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Autism and autism risk in siblings of children with specific language impairment

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Abstract

Background: Several studies have shown that family members of children with autism have elevated rates of spoken and written speech and language problems.
Aims: This study asked whether there was also a greater rate of siblings with autism among probands with specific language impairment.
Methods & Procedures: The probands in this study were 158 children with specific language impairment and 132 children with normal language status. These probands had 522 siblings who were examined for risk of autism using the Autism Behavior Checklist. Siblings found to be at risk were then examined using the Autism Diagnostic Interview—Revised and the Autism Diagnostic Observation Schedule-G.
Outcomes & Results: A concentration of siblings with risk for a diagnosis of autism was found in association with probands who had poor spoken language skills. Four siblings of the 522 (0.8%) met the diagnostic standards for autism. All the probands of these siblings had spoken language scores below −1 SD and three had diagnoses of spoken language impairment.
Conclusions: These data provide additional support for a familial association between autism and spoken language impairment.

Keywords: autistic disorder, specific language impairment, siblings, developmental psychopathology.

Introduction

Specific language impairment (SLI) is a common neurodevelopmental disorder that involves persistent limitations in the acquisition and use of spoken language. The aetiology of SLI is unknown. However, in the past few years there has been a growing body of evidence supporting a genetic contribution (Tomblin 1996). The

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diagnostic standards for SLI involve language achievement levels below cut-off values of −1 to −1.5 standard deviations (SD) of age expectations and the absence of developmental or sensory impairments. Additionally, minimum non-verbal IQ levels as high as 85 have often been employed, although this standard has been questioned by several researchers (Plante 1998, Tager-Flusberg and Cooper 1999).

Autism represents another neurodevelopmental disorder that also has a fundamental feature of impaired spoken-language development and use. Unlike SLI, children with autism also present additional impairments of social development and ritualistic/repetitive behaviour (American Psychiatric Association 1994).

Because autism is an exclusionary condition for SLI, diagnosis of SLI disallows the diagnosis of autism in the same child. Thus, these two neurodevelopmental disorders have usually been viewed as distinctly different. As early as 1967, Rutter hypothesized that language impairment served as the primary abnormality in autism and that this abnormality might be similar to that of a severe receptive aphasia. Rutter and colleagues contrasted the language and cognitive characteristics of children with receptive aphasia and children with autism (Bartak *et al.* 1975, 1977). Although they found that these two clinical groups were similar with regard to deficits in vocabulary and syntax, differences in the communication profiles of these children were found with respect to the use of gestures and spoken language for social communication purposes. Numerous other studies have demonstrated the marked impairment of social aspects of language in children with autism (Baltaxe 1977, Tager-Flusberg 1994). In contrast, children with SLI have often been characterized as having only mild if not preserved social communication (Fey and Leonard 1983, 1984). These results from research on both SLI and autism have provided support for the belief that they are separate disorders.

Diagnostic overlaps in children with SLI or autism

This view, that SLI and autism reflect distinctly different neurodevelopmental disorders, has been challenged from several perspectives. The first has its roots in the work of Rapin and colleagues (Rapin and Allen 1983, 1998, Rapin *et al.* 1992). Within their nosological system for developmental language disorders (DLDs) was a subtype referred to as semantic–pragmatic deficit syndrome. These children were described as having intact phonological and grammatical performance but difficulties in the use of language for social discourse. Semantic–pragmatic disorders were observed in a variety of children with language impairments including those with autism.

Recently, Bishop and colleagues (Adams and Bishop 1989, Bishop and Adams 1989, Bishop 2000) have revised Rapin and Allen's notion of semantic–pragmatic disorder by describing children with pragmatic disorder. Bishop described these children as having difficulties in the appropriate social use of language, but relatively preserved phonological and grammatical abilities, and has referred to this condition as Pragmatic Language Impairment (PLI). These children continue to be the subject of ongoing discussions about whether they represent a variant autism distinct from SLI, a variant of SLI distinct from autism or an intermediate form between SLI and autism (Boucher 1998). Bishop (2000) and Bishop and Norbury (2002) argued that PLI was an intermediate diagnostic form between SLI and autism. Bishop and Norbury provided evidence that many children with PLI do not meet the criteria for autism diagnoses or more generally pervasive developmental disorder not

otherwise specified (PPNOS). Also, Bishop *et al.* (2000) noted evidence of poor pragmatic skills among some children with SLI, as well as other research demonstrating social difficulties in children with autism (Fujiki and Brinton 1994, Redmond and Rice 1998). Thus, overlap between SLI and autism in the form of intermediate cases was not restricted to children with PLI. Bishop (2000) concluded that the contrasts between autism, SLI, pragmatic disorder and Asperger's syndrome were actually reflective of an underlying two-dimensional space formed by the systems of social use of language and the structural aspects of language (syntax and morphology). The particular diagnosis for a given child will depend upon the extent of pragmatic and structural language deficits the child presents.

The potential of overlap between the language characteristics of SLI and autism has also been noted by Kjelgaard and Tager-Flusberg (2001). They focused on the phonological, lexical and grammatical skills of a large group of children with autism. Their data showed considerable variation in their sample with respect to lexical and grammatical development, while phonological abilities in the form of speech sound production were usually normal. This variability revealed some children with autism who had normal levels of vocabulary and grammar; however, the majority of their participants had borderline or impaired levels of language. The profiles of the children with relatively poor language abilities were similar to those found for children with SLI. This similarity in profile characteristic was viewed as suggestive of an overlap between SLI and autism.

Additional evidence of overlap in the behavioural characteristics of individuals with SLI and autism can be found in the results of a follow-up study into adulthood of children with either receptive language disorder or autism (Howlin *et al.* 2000). These authors found that despite clear differences in the social-behavioural characteristics of these children when diagnosed at ages 7 or 8, there was considerable overlap in social and behavioural problems in adulthood.

Overlap between SLI and autism in family members of affected probands

Additional evidence supporting a relationship between autism and SLI has also been found in studies of family members of individuals with autism. Folstein and Rutter (1977) reported elevated rates of language problems in non-autistic co-twins of autistic probands. More recently, Le Couteur *et al.* (1996) reported additional support for these results in a follow-up of the Folstein and Rutter twin study with a new sample of twins. Similar findings of familial loading of communication deficits in non-autistic family members of probands with autism have been found in studies of the Broader Autism Phenotype hypothesis (Cox *et al.* 1975, Landa *et al.* 1992, 1996, Bolton *et al.* 1994, Le Couteur *et al.* 1996, Fombonne *et al.* 1997, Folstein *et al.* 1999, Szatmari *et al.* 2000). The Broader Autism Phenotype (BAP) has been described by Bolton *et al.* (1994) as dysfunction in communication, social skills or stereotyped behaviours and thus it represents a lesser variant of autism that will be expressed in one or more of these domains. Although the above studies found that communication deficits were a part of the BAP, it should be recognized that some studies have not found such evidence (Fombonne and Du 1992, Gillberg *et al.* 1992, Szatmari *et al.* 1993).

Whereas there have been several studies showing elevated rates of language impairments among relatives of children with autism, very little data exist about the rate of autism in families of children with SLI. Rapin (1996), as a part of a

large-scale study of children with DLDs or autism, reported that 2.1% of the families of children with DLD had one or more immediate family member reported to have autism. This rate was 3.9% for the families of children with high functioning autism and 5.0% for the families of the children with low functioning autism. This study did not contain a control population of probands with no developmental disorders but did include a non-autistic low IQ group where the rate of families with positive histories of autism was 0%. Rapin interpreted these data as supporting the view that autism and DLD were aetiologically distinct. However, these results may also be interpreted as suggesting that that the rate of autism in the families of children with SLI is somewhat intermediate between that found in children with autism and that found in a more diverse sample of children with poor IQ.

The evidence of an association between SLI and autism motivated this study. If SLI and autism share phenotypic features and intermediate cases can be found and if factors contributing to SLI increase the liability for autism in families, then it should be expected that there would be an elevated rate of autism in the near relatives of children with SLI. This study was conducted to determine if there was evidence of elevated rates of autism or behaviours characteristic of autism in the near relatives of children with SLI when compared with children with more typical language development. In this case, the study was limited to an examination of the siblings of the probands. This study made use of an existing cohort of children who were participating in a longitudinal study of SLI and therefore is presented as a preliminary inquiry.

Methods

Participants

The probands for this study were participants in an ongoing longitudinal study of 604 children with and without language impairment who were being followed from kindergarten and periodically evaluated for language, cognitive and social outcomes. These children had been initially ascertained in kindergarten as a part of an epidemiological study of SLI (Tomblin *et al.* 1997). The longitudinal cohort was comprised of all children in the epidemiological study who had been found with language impairment and whose parents agreed to participate ($n = 231$) and a random sample of those children in the epidemiological study who had spoken language in the normal range whose parents agreed to participate ($n = 373$) (Tomblin *et al.* 1996). All children had normal hearing and did not present a history of any developmental disorder when they were initially enrolled as kindergartners. The 350 probands recruited were those children from the total cohort of 604 children who had at least one sibling.

The parents of each of proband were contacted via a mailing when the probands were in fourth grade. They were asked to complete a questionnaire about the behaviours of each of the proband's biological siblings (full or half). Responses were received from 290 of 350 parents yielding an 83% response rate. Completed questionnaires described 522 full or half siblings of the 290 probands. Table 1 shows information about gender, the mother's education and the race of these probands according to their language status in kindergarten.

Table 1. Characteristics of probands by kindergarten language status

	Proband group	
	Specific language impairment	Normal language control
Number	158	132
Per cent Males	54	64
Per cent White	80	93
Average highest grade attended by proband's mother	12.9 (1.93)*	14.3 (2.19)

*Standard deviations.

Measures of autism in siblings

Autism Behavior Checklist (ABC)

The ABC is a weighted behaviour checklist designed to be a quick autism screener for educators and parents (Krug *et al.* 1980). This instrument was used to screen for high rates of behaviours associated with autism in the siblings of probands. A cut-off criteria of ≥ 57 was used as a screening cut-off for autism. This cut-off value represented 1 SD below the mean for those children with autism reported in the ABC manual (Krug *et al.* 1980). The authors of the ABC recommended a cut-off of 67 for a high risk of autism; however, a lower value was selected in this case to improve the predictive value positive of this instrument as a screening tool. In addition to items of the ABC, the questionnaire also asked the parent to indicate if each sibling had been identified as mentally handicapped, autistic, learning disabled, hearing impaired or other disability. The parent was provided with a space to indicate the nature of each of these disabilities separately. In order to reduce response bias regarding autism, the survey was renamed a 'Sibling Behavior Checklist' and the parents were not informed that the study was specifically concerned with autism.

Autism Diagnostic Interview—Revised (ADI-R)

The ADI-R was administered to parents of each sibling who failed the ABC screen or had a reported history of autism. The ADI-R is a standardized investigator-based instrument designed for use in the differential diagnosis of autism and pervasive developmental disorders (Lord *et al.* 1994). It was also used to examine for autism in the probands who had siblings at risk for autism. Evidence for a diagnosis of autism was based on the algorithm provided by Lord *et al.* (1994) where the cut-off score for reciprocal social relationships was 10 and that for restricted and repetitive behaviours was 3. The cut-off on communication for verbal children was 8 and for non-verbal children was 7. Additionally, all children were required to have presented abnormal development before the age of 3 years. In all cases, the interviewer was blind to the proband's status.

Autism Diagnostic Observation Instrument—Generic (ADOS-G)

The ADOS-G (Lord *et al.* 2000) was also administered to all but one sibling with elevated ABC scores and/or a history of autism. It is a standardized observation

schedule for the diagnosis of autism and was used to provide direct observational diagnostic information to complement the ADI-R. It was given to provide confirmation of the ADI-R results based on direct observation of behaviours in the children. Each child was administered the ADOS-G module appropriate to his/her verbal abilities and the algorithm appropriate for the module was used for assignment of autism diagnoses. As shown in table 2, Modules 1–4 were administered. The algorithm for Module 1, which was administered to children with no speech, provided cut-off values of 7 for communication, 4 for social interaction and 12 for the total of the two. Module 2, used for children with flexible three-word phrases, had cut-off values of 5 for communication, 6 for social interaction and 12 for the total of these. Modules 2 and 3 were used with verbally fluent children and adolescents, respectively, and used the cut-off values of 3 for communication, 6 for social interaction and 10 for the total of these.

Measures of spoken language in probands

The probands were examined as kindergartners with regard to the following: (1) battery of language tests comprised of the *Test of Oral Language Development Primary—2: Primary* (Newcomer and Hammill 1988) and a narrative comprehension and production task (Culatta *et al.* 1983), (2) performance IQ estimates determined by administration of the Block Design and Picture Completion subtests of the *Wechsler Preschool and Primary Scale of Intelligence—Revised (WPPSI)* (Wechsler 1989), (3) a pure tone hearing test and (4) parental report of developmental, sensory and neuromotor problems. Children were diagnosed as having SLI if they scored at or below -1.25 SD on at least two of five language test domains, if they obtained a performance IQ >70, if they had normal hearing and if they had no parental report of mental retardation, autism, visual impairment, cerebral palsy or severe head injury. See Tomblin *et al.* (1996) for a complete description of the test battery and for discussion of the diagnostic standard used for language impairment in this study.

Procedure

The parents of the eligible probands were mailed the ABC and upon return these forms were scored without knowledge of the proband status. If a sibling met the screening criteria for autism on the ABC (weight score ≥ 57) or the parent had

Table 2. Summary of results of autism status in siblings of 290 probands with or without specific language impairment (SLI)

Sibling group	Total	Number of siblings ascertained through SLI proband
Number of siblings of probands	522	292
Number of siblings with only ABC scores ≥ 56	7	3
Number of siblings with only parental report of diagnosed autism	1	1
Number of siblings with both ABC scores ≥ 56 and diagnosed autism	3	3
Total number of siblings with risk for autism	11	7
Number of siblings with diagnoses of autism based on ADI-R	4	3

indicated a history of a diagnosis of autism for that sibling, the ADI-R was administered by a trained, reliable interviewer. Additionally, the ADI-R was obtained for the probands of siblings at risk for autism in order to rule out autism. For the purposes of this study, the ADI-R results were used as evidence of autism among the siblings and probands. The ADOS-G was given to all children found to have autism with the ADI-R in order to confirm the ADI-R findings based on direct observation of the child.

Results

Parental report of autism behaviours and history of autism diagnoses were obtained for 522 siblings of the 290 probands. The average age of these siblings was 10.9 years (SD = 5.0) and 56% were male.

Risk of autism in siblings

The mean ABC score of the 522 siblings was 7.11 (SD = 14.12). These ABC scores ranged from 0, which was also the modal and median response to a maximum score of 123. The characteristics of the distribution of the ABC scores indicated a highly skewed distribution. This same feature was seen in the distribution of scores for both the siblings of control and SLI probands. Figure 1 shows the mean ABC scores for the siblings of the SLI and control probands. The mean score for the

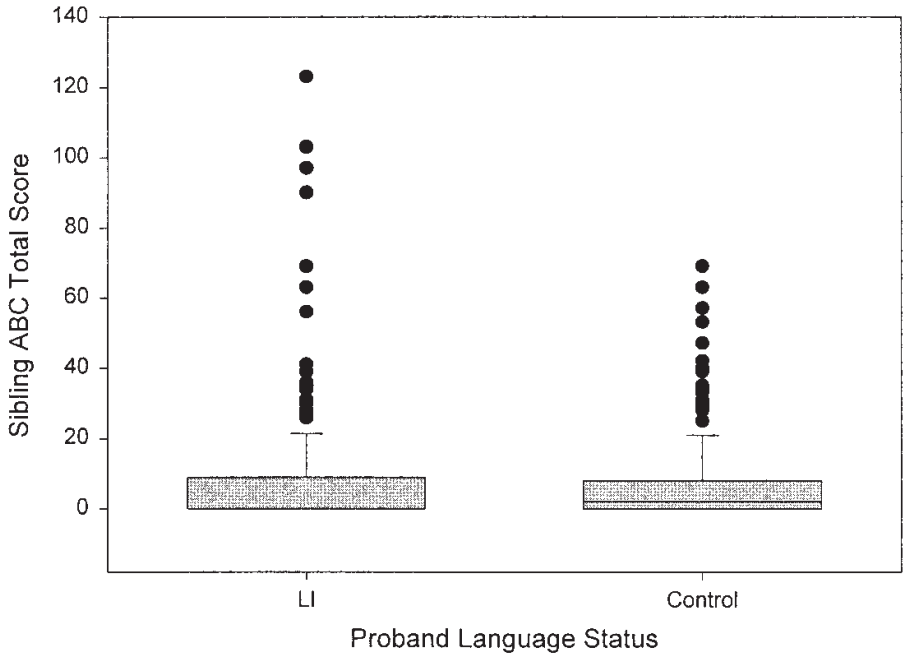

Figure 1. Box plot showing the distribution of scores on the Autism Behavior Checklist (ABC) for siblings of probands with language impairment (LI) and siblings of normal control probands. Each box represents the interquartile range and the line within the box is the median.

230 siblings of the control probands was 6.15 (SD = 9.8). The mean score for the 292 siblings of the SLI probands was 7.79 (16.4). A Mann–Whitney *U*-test indicated that the distribution of ABC scores for the siblings of the two groups was not different ($U = 30569$, $p = 0.23$).

Table 2 provides a summary of the number of siblings found to be at risk for autism based upon either a high ABC score or the presence of a diagnosis of autism reported by the parents. Ten siblings from seven sibships were identified as being at risk for autism when a cut-off on the ABC checklist of ≥ 57 was used as an index for a high probability of autism. An examination of the number of siblings with ABC scores ≥ 57 showed that of the 10, six were siblings of SLI probands. In addition, of these 10 siblings with high ABC scores, three were reported by the parents as having been previously diagnosed as autistic and all these were siblings of probands with SLI. An additional sibling was reported as having a history of autism, although this child had an ABC score of 35. This child also was a sibling of an SLI proband. These 10 children with ABC scores of ≥ 57, plus the additional sibling with a history of autism only, were considered to be at risk for autism, and seven of the 11 (64%) were siblings of probands with SLI. The total sample of siblings was composed of 292 (56%) siblings of probands with SLI. Thus, although the risk for autism was higher in the siblings of the SLI probands, the relationship was not significant ($\chi^2 = 0.26$, d.f. = 1, $p = 0.611$).

Owing to the fact that several of these siblings with risk for autism were either full or half siblings themselves, the observations based on siblings cannot be assumed to be independent, particularly because these data were based on a single informant for each sibship. Thus, these data were recast to examine these rates by sibship. When the rate of autism in siblings was examined by sibship rather than individual siblings, five (3.16%) of the sibships formed by SLI probands had one or more siblings at risk for autism and two (0.74%) of the sibships formed by normal control probands had one or more siblings at risk for autism. Thus, again the rate of autism risk was greater in the sibships of SLI probands than for the sibships of the control probands. However, this difference was not significant ($\chi^2 = 0.0.248$, d.f. = 1, $p = 0.62$).

The above analysis treated the trait of language proficiency in the probands categorically; differentiating between children with z-scores at or below -1.25 SD in two of five areas of language function. This cut-off was established based on research examining the correspondence of clinician decisions and different cut-off scores. However, it must be recognized that there is no evidence that this cut-off distinguishes two qualitatively different groups of language learners (Tomblin and Zhang 1999). As a result, we also examined the relationship between quantitative composite language scores of the probands and the risk status of the siblings. Figure 2 shows a scatter plot of these data. Those probands with siblings having high ABC scores had composite kindergarten scores no higher than –0.98 and no lower than -2.07 in z-score units. Additionally, the one child with a history of autism who also had an ABC score of 35 is shown and the proband for this child also had a composite language z-score of -1.4. Thus, risk for autism was only found in siblings of probands with poor spoken language status in kindergarten.

Autism status in siblings

The analysis thus far concerned the association of SLI with risk of autism. In order to examine the association of diagnosed autism with SLI, those siblings determined to be at risk for autism were evaluated using the ADI-R and the ADOS-G.

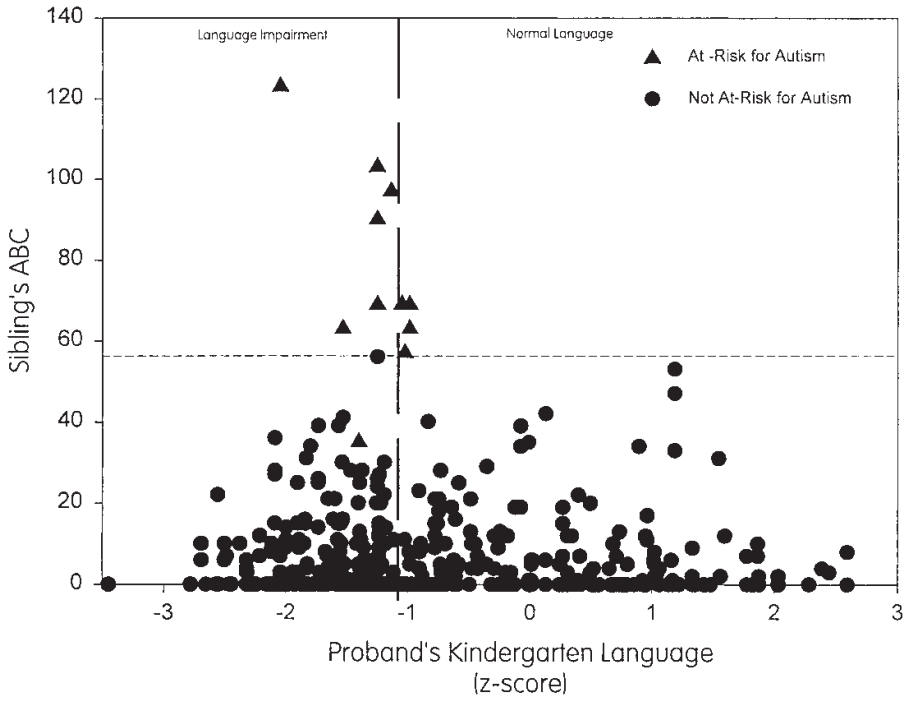

Figure 2. Scatter plot of composite language scores obtained by probands plotted against scores on the Autism Behavior Checklist (ABC) obtained by their siblings. The triangles denote siblings who were at risk for autism owing to an ABC score greater than 56 or to a history of diagnosed autism.

As shown in table 3, four of the 11 siblings met the ADI-R criteria for autism (A, C-1, C-2, D-2). (The letters of the identifier refer to sibship and, thus, children with the same letter are siblings.) Two of the children (C-1, C-2) who met ADI-R criteria for autism were siblings. This resulted in three autism sibships in which one or more sibling coded for autism using the ADI-R algorithm. The ADOS-G was given to all but one of the siblings who met criteria for autism with the ADI-R. One parent refused to consent to further participation by the sibling or the proband. Table 3 contains the results of the ADOS-G. These data showed that siblings C-1, C-2 and D-2 obtained scores that met criteria for autism. The same children who met criteria using the ADI-R also met criteria for the ADOS-G. Although sibling A-1 was not given the ADOS-G, the child did have an existing diagnosis of autism according to parental report and unequivocally met ADI-R criteria. Therefore, A-1 was also coded for autism. The fact that eight of the 11 siblings had high ABC scores, but received rather low ADI-R scores, is likely to be due in part to the fact that the ADI-R uses a very intensive interview method. Determination of whether a particular behaviour indicative of autism is made by the trained examiner and only when a sufficient amount of information has been provided by the parent. Thus, the discrepancy between these two parental report schemes is likely to be due at least in part to differences in method.

Four of 11 siblings determined to be at risk for autism met the current diagnostic

Table 3. ADI-R and ADOS-G scores for 11 siblings with elevated ABC scores and/or history of autism

	ADI-R				ADOS-G		
Sibship–sibling*	Social	Verbal communication	Non-verbal communication	Repetitive and stereotyped behaviour	ADOS-G module	Communication	Reciprocal social interaction
A	**25**	**20**	**0**	**8**	NA	NA	NA
B	1	1	0	0	3	1	2
C-1	**28**	**0**	**13**	**4**	**1**	**8**	**9**
C-2	**27**	**22**	**0**	**8**	**3**	**6**	**8**
D-1	7	3	0	2	3	0	0
D-2#	**27**	**0**	**12**	**6**	**1**	**6**	**14**
E	19	16	0	2	3	0	0
F#	4	4	0	0	2	0	0
G-1#	2	3	0	2	3	1	0
G-2#	4	2	0	1	3	2	3
G-3#	0	2	0	2	2	0	0

*Children with the same letter in their sibship identification belong to the same sibship.
#Half siblings to proband, all other siblings are full siblings of the proband.
Bold font indicates a child met the instrument algorithm for the diagnosis of autism.

standards for autism. Thus, the rate of autism in the sample of 522 siblings was 0.77% (95% confidence interval = 0.22–2.03). A critical ratio test revealed that this proportion was significantly different ($z = 4.165$, $p < 0.0001$) from an estimated population prevalence rate of autism of 0.1% (Gillberg and Wing 1999). Because the determination of autism was not based on a common parent reporting for each member of the sibship, the observations within a sibship may be viewed as independent so long as a shared aetiology is not considered. Thus, it could be concluded that the rate of autism in the total sample of siblings exceeded common estimates for the prevalence of autism in the general population.

The data in table 3 also show that three of the four siblings with autism had a sibling who met the diagnostic criteria for SLI. There were 292 siblings of probands with SLI, thus the rate of autism among these children was 1% (95% confidence interval = 0.21–3.12%). This rate was 10 times greater than the population estimate of 0.1% and was also significantly different from this population estimate ($z = 3.95$, $p < 0.0001$) using a critical ratio test. In contrast, one sibling with autism had a sibling who was not SLI. Thus, the rate of autism in the siblings of the control group was 0.4% (95% confidence interval = 0–2.67%). The fact that this interval includes 0.1% indicates that this rate was not significantly greater than the rate expected in the population in general. These two analyses for the siblings of SLI probands and the controls are not independent of the previous analysis showing elevated rates of autism in the total sample of siblings. This dependency between these two results shows that the elevated rate of autism in the total sample of siblings is likely to be the result of the subsample of siblings of SLI probands. As noted above, there was one case of an autistic sibling of a proband whose language status did not meet the diagnostic criterion of two tests below −1.25 SD. This proband, however, did have a composite language score below −1 SD. Thus, this proband would be considered a borderline case for SLI. When this case is included, all siblings with autism could be reasonably viewed as having probands with poor language, if not SLI.

Autism status of SLI probands

All the probands in this study had been selected because they did not present any sensory or developmental disorders other than language impairment. Thus, by history, none of the probands presented with autism. To insure that the probands who had siblings at risk for autism were not autistic themselves, the ADI-R was administered to the parents of seven of the eight probands. The results of these interviews are shown in table 4. These data indicate that none of the scores met the criteria for autism. The proband in sibship A was not evaluated using the ADI-R owing to the parent's desire to end participation in the study. Before this decision, the parent had provided information about this child on the Child Behavior Checklist (CBCL). The parent did not report any illnesses or disabilities including autism, nor were any special education services provided to this proband. The child's total problems score was 55 and the social problems scale was 50, where 50 is considered average. Thus, it is unlikely that this proband would meet the criteria for autism.

Table 4. ADI-R and language scores for seven probands of siblings with elevated ABC scores and/or history of autism

			ADI-R			
Sibship	Composite language z-score	Language diagnosis	Social	Verbal communication	Non-verbal communication	Repetitive and stereotyped behaviour
A	−1.40	SLI	NA	NA	NA	NA
B	−2.05	SLI	1	3	.	0
C	−1.25	SLI	1	0	.	0
D	−1.03	Normal	4	1	.	2
E	−1.25	SLI	4	5	.	0
F	−1.50	SLI	6	3	.	3
G	−0.99	Normal	3	5	.	2

SLI, specific language impairment.

Discussion

This study was conducted to determine if there was an increased rate of autism among the siblings of probands with SLI. To accomplish this purpose, an existing sample of children who had participated in an epidemiological study of SLI was used. The sample size of 290 probands and 522 siblings used was not designed to provide the power needed to test the association fully. In order to detect an elevated rate of autism or even the risk for autism, the sample size would need to be large or the effect that confers risk would need to be strong. In this regard, it is surprising that this study had sufficient power to detect an elevated prevalence rate in the siblings of probands with poor language.

The data showed that among the 11 siblings at risk for autism, four were confirmed to be autistic using conventional research-based diagnostic methods. The data revealed a pattern of high concentration of autism in a small set of sibships. Among the seven probands with siblings who had high ABC scores were four probands who had more than one sibling at risk for autism. In one case, proband G, three half-siblings received high ABC scores. The cause of this concentration of autism behaviours in these families may be explained in part by the fact that the ABC is a parent report measure and, therefore, the parental reports were likely to be correlated owing to a common reporter. This may have been a factor in the loading of autism risk in the siblings of proband G, particularly given that none of these children were confirmed as autistic on the ADI or ADOS. This effect, however, is not likely to explain all of the familial concentration of autism in these sibships because in one of these four sibships (C) both members were confirmed to be autistic using the research-based instruments including the ADOS-G that does not involve parental report. In sibship D, one of the two members with high ABC scores was confirmed with the ADOS-G providing partial validation of this parent's report.

Although the ABC results may be affected by shared parental reports, it was surprising to see the relationship between high ABC scores or a history of autism in the siblings and poor language skills in the probands. If these high ABC scores are viewed as signs of autism spectrum behaviours for autism and the borderline poor language scores of the probands are viewed as spectrum behaviours for SLI,

then the data show that concentration of these spectrum behaviours of autism and SLI aggregate in families.

This aggregation of autism spectrum in sibships of probands with SLI spectrum is further supported by evidence of aggregation of autism in probands with SLI. Three of the four sibships containing individuals with autism had been ascertained via an SLI proband. These findings provide support for a familial association between SLI and autism itself. The three cases of autism in the siblings of probands with SLI represented 1% of the siblings of SLI probands. This represents a 10-fold increase in the rate of autism over the expected population rate of 1:1000 (Gillberg *et al.* 1999). Several studies have shown that the risk for autism to siblings of probands with autism is between 2 and 6% (Ritvo *et al.* 1989, Bolton *et al.* 1994, Fombonne *et al.* 1997). Thus, the recurrence rate of autism in siblings of SLI probands is between 16 and 50% lower than it is for siblings of probands with autism. Rapin (1996) reported a similar relative rate of autism in the first-degree relatives of her children with SLI and the relatives of children with autism. This pattern of reduced risk in the SLI families when compared with autism families, but an elevated risk when compared with control families, is very consistent with the concept of the BAP. The notion of the BAP is that aspects of the autism phenotype can be found in family members. These manifest in milder levels and are more restricted to a specific aspect of autism such as social difficulties, rigidity or language deficits. In this respect, autism can be splintered and moderated in family members. The findings of this study may suggest that SLI and the language behaviours used to define it can represent one of these splinters within the BAP.

This pattern of association between SLI in probands and autism in siblings is consistent with those data in which family members of probands with autism were found to have elevated rates of language and reading problems (Bartak *et al.* 1977, Folstein and Rutter 1977, Le Couteur *et al.* 1996). Thus, the SLI risk to family members from having a relative with autism or the autism risk to family members from having a relative with SLI appears to be bi-directional. Thus, it may be that these two conditions run together in families.

If SLI does represent a portion of the BAP, one of the important implications pertains to the potential of a partial shared aetiology for SLI and autism. The aetiology of SLI is unknown. There have been several studies demonstrating high levels of heritability (Lewis and Thompson 1992, Bishop *et al.* 1995, Tomblin and Buckwalter 1998). Furthermore, a segregation analysis of a quantitative trait representing the likelihood of SLI has suggested a complex non-Mendelian mode of transmission (Tomblin *et al.* 1999). Given the complexity of the phenotype and the results of the segregation analysis, it is likely that the genetic contributions to SLI are complex and, at best, oligogenic. Similarly, there is substantial evidence that autism has a complex genetic basis (Folstein and Mankoski 2000). Given the familial co-morbidity of these two genetically complex traits, it is reasonable to offer the hypothesis that there is an overlap in the genetic basis of these two conditions.

We view this study as an initial effort. It had the advantage of employing a cohort that had been sampled via population sampling methods. Thus, the aggregation of affected siblings is not likely to be the result of patterns of clinical service seeking by parents. The sample size of the sibships of this cohort, however, would have to be viewed as minimally sufficient for identifying elevated rates of autism in a population, even one at risk. The results need to be replicated using a larger sample of siblings. In such a replication, it would appear appropriate to concentrate

the sampling on probands with language levels at or below −1 SD. Additionally, it would be useful to 'over sample' probands with language skills below −1.5 SD in order to determine if the risk to siblings of these probands was comparable with that of siblings with milder language impairment. Until such a study is conducted, the current study will need to stand in its place and provide encouraging support for a familial and aetiological association between SLI and autism.

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