Congenital Glaucoma: A Case Series

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ABSTRACT

Primary Congenital Glaucoma is an inherited condition which occurs during development of eye. There is defect of trabecular meshwork as well as anterior chamber angle of eye, with consequent high intra ocular pressure of eye. Buphthalmos (Greek for ox eye) was coined in 4th century BC by Hippocrates, this name suggests large eyeballs arising from chronic IOP elevation. Primary congenital glaucoma (PCG) is most common non-syndromic glaucoma occurring in infancy, which can lead to blindness or can offer a lifetime of vision if diagnosed and treated properly. Congenital glaucoma presents with classic trio of symptoms i.e. photophobia, epiphora, blepharospasm and typical signs comprising of corneal clouding, megalocornea, and buphthalmos.[1] Delayed diagnosis, limited treatment and inadequate follow up leads to a heavier burden on the person and community in developing world.[2] Most of the primary congenital glaucoma(PCS) cases are sporadic, while 10-40% familial association, Autosomal recessive with incomplete or variable penetrance pattern and infrequently autosomal dominant. PCG is more common in populations with a larger prevalence of consanguinity and shows variable expressivity and phenotypes in association with CYP1B1 *gene* mutations. Definitive treatment is early surgical intervention.[3]
The purpose of this study is to report three cases of congenital glaucoma presenting to a tertiary care hospital. First case is a neonate with incidental finding of buphthalmos which was then investigated, second case is about a month-old infant with congenital glaucoma and the third case deals with the detailed examination, treatment given for the same.

Keywords: *Blue eye, Buphthalmos, Congenital glaucoma, Intra-ocular pressure, Primary Congenital glaucoma*

INTRODUCTION

Primary Congenital Glaucoma is a inherited condition which occurs during development of eye. There is defect of trabecular meshwork as well as anterior chamber angle of eye, with consequent high intra ocular pressure of eye. Congenital glaucoma presents with classic trio of symptoms i.e. photophobia, epiphora, blepharospasm and typical signs comprising of corneal clouding, megalocornea, and buphthalmos.[1] PCG is commonly Autosomal recessive condition, hence common in consanguinity shows CYP1B1 *gene* mutations. Early surgical intervention is the mainstay treatment.[3] Based upon the age of onset of disease, the primary congenital glaucoma is divided into following types.[4]

1. True congenital glaucoma – known as newborn glaucoma. Here presentation of ocular enlargement or enlargement of eyes at birth itself or within one month of life. Some studies suggest that raised IOP is present in the intrauterine life itself in approximately 25% cases.
2. Infantile glaucoma – This occurs between 1 and 36 months of life. 65% of

cases with primary congenital glaucoma are present with it.

3. Juvenile glaucoma – When raised intraocular pressure is seen after three years of age but before adulthood. It is seen in 10% of cases.

Hoskin classification is a classification system based on the area of dysgenesis as follows:[5]

1. Trabeculodysgenesis – Defect in development of the trabecular meshwork
2. Iridotrabeculodysgenesis – comprises of stromal hypoplasia or hyperplasia, anomalies of iris vessels, or structural defects like coloboma or aniridia.
3. Corneotrabeculodysgenesis – It includes complex conditions or syndromes of congenital glaucoma like Axenfield, Rieger, or Peters anomaly.

Hoskin's classification type 1 is termed as primary congenital glaucoma, whereas types 2 and 3 are termed as secondary congenital glaucoma.[6]

CASE SERIES

CASE 1

1-day old female full-term baby born to a G2P1L1 mother via LSCS indicated due to previous LSCS with birth weight of 2.6kg and did not cry after birth and was referred in view of Birth Asphyxia. The mother had non consanguineous marriage with 2 pregnancies out of which live births were 2. She was on calcium and iron supplements at the time of gestation. No history of taking teratogenic drugs. No any illness to mother during pregnancy. She also had history of Polyhydramnios during the delivery. Birth weight was 2.6 kg & other anthropometry of child at birth was normal. On head-to-toe examination, patient had increased corneal size and corneal haze was noted.

Baby had birth asphyxia for which baby was admitted in NICU, taken on CPAP mode initially and then taken on O2 nasal prongs after 10 days then tapered onto O2 nasal prongs. Baby was transfused with 2 pints of PCV in view of anaemia. Then started with OGT feeds, gradually increased and reached to full feeds. Baby was then shifted to mother side and tolerated oral feeds well, passing urine- stool well. Baby was clinically stable and discharged from our side. Baby was appertained to higher centre for congenital glaucoma treatment.

Fig.1: Megalocornea, Buphthalmos

CASE 2-

A 1-month-old boy presented with enlarged eyes and an intermittent right exotropia, without tearing or photophobia. Examination also suggested high myopia and an optic nerve cup-to-disc ratio larger in the right than the left eye. Referral to a paediatric ophthalmologist was initiated. On

the first examination under anaesthesia (EUA), the child was diagnosed with bilateral megalocornea with a normal IOP. He had other definitive signs and symptoms which led to a diagnosis of primary congenital glaucoma based on the new appearance of Haab's striae, further enlargement of the cornea, and an elevated IOP.[7] At this point, medical management was rendered at higher centre.

Fig 2: Bilateral Megalocornea

CASE 3-

3-month-old female came for evaluation of increased size of bilateral eyes.

The patient was first noted to have tearing, and was treated with antibiotics. At 1 month of age, the parents noted slightly increased size of the eyes, OS. When examined at age of 3months, eye size became obvious and corneal haze was noted clearly. No previous ocular history. No eye surgery nor eye trauma. Medical history is unremarkable. Family and Social History is non-contributory. Ocular examination shows General: well-appearing child in no acute distress. Visual Acuity: OD-- Fix/follow; OS--Fix/follow, Intra-ocular pressure (Perkins): 11 mmHg, OD; 33 mmHg, OS, External and anterior segment examination, Normal; OS: Corneal stromal haze and oedema.

Fig.3: Bilateral congenital Glaucoma

Gonioscopy: OD, Normal; OS, high iris insertion, Dilated fundus exam (DFE): OD--Normal; OS--Raised cupping. Horizontal corneal diameter: OD, 10 mm; OS, 12 mm. Axial eye length (echography): OD, 20.8 mm; OS 24.4 mm. This case had raised intraocular pressure, corneal oedema, increased horizontal corneal diameter, also increased axial eye length that point towards the diagnosis of buphthalmos which is suggestive of primary congenital glaucoma. Typically, the onset of primary congenital glaucoma is around 3-6 months of age, here it is at 1 month of age. There is no evidence of any angle recession, Peter's anomaly, or aniridia posterior embryotoxon /iris hypoplasia /corectopia/iris strands, which confirms this diagnosis. The patient was surgically managed with surgery of trabeculotomy. After the surgical procedure, the intraocular pressure in the affected eye returned to normal (15 mm Hg). No further surgery was required and no IOP-lowering drops were required to control the intraocular pressure thereafter. [OD-right eye, OS-left eye][8]

DISCUSSION

Structures which make the drainage angle are derived from neural crest as well as mesoderm lineage. Trabecular meshwork formation usually occurs around 12–22 weeks of gestation, along with mesenchymal cells resulting in a wedge-shaped structure between the corneal endothelium and the deeper stroma.[9] Schlemm canal is visualised first at 16 weeks of gestation, which is formed from a venous plexus anterior to trabecular angle, becomes easily recognisable with intercanal links by 36 weeks.[10]

Maximum growth of human eye occurs in the first 5 years of life where axial length increases highest in starting first 4 years. High intraocular pressure increases the axial length and corneal diameter. Extreme softness as well as elasticity of the infantile eyeball which can lead to lengthening of axial diameter leading to axial myopia and increasing corneal diameter might cause thinning of cornea that causes breaks in Descemet's membrane. These breaks in Descemet's membrane are called Haab's striae which is a classical finding in buphthalmic cases. A decrease in the number of endothelial cells in the cornea is also seen. Severe congenital glaucoma is associated with corneal haze, and studies also show a correlation between corneal haze and other factors which determine the severity of the disease, increased intraocular pressure, CD ratio, and corneal diameter.

Congenital glaucoma is diagnosed through a full eye examination that is done under anaesthesia in operating theatre in the case of babies and children under three years of age. The examination consists of:

- **Examination of front part of the eye:** to view and assess condition of cornea, its angle, to determine the most appropriate surgical procedure for each case.
- **Examination of the fundus:** After dilating the pupils with eye drops, the ophthalmologist views through a special magnifying lens to examine the retina and optic nerve and identify any signs of damage. Glaucoma leads to a progressive loss of nerve fibres from the optic nerve, forming a gap (excavation) which increases in size as the disease progresses.
- **Tonometry:** for measuring IOP, the ophthalmologist applies some numbing drops in the eyes, then places an instrument on the surface of eye. Normal ocular pressure ranges between 11-20 mm of mercury.

Prognosis of vision usually depends upon the severity of disease at diagnosis and the response to intervention with successful monitoring of IOP on follow-up. Often, patients with earlier onset of clinical manifestation (i.e., less than 1 month of age) have serious disease that responds poorly to angle surgery [11]. It is recommended to promptly proceed to more effective surgery

such as tube drainage surgery in these children.

- **Medical Treatment-** Medically cases are treated with topical beta-blockers, carbonic anhydrase inhibitors, or prostaglandin analogues.
- **Surgical Treatment- Goniotomy:** In this procedure, openings are created in the trabecular meshwork, thereby decreasing resistance to outflow. Trabeculotomy: The trabecular meshwork is incised by cannulating Schlemm's canal with the use of a probe. Trabeculectomy: A part of trabecular meshwork and Schlemm's canal is removed underneath a partial thickness scleral flap, thereby resulting in a fistula draining aqueous to the subconjunctival space, Combined trabeculotomy and trabeculectomy: Involves removal of a block of sclera after performing trabeculectomy, Glaucoma drainage implants, Cyclodestructive procedures [11].

There are some conditions which can cause an abnormal appearance of the eye that can be mistaken for buphthalmos. A complete eye exam is required to rule out these other diseases: Aniridia, Coat's disease, Dysplasia of retina, Endophthalmitis, Inflammatory cyclitic membrane, Neurofibromatosis type 1, Persistent hyperplastic primary vitreous, Retinoblastoma, Sturge weber syndrome, Toxocariasis. The immature angle appearance of PCG results from arrested development of tissues of neural crest origin in third trimester of gestation.[12]

CONCLUSION

1. In conclusion, Congenital glaucoma presents with classic trio of symptoms i.e., photophobia, epiphora, blepharospasm. There is also presence of typical signs like corneal clouding, megalocornea, buphthalmos. The immature angle appearance of PCG results from arrested development of tissues of neural crest origin in the third trimester, with the severity of abnormality varying.
2. PCG is commonly Autosomal recessive condition, hence common in consanguinity shows CYP1B1 *gene* mutations.
3. Early surgical intervention is the mainstay treatment. PCG is most common non-syndromic glaucoma in infancy period of life.
4. Prognosis of vision usually depends upon the severity of disease at diagnosis and the response to intervention with successful control of IOP on follow-up.

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