Original Article

# Etiology of Congenital Cataract in a Tertiary Hospital, Lahore

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## ABSTRACT

**Purpose:** To identify the etiology of congenital cataracts in a tertiary care center of Lahore.

**Study Design:** Cross-sectional.

**Sampling Technique:** Non-probability purposive sampling.

**Place and Duration of Study:** Department of pediatric ophthalmology The Children's hospital and Institute of Child Health, Lahore from May 2018-October 2019.

.**Methods:** Children one year and younger diagnosed with congenital cataract were included in the study. A detailed ocular examination was done including B-scan. Blood samples of all the infants were tested for presence of specific immunoglobulin M (IgM) and immunoglobulin G (IgG) antibodies by the enzyme-linked immunosorbent assay (ELIZA). Specific blood tests were done to rule out any metabolic disorder. The children were referred to pediatrician for detailed systemic examination. Data were collected, and statistical analysis was done using Excel (Microsoft 2015, version 15.15) and Stata (version 13).

**Results:** A total of 384 infants were included in the study. Mean age of the patients was 5.9 ± 5 months. No cause could be identified in 39.06% and familial pattern was seen in 28.12%. Consanguinity was seen in all familial cases. Rubella was found to be the causative agent in 17.7% participants of the study. Associated ocular problem was seen in 89 (23.17%) infants and microphthalmia being the most frequent. Developmental delay was seen in 126 (6.7%) children.

**Conclusion:** Congenital cataract represents a complex and challenging ophthalmic condition that may result in significant visual impairment. Early identification and timely intervention are pivotal for achieving a positive visual outcome.

**Key Words:** Cataract, Congenital, Rubella, Metabolic disorder.

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## INTRODUCTION

There is a significant impact of blindness on an individual's life, affecting their ability to work, learn and perform everyday task.[1] Children who are born blind or become blind at a young age face additional challenges in terms of social and emotional development as well as educational opportunities.[2] According to an estimate, there are about 1.4 million blind children globally, with approximately 75% of them residing in the impoverished areas of Africa and Asia, where proportion of blindness is high.[3] Preventing visual impairment in children presents a unique challenge that differs from those of managing blindness in adults.[4] Since children have an underdeveloped visual system at birth, clear and focused images must be transmitted to their higher visual centers for normal visual development to occur. If visual maturation is interrupted it cannot be corrected in adulthood, adding urgency to the treatment of childhood eye conditions.[5] Unlike adult conditions, there is a pressing need to address childhood eye diseases to prevent lifelong visual impairment. Prevention, early detection, and treatment of these conditions are crucial to prevent and mitigate

the negative impact of blindness on the individual, families and society.

In this decade, significant progress has been achieved in reducing childhood blindness through public health programs targeting measles and vitamin A deficiency.[6] As a result, pediatric cataract has emerged as a major cause of avoidable blindness in developing countries. Out of an estimated 19 million visually compromised children globally, the incidence of lenticular pathology varies depending on the location in the world map, highest in Africa 22.5%, followed by western Pacific 21.3% and South-east Asia 13.6%.

The lens is composed of specialized cells called lens fibers that are arranged in concentric layers. The transparency of the lens is critical for its function. Any disturbance in the arrangement of the lens fibers can lead to the formation of cataract.[7] Genetic causes of congenital cataract can result from mutations in genes that regulate lens development and growth.[8] More than fifty genes have been found to be associated with congenital cataract.[9] Environmental factors such as maternal infections, exposure to toxins, and metabolic disorders can also result in formation of cataract in the newborn.[10,11] Understanding the underlying causes of congenital cataract is critical for developing effective prevention strategies. The aim of this study is to identify and highlight the preventable causes of congenital cataract in children which are 45% of Pakistan's population and open further research and policy making avenues.

## METHODS

A cross-sectional descriptive study was conducted in the department of pediatric ophthalmology at The Children's hospital and Institute of Child Health, from May 2018 to October 2019. The study included children aged one year and younger who were diagnosed with lenticular change. Sample size was calculated by using the following formula.

Sample size n = (Z^2*p*q)/E^2

Z is the critical value from the standard normal distribution corresponding to the desired confidence level (95% corresponds to Z = 1.96).

P is the proportion of population with the characteristics of interest (unknown so we assume p=0.5 for maximum sample size).

Q is the complement of p, which is 1-p

E is the desired margin of error.

Z=1.96

P=0.5

Q=1-p=0.5

E=0.05

E=799

n= (1.96^2*0.5*0.5)/0.05^2=3.84.16 (rounded off to 385)

Purposive sampling technique was used to collect sample. History, including age of presentation, duration of symptoms, and presenting complaints, were noted. A thorough anterior segment examination followed by a dilated posterior segment evaluation was done in all children. In case of dense media opacity, B-scan was performed. Blood samples of all the infants were tested for the presence of specific immunoglobulin M (IgM) and immunoglobulin G (IgG) antibodies by the enzyme-linked immunosorbent assay (ELIZA).

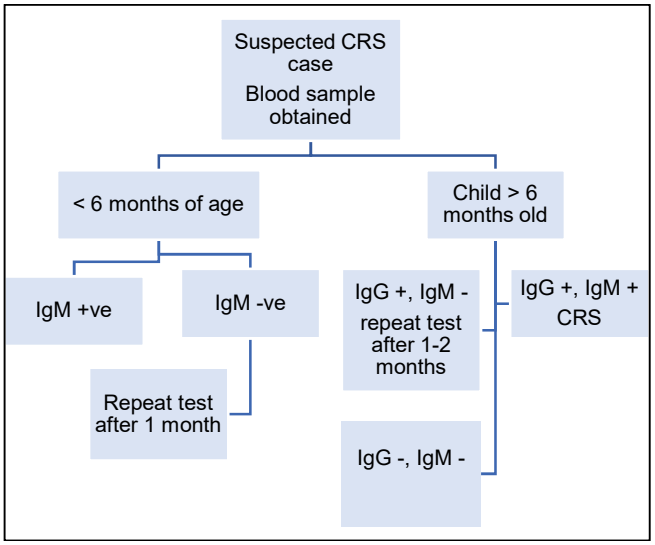


**Figure 1:** Protocol for blood test to rule out CRS (Congenital Rubella Syndrome).

Specific investigations for serum calcium, phosphorus, and glucose were done based on systemic examination. Reducing agent in urine was measured by Benedict's test. Blood glucose level was measured by glucose oxidase–peroxidase method using commercially available kit. A detailed assessment was

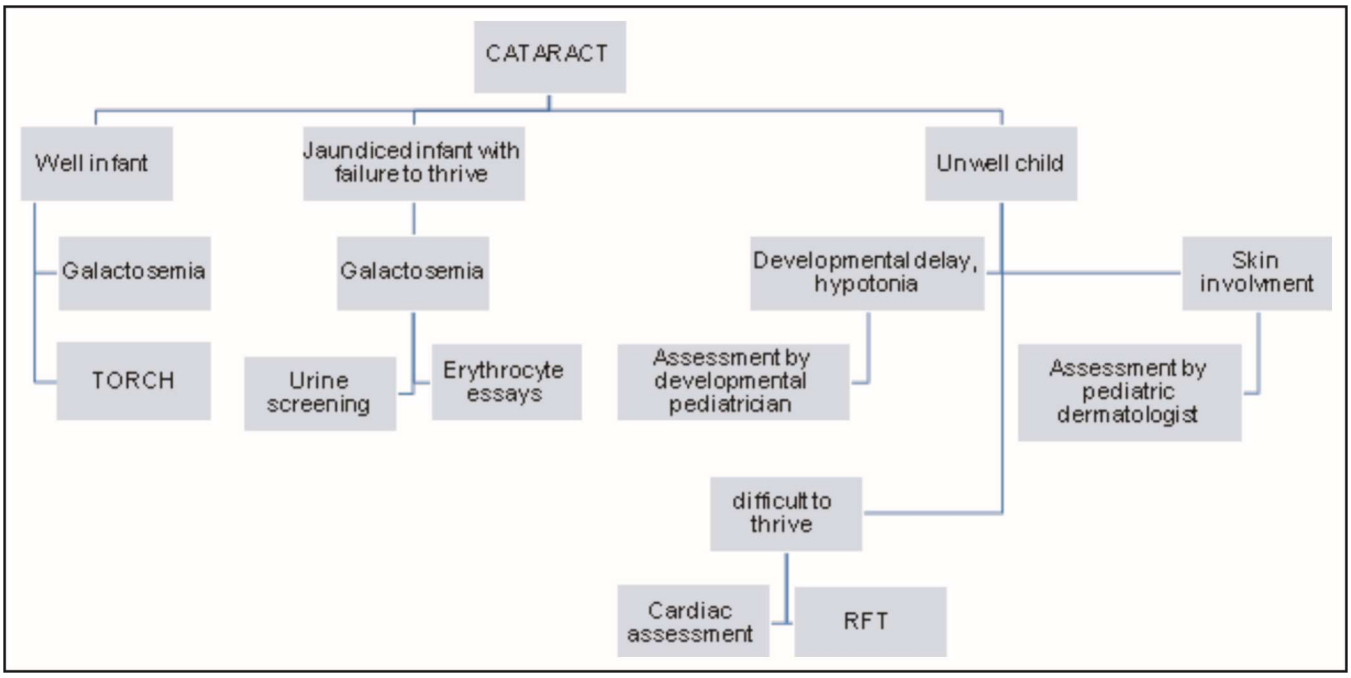


**Figure 2:** Protocol for systemic evaluation of the child (RFT: Renal function test).

asked from pediatric cardiologist to rule out any associated congenital cardiac disease. Deafness was ruled out with the help of audiometry.

Data was gathered and entered into the password-secured personal computer of the prime researcher. A code was assigned to each participant instead of any identifying information. Once data was collected, and the survey closed, statistical analysis was conducted in Excel (Microsoft 2015, version 15.15) and Stata (version 13).

## RESULTS

A total of 384 children one year or younger were included in this study. There were 215(56%) boys and 169 (44%) girls. Mean age was 5.9 ± 5 months. Table 1 shows etiology of cataract in our sample. Consanguinity was positive in 108 (28.1%) patients and I26 (6.7%) patients had associated developmental delay.

There were 28 patients with Down syndrome and two were diagnosed with Turner syndrome. Syndactyly was seen in four patients. Table 2 shows associated ocular findings. Eighty nine (23.17%) patients had associated ocular findings whereas 295 (76.8 %) had isolated cataract.

**Table-1:** *Etiology of congenital cataract.*

| Etiology | N-number Patients |
|---|---|
| Familial | 108 (28.12%) |
| CRS | 68 (17.7%) |
| Idiopathic | 150 (39.06%) |
| Associated with chromosomal abnormalities | 30 (7.8%) |
| Metabolic | 2 (0.26%) |
| Associated with skin involvement | 5 (1.3%) |

**Table 2:** *Associated ocular findings.*

| Associated Ocular Findings | Number (Percentage) |
|---|---|
| Microphthalmia | 31 (8.0%) |
| Microcornea | 16 (23.1%) |
| Raised IOP | 10 (2.6%) |
| Non-dilating pupil | 35 (9.11%) |
| Persistent pupillary membrane | 8 (2.08%) |
| Anterior segment dysgenesis | 11 (2.86%) |
| No associated ocular finding | 295 (76.8%) |

## DISCUSSION

In less developed countries, congenital and developmental cataract, are becoming increasingly prevalent as cause of blindness in children.[12] Cataract is responsible for lifelong visual impairment in approximately 200,000 children/year worldwide.[13] The declining incidence of childhood related factors and the corresponding increase in intrauterine and genetic

factors underscore the need to reevaluate research, training and programmatic priorities.[14]

Out of 384 infants, there were 215 (56%) boys and169 (44%) girls in our study. This was in accordance with a study conducted by Kabylbekova etal in Kazakhstan.[15] A study conducted in a tertiary care hospital in Kathmandu, Nepal found male to female ratio of 1.3:1.[16]

In a study done in Turkey, genetic factors accounted for approximately 29% of congenital cataract.[17] Research suggests that hereditary cataracts may account for a significant proportion, ranging from 8.3% to 25% of cases of congenital cataracts.[9] Familial cataract had a high incidence (28.12%)in our study and consanguinity was seen in all of these cases. A Sharma in his study established that 15.83% patients had a familial history of congenitalcartaract.[16] Rana A et al, from Pakistan reported consanguinity in 69.6% of their cases.[18]

Congenital cataracts may also be present as a component of multisystem genetic disorder. In our study, five patients (1.3%) were diagnosed as suffering from ichthyosis of whom two were siblings.

Literature shows that patients with Down syndrome are at an increased risk of developing cataract.[19] A significant numbers of children (7.8%) in our study were diagnosed with chromosomal abnormalities, (Down syndrome; 29 patients, Turner syndrome; one). Syndactyly (figure 3) was seen in four patients. In a study conducted by S Adhikari et al, 37.5% had hereditary cataract with 8% having systemic syndromes.[20]

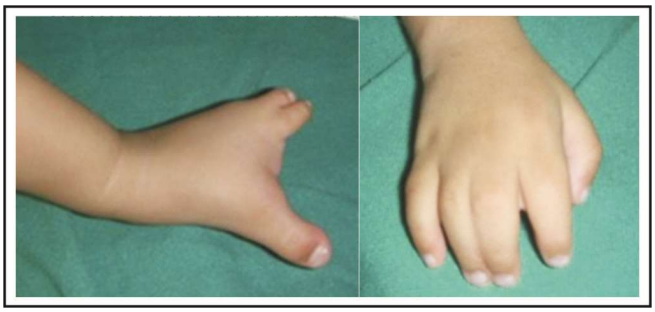

**Figure 3:** Syndactyly in a patient with congenital cataract.

Cataract can manifest in isolation, affecting the lens exclusively, or co-occur with other ocular anomalies,[7] such as microphthalmia, aniridia, anterior chamber developmental anomalies, or retinal degeneration. In our study 89patients (23.17%) had associated ocular findings. A study conducted in Bosnia found that 29.31% of their patients diagnosed with cataract had associated ocularanomalies.[21] Microcornea was seen in 28.3% patients in a study done by A Sharma etal.[16]In our study 23.1% patients had microcornea.

Congenital Rubella Syndrome (CRS) can result in a range of birth defects and health complications, including eye involvement, deafness, heart defects and intellectual disabilities. Vaccination against rubella is an effective preventive measure against CRS, and vaccination campaigns have contributed to a significant reduction in the incidence of CRS globally.[10] Nevertheless, CRS remains a concern in areas with low vaccination coverage, emphasizing the ongoing importance of vaccination programs and awareness raising efforts to prevent this serious and potentially life-threatening condition.[22] A comprehensive understanding of actual prevalence and burden of CRS in Pakistan is currently unavailable. Existing research has primarily focused on institutional and hospital-based studies. Moreover, no research has assessed the prevalence of CRS within the general population, with all the studies thus far evaluating the burden of CRS in symptomatic cohorts of children. In CRS, the foetus synthesizes its own immunoglobulin (IgM antibodies) persisting for 18 months postnatally. However, the sensitivity of IgM estimation for the diagnosis of CRS reduces from 100% before five months, 60% up to 12 months and 40% by 18 months. In our study, 68 (17.7%) infants were diagnosed with CRS by five months of age when the sensitivity of the test is expected to be the highest. Similar results were reported by Sharma A.[16] Javaid M et al, in their study reported 21.7% of cases to be positive for Rubella IgG.[23] Puja Dewan in her meta-analysis reported that 10-15 % of CRS cases had Cataract.[24] Recognizing the significant impact of CRS as a preventable cause of morbidity, particularly childhood blindness and deafness with lifelong special health and social needs, WHO has strongly recommended use of rubella-containing vaccines in many countries.[25] The sustained implementation of vaccination strategies has been successful in drastically reducing rubella cases in the western hemisphere. However, the developing countries have lagged behind in the inclusion of Rubella vaccine in their immunization programs. Pakistan however in collaboration with WHO has included rubella

vaccination in their immunization program and has taken a positive stride towards eliminating Rubella from the country.

Underlying genetic and environmental factors that contribute to congenital cataract may also result in developmental delay, particularly, in areas of language, motor skills, and cognitive functioning. In our study 26 (6.7%) children had developmental delay and were referred to the developmental pediatric department for assessment and management. Early identification and intervention for developmental delay can improve outcomes for these children.

Congenital cataract can be accompanied by various ocular and systemic abnormalities, underscoring the importance of distinguishing between isolated and associated cases. Such differentiation is essential for accurately predicting the potential visual outcomes, as well as for the prompt diagnosis and management of co-morbidities that may contribute to morbidity and mortality. A multidisciplinary approach that involves ophthalmologists, pediatricians, pediatric cardiologist, dermatologist and other specialties as needed is crucial for the effective management of congenital cataract and associated developmental and systemic issues. Geneticists and genetic councilors also have an important role in the management team. There should also be inclusion of school and child visual support services in order to optimize visual potential and help parents and families to access best supportive care.

## CONCLUSION

Congenital cataract presents a complex and challenging ophthalmic condition that may result in significant visual impairment. Typically, the diagnosis of congenital cataract is established either at birth or during the initial year of life. Early identification and timely intervention are pivotal for achieving a positive visual outcome. Consanguinity and CRS have been seen to be a significant cause of congenital cataract. Measures should be taken by the policy makers to look into this phenomenon. Awareness campaigns should be regularly carried out for the society as a whole.

**Conflict of Interest:** Authors declared no conflict of interest.

**Ethical Approval:** The study was approved by the Institutional review board/Ethical review board (OSP-IRB/008-2023).

## REFERENCES


1. **Rainey L, Elsman EBM, van Nispen RMA, van Leeuwen LM, van Rens GHMB.** Comprehending the impact of low vision on the lives of children and adolescents: a qualitative approach. Qual Life Res. 2016;**25(10):**2633-2643. Doi: 10.1007/s11136-016-1292-8.
2. **Ozturk T, Er D, Yaman A, Berk AT.** Changing trends over the last decade in the aetiology of childhood blindness: a study from a tertiary referral centre. Br J Ophthalmol. 2016;**100(2):**166-171. Doi: 10.1136/bjophthalmol-2015-306737.
3. **Lenhart PD, Courtright P, Wilson ME, Lewallen S, Taylor DS, Ventura MC, et al.** Global challenges in the management of congenital cataract: proceedings of the 4th International Congenital Cataract Symposium held on March 7, 2014, New York, New York. J AAPOS. 2015;**19(2):**e1-8. Doi: 10.1016/j.jaapos.2015.01.013.
4. **Gilbert C, Foster A.** Childhood blindness in the context of VISION 2020--the right to sight. Bull World Health Organ, 2001;**79(3):**227-232.
5. **Kabylbekova A, Meirmanov S, Aringazina A, Orazbekov L, Auyezova A.** Age at recognition and age at presentation for surgery for congenital and developmental cataract in Kazakhstan. Ann Med. 2022;**54(1):**1988-1993. Doi: 10.1080/07853890.2022.2091156.
6. **Bruins M, Kraemer K.** Public health programmes for vitamin A deficiency control. Community Eye Health. 2013;**26(84):**69-70. PMID: 24782583; PMCID: PMC3936688.
7. **Shiels A, Hejtmancik JF.** Genetic origins of cataract. Arch Ophthalmol. 2007;**125(2):**165-173. Doi: 10.1001/archopht.125.2.165.
8. **Jakobs PM, Hess JF, FitzGerald PG, Kramer P, Weleber RG, Litt M.** Autosomal-dominant congenital cataract associated with a deletion mutation in the human beaded filament protein gene BFSP2. Am J Hum Genet. 2000;**66(4):**1432-1436. Doi: 10.1086/302872.
9. **Hejtmancik JF.** Congenital cataracts and their molecular genetics. Semin Cell Dev Biol. 2008;**19(2):**134-149. Doi: 10.1016/j.semcdb.2007.10.003.
10. **Vijayalakshmi P, Kakkar G, Samprathi A, Banushree R.** Ocular manifestations of congenital rubella syndrome in a developing country. Indian J Ophthalmol. 2002;**50(4):**307-311. PMID: 12532496.
11. **Nadeem S, Ayub M, Fawad H.** Congenital Cataract: Morphology and Management. Pak J Ophthalmol. 2013;**29(3):**151-155.
12. **Gogate P, Kalua K, Courtright P.** Blindness in childhood in developing countries: time for a reassessment? PLoS Med. 2009;**6(12):**e1000177. Doi: 10.1371/journal.pmed.1000177

13. **Micheal S, Niewold ITG, Siddiqui SN, Zafar SN, Khan MI, Bergen AAB.** Delineation of Novel Autosomal Recessive Mutation in GJA3 and Autosomal Dominant Mutations in GJA8 in Pakistani Congenital Cataract Families. Genes (Basel). 2018;**9(2):**112. Doi: 10.3390/genes9020112.
14. **Pichi F, Lembo A, Serafino M, Nucci P.** Genetics of Congenital Cataract. Dev Ophthalmol. 2016;**57:**1-14. Doi: 10.1159/000442495.
15. **Kabylbekova A, Meirmanov S, Aringazina A, Orazbekov L, Auyezova A.** Age at recognition and age at presentation for surgery for congenital and developmental cataract in Kazakhstan. Ann Med. 2022;**54(1):**1988-1993. Doi: 10.1080/07853890.2022.2091156.
16. **Sharma AK, Shah DN, Upadhyay MP, Thapa M, Shrestha GS.** Demography and etiology of congenital cataract in a tertiary eye centre of Kathmandu, Nepal. Health Renaissance, 2014;**12(1):**3-10.
17. **Taylan Şekeroğlu H, Utine GE.** Congenital Cataract and Its Genetics: The Era of Next-Generation Sequencing. Turk J Ophthalmol. 2021;**51(2):**107-113. Doi: 10.4274/tjo.galenos.2020.08377.
18. **Rana AM, Raza A, Akhter W.** Congenital Cataracts; Its Laterality and Association with Consanguinity. Pak J Ophthalmol. 2014;**30(4)**.
19. **Santoro SL, Atoum D, Hufnagel RB, Motley WW.** Surgical, medical and developmental outcomes in patients with Down syndrome and cataracts. SAGE Open Med. 2017;**5:**2050312117715583. Doi: 10.1177/2050312117715583.
20. **Adhikari S, Badhu BP, Bhatta NK, Jha CB, Baral N, Kumari N.** Etiology and clinical profile of pediatric cataract in a tertiary care center of Eastern Nepal. JNMA J Nepal Med Assoc. 2007;**46(167):**94-98. PMID: 18274562.
21. **Halilbašić M, Zvorničanin J, Jusufović V, Čabrić E, Halilbašić A, Mušanović Z, et al.** Pediatric cataract in Tuzla Canton, Bosnia and Herzegovina. Med Glas (Zenica). 2014;**11(1):**127-131. PMID: 24496353.
22. **Wondimeneh Y, Tiruneh M, Ferede G, Denekew K, Admassu F, Tessema B.** Hospital based surveillance of congenital rubella syndrome cases in the pre-vaccine era in Amhara Regional State, Ethiopia: A base line information for the country. PLoS One, 2018;**13(11):**e0207095. Doi: 10.1371/journal.pone.0207095.
23. **Javaid M, Masud H, Rathore HS, Ali D, Javaid H.** Identification of torch pathogens in congenital cataract patients visiting armed forces institute of ophthalmology. Pak Armed Forces Med J. 2021;**71(5):**1515-1518.
24. **Dewan P, Gupta P.** Burden of Congenital Rubella Syndrome (CRS) in India: a systematic review. Indian Pediatr. 2012;**49(5):**377-399. Doi: 10.1007/s13312-012-0087-4.
25. **Wondimeneh Y, Tiruneh M, Ferede G, Denekew K, Admassu F, Tessema B.** Hospital based surveillance of congenital rubella syndrome cases in the pre-vaccine era in Amhara Regional State, Ethiopia: A base line information for the country. PLoS One, 2018;**13(11):**e0207095. Doi: 10.1371/journal.pone.0207095.

## Authors' Designation and Contribution

Ashvina Qayyum; Consultant Ophthalmologist: *Concepts, Literature Search, Data Acquisition, Data Analysis, Statistical Analysis, Manuscript Preparation, Manuscript Editing.*

Seema Qayyum; Professor: *Concepts, Data Acquisition, Data Analysis, Statistical Analysis, Manuscript Editing, Manuscript Review.*