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BRIEF REPORT

Brief Report: Incidence of Ophthalmologic Disorders in Children with Autism

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Abstract *Purpose* To determine the incidence of ophthalmologic disorders in children with autism and related disorders. *Design* Retrospective chart review. Four hundred and seven children diagnosed with autism or a related disorder between 1998 and 2006. one hundred and fifty-four of these children completed a comprehensive ophthalmology exam by a pediatric ophthalmologist. *Results* Ophthalmologic pathology was found in 40% of patients with autism or a related disorder with 29% having significant refractive errors, 21% demonstrating strabismus, and 10% having amblyopia. *Conclusions* Children with autism or a related disorder will frequently have an ophthalmologic abnormality. Since cooperation with vision screening is understandably limited in these children, a comprehensive eye examination by a pediatric ophthalmologist is recommended for all such children.

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Introduction

Autism is a group of disorders characterized by impaired socialization, flawed verbal and nonverbal communication skills and repetitive patterns of behavior. Autism typically develops before the age of 3 years and is often able to be diagnosed as early as 18 months of age. According to the Centers for Disease Control and Prevention (2010) and the American Academy of Pediatrics (Johnson and Myers 2007) there are currently 1 in 166 children in the United States diagnosed with an Autism Spectrum Disorder. A 3–4:1 male predominance has been reported. Multiple causes have been implicated, including genetic and environmental factors, Fragile X syndrome, tuberous sclerosis, congenital rubella, and phenylketonuria. Current screening recommendations for children diagnosed with autism include a formal audiologic evaluation, lead screening, genetic testing, metabolic testing, electrophysiologic testing, and neuroimaging. However, these children also need to be screened for ophthalmic disorders. Indeed, many children with autism are not capable of cooperating for routine school or pediatrician vision screenings, or are in an age range that makes such screening difficult. The incidence of ophthalmic disorders in children with autism is not well established, as there have been few published studies in the literature (Denis et al. 1997) documenting ophthalmic disorders in children with autism. The purpose of this study was to determine the incidence of ophthalmologic disorders in children with autism, whether or not these disorders led to a complaint by the child or family.

Methods

All children diagnosed with autism at Saint Louis University/Cardinal Glennon Children's Medical Center are routinely recommended to have a comprehensive ophthalmic evaluation by a pediatric ophthalmologist. A comprehensive ophthalmic exam includes assessment of fixation or visual acuity, an external exam, a pupillary exam, an anterior segment exam, a sensorimotor exam and a dilated fundus exam with cycloplegic refraction by retinoscopy. A retrospective chart review from 1998 to 2006 found 407 patients given a diagnosis on the autism spectrum including mild to moderate autism, moderate to severe autism, severe autism or another related disorder (i.e. autism spectrum disorder, pervasive developmental disorder, Asperger's disorder, Rett syndrome). Of these children with an established diagnosis, 154 patients were documented to have completed a comprehensive ophthalmic exam by one of two pediatric ophthalmologists at our institution.

Data collected from the charts of these patients included the type of autism diagnosed, age at diagnosis of autism, sex, race, history of prematurity (defined as less than 37 weeks gestation) and history of mental retardation. Data on comorbidities such as cerebral palsy, epilepsy, and cerebral malformation was not collected as part of this study. Ophthalmologic data collected included age at first eye exam, presence of visual complaints, cooperation with recognition acuity testing, and the presence of strabismus, amblyopia, significant refractive errors (defined as >3.00 diopters (D) of myopia or hyperopia, >2.00 D of astigmatism, or anisometropia of ≥1.00 D in spherical equivalent or ≥1.50 D difference in astigmatism in any meridian) and other ocular disorders. These refractive errors are considered significant in that spectacles would routinely be encouraged by the authors to prevent amblyopia, even in sensory-adverse patients. (The Pediatric Eye Disease Investigator Group 2002) Data analysis was performed using the SPSS statistical program (SPSS Inc. Chicago, IL). Pearson's Chi-square tests were used to evaluate whether any of these variables were independently associated with autism.

Results

Of those 154 patients with a diagnosis on the autistic spectrum and documentation of an ophthalmologic exam by a pediatric ophthalmologist, autism was first diagnosed at a mean age of 3 years, 3 months (Fig. 1). One hundred twenty-two (79%) of these patients were male and 21% were female. Seventy-five percent of these children were white/Caucasian, 19% were black, 4% were Asian/pacific islander, 1% were Middle Eastern, and the remainder were Hispanic or of an uncategorized racial makeup. The largest subgroup of patients in the autistic spectrum were those with mild to moderate autism (44%) followed by those with pervasive developmental disorder (20%) (Fig. 2). 23% of patients were born prematurely and 18% of patients were documented to have some level of mental retardation.

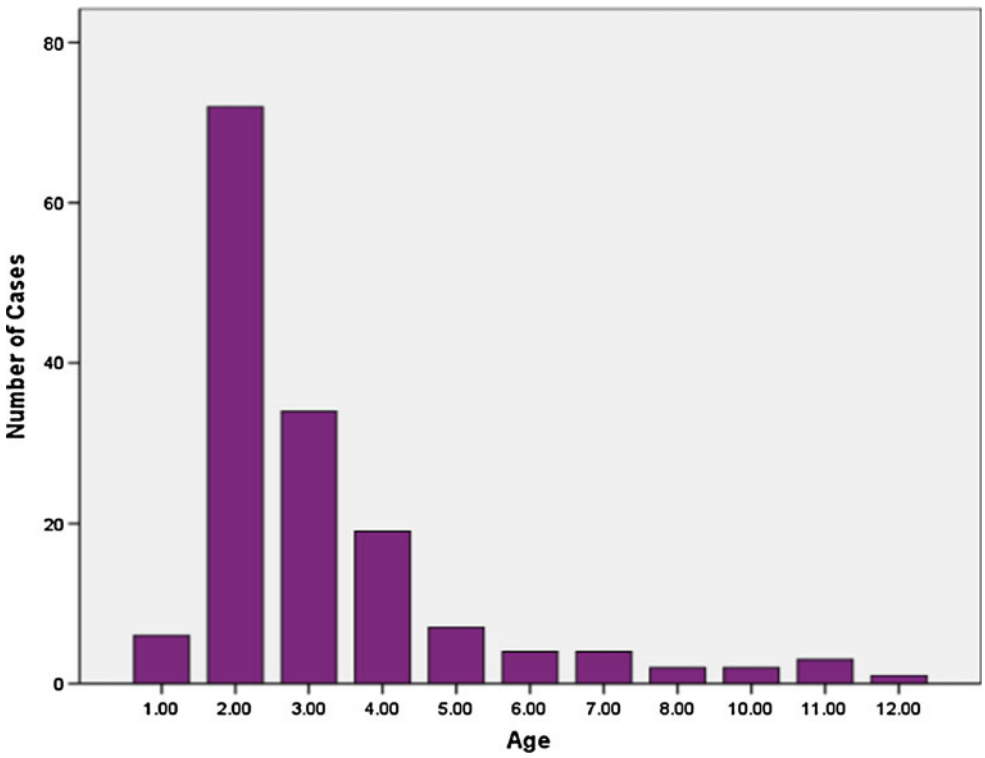


Fig. 1 Age at diagnosis

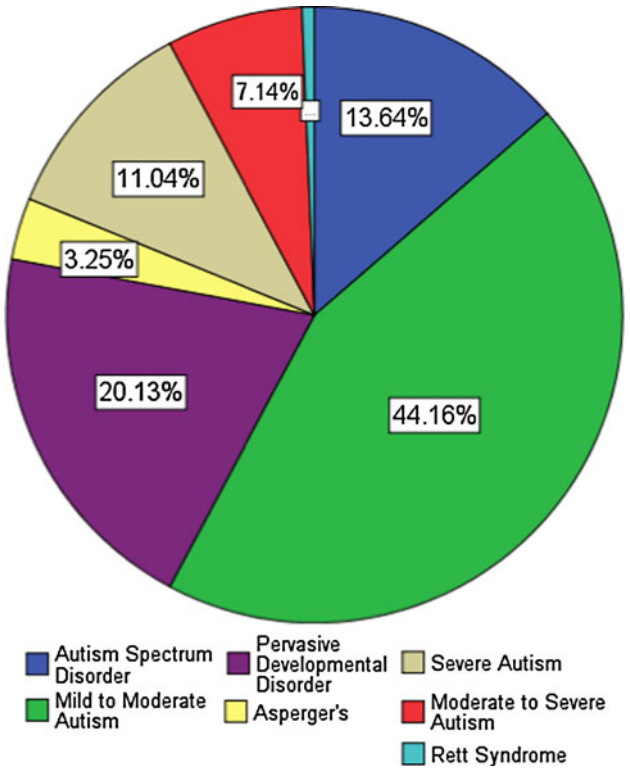


Fig. 2 Types of autism

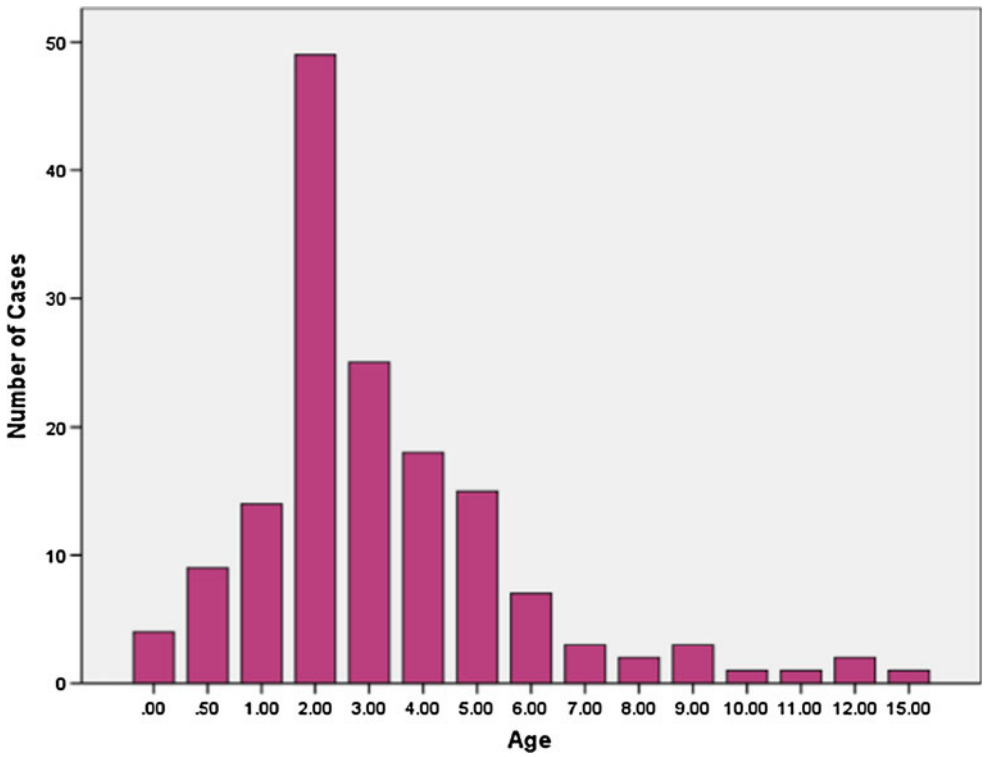
The average age of the first eye exam was 3 years, 3 months (Fig. 3). The majority of patients (57%) presented with an eye-related concern, typically noted by a parent. Only 40% of patients were cooperative for recognition visual acuity testing. Of the 60% who were uncooperative, 36 (39%) were found to have an ophthalmologic disorder. Sixty-one (40%) of the autistic patients had some type of identifiable ophthalmic problem (Fig. 4). Thirty-two (21%) patients had strabismus, with accommodative esotropia most commonly noted. Of those with strabismus, 14 had accommodative esotropia, 11 had exotropia, 4 had non-accommodative esotropia, 2 had infantile esotropia (presenting before 6 months of age), and 1 demonstrated a vertical strabismus. Sixteen (10.4%) patients had amblyopia, with strabismus as the leading cause in 12 of these patients, followed by ametropia in 2 patients, anisometropia in 1 patient, and deprivational amblyopia in 1 patient. Forty-four (29%) patients had significant refractive errors, most commonly hyperopia (26 patients) followed by high myopia in 9 patients, 6 patients with high astigmatism and 3 patients with anisometropia. Several patients had other ophthalmologic disorders (Table 1). Six children who presented without an eye-related concern were identified to have an ophthalmologic problem. Children with autism and mental retardation were more likely to have an ophthalmic problem (any ophthalmic problem, $p = .002$; strabismus, $p = .001$; amblyopia, $p = .127$; refractive error, $p = .044$) but no relation was found for children with autism who also had a history of prematurity <37 weeks gestation.

Discussion

Our findings suggest that ophthalmologic disorders are common in children with a diagnosis in the autism spectrum. Forty percent of these children were discovered to have some type of identifiable ophthalmic problem, most commonly significant refractive error, strabismus, and amblyopia in order of likely occurrence. Each of these disorders is usually treatable, with a combination of spectacle correction, occlusion therapy or atropine penalization, and surgery. Amblyopia is defined as loss of vision in one or both eyes caused by conditions that impair the normal visual input during the period of visual development, such as misaligned eyes (strabismus), uncorrected refractive errors, or occlusion of the visual axis as may be caused by ptosis or cataracts.

The challenge in children with autism is identifying these ophthalmologic problems, and identifying them in a timely fashion. The American Academy of Pediatrics (AAP), the American Association for Pediatric Ophthalmology and Strabismus (AAPOS), and the American Academy of Ophthalmology (AAO) have endorsed specific vision screening guidelines for infants, children and young adults. (Committee on Practice and Ambulatory Medicine, Section on Ophthalmology, American Association of Certified Orthoptists, American Association for Pediatric Ophthalmology and Strabismus 2003) At our institution, it is recommended that all former premature infants meeting criteria for retinopathy of prematurity screening undergo a comprehensive ophthalmic exam at 6 months of age (Fig. 3). Otherwise, while vision screening should be performed as early as is reasonably practical, it should also be carried out as part of the regular plan for continuing care beginning at 3 years of age. Various tests are available for primary care givers to assess vision at this age, though most either require recognition and communication of visual symbols (Snellen letters, Allen Symbols, Lea symbols, tumbling E, HOTV testing) or physical cooperation to get a reliable test (screening machines, photoscreeners, autorefractors). Children with autism often have impaired communication skills or avoid physical interaction with providers to the extent that these normal screening methods are usually insufficient or impractical. Pediatric ophthalmologists are specifically trained to identify structural and functional problems in children without necessarily depending on their cooperation. For instance, amblyopia may be identified by the trained examiner looking for fixation preferences. Refractive errors can be objectively detected with cycloplegic retinoscopy which requires no verbal response from the autistic child.

Fig. 3 Age at first eye exam



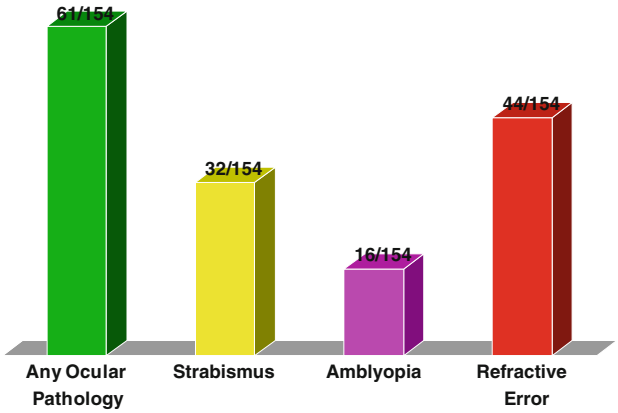


Fig. 4 Frequency of ocular disorders

Table 1 Other ophthalmic problems

Disorder	Frequency	Percent
Nystagmus	6	3.9
Ptosis	5	3.2
Nasolacrimal duct obstruction	2	1.3
Retinopathy of prematurity	2	1.3
Cataracts	1	.6
CN IV palsy	1	.6
Uveal coloboma	1	.6
Euryblepharon	1	.6
Optic nerve cupping	1	.6

In our patient population, 57% of autistic children presented with a suspected visual problem. These visual concerns ranged from the overtly obvious misaligned eye to the subtle observation that the child was squinting or getting closer to objects than would normally be expected. Approximately 9% of autistic children without prior suspicion of an eye-related concern were found to have an ophthalmologic problem. Perhaps more importantly, 39% of children who were not considered cooperative for exam were diagnosed with significant ophthalmic pathology.

Our findings also showed that, when accompanied by mental retardation, children with autism had a particularly high risk of strabismus. This is not unexpected since it is well known that children with cerebral palsy or intellectual disability have an increased incidence of strabismus as well. (Sandfeld et al. 2008; Erkkila et al. 1996; Akinci et al. 2008) However, a history of prematurity alone did not increase the incidence of ophthalmologic problems in children with autism. This is somewhat surprising as children with prematurity are considered at increased risk of strabismus, amblyopia and significant refractive errors, particularly myopia. (O'Connor and Fielder 2008; O'Connor et al. 2007) A relation between the level of autism and the presence of an ophthalmic disorder could not be determined.

Demographically, our sample reflects the typical population of children diagnosed with autism with regards to distribution of patients across the autistic spectrum, age at time of diagnosis and male predominance. In general, there

was a short duration between time of autism diagnosis and ocular examination. However, our study is limited by the nature of a retrospective chart review. While a complete ophthalmologic exam by a pediatric ophthalmologist was recommended for every child given a diagnosis in the autistic spectrum at our institution, only 38% of these patients had a complete ophthalmologic exam documented at our institution. No information is available on those children not examined. As an inner-city medical center with a large indigent population, patients may face a number of socio-economic challenges to following through with medical recommendations, particularly when a vision problem is not suspected by parents or guardians. It is possible that those children suspected of having some type of ophthalmologic disorder were more likely to follow-through with this recommended examination, which would lead to an overestimation of the incidence of specific ophthalmologic disorders in this group of patients. As a tertiary referral center, we may also be more likely to see patients with more difficult problems and those who were unable to undergo vision testing elsewhere.

While a large prospective study is needed to establish the exact incidence of ophthalmic disorders in children with autism, our study clearly demonstrates that these children will frequently be found to have some significant ophthalmologic abnormality. Cooperation with vision screening is understandably limited in many children with autism. Therefore, we recommended a comprehensive eye examination by a pediatric ophthalmologist, optimally as part of a multidisciplinary clinic, for all children diagnosed with autism and related disorders. Identification and treatment of ophthalmic disease may have positive impact on children with autism, particularly with regards to how they visually interact with the world around them.

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Conflicts of interest There are no financial conflicts of interest.

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