

# Congenital Cataracts; Its Laterality and Association with Consanguinity

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*Pak J Ophthalmol 2014, Vol. 30 No. 4*

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**Purpose:** To study the frequency of laterality (bilateral vs. unilateral) and its importance among congenital cataracts. We also investigated consanguinity as a risk factor in congenital cataract cases.

**Material and Methods:** This study was conducted in Ophthalmology department, Holy family hospital, Rawalpindi, from 2<sup>nd</sup> January 2013 to 2<sup>nd</sup> February 2014. A total of 112 eyes and 86 patients in age range from 3 months to 26 years and all types of visually significant congenital cataracts total or partial without prior history of ocular trauma and syndromic association were recruited for the study. Frequency distribution, test of significance was carried out using Statistical Package for Social Sciences Version 20.0.

**Results:** A total of 112 cases (61 males, 51 females) were recruited in the study. There was no statistically significant difference between different age groups and gender ( $p=0.2$ ). The unilateral cases were 19.6% and bilateral were 80.4%. Consanguinity was present in 69.6% ( $n=78$ ) and absent in 30.4% ( $n=34$ ). The difference was statistically significant ( $p=0.00$ ).

**Conclusion:** Bilateral congenital cataract is a more common presentation as compared to unilateral cataract. Consanguinity is an important risk factor for congenital cataract especially bilateral cataracts.

**Key Words:** Congenital Cataracts; ocular trauma, Syndromic association

Congenital cataract is an important cause of preventable visual deprivation in children accounting for 5%-20% of blindness in children worldwide.<sup>1,2</sup> World - wide, the number of children who are blind is estimated to be 1.4 million, 190,000 of them from cataract.<sup>3</sup> Cataract in children can be classified as congenital, developmental or traumatic.<sup>4</sup>

Congenital cataract presents either from birth or shortly thereafter, while developmental cataract usually refers to cataract that appears after the age of two.<sup>5</sup> Pediatric cataracts are responsible for more than 1 million childhood blindness in Asia.<sup>6</sup> The prevalence of cataract in children has been estimated about 3 in 10,000 live births.<sup>7</sup> Ocular morbidity is mainly caused by obstruction to development of the visual system and it has great physical, social economical and psychological impact.

Prevention of visual impairment and blindness in childhood due to congenital and infantile cataract is an important international goal<sup>4</sup> and is a priority for vision 2020.<sup>8</sup> Epidemiology of congenital cataract is not fully understood because it's not a specific entity but combination of multiple factors, including many associated ocular pathologies.

Density and laterality of congenital cataract are one of the most important parameters in terms of visual outcome, others are type of cataract, associated ocular pathology and delay in presentation to hospital. Unilateral dense cataract is a definite indication for early cataract surgery (preferably within days) which is followed by aggressive amblyopia treatment, even then the results mostly remain poor.<sup>9</sup> Unilateral cataracts are generally sporadic, with no family history of cataract or systemic illness, and affected infants have history of full-term and normal health.<sup>9</sup>

Genesis of congenital cataract is still not explored well and very little is known because of modern techniques, long term accurate data needed and lack of sensitive investigative procedures. Genetic factors are important in the etiology of congenital cataract, up to half of childhood cataracts are genetic in origin<sup>10</sup>. Nearly, one-third of total congenital cataract cases are familial.<sup>11</sup> These types of cases are mainly because of genetically induced developmental alterations among the crystalline lens and surrounding tissues. There are a lot of ongoing epidemiological studies to find out risk factors like intrauterine infections, certain enzymes deficiency, and sporadic. The knowledge about the causes is important to develop appropriate planning strategies, which are not available for many regions of the world and where these are available, has been obtained mostly from studies of selected populations, or from routine sources which are often based on small numbers of cases.<sup>12</sup>

Routine ocular examination of young infants is widely recommended to ensure that treatment, genetic counseling, and other advice and support are offered at the earliest opportunity. The parents and any siblings should be examined thoroughly even in the absence of positive family history. In this study we are trying to analyse frequency of laterality among congenital cataract and to investigate consanguinity as risk factor among hospital data of congenital cataract in patients attending our ophthalmology department.

## MATERIAL AND METHODS

Our study includes patients with congenital / infantile cataract presenting to Ophthalmology department, Holy family hospital, newly diagnosed during the 12-month period from 2<sup>nd</sup> January 2013 to 2<sup>nd</sup> February 2014 identified prospectively. It include 112 eyes and 86 patients in age ranging from 3 months to 26 years and including all types of visually significant cataracts total or partial without prior history of ocular trauma and syndromic association. All affected individuals underwent a detailed history and ophthalmological examination. Morphological details of cataract including other ocular associations and also detailed dilated fundus examination where possible were recorded. Informed consent was obtained and detailed medical and family history with especial emphasis on consanguinity was obtained by taking detailed history from parents or guardian of children on admission using a standardized questionnaire. Ophthalmic examination included assessment of the pupillary red reflex with a direct ophthalmoscope, visual acuity or

fixation and following behavior checked according to age of patients, complete anterior segment examination with slit lamp and retinoscopy was done, B-scan was also done where required. All patients underwent irrigation and aspiration of cataract with or without IOL followed by aphakic correction where required according to latest recommendations. Laterality and association of consanguinity with congenital cataract was noted and assessed.

Statistical analysis was performed using Statistical Package for Social Sciences Version 20. Fisher exact test was performed to determine statistically significant differences in the gender of the population. A p value of <0.05 was taken to be significant in all analysis.

## RESULTS

Congenital cataract characteristics and demographics of the cases are shown in table 1 while table 2 shows laterality with age distribution, picture 1 showing consanguinity with laterality while picture 2 showing different morphological presentations of congenital cataract with gender distribution.

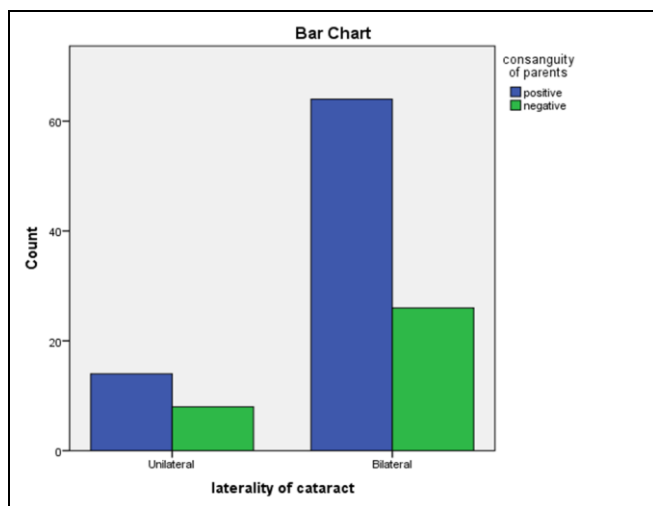
A total of 112 cases (61 males, 51 females) were recruited in the study. The distribution of congenital cataract cases for different age groups in our study was as follows for less than 1 year age group 24.4% (n=15 males, n=11 females), age group 1-5 years 25% (n=19 males, n=7 females), age group 6-10 years 19.6% (n=12 males, n=10 females), age group 11-15 years 17.9% (n=8 males, n=12 females), age group 16-20 years 9.8% (n=4 males, n=7 females) and age group more than 20 years 6.3% (n=3 males, n=4 females). There was no statistically significant difference between different age groups and gender (p=0.2). The bilateral cataracts (n=90) included 48 (53.33%) males and 42 (46.66%) females, while unilateral cataract (n=22) comprised of 13 (59.09%) male and 9 (40.91%) female cases. In both bilateral and unilateral cataract groups males were more as compare to females. This difference was not statistically significant (p=0.093). The cumulative unilateral cases were 19.6% and bilateral were 80.4%. In age group less than 1 year 18% were unilateral and 24% were bilateral, in group 1-5 years unilateral were 9% and bilateral were 26%, in age group 6-10 years unilateral was 23% and bilateral were 18%, in age group 11-15 years unilateral were 18% and bilateral were 18%, in age group 16 - 20 years unilateral were 14% and bilateral were 9%, in age group more than 20 years unilateral were 18% and

**Table 1:** Gender distribution with Laterality of congenital cataract

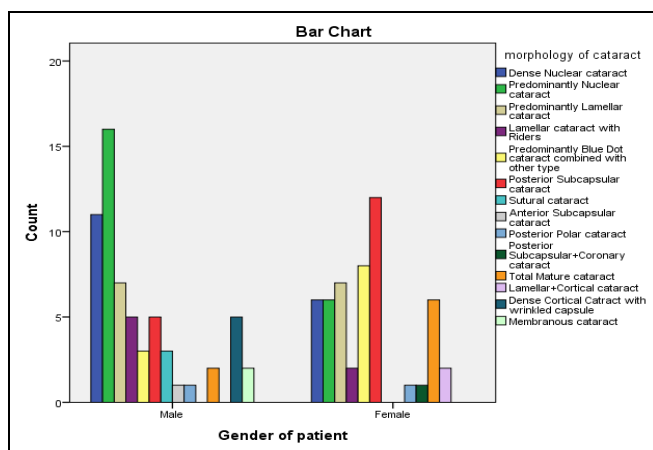
Characteristic	Unilateral Congenital Cataract Group (n = 22)		Bilateral Congenital Cataract (n = 90)	
	Males	Females	Males	Females
Gender	Males 13 (59.09%)	Females 9 (40.91%)	Males 48 (53.33%)	Females 42 (46.66%)

**Table 2:** Laterality vs. age group of congenital cataract

	Laterality of cataract	Age of Patient						Total
		< 1 yr	1 - 5 yrs	6 - 10 yrs	11 - 15 yrs	16 - 20 yrs	> 20 yrs	
	Unilateral	4	2	5	4	3	4	22
	Bilateral	22	24	17	16	8	3	90
	Total	26	26	22	20	11	7	112



**Fig. 1:**



**Fig. 2:**

bilateral were 3%, there was no statistical significance ( $p=0.093$ ) between age of presentation of congenital cataract and laterality.

Consanguinity was present in 69.6% ( $n=78$ ) and absent in 30.4% ( $n=34$ ). The difference was statistically significant ( $p=0.00$ ). Out of total cases with positive consanguinity 18% ( $n=14$ ) were unilateral and 82% ( $n=64$ ) were bilateral while with absent consanguinity 24% ( $n=8$ ) were unilateral and 76% ( $n=26$ ) were bilateral. There was no statistically significant difference between the two groups ( $p=0.49$ ).

In our study we also observed different morphologies of cataracts. The frequencies of different types of congenital cataract were; dense nuclear cataract 15.2% ( $n=11$  males,  $n=6$  females), predominantly nuclear cataract 19.6% ( $n=16$  males,  $n=6$  females), predominantly lamellar cataract 12.5% ( $n=7$  males,  $n=7$  females), lamellar cataract with riders 6.3% ( $n=5$  males,  $n=2$  females) f, predominantly blue dot cataract 9.8% ( $n=3$  males,  $n=8$  females), posterior sub-capsular cataract 15.2% ( $n=5$  males,  $n=12$  females), sutural cataract 2.7% ( $n=3$  males,  $n=0$  females), anterior sub-capsular cataract 0.9% ( $n=1$  males,  $n=0$  females), posterior polar 1.8% ( $n=1$  males,  $n=1$  females) , sub-capsular coronary cataract 0.9% ( $n=0$  males,  $n=1$  females) , total mature cataract 7.1% ( $n=2$  males,  $n=6$  females), cortical cataract 1.8% ( $n=0$  males,  $n=2$  females) , cortical cataract with wrinkle 4.5% ( $n=5$  males,  $n=0$  females), and membranous cataract 1.8% ( $n=2$  males,  $n=0$  females). There was statistically significant difference between gender and morphology of cataract ( $p =0.01$ ).

## DISCUSSION

Congenital cataract is a major cause of blindness in children. Congenital cataract is important in the regards that it blurs the retinal image as well as disrupts the development of the visual pathways in the central nervous system. Congenital cataract is a rare disease, but it is a major cause of low vision or blindness among both developed<sup>13</sup> and developing<sup>14</sup> countries. The causes for most of the congenital cataracts remained unknown.<sup>15,16</sup> Prevention of visual impairment due to congenital and infantile cataract is an important component of world health organization's international program for elimination of avoidable blindness by 2020<sup>17</sup>. Surgical removal of the opacified lens with and without intraocular lens implantation is the only treatment available for congenital cataract.<sup>18</sup>

In our study male were 55% (n=61) and females were 45% (n=51). Male to female ratio was similar to the study of Mwendu J et al<sup>19</sup>, who had 55% (n=99) males and 45% (n=81). In the same study bilateral cataracts were 66% and unilateral were 34%, while in our study it was 80.4% and 19.6% respectively. Rahi JS et al<sup>20</sup>. In their study also the same ratio of laterality 66% and 34% was observed respectively. The difference was not statistically significant in both studies. The difference between our study and the two groups was because of the included age group which was more in our study from 3 month to 26 years while in the rest of the two studies it was 1 year of age. Ruddle JB et al,<sup>21</sup> also observed in their study that there was no significant difference between laterality of cataract (bilateral 45.5% vs. unilateral 55.5%) or gender ( $p = 0.068$ ). Laterality is one of the most important parameters in terms of management. Unilateral cataracts have poor prognosis as there are much more chances of amblyopia as compare to bilateral. In unilateral congenital cataract prognosis for visual outcome after cataract surgery depends on early clearance of visual axis, aphakic correction, and aggressive amblyopia treatment. Congenital cataracts ideally should be operated before three months of age.<sup>19</sup>

In our study cases presented before one year of age group was 24.4% including 18% unilateral and 24% bilateral. After one year of age 75.6% cases presented including 82% unilateral and 76% bilateral. As we observed in our study that small number of cases presented before one year of age and unilateral cataracts were less in numbers. The reason was the early appreciation of reduced vision in bilateral cases.

Management of congenital cataract depends on the etiology, degree of visual interference and laterality of cataract. The outcome of cataract surgery after congenital cataract is 20 times worse than developmental cataracts, especially for those cases which are operated after one year of age.<sup>22</sup> The visual system can get the opportunity to develop and mature after surgery while its progress remains halted by the development of cataract and visual system cannot develop at all in presence of dense congenital cataract<sup>23</sup>. That's why early cataract surgery is important in congenital cataract. Especially for severe bilateral cataracts which are causing significant obscuration of the visual axis, surgery is recommended as early as possible.

In developing countries delay in presentation and inadequate use of surgical services are the major causes of blindness secondary to congenital cataract<sup>24</sup>. The visual outcome depends upon the duration between onset of visual impairment and surgery, the shorter the duration, higher likelihood of good visual outcome. Early presentation is important for visual outcome, regardless the type of cataract. The reasons of excessive delay of presentation in our study population were few barriers to presentation, which include lack of awareness about the disease, difficult access to health services, or acceptance of services (lack of education).

In the language of clinical genetics, a consanguineous marriage is defined as a union between two individuals who are related to each other as second cousins or closer, with the inbreeding coefficient (F) equal or higher than 0.0156, where (F) is a measure of proportion of loci at which the offspring is expected to inherit identical gene copies from both partners. Among Arabs and south Indian communities the inbreeding coefficient (F) is highest where it reaches up to 0.125.<sup>25</sup>

In our study we observed statistically significant high rate of positive history of first cousin marriage and among the positive cases bilateral cataracts were more common as compare to unilateral cataracts. This high rate of observed consanguinity may be considered as one of the risk factor for congenital cataract. At the same time this aspect could not be overlooked that consanguinity is very common in Pakistani families and this relationship of consanguinity with congenital cataract as risk factor may be an incidental finding as number of our patients were limited.

A significant positive association has been consistently demonstrated between consanguinity and morbidity, although consanguinity associated blindness is less frequent but an increased rate of congenital cataracts has been reported in several populations.<sup>26</sup> One billion people are currently living in those countries where consanguineous marriages are customary, and among them, one in every three marriages is cousin marriage, with a deeply rooted social trend. Public awareness is rising about preventive measures of congenital disorders which has led to a trend that the number of couples who are seeking for preconception and premarital counseling on consanguinity are increasing gradually.<sup>27</sup>

The morphology of congenital cataracts is also very helpful in establishing their etiology and prognosis. Congenital cataract is inherited in all three Mendelian forms: autosomal dominant, autosomal recessive, and X-linked. In view of association of congenital cataract with consanguinity in literature, and the need to identify and delineate the variability in congenital cataract, the present study was undertaken to ascertain the role of consanguinity in congenital cataract patients.

The prospective study of laterality and consanguinity in congenital cataract has several limitations. Although we believe that all patients included in our study had congenital cataract not all patients were seen from time of birth. These cataracts showed many different patterns. The underlying and associated factors in patients with congenital cataract in this study were diverse. This complex pattern including variable differences between unilateral and bilateral cataracts has implication for further etiological research.

## CONCLUSION

Bilateral congenital cataract is a more common presentation as compare to unilateral congenital cataract. Consanguinity is an important risk factor for congenital cataract especially for bilateral cataracts.

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