Psychiatry and Clinical Neurosciences (2007), **61**, 39–44 doi:10.1111/j.1440-1819.2007.01608.x

Regular Article

Autism in visually impaired individuals

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Abstract The aim of the present study was to assess the prevalence and associated risk factors of autism in a sample of visually impaired children and adolescents. A total of 257 blind children and adolescents (age range: 7–18 years) were examined for autism using a three-stage process. The first stage estimated probable cases of autistic disorder based on the Autism Behavior Checklist and the second stage by direct observation of the subjects in different settings. In the third stage, subjects with the probable diagnosis of autistic disorder were asked to undergo psychiatric examination. A final diagnosis of autistic disorder (based on the criteria in DSM-IV) was given after interviewing the caregivers and clinical observation. Thirty of 257 subjects met the criteria for autistic disorder. Comparison of the characteristics of the two groups (autistic and non-autistic) with χ^2-squared and independent sample *t*-tests revealed a statistically significant difference in terms of severity of blindness ($P = 0.015$), cerebral palsy ($P = 0.02$) and intellectual level ($P = 0.001$). The results of the present study suggest that subjects with blindness plus autism have greater neurological impairment (as suggested by the presence of lower intellectual level and cerebral palsy), and more severe visual impairment than the subjects with blindness only.

Key words autism, visual impairment.

INTRODUCTION

Congenitally blind children are generally reported to be at risk for serious behavioral and psychological problems, such as withdrawal, isolation, and autism.[1] Several studies have described the coexistence of autism or autistic behaviors in visually impaired individuals; however, there is no agreement about the prevalence, nature and role of the contributing risk factors such as severity of visual impairment (VI), type of blindness, age at onset and other associated handicaps. Evidence relating to the prevalence of autistic behavior/disorder in blind people comes from intensive studies of groups of relatively small numbers of blind children, sometimes with specific diagnoses.[2–6] However, studies of groups of relatively large numbers of partially sighted and blind children of heterogeneous etiology[7,8] have not reported a high prevalence of the autistic features.

Regarding the nature of this co-occurrence, there are two main approaches. Some researchers focus on common organic etiological factors that lead to blindness and autism, while others suggest that focusing on the cause of blindness is irrelevant because irrespective of the ophthalmological diagnosis, blindness has understandable developmental consequences that include autistic-like features.[9]

The first report of the co-occurrence of blindness and autistic behavior was by Keeler.[2] He described autistic behaviors in five children with retrolental fibroplasia (RLF) and 35 children with the same medical disorder but demonstrating milder behavioral difficulties. He speculated that a combination of total or near-total blindness from birth, emotional neglect and perhaps brain damage might account for the autistic behavior in this group.[2]

Later, Chase studied 263 subjects with RLF and noted autistic-like behaviors in the sample, but no case with a clear diagnosis of infantile autism. He mentioned the strong relationship between the neurological findings and autistic behavior.[10]

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Received 1 December 2005; revised 3 August 2006; accepted 13 August 2006.

Recently, Ek *et al*. evaluated 27 cases with retinopathy of prematurity (ROP, the same diagnosis with RLF) and compared them to a group ($n = 14$) with congenital blindness resulting from hereditary causes.[6] They reported that 15 of 27 cases with ROP met the criteria for a diagnosis of autistic disorder (AD). In addition, four had autistic-like behaviors. They concluded that 'in preterm children with severe ROP, hypoxia might be affecting: (i) important areas of the brain such as the white matter adjacent to the ventricles, the striatum, or other parts implicated in the pathogenic chain of events in autism; and (ii) the retina'.

Chess gathered behavioral data on 243 children with congenital rubella.[5] Ten of those children were described as having classical autism and eight as having partial autism. Chess believed that the common component accounting for autism in these cases was brain damage.[5]

Another ophthalmological disorder that was reported in connection with autism is Leber's congenital amaurosis. Rogers and Newhart-Larson described autism in five preschool children with Leber's congenital amaurosis, compared them with a control group with congenital blindness (in the majority of cases with ROP) and reported that no child in the control group received the diagnosis of AD.[4] They suggested that the 'existence of cerebellar deficit in some Leber's patients may provide the neurological basis for the behavioral similarities seen in patients with Leber's amaurosis and sighted autistic patients'.

In addition to the studies suggesting common organic etiological factors in the co-occurrence of autism and blindness, some researchers emphasize the role of sensory deprivation and probable social deprivation. This group of studies mentions self-stimulatory behaviors,[1] limitation in theory of mind,[11] limitation in creative representational play, context-sensitive language, and flexible planning and thought[12] and the delay in distinction between oneself and others[3] in children with blindness. The role of sensory deprivation in the development of this behavioral pattern similar to autism was emphasized.

In summary, studies that have looked at the relationship between blindness and autism have varied in their methodology. While some studies, focusing on the relationship between autism and specific types of ophthalmological disorder, attempt to discuss this co-occurrence in terms of common organic etiological factors, other studies emphasize the role of sensory deprivation and associated environmental risk factors in the emergence of autism/autistic-like behaviors. However, there are no consistent results in terms of the relationship between specific types of ophthalmological problems, severity of VI, time of onset of VI and the role of associated handicaps, such as hearing deficit, cerebral palsy, epilepsy and other intellectual disabilities in cases with comorbidity of autism and VI.

The present study aimed to assess: (i) the prevalence of AD in a relatively large and heterogeneous population with VI and (ii) the associated risk factors, such as hearing deficit, cerebral palsy, epilepsy, intelligence level, type of blindness, degree of VI and age of onset of VI, by comparing the autistic group with the non-autistic group.

MATERIALS AND METHODS

Subjects

A total of 257 children and adolescents (77 girls and 180 boys aged 7–18 years (12.08 ± 2.85 years)), attending to two schools for visually impaired individuals in Istanbul, Turkey, were evaluated. Both schools had dormitories and most of the students stayed in the dormitories on weekdays.

Procedure

Legal permission for the study was obtained from an Istanbul attorney, the Ministry of Education and the ethical committee of the Istanbul University. The study consisted of three stages.

The first stage was to identify probable subjects by reviewing school records, have teachers complete the Autism Behavior Checklist (ABC)[13] and caregivers complete the developmental history form, and to assess the visual acuity of all students. After reviewing school records, meetings were arranged with the teachers. The meetings were intended to provide information about the aim and protocol of the study, to request the teachers' collaboration in collecting data and to instruct them on completing the ABC. Written informed consent was obtained from the caregivers and they were provided information about how to complete the developmental history forms. Visual acuity of all students was assessed by one of the authors (ST).

The second stage aimed to identify probable cases by observation of the children during a lesson (45 min) and in different non-structured situations such as school recess, dormitory time and lunch time. Each classroom consisted of six to eight students, therefore, there was sufficient time to observe students individually and to follow their behaviors. The focus of the observation was the child's social interaction, communication in different settings with teachers or peers, and other behavioral aspects. The objective was to identify any combination of problems in these domains that

might give rise to a suspicion that the child might suffer from AD or have autistic features. With the school having a dormitory it enabled more information to be collected. All researchers were experienced in the evaluation of AD, with a mean 6 years' (2–14 years) experience working with children and adolescents with autistic spectrum disorders. In addition, one author (NMM) had 5 years' clinical experience in the psychiatric evaluation of visually impaired individuals and is the founder of the Pervasive Developmental Disorders Clinic, Istanbul University, Istanbul, Turkey.

After obtaining the ABC from the teachers and the developmental history forms from the caregivers, subjects with total score over 45 in the ABC and those showing autistic behaviors during observation by the clinicians were evaluated in psychiatric interviews. This was the third stage of the study. The children were diagnosed according to DSM-IV criteria.[14] In addition, Childhood Autism Rating Scale (CARS) was completed in order to assess the severity of symptoms.[15]

Assessments

Autism Behavior Checklist

The ABC[13] consists of 57 behaviors that seem more common in autistic children than in children with other handicaps. It has been translated into Turkish and reliability and validity studies have been conducted by Gurkan and Sutcu.[16] They examined a sample of 490 subjects (219 with autism, 97 with MR and 174 normal controls) between 3 and 15 years of age, and the results showed that Cronbach's alpha reliability coefficient for the ABC total score was 0.96 and Spearman-Brown two half-split coefficient was 0.96. Criterion-related validity was analyzed by comparing autistic and non-autistic children's ABC scores and the results indicated a significant difference between autistic and non-autistic children. Severity of the problem was used as another external criterion and the result of the analysis of variance supported criterion-related validity of the instrument. The authors concluded that the validity and reliability of the ABC were satisfactory for the Turkish sample.

The total score generated in ABC ranges from 0 to 158. A total score of 67 or above was considered to indicate autism with 'high probability'. The researchers reported a significantly lower total score for the deaf blind group than for the sighted autistic group. Previous studies on AD among VI or other disabilities reported lower cut-off scores of the ABC.[4,8,17] Because of the results of these reports and due to some of the items related with vision (items number 6, 17, 24, 52 and 57) being missing, we accepted cases with total scores over 45 as probable cases and included them in the third stage for observation and interview.

Developmental history form

The developmental history form, which evaluated psychomotor development, type of blindness, onset of blindness, associated medical disorders and other observations about the child's behavior, was completed by the caregivers.

School records

School records included medical information, intellectual level, behavioral problems of the child and other observations made by the teacher and staff. Intellectual level was determined according to the clinical judgement of the clinicians, performance at school, teacher reports and the assessment done by the school psychologist at the time of school registration, using the verbal items of the Stanford-Binet Intelligence Scale and Wechsler Intelligence Scale for Children-Revised. IQ scores 90 and over were defined as normal, between 70 and 90 as borderline and below 70 as mentally retarded.

Childhood Autism Rating Scale

CARS is an autism diagnostic schedule covering 14 different functional areas that may be comprised in autism plus a final category referring to 'degree of autism'.[15] Turkish translation, reliability and validity studies have been done by Sucuoglu *et al*. on 23 autistic children aged between 5 and 15 years.[18] Reliability studies for internal consistency revealed that the Cronbach's alpha was 0.86. Item-total correlations were generated and the correlations for the 15 items except item 14 were between 0.60 and 0.91. For discriminant validity, analysis showed that $P < 0.005$ for 11 items and $P < 0.05$ for three items. Although the study was conducted on relatively small number of subjects and the results were not compared with normal and mentally retarded groups, CARS is widely used to determine the presence and degree of autism in the absence of more validated instruments.

As item 7 of CARS is related to vision, this item was not included during evaluations. The total scores range from 15 to 60. Scores 30–36 indicate mild to moderate autism and scores above 36 indicate severe autism.

DSM-IV criteria for autistic disorder

Final diagnosis of AD was reached using DSM-IV criteria[14] with the consensus of two clinicians. Clini-

cians evaluated and observed the probable cases and interviewed their caregivers in a standard 90-min psychiatric evaluation.

Assessment of severity of visual impairment

Visual impairment was evaluated using data on low vision and blindness as defined in the ICD-10.[19] Visual acuity was assessed by an ophthalmologist (ST) using the Snellen E chart with the current spectacle correction.

The definition of blindness was taken to be: 1, total blindness (no light perception – NLP); 2, near blindness (visual acuity in their better eye less than 20/1000); 3, profound VI (visual acuity in the better eye between 20/500 and 20/1000); 4, severe VI (visual acuity in the better eye between 20/200 and 20/400).

Ophthalmological disorders that cause VI, and also additional disabilities, were recorded according to the observation and the child's previous medical records.

Statistical analysis

Two groups (non-autistic and autistic visually impaired) were compared with independent sample *t*-test and χ^2-squared tests. The differences between groups for parametric data such as chronological age and age of onset of blindness were evaluated with independent sample *t*-tests. Non-parametric data such as gender, severity of blindness, intellectual level, presence of epilepsy, hearing deficit and cerebral palsy were evaluated with χ^2-squared test.

RESULTS

Fifty-seven subjects were accepted as probable cases of AD according to the teacher-rated ABC (over 45) and clinician observation in the classroom.

In the final stage of the study, only 30 achieved AD diagnosis as determined by the clinician interview with the caregiver and observation of the child. Non-autistic cases had some stereotypical, withdrawal, and social contact problems, with CARS total scores between 17 and 25, and did not meet criteria for AD.

There was no statistically significant difference between the autistic and non-autistic groups in terms of age (autistic group: 12.86 ± 3.05 years; non-autistic group: 11.98 ± 2.8 years; $P = 0.11$), age of onset (autistic group: 7.81 ± 20.85 months; non-autistic group: 8.16 ± 19.59; $P = 0.94$) and gender ($P = 0.96$).

The two groups were similar in the distribution of ophthalmological disorders (Fig. 1).

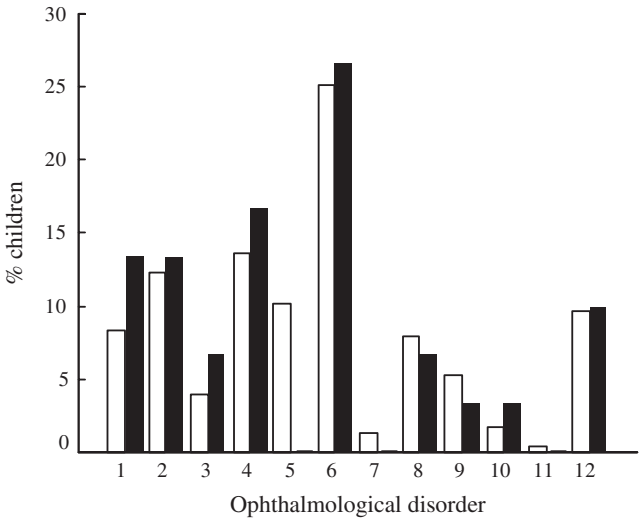

Figure 1. Non-autistic (□) and autistic (■) children with ophthalmological disorders. 1: Congenital/developmental abnormalities of the globe and orbit [non-autistic, 19 (8.37%); autistic, four (13.33%)], 2: congenital cataract [28 (12.33%); four (13.33%)], 3: acquired diseases of globe (phthisis bulbi) [nine (3.96%); two (6.66%)], 4: congenital/acquired diseases of the optic nerve [31 (13.65%); five (16.66%)], 5: congenital glaucoma [23 (10.13%); none (0%)], 6: diseases of retina/macula, [57 (25.11%); eight (26.66%)], 7: diseases of the visual cortex [three (1.32%); none (0%)], 8: movement disorders of the eye [18 (7.92%); two (6.66%)], 9: diseases of cornea/iris [12 (5.28%); one (3.33%)], 10: intraocular tumors (retinoblastoma) [four (1.76%); one (3.33%)], 11: diseases of vitreous [one (0.44%); none (0%)], 12: other (e.g. trauma, strabismus, refractive disorders, etc.) [22 (9.69%); three (10.00%)].

Comparison of the two groups revealed a statistically significant difference in terms of severity of blindness, cerebral palsy and intellectual level (Table 1).

DISCUSSION

Thirty of 257 children with VI with heterogeneous ophthalmologic disorders had AD. The CARS score of the autistic group ranged between 30 and 44.5; 14 had scores between 30 and 36, indicating mild to moderate autism, and 16 had scores between 37 and 44.5, showing severe autism.

The prevalence of autism in the study group seems higher than in previous studies conducted in large-heterogeneous blind groups.[7,8] It could reflect differences in methodology between studies.

Comparison of the groups with regard to intellectual level, type and severity of blindness, time of onset, presence of hearing deficit, cerebral palsy and epilepsy revealed that they differed significantly in terms of

Table 1. Associated risk factors in autistic and non-autistic groups

	Autistic group	Non-autistic group	χ^2-squared and *P*-value		
	n (%)	*n* (%)	χ^2	d.f.	*P*
Severity of visual impairment	–	–	10.44	3	0.015
Total blindness	15 (50)	52 (23)	–	–	–
Near blindness	11 (37)	115 (51)	–	–	–
Profound vision loss	1 (3)	21 (9)	–	–	–
Severe vision loss	3 (10)	39 (17)	–	–	–
Intellectual level	–	–	39.21	2	0.001
Normal	3 (10)	156 (69)	–	–	–
Borderline	9 (30)	20 (9)	–	–	–
Mental retardation	18 (60)	51 (22)	–	–	–
Epilepsy	7 (23)	39 (17)	0.328	1	0.57
Cerebral palsy	10 (33)	29 (13)	–	–	0.024
Hearing deficit	2 (7)	15 (7)	–	–	1
Sex	–	–	0.002	1	0.96
Male	18 (60)	131 (58)	–	–	–
Female	12 (40)	96 (42)	–	–	–

Significance level, $P < 0.05$.

severity of blindness, intelligence level and presence of cerebral palsy.

The present study failed to show a significant relationship between the types of ophthalmological problems and presence of AD. The result is different from the majority of previous studies that had reported autism to be present in subjects with specific types of ophthalmological problems. While some of these studies focused on comorbidity of autism with ROP,[2,6] some reported autism in other groups such as Leber's amaurosis[4] and congenital rubella.[5] However, all share the view that common brain damage/dysfunction and associated handicap lead to the co-occurrence of autism and blindness. The statistically significant difference in terms of cerebral palsy and the intelligence level of autistic and non-autistic subjects in the present study is consistent with this view. A strong relationship between autistic-like behavior and neurological findings was reported by Chase in cases with ROP.[10] In Ek *et al.*'s series of ROP children, six of 15 cases with autism and two of four cases with autistic-like behavior had coexisting cerebral palsy, while no non-autistic case with ROP had cerebral palsy.[6] In addition, all of their ROP cases with AD had mental retardation, while all non-autistic cases were in the normal IQ range.

It could, therefore, be interpreted that the presence of additional handicaps seems to lead to an increase in neuropsychiatric problems.[19] Moreover, it could be concluded that regardless of the type of ophthalmological problem, brain damage/dysfunction has an important effect contributing to autism.

Although the present study failed to find a statistically significant difference between the two groups in terms of time of onset of blindness, the severity of VI was significantly different between groups. It could be concluded that even a minimal degree of vision may make a difference in the socio-emotional development of VI individuals. The importance of severity of VI has been mentioned in previous studies. Keeler compared the incidence of developmental arrest and autistic-like behavior in three groups of children. Those children being totally blind due to ROP were at greater risk for autistic behavior than a group of congenitally blind children with less severe VI, while a third group of children, blinded postnatally, seemed to be relatively protected.[2] Cass *et al.* studied retrospectively the developmental progress of 615 severely VI children during a 15-year period and reported a statistically significant relationship between visual status and developmental outcome; children who were blind throughout their lives were at greater risk for adverse developmental outcome than those whose vision was improved and were at substantially greater risk than those with better vision throughout their lives.[20]

In summary, it seems that the factors that most likely account for the comorbiditiy of AD in blind children are severity of VI, brain damage and mental retardation. Therefore, in addition to treatment and rehabilitation of their ophthalmological and neurological

problems, these children need early intervention programs for their socio-emotional development, language and behavioral problems.

ACKNOWLEDGMENT

This study was supported by Istanbul University Research fund (UDP:435/10032005).

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