is flat bilaterally with no evidence of radial perivascular retinal degeneration or lattice degeneration, and no retina tear or break seen. Refraction showed bilateral high myopia with -20.0 diopteric spherical power in both eyes.

DISCUSSIONS

Stickler syndrome is believed to be a direct result of abnormalities in the production of collagen types II, IX and XI. Normal collagen fibrils are composed of three identical (homotrimer) or differing (heterotrimer) polypeptide chains [3]; genetic mutations affecting the ability of constituent polypeptide chains to successfully form stable trimers therefore prevent the production of mature collagen and subsequently produce the clinical manifestations of Stickler syndrome. The condition is commonly associated with ophthalmologic manifestations including vitreous abnormalities [4], congenital megalophthalmos, radial perivascular retinal degeneration and retinal detachment, in addition to systemic findings which may include or facial, auditory and musculoskeletal abnormalities. It is a progressive disorder and likely to become more severe with age.

The condition has been divided into 4 subgroups based on ocular and systemic clinical findings, with each subgroup also corresponding to a specific genetic defect [5]. Type 1 Stickler Syndrome is due to mutation of COL2A1 gene and characterized by membranous vitrous, congenital megalophthalmos, deafness, arthropathy and cleft palate. Type 2 Stickler Syndrome is due to mutation of COL11A1 gene and characterized by beaded vitrous, and otherwise other findings are similar to Type 1. Type 3 Stickler Syndrome on the other hand is due to mutation of COL11A2 gene with clinical characteristic similar to Type 1 but with no ocular involvement , while Type 4 Stickler Syndrome occurs due to mutation of COL9A1 and COL9A1 gene, which ocular only manifestation with no systemic findings. Our patient is suffering from Type 1 Stickler Syndrome.

This case report highlights the presence of congenital megalophthalmos in Stickler syndrome Type 1 which leads to bilateral severe lagophthalmos and severe exposure keratopathy on the right eye which causing right eye cornea perforation, a rare manifestation of Stickler syndrome.

The congenital megalophthalmos, with narrowing of orbital cavity associated in this syndrome make the patient eyes prominent. On top of that, congenital abnormalities of the anterior chamber drainage angle which lead to secondary glaucoma in this patient worsen the pre-existing buphthalmos that the patient had and these further impaired the proper lid closure mechanism for this patient. The cornea, in particular, predisposed to drying effect due to insufficient lipid layer and evaporative effect of aqueous layer of the tear film. The dry cornea was also unhealthy and predisposed to infection and cornea ulceration. Hence, that explained why the child has right eye cornea perforation with iris plugging with cornea abscess and hypopyon and the left eye also had inferior cornea epithelial defect.

Management of this patient with right eye tectonic penetrating keratoplasty and bilateral tarsorraphy was aimed to preserve the globe integrity and to allow proper healing of the epithelial defect of the cornea. Subsequently, as from EUA the left cornea thickness was thin (394um) and with intraocular pressure of 18mmHg, left eye trabeculectomy was performed. This, in particular was to prevent further buphthalmic of the eye as this will lead to further risk of globe/ cornea perforation. The tarsorraphy was aimed to narrow the palpebral aperture to reduce the further risk of exposure keratopathy. Patient was still able to see from the opening part of the tarsorraphy for some process of visual stimulation and development.

As we are happy after the right eye cornea perforation sealed, with latest EUA shows bilateral cornea were clear and the intraocular pressure for both eyes within the desired range value, that is not the ending story of this child. The high refractive error (-20D for both eyes) for this child need optical correction to optimize visual stimulation and development. The defective collagen production in this syndrome with presence of membranous vitrous in this child might predispose her to rhegmatogenous retinal detachment, the most well-known ocular risk in Stickler syndrome. In a large study of the genetically confirmed type 1 subgroup, 60-80% had lifetime risk of retinal detachment [5]. That is the reason for subsequent regular EUA and proper surveillance is crucial for this patient to anticipate further complications and to prevent or limit total visual loss.

This case report highlights the occurrence of Stickler Syndrome related to a genetic abnormality [5] as there is a strong family history of ocular problem in this patient with her elder brother also has Stickler syndrome with buphthalmic eyes and secondary glaucoma, her father is blind due to bilateral eye advanced glaucoma and her mother also has pthysical

right eye with low vision of the left eye. Genetic counseling is important as this syndrome has important medical and personal consequences, for patients and their family. Managing ocular problem in Stickler Syndrome is challenging. Multi-discipline team approach – including Ophthalmology, Paediatrics, Genetics, Otorrhinology, Orthopedic Surgery, Oral Surgery are all important to get involved in managing patient with Stickler Syndrome to help them to get the best health and quality of life.

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