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OCULAR MANIFESTATION OF DOWN SYNDROME IN INDIAN CHILDREN

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Conflicts of Interest: Nil

ABSTRACT

Introduction: Manifestation in Down syndrome children has strong racial variation. The aim is to study ocular manifestation in children with Down Syndrom

Patients and Methods: This observational study was done in Outpatient clinic of Department of Pediatrics and Ophthalmology, Government Medical College Jammu from August 2017 to July 2018. Children between the age group of 6 months to 14 years with confirmed diagnosis of Down syndrome on karyotyping were includes in study. A total of 68 patients (42 boys and 26 girls) of Down syndrome were includes in study. The mean and median age was 5.8 years and 4.5 years respectively. Detailed ophthalmological examination was done in each case

Results and Conclusion: Upward slant and epicanthal folds were the two commonest finding at 94% and 80% repectively. Emmetropia was seen in 47% children, whereas hyperopia, myopia and astigmatism were seen in 27.9%, 25% and 35.2% respectively. No Brushfield's spots, focal retinal pigment hyperplasia, glaucoma, corneal opacities or keratoconus is seen.

Key words: Down syndrome, upward slant, epicanthal folds, epiblepharon.

Introduction

Down syndrome is most common cause of intellectual disability due to chromosomal disorder. The Center for Disease Control (CDC) data shows that the incidence of Down syndrome is 14.47 per 10,000 live births.(1) The studies in Caucasian and oriental population have shown that Down syndrome is associated with various ocular abnormalities which includes abnormalities of the eyelid, cornea, iris, lens, retina, and optic disc, as well as ametropia, amblyopia, strabismus, and nystagmus.(2-17) Study by Kim et al has shown that the Asian children with Down syndrome demonstrate unreported, high incidence of epiblepharon, the high rate of exotropia, and essentially no notable Brushfield spots.(18) Whether similar findings exist in Indian children is not known. This study was done to determine the ocular manifestation in Indian children

Patients and Methodology

This observational study was done in Outpatient clinic of Department of Pediatrics and Ophthalmology, Government Medical College Jammu from August 2017 to July 2018. Children between the age group of 6 months to 14 years with confirmed diagnosis of Down syndrome on karyotyping were includes in study. A total of 68 patients (42 boys and 26 girls) of Down syndrome were includes in study. The mean and median age was 5.8 years and 4.5 years respectively. Children presenting in Pediatric outpatient department were sent to ophthalmic outpatient department for detailed ophthalmological examination. Children were tested for visual acuity, slit lamp biomicroscopy,

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ocular motility using alternate cover uncover test with/without prism, cycloplegic refraction and ophthalmolscopy. The palpebral fissure was evaluated by placing a clear plastic straight-edged ruler across the bridge of the nose at the level of both inner canthi, and measuring the vertical displacement of the outer canthi. Upward slanting fissures were defined as 2 mm or more above the horizontal line. Cycloplegic refraction was done 45 min after three to five instillations of one drop of cyclopentolate 1%. Emmetropia was defined as a refractive error between -0.75 diopter (D) and+0.75 D spherical equivalent. Myopia was defined as less than-0.75 D spherical equivalent, and hyperopia was defined as more than +0.75 D spherical equivalent. Astigmatism was defined as refractive error more than±0.75 D of the cylinder.(18)

Results

The ocular manifestations are summarized in Table 1. Upward slant was present in almost 94% children. Epicanthal fold was the second commonest finding. Refractive status of patients is summarized in

Table **2**. Emmetropia was seen in 47% children, whereas hyperopia, myopia and astigmatism were seen in 27.9%, 25% and 35.2% respectively. Strabismus was seen in 16 (23%) children; esodeviation in 10 (14.7%) and exodeviation in 6 (8.8%). Nystagmus was seen in one child. Nystagmus was horizontal-pendular type. No Brushfield's spots, focal retinal pigment hyperplasia, glaucoma, corneal opacities or keratoconus is seen.

Table 1: Ocular manifestation of Down syndrome in 68 children

Ocular abnormality	Number (%)
Slanting fissure	64 (94)
Epicanthus	55 (80)
Epiblepharon	39 (57)
Astigmatism	24 (35)
Hyperopia	19 (28)
Myopia	17 (25)
Strabismus	16 (23)
Nasolacrimal duct obstruction	10 (15)
Blephraroconjuctivitis	9 (13)
No. of retinal vessels ≥18	6 (9)
Nystagmus	1 (1)
Lens opacities	1 (1)
Focal retinal pigment epithalial hyperplasi	0
Glaucoma	0
Cornal opacities	0

Table 2: Refractive status of Children with Down syndrome

Refractive status	N=68 (%)
Emmetropia (-0.75D to +0.75D)	32 (47)
Hyperopia: +1D to +2.75D +3D to +5.75D ≥+6D	19 (27.9) 12 (17.6) 6 (8.8) 1 (1.4)
Myopia -1D to -2.75D -3D to -5.75D ≥-6D	17 (25) 11 (16.1) 5 (7.3) 1 (1.4)
Astigmatism ±1D to ±2.75D ≥±3D	24 (35.2) 20 (29.4) 4 (5.8)

Table 3: Comparison of various studies on ocular manifestation of Down Syndrome

Parameter	Present study	Kim et al(18)	Wong and Ho(17)	Da Cuhna et al (5)	Berk et al (2)	Caputo et al (3)	Shapiro et al (13)
Number of the patients	68	123	140	152	55	187	53
Nationality	Indian	Korea	Hong Kong	Brazil	Turkey	US	US
Mean Age (years)	5.8	6.5	3.74	-	7.2	5.8	17.4
Upaward Slant (%)	64 (94)	78(63)	140	125 (82)	-	-	47 (89)
Epicanthus	55 (80)	75 (61)	140	92 (61)	13 (24)	-	-
Epiblepharon (%)	39 (57)	66 (54)	-	-	-	-	-
Refractive errors (%)	36 (52.9)	104 (85)	137 (98)	149 (98)	60	122 (65)	35
Hyperopia	19	35	42	39	29	39	17
Myopia	17	31	12	19	7	42	18
Astigmatism	24	38	8	91	24	41	12
Strabismus (%)	16 (23)	31 (25)	28 (20)	57 (38)	12 (22)	107 (57)	23 (43)
Esotropia	10	18	-	51	11	97	22
Exotropia	6	13	-	0	1	4	

Nystagmus (%)	1 (1.4)	27 (22)	15 (11)	28 (18)	7 (13)	55 (29)	5 (9)
Nasolacrimal duct blockage (%)	10 (15)	21 (17)	-	46 (30)	12 (22)	9 (5)	-
Blepharitis / Conjuctivitis	9 (13)	20 (16)	8 (7)	45 (30)	19 (35)	-	25 (47)
Number of Retinal vessels ≥18 (%)	6 (9)	16 (13)	16	42	21 (38)	-	-
Lens opacity (%)	1 (1.4)	4 (3)	4	20 (13)	11 (20)	21 (11)	7 (13)
Focal RPE hyperplasia (%)	0	2 (2)	-	-	-	-	-
Glaucoma (%)	0	1 (0.8)	1	-	-	10 (50)	-
Corneal Opacities (%)	0	1 (0.8)	-	-	-	-	-
Keratoconus (%)	0	0	-	-	-	-	8 (15)
Brsuhfield's spot (%)	0	0	0	79 (52)	20 (36)	-	43 (81)

Discussion

Table 3 summarizes the difference in manifestation in various studies. High prevalence of upward slant is seen in our study as compared to study by Kim et al.(18) Similar, high prevalence of upward slant was observerd in study by Da Cunha et al (5) and Shapiro et al (13). The second commonest finding of epicanthal fold was seen in 80% children. Other studies have reported variable prevalence of upward slant. Some have reported prevalence to as low as 9% (4), while other have seen it in 100% children.(16)

The prevalence of epiblepharon was high in our study (57); almost similar to study by Kim et al at 54%. (18) the preavalenc of epiblepharon is lower in causcasion population. Refractive errors were seen in 52.9% children in our study. The prevalence is high in other study 65-98% (see Table 3). The reason for this difference is not known.

The prevalence of strabismus was 23% in our study which is similar to other studies (see Table 3). The proportion of exotropia was 37.5% which was similar to study by Kim et al. However, other studies have reported the proportion of exotropia around 0-19 %.(Table 3)(2-5, 7, 8, 10-14) This could be reflection of the fact that Asian race has overall prevalence of exotropia in general population as compared to Caucasion and Africans.(19)

Nystagmus was seen in only one child in our our study. This was in contrast to findings in other studies which have reported the prevalence of 4-30%.(2, 3, 5, 8, 14) Similarly, lens opacity was seen in only one child. Studies by Kim et al(18) and Roizen et al (12) have reported the prevalence of 4% and 5% respectively, whereas other studies have seen high prevalence of 11-86% (Table 3)(2, 3, 5-7, 9-13, 15, 16) The could be lower mean age of our children. The lens opacity is a developmental phenomenon and usually seen adolescents and adults.

Other previous reported findings like corneal opacities, hyperplasia of Retinal Pigment Epithelium and Glaucoma was not seen in our study. Brushfield's spots, commonly seen in Caucasian population were not seen in our study.

In summary, Indian children with Down syndrome present with high prevalence of upward slant, epicanthal folds and epiblepharon. Prevalence of refractive errors, nystagmus and

strabismus was low. No corneal opacity, RPE hyperplasia, glaucoma or Brushfield spots were seen as compared to other studies.

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