Ocular Abnormalities in Children with Down's Syndrome Attending a Semi Urban Tertiary Care Centre in India - A Cross Sectional Study

Sujatha Nambudiri[1], Sinumol Sukumaran Thulaseedharan[2], Seena T.V.[3]

[1, 2, 3] Department of Ophthalmology, Government Medical College, Thrissur, Kerala, India.

ABSTRACT

BACKGROUND

Down syndrome, first described in 1866, is the most common chromosomal derangement in live births. More than half of the patients with Down syndrome have ophthalmic manifestations. We wanted to evaluate the most common ocular abnormalities in children with Down syndrome in South India.

METHODS

All children with positive chromosomal analysis report for Down syndrome between 2 - 18 years attending a semi-urban, tertiary medical care centre from 1/1/2013 to 1/1/2015 were included in this hospital based clinical cross sectional study. Detailed ocular examination included visual acuity assessment using age specific tests, diffuse light examination, assessment of ocular alignment, motility, cover tests, slit-lamp biomicroscopy, cycloplegic refraction and direct and indirect ophthalmoscopy.

RESULTS

In our study, 95 % of children had ocular abnormalities. The most important defects were refractive errors (83.33 %), hyperopia (35 %), myopia 21 % of whom 2 children had high myopia of more than - 6D, and astigmatism (15 %). Among the refractive errors, hyperopia was the commonest. Other ocular abnormalities were cataract (13.33 %), esotropia (13.33 %), exotropia (3.33 %), nystagmus (6.67 %), nasolacrimal duct obstruction (8.3 %) and optic disc coloboma (1.6 %).

CONCLUSIONS

Refractive errors, strabismus were the most common and significant visual defects identified in children with Down syndrome in our study. Early detection of refractive errors, strabismus and prompt and appropriate intervention like glasses for refractive errors and surgical correction of strabismus is absolutely necessary to prevent development of amblyopia. Improvement of vision accelerates the overall development of the child. Management of Down syndrome children should be a team approach with ophthalmologist playing an important role. These children should have early and regular ophthalmological evaluations to maximise the benefit.

KEYWORDS

Down Syndrome, Ocular Abnormalities, Visual Acuity in Children

Corresponding Author:
Dr. Sinumol Sukumaran Thulaseedharan,
Department of Ophthalmology,
Associate Professor,
Government Medical College,
Thrissur, Kerala, India.
E-mail: drsinuthulasi@gmail.com

DOI: 10.18410/jebmh/2021/15

How to Cite This Article:
Nambudiri S, Thulaseedharan SS, Seena TV. Ocular abnormalities in children with down's syndrome attending a semi urban tertiary care centre in India - a cross sectional study. J Evid Based Med Healthc 2021;8(02):75-79. DOI: 10.18410/jebmh/2021/15

Submission 26-08-2020,
Peer Review 01-09-2020,
Acceptance 23-11-2020,
Published 11-01-2021.

BACKGROUND

Down syndrome is the most prevalent genetic disorder associated with intellectual disability. It is due to the presence of complete or partial triplication of chromosome 21[1]. Incidence of Down syndrome ranges from 1 in 800 to 1 in 1200 live births. In addition to cognitive impairment, it is associated with congenital anomalies and characteristic dysmorphic features. Most reliable signs are hypotonia, brachycephaly, upward slant of the eye and transverse line in the palm of the hand (Simian crease). They are more prone to congenital heart defects, gastrointestinal anomalies and hypothyroidism. About 60 % of the patients with Down's syndrome has ophthalmic manifestations like refractive errors, strabismus, nystagmus, anomalies and infections of lids, corneal ectasia, brush field spots, presenile cataract & glaucoma. The high prevalence of ophthalmic disorders highlights the need for periodic evaluations and individualized treatment plans for children with Down's syndrome.[2] Early detection and treatment is critical for prevention of amblyopia in many cases. It is also expected that their overall development and quality of life could be enhanced by early ophthalmological interventions.

We wanted to identify the most common ocular abnormalities in children with Down's syndrome attending a public, semi urban, medical tertiary care centre in South India and determine the percentage of each abnormality.

METHODS

This is a hospital based clinical cross-sectional study. A monthly Down syndrome clinic is being conducted by the Department of Paediatrics in our institution. This study involved 60 children with Down's syndrome who attended the clinic during the study period.

Inclusion Criteria

All children with Down syndrome aged between 2 - 18 years attending the Down syndrome clinic of our hospital, from 1/1/2013 to 1/1/2015 were included in the study. The clinical signs we considered for diagnosis of Down syndrome were brachycephaly, hypotonia, flat face, upward slant of the eyes, transverse line in palm "Simian crease", small ears, wide space in between the 1st and 2nd toe ("sandal gap"), small inter nipple distance, Brushfield spots and nuchal skin fold. The diagnosis of Down syndrome was confirmed by the paediatrician on the basis of consistent clinical characteristics and also by chromosomal study.

Exclusion Criteria

Patients less than 2 years and more than 18 years were excluded. Children under 2 years were excluded from this study as a reliable visual acuity assessment was difficult.

Medical ethics committee clearance was obtained for the study. Informed consent was obtained from the parents of the affected children prior to enrollment in the study. The enrolled children were brought to our paediatric ophthalmology clinic for detailed ophthalmic evaluation. Ocular examinations included visual acuity assessment, diffuse light examination, assessment of ocular alignment, motility, cover tests, slit-lamp biomicroscopy, cycloplegic refraction and direct and indirect ophthalmoscopy. Mild sedation was used in children who did not cooperate for evaluation of refraction and ophthalmoscopy.

The visual acuity assessment was done using picture charts and Snellen's E chart. Prior to data collection, the children were encouraged to match or name specified optotypes on each chart (Figure 1) under binocular conditions at near distance. Then one eye was covered by an eye patch or occluder and assessment was done. Due to varying degrees of cooperation, many times, the visual acuity had to be assessed binocularly rather than each eye separately.

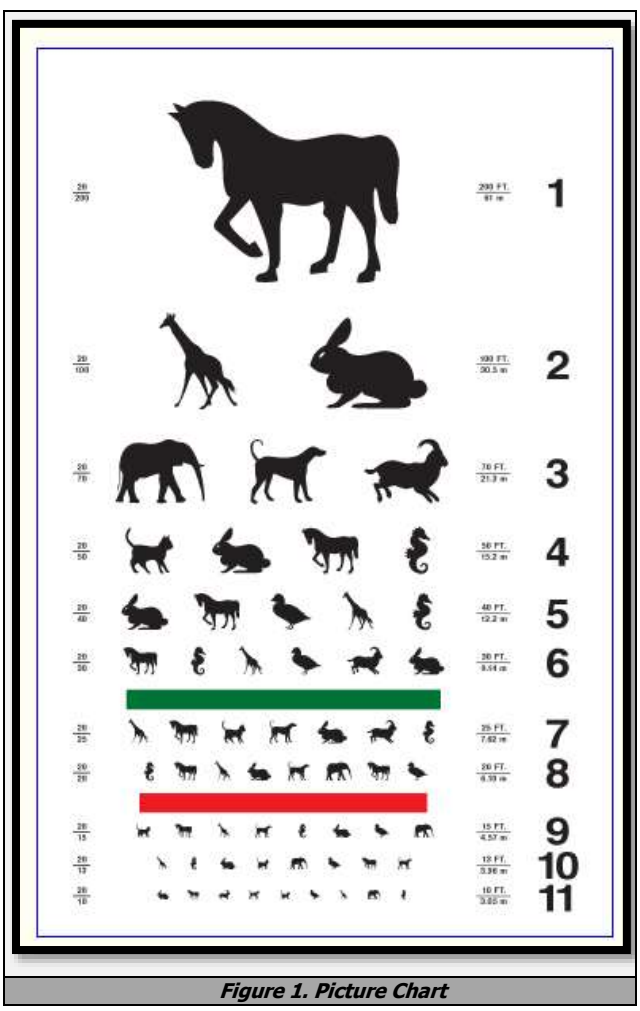


Figure 1. Picture Chart

CSM (Central Steady and Maintained Fixation) was taken as a rough assessment of visual acuity where these methods failed. CSM involves covering one eye while the child is fixating on an object. The non-covered eye should maintain central, steady fixation, which is maintained through a blink.

Diffuse light examination included the Hirschberg test (the corneal light reflex test) and cover tests for strabismus. Ocular movement assessments were done meticulously. Abnormalities like ptosis, microphthalmos and

nystagmus were specifically looked for. Ptosis (blepharoptosis) is an abnormal drooping of the upper eyelid covering more than 1 / 6th of the cornea, confirmed by measurements. Microphthalmos is a severe developmental disorder of the eye in which one or both eyes are abnormally small and have anatomic malformations. A scan was done for measurement of axial length and confirmation. Nystagmus is defined as a regular and rhythmic to and fro involuntary oscillatory movement of eyes. Examination for nystagmus was done in primary position and also during extra ocular movements.

Epicanthal fold, a small fold of skin across the inner angle of the eye was looked for (Figure 2). The diagnosis of nasolacrimal duct obstruction was made when there was a history of watering or recurrent mucopurulent discharge from the eyes since infancy and also by demonstration of reflux of mucus through the puncta on pressure over the lacrimal sac area on the side of the nose.

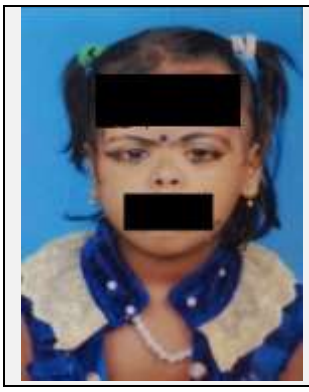


Figure 2. Medial Epicanthal Folds and Pseudo Esotropia

The palpebral fissure was evaluated by placing a clear plastic ruler across the bridge of the nose at the level of both the inner canthi, and then measuring the vertical displacement of the outer canthi. Displacement of 2 mm or more above the horizontal line was taken as the upward (or oblique) slanting of fissures (Fig. Curvature of the cornea was evaluated by keratometry to rule out keratoconus. Diffuse light examination of eyelid margin and conjunctiva were done to rule out trichiasis, blepharitis and conjunctivitis. Slit lamp biomicroscopy was done to rule out the presence of cilia touching the cornea, corneal erosions (by fluorescent staining in suspicious cases), iris features such as Brushfield's spots and stromal hypoplasia. Brushfield spots are white to yellow, slightly raised, discrete pinpoint to pinhead sized areas in the iris periphery.

A thorough examination of the lens regarding its shape, position and presence of any opacity was done. The type of cataract, if any, was identified and whether the opacity is involving the visual axis was also assessed. Dilated fundus examination was done using direct and indirect ophthalmoscopes. Detailed optic disc evaluation for any optic disc coloboma, pit, optic disc hypoplasia, tilted disc, temporal pallor, glaucomatous cupping etc. were done with direct ophthalmoscope. Evaluation for any peripheral retinal degeneration was done with indirect ophthalmoscopy.

Age specific cycloplegic refraction was performed in all patients. Atropine was used for children up to 4 years of age, homatropine in the 5 - 12 years age group and tropicamide in patients more than 12 years of age. A refractive error between + 0.75 diopter (D) and - 0.75 diopter (D) spherical equivalent was taken as emmetropia, less than - 0.75 D spherical equivalent was taken as myopia and more than + 0.75 D spherical equivalent was taken as hyperopia. More than ± 0.75 D of the cylinder was taken as astigmatism. Greater than - 6.0 D of spherical equivalent was taken as severe or high myopia.

Statistical Analysis

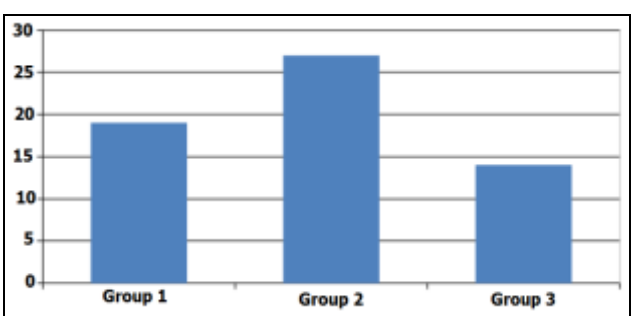
Analysis of data was done by Epi Info software.

RESULTS

60 children were studied out of whom 24 (40 %) were males and 36 (60 %) were females. The mean and median ages of the children were 7.1 and 6 years respectively. Majority of our study population were children less than 6 years of age.

Visual Acuity

In majority of children i.e. Group 2 & 3 (n = 41), visual acuity was assessed by picture charts and Snellen's E chart (Figure 2). There was a group of children in whom visual acuity could not be assessed by the above methods and they were tested by checking the central steady and maintained fixation (CSM) (n = 19) (Graph 1).



Graph 1. Visual Acuity

Group 1 - Vision assessment by checking the CSM (n = 19).
Group 2 - Vision between 6 / 6 and 6 / 36 (n = 27).
Group 3 - Vision ≤ 6 / 36 (n = 14)

Refractive Errors

The most common and important ophthalmological problem identified was refractive errors (83.33 %), among which hyperopia was the commonest followed by myopia and astigmatism (Table 1). Two children had severe pathological myopia with more than six diopters of power.

Refractive Error	Number of Eyes (%)
Emmetropia	10 (16.66 %)
Hyperopia	21 (35 %)
Myopia	13 (21.67 %)
Astigmatism	9 (15 %)
Mixed	7 (11 %)

Table 1. Refractive Status of 60 Patients with Down's Syndrome

Ocular Abnormalities

The most frequent ocular finding in our study group was upward slanting of the palpebral fissures, noticed in 86 % patients (Table 2). Epicanthal folds were another common finding found in 83.3 % patients (Table 2). But these abnormalities have no functional significance. Important structural anomalies detected were congenital cataracts, strabismus, nystagmus and posterior segment malformations like pathological myopic degenerations and coloboma (Table 2) with significant functional and therapeutic implications. Early detection, intervention and follow ups are important in these cases for the proper development of visual systems and prevention of amblyopia and other complications.

Ocular Abnormalities	Number of Patients (%)
Upward slanting of the palpebral fissures	52 (86 %)
Epicanthal folds	50 (83.3 %).
Cataract	8 (13.33 %)
Esotropia	8 (13.33 %)
Exotropia	2 (3.33 %)
Nystagmus	4 (6.67 %)
Nasolacrimal duct obstruction	5 (8.3 %)
Myopic fundus	2 (3.3 %)
Optic disc coloboma	1 (1.6 %)

Table 2. Distribution of Ocular Abnormalities

DISCUSSION

Ocular abnormalities are commonly associated with Down's syndrome but may be overlooked because of the presence of other severe systemic problems. Some children may be receiving suboptimal care because of their condition. Not all patients are referred for ophthalmological evaluation. Also difficulties are encountered for accurate visual acuity assessment and other tests.

Our study found out that 95 % of children with Down's syndrome are having ocular abnormalities. This is a significant increase in incidence when compared with other similar studies. The different criteria used in the definition of ocular abnormalities and the variations in selection of the target population may be the cause.

The most frequent ocular finding was upward or oblique slanting of the palpebral fissures, present in 52 patients (86 %) (Table 2). Epicanthal folds were another common finding found in 50 patients (83.3 %) (Table 2). These findings were comparable to studies done in Hong Kong[3] and Korea.[4] Asian children have distinctive eyelid structures which made these eyelid abnormalities more noticeable. The prevalence of these two abnormalities have been reported as low as 9 %[5] and as high as 100 %[6]. This large variation may be related to age and racial factors. Several authors have reported a decreasing prevalence with increasing age.[7]

83 % cases had refractive errors, with hypermetropia (35 %) being the most frequent, followed by myopia (21.67 %). Two children had severe / high myopia with power greater than - 6 diopters. 15 % of the children had astigmatism, of which 7 (11 %) were of mixed astigmatism. 16.66 % (n = 10) of children were emmetropic. This is comparable with the studies conducted by Stephen E, Dickson J, Kindley AD, Scott CC[8] and Fimiani F, Iovine A, Carelli R, Pansini M, Sebastio G, Magli A[9] where hypermetropia was demonstrated as the most frequent refractive error among children with Down's syndrome. Thus, all studies, disregarding certain minor differences, agree that there is significantly higher prevalence of refractive errors in Down's syndrome compared to the normal population. Hypermetropia is a refractive error which often reduces spontaneously in normal children, but is likely to persist beyond infancy in Down's syndrome.[10] Hyperopia must be corrected in these children due to their reduced accommodation. During this study, we were able to pick up the undetected refractive errors in many children and prescribe appropriate corrections. With proper counseling of the parents, many children started using spectacles regularly.

The percentage of strabismus was 16.6 % in our study. Esotropia was more common (13.33 %) than exotropia (3.33 %) which was comparable to previous reports.[5,9] The study conducted in Nigeria[11] also reported similar results (18.1 %). There has been no explanation for the prevalence of strabismus in Down's syndrome. Possibly a combination of factors such as decreased fusional capacity, decreased visual resolution capacity and a failure to develop an adequate accommodative convergence mechanism may be responsible.

Nystagmus was found in 6.67 per cent (n = 4) of the children, which was similar to previous reports conducted by Fimiani F, Iovine A, Carelli R, Pansini M, Sebastio G, and Magli A.[9] All had manifested nystagmus of horizontal type. Statistical analysis showed that there is no statistically significant association between refractive errors and nystagmus (Fisher Exact - 7.9825 and 2-tailed p 0.3545402346). Occurrence of nystagmus is not of much significance as nystagmus in patients with Down's syndrome was not always associated with significant decrease in visual acuity as pointed out by Wagner SW, Caputo AR and Reynolds DR.[12]

The percentage of lens opacities (13.33 %) in our study was slightly higher than that in the studies of Roizen NJ, Patterson D (5 %)[13] and Wong and Ho (4 %).[3] These varying incidence rates might be related to the differences in age distribution and diagnostic criteria. Nine patients had blue dot cataract and one had combined sutural and blue dot cataract in our group, but none needed surgery as the visual axis remained clear.

The incidence of keratoconus reported in children with Down's syndrome varies between 0 to 30 % in our study. This may be due to the low median age of our patients. Brushfield spots or iris hypoplasia was not detected in any of our patients. This can be explained by the dark-brown irises in our population. These results were also comparable to that of other Asian studies.[3, 4]

The percentage of nasolacrimal duct obstruction ranges from 5 to 30 % according to the age distribution of each study.[2,4,6] In our study, it was found in 5 patients (8.3 %) (Table 2). Retinal examination showed myopic degenerative changes in 2 cases and optic disc coloboma in one case in our study population. In the study by Kim JH, Hwang JM, Kim HJ, Yu YS, they found that 15 % had retinal abnormalities.[4] Not many studies evaluated the retinal

abnormalities of Down's syndrome. More research is needed in this aspect.

Limitations of the Study

The smaller study population, we could get, is a major limitation of our study. As this was a hospital-based study, only children getting regular medical attention were picked up among a large iceberg below. The different criteria used in the definition of ocular abnormalities and the variations in selection of the target population in various studies made the comparison between studies difficult.

CONCLUSIONS

Refractive errors, strabismus and cataract were the most common visual defects identified in children with Down syndrome in our study. Early detection and appropriate intervention is absolutely necessary to prevent amblyopia and also to promote overall healthy development of the child. Hence, these children should have early and regular eye examinations to maximise the benefit. Though assessing vision and performing ocular examinations in these children are challenging, it is possible to obtain results if the techniques are chosen according to their capacity and they are approached with kindness and consideration. We recommend early and regular eye examinations for all children with Down syndrome to diagnose problems and provide appropriate intervention. This may improve the academic performance and overall quality of life of these individuals.

Data sharing statement provided by the authors is available with the full text of this article at jebmh.com.
Financial or other competing interests: None.
Disclosure forms provided by the authors are available with the full text of this article at jebmh.com.

REFERENCES

[1] Down JL. Observations on an ethnic classification of idiots. 1866. Ment Retard 1995;33(1):54-56.
[2] Akinci A, Oner O, Bozkurt OH, et al. Refractive errors and strabismus in children with Down syndrome: a controlled study. J Pediatr Ophthalmol Strabismus 2009;46(2):83-86.
[3] Wong V, Ho D. Ocular abnormalities in Down syndrome: an analysis of 140 Chinese children. Pediatr Neurol 1997;16(4):311-314.
[4] Kim JH, Hwang JM, Kim HJ, et al. Characteristic ocular findings in Asian children with Down syndrome. Eye (Lond) 2002;16(6):710-714.
[5] Solomons G, Zellweger H, Jahnke Pg. Four common eye signs in Mongolism. Am J Dis Child 1965;110:46-50.
[6] Wagner RS, Caputo AR, Reynolds RD. Nystagmus in Down's syndrome. Ophthalmology 1990;97(11):1439-1444.
[7] Lowe RF. The eyes in mongolism. Br J Ophthalmol 1949;33(3):131-174.
[8] Stephen E, Dickson J, Kindley AD, et al. Surveillance of vision and ocular disorders in children with Down syndrome. Dev Med Child Neurol 2007;49(7):513-515.
[9] Fimiani F, Iovine A, Carelli R, et al. Incidence of ocular pathologies in Italian children with Down syndrome. Eur J Ophthalmol 2007;17(5):817-822.
[10] Haugen OH, Høvding G. Strabismus and binocular function in children with Down syndrome. A population-based, longitudinal study. Acta Ophthalmol Scand 2001;79(2):133-139.
[11] Ebeigbe JA, Akpalaba R. Ocular health status of subjects with Down's syndrome in Benin City, Nigeria. Afr J Med Med Sci 2006;35(3):365-368.
[12] Wagner RS, Caputo AR, Reynolds RD. Nystagmus in Down's syndrome. Ophthalmology 1990;97(11):1439-1444.
[13] Roizen NJ, Patterson D. Down's syndrome. Lancet 2003;361(9365):1281-1289.