Specific Language Impairment in Families: Evidence for Co-Occurrence With Reading Impairments

Judy F. Flax
Teresa Realpe-Bonilla
Linda S. Hirsch
Rutgers, The State University of New Jersey, Newark

Linda M. Brzustowicz
Christopher W. Bartlett
Rutgers, The State University of New Jersey, Newark/Piscataway
University of Medicine and Dentistry of New Jersey, Newark/Piscataway

Paula Tallal
Rutgers, The State University of New Jersey, Newark
Scientific Learning Corporation, Oakland, CA

Two family aggregation studies report the occurrence and co-occurrence of oral language impairments (LIs) and reading impairments (RIs). Study 1 examined the occurrence (rate) of LI and RI in children with specific language impairment (SLI probands), a matched control group, and all nuclear family members. Study 2 included a larger sample of SLI probands, as well as their nuclear and extended family members. Probands and their family members who met specific criteria were classified as language and/or reading impaired based on current testing. In Study 1, the rates of LI and RI for nuclear family members (excluding probands) were significantly higher than those for control family members. In the SLI families, affected family members were more likely to have both LI and RI than either impairment alone. In Study 2, 68% of the SLI probands also met the diagnostic classification for RI. The language and RI rates for the other family members, excluding probands, were 25% and 23% respectively, with a high degree of co-occurrence of LI and RI (46%) in affected individuals. Significant sex ratio differences were found across generations in the families of SLI probands. There were more male than female offspring in these families, and more males than females were found to have both Us and Rs. Results demonstrate that when Us occur within families of SLI probands, these impairments generally co-occur with Rs. Our data are also consistent with prior findings that males show impairments more often than females.

KEY WORDS: specific language impairment, reading impairment, family aggregation, sex ratio

Familial Aggregation Studies of Language Impairments

Familial aggregation of oral language impairment (LI) has been examined using several methods (see Tallal et al., 2001, for a complete review). Case history studies of large families with a history of multiple members with LI have reported ranges of 24%–63% of children with LI having at least one other impaired family member (Byrne, Willerman, & Ashmore, 1974; Gopnik & Crago, 1991; Hurst, Banaitser,

Some studies describe their participants as language impaired (LI) while others use the term specific language impaired (SLI); the terms, therefore, will be used interchangeably while reviewing the literature. Probands in this study are described as SLI, as they were selected based on both inclusionary and exclusionary criteria. Other family members are described as LI, as no IQ exclusion was used in classifying them as LI.
Auger, Graham, & Norell, 1990; Ingram, 1959; Luchsinger, 1970; Robinson, 1987). Case-control family studies have been used to determine the prevalence of LI in the family members of LI probands as compared to the general population. In most cases, the proband was clinically identified while information about the family was collected by questionnaire (Neils & Aram, 1986; Rice, Haney, & Wexler, 1998; Tallal, Ross, & Curtiss, 1989a; Tomblin, 1989). Across studies, the incidence of specific language impairment (SLI) in the family members of SLI probands was approximately 20%–25% as compared to 3%–7% for control families. When researchers used a broader phenotype (behavioral profile) that included language and learning disabilities, rates of impairment were 42% in SLI proband families and 19% in control families (Tallal et al., 1989a).

**Family Aggregation Studies of Reading Impairment**

When one family member is diagnosed with a reading impairment (RI), there is a high probability that other family members also have some type of RI (Childs & Finucci, 1979; Halgren, 1950; Sladen, 1970). The family members of reading impaired probands have been found to have a significantly higher rate of RI than the general population (Pennington, 1991). Pennington and Smith (1988) reported that in families with an affected parent, 40% of sons and 18% of daughters also had a reading problem. Scarborough (1998) reviewed eight studies in which RI in parents of probands was examined, finding that rates ranged from 25% to 60%, with a median of 46% among fathers and 33% among mothers (see Grigorenko, 2001, for a complete review).

**Sex Differences**

Results of previous family aggregation studies are not consistent in reporting sex differences in LI and/or RI. Although the majority of studies found unidirectional sex differences (more impaired males than females), rates of impairment among various family members (mothers, fathers, sisters, brothers) differed across studies. When Tomblin and Buckwalter (1994) tested the parents and siblings of SLI probands, they found that fathers and brothers had a higher incidence of LI (40% and 24%, respectively) than did mothers and sisters (15% and 6%, respectively). Lewis (1992) found that brothers of probands showed the highest incidence of LI (42%), while sisters had a rate of 22%. They did not find any differences between rates in mothers and fathers. Tallal et al. (2001) also found similar rates of impairment in mothers and fathers as well as higher rates for brothers than for sisters of SLI probands. On the other hand, Rice et al. (1998) failed to find significant sex differences in the families of SLI probands. Variations in proband ascertainment, impairment definitions, sample size, and testing methods (questionnaire vs. current testing) may all contribute to differences across studies.

**Genetic Linkage Studies**

Over the past 10 years, several studies have identified potential genetic loci based on specific phenotypic data for dyslexia and, more recently, for SLI. Significant linkage on Chromosomes 2, 6, 15, or 18 has been found in studies of individuals with dyslexia (Cardon et al., 1994; Fisher et al., 1999; Gayan et al., 1999; Grigorenko et al., 1997). Recently, two genome scans for SLI-susceptibility loci found genome-wide suggestive evidence for loci on 16 and 19 (The SLI Consortium, 2002) and significant evidence for linkage to 13 (Bartlett et al., 2002). Importantly, there was no evidence in either sample of SLI families for linkage to 7 in the region where FOXP2, the gene implicated in a severe form of oral-motor apraxia and speech impairment (which may also include a major locus for autism) was cloned (Collaborative Linkage Study of Autism, 1999/2001; International Molecular Genetic Study of Autism Consortium, 1998; Lai, Fisher, Hurst, Vargha-Khadem, & Monaco, 2001; Newbury et al., 2002). Furthermore, there was no evidence of linkage on Chromosomes 2, 6, 15, or 18, as has been reported for dyslexia.

Although behavioral studies have demonstrated a relationship between SLI and dyslexia, and many of these have also implied that there may also be a familial genetic relationship, no published studies to date have identified similar genetic loci. This may not be surprising due to substantial sampling differences and assessment strategies among studies, as well as the differences in phenotype classification used for linkage. To address this issue, Bartlett et al. (2002) recently used a reading phenotype to detect gene linkage in SLI families and found significant linkage on Chromosome 13. Chromosome 13 yielded a test statistic of 3.92 (Log10 likelihood ratio) with an associated p value of less than .01 after correction for multiple tests across the genome and accounting for multiple phenotypic classifications. This was the first study to report a significant genetic linkage for a reading phenotype in families identified based on an oral language phenotype.

Despite large amounts of behavioral research supporting relationships between RI and oral LI, there is little evidence at present of similar genetic loci, at least when comparing genes linked for dyslexia with those.
linked for speech or language disorders. However, Bartlett et al.'s (2002) finding suggests that careful phenotyping of literacy and oral language skills within the same families may someday identify genes common to both disorders.

**Early Oral Language and its Relation to Later Reading Difficulties**

Longitudinal research has indicated that children who demonstrate difficulty in developing oral language during the preschool years are at increased risk for later language, reading, and general academic difficulties. In a prospective study of early language in relation to later language and reading skills, Bishop and Adams (1990) observed that those children with both phonological difficulties and more generalized language problems had the poorest outcomes. Scarborough (1990) reported that children who exhibited poor syntactic skills and phonological production at 30 months were later identified as poor readers. In an extensive review of follow-up studies of preschool children with speech and language problems published between 1965 and 1987, Aram and Hall (1989) reported that anywhere from 40% to 100% continued to have oral language problems during the school-age years, while 50% to 75% reported having reading and other academic difficulties. In a large prospective longitudinal study, Tallal, Allard, Miller, and Curtiss (1997) annually assessed the language and emerging academic skills of 100 LI children and their age-matched controls from age 4 to age 9. The LI children showed marked deficits in math, spelling, decoding, reading vocabulary, and reading comprehension as compared to the controls, with the gap between groups widening over time.

Early LI s have been found to affect oral and/or written language abilities throughout the life span so that language and/or academic difficulties can extend through adolescence and well into adulthood. Longitudinal studies demonstrate that, unfortunately, even individuals whose early oral language difficulties appear to have been resolved are still at risk for later language-based deficits in phonological processing, literacy, and general learning difficulties (Rissman, Curtiss, & Tallal, 1990; Scarborough & Dobrich, 1990; Snowling, Bishop, & Stothard, 2000). Stothard, Snowling, Bishop, Chipchase, and Kaplan (1998) found that children whose oral language problems had been resolved by age 5 continued to perform well as adolescents in the area of language comprehension; they evidenced problems, however, in phonological processing and literacy skills. Those children who continued to have significant language problems at age 5 continued to have both oral and written language problems as adolescents.

Recent studies have shown that infants born into families with a history of language or reading problems run a greater risk of developing the same kinds of problems themselves (Benasich & Tallal, 2002; Choudhury & Benasich, 2003; Gallagher, Frith, & Snowling, 2000; Spitz, Tallal, Flax, & Benasich, 1997). In a study of children born into families with a history of language-based learning disabilities, Spitz et al. (1997) found that by age 3 years, these children scored significantly lower as a group than their matched controls on measures of language, while showing no difference in nonverbal skills. In a related family aggregation study, Choudhury and Benasich (2003) found that 3-year-olds from families with histories of SLI scored significantly lower on standardized measures of language and were more likely to fall 1 or more SDs below the mean than were those from a matched control group with no family history of SLI (28% vs. 8%). In another prospective study, Gallagher et al. (2000) studied children at risk for future reading problems because of an identified reading problem in a first-degree relative. At 45 months the “at risk” children scored more poorly than their matched controls on measures of receptive and expressive vocabulary, expressive language, nonword repetition, rhyming, digit span, and letter knowledge. By age 6 years, 57% scored more than 1 SD below the mean on measures of literacy skills.

**Co-Occurrence of RIs and LIs in Individual Children**

Recent research has addressed the issue of co-occurrence of language and reading problems in the same individual. In a review of studies that report incidence of LI in children identified as reading impaired, McArthur, Hogben, Edwards, Heath, and Mengler (2000) reported ranges of 19% to 63%, depending on the criteria used. Of those children who were identified as having SLI, 12.5% to 85% also demonstrated a concurrent RI. In their own study, McArthur and colleagues found that 55% of 110 children classified as reading impaired also had an oral language problem, while 51% of 102 children classified as having SLI had a reading disability. Bishop (2001) reported on the literacy skills of a group of twins with SLI using a twin study design to investigate possible genetic influences on language and reading. Twins with SLI who were impaired in two or three domains of language (as defined by the Diagnostic and Statistical Manual of Mental Disorders (4th ed.; American Psychiatric Association, 1994) had significantly lower literacy scores and a higher incidence of RI than controls. These studies raise the possibility that specific oral LI or specific RI may not actually be separate disorders, but rather both disorders may share some common core elements that influence their co-occurrence in many individuals, with patterns of deficit changing across development.

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Aim of Study

Previous research has suggested that oral language problems, as well as reading problems, aggregate within families (Bishop, North, & Donlan, 1995; Brzustowicz, 1996; Gilger, Pennington, & DeFries, 1991; Gopnik & Crago, 1991; Lahey & Edwards, 1995; Pennington, 1990; Rice et al., 1998; Scarborough, 1989; Tallal et al., 1989a, 2001; Tomblin, 1989; see Stromswold, 1998, 2001, for review), and that males present with these problems more often than females (Beitchman, Hood, & Inglis, 1992; Lewis, 1992; Lewis, Ekelman, & Aram, 1989; Tallal, Ross, & Curtiss, 1988b; Tomblin & Backwarter, 1994). However, the majority of these data were derived from self-report questionnaires rather than from objective standardized tests. In a recent family aggregation study (Tallal et al., 2001) in which both questionnaire and current test data were collected for all nuclear family members of SLI probands, as well as matched controls, overall familial impairment rates for SLI based on case history questionnaires were found to be similar to those based on current testing. However, examination of individual participants' data showed that different family members were identified as language impaired based on questionnaires than were identified using current testing.

In this article, we examine current test data for each family member of SLI probands to investigate familial aggregation for RI as well as co-occurrence of LI and RI. Although there is strong support for a relationship between LI and subsequent reading problems, it is clear that not all reading impaired individuals have a history of oral LI and that not all language impaired individuals develop reading problems.

In the current studies we report rates of LI, RI, and their co-occurrence within individuals from two family aggregation studies. Study 1 presents the results of current language and reading tests for SLI probands and their nuclear family members, as well as for matched control families. Study 2 presents the results of testing both nuclear and extended families that were identified as having two or more individuals in the family qualifying as SLI. This extended family study is the first of its kind to test all nuclear and extended family members with the same battery of tests.

Both studies use only a portion of a much larger database that includes language, reading, and perceptual measures, as well as DNA samples that have been collected from this cohort of families over a 10-year period (1992–2002). Other data from this genetic family study have been reported previously in Tallal et al. (2001) and Bartlett et al. (2002). Examining the relations among language and reading in families may enhance our understanding of the extent to which each of these impairments may co-occur in the same individual, how sex may affect the pattern of these impairments, and how family history may influence the potential risk of having either or both impairments.

Study 1: Case Control Study of Language and Reading Abilities

Method

Participants

Participants were 22 SLI probands, 26 matched controls, and nuclear family members (n = 174) participating in a family aggregation study of SLI. The proband group, consisting of students receiving speech/language services for SLI, and a comparable, nonimpaired control group, and the families of both, were ascertained from schools in suburban New Jersey. Specific ascertainment procedures, descriptive data, and detailed results of language data can be found in Tallal et al. (2001).

In the current study, SLI probands met the following inclusionary and exclusionary criteria:

1. Spoken language quotient (SLQ) standard scores of less than or equal to 85 on an age-appropriate test of language development (Test of Language Development–Primary: Second Edition [TOLD-P-2]; Newcomer & Hammill, 1988; Test of Language Development–Intermediate: Second Edition [TOLD-I-2]; Hammill & Newcomer, 1988; Test of Adolescent Language–Second Edition [TOAL-2]; Hammill, Brown, Larsen, & Wiederheld, 1987), or a score of less than or equal to 85 on the mean of the SLQ and the standard score (SS) on an age appropriate version of the Token Test$^3$[(SLQ + Token SS)/2].

2. Performance IQ (PIQ) of at least 80 on one of the following: Wechsler Preschool and Primary Scale of Intelligence–Revised (WPPSI-R; Wechsler, 1989), Wechsler Intelligence Scale for Children–Revised (WISC-R; Wechsler, 1974), Wechsler Adult Intelligence Scale–Revised (WAIS-R; Wechsler, 1981), or Wechsler Abbreviated Scale of Intelligence (WASI; Wechsler, 1999). For all probands, PIQ was greater than SLQ.

$^3$Z scores were used to convert scores on the Token Test for Children (DeSimoni, 1978) to a scale with a mean of 100 with a standard deviation of 15. Tomblin, Freese, and Records (1992) administered an adapted version of the Token Test for Adults (DeRenzi & Paglioni, 1978; DeRenzi & Vignolo, 1962) to 35 adults diagnosed as LI and 35 controls. These adaptations were used in this study. Scores, expressed as percentage correct, for the two groups were compared. One standard deviation below the mean for the controls corresponded to one standard deviation above the mean for the LI group, indicating that this test is clearly able to distinguish between LI and normal adults. This information was used to translate scores in current study to the same scales with a mean of 100 and a standard deviation of 15.
3. Hearing within normal limits (positive identification at 500 Hz at 30 dB SPL, and 1000, 2000, and 4000 Hz at 20 dB SPL).
4. No motor impairments or oral structure deviations affecting speech or nonspeech movement of the articulators as assessed by a speech-language pathologist.
5. No history of autism or frank neurological disorder such as mental retardation, seizure disorder, or brain injury, as determined by parental report.
6. Native English speaker with English as the primary language spoken in the home.

Control children were matched by age and required to meet Criteria 2–6. Only language criteria differed in that control children had to score within the normal range (standard score > 90 on the language test battery).

Procedure

All participants (probands, matched controls, and their nuclear families) received all the standardized language and cognitive tasks included above (SLQ, PIQ, and Token Test). In addition, all participants who were of age received two decoding subtests (Word Attack and Word Identification) of the Woodcock Reading Mastery Tests–Revised (Woodcock, 1987). Four probands and 11 of their siblings, as well as 4 control participants and their 5 siblings, did not receive the reading tasks because they were too young.

Standardized test scores were examined for each participant in order to determine which family members met the diagnostic criteria set below for current language and/or RIs.

Language Impairment (LI). An SLQ less than or equal to 85 (1 or more SDs below the mean) on an age-appropriate form of the comprehensive language development test (TOLD-P:2, TOLD-I:2, and TOAL-2) or less than or equal to 85 on the mean of the SLQ and the standard score on the age appropriate version of the Token Test.

Reading Impairment (RI). A standard score of less than or equal to 85 (1 or more SDs below the mean) on the Word Attack (nonword reading) or Word Identification (single-word reading) subtests of the Woodcock Reading Mastery Tests–Revised.

Results

Table 1 is a comparison of the SLI and control groups on age at testing and standardized measures of PIQ, language, and reading. Data are presented separately for probands, siblings, and parents. All members of the SLI group (probands, parents, and siblings) scored significantly lower on PIQ than respective members of the control group. Siblings and parents, but not probands, in the SLI groups were also significantly younger than respective members of the control groups. To eliminate the possibility that differences between the two groups on measures of language and reading could be instead attributed to differences in PIQ or age, the relationships between PIQ, age at testing, and each measure of language and reading were examined. PIQ, but not age at testing, was significantly correlated with all measures of language and reading for probands, except Word Identification. As a result, all comparisons of language and reading for probands, except Word Identification, between the SLI and control group were performed using analysis of covariance to adjust for differences in PIQ. Results indicated significant differences between groups on all measures for probands and for all measures except the Token Test for siblings and parents (see Table 1).

### Table 1. Study 1: Comparison of SLI and control groups.

<table>
<thead>
<tr>
<th></th>
<th>SLI</th>
<th>Control</th>
<th>p</th>
<th>Effect size</th>
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<tr>
<td></td>
<td>M</td>
<td>SD</td>
<td>M</td>
<td>SD</td>
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<tr>
<td>PIQ</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Probands</td>
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<td>12.2</td>
<td>111.6</td>
<td>14.2</td>
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<tr>
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<td>14.6</td>
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<tr>
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</tr>
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<td>113.0</td>
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<tr>
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<td>19.6</td>
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</table>

Note. SLI = specific language impairment; PIQ = performance IQ; SS = standard score. Asterisks indicate adjusted for differences in PIQ.
Significant differences in the rates of LI and RI for all family members (excluding probands) were also found between the SLI and control group. For this same population, the overall rate of LI for the SLI group was 30% compared to 7% in the control group, $\chi^2(1, N = 222) = 15.9, p < .01$ (Tallal et al., 2001). This rate can be compared to the overall rate of RI for the SLI group, which was 36% compared to 4% in the control group, $\chi^2(1, N = 208) = 25.9, p < .01$, in the current study. Tests of the correlation between LI and RI within each group were significant within the SLI group, $\chi^2(1, N = 101) = 32.8, p < .01$, but not within the control group, $\chi^2(1, N = 107) = 3.0, p = .08$. Of those family members in the SLI group who received both the language and reading measures, 26% had both LI and RI, while the rate was only 7% for LI alone and 9% for RI alone. This was not true in the control group. Only 1% of family members of the control group were found to have both impairments, while 6% were found to have LI alone and 3% had RI alone.

**Discussion**

Probands differed significantly from their matched controls on all standardized measures of language and reading, and their nuclear family members (parents, siblings) scored significantly lower on most measures of language and reading. For both oral language and reading the rates of impairment in the SLI families were significantly higher than the rates of impairment in the case control families. A significant relationship between the rates of LI and RI was found in the SLI families, indicating a high degree of co-occurrence within individuals. This was not true for the control families. However, the very low rates of occurrence in the control families, particularly co-occurrence of LI and RI, make any group difference in the pattern of co-occurrence extremely difficult to interpret.

Both SLI and RI are considered common disorders in the general population (i.e., they have high base rates). Tomblin et al. (1997) found that 7.4% of an unselected sample of kindergarten children met criteria for SLI, while RI among elementary school children has been estimated to be anywhere from 5% to 10% depending on criteria used (Yule, Rutter, Berger, & Thompson, 1974; Shaywitz, 1998; Shaywitz, Shaywitz, Fletcher, & Escobar, 1990). In the current study, the rates of LI and RI in the family members of SLI probands were 30% and 36%, respectively. These rates far exceed the rates found in the control families as well as those found in the general population, and they are significantly higher than would be expected to occur by chance if these were unrelated disorders. This demonstrates that there is a strong familial component, as well as co-occurrence, for LI and RI.
families had at least one other family member with an LI and agreed to participate in the extended family study. Ten additional extended families were recruited as part of our genetic linkage study for a total of 25 families. All participants (SLI probands, nuclear, and extended family members) received the same battery of standardized tests as in Study 1 and were classified as language and/or reading impaired based on the same criteria.

### Results

#### Missing Data

Of the 25 probands, 3 were too young to receive measures of reading. Of the 68 siblings, 10 were too young to receive measures of reading. Of the 103 adult extended family members, 2 (1 male and 1 female) did not take the Token Test, and this female did not receive the reading measures. Of the 52 children in the extended families, 7 (2 boys and 5 girls) did not take the Token Test and 20 (11 boys and 9 girls) were too young to receive measures of reading.

Passage Comprehension, a subtest of the Woodcock Reading Mastery Tests–Revised that assesses reading comprehension, was given to some but not all participants. Because of the high percentage of missing data (33%) on this measure, Passage Comprehension could not be used in the classification of RI. In order to determine the extent to which classification as RI might change based on the inclusion of reading comprehension, the relationships among Passage Comprehension, Word Identification, and Word Attack were examined for the participants (n = 198) for which Passage Comprehension data were available. When RI, defined by Word Attack and/or Word Identification, was compared to RI defined by Passage Comprehension, there was 89% agreement, which demonstrates the high correlation known to occur across various components of reading. Of the 11% disagreement, only 2% were participants identified as reading impaired based on Passage Comprehension but not Word Attack or Word Identification. Word Attack or Word Identification identified the remaining 9% not identified by Passage Comprehension. Thus, we can estimate that the rates of RI reported in this study based on Word Attack and Word Identification may underidentify by approximately 2% the rate of RI that would have been found in this population had reading comprehension also been included in the classification.

#### Incidence of Familial Impairment for Probands Ascertained as Having SLI

By definition, all probands (100%) met the diagnostic classification for SLI. In addition, 68% of the probands also met the diagnostic criteria for RI. Of the nuclear and extended family members (excluding probands) who took both tests of language and reading (N = 242), 25% met the diagnostic criteria for LI and 28% met the diagnostic criteria for RI. Thirty-six percent of the parents of the SLI probands met criteria as having LI and 36% met criteria as having RI. The incidence of impairment for the siblings of the SLI probands was 37% for LI and 34% for RI. The overall incidences of impairment for the nuclear

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**Table 2. Study 2: Demographic data for nuclear and extended families.**

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<tr>
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<th>Nuclear families</th>
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</tr>
<tr>
<td>Total</td>
<td>83</td>
<td>60</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

|                         | Extended families |             |           |              |
|                         | (relation to      | Male        | Female     |              |
|                         | probands)         | M           | SD         |              |
| Adultsa                 | 43   | 60  | 42  | 58          | 37.9    | 14.6       | 35.4    | 14.4       |
| Childrenb               | 34   | 18  | 65  | 35          | 7.7     | 2.3        | 7.2     | 3.3        |
| Total                   | 77   | 78  |     |             | 109.6   | 10.8       | 109.1   | 12.4       |

*a Adult relatives to probands consist of aunts, uncles, adult cousins, grandparents. b "Children" relatives to probands consist of cousins under 13; second cousins, nieces, and nephews.
family members (excluding probands) were 36% for LI and 35% for RI. The incidences of impairment for extended family members were somewhat lower, 16% for LI and 13% for RI (see Table 3).

**Co-Occurrence of Impairments**

Incidences of familial impairment for LI and RI do not necessarily reflect the actual number of people in whom these impairments co-occur. To examine the incidence of co-occurrence within individuals and across diagnostic classifications, all family members were classified according to whether or not they met the impairment criteria, first for LI and then for RI. Next, individual participants who met the diagnostic criteria for both RI and LI were identified to determine the incidence of co-occurrence of these impairments.

**Table 3. Study 2: Percentage of language and reading impairment by family group and sex.**

<table>
<thead>
<tr>
<th>Language impaired</th>
<th>Reading impaired</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Male</td>
</tr>
<tr>
<td>Probands</td>
<td></td>
</tr>
<tr>
<td>LI = 100%</td>
<td>100</td>
</tr>
<tr>
<td>RI = 68%</td>
<td></td>
</tr>
<tr>
<td>Parents</td>
<td></td>
</tr>
<tr>
<td>LI = 36%</td>
<td>48</td>
</tr>
<tr>
<td>RI = 36%</td>
<td></td>
</tr>
<tr>
<td>Siblings</td>
<td></td>
</tr>
<tr>
<td>LI = 37%</td>
<td>51</td>
</tr>
<tr>
<td>RI = 34%</td>
<td></td>
</tr>
<tr>
<td>Nuclear family (excluding probands)</td>
<td></td>
</tr>
<tr>
<td>LI = 36%</td>
<td>50</td>
</tr>
<tr>
<td>RI = 35%</td>
<td></td>
</tr>
<tr>
<td>Extended family</td>
<td></td>
</tr>
<tr>
<td>LI = 16%</td>
<td>16</td>
</tr>
<tr>
<td>RI = 13%</td>
<td></td>
</tr>
<tr>
<td>Adults</td>
<td></td>
</tr>
<tr>
<td>LI = 13%</td>
<td></td>
</tr>
<tr>
<td>RI = 11%</td>
<td></td>
</tr>
<tr>
<td>Children</td>
<td></td>
</tr>
<tr>
<td>LI = 21%</td>
<td></td>
</tr>
<tr>
<td>RI = 19%</td>
<td></td>
</tr>
<tr>
<td>Overall (including probands)</td>
<td></td>
</tr>
<tr>
<td>LI = 31%</td>
<td>43</td>
</tr>
<tr>
<td>RI = 27%</td>
<td></td>
</tr>
<tr>
<td>Overall (excluding probands)</td>
<td></td>
</tr>
<tr>
<td>LI = 25%</td>
<td>34</td>
</tr>
<tr>
<td>RI = 23%</td>
<td></td>
</tr>
</tbody>
</table>

Figure 1 shows the occurrence and co-occurrence of LI and RI for (a) probands only, (b) nuclear family members (excluding probands), (c) extended family members (separately), and (d) all family members combined (excluding probands). Only participants who received measures of both reading and language were included.

Of the SLI probands, only 32% had an LI alone, while 68% met criteria for both LI and RI. For nuclear family members only (excluding probands), 55% had no LI or RI, 10% met the diagnostic criteria for LI alone, 8% met criteria for RI alone, and 27% met criteria for both LI and RI. In the 45% of all nuclear family members with an LI and/or RI, these impairments co-occurred 59% of the time.

For the extended family members, 78% had no LI or RI, 9% met the diagnostic criteria for LI alone, 7% met criteria for RI alone, and 6% met criteria for both LI and RI. In the 22% of extended family members (excluding probands) with an LI and/or RI, these impairments co-occurred 27% of the time.

For all nuclear plus extended family members combined (excluding probands), 68% had no LI or RI, while 10% met the diagnostic criteria for LI alone, 7% met criteria for RI alone and 15% met criteria for both LI and RI. In the 32% of all family members (excluding probands) with an LI and/or RI, these impairments co-occurred 46% of the time.

**Family Member and Sex Differences**

There were significantly more male than female offspring in these families (total males = 105, total females = 74; probands: male = 21, female = 4; siblings: male = 37, female = 31; extended family members: male = 47, female = 39), \( \chi^2(1, N = 179) = 5.1, p = .02 \).

Table 4 is a summary of the mean oral language and reading scores for probands and their family members, separately by sex. To examine differences in the two measures of language (SLQs and Token Test standard scores) and the two measures of reading (Woodcock Word Attack and Word Identification) among the five family member groups (probands, parents, siblings, extended adults, and extended children) and between sexes (males and females), a series of four 5 x 2 analyses of variance were performed. Results demonstrated significant main effects for family member group on all four measures, SLQ: \( \text{F}(4, 288) = 13.7, p < .01 \); Token Test: \( \text{F}(4, 273) = 9.2, p < .01 \); Word Identification: \( \text{F}(4, 253) = 6.4, p < .01 \); Word Attack: \( \text{F}(4, 254) = 5.1, p < .01 \), and significant main effects for sex on both measures of language and reading.

\footnote{Offspring include Generations 3 and 4 (probands, siblings, and extended family first and second cousins, where \( n = 94 \) under 13 years old and \( n = 13 \) over 13 years old).}
Figure 1. Rates of occurrence and co-occurrence of language impairment (LI) and reading impairment (RI).

a). SLI Probands Only \( (N = 22) \)

- LI only: 68%
- RI only: 32%
- LI + RI: 3%
- None: 0%

b). Nuclear Families Only: Probands excluded \( (N = 108) \)

- LI only: 55%
- RI only: 8%
- LI + RI: 27%
- None: 0%

c). Extended Families Only: Probands excluded \( (N = 134) \)

- LI only: 78%
- RI only: 7%
- LI + RI: 9%
- None: 6%

d). All Family Members: Probands excluded \( (N = 242) \)

- LI only: 58%
- RI only: 7%
- LI + RI: 10%
- None: 15%

Language, SLQ: \( F(1, 288) = 5.3, \ p = .02 \); Token Test: \( F(1, 273) = 3.6, \ p = .05 \), but only one measure of reading, Word Attack: \( F(1, 254) = 4.4, \ p = .04 \); Word ID: \( F(1, 253) = 2.3, \ p = .12 \). No significant interaction effects were found.

Probands (male and female) scored significantly lower than all other family members on all measures of language and reading (see Table 4). Extended family members (adults and children) scored significantly higher than all other family members on measures of language and reading. Parents and siblings scored significantly higher than probands and significantly lower than extended family members, but were not significantly different from each other. Females scored significantly higher than males on both measures of language and on Word Attack.

Discussion

Direct testing of SLI probands, as well as their nuclear and extended family members, revealed a significant number of participants who met the study diagnostic criteria as language and/or reading impaired. Probands were selected to meet the criteria generally used in research and clinical settings to diagnose SLI, yet results showed that for both probands and their affected family members, language, and RIs were much more likely to co-occur in the same individual than to occur in isolation. The high incidence of both oral LI and RI in both the SLI probands and their nuclear and extended family members lends strong support to the hypothesis that there is a high incidence of co-occurrence among LI and RI, at least in families of SLI probands.

The problem of ascertainment bias in family genetic studies has been recognized for some time (Fisher, 1934). Ascertainment bias results when sampling is assumed to be random when in fact it is not (as in our studies). To account for this bias, we have applied what is termed single ascertainment corrections. For single ascertainment corrections to be valid, it is assumed that not all families segregating SLI have been ascertained (if all the SLI families were ascertained this would be termed "complete ascertainment"). Furthermore, the probability of ascertainment is assumed to be proportional to
Table 4. Study 2: Mean scores for language and reading measures by family member and sex.

<table>
<thead>
<tr>
<th></th>
<th>Probands</th>
<th>Parents</th>
<th>Siblings</th>
<th>Adult</th>
<th>Child</th>
<th>p value</th>
<th>Family member</th>
<th>Interaction</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Male</td>
<td>Female</td>
<td>Male</td>
<td>Female</td>
<td>Male</td>
<td>Female</td>
<td>Male</td>
<td>Female</td>
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<tr>
<td>Language</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>M</td>
<td>78.4</td>
<td>79.3</td>
<td>89.7</td>
<td>98.1</td>
<td>88.0</td>
<td>96.6</td>
<td>104.5</td>
<td>107.2</td>
</tr>
<tr>
<td>SD</td>
<td>9.3</td>
<td>6.6</td>
<td>28.5</td>
<td>16.7</td>
<td>16.0</td>
<td>15.7</td>
<td>14.5</td>
<td>14.5</td>
</tr>
<tr>
<td>Token</td>
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<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>M</td>
<td>77.2</td>
<td>79.0</td>
<td>79.7</td>
<td>92.4</td>
<td>86.8</td>
<td>96.9</td>
<td>102.7</td>
<td>100.8</td>
</tr>
<tr>
<td>SD</td>
<td>17.9</td>
<td>15.7</td>
<td>30.2</td>
<td>21.9</td>
<td>24.4</td>
<td>19.5</td>
<td>14.1</td>
<td>16.1</td>
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<td>Reading, Woodcock</td>
<td></td>
<td></td>
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<td></td>
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<tr>
<td>Word ID</td>
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<td></td>
<td></td>
</tr>
<tr>
<td>M</td>
<td>82.2</td>
<td>91.3</td>
<td>89.6</td>
<td>92.9</td>
<td>92.1</td>
<td>96.2</td>
<td>99.3</td>
<td>101.6</td>
</tr>
<tr>
<td>SD</td>
<td>16.8</td>
<td>10.7</td>
<td>18.0</td>
<td>9.7</td>
<td>16.1</td>
<td>13.6</td>
<td>10.5</td>
<td>9.5</td>
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<td>Word Attack</td>
<td></td>
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<td></td>
<td></td>
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<td></td>
<td></td>
</tr>
<tr>
<td>M</td>
<td>83.2</td>
<td>90.3</td>
<td>89.4</td>
<td>95.0</td>
<td>87.8</td>
<td>98.6</td>
<td>99.6</td>
<td>104.1</td>
</tr>
<tr>
<td>SD</td>
<td>14.9</td>
<td>13.8</td>
<td>22.5</td>
<td>15.6</td>
<td>19.8</td>
<td>16.5</td>
<td>13.8</td>
<td>13.1</td>
</tr>
</tbody>
</table>

*Standard scores with $M = 100$, $SD = 15$. 

the number of affected persons in the pedigree. As both of these assumptions are approximately correct for Study 1, simple exclusion of the proband from the analysis corrected the potential bias in co-occurrence rates. However, Study 2 represents two additional complexities for the ascertainment process that are not easily accounted for. Additional families who participated in Study 2 were recruited for a gene linkage study. Ascertainment was based on at least two affected family members (instead of just one affected family member as in Study 1) and also represented extended familial relationships. The proportion of genes shared by extended family members drops off monotonically from the proband. Taking this into account, the rates from Study 2 may not generalize to the general population. They do, however, indicate the same trends seen in the nuclear families from Study 1, which indicates that our ascertainment corrections, while not strictly correct, do approximate the true ascertainment correction. Furthermore, it will be important to determine in future studies whether this pattern of co-occurrence of LI and RI also occurs in families of a reading impaired proband.

Tallal et al. (1989a) was the first family aggregation study to address the possibility of co-occurrence of language and reading deficits in family members of SLI probands. Based on questionnaire data, family members of SLI probands were classified as "impaired" if they reported a personal history of LI, RI, and/or academic failure. Using this broader classification, a much higher incidence of impairment (42%) was found in nuclear family members of SLI probands than is generally reported in the literature based on a more focused definition of SLI alone (about 25%). In the current study, 45% of the nuclear family members (excluding probands) were found to meet criteria as language and/or reading impaired based on current testing, a similar incidence rate to that found in Tallal and colleagues.

A lower incidence of LI and/or RI (22%) was found in the extended family members. Previous studies (Lewis, 1992; Rice et al., 1998) have also reported lower incidence of impairment in extended family members than nuclear family members, and have suggested that when family history questionnaire information is used to determine the status of extended family members, rates might be influenced by how well the informant (usually the parent of the proband) knew the extended family member. However, different concerns arose in the current study. Whereas every first-degree relative of the proband was tested, only some extended family members agreed to be tested. Family members who did not agree to be tested may have included a disproportionate number who are impaired or not impaired, causing true impairment rates to be over- or underestimated. Furthermore, as shared genetics decreases with distance from the proband, so does disease risk when the underlying disease liability is largely genetic. Extended family members have spouses who are genetically unrelated.
to the nuclear family and, consequently, the gene pool is even further diluted once they have offspring.

These data may also be disproportionately influenced by families with both parents affected. In a pedigree analysis that demonstrated a high degree of co-occurrence of language, reading, and auditory rate processing impairments in a large multigenerational family, Flax et al. (2001) found a high degree of assortative mating together with a higher affection rate. Likewise, Tallal et al. (1989a, 2001) found a higher incidence of impairment in offspring when both parents were impaired. These data suggest that differences in the proportion of families with both parents affected in one study may alter the affection rate reported across studies.

In the current study the overall familial incidence of impairment (excluding probands) was significantly higher for males than for females (43% vs. 15% for language; 28% vs. 18% for reading), consistent with previous studies that have reported sex differences in LI and RI (Lewis, 1992; Tomblin, 1989; Tomblin & Buckwalter, 1994). However, there is still considerable disagreement as to whether there is truly a sex difference in these disorders. Shaywitz et al. (1990) challenged the presence of true sex differences in language or reading disorders by suggesting that reported sex differences actually reflect ascertainment bias. In an epidemiological study, they found that when children were identified as having an RI based on clinical or school referrals, boys were 2 to 4 times more likely to be identified as having an RI than girls. However, there was no significant sex difference when a comprehensive battery of standardized measures was used to identify children with RIs based on a nonselected sample. They concluded that sex differences reported in clinical samples may result from boys being referred for services more often than girls, based on behavioral rather than reading problems.

Although gender of the affected individual clearly may influence the chance that a proband will be referred for study, resulting in ascertainment bias, it is unknown how these factors in affected relatives may influence the chance that a family will be referred for study or that members of the extended pedigree will agree to participate. Therefore, although our findings are consistent with the prior reported increased rates of impairment in males, it is possible that our resulting gender difference was also influenced by ascertainment bias.

There remains considerable disagreement in the literature pertaining to the exact relationship between oral LIs (i.e., SLI) and RIs (i.e., dyslexia). Evidence from this family genetic study demonstrating the co-occurrence of oral LI and RI among family members of SLI probands, coupled with the fact that these deficits occur together in the same individual considerably more frequently than they occur alone, lends strong support to the idea that language and reading may be interrelated impairments stemming from a combination of genetic and/or environmental factors. Children diagnosed with language difficulties in preschool are known to be at greater risk not only for continued oral language problems but also for the subsequent development of literacy problems (reading, writing, and spelling) once they enter school (Aram & Hall, 1989; Bishop & Adams, 1990; Tallal et al., 1997). These facts, combined with the co-occurrence of LI and RI in families of SLI probands, are of considerable importance when looked at from the perspective of early identification and treatment of children at risk for language/learning deficits.

Family aggregation studies have consistently demonstrated that a positive family history of LI puts each child born into these families at considerably higher risk of developing similar deficits (Scarborough, 1990; Spitz et al., 1997). These findings suggest that information pertaining to family history of both LI and RI may provide valuable information to consider in making decisions pertaining to early intervention for both LI and RI. In addition, these findings highlight the importance of evaluating the oral language abilities of all children experiencing difficulty learning to read and (when appropriate) providing interventions that address both their oral and written language problems.

Comi-Ramsden and Botting (1999) suggested that "SLI is not a unitary, static condition, but a dynamic difficulty that evolves with developmental time" (p. 1202). The considerable degree of co-occurrence between LI and RI in family members of children with SLI, across generations, supports this view of developmental language impairments. Furthermore, these findings demonstrate the interrelatedness of these mental functions, both within individuals and across generations within families.

Aggregation studies alone cannot determine the extent to which genetic and/or environmental factors contribute to this pattern of co-occurrence of LI and RI in SLI probands and their family members. Expression and penetrance of spoken language disorders are rarely complete and may involve complex interactions between genes and the environment (Stromswold, 1998). This is evidenced by the fact that in the current study phenotypic profiles differed among affected family members. Gene linkage analyses currently in progress are attempting to link the phenotypic data to DNA derived genotypes from these families. Based on the pattern of frequent co-occurrence of LI and RI evident in the phenotypes of these families, both distinct genes for language impairment and genes that may show linkage for RI are currently being sought.

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Contact author: Judy F. Flax, PhD, Center for Molecular and Behavioral Neuroscience, Rutgers, The State University of New Jersey, 197 University Avenue, Newark, New Jersey 07102. E-mail: flax@axon.rutgers.edu