

Anomalies of Refraction, Accommodation and Binocular Single Vision in Down Syndrome

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Abstract:

Down syndrome is one of common chromosomal abnormalities. Many ophthalmic features are studied and reported previously. The aim of this research is to investigate and thoroughly study the anomalies of refraction, accommodation and binocular single vision (BSV) in Down syndrome along with strabismus and ocular abnormalities.

Methods:

Total of 40 children with Down syndrome, age ranging from 6 years to 14 years old underwent ocular assessment including visual acuity assessment, non Cycloplegic refraction, ophthalmoscopy, ocular alignment and ocular motility tests and tests for binocular single vision.

Results:

Among population of 40 children with Down syndrome. 40% children had hypermetropia and 37.5% had Astigmatism and 5% had myopia being least common. Esotropia being the most common deviation with 60% population size. Binocular single vision was found to be effected as well. It was completely absent in 12.5% of population where as 25% population had only grade I presented whereas 17.5 had grade II presented. Other ocular finding were also noted and prevalence's as Nystagmus in 15%, Cataract in 25%, Blephritis 10%, conjunctivitis in 12.5%, keratoconus 5% and iris abnormalities in 10% population.

Conclusions:

In our study, the anomalies of refraction were found to be hyperopia and astigmatism. The

reduce near point of accommodation was noted hence reduced amplitude of accommodation is identified where as weak accommodation should take in to consideration when dealing with Down syndrome. Binocular single vision is also found to be disturbed in the presence of strabismus. Whereas other ocular abnormalities like cataract, nystagmus and blephritis must be also considered. So further studies are required to identify other anomalies which can be related to vision and must be diagnosed and treat with best treatment regime available.

Key Words:

Down syndrome, Refractive error, Binocular single vision, Accommodation.

Introduction:

Down syndrome is among most common genetic disorders in which a person has 47 chromosomes with an extra chromosome at pair 21. This additional chromosome hinders normal body development and leads to abnormal physical, intellectual and medical development in a person¹. It occurs in every 1 out of 700 live births². Overall Down syndrome is further divided in 3 types based on the reasons behind mutation occurring at 21st pair of chromosome. Among all, 95% of Down syndrome's population has trisomy at pair 21 (three pair of chromosomes instead of two). This form of mutation occurs due to non-disjunction at the 21st pair of chromosome in either mother's ovum or father's sperm. Another 4% may carry translocation Down syndrome which occurs due to attachment of 21st pair of chromosome in the

cell. The remaining 1% has mosaic behavior for chromosome 21 which means that only some of their cells will contain trisomy at 21st pair of chromosome whereas in very rare conditions a very small portion of chromosome 21 is duplicated³. Where in Pakistan, no such study is available for prevalence but it was estimated to be 0.2% out of 1000 women to have children with Down syndrome in Karachi city only⁴.

There are certain factors that lead to trisomy at 21 Chromosome. The most common factor for giving birth to a child with Down syndrome is maternal age. 1 out of 350 women aged 35 and 1 out of 25 women aged 45 or above are at risk of delivering a baby with Down syndrome⁵.

Apart from age there are some more factors that are related to mothers. These factors are fertility regulations, previous abortions, maternal mortality sexually transmitted disease and infertility. Whereas spontaneous abortions and history of many still births increase the risk for chromosomal non disjunctions. These risk increases because of lack of proper diet and nutrition during pregnancy. Besides these known risk factors, consanguinity, region (Rural/Urban) of residence of parents, exposure of parents to chemicals, education status of parents, habits of father like smoking and alcohol consumption, prenatal scanning and reproductive performance of mother are also the possible risks for chromosomal mutation⁶. The mutation at chromosome pair 21 leads to different appearance of child than normal children. Exhibiting the typical features for Down syndrome can vary among children. The most common physical features are: low muscle tone, the child appears to be floppy, flat facial features including small skulls flat nasal bridge and flat back part of head, almond shaped eyes (upward slanting) with epicanthal folds, small ears, single deep crease across the center of palm, enlarged tongue that tends to stick out usually⁸, presence of excessive skin at nape of the neck and separated sutures between bones of

skull and smaller mouths⁹.

Down syndrome is a multi-system disorder but the extent to affect different organ systems can vary from person to person. The major medical conditions that are associated with Down syndrome are heart defects, skeletal defects, obesity, and ocular diseases¹⁰.

Ocular diseases are also common in people with Down syndrome. About 60% of DS population represents with ophthalmic manifestations¹⁵. The ocular anomalies in Down syndrome have significant manifestations, including refractive errors, binocular vision disorders, strabismus, accommodation anomalies, eyelid abnormalities, glaucoma and retinal disorders¹⁶. The prevalence for refractive error including myopia and astigmatism tends to be 25.3%, cataract includes 42.0%, conjunctivitis includes 13.4%, strabismus including esotropia and exotropia tends to be 21.1% and blephritis included 10.1%¹⁷.

Studies have suggested that many ocular anomalies are associated with Down syndrome and proper evaluation is mandatory before applying any management strategies. A study was conducted by Han, Kim and Paik in Korea to examine the characteristics of refractive error and strabismus in Down syndrome in Korean population. This study suggested that Esotropia and hypermetropia were more common in patients with Down syndrome. Whereas hypermetropia and accommodation weakness were suggested to be root cause of Esotropia. They thus advised to add the measurement of near point of accommodation to routine examination when dealing with children with down syndrome.

A similar research was done in Japan by Morton was to identify characteristics and ocular findings in children with Down syndrome. The outcomes implied that Hypermetropia and astigmatism were common refractive errors. Strabismus, especially esotropia was found in majority of children and other ocular findings were cataract, corneal disease, nystagmus, entropion of eyelids

and chorioretinal degeneration were also found. This study proposed that best ophthalmic investigation is important for children with Down syndrome at the time of infancy.

Methods:

Patients were recruited from Shaadab training Institute for Mentally Retarded children, Iqbal Town Lahore from 23 October, 2015 to 30 November, 2015. Inclusion criteria was children with Down syndrome age ranging from 5 to 25 years old. Exclusion criteria was any history of trauma that had effect on eyes, DS children with severe mental retardation and age less than 5 years and greater than 25 years. Visual Acuity was assessed through Snellen charts and Kay picture charts. Non Cycloplegic retinoscopy was performed for studying anomalies of refractive errors, accommodation was assessed with push up tests and amplitude of accommodation was determined according to age and near point of accommodation. Grades of Binocular Single vision were assessed with wearing best refractive correction. Strabismus was examined with Hirschberg test which is based on corneal reflection test and cover/uncover test was performed to identify both latent and manifest squints. Ophthalmoscopy was performed for other ocular findings that can reduce ocular performance.

Results:

Table 1: Percentage distribution of Refractive Errors

Type of Refractive Errors	Frequency	Percent
'Myopia'	2	5.0
'Hypermetropia'	16	40.0
'Astigmatism'	15	37.5
'Emmetropia'	7	17.5
Total	40	100.0

Table 1: Shows that among 40 subjects, 25%

showed Myopia, 40% showed Hyperopia, 37% showed Astigmatism and 17.5% showed Emmetropia.

Table 2: Percentage distribution of Amplitude of Accommodation

Amplitude of Accommodation	Frequency	Percent
Less than Normal Range	18	45.0
Within Normal Range	11	27.5
Greater than Normal Range	11	27.5
Total	40	100.0

Table 2 Highlights that among 40 subjects, 45% subjects had amplitude of accommodation Less than Normal Range, 27.5% had amplitude of accommodation Within Normal Range and 27.5% had amplitude of accommodation Greater than Normal Range.

Table 3: Percentage distribution of Binocular Singular Vision Grades

Binocular Singular Vision	Frequency	Percent
Simultaneous Perception	10	25.0
Simultaneous Perception, Fusion	7	17.5
Simultaneous Perception Fusion Stereopsis	18	45.0
No BSV	5	12.5
Total	40	100.0

Table 3: shows that among 40 subjects, 25% subjects had only Simultaneous Perception, 17.5 % had Simultaneous Perception and Fusion, 45% had Simultaneous Perception, Fusion and Stereopsis and 12.5% had No BSV.

Table 4: Percentage distribution of Types of Strabismus

Strabismus	Frequency	Percent
No Deviation	7	17.5
Esotropia	2	60.0
Exotropia	4	15.0
Esophoria	6	2.5
Exophoria	12	5.0

Table 4: shows that among 40 subjects, 17.5% had No Deviation, 60% had Esotropia, 15% had Exotropia, 2.5% had Esophoria and 5% had Exophoria.

While other ocular findings include Nystagmus (15%), Cataract (25%), Blephritis (10%), Conjunctivitis (12.5%), Keratoconus (5%), Iris abnormalities (10%) and 22.5% subjects had no ocular abnormalities.

Discussion:

Down syndrome is associated with ocular anomalies. Visual Acuity is found to be reduced in children with Down syndrome through Snellen chart. The chart selection was taken according to cooperation and familiarity level of child. Both eyes showed significant reduced visual acuity in both eyes. About 60% population has visual acuity less than 6/18. The reduced visual acuity is due to high refractive errors and which are often associated with Nystagmus and squint 56. Refractive errors in Down syndrome are very common. Upon non-Cycloplegic refraction, 82.5% were found to have refractive errors where as 17.5% has no any refractive error. Among 33 children the results came out to be slightly dual. Incidence of both Astigmatism and Hypermetropia were in higher but Hypermetropia was found to be slightly higher as 40% whereas Astigmatism as 37.5%. Whereas myopia was found in very little population which is about 5%. Thus refractive errors must be watched out carefully when dealing with children with Down syndrome¹⁰. Accommodation was thoroughly observed in our study. Near point of Accommodation was evaluated through pushup examination through RAF rule. The normal ranges

for near point of accommodation were defined according to age so as amplitude of accommodation was also evaluated according to age. According to Elkington AR58, the new born infant has amplitude of accommodation around 14 diopters which is gradually reduces to 1 Diopters at 60 years. While observing amplitude of accommodation in Down syndrome the reduction in amplitude was not gradual. 45% children had amplitude of accommodation less than 9 Diopters who are less than 25 years of age. This marked reduction was explained by Haugen, Hovding and Eide⁵⁹ that central lens was thinner in population with Down syndrome when compared with control group thus effecting mark reduction in power of lens but when mechanism of lens zonules and ciliary muscles were studied that were exactly according to normal control group thus they suggested that expected lag in accommodation was based solely on decrease in power of lens. However this explanation is further assisted by the characteristics of predisposition to earlier presbyopia due to changes in crystalline lens that can be structural as well as mechanical changes. The marked reduction in Amplitude of accommodation has to go through further research and investigation to find associated anomalies of accommodation¹¹. Binocularity found to be effected in Down syndrome as well. Binocular Single vision was assessed through all its grades. Simultaneous perception was examined from Visual acuity assessment and worth 4 dot test for both near target and distance targets whereas fusion was examined through worth four dot test and stereopsis was performed through Lang II pencil test and Titmus fly test. These all tests were performed with child wearing proper correction. Among sample population, 45% children had Normal retinal correspondence and all grades of Binocular single vision were presented and 12.5% population had no BSV at all and which was found to be related with presence of significant deviations. 25% population has only

simultaneous perception presented and remaining 17.5% population size had both simultaneous perception and fusion presented but stereopsis was missing.

Binocular single vision is much effected by strabismus and this is evident through extra ocular moments performed in all diagnostic position and through Hirschberg test, cover uncover test and alternate cover test. Among our sample population, 17.5% population had no strabismus and eyes were straight. Esotropia is very common and found to be 60% of population and Exotropia was found in 15% population. According to Yurdakul NS *et al.*, 60 Esotropia is common in people with Down syndrome along with higher incidence of hypermetropia¹².

It has been shown that reduction in vision in Down syndrome is also due to certain ocular conditions. During our study ophthalmoscopy was performed and ocular examination was done. Cataract was among 25% of population, Nystagmus was seen in 15%, both blephritis and Iris abnormalities were found in little percentage of 10% each whereas conjunctivitis was found in 12.5% and 5% in keratoconus remaining 22.5% had no ocular problems¹³. Cataract being very common in the population sample and was very common in older ages than younger children in population sample. Blepharitis and conjunctivitis being inflammatory disorders of eye were also common in children with Down syndrome. The increment in susceptibility to infections of eyes is often associated with impact of trisomy 21 on immune system¹⁴.

All of the above discussion concluded that Down syndrome is more than often associated with refractive errors, reduced accommodation, abnormal binocular single vision, manifest strabismus and other ocular anomalies. These anomalies are the reason for decrease visual performance and they have to be managed at early age of child¹⁵.

Conclusions:

The results from this study have important implications with respect to the ophthalmic care of Down syndrome children. The higher prevalence of ophthalmic disorders have indicated the periodic evaluations of children with Down syndrome especially right after birth and regular examinations must be conducted because this study has confirmed the presence of ocular abnormalities in children with Down syndrome.

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