Ocular Manifestations in Down's Syndrome

Nanda L[1], Adarsh[2], V.K. Srivastava[3], Nithisha T M[4], Shivakumar M[5], Garima Yadav[6]

ABSTRACT

Introduction: Down syndrome is also known as trisomy 21. It is a genetic disorder caused by presence of all or part of 3rd copy of chromosome 21. It is typically associated with delay in physical growth, characteristic facial features and mild to moderate intellectual disability. The parents are genetically normal and the extra chromosome occurs by chance. Ocular abnormalities like cataract, strabismus, and refractive errors are common. The aim of the present study was to report clinically significant ophthalmic abnormalities in children with Down's syndrome.

Material and methods: A prospective study was done in sixty four children with Down syndrome between the age group 1 year to 14 years. All children underwent ocular examination which included visual examination, slit lamp examination and fundus examination.

Results: Sixty four patients with Down syndrome (mean age, 8 years; range, 1year 6months to 14 years) underwent eye examinations. Clinically significant refractive errors were present in 32% of the subjects, strabismus in 32%, nystagmus in 3%, and cataract in 7%.Six year and younger patients showed a higher prevalence of hyperopia than those who are in older age groups; pts between 6-10 years old had a higher prevalence of astigmatism.

Conclusion: This study shows early detection of ocular abnormalities in children with Down syndrome has greater importance in reducing visual abnormalities.

Keywords: Refractive error, strabismus, cataract, visual acuity, retinal abnormalities

INTRODUCTION

Down's syndrome is the most common genetic chromosomal disorder of chromosome 21(trisomy 21) and is associated with significant ocular morbidity. It varies in severity, causes lifelong intellectual disability and developmental delays. Most common features are flattened facial features, small head, short neck, protruded tongue, upward slanting eyes, small eyes, poor muscle tone, broad short hand with single crease in the palm. The abdomen is often protuberant and cardiac malformations are common. Ocular findings include strabismus, cataract, refractive errors, accommodative insufficiency, blephritis, retinal abnormalities, epicanthal folds.[1,2] Most reported studies of ocular findings in Down syndrome have been performed in Caucasians.[2,3] In the present study, our aim was to study the patients with Down syndrome to identify the characteristic ocular findings and to find the prevalence rate.

MATERIAL AND METHODS

The study was done in the department of ophthalmology and in coordination with department of pediatrics, RRMCH Bangalore. It was a cross sectional study conducted for the duration of 1 year 6 months (January 2014 to September 2015). The study subjects includes all the patients diagnosed with Down Syndrome during the period of study with age between 1 year to fourteen years and patients below the age of 1year and above age of 14 years were excluded from the study. Total sixty four patients were examined. Informed consent from patient's attendees was taken and ethical clearance from the ethical committee was obtained. Clinical examination of the eye included visual assessment with cycloplegic refraction, ocular motility, ocular adenexa, slit lamp examination, fundus examination, glaucoma evaluation and systemic examination.

A clinical history was obtained from parents regarding patients age, maternal age of conception, history of wearing glasses, onset of strabismus and/ or nystagmus, occlusion therapy for amblyopia, previous external infections, watering, photophobia, treatment modalities, previous history cataract or strabismus surgery inquired. All details about previous cardio-vascular surgery any complications related to Pulmonary, endocrine, GI examination, neurological examinations were inquired.

The visual acuity was evaluated according to the patient's intelligence and responsiveness. In a non verbal patient vision is evaluated in terms of location (eccentric or central fixation) and duration. In verbal patients it is tested using optotypes (snellens chart, tumbling E chart, Tellen cards) few were tested with pattern vep.[3,4] Palpebral fissure was measured with the help of a straight ruler which was placed over the bridge of nose at the level of inner and outer canthus. Horizontal and vertical displacement was measured.

The lid margins and conjunctiva were assessed for abnormalities such as blepharitis, hordeola, chalzion and conjunctivitis and some pts may have nasolacrimal duct obstruction.[5,6] The diagnosis of nasolacrimal duct obstruction was based on history of epiphora or recurrent mucopurulent discharge since infancy and by the reflux of mucus with pressure over lacrimal sac. The presence of keratoconus, keratoglobus and iris abnormalities such as Brushfield's spots and stromal hypoplasia was also evaluated.[7] Lens was evaluated for developmental or congenital cataract. Cycloplegic refraction was performed in all patients, regardless of age, 45 min after three to five instillations of one drop of cyclopentolate1%. Emmetropia was defined as refractive error between -0.75 diopter (D) and +0.75 D spherical equivalent.[8,9] Hyperopia

[1]Senior Resident, [3]Professor and HOD, [4]Associate Professor, [5]Professor, [6]Junior Resident, Department of Ophthalmology, [2]Professor and HOD , Department of Paediatrics, RRMCH, India

Corresponding author: Garima Yadav, Junior Resident, Department of Ophthalmology, RRMCH, India

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was defined as more than +0.75 D spherical equivalent and myopia was defined as less than -0.75 D spherical equivalent. Astigmatism was defined as refractive error more than +- 0.75 of the cylinder.[10,11]

Direct and indirect ophthalmoscopy after cycloplegic retinoscopy was used to examine the retina, choroid and optic disc, and included a full assessment of vessels in relation to optic disc.

STATISTICAL ANALYSIS

Data was analyzed using SPSS version 19. Results are based on descriptive statistics.

RESULTS

Sixty four patients with Down syndrome (mean age, 8 years; range, 1year 6months to 14 years) underwent eye examinations. Clinically significant refractive errors were present in 32% of the subjects, accommodative insufficiency in 34%, nystagmus in 3%, and cataract in 7%, retinal abnormality in 10%, epicanthal folds in 90%, lacrimal system obstruction in 12%, blephritis in 18%, iris abnormalities in 12%, upward slanting of palpebral fissure with outer canthus in 93%. Strabismus was present in 21 patients (32%), 14 of whom had esodeviations and 7 of whom had exodeviations. Nystagmus was observed in 2 patients (3%), usually in the horizontal-pendular type (table-1). Six year and younger patients showed a higher prevalence of hyperopia than those who are in older age groups; patients between 6-10 years old had a higher prevalence of astigmatism. Patients older than 10 years had more cataract, strabismus, iris abnormalities. Myopia is more common in patients with cardiac abnormalities. Patients develop amblyopia due to strabismus and refractive error. Brushfield spots and keratoconus were not found.

We observed that majority of the patients had upward slanting of the palpebral fissure. On examining the fundus it showed numerous vessels >18 crossing the optic disc margin and extending towards retinal periphery. In one patient retinal pigment epithelium showed focal hyperplasia. In the bar chart below incidence of ocular abnormality in Down syndrome is shown (figure-1)

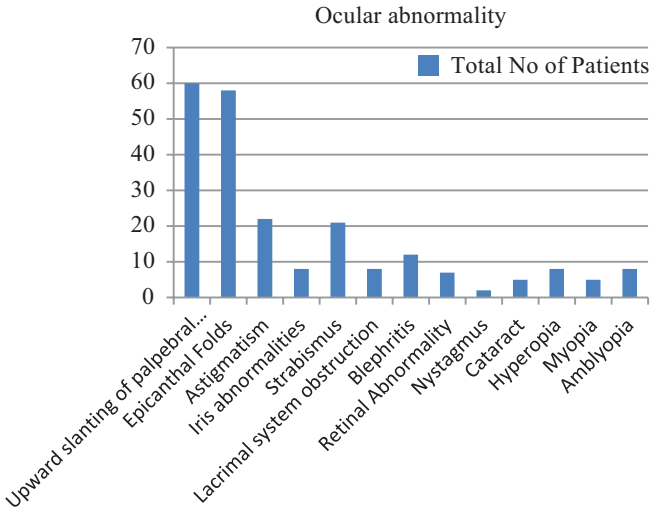

Figure-1: Incidence of ocular abnormality in Down syndrome

DISSCUSSION

The incidence of strabismus in our study was 32% which is similar to that in other studies from da Cunha and Moreira (38%)[12] or Lowe (33%)[13] or Hiles et al (34%).[14] Asians are shown to have higher prevalence of exotropia as compared to Caucasians.[15] Racial factors may play a role in this strikingly high incidence. Upward slanting of palpebral fissure, the most frequent ocular finding, is present in 60 patients(93%). Epicanthal folds, the second most prevalent feature, were found in 58 patients (90%).The prevalence of these two abnormalities has been reported as low as 9% and as high as 100%. This variation might be related to age and racial factors. Several authors have reported a decrease in prevalence with increased age as shown in table 2.

Nystagmus was present in 2 patients (3%), which is in accordance with previous reports of 4-30%.The patients having nystagmus in the present study usually had refractive errors, which are in accordance with other studies reporting nystagmus associated with refractive errors.[16] The incidence of cataract (7%) was similar to that in the studies done by Shapiro and France (7%)[16] and Roizen et al (5%)[17], but quite lower than 11-86% of other reports by Berk et al (11%)[18] and da Cunha and Moreira (20%).[12] This varying incidence rate might be related to the differences in age distribution and diagnostic criteria.

The incidence of keratoconus varies between 0 and 30%. But

Sr. No.	Ocular Abnormality	Sample Size	Positive Findings	
1	Upward slanting of palpebral fissure with outer canthus 2mm or higher than inner canthus	64	60	93.75%
2	Epicanthal Folds	64	58	90.63%
3	Astigmatism	64	22	34.38%
4	Iris abnormalities	64	8	12.50%
5	Strabismus	64	21	32.81%
6	Lacrimal System Obstruction	64	8	12.50%
7	Blephritis	64	12	18.75%
8	Retinal Abnormality	64	7	10.94%
9	Nystagmus	64	2	3.13%
10	Cataract	64	5	7.81%
11	Hyperopia	64	8	12.50%
12	Myopia	64	5	7.81%
13	Amblyopia	64	8	12.50%

Table-1: This table shows ocular abnormality with prevalence rate

Comparison of ocular findings in previous studies with our findings	Present study	Wong and Ho (1997)[19]	Da Cunha and Moreira (1996)[12]	Berk et al (1996)[18]	Caputo et al (1989)[20]	Shapiro and France (1985)[16]
Number of patients	**64**	**140**	**152**	**55**	**187**	**53**
Nationality	Indian	Hong Kong	Brazil	Turkey	US	US
Range of age (years)	1 yr 6month - 14	0-13	0-18	0-25	0-26	7036
Mean age (years)	8	3.74	-	7.2	5.8	17.4
Upward slanting (%)	60(93)	140	125(82)	-	-	47(89)
Epicanthus (%)	58(90)	140	92(61)	13(24)	-	-
Refractive errors (%)	35(54)	137(98)	149(98)	60	122(65)	35
Hyperopia	8	42	39	29	39	17
Myopia	5	12	19	7	42	18
Astigmatism	22	8	91	24	41	12
Strabismus (%)	21(32)	28(20)	57(38)	12(22)	107(57)	23(43)
Esotropia	14		51	11	97	22
Exotropia	7		0	1	4	
Hypertropia	0		4	0	6	
Nystagmus (%)	2(3)	15(11)	28(18)	7(13)	55(29)	5(9)
Nasolacrimal duct obstruction (%)	8(12)	-	46(30)	12(22)	9(5)	-
Blepharitis (%)	12(18)	8(7)	45(30)	19(35)	-	25(47)
Number of retinal vessels >= 18 (%)	6(9)	16	42	21(38)	-	-
Lens opacities (%)	5(7)	4	20(13)	11(20)	21(11)	7(13)
Focal RPE hyperplasia (%)	1(1.5)	-	-	-	-	-
Glaucoma (%)	0	1	-	-	10(5)	-
Corneal opacities (%)	0	-	-	-	-	-
Keratoconus (%)	0	0	-	-	-	8(15)
Brushfield spots (%)	0	0	79(52)	20(36)	-	43(81)

Table-2: Comparison of ocular findings in previous studies with our findings

no keratoconus was seen in our study because the median age was very low. The children might be young so keratoconus might not have developed but as their age increases the chances it might occur. Unlike higher prevalence rate up to 90%, our study showed iris abnormality up to 12.50% and no brushfield spots were seen. This can be explained by dark brown and black irises in our children. Wong and Ho[19] also reported that non of Hong Kong children showed these conditions either.

As shown above in the results our study results coincide with that of other studies done as shown in table 2.

CONCLUSION

Early awareness and detection of clinical features of Down's syndrome will decrease the complications and sight threatening conditions. This study suggests that children having Down syndrome are at a greater risk of visual impairment and therefore, early detection should be emphasized to prevent ocular related problems. This article provides a more information of the prevalence and severity of the complications in patients with Down's syndrome.

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