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Ophthalmic Manifestations in Childrens Presenting With Down Syndrome

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Abstract

Purpose - *To study the ophthalmic manifestations in childrens presenting with Down syndrome.*

Methods- *This was a prospective observational study that involved 30 eyes of 15 childrens of Down syndrome presenting with low visual acuity, strabismus, nystagmus, blephritis, large epicanthal folds, nasolacrimal duct obstruction , etc.*

Results-*There were 9 males and 6 females and the age group taken was 1 to 10 years.*

Most common presentation in down syndrome patients is low visual acuity (<6/18) in 74% patients followed by strabismus in 68 % patients , nystagmus in 52% patients , upward slanting palpaberal fissure in 51% patients , blephritis in 34% patients, nasolacrimal duct obstruction in 32% patients ,brushfield spots in 14% patients. Most common refractive error in down syndrome children is myopia in 46% patients followed by astigmatism in 32% patients and hyperopia in 22% patients. Fundus finding are rare in downs syndrome patient but include cupping of disc and optic atrophy. Other features of down syndrome are cerebral palsy, autism , hypertelorism, flat nasal bridge, alopecia and macroglossia.

Conclusion - *Down syndrome is common genetic disease presenting usually with multiple systemic features. Most common ocular manifestation is low visual acuity followed by strabismus, nystagmus , blephritis , nasolacrimal duct obstruction and brushfield spots.*

Most common refractive error is myopia but usually difficult to correct due to nystagmus and strabismus. Assessment and regular follow-up by the pediatric ophthalmologist is important to detect abnormalities otherwise patient may develop amblyopia.

Keywords: *down syndrome, low visual acuity, strabismus ,myopia*

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I. Introduction

Down syndrome is the most common chromosomal abnormality [1]. Down syndrome occurs in approximately 1 in 700 births, and the frequency rises with maternal age due to an increase in nondisjunction events. Roughly 5,400 children with Down syndrome are born each year in the United States.[2]

The reported prevalence of ophthalmic disorders in Down syndrome patients ranges from 46% to 100%.[3] The incidence of associated eye disease requiring monitoring or intervention increases with age, from 38% of Down syndrome children < 1 year old to 80% of 5 to 7 year-old children.[4]

Ophthalmic Features

Strabismus

Strabismus, especially esodeviation, is common in children with Down syndrome. In most of the patients strabismus type is esotropia, although in few children of Down children with strabismus were found to have exotropia [5] Only one study of Down syndrome children mentions hyperdeviation .[6]

Nystagmus

Nystagmus is common presentation of children with down syndrome and usually it is rapid horizontal nystagmus[7] Eighty to 89% of Down syndrome children with nystagmus have a manifest nystagmus while a smaller cohort have a latent or manifest-latent nystagmus. Nystagmus in Down syndrome children has been associated with lesser visual acuities.[8]

Refractive Error and Astigmatism

Down syndrome children have a higher prevalence of myopia , hyperopia and astigmatism. Failure of emmetropization has been proposed as the etiology of the increase in prevalence of refractive error after

infancy[9]. In infants up to 12 months old without Down syndrome, clinically significant astigmatism is common. The prevalence of astigmatism then declines by 4 years of age [10,11] . But in down syndrome children astigmatism continues to increase with increasing age.

Infants with Down syndrome generally have "with the rule" astigmatism. The type of astigmatism often changes to an oblique type of astigmatism.[12,13,14] The development of oblique astigmatism has been proposed to be caused by the oblique, upslanting palpebral fissues in Down patients as the axis of astigmatism has been correlated with the angle of the palpebral fissure.

Visual Acuity

Best corrected visual acuity (BCVA) in Down syndrome patients is on average less. There are multiple causes for this decrease in BCVA. One major cause is amblyopia followed by strabismus is also common in Down syndrome, early and regular assessment and treatment can help minimize visual loss due to amblyopia. Nystagmus may also account for a substantial extent of visual acuity loss. Other common causes of decreased visual acuity in Down syndrome patients include cataracts and high refractive errors.

Accommodation

Poor accommodative ability has been found in Down syndrome children. Using dynamic retinoscopy, studies have found that of Down syndrome children have an abnormally low ability to accommodate, inaccurate accommodation has been found to be associated with hypermetropia and strabismus[13].

Eyelids and Midface

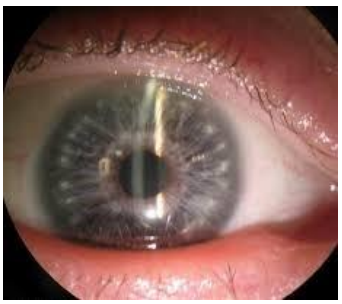
The most common findings are upward slanting palpebral fissures and large epicanthal fold. Blepharitis is also a common finding in Down syndrome patients. The high rate of blepharitis has been speculated to be due to impaired immune or due to skin abnormalities in Down syndrome individuals. Other disorder of the eyelids include epiblepharon, which is less common.

Lacrimal

Lacrimal duct obstruction is common with bilateral involvement in most of patients. The higher rate of nasolacrimal duct obstruction is related to the facial morphology of Down syndrome patients and may be due to an abnormal persistence of a membrane or bony obstruction blocking the nasolacrimal duct[15].

Iris

Brushfield spots are white, gray or brown spots spaced around the periphery of the iris. They are focal areas of iris stromal tissue connective tissue hyperplasia which are of no functional importance. They are not pathognomonic for Down syndrome and can be seen in children without disease. Brushfield spots are more common on blue, green or hazel irides compared to dark irides.



Lens

The frequency of congenital cataracts in Down syndrome patients was less than 1%, and the frequency of developing a cataract before age 18 was 1.4%. Cataract is another important cause for low visual acuity.

Retina

A common retinal finding in Down syndrome is a spoke-like appearance with a high number of vessels crossing the margin of the optic nerve. Although not associated with vision loss.This has been proposed to be a consequence of a deficiency in systemic angiogenesis. Retinoblastoma has also been speculated to be connected to Down syndrome, possibly caused by oncogenic genes overexpressed on chromosome 21, although there have only been a few recorded cases of this disease in Down patients.

Optic Nerve

Optic nerve anomalies in Down syndrome are rare. Optic disk elevation and in some cases optic nerve hypoplasia has been reported , but may represent coincidental findings.

II. Method And Material

This was a prospective observational study that involved 30 eyes of 15 patients of down syndrome presenting with low visual acuity, strabismus, nystagmus, blephritis, large epicanthal folds, nasolacrimal duct obstruction. Patients were recruited from the OPD of MLB MEDICAL college, Jhansi ,Uttar Pradesh and were followed from 1st july 2020- 1st march 2021 . It was performed under the Helsinki Declaration of 1975, as revised in 2000. The necessary permission from the Ethical and Research Committee was obtained for the study.

Inclusion criteria

1. All patients between the age group 1 to 10 years who presented to the OPD of MLB medical College Jhansi with diagnosed case of down syndrome presenting with low visual acuity, strabismus, nystagmus, blephritis, large epicanthal folds, nasolacrimal duct obstruction

Exclusion criteria

1.Patients outside the age group of 1 to 10 years.
2.Patients with any other corneal pathology.
3.Patients with other conjunctival diseases.
4.Patients with any other ocular pathology.
5.Patients with history of ocular trauma.

All patients were subjected to a detailed history taking, complete ophthalmic examination in diffuse and focal light .

III. Results

A total of 30 eyes of 15 down syndrome patients were studied. We included eyes with a complaint of low visual acuity, strabismus, nystagmus, blephritis, large epicanthal folds, nasolacrimal duct obstruction. There were 9 males and 6 females and 60% of the studied eyes were right eyes.

Table2 :Type of refractive error in childrens of down syndrome

	Refractive error	% of patients
1	Myopia	46
2	Astigmatism	32
3	Hyperopia	22

Table1: Ocular manifestation in childrens of down syndrome

	Manifestation	% of patients
1	Low visual acuity (<6/18)	74
2	Strabismus	68
3	Nystagmus	52
4	Upward slanting palpaberal fissure	51

5	Blephritis	34
6	Nasolacrimal duct obstruction	32
7	Brushfield spots	14

IV. Discussion

A spectrum of ocular anomalies is prevalent in DS and has been described extensively in the literature across different racial groups. Liyanage S, Barnes J et al. have established that refractive errors and strabismus are more prevalent among children and young adults with down syndrome. In children with down syndrome, heart defects have been reported to be associated with myopia, although the mechanism is not clear. For refractive errors, our finding of high prevalence of myopia is comparable to findings with Tsiaras WG, Pueschel S, Keller C, Curran R, Giesswein S study on Amblyopia and visual acuity in children with Down's syndrome. The high incidence of refractive errors among children with down syndrome is believed to be caused by failure of emmetropisation. Visual acuity was improved to a statistically significant degree in the down syndrome group after refraction . The prevalence of strabismus in the down syndrome children in our study was higher than findings reported by Paudel et al, [16] but lower than in other studies.As in the current study, esotropia has been reported to be more prevalent than exotropia in down syndrome children. Bromham et al [17] reported that nystagmus was a feature of down syndrome in his series. Kim et al showed prevalence of conjunctivitis and blepharoconjunctivitis in his study while there were no significant cases of conjunctivitis in our study.. Early cataract formation, which may reflect the faster aging process in many persons with down syndrome was not prevalent in our sample. Cupping of the discs and optic atrophy, however, were less prevalent in the down syndrome.

V. Conclusion

Down syndrome is common genetic disease presenting with multiple systemic features. Most common ocular manifestation is low visual acuity followed by strabismus, nystagmus , blephritis , nasolacrimal duct obstruction and brushfield spots.Most common refractive error is myopia.Our study spans over a period of 9 months and is prospective in nature focusing on ophthalmic manifestations of childrens of down syndrome. Childrens should be evaluated for strabismus, cataracts, and nystagmus along with visual acuity testing within the first 6 months of life followed annually until age 5 with evaluation for refractive error, strabismus, or other conditions which could result in amblyopia followed biannually from 5-13 years old to evaluate for the onset of new ophthalmic disorders.

References:

[1]. Malt EA, Dahl RC, Haugsand TM, et al. Health and disease in adults with Down syndrome. Tidsskr den Nor Laegeforening 2013;133:290–294.
[2]. Sherman SL, Allen EG, Bean LH, Freeman SB. Epidemiology of Down syndrome. Ment Retard Dev Disabil Res Rev 2007;13:221–227.
[3]. Creavin AL, Brown RD. Ophthalmic abnormalities in children with Down syndrome. J Pediatr Ophthalmol Strabismus 2008;46:76–82
[4]. Roizen NJ, Patterson D. Down's syndrome. Lancet 2003;361:1281–1289.
[5]. Fimiani F, Iovine A, Carelli R, et al. Incidence of ocular pathologies in Italian children with Down syndrome. Eur J Ophthalmol 2007;17:817–822.
[6]. Da Cunha RP, Moreira JB. Ocular findings in Down's syndrome. Am J Ophthalmol 1996;122:236–244.
[7]. Ljubic A, Trajkovski V, Stankovic B. Strabismus, refractive errors and nystagmus in children and young adults with Down syndrome. Ophthalmic Genet 2011;32:204–211.
[8]. Felius J, Beauchamp CL, Stager DR. Visual acuity deficits in children with nystagmus and down syndrome. Am J Ophthalmol 2014;157:458–463.
[9]. Doyle SJ, Bullock J, Gray C, et al. Emmetropisation, axial length, and corneal topography in teenagers with Down's syndrome. Br J Ophthalmol 1998;82:793–796.
[10]. Mohindra I, Held R, Gwiazda J, Brill J. Astigmatism in infants. Science 1978;202:329–331.
[11]. Gwiazda J, Scheiman M, Mohindra I, Held R. Astigmatism in children: Changes in axis and amount from birth to six years. Investig Ophthalmol Vis Sci 1984;25:88–92.
[12]. Antonela L, Vladimir T. Refractive errors in children and young adults with Down's syndrome. Acta Ophthalmol 2011;89:324–327.
[13]. Haugen OH, Høvding G, Lundström I. Refractive development in children with Down's syndrome: a population based, longitudinal study. Br J Ophthalmol 2001;85:714–719.
[14]. Cregg M, Woodhouse JM, Stewart RE, et al. Development of refractive error and strabismus in children with Down syndrome. Investig Ophthalmol Vis Sci 2003;44:1023–1030
[15]. Baran F, Kelly JP, Finn LS, et al. Evaluation and treatment of failed nasolacrimal duct probing in Down syndrome. J Am Assoc Pediatr Ophthalmol Strabismus 2014;18:226–231.
[16]. Paudel N, Leat SJ, Adhikari P, Woodhouse JM, Shrestha JB. Visual defects in Nepalese children with Down syndrome. Clin Exp Optom. 2010;93:83–90. [PubMed] [Google Scholar]
[17]. Bromham NR, Woodhouse JM, Cregg M, Webb E, Fraser WI. Heart defects and ocular anomalies in children with Down's syndrome. Br J Ophthalmol. 2002;86:1367–1368. [PMC free article] [PubMed] [Google Scholar]