Ophthalmic Manifestation of Trisomy 21 in a Tertiary Eye Hospital- a Hospital Based Study

Sidratul Muntaha Naznin[1], Mohammad Mostafa Hossain[2], Quazi Sazzad Iftekhar[3], Tarikul Ahasan[4], Bipul Kumer De Sarker[5]

Abstract

Introduction: *Trisomy 21 is a genetic disorder caused by abnormal cell division results in an extra full or partial copy of chromosome 21.It is commonly known as Down syndrome. 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 manifestation in children with Down's syndrome.*

Material and methods: *A prospective study was carried out in one hundred and twenty children with Down syndrome between the age group of one year to 16 years since January 2018 to September 2019. All children underwent ocular examination which included vision assessment, cyclo refraction, slit lamp examination and fundus evaluation.*

Results: *One hundred and twenty patients with Down syndrome (One year six months to 16 years) underwent ocular examinations. Average age of presentation was +/-8 years. Most of the down patient was male 63.3%. Clinically significant refractive errors were present in 72%, cataract in 80%, retinal abnormality in 53.3%, epicanthal folds in 92%, lacrimal system obstruction in 68.3%, iris abnormalities in 26.6%. Strabismus was present in 74 patients (61.6%).*

Conclusion: *This study concludes that the early detection of ocular abnormalities in children with Down syndrome has greater importance in reducing visual morbidity and better life expectancy.*

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

Introduction

Down's syndrome is the most common genetic disorder of chromosome 21(trisomy 21) and is associated with significant ocular morbidity. Down syndrome is a chromosomal disorder caused when abnormal cell division results in an extra full or partial copy of chromosome 21. This extra genetic material causes the developmental changes and characteristic physical features of Down syndrome. The frequency of Down syndrome is approximately 1 in every 800 births; the rate is increased in older mothers. It is one of the most common chromosomal abnormalities in live-born children. The improved quality of medical care and educational resources have allowed for productive and longer life expectancy.

It varies in severity, caus-es lifelong intellectual disability and developmental delays. Most common features are flattened facial features,

1. Associate Professor cum consultant, Pediatric department, Ispahani Islamia Eye Institute& Hospital
2. Professor cum Senior consultant, Pediatric department, Ispahani Islamia Eye Institute & hospital
3. Associate Professor cum senior consultant, Pediatric department, Ispahani Islamia Eye Institute & Hospital
4. Assistant Professor cum consultant, Pediatric department, Ispahani Islamia Eye Institute& Hospital
5. Associate Professor and Consultant, Ispahani Islamia Eye Institute& Hospital

Address of Correspondence: Dr. Sidratul Muntaha Naznin, Consultant cum Associate Professor, Department of pediatric ophthalmology & strabismus, Ispahani Islamia Eye Institute & hospital, Farmgate, Dhaka, Cell : 01316100916, E-mail: nsidratulmuntaha @gmail.com

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 insuf-ficiency, blepharitis, retinal abnormalities, epicanthic folds.1, 2 Most of the 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.

It also commonly causes other medical abnormalities, including heart and gastrointestinal disorders. Better understanding of Down syndrome and early interventions can greatly increase the quality of life for children and adults with this disorder and insure a better life expectancy.

Material And Methods

The study was carried out in the department of pediatric ophthalmology and strabismus in Ispahani islamia eye institute & hospital, Dhaka, Bangladesh. It was a prospective study conducted for the duration of 1 year 6 months (January 2018 to September 2019). The study subjects includes all the patients diagnosed with Down Syndrome during the period of study with age between one year to 16 years and patients below the age of one year and above age of 16 years were excluded from the study. Total one hundred and twenty patients were finally included in the study. Informed consent from patient's attendees was obtained. Clinical ex-amination of the eye included visual assessment with cylo-plegic refraction, anterior segment examination, glaucoma evaluation, background fundus examination and systemic examination was conducted.

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

The visual acuity was evaluated according to the patient's intelligence and responsiveness. In a nonverbal patient vi-sion is evaluated in terms of location (eccentric or central fixation) and duration. In verbal patients it is tested using op-totypes (snellens chart, tumbling E chart, Tellen cards) 4, Palpebral fissure was meas-ured 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 abnormal-ities such as blepharitis, hordeolam, chalazion and conjunctivi-tis and significant number of patients 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 stro-mal 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 was defined as more than +0.75 D spherical equivalent and myopia was defined as less than -0.75 D spherical equiva-lent. Astigmatism was defined as refractive error more than +- 0.75 of the cylinder.10,11

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

Results

One twenty patients with Down syndrome (average age of presentation +/- 8 years; range one year six months to 16 years) underwent ocular examina-tions. Average age of presentation was 6-10 years. Most of the down patient was male 63.3%. Clinically significant refractive errors were present in 72% of the subjects, cataract in 80%, retinal abnormality in 53.3%, epicanthal folds in 92%, lacrimal system obstruction in 68.3%, and iris abnormalities in 26.6%. Strabismus was present in 74 patients (61.6%), 48 of whom had esodeviations and 26 of whom had exodeviations. Nys-tagmus was observed in 58 patients (48.3%), usually in the jerky type. 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. My-opia is more common in patients with cardiac abnormalities. Patients develop amblyopia due to strabismus and refractive error. Brush field spots and keratoconus were not found.

Current study revealed that that 100% of the patients had upward slant-ing of the palpebral fissure. On examining the fundus it showed numerous vessels >18 crossing the optic disc mar-gin and extending towards retinal periphery. In one patient retinal pigment epithelium showed focal hyperplasia. Important ophthalmic finding are showed in following tables.

Table I : *Basic Demographic Data*

Variable	Range	Number	Percentage
Age of presentation	0-5 Years	20	16.67%
	6-10 Years	80	66.67%
	11-16 Years	20	16.67%
Gender	Male	76	63.3
	Female	44	36.7
Area	Urban	102	85%
	Rural	18	15%
Positive Family History	Positive	22	18.3%
	Negative	98	82.7%

Table II: *Refractive error in study subject*

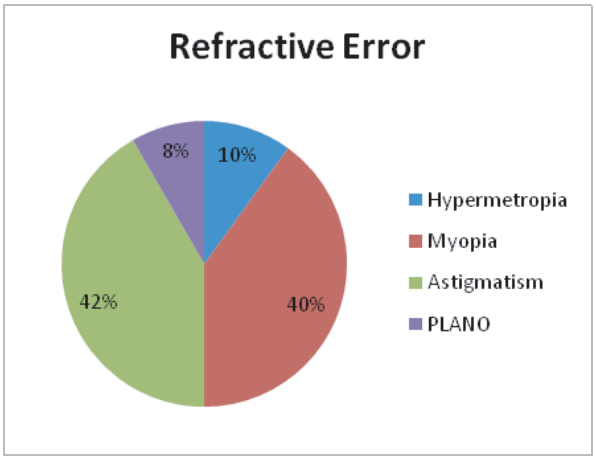


Table III: *Visual acuity in study subject*

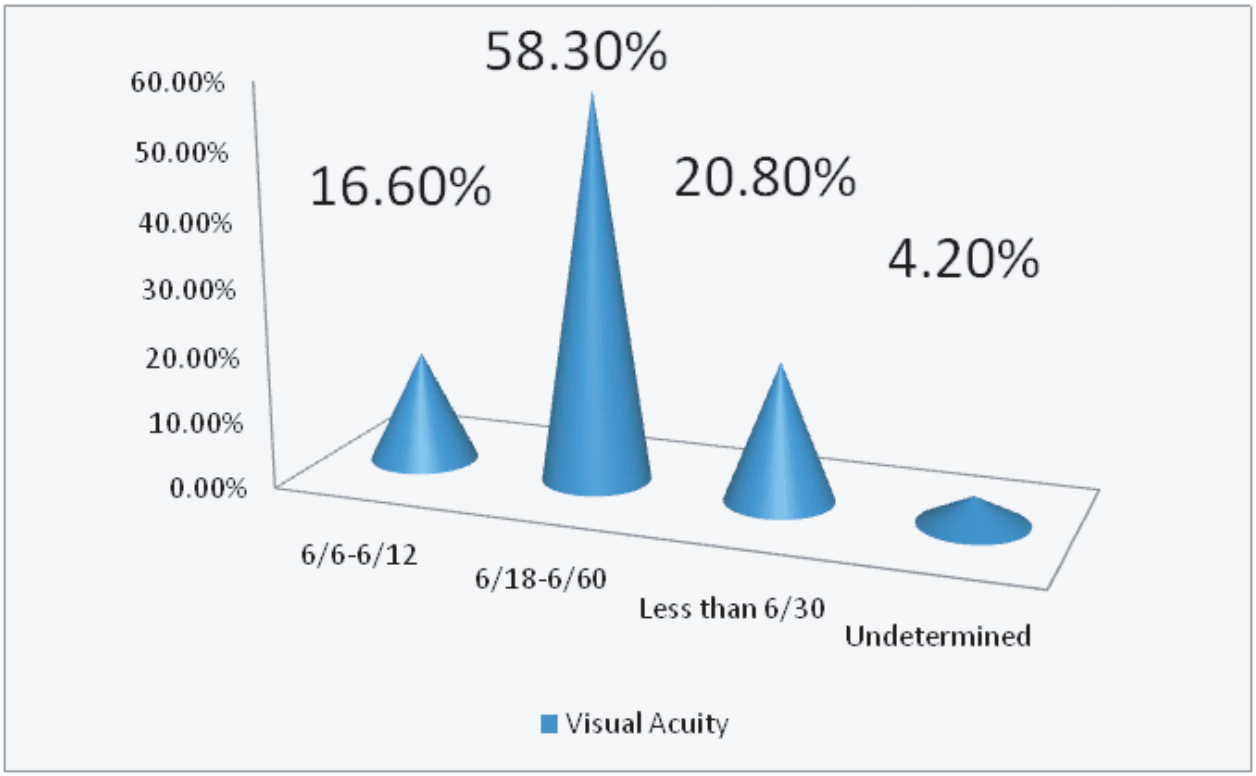
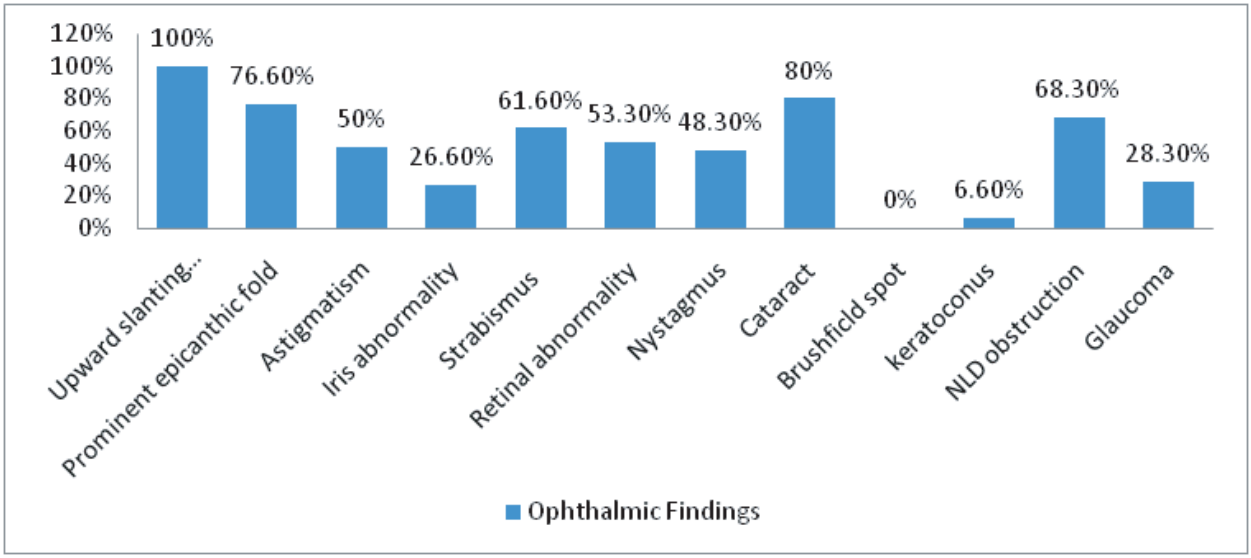


Table IV: *Ophthalmic finding in study population*



Disscussion

Upward slanting of palpebral fissure, the most frequent ocular finding, is present in almost all of the patients (100%). Epicanthic folds was the another prevalent feature found in 92 patients (76.6%) The incidence of strabismus in our study was 61.6% which is differ to that in other studies from da Cunha and Moreira (38%). Asians are shown to have higher prevalence of exotropia as compared to Caucasians. Racial factors and increased maternal age may play a role in this striking-ly high incidence.

Nystagmus was present in 58 patients (48.3%), which is in ac-cordance with previous reports of 4-30%.The patients hav-ing nystagmus in the present study usually had refractive errors, which are in accordance with other studies reporting nystagmus associated with refractive errors.9The incidence of cataract (80%) was significantly high to that in the studies done by Shapiro and France (7%)16 and Roizen et al (5%)7, 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 no keratoconus was seen in our study because the median age was very low. The children might be young so keratoco-nus 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 26.6% and no brushfield spots were seen. This can be explained by dark brown and black irises in our children. Wong and Ho19 also reported that none of Hong Kong children showed these con-ditions either.

Conclusion

This study suggests that children having Down syndrome are at a greater risk of visual impairment and therefore, early detection should be emphasized to pre-vent ocular related complication. when abnormal cell division results in an extra full or partial copy of chromosome 21As children with Down syndrome have social, behavioral and emotional difficulties, visual impairment significantly reduces their overall quality of life. Therefore, the pediatric ophthalmologist must strive to provide the best advice and intervention for improved visual acuity and to minimize ocular complications in all children with Down syndrome. Further, parents and teachers must be aware that despite refractive correction children with Down syndrome may have poor visual acuity which may lead to failure in performing intellectual tasks and be erroneously attributed to their cognitive impairment. Therefore; early awareness and detection of clinical features of Down's syndrome will reduce the visual morbidity and insure better life expectancy.

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