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Ophthalmologic Manifestations in Children with Down Syndrome Attending a Child Development Centre in Bangladesh

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ABSTRACT

Background: *This study was conducted to establish the background of importance of the routine ophthalmological check-up in children with Down Syndrome (DS) in this region which is very rarely done in Bangladesh.*

Objectives: *To find out the oculo-visual manifestations of Down Syndrome in children who undergo follow-up regularly at a Child Development Centre in Bangladesh, which has significant role for overall development and rehabilitation.*

Methods: *This observational study was done to examine the ocular status of 54 children with Down syndrome who attended the Child Development Centre, Institute of Child and Mother Health (ICMH), Dhaka. After confirming the diagnosis by clinical features and karyotyping, a complete ophthalmic examination was performed by a single specialist pediatric ophthalmologist in two tertiary eye care centers (he served in both) on an out-patient basis. Records were kept and analyzed later by SSSS version 22.0.*

Results: *Examination of the participants revealed that no single case was free of eye problem. About 67% of the participants had significant refractive error. Most of them had hypermetropia (40.7%), followed by astigmatism (16.6%) and myopia (9.3%). Other ocular findings were the following: lacrimal system obstruction (38.9%), strabismus (18.5%), blepharitis (18.5%), nystagmus (16.6%), cataract (7.4%), amblyopia (7.4%), chalazion (3.7%), ptosis (3.7%) and retinal abnormalities (1.9%).*

Conclusion: *Identification, close observation and management of oculo-visual disorders by a pediatric ophthalmologist is mandatory for children with Down syndrome as a whole. The early and correct diagnosis of the ocular abnormalities will help to improve their visual, functional, psychological, and social aspects.*

Keywords
Down syndrome, Eye problems in children, Refractive error.

Introduction
Down syndrome is one of the most common genetic diseases. The phenotype of Down syndrome is caused by a triplication of all or part of chromosome 21. This syndrome is found in all races, nationalities, and socioeconomic strata [1]. Worldwide Down syndrome occurs in approximately 1 in 700 births. Advanced maternal age was identified as the major risk factor for it. Subsequently, maternal parity and genetic predisposition for chromosomal nondisjunction were established as additional independent risk factors [2-5].

Children with Down syndrome are at increased risk for a variety of ophthalmic disorders. The ocular features of the disorder, first

described by Langdon Down in the mid-19th century, include epicanthus, obliquely positioned and narrow palpebral fissures and hypertelorism. Multiple publications [6-9] have expanded the ocular phenotype to include common pediatric conditions such as blepharoconjunctivitis/conjunctivitis, nasolacrimal duct obstruction, significant refractive errors and strabismus. Less common diseases, such as glaucoma, cataract, optic nerve hypoplasia or dysplasia, nystagmus, amblyopia, keratoconus and hypotelorism have also been reported. Visual acuity of children with Down syndrome is generally less than other children of same age.

The reported prevalence of ophthalmic disorders in Down syndrome patients ranges from 46% to 100% [10]. The incidence of associated eye disease increases with age. Children with Down syndrome under 1-year-old with ophthalmic disorders have a rise from 38% to 80% in 5-7 years' age that need monitoring or intervention [11]. So, close observation and management of ocular disorders by a pediatric ophthalmologist is necessary, as visual impairment can be a significant impediment to quality of life. Eye care for children with Down syndrome is still inadequate in the LMIC countries. Despite several studies being conducted, none assessed the ocular features of children with Down Syndrome in the Bangladeshi population. So identification of unique ocular anomalies in these children is important.

Methodology

This cross-sectional, non-interventional observational study was carried out from July to December 2022 on 54 children (Age 0-18 years) with Down syndrome who attended the CDC of the Institute of Child and Mother Health (ICMH), Dhaka. After taking informed consent from the parents/guardian, a pre-tested questionnaire was filled up that included the participant's history and relevant clinical examination. A complete ophthalmic examination was performed on each patient by a single specialist pediatric ophthalmologist in two tertiary eye care centers (he served in both) on an out-patient basis. The techniques used were: visual acuity (Snellen illiterate chart and Lea picture charts, CSM), Slit Lamp examination, Refraction (Streak retinoscopy with cycloplegia), ocular alignment (cover test), near point of convergence (pen and ruler), external examinations and Dilated fundoscopy. The findings of the examinations were noted down. Statistical Package for the Social Sciences (SPSS) version 22.0 for Windows was used to analyze the data.

Results

A total of 54 children (27 males, 27 females) with Down syndrome (mean age: 6.3 years; range: 7 months to 17 years) underwent eye examinations in this study. One-third (33%) of the mothers were >40 years of age and one-third of the fathers were >50 years of age. Here, consanguinity was found in 29% and around 9% children had a family history of Down syndrome. More than two-thirds (68%) of mothers were multiparous. Karyotyping showed most (90%) had nondisjunction of chromosome 21. The majority of participants came from mid to high income groups (Table 1).

Table 1: Baseline characteristics of the study participants (n=54).

Background Characteristics	Number	(%)
Gender		
Male	27	50%
Female	27	50%
Age group		
≤1 yr	5	9.30%
>1-5 yrs	32	59.30%
>5-10 yrs	11	20.40%
≥10 yrs	6	11.10%
Maternal age		
20-30 yrs	13	24.10%
>30-40 yrs	23	42.60%
>40-50 yrs	16	29.60%
>50 yrs	2	3.70%
Paternal age		5.60%
20-30 yrs	3	33.30%
>30-40 yrs	18	29.60%
>40-50 yrs	16	31.50%
>50 yrs	17	
Family h/o Down syndrome		
Yes	5	9.30%
No	49	90.70%
Consanguinity		
Yes	4	7.40%
No	50	92.60%
Maternal parity		
Single	17	31.50%
Multiple	37	68.50%
Karyotyping		
Nondisjunction (47, XX+21/47, XY+21)	49	90.70%
Translocation (t, 21/21)	4	7.40%
Mosaic variety	1	1.90%
SE status		
Low income (<10,000 tk/mo)	4	7.40%
Middle income (10,000-30,000 tk/mo)	24	44.40%
Higher income (>30,000 tk/mo)	26	48.14%

The ocular features are summarized in Table 2. Twenty-four children (44.4%) had at least one abnormal ocular finding identified at this single visit, the rest were diagnosed with multiple problems. Refractive errors were the commonest (66.6%) finding, followed by Lacrimal system obstruction (38.9%), Strabismus or squint (18.5%), Blepharoconjunctivitis (18.5%), Nystagmus (16.6%), infantile or juvenile Cataract (7.4%), Chalazion and Ptosis (each 3.7%).

Visual acuity could not be checked conventionally in eleven (11) subjects due to poor response/ poor understanding of the test, but four (04) of the participants seemed to be amblyopic (7.4%). Retinovascular anomaly was found in only one case (1.9%). Brushfield spots and keratoconus was not found in any of these study participants.

In our study, we found that myopia was more prevalent in subjects under 5 years of age (05 cases among 36 cases of refractive error,

9.3%). Hypermetropia was found in all age groups (total 22 cases; 40.7%) and it seemed to increase in subjects around 5 years and above. Astigmatism (total 16.6%) was seen more in the children aged over 7 years (Figure 1).

Table 2: Ophthalmologic Manifestations in children with Down syndrome.

Ophthalmologic Manifestations* at examination	Distribution of participants by age				
	≤1 yr (n = 05)	>1-5 yrs (n = 32)	>5-10 yrs (n = 11)	>10 yrs (n = 06)	Total n (%)
Refractive errors	3	15	12	6	36(66.6%)
Lacrimal system obstruction	5	10	3	3	21(38.9%)
Strabismus/squint	2	3	2	3	10(18.5%)
Blepharoconjunctivitis	3	5	2	0	10(18.5%)
Nystagmus	0	5	2	2	9(16.6%)
Cataract	0	3	0	1	4(7.4%)
Amblyopia	0	2	2	0	4(7.4%)
Chalazion	0	2	0	0	2(3.7%)
Ptosis	0	2	0	0	2(3.7%)
Retinovascular anomaly	1	0	0	0	1(1.9%)

*multiple response

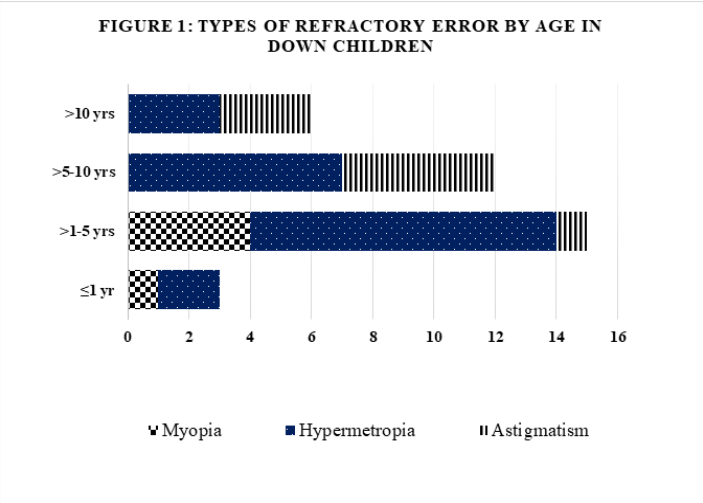

Figure 1: Bar graph showing types of refractive errors by age in children with Down Syndrome.

Discussion

Children with Down syndrome often do not complain about their vision problems, either because they don't identify the problem or they can't communicate the problem well enough. So there is always a chance of increased prevalence of amblyopia, due to uncorrected refractive error and squint with Down's syndrome. It is of paramount importance that these children undergo ophthalmologic examination early in life, to be followed up regularly and treated appropriately. This study focused on the oculo-visual findings of young children with Down syndrome that would not only support the provision of multidisciplinary services (e.g., eye screening) but also make them mandatorily available. This, in the long run, will reduce the risk of less successful development and promote their optimal outcome.

Children with Down syndrome are very prone to Blepharitis, an inflammation of the eye lash follicles that causes debris to collect along the margins of the eyelids and can cause irritation of the eyes. It can be a source of discomfort and itching, which may be a reason for not co-operating with activity and it can result in scarring of the follicles and in-growing eyelashes. Blepharitis responds very well to simple treatment and parents should be encouraged to learn about this and can help them so they are symptom free comfortable and participate in activities.

There was a high prevalence of strabismus among children with Down syndrome. In normally developing children, strabismus is associated with hypermetropia. The classic assumption has always been that normally developing children with high hypermetropia over-accommodate to obtain a clear image. This excessive accommodation results in a high degree of accommodative-convergence and is believed to cause esotropia/internal squint. However, this hypothesis does not explain the findings in children with Down syndrome. The high prevalence of strabismus in children with Down syndrome cannot be attributed to the presence of hypermetropia. The sign or magnitude of the refractive error appears irrelevant.

These DS children tend to under-accommodate by quite a large amount. For that reason, they focus very poorly at near. This is consistent for any Down's child, and persists even when the child wears the glasses to correct his/her sight. It may be explained this way: whenever they try to do any near work (i.e. reading or writing), it becomes difficult for them because it is out of focus but they cannot realize it by themselves; and most of the times parents also fail to identify the problem. This is one important reason why many children with DS are not interested at any near activity. Though the exact reason for the poor focusing is yet to be fully explained, however, it is observed that their focusing improves with bifocal spectacles [12]. Children with Down's syndrome have smaller noses and a shorter distance from ears to face than normal children, and so they have difficulty in fitting the glasses. Not addressing this issue many a times is the cause of not wearing the glasses regularly. It is important to adjust a frame, replace pads if needed and shorten the sides etc., so that glasses fit properly.

These ocular abnormalities in subjects afflicted with DS have been widely reported and are very well known and our data revealed similar findings to those in the published literature. Table 3 showed a comparison of ocular findings in previous studies with our findings in children with Down Syndrome. The incidence of strabismus in our study was 18.5% and was in agreement with the previous reports in the given table (ranging 20%–40%). The rates of nystagmus, blepharoconjunctivitis and lens opacities/ cataract were also comparable among different countries and times reported in those studies.

The percentage of subjects with lacrimal system obstruction was higher (38.9%) than that reported in previous studies (range: 05%–30%). On the other hand, retino-vascular anomaly was much less reported (1.9%) in our participants than that of those studies. A good portion (55.6%) of participants had the combination of strabismus, nystagmus, lacrimal system obstruction and refractive errors.

Numerous studies found the increased frequency of refractive errors and lower visual acuity in subjects with DS. It was widely reported that hypermetropia was more common than myopia. Hypermetropia in Down Syndrome children has been found to range from 4% - 65% [10,16]. Although the prevalence of myopia ranges from 8% - 41% in Down children, it has not been found to be significantly higher than in controls [10]. In the present study, hypermetropia was observed in the majority of the subjects (40.7%), while myopia was less frequently observed (9.3%) and only found in infants and younger children (<1-5 yrs). This variation might be related to the small sample size.

Infants with Down syndrome generally have "with the rule" astigmatism. The type of astigmatism often changes, with 26% - 55% of Down children having an oblique type of astigmatism [17,18]. The development of oblique astigmatism has been proposed to be caused by the oblique, up slanting palpebral fissures in Down patients [19] as the axis of astigmatism has been correlated with the angle of the palpebral fissure. A cross sectional population study of Down syndrome children by age got the prevalence of oblique astigmatism low in infancy, then 7.1% of DS children with astigmatism at one year of age, which raised to 45% of 15-year-old DS children with astigmatism [20]. With increasing age, astigmatism appeared to be increased also in our study with DS children. The percentages of astigmatism in our study did not agree with most of that in previous studies mentioned in Table 3. This varying rate might be related to the differences in age distribution and diagnostic criteria.

Absolute normal visual acuity is nearly impossible for one with Down syndrome. Again, these children are mentally retarded to some extent (mild to severe), and an additional ophthalmological cause of handicap or sensory impairment may further limit their overall functioning and as a result, may refrain the children from participating in significant learning processes.

Conclusion

Vision has a cardinal role in a child's overall development but eye care for children with Down syndrome is still inadequate in our country. The results of this study suggested that children having Down syndrome are at a greater risk for a variety of visual impairments. The early detection and correction/management of these ocular disorders with regular follow-up should subsequently improve the capabilities and the quality of life of this developmentally challenged population.

Limitations of the study

1) The small sample size and poor response/inattention from some of the participants may affect the applicability of the study findings.
2) Not being a follow-up study, this could not illustrate changes in the course of visual acuity and refraction related to patient-age in the subjects with DS.
3) The use of visual evoked responses could have been beneficial in further categorizing some findings, but the facility was not available at the centre at the time the study was conducted.

Recommendations

1) Pediatric ophthalmologists must be considered part of the multidisciplinary team in CDC of Bangladesh for all Down syndrome children's care who can greatly assist in reaching their full health and developmental potential.
2) All children with Down syndrome should be evaluated for strabismus, cataracts and nystagmus along with visual acuity testing within the first 6 months of life.
3) They should be followed up annually until age 5 with evaluation for refractive error, strabismus, and later to evaluate for the onset of new ophthalmic conditions which could result in amblyopia.

Table 3: Comparison of ocular findings in previous studies with our findings in children with Down Syndrome.

Variables	Present study (2020)	Tomoko, et al. [12]	Nanda L, et al. [13]	Wong and Ho [14]	Da Cunha, et al. [15]	Caputo, et al. [8]
Country	Bangladesh	Japan	India	Hong Kong	Brazil	USA
Number of patient	54	222	64	140	152	187
Range of age	7 month-17 yr	3 month-19 yr	18 month-14 yr	0-13 yr	0-18 yr	0-26 yr
Refractive error	36(66.6%)	-	35(54%)	137(98%)	149(98%)	122(65%)
Hypermetropia	22(40.7%)	~80%	08(12%)	42(30%)	39(26%)	39(21%)
Myopia	05(9.3%)	10-40%	05(7%)	12(8.6%)	19(13%)	42(22%)
Astigmatism	09(16.6%)	71-85%	22(34.4%)	08(7%)	91(60%)	41(21%)
Strabismus	10(18.5%)	87(39.2%)	21(32%)	28(20%)	57(38%)	107(57%)
Nystagmus	09(16.6%)	46(20.7%)	02(3%)	15(11%)	28(18%)	55(29%)
Blepharoconjunctivitis	10(18.5%)	29 (13.1%)	12(18%)	08(7%)	45(30%)	-
Lens opacities/cataract	04(7.4%)	25 (11.3%)	05(7%)	04(2.9%)	20(13%)	21(11%)
Lacrimal system obstruction	21(38.9%)	12 (5.4%)	08(12%)	-	46(30%)	09(5%)
Retinovascular anomaly	01(1.9%)	13(5.9%)	06(9%)	16(14%)	42(28%)	-

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