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Frequency and ethiological frequency of congenital cataract associated with microphthalmia and postoperative visual results.

Frequência da microftalmia associada à catarata congênita, sua frequência etiológica e o resultado visual pós-cirúrgico

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ABSTRACT

Objective: *To determine the frequency of microphthalmia associated with congenital cataract and its etiological frequency. Compare the result of visual acuity in aphakic microphthalmus eyes, with the visual acuity result obtained in non microphthalmus eyes.* ***Methods:*** *Retrospective study of 76 patients with microphthalmia and congenital cataract, selected after analysis of 1050 medical records of patients seen in congenital cataract clinic of UNIFESP. All patients underwent complete ophthalmologic examination and microphthalmia determined by ultrasound biometry. Investigations were made to clarify the etiological cause. The postoperative visual outcome of Group I (with microphthalmia) was faced with the visual results obtained in Group II (control group without microphthalmia).* ***Results:*** *The anteroposterior diameter of microphthalmus eyes ranged from 13 to 21 mm. The etiological frequency of microphthalmia and congenital cataract was distributed as follows: infectious diseases (55.3%), idiopathic (26.3%), colobomas (7.9%), hereditary (6.6%), persistent hyperplastic vitreous (2.6%) and linked to the Lenz's syndrome (1.3%). The visual acuity in aphakic eyes that reached better view and or equal to 20/200 was 68.3%.* ***Conclusion:*** *The frequency of microphthalmia associated with congenital cataract was 7.23%. The etiological occurred more frequently in infectious disease (55.3%). The aphakics eyes with microphthalmia tend to have worse visual acuity results than the eyes without microphthalmia. If we consider the visual results same and above 20/200 as successful in this search, aphakic eyes with microphthalmia that hit these indices are 68.3%.*

Keywords: *Cataract/congenital; Congenital abnormalities; Aphakics , postcataract; Microphthalmus*

RESUMO

Objetivo: Determinar a frequência da microftalmia associada à catarata congênita e sua frequência etiológica. Comparar o resultado visual após a cirurgia da catarata congênita em olhos microftálmicos, com o resultado visual obtido em olhos não microftálmicos. **Método:** Estudo retrospectivo de 76 pacientes portadores de microftalmia e catarata congênita, selecionados após análise de 1050 prontuários dos pacientes atendidos no ambulatório de catarata congênita da UNIFESP. A microftalmia foi determinada pela ecobiometria ultrassonica. Exames oculares e complementares foram feitos para esclarecer a causa etiológica. O resultado visual pós- operatório do Grupo I (com microftalmia) foi confrontado com o resultado visual obtido no Grupo II (sem microftalmia). **Resultados:** O diâmetro ântero-posterior dos olhos microftálmicos variou de 13 à 21 mm. A frequência etiológica da catarata congênita associada aos olhos microftálmicos foi assim distribuída: doenças infecciosas (55,3%); seguidos de idiopáticas (26,3%), colobomas (7,9%), hereditárias (6,6%), persistência do vítreo primário hiperplásico (2,6%) e associada à síndrome de Lenz (1,3%) .A frequência da microftalmia foi de 7,23 %. 68,3% de olhos afácicos microftálmicos atingiram visão melhor e ou igual à 20/200. **Conclusão:** A frequência da microftalmia associada à catarata congênita foi de 7,23%. A maior frequência etiológica ocorreu nas doenças infecciosas (55,3%), Embora os olhos microftálmicos tenham tendência para piores resultados visuais quando comparados aos não microftálmicos, nesta pesquisa os olhos microftálmicos afácicos que atingiram visão melhor ou igual a 20/200 foram de 68,3%.

Descritores: Catarata/congênito; Anormalidades congênitas; Afacia pós- catarata; Microftalmia

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Introduction

Microphthalmia is a congenital malformation in which the volume of the ocular bulb is reduced. A microphthalmic eye is one whose axial diameter is less than 16 mm in the newborn.[1]

It can be classified according to the appearance of the ocular bulb. We call simple microphthalmia those eyes that are anatomically normal, except for the reduced antero-posterior diameter with high hypermetropia, and complex microphthalmia those eyes that, in addition to the reduced diameter, present either anterior segment malformation or anomalies in the posterior segment.[2-4]

Although all microphthalmos originate in embryonic life, they can differ in the time of onset in the embryogenesis, generating distinct anomalies between the affected eyes. Thus, we have three possibilities for the onset of microphthalmia during ocular formation. The first possibility arises when crystalline malformation occurs. The presence of the crystalline is a determinant factor for ocular growth in embryogenesis, and factors interfering in its formation can generate microphthalmic eyes early in pregnancy. A second possibility of microphthalmia onset is at the time of embryonic fissure closure, which is incomplete and causes coloboma to interfere with ocular growth. Finally, the third possibility of microphthalmia onset would be the persistence of the primary hyperplastic vitreous that occurs at later embryonic time.[5-8]

It is known that visual deprivation during the first months of the child's life caused by the presence of congenital cataract leads to an amblyopia of difficult reversion if not operated during the critical period of the development of the fixation reflex. When congenital cataract is associated to microphthalmia the treatment becomes even more difficult, discouraging many ophthalmologists to invest surgically in the visual recovery. [9]

This study aims to determine the frequency of microphthalmia associated to congenital cataract and its etiological frequency, and to evaluate the outcome of visual acuity after congenital cataract surgery in these microphthalmic eyes.

Methods

The patients included in this retrospective study came from Ambulatório de Catarata Congênita of Universidade Federal de São Paulo (UNIFESP). From December 1989 to December 1998, 1050 children were treated, and 76 of them had congenital cataract associated to unilateral or bilateral microphthalmia. Within this group of 76 patients, 38 children could not be operated because they had very small eyes, colobomas compromising the macula, and inoperable retinal detachment. However, the other 38 children (60 eyes) were operated on and composed Group I of this study. Group II control consisted of 31 children (51 eyes) of the same ambulatory who had unilateral or bilateral congenital cataract, but without microphthalmia or other ocular conditions. Group control underwent the same surgical techniques, had free visual axis and a minimum follow-up of 3 years.

All patients underwent complete ophthalmologic examination, as well as measurement of antero-posterior diameter by biometry (A-scan ultrasound echobiometry, Humphrey Ultrasound biometre model 820, Humphrey Instruments, USA). An average reading was obtained after 3 readings of the antero-posterior diameter of the eye, and compared with the Sampaolesi axial length frame for normal children.[10,11]

Microphthalmic eyes were those whose measurement was two standard deviations below the normal range for the age. The horizontal diameter of the cornea was measured by a millimeter ruler.

All children underwent preoperative exams and investigation of the possible etiological causes of cataract and microphthalmia, including serologies, metabolic exams and genetic search.

The surgical techniques used in the microphthalmic eyes were extra capsular facectomy without intraocular lens implantation and with small primary posterior capsuloctomy without anterior vitrectomy in 55 eyes, and lensectomy with anterior vitrectomy without intraocular lens implant in 5 eyes. Group II control underwent lensectomy without prior vitrectomy and without intraocular lens implant.

In the postoperative period, antibiotic and corticoid eyedrops, cyclopentolate, and dorzolamide hydrochloride hypotensive eyedrops were used for one month after surgery. In bilateral cataracts, eyeglasses were prescribed in the first week after surgery, and for monoculars gelatinous contact lens, daily use of diameter 10.5 mm in 3 options of base curves, ranging from 38.00 to 48.00 diopters (Solótica, São Paul), or eyeglasses to wear during the period of occlusive treatment. The visual acuity was evaluated with the best optical correction on Teller cards for nonverbal children, and on the Snellen table for verbal children. The longest segment was 10 years, and the shortest segment was 8 months.

The statistical methods applied were:

- Analysis of variance by Kruskal-Wallis stations to compare the values of the antero-posterior diameter of the eyes between the different etiologies. This analysis was supplemented, where necessary, by the multiple comparison test.

-Chi-square test or Fisher's exact test to compare Group I and Group II

-Non-parametric test for "k" Kruskal-Wallis independent samples supplemented, where necessary, by the multiple comparison test to evaluate possible differences in visual acuity in each one of Groups I and II in different etiologies, age group and laterality

Non-parametric test for two independent Mann-Whitney samples to compare Groups I and II for age and laterality in each etiology.

The level of rejection for the null hypothesis was always set at a value less than or equal to 0.05%.

Results

Of the 1,050 patients seen at the congenital cataract ambulatory of UNIFESP- Universidade Federal de São Paulo, 76 presented complex microphthalmia. The frequency of microphthalmia associated to congenital cataract was 7.23%.

Of the 76 patients with this condition, 40 (52.6%) were males and 36 (47.4%) were female.

Thirty-nine patients had microphthalmia and bilateral congenital cataract (78 eyes), and 37 unilateral patients, with a total of 115 eyes.

The horizontal diameter of the cornea ranged from 5 to 10 mm: 15 eyes (13%) from 5 to 6 mm; 62 eyes (54%) from 7 to 8 mm, and 38 eyes (33%) from 9 to 10 mm. The antero-posterior diameter of the ocular bulb of these same eyes ranged from 13 to 21 mm, with the following distribution: 3 eyes (2.7%) from 13 to 14 mm; 53 eyes (46%) from 15 to 17 mm; 49 eyes (42.6%) from

18 to 19 mm, and 10 eyes (8.7%) from 20 to 21 mm.

The frequency of etiological diagnoses of patients with microphthalmia and congenital cataract was: infectious diseases with 42 (55.3%) cases; 20 (26.3%) idiopathic cases; 6 cases (7.9%) associated to coloboma; heredity with 5 (6.6%) cases; persistence of the primary hyperplastic vitreous with 2 (2.6%) cases, and associated to syndromes with 1 (1.3%) case. The descending order of the average of antero-posterior diameters (mm) of the microphthalmic eyes was: hereditary 18.80; persistence of the primary hyperplastic vitreous 18.74; colobomas 18.08; idiopathies 18.05; Lenz's syndrome 17.82; rubella 17.77; toxoplasmosis 17.26, and cytomegalovirus 15.30.

Table 1 shows the antero-posterior diameters of microphthalmic eyes separated by etiology; the statistical analysis showed

Table 1

Values of the antero-posterior diameter (in millimeters) of the eyes with microphthalmia and congenital cataract in different etiologies

Rubella	Toxoplasmosis	Hereditary	Colobomas	Idiopathic	CMV	PVPH	Lenz Sind.
Average							
17,77	17,26	18,81	18,08	18,05	15,30	18,74	17,82

Analysis of variance by Kruskal-Wallis stations
(Rubella x Toxoplasmosis x Hereditary x Coloboma x Idiopathic x CMV x PVPH x Syndrome)

H calc=14.27* H crit.= 14.07

Multiple comparisons

CMV lower hered and PHPV

CMV- Citomegalovirus PHPV - Persistent hyperplastic primary vitreous

Table 2

Frequency of patients with microphthalmia and congenital cataract (Group I) and congenital cataract without microphthalmia (Group II), according to the etiology. In parentheses the percentage of participation of each Group

Etiologies	Group I		Group II		Total	
	N	%	N	%	N	%
Rubella	17	(44,73)	8	(25,80)	25	(36,23)
Toxoplasmosis	6	(15,78)	0	(0,0)	6	(8,70)
Syndrome	1	(2,63)	1	(3,23)	2	(2,90)
Hereditary	4	(10,53)	6	(19,35)	10	(14,49)
Idiopathic	10	(26,32)	16	(51,61)	26	(37,68)
Total	**38**	**(100,0)**	**31**	**(100,00)**	**69**	**(100,0)**

Table 3

Frequency of patients with microphthalmia and congenital cataract (Group I) and congenital cataract without microphthalmia (Group II), according to the morphological types of cataract. In parentheses the percentage of participation of morphological types in each Group

Morphological types of cataract	Group I		Group II	
	N	%	N	%
Lamellar	10	(16,67)	8	(15,69)
Nuclear	10	(16,67)	6	(11,76)
Polar	4	(6,67)	4	(7,84)
Total	36	(60,00)	33	(64,71)
Total of eyes	**60**	**(100,00)**	**51**	**(100,00)**

Chi-square test

X^2 calc=0,63 N. S. X^2 critical= 7,82

that the average of the antero-posterior diameter of the eyes with cytomegalovirus (15.30 mm) was significantly lower than the average of the hereditary microphthalmia (18.80 mm) and the eyes with persistence of the primary hyperplastic vitreous (18.74 mm).

Tables 2, 3 and 4 present the characteristics of patients with microphthalmia and congenital cataract (Group I), comparing them to those with congenital cataract without microphthalmia (Group II), according to the etiology, morphological type of congenital cataract, and the type of strabismus, respectively.

Table 5 compares Groups I and II that underwent congenital cataract surgery according to the antero-posterior diameter of the eye and the horizontal diameter of the cornea. In Group I, the antero-posterior diameter was significantly lower (average 18.18 mm) than the eyes in Group II (average 21.80 mm). The horizontal diameter of the corneas of the operated microphthalmic eyes was also statistically smaller (average 8.15 mm) than the non-microphalic eyes (average 10.31 mm).

Table 6 shows the results of the bilateral aphakic eyes of the Group I versus Group II operated until the fourth month of life in the different etiologies. Similar results were obtained between the two Groups for rubella, heredity and idiopathic etiology. Group II

Table 4

Frequency of patients with microphthalmia and congenital cataract (Group I) and congenital cataract without microphthalmia (Group II), according to the types of strabismus. In parentheses the percentage of participation of types of strabismus in each Group

Types of strabismus	Group I		Group II	
	N	%	N	%
Esotropia	20	(52,63)	9	(29,03)
Exotropia	10	(26,31)	9	(29,03)
Centered reflection	8	(21,05)	13	(41,93)
Total of cases	**38**	**(100,00)**	**31**	**(100,00)**

Chi square test

X^2 calc= 4,75 N. S. X^2 critical=5,99

Table 5

Antero-posterior (AP) diameter of the eye and horizontal (H) diameter of the cornea (in millimeters) of patients with microphthalmia and congenital cataract (Group I) and with congenital cataract without microphthalmia (Group II)

Diameter	AP of the eye (mm)		H diameter of the Cornea (mm)	
	Group I	Group II	Group I	Group II
Average	18,18	21,80	8,15	10,31

Mann- Whitney Test

AP Diameter of the eye
Z calc= 6,492*

Horizontal diameter of the cornea
z calc= 9,155*

Table 6

Comparison of the average visual acuities of microphthalmic, bilateral aphakic eyes (Group I) and non-microphthalmic eyes (Group II) operated until the fourth month of age in different etiologies

	Group I	Group II	Mann Whitney VC	U	SIG.
Rubella	20/166	20/97	8,0	14,0	NS
Hereditary	20/305	20/315	0,0	4,0	NS
Idiopathic	20/186	20/78	2,0	3,0	NS
Syndrome	20/2000	20/85	0,0*	0,0	*

VC= value calculated U= critical value SIG.= significance NS= non-significant

presented a significantly better result in Down's Syndrome than in Lenz's syndrome, which was microphalic. There is a tendency for the best visual result of rubella patients in Group II when compared to Group I, suggesting a worse visual result in microphthalmic eyes. This trend was also observed in the idiopathic etiology, as a worse visual result of Group I (microphthalmic) was observed compared to Group II (non-microphthalmic).

Table 7, compares the results of the visual acuities of microphthalmic eyes in Group I and non-microphthalmic in Group II bilaterally operated until the fourth month of age, showing statistically similar visual results. Heritable etiologies and the other etiologies could not be compared due to the small number of the sample.

Table 8 presents the visual results of aphasic eyes in Group I versus Group II operated after 4 months of age. Due to the very large variability of results, they could not be analyzed. Regarding rubella, Group II (non-microphthalmic) showed a tendency to present better results than Group I (microphthalmic).

Table 9 presents the percentage of apophysic microphalic eyes in the various etiologies that reached visual acuity greater or equal to 20/200, and which was considered as a good result of visual acuity in these microphthalmic eyes. The greatest number of good results occurred in rubella (82.7%). In hereditary, the index was (75%), in idiopathic (58.3%), and in toxoplasmosis (44.4%).

Discussion

It is believed that the frequency of microphthalmia associated to congenital cataract (7.23%) found in this study is due to Ambulatório de Catarata Congênita of UNIFESP be a specialty and reference service for treatment. To date, no data on the incidence and/or prevalence of microphthalmia associated to congenital cataract has been known in other medical services

Table 7

Comparison of the average visual acuities of microphthalmic, bilateral aphakic eyes (Group I) versus non-microphthalmic eyes (Group II) operated until the fourth month of age in different etiologies

	Group I	Group II	Mann VC	Whitney U	SIG.
Rubéola	20/145	20/180	17,0	36,0	NS
Hereditária	20/92	20/173	5,0	10,0	NS

VC= value calculated U= critical value SIG.= significance NS= non-significant

Table 8

Comparison of the average visual acuities of microphthalmic, unilateral aphakic eyes (Group I) versus non-microphthalmic eyes (Group II) in patients operated until the fourth month of age in different etiologies

	Group I	Group II	Mann VC	Whitney U	SIG.
Rubella	20/2598	20/72	0,0	0,5	NS
Idiopathic	20/350	20/778	4,0	14,0	NS

VC= value calculated U= critical value SIG.= significance NS= non-significant

Table 9

Percentage (%) of aphakic microphalic eyes that reached visual acuity greater or equal to 20/200 in the various etiologies

Etiologies	Nº of patients	Nº of eyes operated	Nº of eyes with vision >20/200	% in each etiology
Rubella	17	29	24	82,7
Toxoplasmosis	6	9	4	44,4
Hereditaries	4	8	6	75,0
Idiopathics	10	12	7	58,3
Syndrome	1	2	0	0
Total	38	60	41	68,3

in Brazil. In this investigation, infectious diseases present a very high frequency (55.3%) in the etiology of congenital cataract associated to microphthalmia, unlike developed countries such as the United States, where the frequency of congenital cataract with microphthalmia is mainly due to heritable causes.[12-14]

The difference between microphthalmic eyes with involvement of the posterior segment or the anterior segment gives distinct characteristics that will interfere with the conduct, the choice of the surgical technique and the visual recovery. Therefore, it is very important to know the etiologic cause of microphthalmia and the degree of involvement of other ocular structures.

Tables 2 and 3 show Groups I (with microphthalmia) and II (without microphthalmia) regarding the etiologies and morphological type of the congenital cataract. Note that in both groups there was a greater number of total cataracts that are more amblyopic, which would determine a worse visual result.[15]

Diagnosing microphthalmia only by the presence of microcornea or high hyperopia is a mistake that confuses the definition itself. The ocular echobiomentry is important to make the differential diagnosis between microcornea and microphthalmia, because the presence of microcornea is not necessarily accompanied by microphthalmia, and the eye may have a normal antero-posterior diameter or even increased for the age group. Table 4 presents the measurements of the horizontal diameters of the corneas of the eyes of Groups I and II, and it shows that the average of the corneas of the microphthalmic eyes was 2.16 mm, smaller than the average of the non-microphthalmic ones. The average antero-posterior diameter of the microphthalmic eyes was 3.62 mm, smaller than the non-microphalic ones. These data are consistent with the literature, in which the antero-posterior diameter of the microphthalmic eye is on average 3.1 ± 0.7 mm smaller than that of the non-microphthalmic eye.[3, 4]

Microphthalmic eyes with congenital cataracts are usually accompanied by microcorneas, probably because in the embryogenesis the crystalline early induces the formation of the anterior epithelium of the cornea. The delay or malformation of the crystalline, in addition to causing the onset of the cataract, induces the formation of microcorneas and the development of sclera in place of the cornea (sclerocornea).[5]

As the incidence of microphthalmia in the population is low, from 0.22/1000 to 1/2,000 live births, and the incidence of microphthalmia associated to congenital cataract (although unknown) should be even lower, it is difficult to have prospective studies.[14,16]

For this reason, this 10-year retrospective study was performed, and although the number of complex microphthalmic eyes was sufficient to verify certain aspects such as the etiological frequency, the antero-posterior diameter of the eye, and the horizontal diameter of the cornea, it was difficult to statistically evaluate the visual result, because there was reduction of the sample when the operated eyes were separated in the several variables that interfere in the final visual acuity after the surgery of congenital cataract.[17]

Another aspect that interfered in the visual result was the fact that children with microphthalmia whose cataracts with a greater chance of a better postoperative visual prognosis arrived after the 4 months of age at the specialized service because they were discouraged to have the congenital cataract surgery of the microphthalmic eye. We know that after 4 months, especially in unilateral cases, the recovery of amblyopia becomes difficult due to the closing of the critical period for the development of the fixation reflex.[18-24]

Table 6, 7 and 8 present the average visual acuities obtained in the different etiologies and regarding laterality in Groups I and Groups II before and after the 4 months of age. Due to the small sample of unilateral cases, it was difficult to statistically evaluate the visual outcome.

It is known that children with bilateral total congenital cataracts who only perceive the existence of light have serious limitations to their development and condition of life. In this study, children with microphthalmia and congenital cataracts who had a postoperative visual result equal to or better than 20/200 in both eyes were considered successful because they had a radical change in their quality of life. They became independent in the activities of their daily tasks and had a satisfactory development, being able to study with simple adaptations to overcome their visual limitations, like sitting in the first row of the class, using pencils and pens with thick tips, and having their lessons with bigger letters. Some have benefited from close additions of more than +3.00 DE.

Table 9 shows the operated microphthalmic eyes, and the majority (68.3%) had a visual result equal to or better than 20/200. Even in monocular cases with 20/200, the child gained visual field.

We emphasize that the microphthalmic eyes are not all of the same nature, and those eyes presenting the anterior segment compromised, i.e., with microcornea, cataract and aniridia, are distinct from the eyes with microphthalmia by alterations in the posterior segment, such as colobomas and persistence of the primary hyperplastic vitreous. Therefore, we must evaluate the surgical limitations of congenital cataract in microphthalmic eyes and their possibilities of visual recovery.

Conclusion

The frequency of microphthalmia associated to congenital cataract was 7.23% in the ambulatory of congenital cataract of UNIFESP, with infectious diseases being the most frequent etiology. Although the microphthalmic eyes operated of congenital cataract tend to have worse visual acuity results than non-microphalic eyes, and if we consider visual results equal to and above 20/200 as successful, in the present study the aphthic microphalic eyes reaching these indices in the various etiologies were 68.3%.

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