Communication and Social Deficits in Relatives of Individuals with SLI and Relatives of Individuals with ASD

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Abstract We investigate two aspects of the autism triad, communication and social difficulties, in relatives of specific language impairment (SLI) probands (with and without additional autistic symptomatology) as compared to relatives of autism spectrum disorder (ASD) and Down’s syndrome (DS) probands. Findings involving 726 first degree relatives of 85 SLI, 99 ASD and 36 DS probands revealed a higher rate of communication difficulties in relatives of both subgroups of SLI probands compared to ASD and DS relatives. Similar levels of social deficits were found in relatives of SLI + ASD and ASD probands. There was a higher than would be expected rate (4.3 %) of ASD, particularly in siblings of SLI + ASD probands. Communication and social deficits appear to breed true in SLI and ASD.

Keywords Communication and social difficulties in ASD · Familial deficits · Specific language impairment (SLI) · Autism spectrum disorder (ASD) · Down’s syndrome (DS)

Introduction

The link between specific language impairment (SLI) and autism spectrum disorders (ASD) has been extensively investigated in recent years. Both groups of individuals are known to have language and communication deficits. In SLI, key areas of difficulties include grammar and phonological short-term memory (Bishop et al. 1996; Rice 2000) whilst in ASD difficulties with pragmatics and the social use of language are more common (Tager-Flusberg et al. 2005), suggestive of different etiologies. However, Tager-Flusberg and colleagues (Kjelgaard and Tager-Flusberg 2001; Tager-Flusberg and Joseph 2003) have presented evidence and argued that there is a distinct subgroup of individuals with ASD who also have similar structural language difficulties to those seen in SLI. They suggest a likely shared etiology between the two disorders. There is also evidence that individuals with ASD can present difficulties with phonological processing (Bishop et al. 2004), similar to individuals with SLI. These data have prompted much debate regarding the etiology and heritability of impairments in SLI and ASD (Whitehouse et al. 2007; Williams et al. 2008). Within this context, family history studies comparing the two disorders are of interest. Understandably, the majority of the research has focused on the overlap in the language and communication difficulties of relatives of SLI and ASD probands. This paper aims to further our knowledge in this area by investigating both communication and social deficits in relatives of SLI probands and compare them with the relatives of probands with ASD.

It has been established that there are wider language and communication difficulties in the relatives of individuals with SLI, specifically language delay, structural language problems and reading difficulties (Falcaro et al. 2008;
Lahey and Edwards 1995; Rice et al. 1998; Spitz et al. 1997; Tomblin 1989). On the one hand, the communication problems in the relatives of individuals with ASD share some common aspects with those of individuals with SLI, such as language delay and reading difficulties (Fombonne et al. 1997), although reading difficulties appeared to be more marked in relatives of individuals with SLI. On the other hand, there appear to be differences in the language deficits observed in the relatives of individuals with ASD and SLI. In particular, the parents of children with ASD exhibit more pragmatic difficulties in language use when compared to parents of children with SLI (Ruser et al. 2007; Whitehouse et al. 2007). Furthermore, parents of probands with ASD do not appear to have phonological short-term memory deficits or more generalised phonological processing problems than those of control families (Bishop et al. 2004). High heritability of phonological short-term memory deficits in SLI as indexed by nonword repetition tasks was first shown by Bishop and colleagues (Barry et al. 2007; Bishop et al. 1996) and has been replicated extensively (Falcaro et al. 2008). Taken together, this evidence suggests potential different etiologies for the communication deficits observed in SLI and ASD.

In addition to communication difficulties, social difficulties have also been documented in the families of ASD probands (e.g. Bolton et al. 1994; Piven et al. 1997). However, relatively sparse attention has been given to social difficulties in the relatives of SLI probands. One study of families identified on the basis of a proband with SLI provides preliminary evidence of a higher rate of autism in the siblings of SLI probands (Tomblin et al. 2003).

Changing Diagnostic Criteria or Developmental Processes?

The debate regarding the shared or distinct etiology and heritability of impairments in SLI and ASD has also been fuelled by findings of subgroups of individuals with autism and language impairment (ALI; Tager-Flusberg 2006) and subgroups of individuals with SLI and autistic symptomatology (SLI + ASD; Conti-Ramsden et al. 2006). Researchers working with cases of diagnosed autism have argued that individuals with ALI have profiles of language difficulties that are similar to those found in individuals with SLI, suggesting some shared etiology in ASD and SLI (Kjelgaard and Tager-Flusberg 2001; Tager-Flusberg 2006). However, it has been pointed out that, although the profiles of language impairment in ALI and SLI may be similar at a particular point in time, the developmental trajectories of language difficulties in individuals with ALI and individuals with SLI are different (Williams et al. 2008). For example, the structural language deficits seen in children with ALI appear, at least to some extent, to resolve over time, whilst deficits in this area continue to be substantial in the majority of individuals with SLI.

From the starting point of cases of SLI, there is increasing evidence that a proportion of children with diagnosed SLI in childhood exhibit autistic symptomatology in later life. Conti-Ramsden et al. (2006) found a higher prevalence of autism at age 14 than what would be expected in the general population in children with SLI recruited at age 7. They also found evidence of broader phenotype ASD symptomatology in some adolescents with SLI (see also Leyfer et al. 2008). This is consistent with previous research indicating an increase in autistic symptoms during adulthood in individuals with a history of language impairment in childhood (Howlin et al. 2000).

The controversy focuses on whether these cases are the result of changes in diagnostic boundaries in autism, whereby broader phenotype autism symptomatology is now considered as evidence for a diagnosis of autism. Bishop et al. (2008) found that a proportion of children diagnosed with SLI before the mid 1990s actually met modern day autism criteria in adulthood. They argue these individuals are likely to have been “missed” cases of autism given that the diagnostic criteria at the time of their diagnosis were more stringent than what is currently used. In effect these authors are suggesting that these cases were very likely, all along, individuals with ALI, although it needs to be noted that these authors do not use the ALI terminology in their study.

Conti-Ramsden et al. (2006) argue against the idea that these individuals are misdiagnosed cases of ALI and against the notion that changes in diagnostic boundaries are responsible for their findings with adolescents with SLI. They propose that some children with SLI develop autistic symptoms in adolescence and are therefore cases of individuals with SLI with autistic symptomatology and not individuals with ALI. Conti-Ramsden and colleagues (St. Clair et al. 2010) suggest that developmental processes and experiences of individuals with persisting language difficulties are likely to be responsible for the emergence of autistic symptomatology in adolescence. It is important to note that the individuals in the Conti-Ramsden et al. (2006) study were recruited in the mid 1990s, after the publication of DSM-III-R which used the broader diagnostic criteria than DSM-III, which was very likely used in the samples studied by Bishop et al. (2008). Furthermore, nearly two-thirds of the individuals in the Conti-Ramsden et al. study had no history of autistic symptoms earlier in life (at 4–5 years of age).

We acknowledge that this controversy is far from being resolved. Nonetheless, given the fact that the individuals with SLI in this investigation were the same individuals as those participating in the studies by Conti-Ramsden et al.
(2006; St. Clair et al. 2010), we use the terminology that has been used previously and refer to these individuals as individuals with SLI with autistic symptomatology (SLI + ASD).

The Present Study

This paper investigates the prevalence of communication and social deficits in the relatives of SLI probands (with and without additional autistic symptomatology) using the Family History Interview (FHI; Bolton et al. 1994). We were particularly interested in the comparison between relatives of SLI probands and relatives of ASD probands. Nevertheless, we used two groups for comparison—the relatives of ASD probands and the relatives of Down’s syndrome (DS) probands. The inclusion of relatives of DS probands provided an opportunity for comparisons of SLI and ASD with DS, a non-heritable disorder. Both the ASD and DS relatives were previously studied in a series of papers by Rutter and colleagues (Bolton et al. 1994; Murphy et al. 2000; Pickles et al. 2000).

This investigation aims to develop our understanding of how SLI relates to autism spectrum disorders both in terms of heritability and etiology. First, we examine whether types of deficits breed true in the two disorders, that is, compared to relatives of ASD probands we expect relatives of SLI probands to have more communication difficulties, but fewer social deficits. The converse is expected with social deficits, so that we expect relatives of ASD probands to have more social deficits and fewer communication difficulties when compared to relatives of SLI probands. Our particular contribution to this analysis is the inclusion of relatives of probands with SLI + ASD and the examination of their communication-social profiles. Second, we examine broad domains of language and communication: articulation, language delay, reading and spelling, in order to examine similarities and differences across groups and potential overlap in etiology between SLI and ASD. Third, we carry out further analyses with the relatives of probands with SLI in order to better understand the patterns of heritability and familial loading in SLI. We investigate the rate of ASD in SLI siblings, with particular reference to examining differences between siblings of SLI-only probands versus siblings of SLI + ASD probands. We also provide more detail on relatives of SLI probands such as receipt of speech therapy, communication milestones, and types of communication difficulties in relatives. We examine familial loading in relation to characteristics of the relatives (gender, parents vs. siblings) and proband characteristics (language ability, nonverbal ability, nonword repetition skills, presence of ASD symptomatology) as well as family clustering.

Methods

Participants

There were 726 first degree relatives related to 85 SLI probands, 99 ASD probands and 36 DS probands. DS is an established control group for family studies, as it is a genetic disorder which is not inherited. Thus, this provides us with a control sample of families who have a disabled child, but no heritable component that could potentially impact other family members, which is the case for both ASD and SLI. The ASD and DS participants were drawn from the database of Bolton and colleagues and are described in Bolton et al. (1994). The SLI probands were part of the Manchester Language Study (Conti-Ramsden and Botting 1999a, b). Relatives were included in this investigation if information was available on both the communication and social domains examined with the FHI (see section below).

Background Information on the Proband Groups

The SLI probands were recruited from 118 language units, which were specialist education units for children with primary language difficulties. Language units generally take children who have nonverbal abilities within the normal range and severe enough language difficulties so that they require intensive support and a statement of special educational needs. A random sample of half of all 7 year old children who were spending more than 50 % of their time in a language unit in England were recruited for the study. Children with a diagnosis of autism were excluded from the study. Eighty-five families of these probands agreed to participate in this follow-up investigation when the probands were 14 years old. For further details see work of Conti-Ramsden and colleagues (Conti-Ramsden and Botting 1999a, b; Conti-Ramsden et al. 1997).

The families of individuals with diagnosed autism were recruited from the clinical cases at the Maudsley Hospital Children’s Department. There were 110 randomly selected possible participants (aged between 5 and 36). Nine families refused to take part in the study and two further probands were excluded (one due to IQ < 30 and the other tested positive for Fragile X syndrome), leaving 99 participating families. The probands with autism already had a clinical diagnosis but this diagnosis was confirmed by administering the Autism Diagnostic Instrument-Revised (ADI-R; Lord et al. 1994) and the Autism Diagnostic Observational Schedule-Generic (ADOS-G; Lord et al. 2000). All probands were confirmed as meeting ICD10 and DSM-III-R criteria for autism. For further details see Bolton et al. (1994).
The Down’s syndrome families were recruited from a sample originally identified by Gath and Gumley (1986). Fifty families were selected from the total 104 eligible families to match the Autism families as closely as possible. Twelve families either could not be traced or were not willing to participate. Two further families were excluded as one of the probands had died and the other family had a recognised further genetic disorder. Therefore, there were 36 Down’s syndrome probands and their families included in this study. Where it was possible to determine from the existing karyotype information, all probands had trisomy 21. See Bolton et al. (1994) for further details.

The SLI probands were assessed using the gold standard diagnostic instruments for autism (ADI-R and ADOS-G), the to establish the presence of autistic symptomatology at 14 years of age as per Conti-Ramsden et al. (2006). There were 56 individuals who did not show any evidence of autistic traits, who are referred to the SLI-only group. However, 26 individuals (the SLI + ASD group) had evidence of some broader ASD features, but did not meet the strict criteria for full diagnoses, i.e., they met criteria for autism or ASD on the ADOS-G and/or displayed an impairment level difficulty in the social domain of the ADI-R in addition to having an impairment on one of the two remaining domains, communication or stereotypical behaviour (Risi et al. 2006). The families of a further three probands, referred to as “other SLI” provided family history but declined to participate in the autism evaluation of the proband. See Table 1 for details of all relatives participating in the study.

For most analyses the SLI sample is split into the SLI-only and SLI + ASD subgroups, but where appropriate all SLI probands are combined. As is customary in family studies, not genetically related relatives, i.e., adopted first degree relatives and partially genetically related relatives, i.e., half-sibs, were excluded.

**Measures**

**Relatives**

Obtaining family history information is a well-known, reliable method used in family studies of psychiatric disorders (Ptok et al. 2001). In this study we used a modified version of the FHI that had been developed for use with the ASD and DS families. This interview has been shown to be a reliable and valid schedule to identify deficits of the autism triad: communication impairment, social dysfunction and stereotypic behaviours (Bolton et al. 1994). The focus of this study was on communication impairments and social deficits. Following Bolton et al. (1994) we used the same items related to difficulties in the areas of language delay (no phrase speech by 33 months), reading (required remedial help), articulation (difficulty being understood/speech therapy), or spelling (frequent spelling errors) to form an indicator of a communication deficit. The social deficit was defined in terms of difficulties in the areas of social dysfunction, impaired social play, impaired friendships, impaired conversation, and inappropriate or odd behaviour. The modified version of the FHI used in this study had some minor changes in the wording of the interview questions. The exact items used with the relatives of the SLI sample as directly compared with the wording used with the ASD and DS families are presented in Table 1.

**Table 1** Distribution and mean ages in years (with SD and range in brackets) of the 726 relatives by proband group (SLI-only, SLI + ASD, ASD and DS)

| Family Type | SLI-only | SLI + ASD | Other SLIa | ASD | DS |
|-------------|----------|-----------|------------|-----|----|
|             | N        | Age (SD)  | Age (SD)   | N   | Age (SD)  | N | Age (SD)  |
| Parents     | 103      | 43.0 (6.0)| 45.0 (6.2)| 5   | 45.8 (7.1)| 193 | 49.9 (8.9)| 70 | 52.3 (8.6) |
|             |          | [28–56]   | [32–63]    |     | [37–52]   |   | [31–79]   |    | [35–79]    |
| Mothers     | 54       | 41.8 (5.7)| 42.8 (5.6)| 3   | 44.0 (7.5)| 97  | 48.5 (8.0)| 35 | 51.5 (8.5) |
|             |          | [28–52]   | [32–56]    |     | [37–52]   |   | [31–65]   |    | [35–68]    |
| Fathers     | 49       | 44.3 (6.1)| 47.6 (6.0)| 2   | 51.0 (–)  | 96  | 51.2 (9.5)| 35 | 53.2 (8.7) |
|             |          | [33–56]   | [40–63]    |     | [32–79]   |   | [32–79]   |    | [38–79]    |
| Siblings    | 79       | 15.9 (3.9)| 16.1 (3.4)| 6   | 21.6 (9.7)| 134 | 21.2 (6.8)| 63 | 25.0 (8.2) |
|             |          | [8–28]    | [10–25]    |     | [8–42]    |   | [8–42]    |    | [8–41]     |
| Sisters     | 34       | 15.5 (2.6)| 16.5 (2.1)| 3   | 15.3 (6.4)| 58  | 21.6 (6.6)| 34 | 25.4 (9.2) |
|             |          | [10–22]   | [12–21]    |     | [8–41]    |   | [8–41]    |    | [8–41]     |
| Brothers    | 45       | 16.2 (4.6)| 15.8 (4.2)| 3   | 31 (–)    | 76  | 20.9 (7.0)| 29 | 24.6 (7.1) |
|             |          | [8–28]    | [10–25]    |     | [8–42]    |   | [8–42]    |    | [8–41]     |
| Total relatives | 182 | 73 | 11 | 327 | 133 |

a Other SLI: those who had FHI but no autism evaluation of the proband
Table 6 in the Appendix. The modified version of the FHI also included additional questions about communication competence which were administered to relatives of SLI probands only (see Table 4 for a summary of these items).

All siblings of SLI probands were screened for autism and other pervasive developmental disorders (PDD) using the Social Communication Questionnaire (SCQ; Berument et al. 1999), completed by the primary caregiver. The siblings scoring over the cutoff for PDD in the SCQ were then assessed for autism using the ADI-R and ADOS-G (Lord et al. 1994, 2000).

SLI Probands

Total language score (TLS) was ascertained via the Clinical Evaluation of Language Fundamentals Third Edition (CELF-3; Semel et al. 1995). The Wechsler Intelligence Scale for Children Third Edition (WISC-III; Wechsler 1992) was used to determine Performance IQ (PIQ). The Children’s test of Nonword Repetition (Gathercole and Baddeley 1996) provided a measure of phonological memory.

Analysis

We fitted binary and ordinal logistic regression models for the majority of the comparisons. Since substantial shared genetic, environmental and reporting factors are likely in family datasets, we accounted for the lack of independence of the datapoints by the use of clustered-by-family robust standard errors throughout all analyses (unless otherwise specified). For analyses which had sparse levels of impaired individuals (affectedness rate below 5%) asymptotic methods could have been unreliable. In these cases we used exact binary logistic regression. These exact models did not allow us to control for the lack of independence. However, the need to control for the lack of independence diminishes with increasingly sparse impairment rates. For the analysis of the siblings SCQ scores (as well as other continuous scores), we used linear regression models accounting for the lack of independence in the data by using clustered-by family robust standard errors. Following the regression, we tested the intercept against the expected level of SCQ score in the general population. All models included covariates of generation and gender of the relative. All analyses were undertaken in Stata/SE (Stata-Corp 2007).

It needs to be noted that although our sample size is large, the power of our cross group comparisons is reduced by intra-familial correlation. With an intraclass correlation of 0.05, for the comparison of relatives of SLI and autism probands we have approximately 80 % power for an effect size of 0.25, or a rate comparison of 10 versus 4 % (two-tailed alpha = 0.05). Thus our comparison of the familial profile of these disorders still lacks precision and further replication would be desirable.

Results

SLI, ASD and DS Relatives

Communication and Social Deficits: Overall Analysis

Elevated rates of communication deficits were found in relatives of probands in the SLI-only group (21.4 %) compared to both ASD (8.6 %) and DS relatives (1.5 %), OR = 3.0, CI [1.64, 5.46], p < .001 for ASD relatives and OR = 19.40, CI [4.55, 82.68], p < .001 for DS relatives. However, differences in communication deficits in the relatives of the SLI-only probands and the SLI + ASD probands (12.3 %) were not significant, OR = 0.51, CI [0.20, 1.28], p = .15. Relatives of the SLI + ASD probands had similar levels of communication deficits as ASD relatives, OR = 1.53, CI [0.62, 3.79], p = .35, but more communication deficits than the relatives of DS probands, OR = 9.94, CI [2.01, 49.04], p = .005. See Table 2 for a summary of the results.

Relatives of SLI-only probands had fewer social deficits (1.1 %) than relatives of SLI + ASD probands (6.9 %) and ASD probands (7.7 %), OR = 7.04., CI [1.36, 36.40], p < .05 and OR = 0.13, CI [0.03, 0.54], p = .005, respectively. The rate of social deficits was not different between relatives of SLI-only probands and DS probands (1.5 %), OR = 0.69, CI [0.10, 4.89], p = .71. There was no difference in rate of social deficits between the relatives of SLI + ASD and ASD probands, OR = 0.90, CI [0.34, 2.41], p = .84, but there were marginally more social impairments in the relatives of SLI + ASD probands when compared to the relatives of DS probands, OR = 4.88, CI [0.94, 25.23], p = .06.

Communication and Social Deficits: Individual Item Analysis

Deficits in the communication domain were most prevalent in relatives of SLI-only and SLI + ASD probands. Thus, we investigated more closely how the SLI relatives (parents and siblings) differed from the ASD and DS relatives on the specific items that make up the communication deficit. As there was no difference in the communication domain between the relatives of the SLI-only and SLI + ASD probands, we combined the relatives of all SLI probands in the following analyses. See Table 3 for a summary of the results of the individual communication items.
There was no difference in the proportion of parents reported to have delayed language, \( p > .1 \). However, the siblings of SLI probands had a higher rate of language delay than the DS siblings, OR = 4.83, CI [1.07, 44.91], \( p < .05 \), though they did not differ from the siblings of ASD probands, OR = 1.63, CI [0.70, 3.89], \( p = .29 \).

Specific language impairment relatives had more reading difficulties than ASD and DS relatives, in both the parent and sibling analysis, parent analysis: OR = 8.65, CI [2.5, 29.85], \( p < .001 \) and OR = 4.62, CI [1.06, 20.10], \( p < .05 \), respectively; sibling analysis: OR = 2.40, CI [1.15, 5.00], \( p < .05 \) and OR = 8.67, CI [2.13, 35.27], \( p < .005 \), respectively.

There were more spelling difficulties in the parents of SLI probands than the parents of ASD probands, OR = 3.03, CI [1.49, 6.15], \( p < .005 \), but the rates were similar for SLI and DS parents, OR = 1.97, CI [0.83, 4.63], \( p = .12 \). The opposite pattern was found in the sibling analysis. Siblings of SLI and ASD probands had similar levels of spelling difficulties, OR = 3.45, CI [1.35, 8.87], \( p = .01 \). There were no significant differences in terms of articulation problems in the three groups of relatives for both the parent and sibling analyses, \( p > .20 \).

The same approach was applied to specific social impairments in the relatives of the probands where these

| Table 2 | Deficits (% affected) in the relatives of SLI-only, SLI + ASD, ASD and DS probands |
| --- | --- | --- | --- | --- | --- |
| Deficit | SLI-only | SLI + ASD | ASD | DS | Comparison |
| | N | % | N | % | N | % | N | % |
| Communication | 39/182 | 21.4 | 9/73 | 12.3 | 28/327 | 8.6 | 2/133 | 1.5 |
| Social | 2/182 | 1.1 | 5/73 | 6.9 | 25/327 | 7.7 | 2/133 | 1.5 |

\( a \) This difference was marginally significant

| Table 3 | Specific communication difficulties (% affected) in the parents and siblings of the SLI, ASD and DS probands |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Communication difficulties | Parents | Siblings |
| | SLI (%) | ASD (%) | DS (%) | SLI (%) | ASD (%) | DS (%) | SLI (%) | ASD (%) | DS (%) |
| Language delay | SLI = ASD | SLI = DS | SLI = ASD | SLI > DS |
| Probable | 0.7 | 2.1 | 0.0 | 0 | 3.0 | 1.6 |
| Definite | 4.0 | 1.6 | 0.0 | 14.8 | 6.7 | 1.6 |
| Reading | SLI > ASD | SLI > DS |
| Probable | 6.6 | 1.0 | 1.4 | 3.5 | 3.0 | 1.6 |
| Definite | 5.3 | 0.5 | 1.4 | 19.1 | 8.2 | 1.6 |
| Spelling | SLI > ASD | SLI = DS | SLI = ASD | SLI > DS |
| Probable | 11.3 | 3.1 | 7.3 | 3.5 | 6.7 | 4.8 |
| Definite | 6.6 | 3.6 | 2.9 | 19.1 | 8.2 | 3.2 |
| Articulation | SLI = ASD | SLI = DS | SLI = ASD |
| Probable | 1.3 | 1.0 | 0.0 | 4.4 | 2.2 | 4.7 |
| Definite | 2.7 | 0.5 | 0.0 | 7.0 | 6.0 | 1.6 |

There was no difference in the proportion of parents reported to have delayed language, \( p > .1 \). However, the siblings of SLI probands had a higher rate of language delay than the DS siblings, OR = 4.83, CI [1.07, 44.91], \( p < .05 \), though they did not differ from the siblings of ASD probands, OR = 1.63, CI [0.70, 3.89], \( p = .29 \).

Specific language impairment relatives had more reading difficulties than ASD and DS relatives, in both the parent and sibling analysis, parent analysis: OR = 8.65, CI [2.5, 29.85], \( p < .001 \) and OR = 4.62, CI [1.06, 20.10], \( p < .05 \), respectively; sibling analysis: OR = 2.40, CI [1.15, 5.00], \( p < .05 \) and OR = 8.67, CI [2.13, 35.27], \( p < .005 \), respectively.
familial deficits were most prevalent, i.e., SLI + ASD and ASD groups. There were no significant differences in the individual items that made up the social deficits between the relatives of SLI + ASD and ASD probands, with all ps > .1.

SLI Relatives

Rate of ASD in SLI Siblings

We next investigated ASD symptomatology in the siblings of the SLI probands. First, the mean SCQ total score of the siblings of SLI probands \((M = 2.94, SD = 5.82)\) was significantly lower than the general population estimate based on this instrument as reported by Chandler et al. \(2007\) \((M = 4.7, F(1,56) = 6.20, p < .05)\). In the same vein, the proportion of individuals scoring above the cutoff \(5.7\,\%\), \(4/70\) was similar to the proportion expected in the general population \(5.3\,\%\); Chandler et al. \(2007\). The SLI-only siblings had lower mean SCQ total score at \(1.87\) \(SD = 3.64\) than the SLI + ASD siblings \(M = 5.10, SD = 8.83\), though this difference was not significant, \(\beta = -3.23, CI [-7.22, 0.77], p = .11\). Both of these means were either similar (SLI + ASD; \(F(1,19) = 0.04, p = .84\)) or lower (SLI-only; \(F(1,34) = 28.19, p < .001\)) than the mean level found in the general population (Chandler et al. \(2007\)).

Second, we examined the rate of ASD. There were two SLI-only siblings \(4.4\,\%, 2/46\) and two SLI + ASD \(9.5\,\%, 2/21\) siblings who qualified for PDD on the SCQ (all from different families), a non-significant difference \(OR = 0.43, CI [0.06, 3.34], p = .42\). Both of the SLI + ASD siblings qualified for autism on the ADI-R and either autism or ASD on the ADOS-G. One of the SLI-only siblings met criteria for autism on the ADOS-G but not on the ADI-R (the other SLI-only sibling did not consent to a full autism assessment). Thus, requiring a diagnosis of autism/ASD on both instruments gave a minimum ASD rate of \(2.9\,\%\) \(CI 0.34–9.94\) among the 70 siblings of SLI probands compared to a rate of \(4.3\,\%\) \(0.89–12.02\) when autism just one of the gold standard diagnosis instruments was required. Both these estimates were higher than what is expected in the general population (Baird et al. \(2006\)).

SLI Sample: Additional Communication Questions

The following descriptive analysis was based on the additional questions found in the modified FHI. Nearly a quarter of the siblings of SLI probands \(22.0\,\%\) and \(6.5\,\%\) of parents received some form of speech therapy. Siblings’ first words were on average evident at 18 months. Twenty-two percent of the siblings were reported to have first words after 18 months \(61\,\%\) of these individuals received speech language therapy. Age at first phrases was around 26 months. Again, \(22\,\%\) of the siblings had first phrases after 33 months \(75\,\%\) of these individuals received speech language therapy).

Table 4 presents rates of siblings and parents of SLI probands reported as having definite or probable difficulties on each of the additional communication questions included in the modified FHI. A brief description of each question is given in the table and presented separately for relatives of SLI-only and SLI + ASD probands. The questions are ordered in terms of rates of sibling impairment (from high to low) for the SLI + ASD probands.

Familial Loading and SLI Relative Characteristics

Male SLI relatives were no more likely than female SLI relatives to have at least one area of deficit \(OR = 1.07, CI [0.45, 2.60], p = .87\) for parents and \(OR = 0.43, CI [0.17, 1.11], p = .08\) for siblings. Deficits were more likely in SLI siblings than in parents, \(OR = 2.16, CI [1.23, 3.82], p < .01\).

Familial Loading and SLI Proband Characteristics

Table 5 provides the distribution of the SLI probands’ gender as a function of type of relative (parent vs sibling) and whether they exhibited social or communication deficits or not (familial deficits versus no familial deficits). Table 5 also provides means and standard deviations for proband language, nonverbal skills (PIQ), and nonword repetition as a function of type of relative and presence of familial deficits. Familial communication or social deficits in parents or siblings were not related to the gender of the SLI proband, \(ps > .10\) (see Table 5). The number of relatives of SLI probands who had social difficulties was too small to carry out statistical analysis of familial loading of this type of difficulties in SLI (see Table 2).

We conducted logistic regressions to determine if proband language, nonverbal and nonword repetition abilities were related to the family loading of communication difficulties. The proband’s TLS, nonverbal IQ, and nonword repetition ability were entered into a stepwise regression. The relative’s gender was entered as a factor at each stage. The dependent or outcome variable was relatives’ communication impairment. For the first logistic regression, the outcome variable was presence or absence of sibling communication impairment. For the second logistic regression, the outcome variable was presence or absence of parent communication impairment. TLS and nonverbal IQ were not predictive of sibling nor of parent communication impairment, \(ps > .10\). However, proband’s performance on the nonword repetition task was poorer in those...
probands who had siblings with communication impairments, $\beta = -0.10$, CI $[-0.18, -0.02]$, $p < .05$. Though the findings for nonword repetition had a similar pattern in the parent analysis, the results were not as clear, $\beta = -0.05$, CI $[-0.13, 0.03]$, $p = .21$.

We also investigated whether the presence of autistic symptomatology in the proband (SLI + ASD or SLI-only status) influenced familial loading of communication difficulties. There was no relationship between the ASD status of the proband and familial communication deficits, $ps > .07$.

**Familial Clustering in SLI**

Of the 36 SLI families affected there were 23 with one non-proband family member affected, 11 families with two members affected, one family with three affected family members and one family with four family members affected. These are similar to the rates reported in Bolton et al. (1994) for the ASD sample. Overall, the relatives with deficits were found across a wide range of SLI families, indicating our findings were not due to only a few highly impaired families.

**Discussion**

In this study we found strong evidence that communication and social deficits breed true in SLI and ASD. Relatives of probands with SLI had higher rates of communication deficits and lower rates of social deficits whilst relatives of probands with ASD had higher rates of social deficits, and
lower rates of communication difficulties. What was of particular interest was the finding that the relatives of probands where SLI is accompanied by additional autistic symptomatology (probands with SLI + ASD) were found to have similar rates of affectedness of social deficits as relatives of probands with ASD. This provides new evidence for the likely heritability of social deficits in some families with a proband with SLI and further specifies that heritability of social deficits is more likely in families where there is a proband who has additional ASD symptomatology.

Communication Difficulties

Overall, more communication deficits were found in the relatives of SLI probands compared to the relatives of ASD probands. As expected, both the SLI and ASD groups were shown to have higher levels of communication deficits in relation to DS probands (Bolton et al. 1994), as the latter is a non-heritable disorder. An examination of the general pattern of affectedness of the particular broad areas of communication difficulties studied here indicated similar level of affectedness in relatives of SLI and ASD probands for language delay and articulation. The lack of difference in language delay is perhaps not surprising, as this feature is common in both SLI and ASD (Falcaro et al. 2008). The similarities in the rate of affectedness of articulation difficulties in relatives of SLI and ASD probands, is more unexpected. It is known that children with SLI can have phonological difficulties in early childhood leading to poor pronunciation (Bishop and Edmundson 1987; Leonard 1998) whilst this is not usually the case for children with ASD (Kjelgaard and Tager-Flusberg 2001). However, the rates of affectedness were low in both SLI and ASD relatives, particularly the parents, and this may have reduced the power to identify differences.

As expected from the work of Lindgren et al. (2009), we observed elevated rates of reading difficulties in the relatives of SLI probands when compared to relatives of ASD probands. We also found this to be the case for spelling difficulties, in the data for parents, although these results were less clear for siblings. These results provide evidence of familial communication deficits in both disorders, but also suggest communication deficits in SLI and ASD are not interchangeable nor necessarily of the same magnitude. In terms of the debate regarding whether SLI and ASD have similar versus distinct etiologies to their communication deficits, these data support the view of likely different etiologies for these disorders. We would have expected to see more similarities in the patterns of difficulties observed in the relatives if the communication deficits that characterise SLI and ASD had similar etiologies (Williams et al. 2008).

Only nonword repetition ability of SLI probands was predictive of their relatives’ communication deficits. Nonverbal intelligence and overall language ability did not distinguish relatives who had communication deficits from those who did not have communication deficits. This finding, taken together with the results with ASD families reported by Bishop et al. (2004) that nonword repetition is not heritable in ASD, and that different patterns of errors are observed in SLI and ASD (Whitehouse et al. 2008) suggests that phonological short-term memory deficits as indexed by nonword repetition tasks are specific to SLI and do not appear to be part of the ASD broader phenotype. In this respect, once again, our data suggest the language deficits observed in SLI and ASD are likely to arise from different etiologies.

Approximately 25 % of SLI siblings received some form of speech therapy. The distribution of age at first words and phrases indicated a developmental shift towards delayed language acquisition in relation to what is expected in the general population. The mean age for first words at 18 months was outside the expected developmental period for typical children (Bloom 1993). Indeed, 79 % of the siblings with age of phrase speech after 33 months went on to have speech-language therapy. All together, this supports previous results indicating that the siblings of probands with SLI are at risk for language delay and are more likely to require specialised language services (Rice et al. 1998; Tomblin 1989). Broadly classified, the SLI siblings had the most difficulties with language understanding, language expression, and social use of language. However, it appears that difficulties with the social use of language were more likely to occur in siblings of probands with SLI + ASD than siblings of SLI-only probands. In particular, relatives of probands with SLI + ASD were reported to more often have an unusual manner of speaking, use unusual words and odd phrases and repeat words that they hear other people say. This is in line with previous research involving types of communication difficulties in parents of probands with SLI compared to probands with ASD, the latter exhibiting more difficulties with the social use of language (Ruser et al. 2007; Whitehouse et al. 2007). However, it needs to be noted that this is preliminary evidence based on small numbers, and requires further research. Furthermore, in this study, we did not have comparable data on the communication questions for relatives of ASD probands. Further research comparing the types of communication difficulties observed in both parents and siblings of SLI probands with and without autistic symptomatology and those of parents and siblings of ASD probands would shed light into the heritability of different aspects of language in these two disorders.

Social Deficits

As expected, there was evidence of social difficulties in the relatives of ASD probands. In contrast, there was no
evidence of higher rates of social difficulties in the relatives of SLI-only probands; the SLI-only and DS relatives were similar. This suggests that social deficits are heritable in ASD but they are less likely to be so in SLI.

However, the pattern was different among relatives of those probands with a history of SLI with autistic symptomatology, who had a higher rate of social deficits than the relatives of SLI-only and similar rates of difficulties to the relatives of ASD probands. Thus, in terms of social deficits the relatives of SLI + ASD were much more similar to the relatives of AS than SLI probands and this was also the case for the individual areas of social difficulties examined which included impaired friendships, impaired conversations and inappropriate behaviour amongst others.

This is an intriguing result that invites at least two potential explanations based on the controversy surrounding commonalities versus distinctiveness of individuals with SLI and individuals with ASD. It is possible that SLI + ASD probands are exposed to more autistic symptomatology in their families, thus having an environmental, developmental influence in the emergence of ASD symptomatology in these individuals from childhood to adolescence. Another possibility is that there is a common heritable element in families of probands with ASD and probands with SLI + ASD, that is, a distinctive basis to at least some of their difficulties they exhibit in social functioning. A direct contrast of families of probands with SLI + ASD and families of probands with diagnosed autism but with an SLI language profile, i.e., ALI, could provide valuable evidence as to how and why autistic symptomatology and SLI comes to be clustered in some families.

Finally, the results of our point estimates are consistent with a higher rate of ASD in the siblings of individuals with SLI than the 1% expected in the UK general population (Baird et al. 2006). Thus, our results support Tomblin et al.’s (2003) finding of a likely higher rate of autism in the siblings of individuals with SLI. Although these findings are in need of replication given that we are examining difficulties that have low occurrence, they suggest that the higher rate of ASD in siblings of probands with SLI is likely to be more evident in siblings of probands with SLI who also have broader phenotype ASD symptomatology.

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Appendix

See Table 6.

### Table 6

Questions used in the current paper from the 1997 version of the Family History Interview for developmental disorders of cognition and social functioning used with the SLI sample and the equivalent questions on the original FHI used with the ASD and DS samples

| FHI for the SLI sample | FHI for the ASD and DS sample |
|------------------------|-------------------------------|
| Was X adopted?         | Relative adopted; adopted?    |
| Sex of relative        | Sex of relative               |
| Month/date/year of birth | Month/day/year of birth      |
| How did X get on with school work? Did he/she have any difficulties with school work? | Difficulties in school performance associated with mental handicap; How did X go on with school work? (Any difficulties? Help required?) |
| How old was X when s/he first used words meaningfully apart from “mama” and “dada”? How old was X when s/he first said something that involved putting words together meaningfully, i.e., using two or three word phrasing? | Age at starting to talk? |
| What was X’s pronunciation (articulation) like during the first 5 years? Were there any sounds or words that he/she frequently mispronounced? | What was pronunciation (articulation) like? |
| Did X ever have any difficulties learning to read? Does s/he enjoy reading? | Difficulties in learning to read? Enjoys reading? |
| Does X have, or has s/he ever had, difficulties in spelling? | Difficulties in spelling? |
| Does/did X show an interest in having relationships with others? | Ever any problems with social relationships in childhood? Difficulty making friends? Involved in social play? Spontaneous affection toward family? Chat into-and-fro manner? Isolated, aloof? |
| Are/were his/her relationships limited because of severe awkwardness in his/her approaches to other people, or because of a failure to recognize and respond to obvious social cues or to appreciate the feelings of others? | |
Table 6 continued

| FHI for the SLI sample | FHI for the ASD and DS sample |
|------------------------|-----------------------------|
| Was X cuddly and affectionate as a young child? How did s/he show affection? | Was X cuddly and affectionate as a young child? How did X show it? |
| During the school years, how is/was X at making friends? Is/was there at least one other child of roughly the same age with whom he has had a real friendship? | Joins in with group of children? Making friends? (Popular? Solitary? Outgoing?) |
| As a child, does/did X converse in a to-and-fro manner? Does s/he initiate topics? | Talkative? Chatty? |
| As an adult, does X show an interest in having relationships with others? | How does X get on with people? Friends? Opposite sex? |
| Does/did X behave in social situations in a way that seems odd, rude or embarrassing (but is not necessarily intended by X to be so)? | Does/did X behave in social situations in a way that seems odd, rude or embarrassing (but is not necessarily intended by X to be so)? |
| As an adult, does X converse in a to-and-fro manner? | As an adult, does X converse in a to-and-fro manner? |
| After age 15, has there been at least one person with whom X has had a close friendship? | After age 15, has there been at least one person with whom X has had a close friendship? |

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