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Case Report

Childhood glaucoma in Down syndrome: a case report and literature review

Puspha Raman*, Visvaraja Subrayan

Department of Ophthalmology, Faculty of Medicine, University of Malaya, Kuala Lumpur 50603, Malaysia

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***Correspondence:**
Dr. Pushpa Raman,
E-mail: puspha@gmail.com

ABSTRACT

Children with down syndrome have a higher prevalence of glaucoma compared to the age matched normal population. Although the management of childhood glaucoma in a down syndrome child is almost similar to primary congenital glaucoma, the outcomes and prognosis of these children vary due to the associated comorbidities. This case illustrates a classical presentation of a 5-month infant with down syndrome with clinical triad of photophobia, blepharospasm, and epiphora. Prompt recognition of the condition and timely surgical management was vision saving in this child. Extended care for visual rehabilitation was given to this child to maintain the visual outcome.

Keywords: Congenital glaucoma, Down syndrome, Goniotomy, Trabeculotomy

INTRODUCTION

Down syndrome (triplicate of all or part of chromosome 21 [HSA21]; OMIM 190685) is a common cause of mental retardation, with a prevalence in western societies ranging from 0.9 per 1000 live births.[1] The reported incidence of down syndrome in one of the largest government hospital in Malaysia was 1:959. The highest incidence was reported among the Malaysians 1:981, followed by the Chinese 1:940 and Indians 1:860.[2]

Children with down syndrome have a higher prevalence of ocular disease compared to the age matched normal population. The most common ocular associations are strabismus, refractive errors and impaired accommodation. An increased prevalence of congenital cataracts, childhood glaucoma, keratoconus and nystagmus also noted in this group of children.[3]

According to Childhood Glaucoma Research Network/World Glaucoma Association, glaucoma in down syndrome children is classified under childhood glaucoma with nonacquired systemic disease. Although the management is almost similar to primary congenital glaucoma, the outcomes and prognosis of these children vary due to the associated comorbidities.

This case illustrates a classical presentation of a down syndrome child with the clinical triad of photophobia, blepharospasm, and epiphora, where prompt recognition and management was vision saving.

CASE REPORT

A 5 months old Indian baby girl was brought to University Malaya Ophthalmology Department with bilateral eye clouding and tearing. According to the mother the eyes were clear at birth and she started to notice the haziness at 3rd month which became progressively worse. Mother noticed that the child getting increasingly fretful in bright light. The child was having persistent tearing in the past one month. Otherwise the parents were unaware of any redness or increasing size of the eyes. The child was clinically suspected to have Down's syndrome (awaiting chromosomal study) and was under Pediatrics Unit's regular follow up. She had a

small Patent Ductus Arteriosus (PDA), which was managed conservatively. Otherwise, the child was feeding well and gaining adequate weight.

The child was born via spontaneous vaginal delivery at 38 weeks of gestation. The antenatal and perinatal history was unremarkable. Maternal age was 30 years old. No history of birth trauma. No family history of glaucoma, blindness or chromosomal diseases.

This is the first child for the parents and they are both working as professionals in a multinational company. The paternal grandmother is a retired staff nurse who is the primary caretaker after the parents left to work.

On general examination, the child has distinct facial dysmorphism in line with the diagnosis of Down syndrome; mongoloid facies, flat nasal bridge, prominent epicanthal fold with a protruding tongue. The child also has other typical features like single transverse palmar crease and hypotonia.

On ocular examination, the child was able to fix on a bright light but not interested to follow the light. The child had bilateral buphthalmos with epiphora. The sclera which was only visible at the temporal side had a bluish appearance. Bilateral cornea was hazy with descemet striae. The globe was firm on palpation. IOP which was taken with Icare device was 44 mm Hg in right eye and 37 mm Hg in the left eye. A clinical diagnosis of infantile glaucoma was made. The child was planned for Examination Under Anaesthesia (EUA) and to proceed with glaucoma surgery on the same settings. While awaiting surgery, the child was started on topical dorzolamide TDS and topical latanoprost on night.

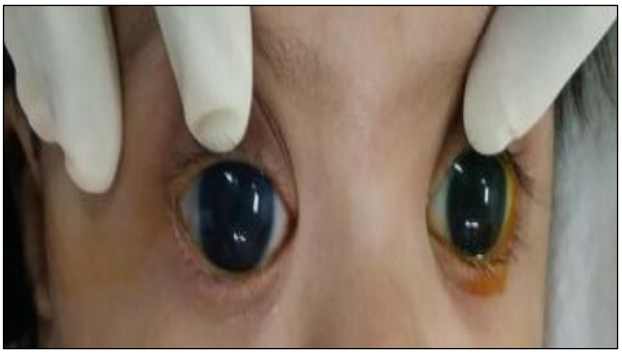

Figure 1: External photograph prior to EUA. Left eye noted to have increased tear meniscus.

The first step in ensuing the success of the treatment is to get the parents understand the implication of the current diagnosis and get them committed with the management plan. Thus, we provided counselling to the parents regarding their utmost important role in ensuring the child getting timely eye drops, attending the frequent follow up visits and also prepare for repeated surgeries if adequate control is not achieved. The parents were very apprehensive but were able to appreciate the wealth of the information and ready to commit to the betterment of the child's health. We planned a multidisciplinary approach for the surgical planning in view of her underlying comorbidities. She was referred to pediatrics team and pediatrics anesthesia clinic to be assessed for fitness for general anesthesia prior to the planned date.

On the day of procedure, the anesthetist was able to intubate the child without complication. Examination under anesthesia was performed. Objective refraction was performed with retinoscopy. The child was -1.00 DS (low myope) bilaterally with no obvious astigmatism. Axial length was 22 mm bilaterally. Bilateral cornea was hazy with moderate corneal edema. Haab's Striae was evident on retro illumination (Figure 1 and 2).

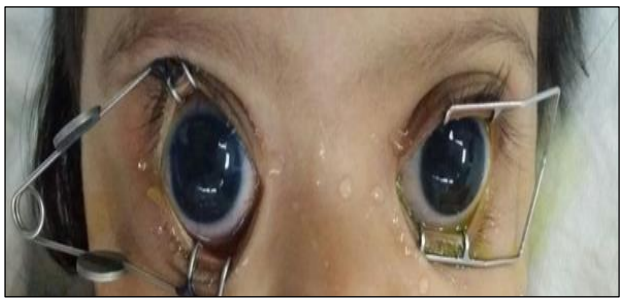

Figure 2: External photograph during the EUA. Note the increased corneal diameter with right corneal haziness

Horizontal corneal diameter was 13 mm on the right eye and 12 mm on the left eye. No other anterior segment anomalies such as aniridia, iridocorneal adhesion or correctopia were noted. Tonometry was performed with Perkins tonometer. IOP was 40 mm Hg and 27 mm Hg for Right and Left Eye respectively. Gonioscopy was performed with Swan Jacob lens. The anterior chamber was deep with high iris insertion. No other angle anomaly was noted. Optic nerve and fundus evaluation was done with indirect ophthalmoscopy. Bilateral optic disc showed increased cupping with the CDR of 0.6 and 0.5. Otherwise fundus was normal with healthy macula.

After the clinical assessment, the child was planned for a combined Trabeculotomy and Trabeculectomy for the right eye and Trabeculotomy alone on the left eye. The surgeon created a fornix based conjunctival flap and hemostasis was secured with cautery. A partial thickness scleral flap was dissected into about 1 mm of clear cornea. A radial incision was then carried out across the inner trabeculectomy site about 2 mm from the limbus, until the Schlemm's canal was entered; evidenced by a gush of aqueous humor. Trabeculotomy ab externo was then performed using the internal arm of trabeculotome probe, first to the left and then to the right to complete about 120 degrees of the circumference. The pre-marked 2×2 mm inner block tissue comprising trabecular meshwork and scleral spur was excised with vannas scissor and peripheral iridectomy was performed. The

partial thickness scleral flap was sutured with interrupted 10-0 Nylon. The conjunctival flap was closed with 8-0 vicryl, continuous stitches. Trabeculotomy ab externo was performed in the left in a similar manner. Subconjunctival dexamethasone injection given and the eye was padded with an overlying eye shield.

The antiglaucoma medications were withheld postoperatively. She was started on antibiotic and steroid eye drops for both eyes. On post-operative day 1, bilateral cornea has cleared and IOP was 15 mm Hg and 17 mm Hg . The child was further seen at 2 weeks and 1 month. She made an uneventful recovery and there was no evidence of infection or bleb related complications.

At 1-month review, bilateral cornea was clear. The IOP was 13 mm Hg and 18 mm Hg. The left eye IOP control was unsatisfactory over the next few visits, fluctuating between 18-24 mm Hg. A decision for goniotomy surgery for left eye was made after discussing with parents.

She underwent her second surgery under anesthesia uneventfully. The angle was visualized with Swan Jacob lens. A needle knife is passed across the anterior chamber and a superficial incision made in the uveal trabecular meshwork, at the Barkan Membrane. Goniotomy was done over nasal area of 180°.

The child made an uneventful recovery. She was followed up for every 2 months. The IOP on follow up was 13 mm Hg and 14 mm Hg without any antiglaucoma medication. She had clear cornea with visible Haab's Striae in the right eye. She had bilateral horizontal nystagmus. No strabismus or abnormal head posture was noted. The CDR was 0.4 bilaterally on the 18th month review. Her cycloplegic refraction revealed +1.00 dioptre bilaterally without any evident of increasing axial myopia.

DISCUSSION

Pediatric glaucoma is a relatively rare, potentially blinding condition caused by elevated intraocular pressure. The goal of preserving a lifetime of vision for these children involves early, prompt control of IOP to a level where progression is unlikely along with correction of ametropia and rigorous amblyopia treatment. Pediatric glaucoma is classified as primary when an isolated idiopathic developmental abnormality of the anterior chamber angle exists and secondary when aqueous outflow is reduced due to either a congenital or an acquired ocular disease or systemic disorder.[4]

The above case illustrates a typical presentation of a Down syndrome child with childhood glaucoma and the timely surgical intervention which was able to halt the progression of the disease.

Down syndrome (Trisomy 13) is one of the recognized syndromes that is associated with childhood glaucoma. In most recent studies conducted in Asia, the prevalence of glaucoma in Down syndrome ranges from 0.4% to 1% of cases, although in one series in Malaysian Down syndrome children, the prevalence was reported to be 6.7%.[5-7] The commonest ocular associations of Down syndrome that are reported in the Malaysian studies are include epicanthic fold (96.7%), nystagmus (33.3%) and strabismus (26.7%). Other findings were bilateral congenital cataract (13.3%) blepharoconjunctivitis (10.0%), eyelid abnormalities (6.7%) nasolacrimal duct obstruction (3.3%) bilateral retinoblastoma (1.7%), bilateral retinal detachment (1.8%) and chronic uveitis (1.7%).[7] Baby T in our case had prominent epicanthic fold and nystagmus. It is interesting to note that iris anomalies such Brushfield spots which are commonly seen in Western Down syndrome children are not noted reported in Asian studies. Our child also did not have any brushfield spots.

At this point it is imperative to highlight the role of primary care providers and pediatricians to screen ocular associations in Down syndrome children which are potentially blinding such as cataract and glaucoma.

This child presented with the classical triad of epiphora, blepharospasm and photophobia. The epiphora of congenital glaucoma is often misattributed to congenital nasolacrimal duct obstruction, which is found in 5-6% of newborns. The hallmark of congenital glaucoma is ocular enlargement, which occurs because the immature and growing collagen that constitutes the cornea and sclera in the young eyes responds to the increasing intraocular pressure by stretching.[8] Clinically, ocular enlargement is most evident as an increase in corneal diameter. Enlargement of the globe, often referred to as buphthalmos, meaning "ox eye". In general, horizontal corneal diameter in the normal neonate is 10 to 10.5 mm and increases from 0.5-1.0 mm during the first year of life.[8] Our child had the corneal diameter of 13 mm and 12 mm for right and left eye respectively, which indicates the rapid progression of the disease in the first few months of life. Corneal edema and haze is a common sign of the disease, as are horizontal or circumferential breaks in Descemet's membrane termed Haab's striae. Haab's striae will remain visible on examination throughout the patient's life, even if the edema resolves with normalization of intraocular pressure. Haab's striae was visible on Baby T's cornea at 18th month visit.

Medical therapy usually provides a supportive role to reduce the IOP temporarily, to clear the cornea, and to facilitate surgical intervention. Laser therapy has a limited role in childhood glaucomas. The most effective and definitive form of treatment of most childhood glaucomas is surgical. Primary surgical treatment is usually with goniotomy or trabeculotomy. Combined trabeculotomy with trabeculectomy may be useful in certain patients who are predicted to have a high risk of

failure with goniotomy or trabeculotomy alone. Refractory pediatric glaucomas may be managed by trabeculectomy with anti-fibrosis drugs, glaucoma drainage implants and cyclodestructive procedures. Most infants with primary congenital glaucoma who present between 3 months and 1 year of age will have their IOP controlled by one or two angle surgeries. If IOP control is not achieved with the first surgery, at least one other angle surgery is attempted before trying another strategy.[9]

In the present case, goniotomy was not the primary choice because of the corneal haziness. The surgeon decided on combined trabeculectomy and trabeculotomy for the worse eye and trabeculotomy alone for the fellow eye. However, many authors do not consider trabeculectomy as a first-line procedure in congenital glaucoma in view of a higher incidence of complications and lower success rate. Problems encountered in performing trabeculectomy in children are thick tenon, thin sclera, difficulty in identifying the limbus and an exuberant healing response. Nevertheless, several reports have documented successful results following primary trabeculectomy for congenital glaucoma, which are comparable to goniotomy or trabeculotomy.[10]

Congenital glaucoma is a clinical challenge for any ophthalmologist, and requires a long-term commitment toward these patients with extended care for rehabilitating these children. A study in Nigeria identified that presentation with features of advanced disease, poor acceptability of surgery, poor follow-up and low surgical rate are problems associated with the management of congenital glaucoma.[11]

CONCLUSION

In conclusion, the main goal in managing childhood glaucoma is early diagnosis and therefore early surgical treatment to normalize IOP, in order to allow possible reversal of cupping and thus to minimize the impact on vision.

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