Original article

Demography and etiology of congenital cataract in a tertiary eye centre of Kathmandu, Nepal

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Abstract

Background: Congenital Cataract is the most important cause of treatable childhood blindness. Rubella is one of the major causes of preventable disease in many countries. There are scanty reports on congenital cataract in Nepal. **Objective:** To find out the demographic and etiological factors of congenital cataract in children. **Method:** In a hospital based cross sectional study, 46 children with congenital cataract were evaluated to find out morphology of cataract, laterality, associated ocular and systemic abnormality, visual status and etiology of cataract. Assessment included antenatal, birth and neonatal history, a detailed eye examination in slit lamp or the operating microscope under general anaesthesia, serum serology for TORCH infections, random blood sugar, urine reducing substance and thyroid profile. **Result:** Among 46 children with congenital cataract, 76.1% children presented before 5 years of age and 78.2% had bilateral onset. Male to female ratio was 1.3:1. Most of the children were legally blind (79.3%) in cataractous eye. Family history of congenital cataract was present in 15.2% cases. The most common mode of presentation was leukocoria in 91.3%. Microcornea (28.3%), resolved uveitis (13.0%), and iris atrophy (8.7%) were the most common ocular associations. Delayed developmental milestone (21.7%) and cardiac anomalies (10.9%) were the most common systemic anomalies. Lamellar cataract (51.3%) was the most common morphology of cataracts observed. The maternal infection was the major cause of congenital cataract in 17.4% cases with predominantly rubella infection in 13% cases. **Conclusion:** Most of the children with cataract are legally blind. Maternal infection in the antenatal period is the major cause of congenital cataract.

Keywords: childhood blindness, congenital cataract, rubella infection

Introduction

The term congenital cataract refers to lens opacity present at birth or that occurs within first year of life.[1] Cataract is one of the most treatable causes of visual impairment and blindness worldwide that accounts for 10% of childhood blindness.[2,3] Recent epidemiological studies report prevalence of congenital cataract at 1.2 to 6.0 cases per 10,000 infants.[4,5] It is estimated that 20,000 to 40,000 neonates are borne with congenital or developmental cataract.[6] In Nepal there are estimated 30,240 blind children and three times as many as having low vision. Cataract is the second leading cause of childhood blindness that accounts for 25 % of childhood blindness in Nepal.[7] Most of the congenital cataract is idiopathic in origin.

Childhood cataract has been reported to be hereditary or associated with rubella infection, metabolic disorders, systemic syndromes and abnormalities.[8,9,10] Maternal infections

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associated with congenital cataract also include rubella, toxoplasmosis, cytomegalovirus, and herpes simplex infections.[11,12] Associated metabolic disorders include galactosemia, hypocalcemia, hypoglycemia, thyroid disorders, while syndromic conditions include Lowe's and Hallermann Strieff Francois syndrome Down's syndrome, etc.[14] Hereditary cataracts are usually transmitted in an autosomal dominant fashion with complete penetrance. The autosomal dominant cataract most commonly present as bilateral lenticular opacities, but variability can be present even within the same pedigree.[15] Autosomal recessive congenital cataract usually results from consanguineous relationship. X-linked congenital cataract is the least common hereditary condition.[16]

The most common metabolic disorder causing congenital cataract is galactosemia.[17] Timely diagnosis of congenital cataract may be helpful in improving useful vision, treating amblyopia and low vision rehabilitation.The knowledge of possible etiological cause of congenital cataract including rubella infection can help in the prevention of childhood blindness. The demography and morphology of cataracts can be helpful in establishing the etiology, surgical outcome and visual prognosis. However, etiology of congenital cataract is not well established and epidemiological data on childhood blindness in Nepalese population is not available in details. Therefore this study was carried out to describe the demographic pattern and aetiological factors of congenital cataract.

Methods

Study design and sample size

In this cross sectional and descriptive study, a total of 6002 children below 15 years of age were examined in Out Patient Department. (OPD) of B.P. Koirala Lions Center for Ophthalmic Studies (BPKLCOS) in the period of one year.

Among them, 46 (0.76%) children having congenital cataract were enrolled in this study. All the children having traumatic cataract, congenital cataract operated prior to this study, severe systemic illness hindering complete eye examination and investigation were excluded. The purpose of the study was clearly explained and verbal consent was received from each parent.

Assessment

Parents or care givers were interviewed with relevant questions in their local language. Relevant information were asked and recorded which included presenting complaint, antenatal, birth history, neonatal history, family history related to congenital cataract. Systemic illness and medications among the mothers during pregnancy were also recorded.

Visual Acuity

Visual acuity of the child was assessed with the help of age appropriate testing methods for verbal and non verbal children. Children below 2 years of age were tested with Follow & Fixate method. Children between 2-5 years of age were tested either by using Cat Ford drum or Lea Symbol chart. Visual acuity of children above 5 years of age were tested by using Snellen's E- chart.

Assessment of strabismus

Extra ocular motility examination was performed using pen torch light in all cardinal gazes. Cover test was performed to detect strabismus and the amount of strabismus was measured with the Krimsky prism test. Careful eye examinations were performed to detect nystagmus by torch light examinations.

A complete eye examination

Gross anterior segment examination of eye included pupillary examination to find out the causes of leukocoria. Detailed slit lamp examination of anterior segment was performed using Haag Streit -900 slit lamp biomicroscope to rule out congenital anomalies. Hand held slit lamp (Carl Zeiss, OSRAM-64222) was also used in infants and uncooperative children. Detailed morphology of cataract evaluation was performed after dilation of pupil with the instillation of eye drop (Tropicamide 0.5% and Phenylephrine 2.5%) for three times at the interval of 15 minutes. Posterior segment examination was also performed after dilation with the help of direct and indirect

ophthalmoscope (Neitz) with Volk +20D lens. Ultrasound B -Scan was also performed at the same time in the children when fundus was not clearly visible. Intraocular pressure was measured with the Perkins tonometer. Mothers as well as siblings of the children were also examined to rule out cataract.

Referral

Suspected children having possibilities of systemic illness or syndrome were referred to a paediatrician. Suspected cases of rubella were referred to specialists for cardiac and ENT evaluation at Tribhuvan University Teaching Hospital (TUTH).

Investigation

Children

Investigations performed in every child included hemoglobin (Hb), TC, DC, ESR, blood sugar, serum calcium, ELISA (IgG/IgM) for toxoplasmosis, rubella, cytomegalovirus, Herpes simplex I and Herpes simplex II, thyroid functions tests (T3, T4, and TSH), urine (RE/ME), and urine for reducing substances and chest X-ray P-A view.

Mother

Detailed investigation in every mother was performed for blood sugar, serum calcium, ELISA (IgG/IgM) for toxoplasmosis, rubella, cytomegalovirus, Herpes simplex I and II and thyroid function tests.

Statistical analysis

All data were evaluated using statistical tools in Microsoft excel 2003. All the variables related to age, gender, laterality, history, ocular and systemic disorders, with relavant investigations were presented in frequency and percentage.

Results

Demographic distribution of children

Demographic distribution of children is presented in table 1. The age of the children ranged from 0-14 years. The youngest one was an infant of 16 days. Most of the children belonged to the age group of five years or less in 76.1%. Male female ratio was 1.3:1. Bilateral congenital cataract was present in 78.2%.

Most of the children belonged to the Central development region of Nepal (54.4%) followed by Western development region (26.1%). Unilateral cataract was present in children above 5 years of age only.

Table 1: Demographic distribution of children

Categories		Frequency	%
Age	≤ 5 years	35	76.1
	>5 years	11	23.9
Sex	Male	26	56.5
	Female	20	43.4
Laterality	Unilateral	10	21.8
	Bilateral	36	78.2
Geographical location	Eastern	4	8.7
	Central	25	54.4
	Western	12	26.1
	Mid-western	3	6.5
	Far-western	2	4.3
Total		46	100

Presenting Visual Acuity

Table 2 represents presenting visual acuity in the participants. Moderate visual impairment was present in seven eyes (8.5%), severe visual impairment was present in 10 eyes (12.2%) and legal blindness to the light perception was present in 65 eyes (79.3%).

Table 2: Presenting visual acuity in cataractous eyes

Visual Acuity	Number of eyes N (%, 95% CI)
Light perception present(LP+)	4 (4.9%)
Follows Light & Fixation at Object	34 (41.5%)
Follows Light & Poor Fixation	27 (32.9%)
3/60 – 6/60	10 (12.2%)
6/60 –6/36	7 (8.5%)
Total	82 (100%)

Antenatal, birth and family history

Table 3 shows antenatal, birth and family history. Based on the recall of history related to pregnancy, eight mothers (17.4%) had history of fever during their first trimester of pregnancy for which they took oral antibiotics and NSAID. Another two mothers (4.3%) had taken unknown herbal medicine for unwanted pregnancy. Fever

with skin rashes, pre-eclampsia, jaundice with chronic diarrhoea was present in each mother out from others three. One mother had night blindness, decreased vision and hearing loss along with bilateral cataract. Forty mothers (87%) had full term normal delivery while other six (13%) had preterm delivery with low birth weight babies (≤ 1500gm). Out of the six children with preterm delivery, four of them had evidences of neonatal asphyxia**.** Among the 46 children, seven (15.2%) had family history of congenital cataract. All the children having positive family history had bilateral congenital cataract.

Table 3: Antenatal, birth and family history

Category	Characteristics features	Frequency	Percentage
Systemic history of mother	Fever without rashes	8	17.4
	Fever with skin rashes	1	2.2
	Pre-eclamsia	1	2.2
	Jaundice with diahorrea	1	2.2
	Night blindness	1	2.2
	UTI	1	2.2
Medication during pregnancy	Antibiotics & NSAID	8	17.4
	Herbal medicine	2	4.3
	Anti hypertensive drug	1	2.2
Delivery	Preterm (< 32 weeks)	6	13.0
Family history	Congenital cataract	7	15.2
Neonatal problem	Neonatal asphyxia & low birth weight	4	8.7
	Low birth weight	2	4.3

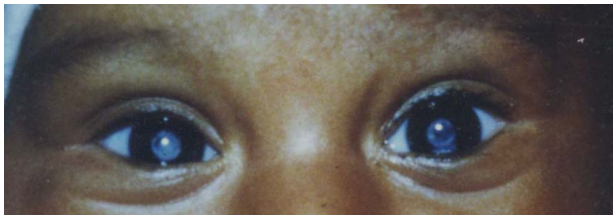
Table 4 shows the systemic and ocular abnormalities. Leukocoria was the commonest presenting sign in 42 cases (91.3%) followed by associated signs of nystagmus in 17 children (36.9%) and strabismus in 12 children (26.1%). Ratio between esotropia to exotropia was 1.4:1. Ocular abnormalities were detected in 32 children (69.6%). They were microcornea (28.3%), sclerocornea (4.3%), resolved uveitis with posterior synechiae (13.0%), iris atrophy (8.7%), rubeosis iridis (2.2%), chorioretinal scars (6.5%), pigmentary retinopathy (2.2%), optic disc hypoplasia (2.2%) and optic atrophy (2.2%).

Table 4: Systemic and ocular abnormalities in children with congenital cataract

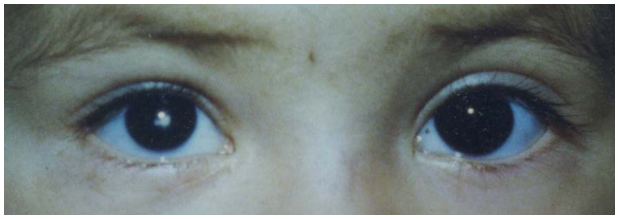
Category	Characteristics features	Frequency	Percentage
Mode of presentation	Leucokoria	42	91.3
	Strabismus	12	26.1
	Nystagmus	17	36.9
Ocular disorders	Micro-cornea	13	28.3
	Sclerocornea	2	4.3
	Resolved uveitis with posterior synechiae	6	13.0
	Iris atrophy	4	8.7
	Rubeosis iridis	1	2.2
	Chorioretinal scars	3	6.5
	Pigmentary retinopathy	1	2.2
	Optic disc hypoplasia	1	2.2
	Optic atrophy	1	2.2

Systemic abnormality	Delayed developmental milestone (DDM)	7	15.2
	Cardiac anomalies	5	10.9
	Ventricular septal defect	*2*	*4.3*
	Pulmonary stenosis	*2*	*4.3*
	Atrial septal defect	*1*	*2.2*
	Hydrocephalous + DDM+ Mental retardation	3	6.5
	Down syndrome	2	4.3
	Sensory-neural hearing loss (SNHL)	2	4.3
	Syndactaly	1	2.2
	Microcephaly	2	4.3

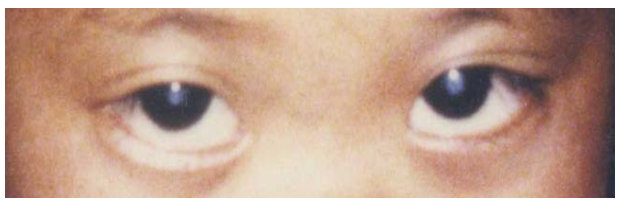
Systemic abnormalities were reported in 22 children (47.8%). Among them, seven children (15.2%) had delayed developmental milestones, two children (4.3%) had clinical features of Down's syndrome, two children (4.3%) had microcephaly, three children (6.5%) had combination of hydrocephalous with delayed developmental milestone and mental retardation, two children (4.3%) had sensory neural hearing loss on audiometric test, One child (2.2%) had syndactaly in his fore arm, and five children (10.9%) had cardiac murmur having revealed atrium septum defect in one, ventricular septum defect in two and pulmonary stenosis in the other two on echocardiography.



Picture 1: A child with bilateral congenital cataract



Picture 2: A child with unilateral congenital cataract



Picture 3: A child with Down's syndrome with bilateral congenital cataract

Morphology of cataract

Morphologically, six types of cataract were observed in children with congenital cataract. Among 36 children (72 eyes) having bilateral cataract, lamellar cataract was most commonly observed in 48 eyes (66.7%) followed by membranous (partially absorbed) in 11.1%, nuclear cataract in 11.1% and total cataract in 11.1% (8 eyes) each. Among 10 children having unilateral cataract, lamellar cataract observed in 6 eyes (60%) was the most common.

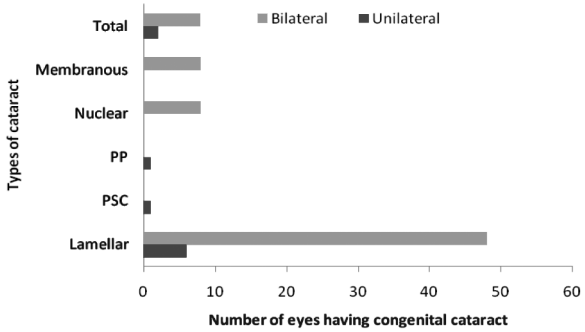


Figure 1: Morphological distribution of congenital cataract. (PP= posterior polar cataract; PSC = posterior sub-capsular cataract)

Serological Test (ELISA) for TORCH infections

Table 5 shows serological test for TORCH infection. Serological test (ELISA) for TORCH infections i.e. toxoplasmosis, rubella, cytomegalovirus infection and Herpes simplex type I and II were performed in both serum samples obtained from the child and mother. ELISA IgM was found negative in all cases. But six (13.0%) children and their mother had a significant IgG positive for rubella, two of them had congenital rubella syndrome. Cytomegalovirus infections showed evidences of past infections in 28.3% children.

Table 5: Seropositivity of different components of TORCH

Serological test	Children N (%)	Mother and child N (%)
ELISA (Ig G) - Toxoplasmosis	1 (2.2)	1 (2.2)
ELISA (Ig G) - Rubella	6 (13.0)	6 (13.0)
ELISA (Ig G) - Cytomegalovirus	1 (2.2)	1 (2.2)
ELISA (Ig G) - Herpes simplex I	0	0
ELISA (Ig G) - Herpes simplex II	0	0

Table 6 shows association of maternal infection and systemic abnormalities. In majority of the children negative for rubella infection, lamellar cataract was found.

Table 6: Association of maternal infection and systemic abnormalities

		Morphology of cataract		Relative risk	95% CI	p-value
		Lamellar	Non-lamellar			
Rubella Infection	Positive	5	1	1.7	0.19-15.9	0.59
	Negative	49	17			
Systemic abnormalities	Present	19	3	2.7	0.69-10.57	0.14
	Absent	35	15			

Table 7 shows association of gender of the child with age of presentation. More of male children were brought to the hospital than female children though the difference was not statistically significant.

Table 7: Association of gender of the children with age of presentation

Gender	Age of presentation		Relative risk	95% CI	P value
	<5 years	>5years			
Male	20	6	1.1	0.28-4.34	0.88
Female	15	5			

Aetiology of congenital cataract

Aetiology of cataract was established in 19 (41.3%) children. Among them, maternal infection was the major cause of cataract in 8 (17.4%) children followed by hereditary cataract in seven (15.2%) children, and associated Down's syndrome in two (4.3%) children. Aetiology of congenital cataract was largely undetermined in 27 (58.7%) children.

Discussion

Our study shows the prevalence of congenital cataract of 0.76% with the male: female ratio of 1.3:1. Most of the children in our study presented with congenital cataract before five years of age. Aetiology of congenital cataract was established in 41.3% children. Cataracts associated with maternal infection (17.4%) and hereditary cataracts (15.2%) were the more common type. Aetiology was largely undetermined in 58.7% like in the other previous studies. Gilbert et al (1997) reported majority of congenital cataract to be idiopathic (46.05%) followed by hereditary in 19.4% cases.[6] Johar et al (2004) reported major aetiology of non traumatic cataract to be idiopathic in 73% followed by 7.2% hereditary associated and 4.6% congenital rubella syndrome associated.[3] Our finding of 4.3% congenital rubella syndrome associated cataract is similar to the findings of Johar et al and that of El Fkih et al (2007). The study reported aetiology of congenital cataract in 62.5% of cases. Hereditary (42.3%) was the most common cause followed by metabolic in 7% and intrauterine infection in 4.7% of cases.[18] However, in the study by Adhikari et al, the major aetiology of non-traumatic cataract was

hereditary in 37.5% followed by idiopathic in 27.7% children, associated with systemic syndrome in 8.0%, due to maternal infection in 4.5%, metabolic disorder in 2.7% and associated with ocular dysmorphology in 9.8%.[19] Most of the recent studies also highlighted the congenital rubella syndrome as one of the major causes of congenital cataract.[12,20,21,22,23] In our study, serological test revealed rubella infection (13.0%) as the most common cause of maternal infection leading to congenital cataract. In the Johar et al study (2004), 67% of the mother had history of illness, and 22% had taken medications during pregnancy in the group of undetermined cases.[3] In our study 13 mothers (28.3%) had febrile illness during pregnancy and eleven of them had taken medications. In our study systemic abnormality was present in 22 children (47.8%) and ocular disorder was preset in 29 children (63%) and bilateral cataract was present in 78.2%. Vijayalakshmi et al (2002) reported bilateral congenital cataract in 89%.[23] In our study the most common mode of presentations were leukocoria, nystagmus and strabismus while the most common associated ocular disorders were microcornea (28.3%), resolved uveitis (13.0%), and iris atrophy (8.7%). Delayed developmental anomaly (21.7%)andcardiac anomalies (10.9%) were the most common associatedsystemicanomaliesinourstudy.Gilbert et al (1997) also reported strabismus (28.9%) and nystagmus (15.8%) as the most common associated ocular abnormalities.[6] Vijayalakshmi et al (2002) reported microphthalmos in 85.1% eyes, iris abnormalities in 58.6% eyes, and pigmentary retinopathy in 37.9% eyes. In the same study, systemic manifestations included cardiac anomalies in 50% and neurological anomalies in 34% children.[23] Low birth weight (below 2000gm) was seen in 30% infants in the Vijayalakshmi et al study where as it was seen in 4.3% in our study. Lamellar cataract (65.8%) was the most common type of congenital cataract in both unilateral and bilateral congenital cataract followed by total cataract in our study. The Vijayalakshmi et al study (2002) reports nuclear cataract in 97.5%.[23] In contrary, nuclear cataracts was present in only 11.1% children with bilateral congenital cataract in our study. The limitations of our study include small sample size. We couldn't subject all children and mothers for complete investigations to determine the possible aetiology of congenital cataracts. Chromosomal and genetic investigations were not done in this clinical study.

Conclusion

Congenital cataract is one of the important causes of paediatric blindness. Majority of the cataract had bilateral presentations with the clinical features of leukocoria, nystagmus and strabismus. Esotropia was more common type of strabismus than exotropia. Majority of the cases were presented to the hospital before five years of age. Lamellar and nuclear cataract was most common morphologic type of congenital cataract. Partially absorbed cataracts and nuclear cataracts were present in rubella positive cases. Common etiological factors were hereditary and maternal infections including rubella. There was no case associated with thyroid disorders.

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