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Are Phonological Processing Deficits Part of the Broad Autism Phenotype?

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Two tests of phonological processing, nonword repetition, and nonsense passage reading, were administered to 80 probands with autistic disorder or PDDNOS (index cases) and 59 typically developing controls, together with their parents and siblings. In addition, parents completed a questionnaire about history of language and literacy problems, and all participants were given tests of verbal (VIQ) and performance IQ (PIQ). Parents also completed the Autism-Spectrum Quotient, which was used to index the broad autism phenotype. Index probands scored well below control probands on the two phonological tests. However, on neither phonological measure did index relatives differ from control relatives. Within the index group, there was no relationship between the proband's level of VIQ, or age at achieving phrase speech, and phonological score of relatives. VIQ was the only measure to show any familiality within the index group. Reported history of language and literacy problems did not differentiate index parents from control parents overall, but those who were categorized as cases of the broad phenotype reported more history of language and literacy problems than did other index parents. However, they did not have poorer scores on the phonological measures. It is concluded that phonological processing deficits are not part of the broad autism phenotype.

KEY WORDS: autism; specific language impairment; phonological processing; family; broad phenotype

INTRODUCTION

Evidence for a Common Etiology in Autism and SLI

Traditionally, autism has been regarded as a pervasive developmental disorder that should be differentiated from specific developmental disorders such as specific language impairment (SLI) or developmental dyslexia. In more recent years, however, it has been suggested that there may be etiological overlap between autism and SLI. A popular contemporary view is articulated by Folstein et al. [1999] as follows: "...each component of the broader autism phenotype is inherited separately and manifested independently in non-autistic family members, who may have only impaired language, social reticence, or rigid/obsessive features." (p. 1118). In effect, the child with autism may be seen as inheriting a particularly detrimental combination of alleles, any of which on its own might cause at most only mild developmental difficulties. The main reason for this shift of perspective is that studies of relatives of people with autism showed that, although core autism is rare in first degree relatives (albeit substantially higher than the general prevalence rate in the population), milder cognitive deficits, involving one or two components of the autistic triad, are more frequent. Language deficits in relatives of people with autism have been described in several studies [Rapin, 1996; Folstein et al., 1999; Szatmari et al., 2000].

This perspective prompts re-examination of the question of how autism relates to SLI. Although both groups are known to have poor language skills, in autism there are substantial difficulties with pragmatics, that is, appropriate use of communication, whereas in SLI there are usually marked problems with language structure, especially grammar and phonology. Tager-Flusberg and Joseph [2003] argued that there is a distinct subtype of autism with the same structural language difficulties as are seen in SLI, and they suggested that the same genes that cause SLI in one child might cause autism in another, provided they were inherited in combination with other risk genes. Consistent with this, Tomblin et al. [2003] reported that the prevalence of autism in sibs of children with SLI (1%) was significantly higher than general population estimates. However, the number of sibs with autism in their sample was very small, and the prevalence rate in sibs of SLI cases did not differ significantly from that of a control group (0.4%), making this finding rather ambiguous. The notion of etiological overlap between autism and SLI has also received indirect support from molecular genetic studies in which linkage signals to sites on chromosomes 2q [Buxbaum et al., 2001; Shao et al., 2002] and 7q and 13q [Bradford et al., 2001] were strengthened when the analysis was confined to multiplex families in which both probands had autism with language delay. In the Bradford et al. [2001] study, parents of probands in this subset of families also reported language delay.

Evidence Against a Common Etiology in Autism and SLI

Bishop [2003] drew attention to several difficulties confronting a theory that treats SLI and autism as having partly overlapping etiologies. First, though many children with autism do poorly on conventional language tests sensitive to SLI, a substantial minority do not. Furthermore, the kinds of abnormality of speech sound production (expressive phonology impairments), which are fairly common in children with severe SLI, especially in the preschool years, are not usually seen in

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verbal autistic children. Third, molecular genetic studies have found no overlap in the loci that show linkage to SLI and those that show linkage to autism [Barnby and Monaco, 2003]. There was initial optimism [Folstein and Mankoski, 2000; Alarcón et al., 2002] that autism may be associated with the specific mutation of the FOXP2 gene on chromosome 7q that cosegregates with severe speech and language disorder in one British family [Lai et al., 2001]. However, this has now been discounted [Wassink et al., 2002]. Finally, there is some controversy as to what extent relatives of people with autism do have a raised prevalence of specific language or literacy impairments. Bailey et al. [1998], in reviewing studies of IQ in relatives of people with autism, noted that no consistent cognitive profile has been found. Some studies report that verbal IQ (VIQ) is lower than performance IQ (PIQ), some report the opposite pattern, and others find no difference. Furthermore, such differences as are found are not seen in all relatives (mothers, father, sisters, and brothers), leading one to suspect that they may arise because the likelihood of type I error is relatively high when several groups of relatives are compared on a range of IQ measures (e.g., VIQ, PIQ, and the verbal-performance discrepancy). Two family studies, one in the UK [Fombonne et al., 1997], one in the US [Piven and Palmer, 1997], compared rates of self-reported language, and literacy impairment in relatives of people with autism and a control group consisting of relatives of people with Down Syndrome (DS). In both studies, parents from autism families reported relatively high rates of developmental disorders of language and literacy in both themselves and their unaffected children. Nevertheless, on direct testing, mean levels of verbal ability among autism parents were, if anything, higher overall than in DS parents, and literacy tests revealed no significant differences.

Verbal IQ tests focus predominantly on verbal knowledge and reasoning rather than the structural language skills that are most impaired in SLI, and so it could be argued they may not be sensitive to characteristics of SLI in relatives. This issue was addressed in a study by Plumet et al. [1995], which used only female autistic probands, and employed a range of tests that predominantly assessed phonological processing in oral and written language. Brothers of autistic probands obtained lower scores than brothers of DS probands, whereas parents and sisters did not differ. However, this result would not have reached statistical significance if correction had been made for multiple comparisons. A more recent small-scale study by Pilowsky et al. [2003] found no deficits in siblings of children with autism on language tests that are used to diagnose SLI.

Aims of the Current Study

The current study was designed to test whether language deficits of the kind seen in SLI form part of the broad autism phenotype. To address this question we first compared the rate of language impairments in probands with autistic spectrum disorder (index cases) with that seen in control probands, to establish how sensitive each language measure was to deficits in autism. Next, we compared language measures in relatives of index cases versus controls, predicting a higher rate in the former group for measures that distinguished index versus control probands. In a further analysis, we considered whether there was any association between poor language skills in the probands and poor language skills in their parents or siblings. This kind of analysis provides a much stronger test of the hypothesis that language deficits are part of the heritable phenotype, because it allows for the possibility that such an association may characterize only a subgroup of those with autistic disorder, as predicted by the theoretical account of Tager-Flusberg and Joseph [2003]. Finally, we considered whether language deficits were related to self-reported early history of language impairment, and to the "broad phenotype" status of parents, as assessed by the Autism-Spectrum Quotient [Baron-Cohen et al., 2001].

Measures of Language Impairment

Language impairment is a broad construct encompassing a range of separate skills. The chances of finding etiological overlap between SLI and autism may depend crucially on the language measures we use. In this study, we considered two types of measure: verbal IQ, and tests of phonological processing. Phonological processing is a skill that is frequently impaired in SLI and specific reading disability [Kamhi et al., 1988]. To assess phonological processing of oral language we used nonword repetition. In this test, the participant hears a spoken polysyllabic nonsense word, such as "nembid" or "perplisteronk" and is asked to repeat it immediately. In the context of genetic studies of autism, nonword repetition appears to be a particularly interesting task for four reasons. First, poor performance is seen not only in children with current evidence of SLI, but also in older individuals who have a past history of SLI but appear to have outgrown their obvious language difficulties [Bishop et al., 1996]. Second, nonword repetition skill is strongly heritable [Bishop et al., 1996, 1999], and is less influenced than other language measures by social and ethnic background [Campbell et al., 1997]. Third, molecular genetic studies have found linkage to a locus on chromosome 16q when nonword repetition is used to define the phenotype of SLI [SLI Consortium, 2002]. And fourth, children with autism often show marked impairments in nonword repetition [Kjelgaard and Tager-Flusberg, 2001], despite the conventional wisdom that verbal mimicry is an area of strength.

The other phonological test that we used was a nonsense passage reading task, taken from Gross-Glenn et al. [1990]. This task was selected because it is sensitive to literacy difficulties in adults who have compensated for childhood reading difficulties. Reading of nonsense words requires application of rules for mapping letters to speech sounds, and so taps phonological skills.

METHODS

Participants

Index families. The data reported here come from the Western Australia Family Study of Autistic Spectrum Disorders. Families with two or more children, where at least one child had an autistic spectrum disorder (index families) were recruited by advertisements sent to centres where children with autism were diagnosed or educated. Families were excluded if the proband had a serious organic condition, such as identified metabolic or genetic disease. The child with an autistic spectrum disorder was designated the proband. A diagnosis of autism was confirmed in the probands in 59 index families using the Autism Diagnostic Interview-Revised [Lord et al., 1994]. A further 23 index probands scored above threshold on one or two of the areas of impairment assessed by the ADI-R and were designated as cases of PDDNOS. In most cases ($N = 19$), they were impaired only on communication and/or social interaction. For the analyses reported here, the 80 autism and PDDNOS index cases are treated together. Autistic symptomatology in siblings of index probands was assessed using the Social Communication Questionnaire [SCQ: Berument et al., 1999], and the ADI-R was administered to any child scoring above 10 (cutoffs for PDD and autism are 14 and 21 respectively). In two index families, more than one child met full ADI-R criteria for autistic disorder, and in these cases, the child who was first referred to the project was

regarded as the proband. Significant autistic symptoms that fell short of meeting algorithm criteria for autistic disorder but met our criteria for PDDNOS were seen in seven families (in one sibling in six families and in two siblings in one family).

All first degree relatives for whom consent was given were included in the study. Data from mothers was available for 99% of families, from fathers for 83% of families, and for one or more siblings for 80% of families.

Control families. Control families were recruited by brochures sent to schools, and mailouts in the Perth Metropolitan Region. The goal was to select a typically developing sample that was similar to the index sample in terms of age and sex distribution. Because reliance on volunteers tends to yield a sample biased in favour of children with above average IQ, some lower ability control probands were recruited by screening IQ in all children in a school for whom parental permission was given, and then inviting parents of less able children to take part in the main study. Control probands were screened for autistic symptomatology using the SCQ. The ADI-R was administered to those who scored above 10 points, and any scoring above the algorithm criterion on any domain were excluded. The final sample consisted of 59 control probands.

All first degree control relatives for whom consent was given were assessed. Data from mothers was available for 98% of families, from fathers for 64% of families, and for one or more siblings for 80% of families. Participation rates were equivalent to that of index families for mothers and siblings, but significantly lower for fathers, $\chi^2(2) = 5.9$, $P = 0.015$.

Comparisons of age and educational background. *T*-tests were run to compare age of relative groups and found to be nonsignificant. For parents, the mean age was 41.4 years for index families and 40.9 years for control families. For siblings, the mean age was 11.32 years for index families and 10.61 years for control families. Index probands were significantly older than the control probands: mean age 10.44 years (SD = 4.51) for index vs. 9.03 years (SD = 3.34) for controls, t (137) = 2.02, $P = 0.045$. Age differences between groups are taken into account in analyses by the use of age-scaled scores. Educational level of parents was coded on a four-point scale, ranging from 1 = up to year 10 high school, to 4 = university degree. In a nonparametric test, index and control mothers and fathers did not differ significantly on this measure.

Psychological Assessment

Probands and their first degree relatives were seen at home for a neuropsychological examination, except for siblings aged below 3.5 years. Parents and other family members aged over 16 years were given the Vocabulary, Similarities, Picture Completion, and Object Assembly subtests of the Wechsler Adult Intelligence Scale—III [Wechsler, 1997], and these were prorated to give short form estimates of Verbal and Performance IQ. Younger participants were given short forms of the Wechsler Scale suitable for their age range.

Measures of Phonological Processing

Nonword repetition. Items from the Nonword Memory Test were tape-recorded by an Australian speaker. These items were devised by Baddeley, Gathercole, and Watson (in preparation) to provide a test of nonword repetition that would be difficult enough to avoid ceiling effects in adults. There are seven items at each of four syllable lengths (2–5 syllables). A monosyllabic practice item ("vonk") was given with feedback to ensure the participant understood what was required, after which all 28 items were presented from audiotape. The task was to repeat back the spoken nonword, and responses were scored on-line as right or wrong.

Nonsense passage reading. The nonsense passage reading test devised by Gross-Glenn et al. [1990] was given to all participants aged 8 years and over. This consisted of two textual passages that incorporated nonsense words, which participants were required to read aloud. For example, the first sentence of passage 1 went as follows: "Once upon a time a tawndy rapsig named Gub found a tix of pertollic asquees."

Self-Report Measures in Parents

Parents were mailed two self-report questionnaires. The first of these was the Autism-Spectrum Questionnaire [Baron-Cohen et al., 2001], a self-report inventory for adults aimed at identifying autistic characteristics. As reported by Bishop et al. [2004], on two of the scales from this questionnaire, social skills and communication, index parents obtained significantly higher scores than control parents. A composite score based on these scales was used to identify cases of the broad phenotype. The second questionnaire was designed for this study to identify developmental language and literacy problems, and can be found on the website: http://www.psych.ox.ac.uk/oscci/dbhtml/referencelists.htm

RESULTS

Verbal and Performance IQ

Comparison of control and index probands. Mean short-form verbal and performance IQs are shown for the whole sample in Table I. IQ scores were markedly lower in the index probands than control probands. A two-way ANOVA was conducted with IQ (verbal/performance) as a repeated measure and group (index vs control) as between subjects factor. The main effect of group gave F (1, 137) = 44.7, $P < 0.001$, with effect size (η^2) = 0.246. There was also a significant interaction between group and IQ, F (1, 137) = 9.03, $P = 0.003$, $\eta^2 = 0.062$, reflecting the fact that PIQ tended to be higher than VIQ in index probands.

Comparison of control and index relatives. Where there were multiple siblings of a proband, all eligible children were tested, and a mean score was computed for the brothers and sisters for each family. To assess comparability of the index and control relatives, their data were entered into a four-way

TABLE I. Mean (SD) Prorated IQ for Family Members in Index and Control Groups

	Index					Control				
	N	VIQ		PIQ		N	VIQ		PIQ	
Proband	80	75.9	(26.03)	82.78	(25.31)	59	105.29	(18.98)	101.14	(20.68)
Mother	80	102.88	(14.38)	106.33	(16.13)	57	105.81	(16.19)	106.79	(17.57)
Father	65	102.09	(17.02)	103.48	(18.33)	39	103.26	(15.07)	109.90	(13.69)
Mean sister[a]	39	102.69	(11.43)	112.64	(16.22)	36	107.80	(11.66)	107.63	(14.15)
Mean brother[a]	49	103.68	(15.78)	104.33	(15.47)	19	106.26	(16.04)	106.16	(17.88)

[a]These means include two brothers of index cases who met criteria for autism, and seven brothers and one sister of index cases who met criteria for PDDNOS. However, means were altered only slightly by their exclusion (sister VIQ = 103.2; PIQ = 113.4; brother VIQ = 106.0; PIQ = 104.8).

ANOVA with IQ (verbal/performance) as a repeated measure, and group (index vs. control), gender, and relationship (parent/sib) as between subjects factors. Siblings who met diagnostic criteria for ASD were excluded from this analysis. The main effect of group was not significant, $F(1, 368) = 0.92$; $P = 0.339$, indicating that relatives of index cases did not obtain lower scores than control relatives. However, there was a significant interaction between group, IQ test and gender, $F(1, 368) = 6.06$, $P = 0.014$, which had not been predicted. The effect size for this interaction was very small: $\eta^2 = 0.013$. This reflected a tendency for female relatives of index cases to have higher PIQ than VIQ.

Familiality of low verbal IQ. If there is a distinct subtype of autism characterised by low VIQ, then we might expect to see verbal deficits in relatives only for those probands with low VIQ. To test this possibility, index probands were subdivided into those scoring 77 or less, and 78 or more on VIQ, and the mean VIQ of relatives of these subgroups was compared (see Table II). Given the lack of consistent findings on VIQ in relatives in previous studies [see Bailey et al., 1998], we deemed it appropriate to use two-tailed tests with Bonferroni correction for multiple comparisons to guard against type I error. Although there was a trend for fathers of low-IQ probands to obtain lower scores than fathers of average-IQ probands, this was not statistically significant when a Bonferroni correction was applied. Nevertheless, it is noteworthy that in all relative groups the mean VIQ was lower when the proband had low VIQ. To give a more powerful analysis that combined information from all relatives, an ANOVA was run on the subset of 56 index families who had data for all types of relative (28 with proband VIQ below 78 and 28 with proband VIQ above 77), treating relative type (mother, father, mean sib) as a repeated measure. This gave a significant effect of proband VIQ, $F(1, 54) = 4.59$, $P = 0.037$, with effect size (η^2) of 0.078. This analysis suggests that there is a reliable association between low VIQ in the proband and VIQ of first degree relatives.

Nonword Repetition

The control sample was used as a basis for converting raw scores to age-scaled scores. Scrutiny of the relationship between age and raw score indicated that for those aged below 11 years, there was a significant linear relationship between log age and raw score (Pearson $r = 0.613$, $N = 83$, $P < 0.001$). Gender did not significantly predict raw score and was not considered further. The regression equation (control participants only) was used to convert raw scores to age-scaled z-scores by subtracting the predicted from the obtained score, and dividing by the standard error of estimate. For those aged 11 years and over (control parents, probands and siblings), there was no effect of age on raw score. Scores of all older participants were converted into z-scores on the basis of the control mean raw score of 19.87, with SD of 3.8. Finally, all z-scores were rescaled with mean of 100 and SD 15, with scale floor at 50.

TABLE II. Mean (SD) VIQ for Parents and Siblings of Index Cases, in Relation to Proband VIQ

	Proband VIQ			
	78 or above	77 or less	*t*	*P*
Mother				
N	40	39		
Mean	105.23	100.51	1.47	0.145
SD	(15.49)	(12.83)		
Father				
N	34	32		
Mean	106.91	97.94	2.14	0.036
SD	(19.03)	(14.63)		
Mean sibling				
N	33	37		
Mean	104.83	101.00	1.18	0.242
SD	(13.64)	(13.47)		

Comparison of control and index probands. Eleven index probands were unable to attempt the task; all of these cases had verbal IQ below 65. The scaled scores for the remaining probands are shown in Table III. Proband data were analyzed in two-way ANOVA, with group and gender as factors. Those in the index group scored significantly lower than those in the control group, $F(1, 124) = 11.88$, $P = 0.001$, $\eta^2 = 0.087$. The main effect of gender, and the interaction between gender and group were nonsignificant, though the small number of female probands meant the study had low power to detect such effects. A subsidiary analysis was conducted comparing index probands who met full criteria for autism with those diagnosed as PDDNOS, but there was no significant difference in means between these two subgroups: autistic group: $M = 83.61$, $SD = 21.7$, $N = 51$; PDDNOS group: $M = 80.71$, $SD = 22.81$, $F(1, 67) < 1$, $P = 0.634$.

The correlation between VIQ and nonword repetition for probands was substantial, Pearson $\underline{r} = 0.72$, $DF = 126$, $P < 0.001$. The proband sample was subdivided according to index/control status and whether VIQ was above 84, and the resulting groups were compared in one-way ANOVA, with Scheffé tests for pairwise comparisons. The mean nonword repetition score did not differ for low-VIQ index probands ($N = 36$, mean = 69.5, $SD = 15.87$) and low-VIQ control probands ($N = 6$, mean = 75.3, $SD = 17.72$). Furthermore, the mean nonword repetition score did not differ for average-VIQ index probands ($N = 33$, mean = 97.4, $SD = 18.06$) and average-VIQ control probands ($N = 53$, mean = 103.3, $SD = 13.56$). In sum, whatever factor leads to low nonword repetition in a subset of individuals with autism appears to be the same factor that causes low VIQ.

Comparison of control and index relatives. Data from relatives were analyzed in a three-way ANOVA, with group (index or control), relative type (sibling or parent) and gender as factors. The family was the unit of analysis, with the mean score for brothers and sisters used in cases where there were multiple siblings. The main effect of group was nonsignificant, $F(1, 368) < 1$, $P = 0.749$, indicating that relatives of index cases obtained scores no different from relatives of controls. As can be seen from Table III, male relatives tended to score lower than female relatives, and the main effect of gender was statistically significant $F(1, 368) = 4.99$, $P = 0.026$, though small in magnitude, $\eta^2 = 0.013$. The interaction between group and gender was nonsignificant, $F(1, 368) < 1$, $P = 0.597$.

Familiality of low nonword repetition. Index probands were divided into those who scored more than 1.5 SD below the mean (scaled score 77 or less), those scoring above this level, and those unable to attempt nonword repetition. On one-way ANOVA comparisons of the three groups, F-ratios were below 1 for all relatives. Thus, although poor nonword repetition scores were common in index probands, this deficit did not appear to be familial.

Speeded Nonsense Passage Reading

Two scores were available from this test: the total time to read the two passages, and the number of reading errors. Both scores were skewed in the control families, with the majority of participants having low scores, and there being a long tail of higher scores. To derive a single score representing performance on this test, both indices were log transformed to normalize the data, and converted to z-scores, using the control

TABLE III. Mean Age-Scaled Scores on Nonword Repetition and Nonsense Passage Reading for Index and Control Probands and Relatives

	Index			Control		
	N	Mean	SD	N	Mean	SD
Nonword repetition						
Proband	69	82.85	(16.26)	59	100.42	(12.89)
Mother[a]	77	100.51	(12.92)	57	100.24	(15.18)
Father	64	95.95	(15.64)	39	95.35	(12.84)
Mean sister	37	102.74	(14.78)	35	103.24	(16.40)
Mean brother	48	101.89	(13.06)	19	99.33	(10.25)
Nonsense reading						
Proband	39	92.21	(14.59)	33	101.96	(12.61)
Mother	80	97.06	(12.44)	57	102.81	(15.74)
Father	65	95.61	(16.63)	39	96.32	(14.06)
Mean sister	29	96.41	(9.92)	24	103.48	(12.16)
Mean brother	35	99.38	(10.92)	13	96.43	(11.33)

[a]Data excluded from two index mothers with hearing loss, and from one where background noise precluded test administration.

participants as the normative standard. These z-scores, which correlated together at $r = 0.674$, were then averaged. Inspection of the distribution of this reading composite relative to age indicated that there was a linear trend for improvement with age up to around the age of 18. For participants aged under 18 years, the regression of the composite reading score on age for the control sample was computed, with the regression equation used to compute an age-adjusted standard score with mean 100 and SD 15, and floor score of 50. For those aged over 17 years, the mean and SD of the composite score were used to compute standard scores.

Comparison of control and index probands. There was a substantial difference between mean scores of index and control probands, (see Table III) $F\ (1,\ 70) = 9.03$, $P = 0.004$, $\eta^2 = 0.114$. As had been done for the nonword repetition test, a further analysis was conducted to consider how poor nonsense passage reading related to VIQ, with index and control probands subdivided according to level of VIQ. The overall correlation between nonsense passage reading and VIQ was 0.70, $DF = 72$, $P < 0.001$. The pattern of results was the same as for nonword repetition. Thus, the reading scores of index cases of low VIQ ($N = 16$, $M = 84.13$, $SD = 14.87$) did not differ from that for controls of low VIQ ($N = 3$, $M = 77.11$, $SD = 5.54$), and the reading scores of index cases of average VIQ ($N = 23$, $M = 97.84$, $SD = 11.66$) did not differ from those of control cases of average VIQ ($N = 30$, $M = 104.45$, $SD = 10.14$).

Comparison of control and index relatives. Data for relatives were analyzed with group (index/control), and relative type (parent/sib) as factors in an ANOVA. All main effects were nonsignificant: group, $F\ (1, 334) = 2.35$, $P = 0.127$; relative type, $F\ (1, 334) = 0.32$, $P = 0.572$; gender, $F\ (1, 334) = 3.02$, $P = 0.083$. There was a small but significant interaction between gender and group, $F\ (1, 334) = 4.76$, $P = 0.030$, $\eta^2 = 0.014$, reflecting lower scores in female relatives of index versus control cases.

IQ and Phonological Processing in Relation to Proband's Language Development

Several recent molecular genetic studies have suggested that evidence for linkage is clearest when the sample is restricted to probands with severe early language delay. Following Buxbaum et al. [2001] we subdivided the index group according to item 13 of the ADI-R, "age of first phrases," into an "average" and "late" phrase speech group, according to whether or not phrase speech was present by 36 months. Proband and relative IQ scores and phonological scores are shown in relation to this subdivision in Table IV. The pattern of results is similar to that obtained when probands were subdivided on VIQ, in that there is a trend for parents of probands with average phrase speech to have higher VIQ than parents of those with late phrase speech, which reaches significance for fathers if no correction is applied for multiple comparisons. However, when data from all relatives were entered into ANOVA with relative type as a repeated measure (38 families of probands with late phrase speech, 18 with average phrase speech), there was no main effect of proband's phrase speech status on relatives' VIQ, $F\ (1,\ 54) = 1.31$, $P = 0.257$. Comparisons of relatives of the two phrase speech groups on PIQ and the phonological measures gave no significant differences. A similar pattern of results was seen when index probands were subdivided according to whether age at first words was below 21 months.

Relationships Between Broad Phenotype Status and Language Measures in Parents

In a previous report on this sample [Bishop et al., 2004], we demonstrated a higher rate of self-reported social-communication difficulties on the Autism Spectrum Quotient [AQ: Baron-Cohen et al., 2001] among parents of children with autism compared with parents of control children. The AQ assesses current rather than past communicative functions, and the difficulties it detects have more to do with appropriate use of communication than with verbal abilities. Table V shows mean scores on language measures for parents in relation to their status on the AQ. Parents with a total score of 1.3 or above on a principal component score from the communication and social skills scales were categorized as cases of broad phenotype (BP+), and those scoring below this score were categorized as not broad phenotype (BP−) for this analysis. The cutoff score of 1.3 which was selected to minimize the number of control parents included as cases of BP+, while still giving adequate numbers of index BP+ cases for analysis, corresponds to a summed raw score on these scales of 11 or more. Data from control parents are shown for comparison, although the number meeting the criterion for broad phenotype is too small to warrant statistical analysis. Within the index group, we found no association between broad phenotype status and scores on any of the verbal measures (all F-ratios <1).

Language development history in relation to broad phenotype status. Language history questionnaires were completed by 78 mothers and 60 fathers of index families, and 53 mothers and 38 fathers of control families. Questions 1–4 were used to exclude parents whose language may have been affected by physical disability or adverse home environment.

TABLE IV. Mean and SD of Test Scores for Index Families Subdivided by Proband Phrase Speech Status at 36 Months

	With phrase speech			No phrase speech				
	N	Mean	SD	N	Mean	SD	F	*P*
VIQ								
Proband	27	91.52	(27.85)	53	68.89	(22.71)	15.21	<.001
Mother	27	106.85	(14.82)	52	100.85	(13.79)	3.20	0.077
Father	24	108.29	(18.1)	42	99.29	(16.49)	4.24	0.043
Mean sib	21	100.14	(14.32)	49	103.94	(13.25)	1.15	0.287
PIQ								
Proband	27	95.04	(24.36)	53	76.53	(23.63)	10.75	0.002
Mother	27	110.74	(18.3)	53	104.08	(14.57)	3.14	0.080
Father	24	103.63	(19.08)	41	103.39	(18.12)	.002	0.961
Mean sib	21	108.94	(10.7)	47	106.21	(16.92)	0.46	0.499
Nonword repetition								
Proband	26	89.44	(22.17)	43	77.73	(22.91)	4.34	0.041
Mother	26	100.67	(13.74)	51	100.44	(12.62)	0.01	0.942
Father	23	97.25	(14.36)	41	95.22	(16.43)	0.25	0.621
Mean sib	21	104.00	(10.31)	46	101.31	(14.5)	0.58	0.448
Nonsense reading								
Proband	14	95.66	(11.51)	26	90.72	(15.78)	1.06	0.310
Mother	27	98.41	(13.21)	53	96.37	(12.1)	0.48	0.491
Father	24	96.18	(18.68)	41	95.20	(15.8)	0.05	0.821
Mean sib	16	97.03	(8.74)	37	98.21	(10.17)	0.16	0.689

None of these items significantly differentiated index from control parents. Overall, 29 of 138 index parents and 11 of 91 control parents were excluded from analysis of language development history on the basis of a positive response to at least one of these questions.

Table V shows mean summary scores on the language history questionnaire for index cases subdivided according to broad phenotype status, and for control cases in the BP− subgroup (the one BP+ control parent who had useable data on the language history questionnaire is excluded). The language/literacy total score was formed by summing responses to items 5 to 10 and 14 to 15. For each item, a report of a definite problem was rated as two, a possible problem as one, and no problem as zero, so a high score corresponds to a history of difficulties. Item 15, which asked about competence in spelling was coded so that a "definitely good speller" response was scored as zero, a "somewhat" response as one, and a "not a good speller" response as two. When the three groups were compared using ANOVA, there was a significant group effect, $F(2, 151) = 3.82$, $P = 0.024$. Scheffé tests indicated that the index BP+ group differed significantly from the other two groups at the 0.05 level, but that the index BP- and control groups did not differ from one another.

DISCUSSION

This study confirmed that probands with an autistic spectrum disorder are impaired relative to typically developing controls on two tests that involve phonological processing of spoken and written words, namely nonword repetition and nonsense passage reading. However, there was no indication of disproportionate phonological difficulties in autistic people of normal verbal ability.

Impairments of nonword repetition were not heritable in probands with autism, insofar as there was no relationship between poor scores of probands and their relatives. This contrasts with studies of specific language or literacy problems, where nonword repetition deficits appear heritable and may be found even in individuals who do not have measurable deficits on other language tests [Bishop et al., 1996, 1999; Raskind et al., 2000].

There was some evidence of familiality on the VIQ measure. Nevertheless, the effect was small, and mean VIQ of relatives of probands with low VIQ was well within normal limits. Parental VIQ (but not sibling VIQ) also showed some relation to delay in early language milestones in index probands, whereas the phonological measures did not.

Unlike previous studies, we did not find an increased rate of self-reported language and literacy difficulties in parents of autistic children in general compared with control parents. However, when the Communication and Social Skills scales of the AQ were used to define the broad phenotype in index parents, there was an increased reportage of early language and literacy problems in those with the broad phenotype. However, those with the broad phenotype did not differ from other index parents on direct measures of verbal IQ, nonword repetition or reading.

There are some similarities between our findings and those of Fombonne et al. [1997], who found that parents of children with autism had higher rates of self-reported language delay than control parents, but did not differ when assessed on objective measures of language and literacy. There are two possible explanations for this pattern of results. One is that self-report yields inflated estimates of communication impairments in relatives of people with autism. It is plausible that parents of a child with autism may become more sensitive to indicators of communicative difficulty after reading about

TABLE V. Mean (SD) Scores of Parents on VIQ and Phonological Tasks in Relation to "Broad Phenotype" Classification on the AQ

	Status	N	VIQ		Nonword repetition		Nonsense reading		Language/literacy history[a]	
Index	BP−	94	104.21	(15.69)	99.28	(17.62)	97.84	(13.28)	1.2	(1.49)
	BP+	20	105.50	(13.15)	99.33	(8.21)	95.75	(14.48)	2.5	(2.20)
Control	BP−	84	103.30	(19.09)	97.95	(14.88)	100.93	(14.87)	1.1	(1.66)
	BP+	3	95.67	(22.50)	95.25	(9.94)	91.02	(14.14)	—	

[a]N = 86, 16, and 80 respectively.

autism and attempting to understand their child's difficulties. An alternative explanation is that the communication difficulties that are identified by self-report are genuine, but are different in kind from those assessed by tests of verbal IQ or phonological processing. As Landa et al. [1991] noted, it is possible to have a normal verbal IQ yet still have marked pragmatic impairments. Unfortunately, pragmatic difficulties are much harder to evaluate objectively than structural language skills and few studies have included pragmatic measures. Landa et al. [1991, 1992] found higher rates of pragmatic impairment in parents of children with autism than in controls, but Pilowsky et al. [2003] failed to find any evidence of pragmatic deficit, or indeed of developmental language difficulties, in siblings of children with autism.

Our results suggest that the kinds of phonological difficulties seen in SLI are not typically part of the heritable phenotype seen in autism. We have confirmed that there are similarities in structural language deficits seen in SLI and autism, but these may be phenocopies rather than evidence of a common underlying etiology. It is possible that we may have more success in identifying communicative features of the broad phenotype if we move away from measures that are sensitive to SLI, and focus more on instruments that assess use of language in social communication.

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