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A Clinical Study on the Etiology, Presentation, and Management of Congenital Cataract

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1. Aims and Objectives

This clinical study was conducted with the following primary objectives:

- To evaluate the incidence, clinical presentation, and etiology of congenital cataracts in patients presenting to a tertiary regional eye care center.
- To analyze the various surgical management modalities utilized for these pediatric cases.
- To document and discuss postoperative visual rehabilitation, outcomes, and complications.

2. Materials and Methods

A prospective cohort of 26 pediatric cases admitted with congenital cataracts was selected from the outpatient department of Sri Padmabhushan P. Sivareddy Regional Eye Hospital. These patients presented directly for ophthalmic checkups or were referred from the pediatric department where they had been admitted for other systemic ailments.

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Clinical History taking

A thorough clinical history was obtained, primarily relying on mothers as the chief informants.

- Symptom Chronology: Onset of symptoms was tracked in days, weeks, months, or years.
- Detection Mode: Documented whether the cataract was detected via routine examination, noticed incidentally by parents/physicians, or identified due to secondary complications.
- Antenatal and Perinatal History: Detailed history of maternal health, trauma, intrauterine infections (especially TORCH), drug intake during pregnancy, radiation exposure, nutritional deficiencies, and birth history (including birth asphyxia and placental insufficiency).
- Pedigree & Family History: Family history of ocular anomalies and parental consanguinity were documented on standardized proforma case sheets.

Inclusion Criteria

Patients were enrolled in the study if they met all of the following criteria:

1. Age Profile: Pediatric patients aged from birth up to 14 years.
2. Diagnosis: Confirmed clinical diagnosis of congenital or infantile cataract (unilateral or bilateral) presenting as any morphological type (e.g., zonular, total, polar, capsular, or coronary).
3. Surgical Candidacy: Children with visually significant lens opacities (e.g., dense central, total, or nuclear opacities) that severely compromised visual function (e.g., limiting vision to fixation/following of light, hand movements, or causing school-related visual difficulties) and required surgical intervention under general anesthesia.
4. Parental Consent: Patients whose parents or legal guardians consented to hospital admission, surgical management, routine postoperative follow-up, and optical rehabilitation protocols.

Exclusion Criteria

Patients were excluded from the study if they presented with any of the following conditions:

1. Mild Ocular Involvement: Patients with incomplete, non-progressive, or minimal central lenticular opacities who maintained good functional peripheral vision and were successfully managed conservatively in the outpatient department with

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pupillary dilation (Atropine 1% ointment) without requiring hospital admission or surgery.

2. Traumatic/Acquired Cataracts: Cataracts resulting directly from mechanical ocular trauma, chemical injuries, or those presenting as true developmental juvenile/senile cataracts outside the infantile classification.
3. Severe Active Ocular Infection: Presenting with acute, active ocular or adnexal infections (e.g., acute dacryocystitis, purulent conjunctivitis) that precluded safe intraocular surgery until resolved.
4. Inability to Complete Surgery or Follow-up: Patients whose parents refused surgical intervention at a young age, or those who were entirely lost to follow-up within the immediate postoperative period.

Pediatric and Systemic Evaluation

All patients underwent systemic and general pediatric examination by a pediatrician. Developmental and anthropometric findings recorded in the proforma sheets included:

- Physical growth parameters (height, weight, mid-arm circumference, and general nutritional status).
- Vital signs (pulse rate, blood pressure).
- Neurological and developmental assessment (mental status, motor milestones).
- Physical anomalies (dental and skeletal abnormalities).
- Systemic evaluation (cardiovascular, respiratory, abdominal, and central nervous system reviews).

Ocular Examination

Ophthalmic assessments were performed while the child was cooperative, naturally asleep, or under systemic sedation advised by the pediatrician.

- External Features: Ocular posture, head posture, facial symmetry, ocular movements, nystagmus type, fixation pattern, and presence of strabismus (squint) were evaluated.
- Anterior Segment: Thorough examination of all anterior segment structures was conducted to check for microphthalmos or other anomalies. Pupillary reactions (both direct and consensual) were carefully recorded.
- Lens Assessment: Morphology, location, density, and extent of the cataract were evaluated. Older children (capable of cooperation) underwent slit-lamp examination to localize lens opacities.

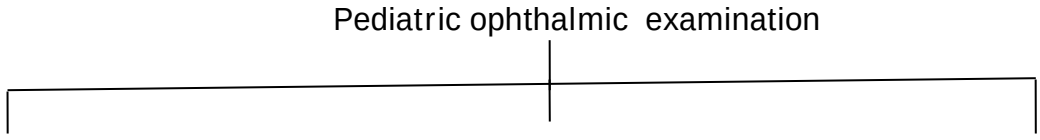
- Visual Acuity:

✓ *Infants and toddlers:* Evaluated by assessing fixation patterns, interest in maternal facial features, and ability to track light or squeaky toys.

✓ *Children > 2 years:* Assessed using common recognizable objects.

✓ *School-age children:* Tested using Snellen's charts or the Tumbling 'E' chart.

- Posterior Segment: Fundus examinations were performed using indirect ophthalmoscopy to assess vitreous, retinal, disc, macular, and vascular integrity. Where dense cataracts precluded direct visualization, B-scan ultrasonography was performed.



behavior/posture	visual assessment	
anterior & posterior		
head posture	fixation/following (infants)	detailed
slit-lamp/ torch		
facial symmetry	common objects (>2 yrs)	papillary
reflexes		
ocular alignment	snellen / 'E'chart (older)	indirect
ophthalmoscopy		
nystagmus/squint		
B-scan ultrasonography		

Preoperative Investigations

Prior to surgery, a battery of routine and specialized diagnostic investigations was completed:

1. Routine Hematology: Complete hemogram.
2. Urinalysis: Routine urine examination.
3. Serology: VDRL testing (to screen for congenital syphilis) and Hemagglutination test for Toxoplasmosis.
4. Biochemical: Fasting blood sugar.
5. Imaging: Chest X-ray, Skull X-ray, and ocular B-scan ultrasonography.

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Management Protocol

- Conservative Treatment: Patients with incomplete or mild central lenticular opacities who retained functional peripheral vision were managed conservatively. They were prescribed Atropine 1% eye ointment (initially twice daily, tapering to once weekly) to maintain pupillary dilation and allow vision around the opacity. Parents were instructed to return if visual function deteriorated.
- Surgical Intervention: Patients with visually significant cataracts underwent surgery under general anesthesia. Intraocular pressure (IOP) and lacrimal sac patency were checked preoperatively.

3. Results and Observations

The clinical data gathered from the 26 operated cases were analyzed across various parameters:

Age Group (Years)	Number of Children (n)	Percentage (%)
≤ 1 Year	2	7.69%
1–3 Years	4	15.38%
3–6 Years	10	38.46%
6–9 Years	6	23.07%
$10+$ Years	4	15.38%
Total	**26**	**100.00%**

Table II: Sex Distribution

A higher proportion of females presented with congenital cataracts compared to males in this cohort.

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Sex	Number of Children (n)	Percentage (%)
Male	11	42.30%
Female	15	57.70%
Total	**26**	**100.00%**

Male to Female Ratio: 0.73:1

Table III: Laterality (Eye Affected)

Bilateral cataracts were highly prevalent, accounting for nearly 70% of the cases.

Eye Affected	Number of Cases (n)	Percentage (%)
Right Eye	3	11.54%
Left Eye	5	19.23%
Bilateral	18	69.23%
Total	**26**	**100.00%**

Table IV: Consanguinity

The cohort was evenly split between patients born to consanguineous couples and those born to non-consanguineous couples.

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Parental Consanguinity	Number of Cases (n)	Percentage (%)
Consanguineous	13	50.00%
Non-Consanguineous	13	50.00%
Total	**26**	**100.00%**

Table V: Associated Ocular and Systemic Diseases

Nystagmus (predominantly horizontal) was the most common associated finding, followed by strabismus (squint) and microphthalmos.

Associated Findings / Pathologies	Number of Cases (n)	Percentage (%)
Horizontal Nystagmus	20	76.92%
Rotatory Nystagmus	3	11.53%
Microphthalmos	5	19.23%
Squint (Esotropia / Exotropia)	12 (6 Esotropia, 6 Exotropia)	46.14% (23.07% each)
Systolic Murmur	1	3.84%
Loss of Hearing	1	3.84%
Mental Retardation	1	3.84%
Failure to Thrive	1	3.84%

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Table VI: Antenatal History and Risk Factors

While most pregnancies were uneventful (76.92%), maternal infections and systemic complications during gestation were present in a subset of cases.

Antenatal / Perinatal Factor	Number of Cases (n)	Percentage (%)
Birth Asphyxia	2	7.69%
Pre-eclamptic Toxemia (P.E.T)	2	7.69%
Toxoplasmosis	1	3.84%
Rubella	1	3.84%
Normal (No history of risk factors)	20	76.92%
Total	**26**	**100.00%**

Table VII: Morphological Types of Cataract (Sex-Wise Distribution)

Zonular (lamellar) cataracts were the most common morphology observed in both male and female patients.

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Morphological Type	Male (n)	Female (n)	Total (n)
Zonular Cataract	5	8	13
Total Cataract	2	4	6
Posterior Polar Cataract	2	0	2
Anterior Polar Cataract	1	1	2
Anterior Capsular	0	2	2
Anterior Capsular Cataract	0	2	2
Coronary Cataract	1	0	1
Total	**11**	**15**	**26**

Table VIII: Morphology vs. Laterality

Among the bilateral cataract cases, zonular cataracts remained the dominant type.

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Morphological Type	Right Eye (n)	Left Eye (n)	Bilateral (n)	Total (n)
Zonular Cataract	0	4	9	13
Total Cataract	1	0	5	6
Posterior Polar Cataract	1	0	1	2
Anterior Polar Cataract	1	0	1	2
Anterior Capsular Cataract	0	0	2	2
Coronary Cataract	0	1	0	1
Total	3	5	18	26

Table IX: Surgical Procedures Performed

Extracapsular cataract extraction (ECCE) with primary posterior chamber intraocular lens (PCIOL) implantation was the most common surgical procedure.

Surgical Procedure	Number of Cases (n)	Percentage (%)
Extra Capsular Cataract Extraction (ECCE) with PCIOL	10	38.46%
ECCE with PCIOL & Primary Posterior Capsulotomy	4	15.38%
Extracapsular Lens Matter Extraction (ECCE alone)	5	19.23%
Needling and Aspiration (with Capsulotomy)	3	11.53%
Needling alone	2	7.69%
Linear Extraction	1	3.84%
Needling and Aspiration with Anterior Chamber IOL (ACIOL)	1	3.84%
Total	26	100.00%

Table X: Postoperative Complications

Posterior capsule opacification (PCO) was the most frequent complication, occurring almost exclusively in eyes that did not undergo primary posterior capsulotomy.

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Postoperative Complication	Number of Cases (n)	Percentage (%)
Posterior Capsule Opacification (PCO)	18	69.23%
Transient Iritis	3	11.53%
Exudative Uveitis	1	3.84%
Decentration of IOL	1 (out of 14 IOL cases)	7.14%

4. Discussion

The study of 26 pediatric patients reveals key patterns in the epidemiology, presentation, and management of congenital cataracts in the region.

Late Presentation and Barriers to Care

The age distribution indicates a significant delay in surgical presentation, with 38.46% of patients presenting between 3 and 6 years of age. This delay often stems from parental ignorance, low socioeconomic status, or failure to notice early visual deficits in infants. The diagnosis was frequently made when the child reached school-going age and faced noticeable difficulties in keeping pace with schoolwork.

Etiology and Prenatal Risk Factors

While 76.92% of cases were idiopathic (unknown etiology), specific prenatal and genetic indicators were identified:

- Consanguinity: Half of the patients (50%) were born to consanguineous parents, suggesting an autosomal recessive genetic influence in this sub-population.
- Antenatal Infections: Maternal Toxoplasmosis (3.84%) and Rubella (3.84%) were linked to congenital lens opacities, underscoring the importance of antenatal screening and immunization programs.

Ocular Comorbidities & Prognostic Indicators

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The presence of nystagmus in 88.45% of cases (76.92% horizontal and 11.53% rotatory) serves as a poor prognostic indicator for postoperative visual recovery. Nystagmus usually signifies early-onset, severe visual deprivation during critical periods of foveal development. Associated microphthalmos (19.23%) and strabismus (46.14% combined esotropia/exotropia) also present additional challenges for successful visual rehabilitation.

Surgical Management and Postoperative Care

Extracapsular cataract extraction (ECCE) combined with PCIOL implantation remains the mainstay of treatment for children older than 2 years.

- Posterior Capsule Opacification (PCO): PCO was the most common postoperative complication, affecting 69.23% of cases. It occurred in almost all cases where a primary posterior capsulotomy was not performed during the initial surgery. Children with intact posterior capsules who later developed PCO were managed with Nd:YAG laser capsulotomy.
- Primary Posterior Capsulotomy: Patients who underwent ECCE + PCIOL combined with a primary posterior capsulotomy showed more stable visual axes and improved long-term visual outcomes.
- Aphakic Correction: Postoperatively, patients were fitted with aphakic spectacle glasses within one week of surgery. Older children (above 5 years) received bifocals to facilitate both near and distance vision. Parents rejected contact lens use due to high costs, handling difficulties, and the demand for frequent follow-up.

Amblyopia Therapy

Unilateral congenital cataracts and late-presenting cases (operated on after 1 year of age) showed more limited visual recovery. To combat deprivation amblyopia, occlusion therapy was initiated as soon as the eye was quiet postoperatively.

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5. Summary and Conclusions

1. Demographics: The study involved 26 children operated on over a one-year period. A higher incidence was noted in females (1:0.73 male-to-female ratio) and in families of lower socioeconomic status.
2. Clinical Presentation: Bilateral cataracts were the most common presentation (69.23%). Zonular (lamellar) cataracts was the most common morphological variant observed (50%).
3. Late Diagnosis: A high percentage of cases were diagnosed late (between 3 and 6 years of age), primarily when school-related visual difficulties arose.
4. Etiological Findings: Consanguinity was present in 50% of the cases. Although most cases were idiopathic, birth asphyxia, pre-eclamptic toxemia, and maternal TORCH infections (Rubella, Toxoplasmosis) were identified as maternal risk factors.
5. Associated Signs: Nystagmus was observed in 88.45% of cases, serving as an indicator of early-onset visual deprivation and a predictor of guarded visual recovery.
6. Surgical Strategy: ECCE with PCIOL is an effective procedure for children over 2 years of age. Performing a primary posterior capsulotomy at the time of surgery reduces the risk of visual axis obscuration.
7. Complications: PCO was the most common complication (69.23%) in eyes that did not undergo primary posterior capsulotomy.
8. Rehabilitation: Postoperative spectacle correction and early, aggressive amblyopia patching therapy are essential to maximize visual rehabilitation, particularly in unilateral cases.

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