

# Article ▶ Ocular Morbidity In Children With Autism

Sonisha Neupane, BOptom, Mechi Eye Institute, Birtamod, Mechi, Nepal

Gopal Bhandari, BOptom, Bharatpur Eye Hospital, Chitwan, Nepal

Gauri Shankar Shrestha, MOptom,

B. P. Koirala Lions Centre For Ophthalmic Studies, Maharajgunj Campus, Kathmandu, Nepal

## ABSTRACT

**Background:** Autism Spectrum Disorder is a range of complex neuro-developmental disorders characterized by social impairment, communication difficulty, and restricted, repetitive, and stereotyped patterns of behavior. Children on the autism spectrum exhibit variable refractive errors, strabismus, oculomotor dysfunction, and atypical gaze. This project was an attempt at early identification of, and intervention for, ocular and visual abnormalities in children on the autism spectrum.

**Methods:** Thirty-six children from four rehabilitation centers in Nepal underwent detailed optometric examinations including refractive, oculomotor, binocular, and disease evaluations.

**Results:** Visual abnormalities were seen in 24 (66%) children that included myopia, hyperopia, and astigmatism. Strabismus was present in eight (22%) children. Amblyopia was present in 11 (31%) children. Amblyopia due to refractive error and strabismus was present in nine (25%) and two (6%) children, respectively. Nystagmus was seen in only one (3%) patient.

**Conclusion:** The relatively high prevalence of visual disorders in this group would indicate that the visual needs of patients on the autism spectrum must be addressed. The need for early and regular eye examinations is warranted.

**Keywords:** amblyopia, autism spectrum disorder, ocular morbidity, refractive error, strabismus

## Introduction

Autism was first described by child psychiatrist Leo Kanner in 1943.<sup>1-3</sup> Since the 1980s, autism has been recognized as a neuro-developmental disorder defined by impairment in social interaction and communication with restricted, repetitive, stereotyped patterns of behavior, interests, and activities.<sup>1,3</sup> Many systemic conditions are linked with autism, including associations with intellectual disability, epilepsy, disruptive behavior, and learning difficulty.<sup>1</sup> Depending upon the author and which paper is read, the autism spectrum has been expanded to include: Autism, Asperger's syndrome, Pervasive Developmental Disorders-not otherwise specified (PDD-NOS), Rett Syndrome, Childhood Disintegrative Disorder, Semantic Pragmatic Disorder, non-verbal learning disabilities, and even some form of Attention Deficit Hyperactivity Disorder.<sup>4,5</sup>

Autism typically appears during the first three years of life. It affects normal functioning of the brain and has an impact on development of social interaction and communication skills. Both children and adults with autism typically show difficulties in verbal and non-verbal communication, social interactions, and leisure or play activities.<sup>6</sup> There is controversy regarding the prevalence of autism. According to the U.S. Department of Education, both the prevalence and incidence of autism are on the rise.<sup>7</sup> Recent studies suggest that the prevalence of autism is one in every 110 births in the

United States and almost one in 70 boys.<sup>6,7</sup> Autism is three to four times more likely to occur in boys than girls.<sup>6,8,9</sup> Multiple factors, including organic, environmental, and genetic causes, have been implicated. Many abnormalities involving the brain have been documented in the autistic population, while possible etiologies of autism include vaccines and genetics.<sup>1,10-13</sup> In a case studied by Taub, the US government agreed to pay a settlement to parents who claimed that vaccination had caused their child to develop autism with the rationale that the vaccine the daughter received at 18 months of age significantly aggravated an underlying mitochondrial disorder that manifested with the features of autism spectrum disorder.<sup>14</sup>

Using the Diagnostic and Statistical Manual (DSM-IV) system, clinicians evaluate three main categories of behavior:<sup>15,16</sup> 1) impairment in social interaction, 2) impairment in communication, and 3) repetitive, stereotyped behavior patterns. A patient can only be considered to have one of the Autism Spectrum Disorders (ASD) if abnormal functioning is observed in at least one of these three areas prior to age three.

Children with autism may be difficult to examine due to limited communication skills and unpredictable behavior. They often exhibit atypical gaze or gaze avoidance and manifest stereotypical behavior such as eye pressing, hand flicking, and light gazing.<sup>16-20</sup> Refractive error and visual acuity are highly variable in this patient base. Strabismus is often present, and pursuits and saccades have been found to be deficient.<sup>16-22</sup>

The absence of eye contact, unresponsiveness to facial gesture, and/or difficulty in sharing joint visual attention are signs of abnormal or atypical visual development. A comparative study with autistic children and a normal cohort showed that looking at the eyes of others was significantly decreased in children with autism, and fixation time on eyes by the children with autism correlated with their level of social disability.<sup>23</sup>

Denis et al.<sup>19</sup> reported on 10 autistic children (six girls and four boys) ranging in age from one to 13 who underwent a detailed ocular examination. Refraction showed hyperopia in seven cases and astigmatism of more than 1D in six cases. The axis of astigmatism was oblique for eight eyes, with-the-rule for six eyes, and against-the-rule for two eyes. Strabismus was present in six cases (four exotropia, two esotropia). Fundus examination showed pallor of the optic disc in four cases.

Scharre and Creedon<sup>18</sup> evaluated 34 autistic children, ages two to 11 years, for ocular alignment, refractive error, visual acuity, ocular motility skills, and stereopsis. None of the children manifested ocular disease, known seizure disorders, or dysmorphic features. Their developmental levels ranged from average intelligence to severely intellectually disabled. Refractive errors ranged from -4.25 to +3.25 D with a near retinoscopy technique. Of the 34 children, 21% were strabismic at distance and 18% were strabismic at near. The Lang stereo test was attempted on all children and completed on 17. Of the 17, all but three exhibited 550 sec arc or better. Only 15% exhibited voluntary pursuit movements, but all the children demonstrated saccadic fixations. Thirty-one children had atypical optokinetic nystagmus (OKN) responses such as delayed onset, short duration, gaze avoidance, or stereotypic behavior. With repeated testing consistent visual responses on OKN and visual acuity were noted.

Elizabeth et al.<sup>24</sup> examined 51 children with autism (44 males, eight to 18 years old) and 44 typically developing controls (13 males, eight to 17 years old). Visual acuity, stereoacuity, vergence, convergence, prism fusion range, near point of convergence, strabismus, ocular motility, and optokinetic response were tested. Thirty-one percent of the children with autism and 11% of the typically developing children had some type of visual impairment. Children with autism displayed significantly poorer visual acuity (although within normal limits) and receded near point of convergence.

The current study was designed to detect ocular abnormalities in Nepalese children who exhibit autistic behaviors with the thought that early intervention to treat their visual anomalies would result in increased sensorimotor skills. Better sensorimotor skills should enhance the quality of life for these individuals.

Although similar studies have been performed with children diagnosed with Down syndrome and cerebral palsy, no such study of the visual defects of Nepalese children with autism has been performed. It is hoped that this study may

serve as a guideline for the visual management of subjects on the autism spectrum.

## Methods

This was a cross-sectional study conducted among children with autism from November 2009 to October 2010 in four rehabilitation centers in the Kathmandu Valley in Nepal. The centers enrolled a total of 36 subjects. All the children were identified as having autism by a pediatrician. Exclusion criteria included: parents who were not willing to allow their children to participate in the study and children who needed general anesthesia for ocular examination. The children were examined with their caretakers nearby, and informed consent was obtained from the guardians and teachers after the details of the study were explained.

Patient demographics including age, gender, and race were recorded with a research form designed for this study. A brief history, including any visual and ocular complaints, previous eye examination, and use of glasses and medications, was recorded.

Visual acuity was assessed binocularly with an OKN drum of 3 cycles/degree rotated at a frequency of approximately 20-50 m/sec. If the child was able to follow the stripes, visual acuity was considered better than 6/60 (20/200).<sup>25</sup> If the child was not able to follow the stripes, the visual acuity was considered to be worse than 6/60 (20/200). For those who did not respond to OKN, the ability to maintain central steady maintained fixation (CSM) was judged and recorded. We attempted other methods of visual acuity assessment (preferential looking chart, Kay-picture chart), but none of the subjects responded.

Ocular motility was tested by moving a colorful object or penlight in the six different cardinal gazes. In unresponsive subjects, the doll's head tilt technique was used to assess ocular motility. The Hirschberg reflex was assessed to detect the presence of strabismus. The laterality and constancy of deviation were noted. The magnitude of deviation was estimated. Nystagmus, if present, was noted as to type and direction.

Cycloplegic retinoscopy was performed on every subject at a working distance of 50cm. One drop of 1% cyclopentolate was instilled three times with the interval of 10 minutes in between. Retinoscopy was performed 45-60 minutes after the instillation of the first drop, ensuring full cycloplegia (Heavener's recommended dose-reference).<sup>26,27</sup> Refractive error was categorized as:<sup>28</sup> myopia if the spherical equivalent was  $\geq -0.50D$ , hyperopia if the spherical equivalent was  $\geq +1.00D$ , and astigmatism for  $\geq 1.00 D$ . Subjective refraction was attempted but not completed due to the limited cooperation of the subjects.

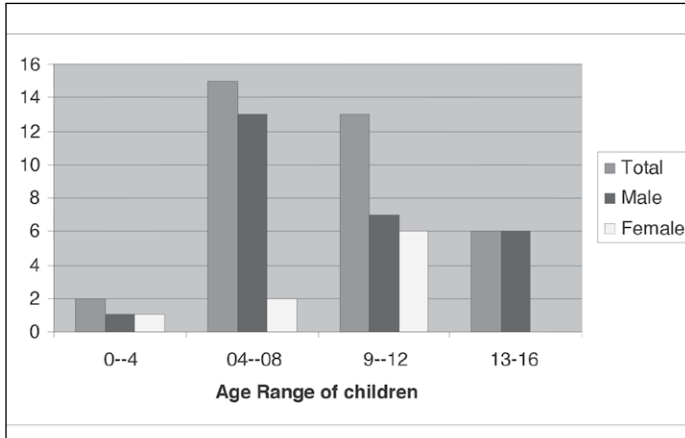
A hand-held biomicroscope was used to examine the anterior segment. A penlight was used for uncooperative subjects. Posterior segment evaluation was performed using a direct ophthalmoscope.

**Table 1: Prevalence of ocular morbidities**

Findings	Total	Gender		Ethnic Group		Age			
		Male	Female	Caucasian	Mongolian	1-4	5-8	9-12	13-16
Congenital Bilateral Ptosis	1	1	0	0	1	0	1	0	0
Chalazion	1	0	1	0	1	0	0	1	0
Horizontal Pendular Nystagmus	1	1	0	1	0	0	1	0	0
Strabismus	8	5	3	5	3	2	2	2	2
Refractive Error	21	15*	6**	15	6	2	9	7	3

\*male 15 (refractive error only-10, refractive error associated with strabismus-4, refractive error associated with nystagmus-1)

\*\*female 6 (refractive error only-3, refractive error associated with strabismus-3)



**Figure 1: Age and gender distribution** Ocular abnormalities were seen in 24 subjects. A description by age, gender, and race can be found in Table 1. The posterior segment was normal in all children.

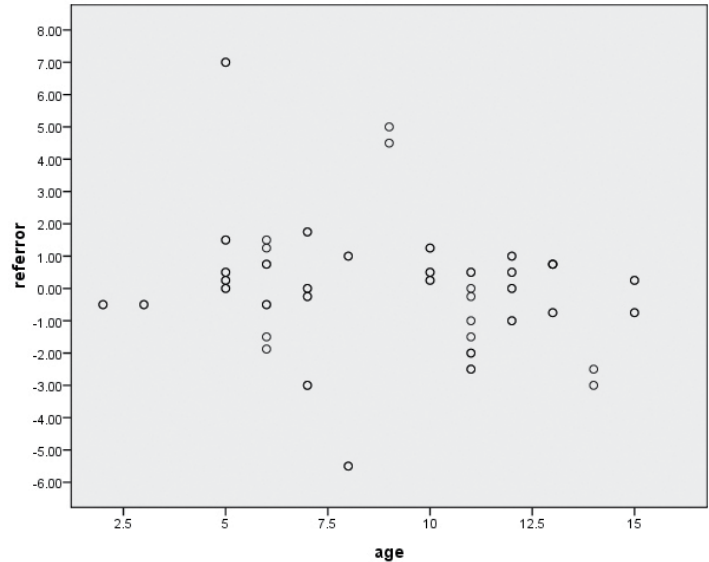
These data were entered into Statistical Package for Social Sciences (SPSS) version 16.0 and Microsoft Excel 2010, and relevant data were analyzed using appropriate descriptive statistical tools.

## Results

Thirty-six total patients were enrolled in the study (nine female and 25 male). The age of the subjects ranged from two to 15 years with a mean age of nine years (Figure 1). The majority of the autistic children were Caucasian (25), and the others were Mongolian (11). Only two had received previous eye care.

Refractive error was the most prevalent finding, occurring either alone or in association with other abnormalities. Significant refractive error was seen in 21 subjects. Two subjects were found to have anisometropia that ranged between 1.00 D and 2.00 D. Figure 2 portrays the refractive condition by age.

Myopia and myopic astigmatism were the most common type of refractive error, occurring in 28 (36%) eyes, and hyperopia and hyperopic astigmatism were present in 21 (22%) eyes. Specifically, simple myopia occurred in 19 eyes (26%), followed by simple hyperopia in 12 eyes (17%). Myopic astigmatism was present in nine eyes (12.5%), and hyperopic astigmatism was seen in four eyes (6%). Sixteen (44%) cases required an optical prescription.



**Figure 2: Scatter diagram of refractive error with reference to age** The mean refractive error was +0.12D and ranged from -5.50 D to +7.00D (median: +0.125 D). Figure 3 shows the range of refractive error for the subject population. The majority of subjects' spherical equivalent refractive error was within  $\pm 2.00$ D. Astigmatism was found in 13 (17%) of 72 eyes. Different type of strabismus was present in 8 (22%) children (Table 2). Amblyopia was suspected in 11 children on the basis of objective findings. The suspected distribution of amblyopia by type is as follows: strabismic (2), isoametropic (3), anisometropic (2), and meridional (4).

## Discussion

This study was a cross-sectional community-based study involving 36 children with autism. Our male to female ratio was 3:1. This finding closely agrees with previously reported male/female ratios in autism of 3-4:1.<sup>6,8-10,15</sup> Theoretically, the genetic influence in females would have to be of greater severity in order to cause autism, as researchers suspect that females have a greater threshold than males for brain dysfunction.<sup>29</sup>

The age of the participants ranged from two years to 15 years with a mean age of nine  $\pm 3$  years. Most children were found to be diagnosed at later years.<sup>30</sup> In our study, patients were usually sent to a rehabilitation center at a later age. Between the ages of 12 and 16, most subjects learned basic skills and were given ongoing education at their homes.

Most of the children were Caucasian. The overall population in Nepal is made up of a greater percentage of Caucasians in comparison to Mongolians.<sup>31</sup> This may also reflect a higher awareness and knowledge about proper referral to rehabilitation for autistic children on the part of Caucasians.

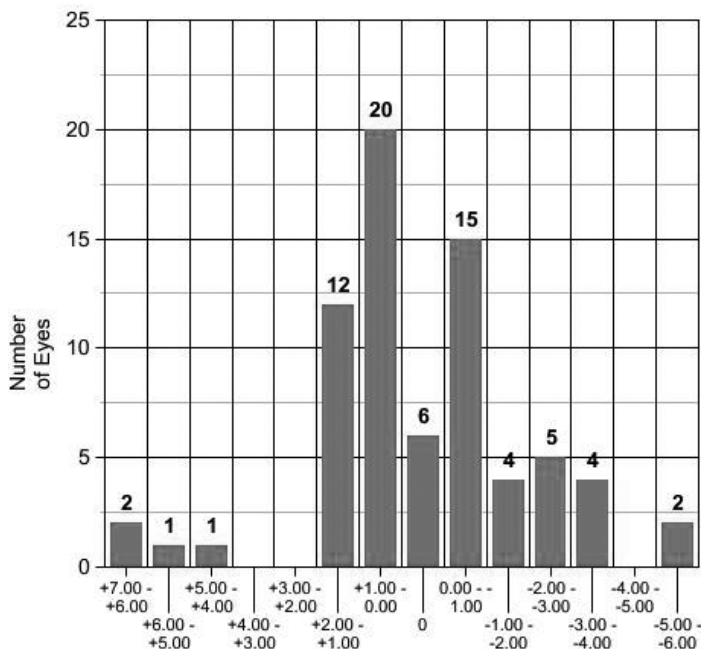


Figure 3: Range of refractive error (72 eyes of 36 patients)

Table 2: Prevalence of Strabismus

Convergent Strabismus	
• Alternate convergent strabismus	1 (13%)
• Right convergent strabismus	0
• Left convergent strabismus	1 (13%)
Divergent Strabismus	
• Intermittent divergent strabismus	3 (37%)
• Alternate divergent strabismus	2 (25%)
• Right divergent strabismus	0
• Left divergent strabismus	1 (12%)
<b>TOTAL</b>	<b>8 (22%)</b>

In contrast, it is also possible that Caucasians simply have a higher prevalence of autism.

Only two subjects had a previous history of ocular examination. Autistic children demonstrate atypical gaze or gaze avoidance, eye pressing, side looking, visual inattention, poor visual awareness of surroundings, and fascination with spinning objects, lights, shadows, and bright metallic objects.<sup>16-22</sup> Before making any assumptions about these atypical looking behaviors, the optometrist must check carefully for uncompensated refractive abnormality by conducting retinoscopy and testing with appropriate lenses. In some instances, high uncompensated ametropias may result in gaze aversion.<sup>32</sup> Hence, when a refractive condition or other visual problem is detected, it may be considered as a characteristic of autism itself.

Accurate assessment of visual acuity in autism was a major challenge. Conventional acuity tests are often impractical and unsuccessful for achieving a reliable measurement. Although we attempted to use the Kay picture and preferential looking

charts, we could not get acceptable responses in all subjects. The concept utilized in preferential looking testing to discriminate between various stimulus pairs, especially faces, are of principal importance when trying to engage patients with autism.<sup>32</sup> However, we were forced to revert to other less reliable and qualitative vision assessment techniques like the OKN drum and CSM. An electro diagnostic technique such as the visual evoked potential may be necessary to quantify the visual status of these children in whom assessment by subjective means is difficult.

The prevalence of ocular abnormalities seen in our study was 67%. We compared our findings with those of the study by Nepal et al. which examined 1100 schoolchildren of the Kathmandu Valley.<sup>33</sup> An ocular morbidity of 11% was found in that study. Refractive conditions were the most common (58%) abnormality. Most children had refractions ranging from -1.00 to +2.00 DS. In comparison, only 8% of schoolchildren of the Kathmandu Valley were reported to have a refractive condition.<sup>33</sup> Though the number of children in our study was significantly less than the number of schoolchildren enrolled in the study by Nepal et al., the prevalence of refractive error could still be considered high in our subjects as we looked at case specific prevalence.

Refraction showed hyperopia in 16 (22%) eyes, myopia in 26 (36%) eyes, and astigmatism more than 1.00D in 11 (15%) eyes. Refractive measures ranged from -5.50 to +7.00 DS, and the median was +0.12 DS. When compared with the general school sample of children from the Kathmandu Valley, myopia was found to be the most common (4%) as opposed to hyperopia (1%).<sup>33</sup> Denis et al.<sup>19</sup> reported hyperopia in seven cases (70%), and Scharre et al.<sup>18</sup> demonstrated refractions ranging from -4.25 to +3.25 D with a median of plano.

The astigmatism prevalence in our study population was 11%. Generally more than 3.00 D of astigmatism, if left uncompensated, is known to be amblyogenic. Denis et al.<sup>19</sup> found astigmatism of more than 1.00D in six cases (60%), bilateral astigmatism in four cases (40%) and unilateral astigmatism in two cases (20%).

Many of the subjects were found to have higher refractive measures. In the age range of birth to four years, the process of emmetropization is still quite active.<sup>34</sup> In the current study, refractive value patterns of older children were similar to the younger children. Hence, we may speculate that the emmetropization process was not functioning properly in these children. Other studies on emmetropization may be necessary to understand refractive error development and emmetropization associated with autism. Such information may help in the timely correction of refractive error and prevention of amblyopia.

We analyzed strabismus based on the Hirschberg method in this group of children. Though it was not a quantitative approach, strabismus was found in eight (22%) children. The most common type of strabismus was exotropia (intermittent or constant) accounting for six (75%) out of eight children.

Amblyopia was suspected in 11 patients based on the presence of strabismus and refractive error that can lead to development of amblyopia. A definitive diagnosis could not be confirmed due to a lack of cooperation. This number could rise if quantitative measurements were possible because there was a chance of missing small angle strabismus. The most common type of strabismus seen in the present study was exotropia (17%). Others have reported 1% to be strabismic at distance and 18% to be strabismic at near, percentages similar to the present study.<sup>18</sup> In the school children of Kathmandu Valley, strabismus was found in 1.6% of cases.<sup>32</sup> Alternate divergent strabismus is the most prevalent type of strabismus (1.4%). Although the prevalence of strabismus was different in autistic and normal children, the pattern of strabismus was similar (divergent squint in our study accounted for 75% of all strabismus, whereas in school-going children it accounted for 87%).

While the posterior segment was normal in all children in the present study, Denis et al.<sup>19</sup> reported pallor of the optic disc in four cases of autism. Leber's congenital amaurosis has also been associated with autism.<sup>19</sup> While the retina may continue to change throughout adulthood, vision loss generally does not occur past childhood in this condition.

The study has significant limitations. The sample was not sufficient to make wide and appropriate conclusions. The examinations were performed in a community setting for most children and the environment may have influenced the results of some tests. Visual acuity could not be taken by the standard clinical methods. Accommodation, convergence, and stereopsis were not evaluated; therefore, the actual status of binocularity was not adequately explored. Finally, the findings were not compared with an age-matched normally developing cohort. Optometrists serving these patients must be prepared to modify assessment and intervention techniques to meet the need of the patients in a meaningful way.<sup>35</sup> As the primary eye and vision care providers, optometrists have an obligation to understand the broader scientific literature regarding vision in individuals with autism.<sup>32</sup>

## Conclusion

Visual and ocular findings in autistic children appear to be mainly uncorrected refractive error and binocular vision problems. Refractive error is the most common and obvious ocular abnormality. Children with autism are significantly more likely to have ocular morbidity than normally developing children. In summary, there is need for early detection as well as methods of prevention for progression of ocular defects in autism. Physical and visual rehabilitation as an early intervention is important for the improvement of visual function. The diagnosis of autism is best made between the ages of one and three years, though diagnosis typically occurs late or not at all in Nepal. During this period, pediatricians that engage in the care and management of autistic children

should be encouraged to refer these children for optometric evaluation.

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