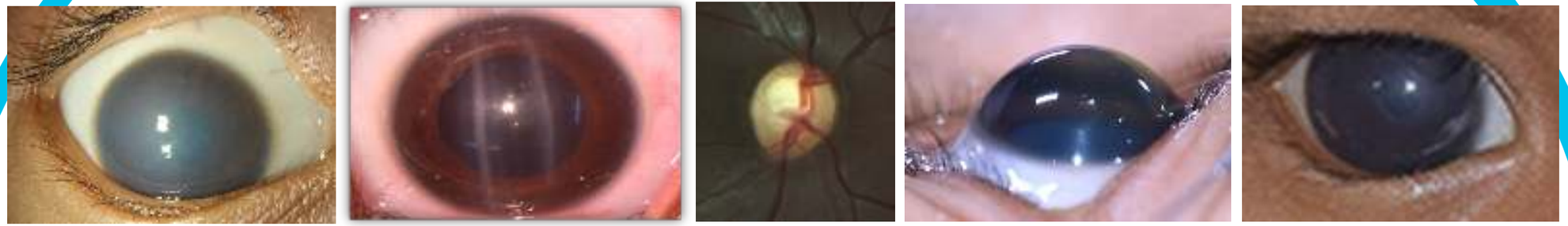
LVPEI
So that all may see

L V Prasad Eye Institute



Is this Congenital Glaucoma?

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L V Prasad Eye Institute
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India

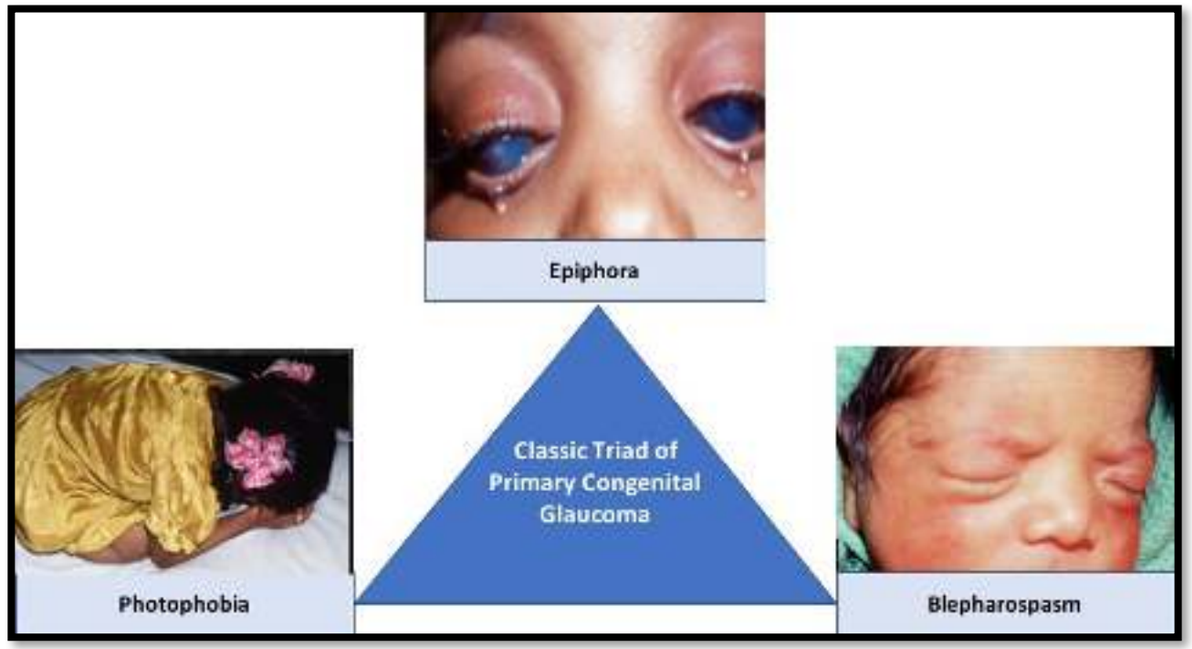
No Financial Disclosures

Images and cases: L V Prasad Eye Institute, Hyderabad
Have consent to use them

Outline

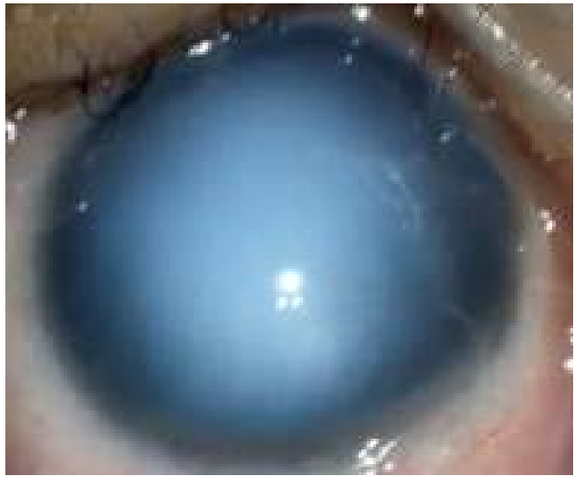
- Classical features of PCG
- Conditions that mimic based on the signs and symptoms
- Clinical scenarios of common mimickers

SIGNS AND SYMPTOMS OF CONGENITAL GLAUCOMA



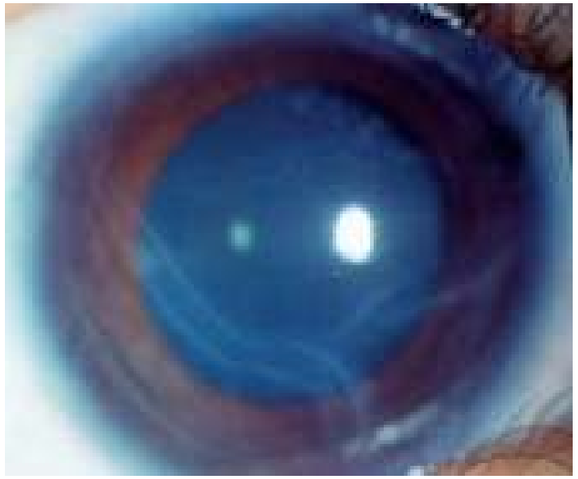
BUPHTHALMOS

MIMICKERS OF PCG



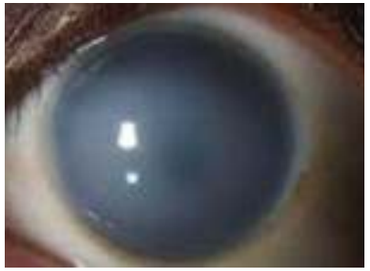
Corneal Haze

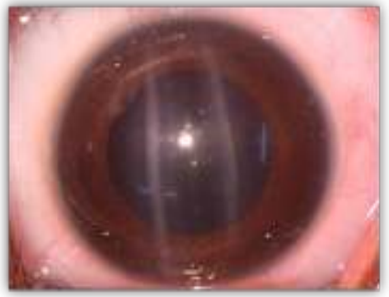
- Sclerocornea
- Trauma
- Ulcer
- Mucopolysaccharidosis
- CHED
- Dermoid



Descemet's membrane tears

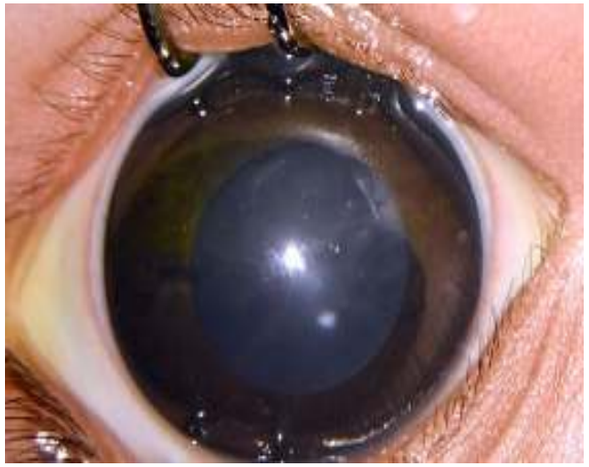
- Birth trauma
- Posterior Polymorphous corneal Dystrophy (PPMD)





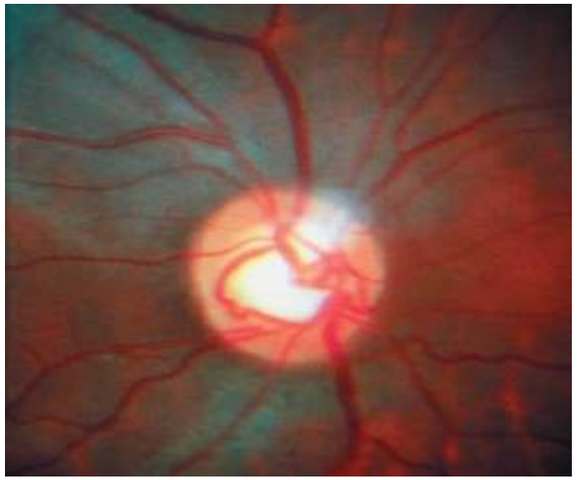
Khan AO. Ophthalmic genetics. 2011

Alobaidy R, Case Reports 2014.



Buphthalmos

- Congenital Megalocornea
- Keratoglobus
- High Myopia

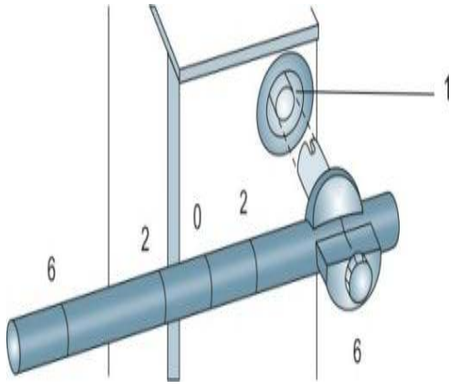


Optic disc cupping

- Hypoxic Ischemic Encephalopathy
- Physiological Cupping

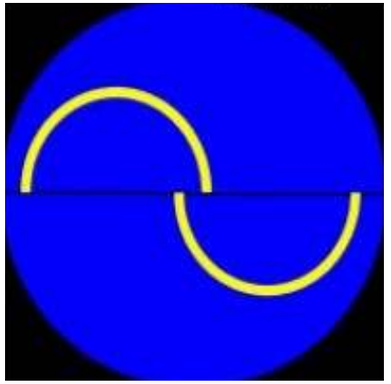
Khan AO. Ophthalmic genetics. 2011

Increase in IOP



Faulty Calibration

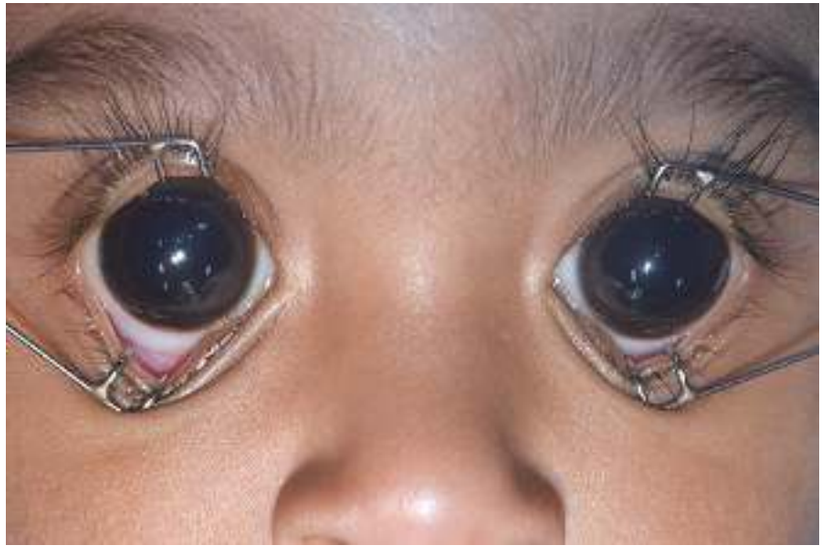
Unable to cooperate



Improper Technique

Picture courtesy:NIH

1



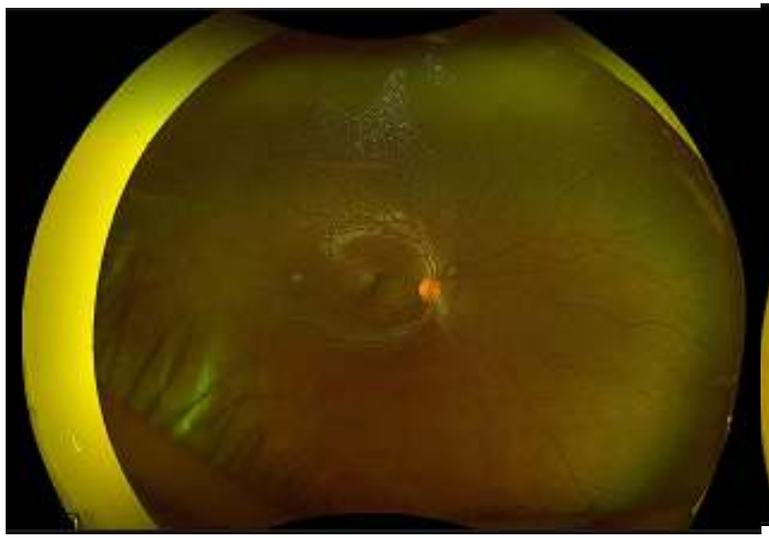
- Increase in the black portion of the eyes since birth
- No other complaints
- Similar complaints in the father and maternal grandfather

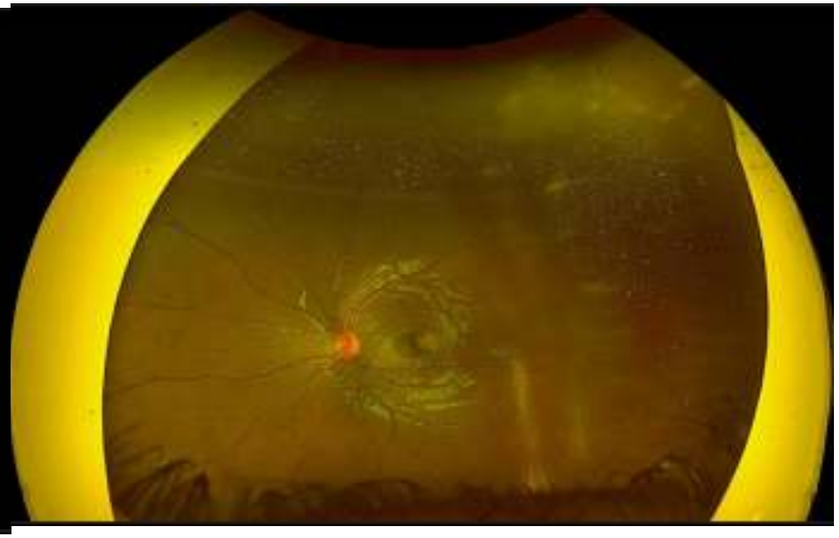
	Right Eye	Left Eye
Corneal diameter	15 mm	14.5 mm
K1	42.5	45
K2	49.25	48
Axial length	19 mm	18.5 mm
CCT	320 microns	404 microns
IOP	02 mm Hg	02 mm Hg
	0.2	0.2

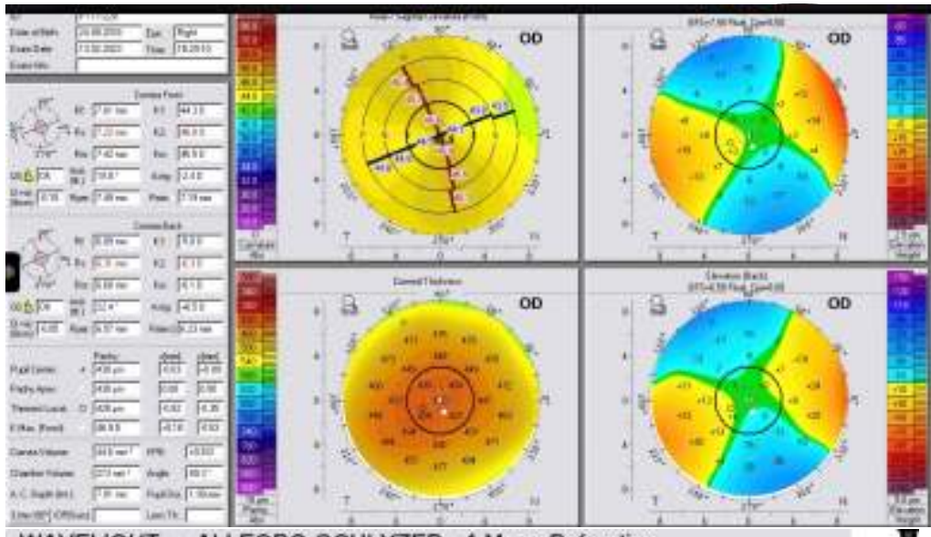
VARIANTS RELEVANT TO INDICATION FOR TESTING

One likely pathogenic variant in the *CHRDL1* gene was identified in this individual. No other variants of relevance to the indication were identified. Please see below for more detailed variant information.

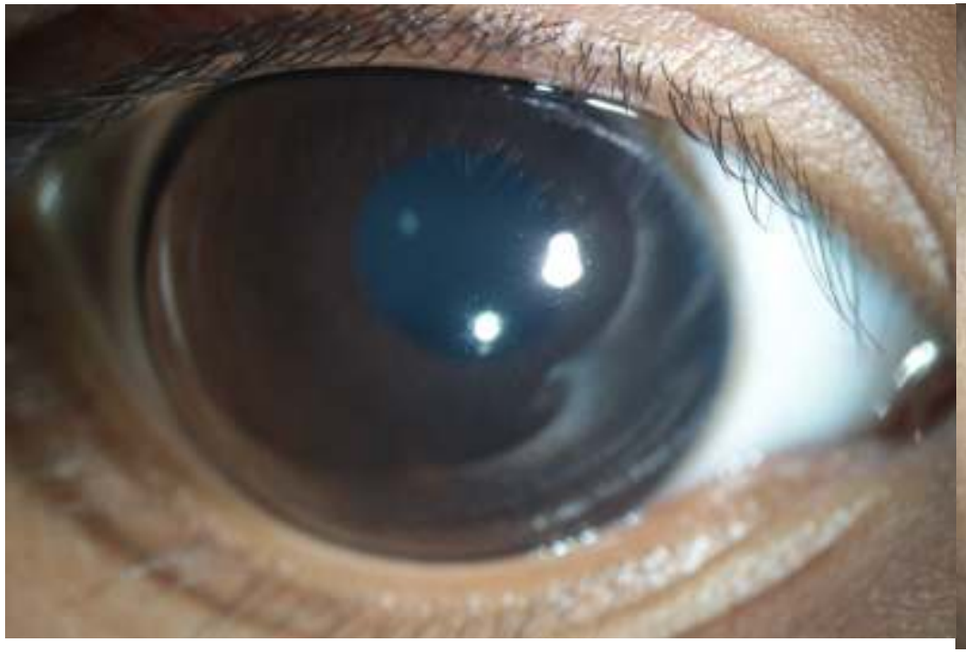
Gene & Transcript	Variant	Zygosity	Location	Disorder	Inheritance	ACMG Classification
CHRDL1 NM_001143981.2	c.94+2dupT p.?	Hemizygous	Intron 2	Megalocornea 1, X-linked [OMIM ID: 309300]	X-linked Recessive	Likely pathogenic PM2 & PP3_Strong

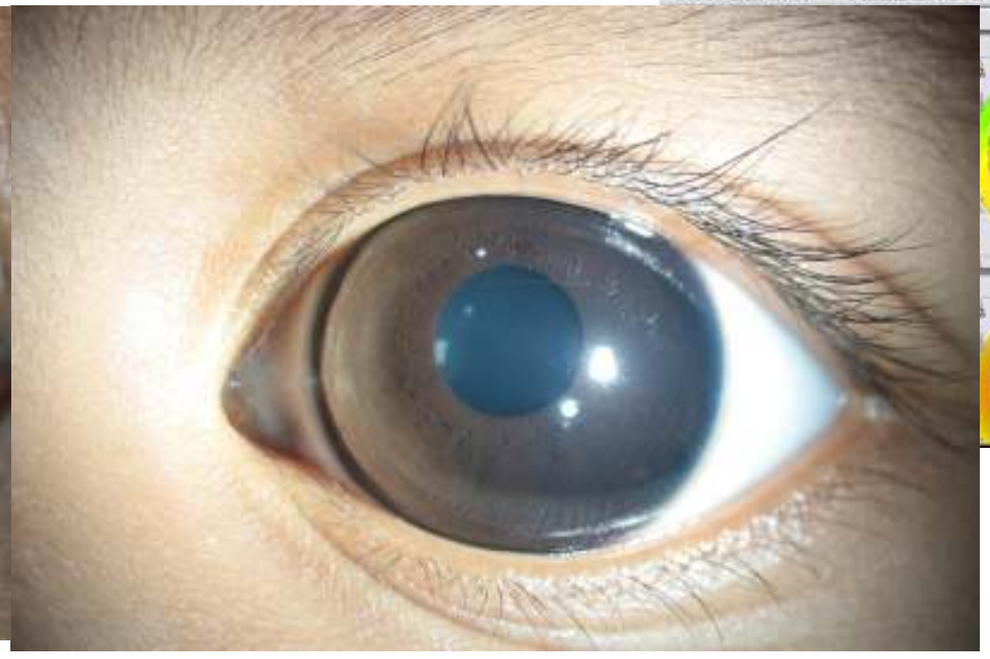


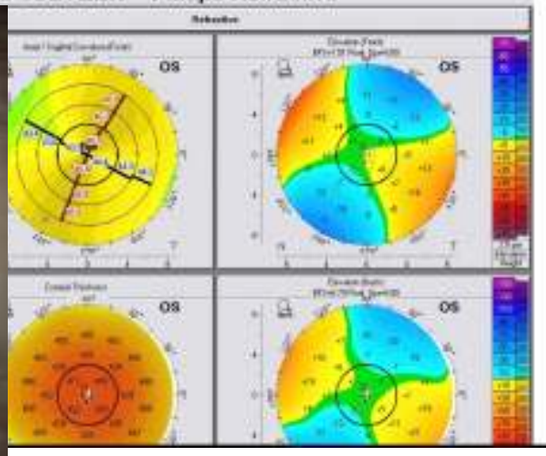




WAVELIGHT - ALLEGRO OCULYZER 4 Maps Refractive





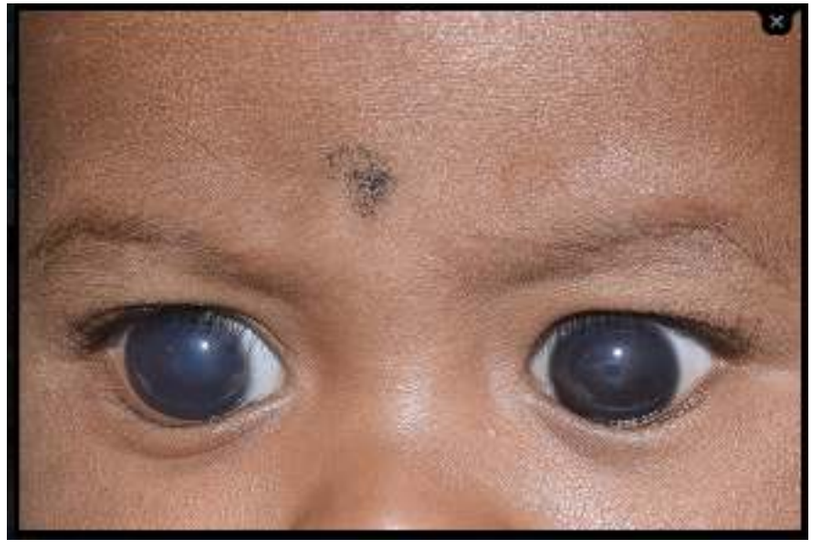


	Congenital Megalocornea	Primary Infantile Glaucoma
Inheritance	X-linked recessive	Autosomal Recessive
Natural History	Non-progressive	Progressive
IOP	Normal	Elevated
Corneal Exam	Enlarged, shagreen	Haab striae; corneal edema; decreased endothelial cells
Corneal diameter	>13 mm; symmetric	Variable; asymmetric
Axial length	Normal	Increased axial length
Optic disc	Normal	Cupping

Follow up every 4-6 monthly

2

- 4-month-old, enlargement of both eyes, photophobia and epiphora since 1-month
- 2nd degree consanguineous marriage
- RE Buphthalmos with corneal haze
- Phocodonesis and iridodonesis



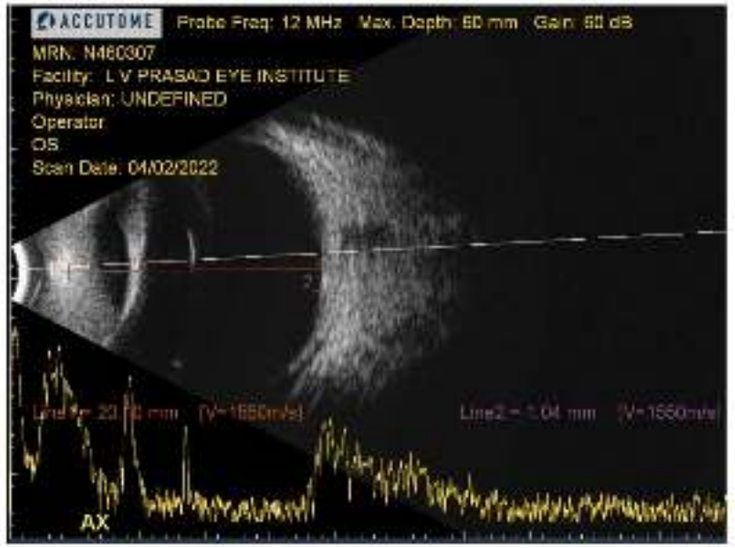
HCD-13.5 mm

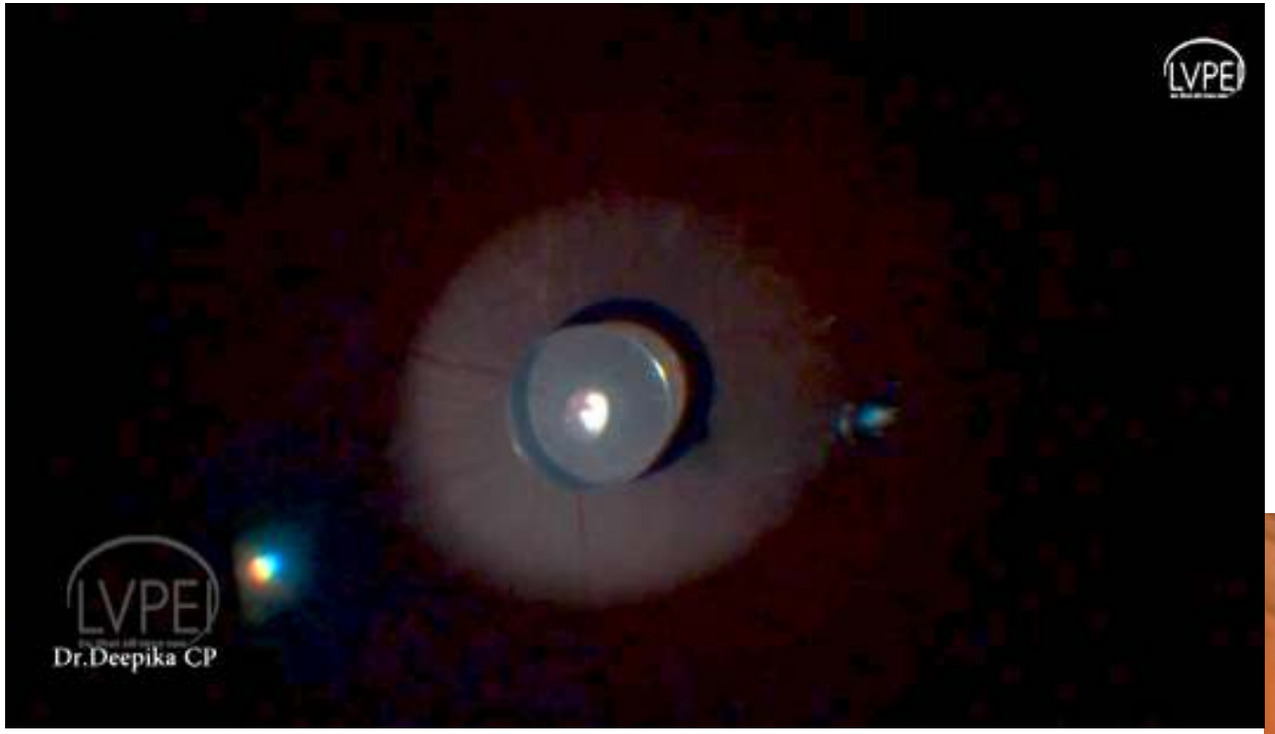
IOP was 02 mmHg in BE under anesthesia

Corneal edema, grossly subluxated lens in BE

BE CDR 0.2

Retinoscopy: -16, -14 D



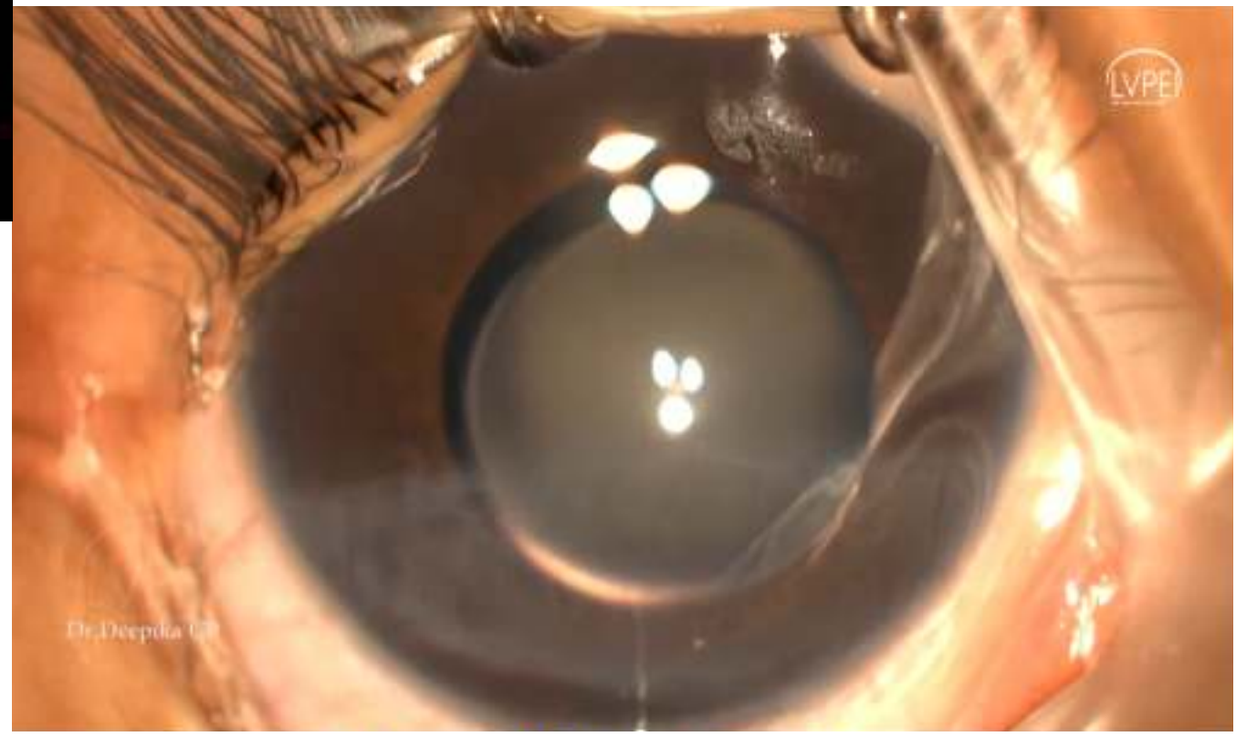


Results:

LP (Likely Pathogenic)	Likely Pathogenic variant has been identified related to the provided phenotype *(ref Table 1)*.

Table 1 : Likely Pathogenic Variant

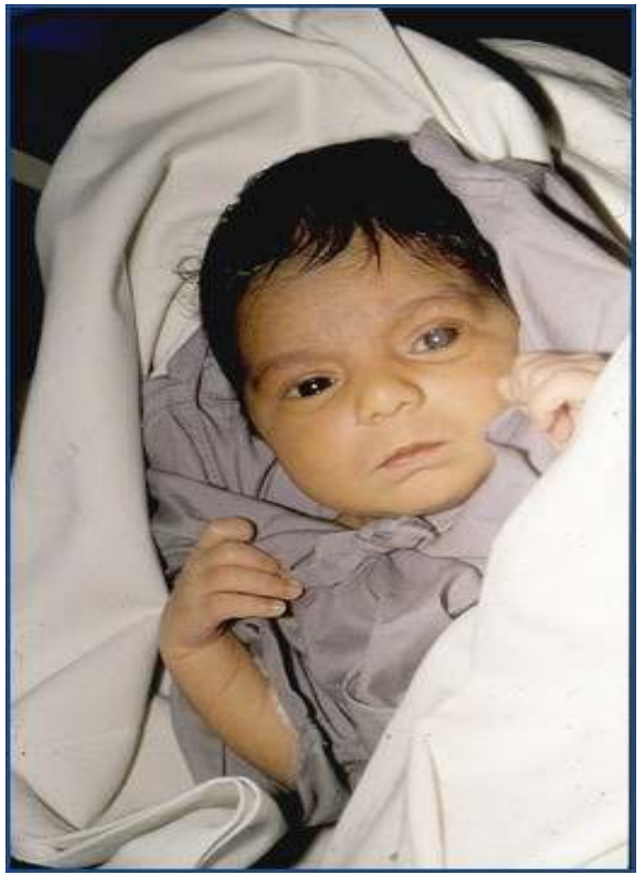
Gene	Genomic Position	Variant change	Zygosity/ Inheritance	Seq. Depth	Variant type	Exon	Associated Disorders
LTBP2	Chr14: 74975560	c.C3499T/ p.Gln1167Ter	Homozygous/ Autosomal Recessive	53x Ref=0 Alt=53	stopgain	23	Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma



- Post PPV+PPL
- IOP 3, 10 mm Hg
- 0.2 CDR
- +13.0 D
- >2 year FU, IOP still normal

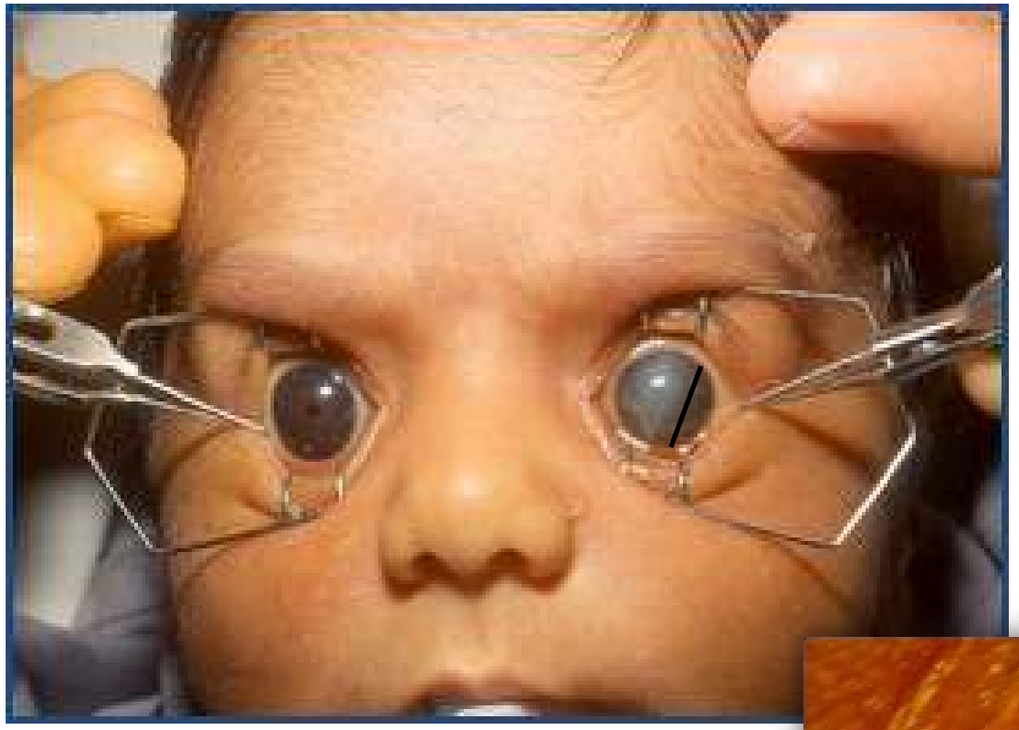
Intraoperative image: Curtsey Dr.Deepika CP, LVPEI

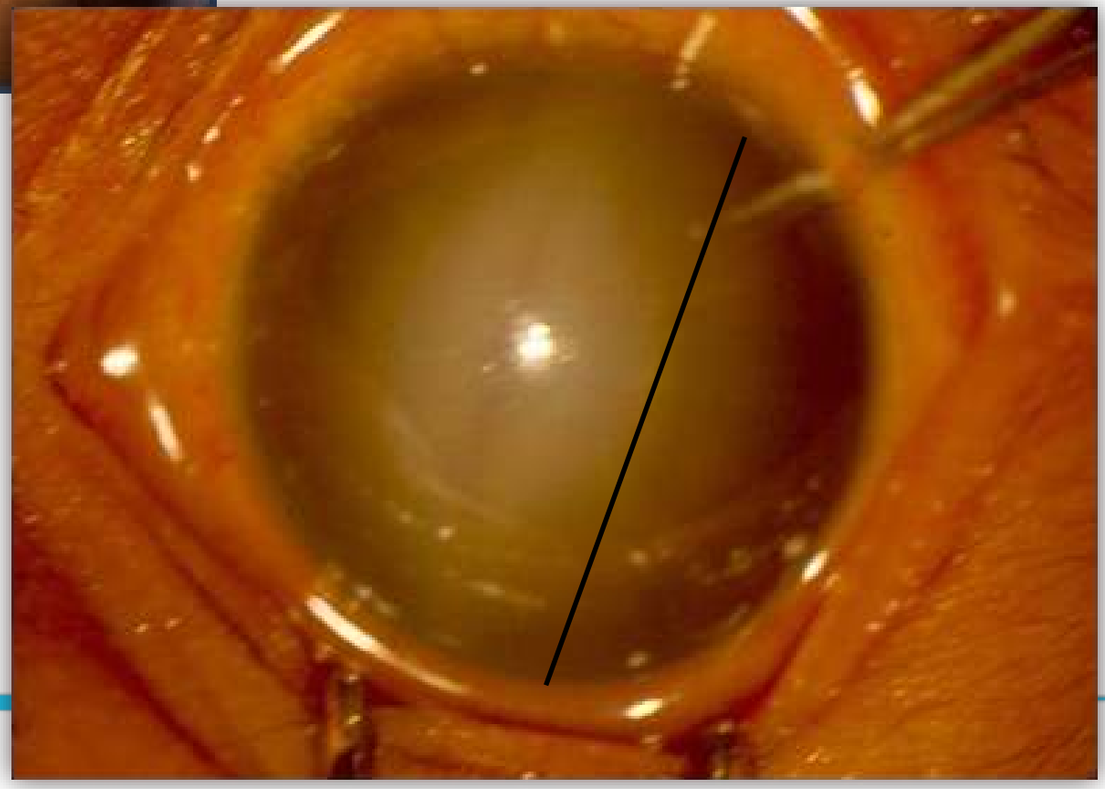
3



- 3-day-old boy
- Clouding of the left eye since birth
- No other symptoms
- No significant family history

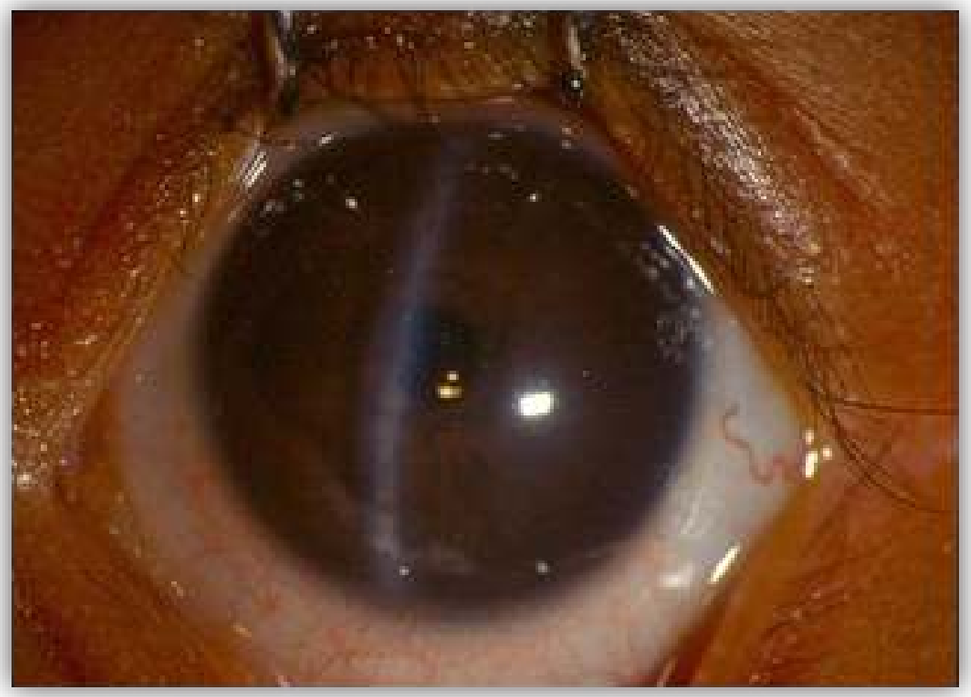
BIRTH TRAUMA

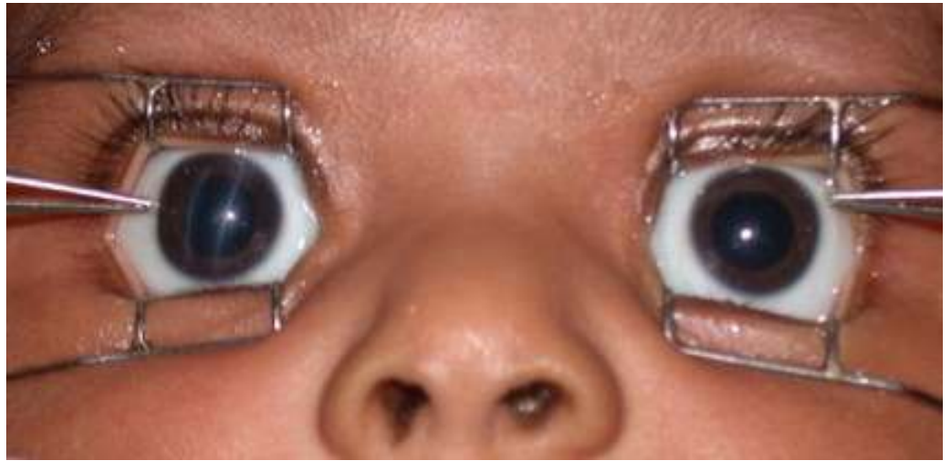


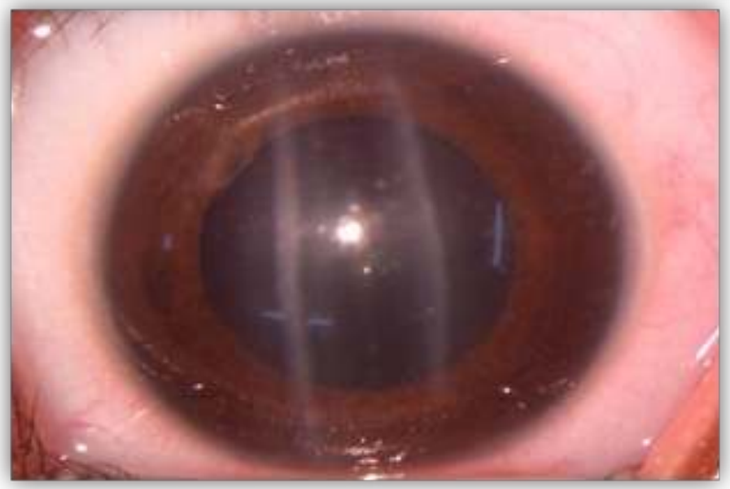


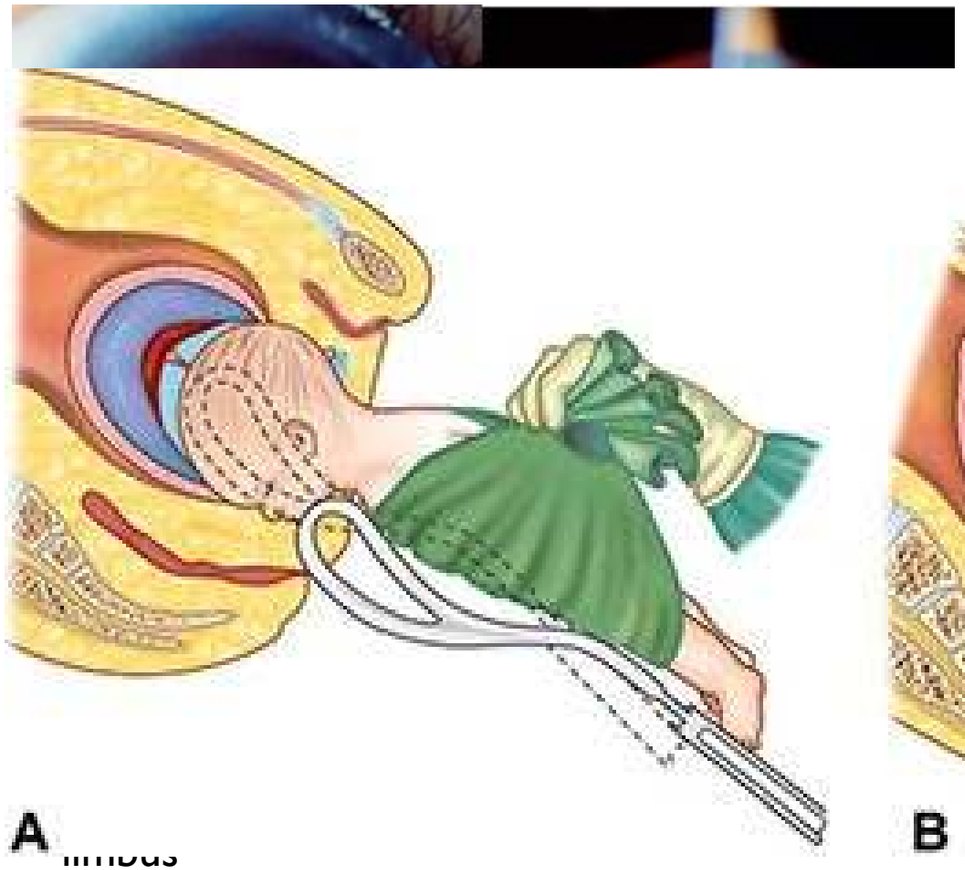
Delivery by forceps

Observe



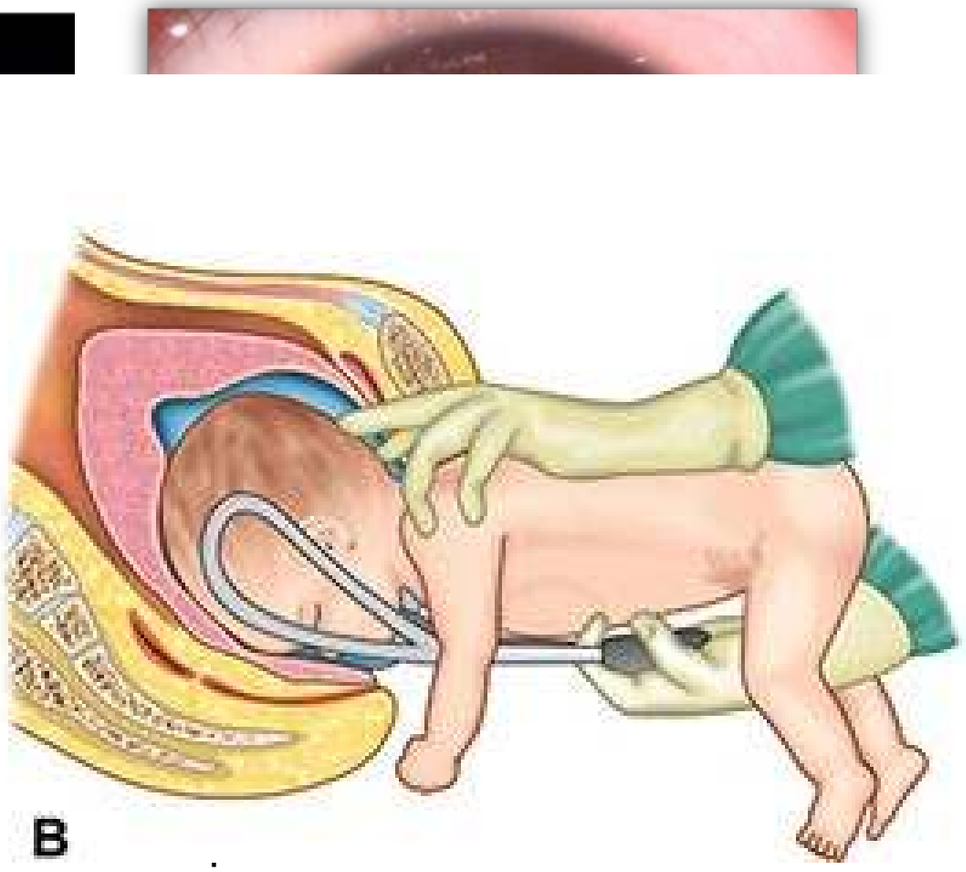






A

- Uncommon in corneas < 12.5 mm



B

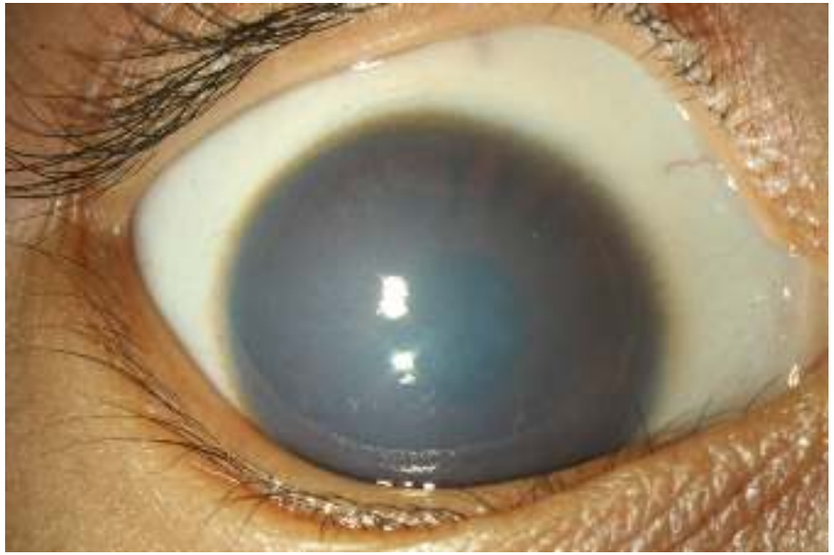
- The blade slips over the inferior orbital rim and compresses the globe vertically

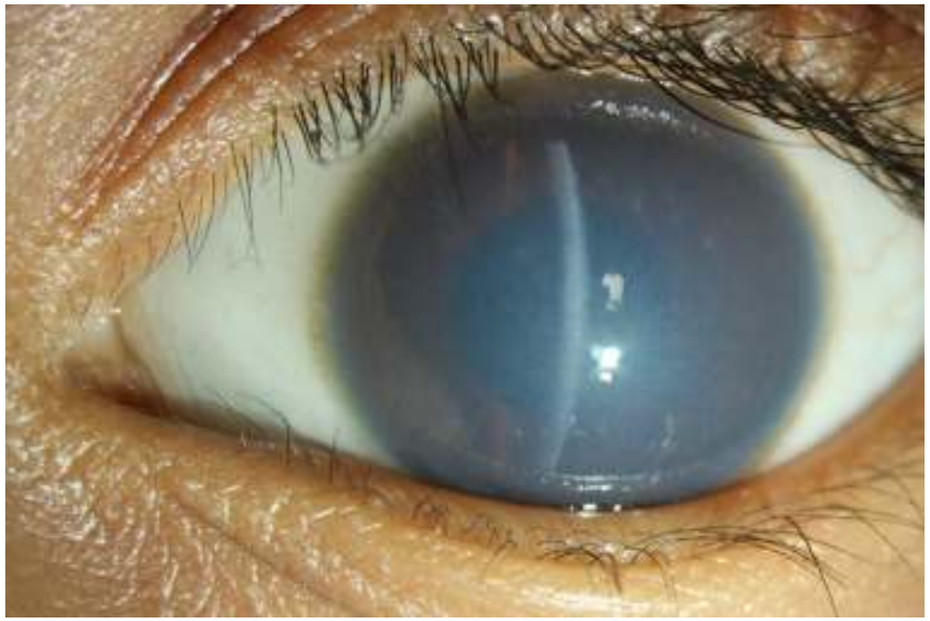
Krachmer JH,et al.. Cornea. 3rd edn Mosby; 2010

4

9-year-old girl

- Clouding of the black portion of the eye since birth
- Inability to see clearly
- Consanguineous marriage of parents
- Elder sister had a similar problem





BE 20/800, nystagmus

Diffuse corneal haze in BE

Corneal diameter 12/11.5mm, H/V

CCT > 1000 microns

IOP: 23 mm Hg in RE, 25 mm Hg in LE

BE Fundus- disc seen hazily- WNL

B scan AXL in BE - 22 mm

CONGENITAL HEREDITARY ENDOTHELIAL DYSTROPHY

	CHED	PCG
Gene	SLC4A11	CYP1B1
HCD Appearance Haab striae	Normal Edema Absent	>12/13mm edema or clear Present
Corneal thickness	Very thick - upto 1mm	Increased (with edema), not too high
Axial length	Normal	Increased
IOP	Normal or High	Often High
Disc	If visible Healthy	Disc changes
Gonio	Normal	Abnormal – anterior iris insertion

CHED

- Autosomal recessive corneal disorder specific for biallelic SLC4A11 mutations
- Characterized by a diffuse mosaic haze and a corneal thickness that can be two or three times normal
- IOP may be high - look for other features of PCG
- RARELY May coexist with glaucoma

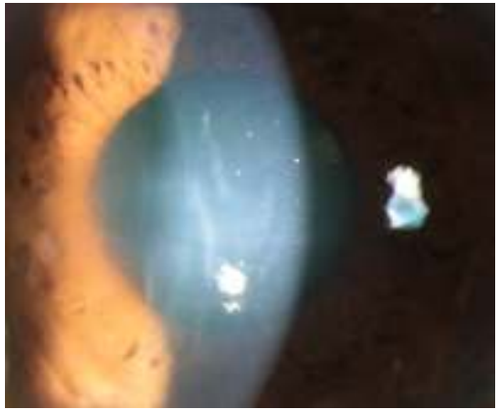
Khan AO. Ophthalmic genetics. 2011
Khan AO et al. Journal of AAPOS. 2016
Ramamurthy B et al. Cornea. 2007

5

6-months-old boy

- BE whitish discoloration of the black portion since 1 day
- H/o watering in BE since 5 days
- No significant family history
- Maternal and birth history - nothing significant

	Right Eye	Left Eye
Lids	Normal	Normal
Conjunctiva	Normal	Normal
Cornea	**Inferior corneal edema**	**Diffuse corneal edema with DM folds**
Corneal diameter (HCD)	**11mm**	**11mm**
Anterior chamber	Deep	Deep
Iris	Normal color and pattern	Normal color and pattern
Pupil	Round, reacting to light	Round, reacting to light
IOP by Perkins	**16 mm Hg**	**10 mm Hg**
Lens	Clear	Clear
Fundus	CDR 0.3, HNRR	No view – corneal edema



Representative photo

History of exposure to the sap of the Calotropis tree

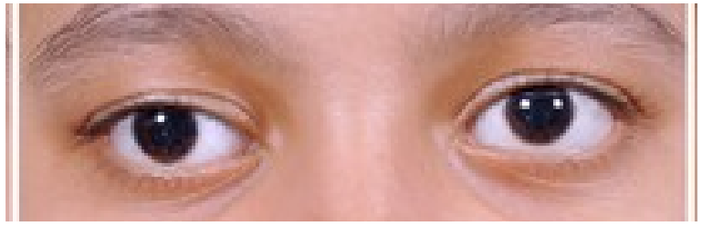
Waikar S, Srivastava VK. Calotropis induced ocular toxicity. Medical Journal, Armed Forces India. 2015 Jan;71(1):92.

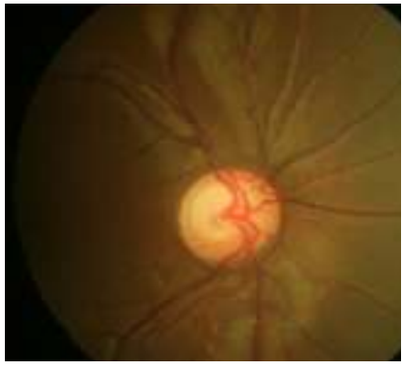
TOXIC KERATOPATHY

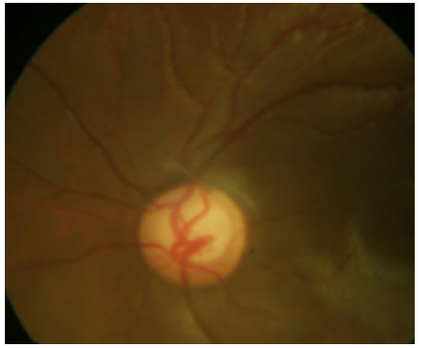
	Toxic keratopathy	PCG
History	History of exposure to chemical	Photophobia, tearing
Laterality	Unilateral/bilateral	Usually bilateral
Cornea Diameter Appearance Haab striae	Normal Edema Absent	>13mm Edema or clear Usually horizontal
Axial length	Normal	Increased
IOP	Normal	High
Disc	Healthy	Disc changes

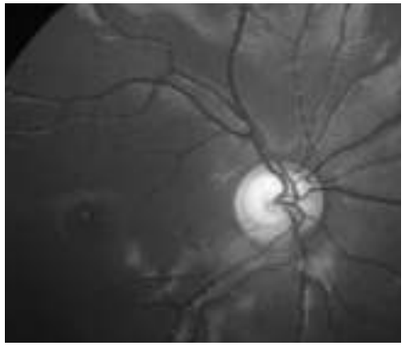
6

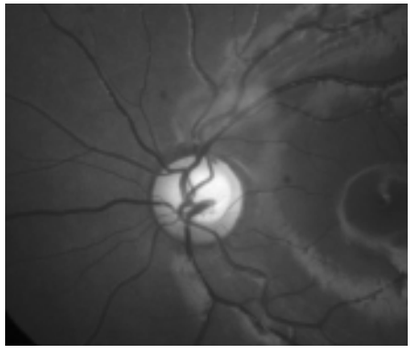
- 11-year-old
- BCVA: 20/159, 20/20
- IOP: 16, 15 mmHg
- CCT 513, 534 microns
- angles open
- She was Pre-term child with low birth weight, was incubated for 5 days, mild delay in attaining milestones
- No H/O consanguineous marriage

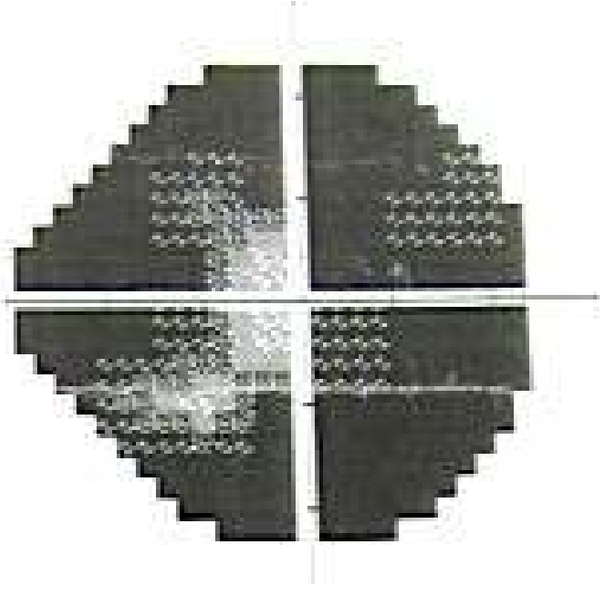


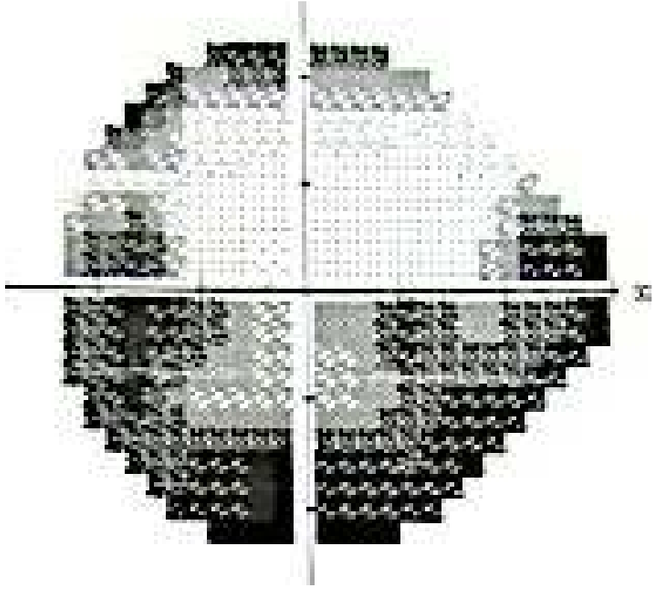




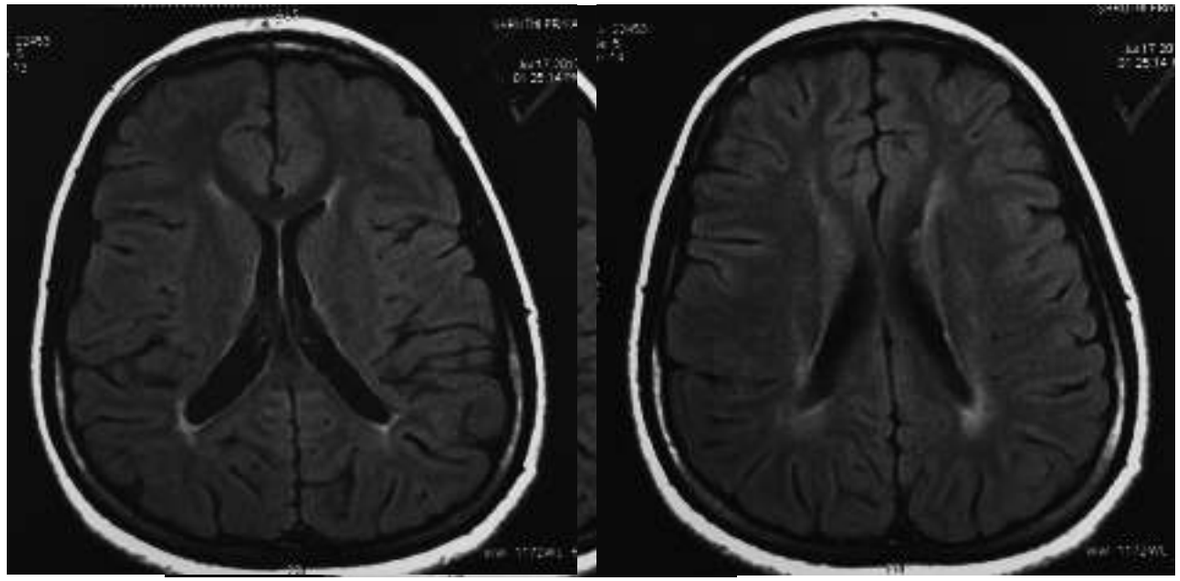




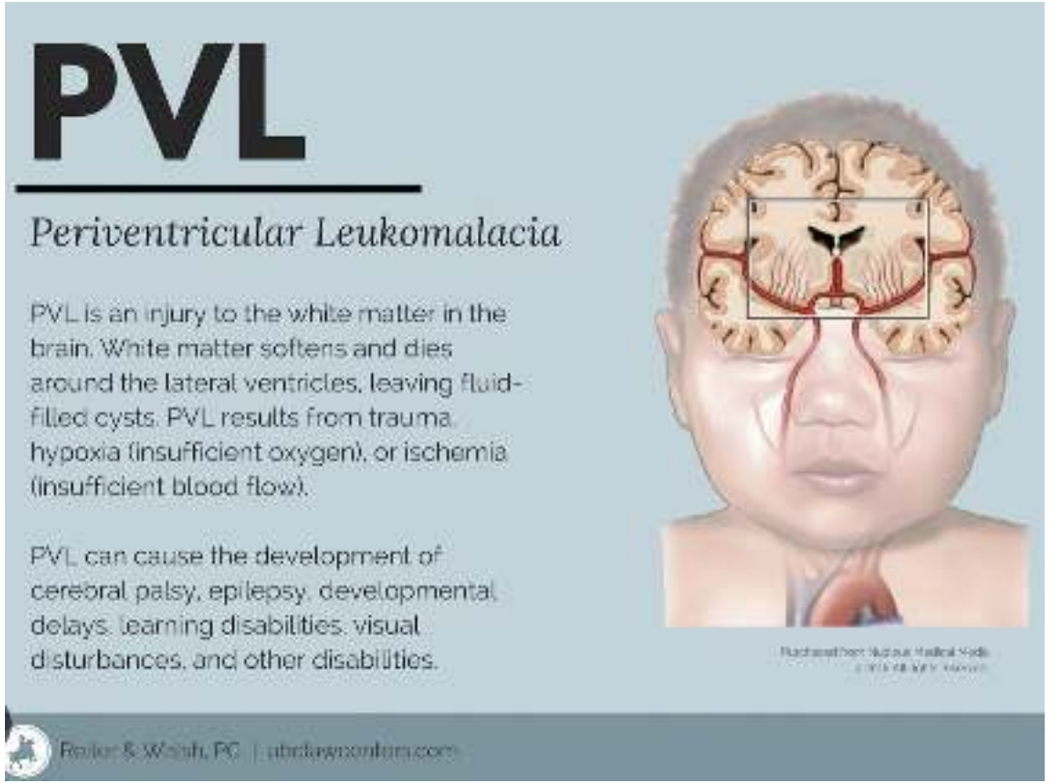




MRI: PERIVENTRICULAR LEUCOMALACIA



Patchy hyperintensity in the periventricular posterior parieto-occipital white matter



Diagnosis

- Hypoxic Ischemic Encephalopathy related optic neuropathy (HIE)
- RE Strabismic amblyopia
- HIE can mimic glaucoma both by disc and visual field changes
- HIE is an important DD of enlarged optic disc cupping in children preterm birth
- HIE is often misdiagnosed or treated as juvenile or congenital Glaucoma
- Neuroimaging is important to confirm diagnosis

Chhablani PP, IJO, 2014

Effect of hypoxia on visual pathway

HYPOXIA

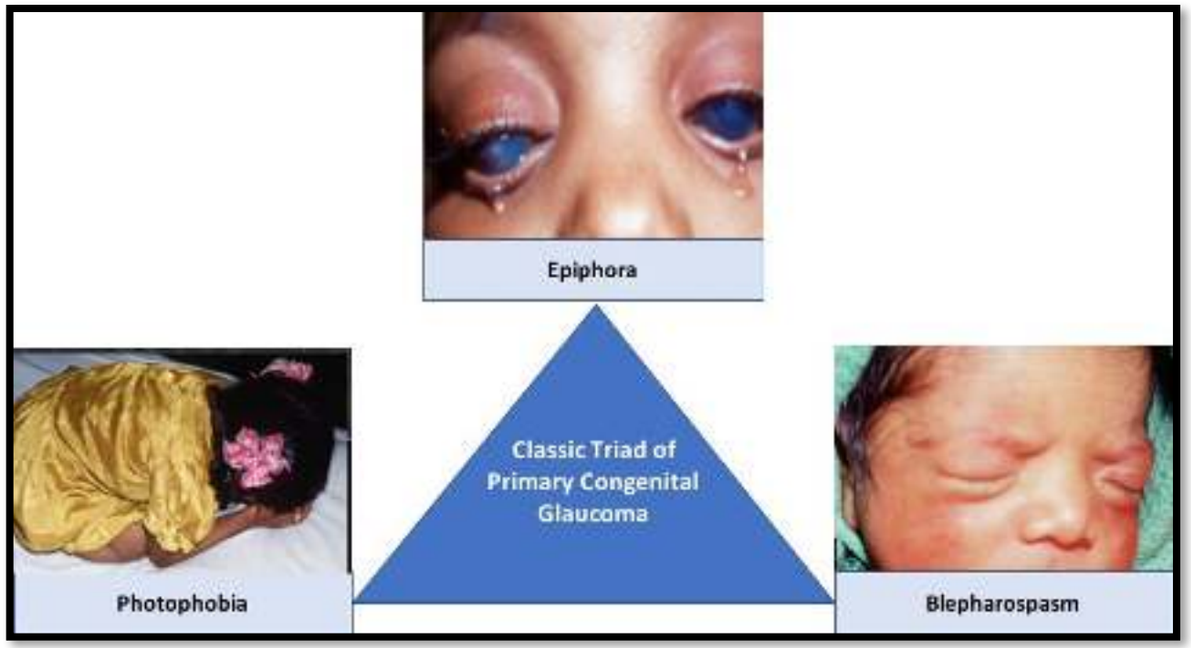
Anterior visual pathway

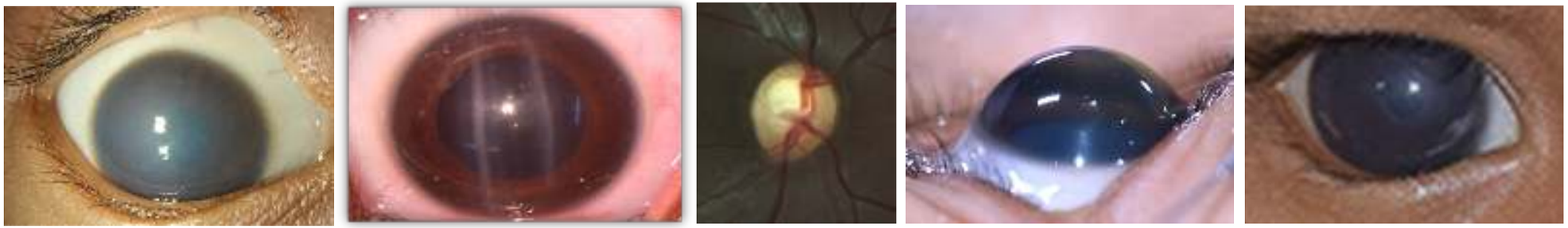
- Optic nerve hypoplasia
- Optic atrophy
- Pseudo glaucomatous cupping

Posterior visual pathway

Cortical visual impairment

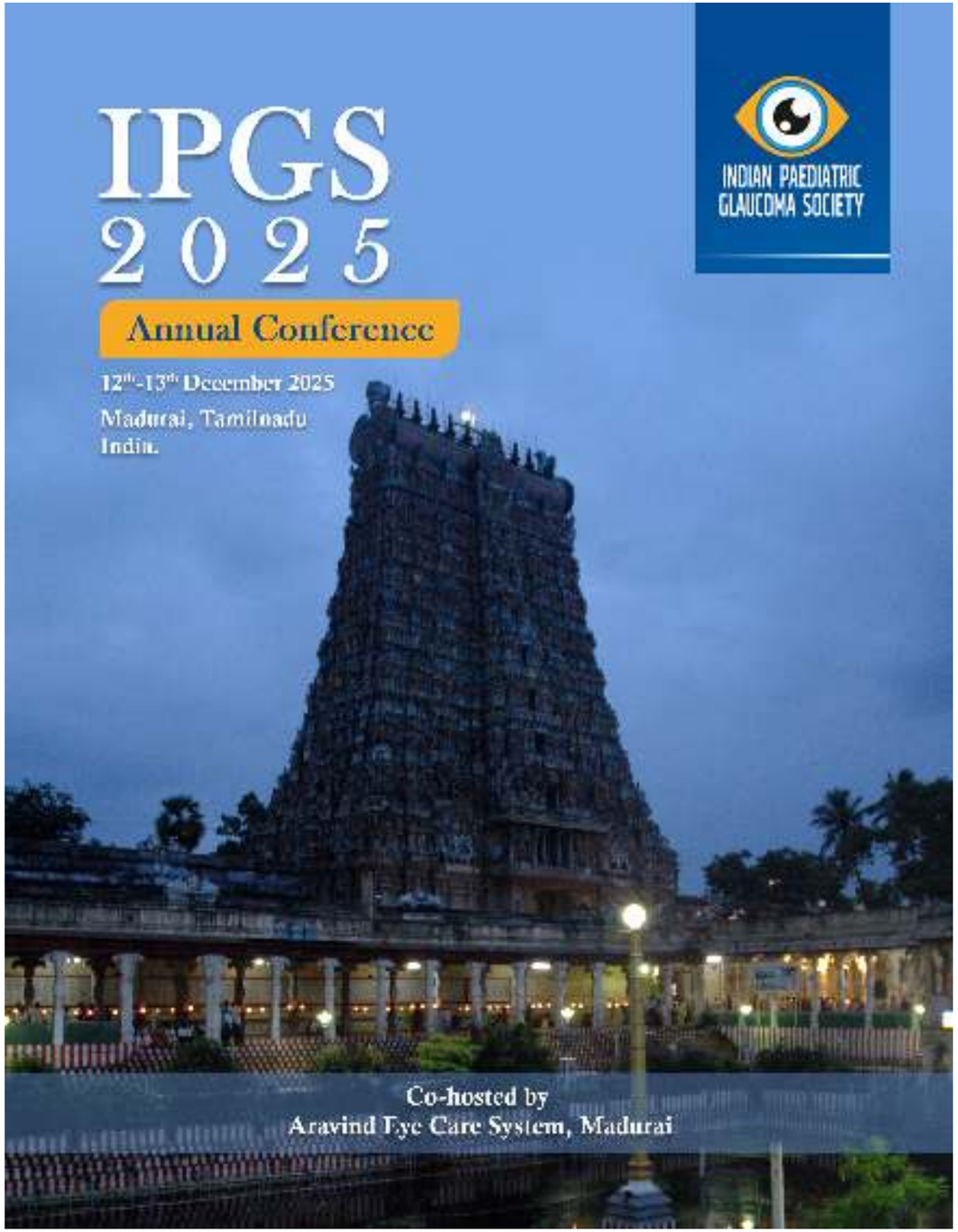
PCG is classical





Summary

- A variety of conditions can mimic PCG
- A detailed history, evaluation and subtle clinical features can help us to differentiate the conditions from PCG
- Implications of unnecessary or inappropriate treatment



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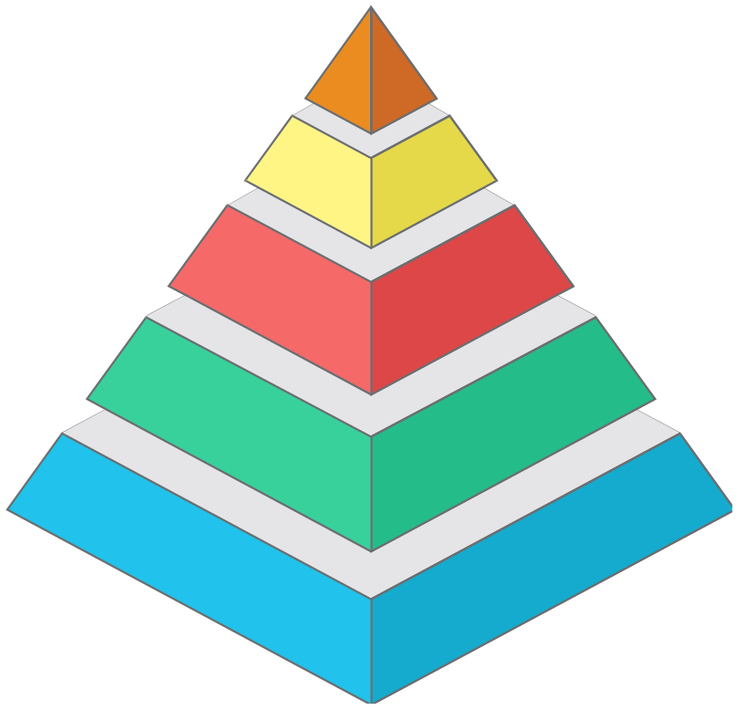
LVPEI
So that all may see

L V Prasad Eye Institute



Thank You