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# Familial Aggregation in Specific Language Impairment

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A case-control family study design, in which the current language-related abilities of all biological, primary relatives (mother, father, siblings) of probands with specific language impairment (SLI) and matched controls were assessed, was used to investigate familial aggregation for language disorders. Current test data from each family member showed the rate of language impairment for mothers, fathers, sisters, and brothers of the SLI probands to be significantly higher than for members of control families. Impairment rates for fathers and mothers were approximately equal, whereas rates for brothers were significantly higher than for sisters. In SLI proband families, Language Impairment (LI) occurred in 13.0% of offspring (excluding proband) with neither parent affected, 40% of offspring with one parent affected, and 71.4% of offspring in families in which both parents were language impaired. Rates of impairment as determined in current testing were compared directly to impairment rates estimated from family-history questionnaires collected from the same families. Group data showed impairment rates estimated from the family-history questionnaires to be similar to the rates based on actual testing. Furthermore, both appeared in line with rates based primarily on questionnaire data as reported previously in the literature. However, case-by-case analyses showed poor intrasubject agreement on classification as language impaired on the basis of current testing as compared to history information.

**KEY WORDS:** family history, specific language impairment, familial aggregation

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Over the past decade, evidence has been collected to support the hypothesis that specific language impairment (SLI)<sup>1</sup> may aggregate in families and, therefore, may contain a genetic component (Bishop, North, & Donlan, 1995; Brzustowicz, 1996; Gopnik & Crago, 1991; Lahey & Edwards, 1995; Lewis & Thompson, 1992; Rice, Haney, & Wexler, 1998; Spitz, Tallal, Flax, & Benasich, 1997; Tallal, Ross, & Curtiss, 1989; Tomblin, 1989; van der Lely & Stollwerck, 1996; see Stromswald, 1998, 2000 for a comprehensive review of family aggregation studies). Studies of how SLI aggregates in families include case-history reports (Byrne et al., 1974; Ingram, 1959; Luchsinger, 1970), large-group reports (Neils & Aram, 1986; Tallal et al., 1989; Tomblin, 1989), and case-control family studies (Neils & Aram, 1986; Tallal et al., 1989; Tomblin, 1989). Others have focused on SLI subtypes within families or examined the concordance of language impairment (LI) in monozygotic/dizygotic twin studies as a means of looking at rates of heritability (Bishop et al., 1995; Lewis & Thompson, 1992; Tomblin & Buckwalter, 1998).

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<sup>1</sup> Some studies describe their subjects as language impaired (LI), whereas others use the term specific language impaired (SLI). These terms will be used interchangeably while reviewing the literature.

To date, most studies have been based on retrospective questionnaire data collected from family members of language-impaired probands. Only one study included concurrent test data of probands as well as all of their primary relatives from families in which a child had been identified as SLI (Tomblin & Buckwalter, 1994).

### **Family Aggregation of Language Impairment**

Very early group studies that looked at family histories of children identified as language impaired reported that 28% to 63% of the children had at least one other family member who was impaired. Unfortunately, none of these studies provided enough information to determine the nature of the language problem(s). Further, the broad range of results likely reflected very different criteria used across studies for classification of impairment. Through case histories, Hurst, Baraitser, Aiger, Graham, and Norell (1990) reported the pedigree of a three-generational family containing 30 members. More than 50% (16) of the family members were affected with speech and language difficulties, the pattern of which was consistent with an autosomal dominant mode of inheritance. Gopnik and Crago (1991) studied this same family and found that of the 20 family members they tested, only 7 appeared to fall into the normal range of language development.

More recent studies have begun to provide evidence of family aggregation in quantitatively assessed populations of SLI children. Bishop and Edmundson (1986) used case history data to demonstrate a significantly increased frequency of affected primary and secondary relatives for a group of language-impaired children compared to a group of normally developing control children. Similarly, using questionnaire data, Robinson (1991) found that 28% of LI children had first-degree relatives with a history of speech delay and 20% had first-degree relatives with a history of learning problems.

Neils and Aram (1986) reported that 20% of their LI children had family members with a spectrum of speech and language problems, as compared with their control group where the rate was only 3%. Tomblin (1989) found similar results when he studied the family histories of 51 LI second-grade children and 146 control children. Family members were considered affected only if they reported having received speech/language intervention. Twenty-three percent (23%) of the LI proband families and only 3% of the control families reported a significant family history for language impairment. Tallal, Ross, and Curtis (1989) found that when families of SLI children were compared with the families of matched controls, based on self-report of either language or learning problems, the families of SLI children were more

likely to have a positive family history, higher average frequency of impairment, a higher rate of affected siblings, and a higher rate of affected mothers and fathers. A considerably higher incidence of affected family members was found in this aggregation study, as a much broader definition of language and learning problems (including general academic failure) was used. Most recently, Rice, Haney, and Wexler (1998) reported that in a case-control study that included the nuclear (first-degree) and extended family members of 31 LI children and 67 control children, using family-history data, the overall affectedness rate was 15% for SLI family members and 6% for control family members. The rates of affected family members were slightly higher within the nuclear families alone: 22% in the SLI families and 7% in the control families.

### **Twin Studies**

Twin studies have been an important source of information about heritability of language impairments. Monozygotic twins should have identical genetic material, and dizygotic twins share an average of 50% of their genetic material, providing an excellent way to examine heritability of language impairment. Monozygotic twins should be concordant for inherited traits approximately twice as often as dizygotic twins. Lewis and Thompson (1992) looked at 57 sets of same-sex twins (32 monozygotic and 25 dizygotic) and found a concordance rate of .86 for monozygotic twins and .48 for dizygotic twins. This study did not look directly at SLI but instead examined a variety of speech, language, and learning difficulties. The impairment rates for other family members ranged from 23% (monozygotic) to 27% (dizygotic), which is consistent with affected rates in other family incidence studies. Bishop, North, and Donlan (1995) reported a concordance rate of .70 for monozygotic and .46 for dizygotic twins, with the effect being stronger for males. Tomblin and Buckwalter (1998) found the highest rate of concordance (.96) for poor language in monozygotic twins and a .69 concordance rate in dizygotic twins.

### **Family History vs. Direct Testing**

The question remains: Would the predicted rate of affected family members change if family members were directly tested rather than relying on family-history questionnaire information? To address this question, Plante, Shenkman, and Clarke (1996) used the test battery developed by Tomblin, Freese, and Records (1992) to directly test the parents of children with SLI and a group of adult controls. When test results were compared with family case-history reports/questionnaire data, test results consistently identified more language-impaired

adults among both the control parents and the parents of children with SLI. Tomblin and Buckwalter (1994) also used the same test battery to assess the parents as well as siblings of SLI probands. Twenty-one percent (21%) of all family members scored at levels consistent with a diagnosis of SLI. Fathers and brothers (40% and 24%, respectively) were more likely to be diagnosed as SLI than mothers and sisters (15% and 6%, respectively).

To date, no study has involved the administration and comparison of the same battery of tests and the same questionnaire form for all probands, matched controls, and their first-degree relatives. Such a study is necessary to determine the relationship between questionnaire data and current performance-based testing in identifying language impairment rates for families with or without an SLI proband. Because accurate phenotypic classification of each family member is necessary for gene-linkage studies, it is important to examine differences when LI classification is based on current testing as compared to a history of disorder.

The purpose of the current study was to validate previous findings of familial aggregation for developmental language disorders using a case-control family study design in which the current language-related abilities of all biological primary relatives (mother, father, siblings) of SLI (Specific Language Impaired) probands and matched controls are evaluated. Specifically, the goal of this study was to determine whether there is increased familial aggregation for language impairments in the families of SLI probands as compared to controls. A second goal was to compare directly the results of family aggregation analyses based on questionnaire data with those derived from current test data, for both group data as well as individual cases.

## Method

### Subjects

Two groups of subjects, a proband group consisting of students receiving school speech/language services for specific language impairment (SLI) and a comparable, non-impaired control group, and their families were ascertained from local schools. Subjects were recruited through the procedures described below as part of a larger family genetic study. SLI and age-matched control probands [4 to 14 years old; mean = 7.6;  $t(46) = .50$ ,  $p = .620$ ] participated in this case-controlled portion of the study. The SLI group consisted of 22 subjects who met the family study inclusion criteria and were identified as SLI.<sup>2</sup> The control group consisted of 26 subjects who met the family-study inclusion criterion but did not meet the criteria for SLI. The socioeconomic status (SES) of the families within the two groups was estimated using the Hollingshead (1975) scale. When SES scores

for the two groups were compared, no significant difference was found [ $t(39) = -.80$ ,  $p = .431$ ] (means: controls = 53.7, probands = 55.8).

### Family Ascertainment Procedure and General Study Design

To ascertain SLI probands, a recruitment letter was sent home to the parents of every elementary or middle school child receiving speech and language services in the participating school system. Control probands were ascertained in a similar manner. Letters were sent home to the parents of every child in one of the elementary schools in each of the school districts involved. For both groups, the letter was nonspecific as to the reason for the study; it emphasized only that we would like to study the learning patterns of the child and to look at similar skills in their first-degree relatives.

When a potential control family responded, it was asked only if the child had any history of learning problems. If the child did not have a learning problem, then the family was accepted as a control family and was asked no other questions about any other family members at that time. Only families with both biological parents and at least two of their offspring (the proband and at least one sibling) available for testing were included.

An extensive family-history questionnaire was completed by each family at the time of induction into the study. The questionnaire solicited pertinent personal, medical, and educational information about all family members. In addition, for each group the proband as well as each of his or her first-degree relatives was assessed using a single age-appropriate battery of tests. The testing battery consisted of both standardized and experimental measures of hearing, general cognition, auditory rate processing, language, and reading. Only the data pertaining to rates of speech and language impairment are reported here. Siblings under the age of 4 years were not tested until after they reached age 4. Infants born into these families over the years of this study were initially entered into another ongoing research project aimed at determining the early predictors of language-learning deficits (Benasich & Tallal, 1998; Spitz et al., 1997). These infants were followed longitudinally and were assessed for the current study once they reached age 4.

<sup>2</sup> Twenty-five SLI probands and their families were inducted into the study and completed the test battery. Two of these families were eliminated from data analysis when scoring errors on the language test battery were found and corrected and showed that two SLI probands failed to reach the study criterion of 85 or below (one proband scored 86 and one 87). A third family was eliminated when a scoring error was corrected on the performance IQ test, revealing that the proband failed to reach the IQ criteria of 80 or above. This LI proband scored 79 on PIQ.

## SLI Proband Inclusion Criteria

In order to be included as an SLI proband, subjects were required to meet all of the following criteria:

1. Both biological parents and at least two children (proband and at least one sibling) available and willing to take the entire test battery
2. Performance IQ (PIQ) of 80 or better on the age-appropriate Wechsler Intelligence Performance Scale (Wechsler, 1974, 1981, 1989)
3. Normal hearing acuity (20 dB at 1000, 2000, 4000 Hz.; 30 dB at 500 Hz)
4. No motor handicaps or oral structural impairment affecting speech and nonspeech movements of the articulators as determined by the consensus of two licensed speech-language pathologists using the Oral Speech Mechanism Screening Examination (St. Louis & Ruscello, 1987)
5. No co-morbid diagnosis of autism, emotional difficulties, or neurological disorders
6. Composite language score on an age-appropriate version of the TOLD/TOAL (TOLD-P2/Newcomer & Hammill, 1988; TOLD-I2/Hammill & Newcomer, 1988; TOAL/Hammill, Brown, Larsen, & Wiederhold, 1987) less than or equal to 85, or the average of scores on the TOLD/TOAL and the age-appropriate version of the Token Test<sup>3</sup> (Token Test for Children/DiSimoni, 1978; Token Test for Adults/Tomblin, Freese, & Records, 1992 originally developed by DeRenzi & Vignolo, 1962, and DeRenzi & Faglioni, 1978) less than or equal to 85.

<sup>3</sup> The Token Test for Children (DiSimoni, 1978), given to all probands and siblings younger than 13 years old, has a mean of 500 and a standard deviation of 5. Raw scores were converted to *z* scores and then converted on a scale of 100 with a standard deviation of 15, translating the Token Test for Children to the same standardized scale as the TOLD/TOAL. An adapted version of the Token Test for Adults (Tomblin, Freese, & Records, 1992; originally developed by DeRenzi & Vignolo, 1962, and DeRenzi & Faglioni, 1978) was administered to family members over the age of 13. To date, this version has not been standardized. Therefore, raw scores from the current sample were also converted to the same standardized scale as the Token Test for Children, as follows: Tomblin, Freese, and Records (1992) administered the adapted version of the Token Test for Adults to 70 adults, 35 of whom had been diagnosed as LI based on other criteria and 35 non-LI controls. Within each of these two groups of adults, scores on the Token Test expressed as percent correct, were normally distributed. When the distribution of scores for the two groups were compared it was found that one standard deviation below the mean of the control group corresponded to one standard deviation above the mean for the language-impaired group, indicating that this test is clearly able to distinguish between language-impaired and normal adults. This information was used to translate raw scores on the Adult Token Test from the current study, expressed as percent correct, into standard scores with a mean of 85 and a standard deviation of 15. *Z* scores were used to again translate these standardized scores to the same standardized scale used for the Token Test for Children ( $M = 100$ ,  $SD = 15$ ).

## Control Proband Inclusion Criteria

Control probands were matched by age and required to meet the criteria noted in items 1–5 above. Only the language criteria (item 6 above) differed between groups. Control probands were given the same language test battery as the SLI probands but had to score within the normal range (>90 on the language test battery) and have a normal history of speech and language development.

## Family-History Questionnaire

The questionnaire was constructed to obtain information concerning the age, sex, and relationship of each family member to the proband. Included were items concerning prenatal development, early development, socioeconomic background, and medical history of all family members (mother, father, and siblings). This paper reports only the data pertaining to history of speech/language problems. For classification, information was obtained concerning the presence of language disorders, speech disorders, and speech/language therapy received. The majority of questions in the questionnaire were presented as yes/no questions, followed by an open-ended question to give parents the opportunity to explain their response.

In most cases, the parents completed the questionnaire during the first testing visit, giving a member of the research team the opportunity to go through the responses to ensure that all questions had been answered and to ask for clarification if necessary. In the majority of cases, fathers completed the questionnaire about themselves, and mothers completed the questionnaire for the rest of the family, but there were a few fathers who responded for themselves and the rest of the family. Furthermore, if a parent had difficulty reading or interpreting a question, assistance was available.

## Classification of Family Members as LI Based on Family-History Questionnaire

For the present analyses, parents were considered affected if they reported (a) a history of any language problems, or (b) having received help or therapy for speech/articulation. Siblings were considered affected if their parents reported that they had current or past problems in language and/or speech development. Because there might have been confusion concerning what constituted a language versus a speech problem, the questionnaire used two methods: (a) an open-ended question asking parents to describe the type of speech/language impairment or the kind of therapy received, and (b) a checklist of speech, language, and learning problems. The same procedure was followed across all informants. Only children who had language problems (with

or without associated speech and learning problems) were classified as LI.

## Classification of Family Members as LI Based on Current Testing

Parents and siblings of both the SLI and control probands were classified as LI<sup>4</sup> if they met the same language criteria (item 6 only) used to classify the probands as impaired. That is, parents and siblings were classified as LI if their composite language scores on the age-appropriate version of the TOLD/TOAL was less than or equal to 85 or their average score on the TOLD/TOAL and the age-appropriate version of the Token Test was less than or equal to 85.

## Results

### Probands

Descriptive statistics for age, sex, measures of language development, and Performance IQ (PIQ) are summarized in Table 1. The control group included 14 boys and 12 girls, and the SLI group included 16 boys and 6 girls, reflecting the higher incidence of males who receive language services in school (Ludlow & Cooper, 1983; Tallal et al., 1989). As expected, the SLI probands scored significantly lower on the Test of Language Development [ $t(46) = 13.4, p < .001$ ] and the Token Test for Children [ $t(28.4) = 5.6, p < .001$ ] than probands in the control group. SLI probands also had significantly lower PIQ scores than control probands [ $t(46) = 3.7, p < .001$ ].

### Siblings

Descriptive statistics for siblings' age, sex, measures of language development, and Performance IQ are also summarized in Table 1. There were 33 siblings in the control group (15 boys and 18 girls) and 45 siblings in

the SLI group (25 boys and 20 girls). At the time probands were inducted into this study, siblings ranged in age from 1 1/2 to 17 years old, with the average age of the siblings in the control group being significantly higher than siblings in the SLI group [ $t(76) = 2.3, p = .024$ ].

Siblings of the SLI probands scored significantly lower on the Test of Language Development [ $t(75) = 4.9, p < .001$ ] and Token Test for Children [ $t(61.1) = 3.5, p < .001$ ] than siblings in the control group. Consistent with probands' PIQ scores, siblings of the SLI probands showed significantly lower PIQ scores than the siblings in the control group [ $t(73) = 3.5, p < .001$ ].

### Parents

Descriptive statistics for parents' age, SES, PIQ, sex, and measures of language development are also summarized in Table 1. Parents ranged in age from 31 to 52 years old, with parents in the control group significantly older than the parents in the SLI group [ $t(94) = 2.9, p = .005$ ]. Parents of the SLI probands scored significantly lower on the Test of Language Development ( $Z = -2.8, p = .003$ ) and the Adult Token Test ( $Z = -2.5, p = .006$ ) than parents in the control group. Performance IQ scores for the parents in the SLI proband group were also significantly lower than the PIQ scores for the parents in the control group [ $t(94) = 2.7, p = .007$ ].

To account for the possibility that group differences on measures of language development could be attributed to differences in Performance IQ between groups, the relationships between PIQ and the measures of language development (age appropriate versions of the Token and TOLD/TOAL) were examined separately for probands, siblings, and parents. More correlations were significant for parents than for probands and siblings (see Table 2). In cases where language measures were significantly correlated with PIQ, analyses were conducted to determine the necessity of including PIQ as a covariate. In all such cases, PIQ was found not to be a significant covariate.

## Family Aggregation Based on Current Testing

Compared to probands in the control group, the SLI probands were significantly more likely to have a positive family history, defined as having at least one first-degree relative identified by current testing as SLI (59.1% vs. 19.2%,  $\chi^2_1 = 8.1, p = .005$ ). The overall impairment rate of family members (excluding the probands) was significantly higher for the proband group than the control group (31.0% vs. 7.1%,  $\chi^2_1 = 15.9, p < .001$ ).

<sup>4</sup> Longitudinal studies of individuals with SLI have shown that IQ scores (including Performance IQ) can decline significantly with age in the face of continued language deficits (Tallal, Allard, Miller, & Curtiss, 1997). That is, the same individual who may meet research or clinical criteria of SLI, with performance IQ within the normal range early in life, may continue to have a language impairment but show a significant drop in performance IQ later in life and thus no longer meet the criteria as SLI. For this reason, in the current study we selected child probands using criteria for SLI that included both a significant language impairment as well as a performance IQ within the normal range. However, relatives were classified as LI if they demonstrated language skills one or more standard deviations below the mean, regardless of their performance IQ score. Our test data showed that all probands and all except 3 relatives (1 sibling, 2 parents) who scored one or more standard deviations below the mean on language testing, also had a performance IQ of 80 or above. Nonetheless, to be inclusive of all relatives' scores, throughout this paper we refer to affected relatives as language impaired (LI), whereas we refer to the probands who met all study criteria as SLI.

**Table 1.** Demographic data and language development scores for control and SLI probands.

	Control		SLI		<i>p</i> value
	<i>M</i>	<i>SD</i>	<i>M</i>	<i>SD</i>	
Number of probands	26		22		
Male/Female	14 / 12		16 / 6		
Number of siblings	33		45		
<b>Probands</b>					
Age at testing	7.7	(1.6)	7.4	(2.2)	.620
Age-Appropriate TOLD (P2 or I2)	113.0	(8.7)	80.7	(7.8)	<.001
Token Test for Children	104.5	(10.4)	77.6	(20.2)	<.001
Performance IQ	111.6	(14.1)	97.4	(12.2)	<.001
<b>Siblings</b>					
Age at testing	10.4	(3.0)	8.7	(3.5)	.024
Age-Appropriate TOLD (P2 or I2)	111.5	(14.5)	93.3	(17.1)	<.001
Token Test for Children	106.7	(10.7)	93.3	(20.7)	<.001
Performance IQ	113.1	(14.9)	101.1	(14.7)	<.001
<b>Parents</b>					
Age at testing	42.2	(4.0)	39.9	(3.9)	.005
TOAL	116.5		108.0		.003
Token Test for Adults	111.4		101.7		.006
Performance IQ	109.1	(12.9)	101.5	(14.3)	.007
Family SES	53.7	(6.4)	55.8	(10.4)	.431

Notes. Because within each group, scores on the Token Test for Children were normally distributed with unequal variance, *t* tests were approximated, adjusting the degrees of freedom, rather than performing nonparametric tests. Within each group, scores on the TOAL and Adult Token were non-normally distributed; therefore large-sample Wilcoxon Rank Sum Tests using *Z* score approximations were used to compare distributions. Medians are reported for measures of central tendency.

Rates of maternal impairment, paternal impairment, and sibling impairment for the entire sample were examined to determine if a larger proportion of all family members identified as LI were from families of the SLI probands. Chi-square tests of independence found that a majority of those family members identified as LI were from families of the LI probands. Overall, 18.8% of mothers were identified as language impaired, more of which were mothers of SLI probands (12.5% vs. 6.3%,

$\chi^2_1 = 1.9, p = .164$ ); 17.0% of fathers were identified as language impaired, the majority of which were fathers of SLI probands (14.9% vs. 2.1%,  $\chi^2_1 = 7.2, p = .008$ ); 20.8% of siblings were identified as language impaired, the majority of which were siblings of SLI probands (18.2% vs. 2.6%,  $\chi^2_1 = 7.6, p = .006$ ).

**Table 2.** Correlations between performance IQ and measures of language.

	Probands	Siblings	Parents
<b>SLI group</b>			
TOLD-P2,-I2, TOAL	.09	.57**	.67**
Token Test	.30	.38*	.69**
<b>Control group</b>			
TOLD-P2,-I2, TOAL	.44*	.33*	.51**
Token Test	.02	.43*	.43**

\**p* < .05

\*\**p* < .01

### Family Aggregation Based on Current Testing Compared to Family-History Questionnaires

Table 3 is a summary of family impairment by group and sex based on current testing compared to family-history questionnaire data. The number of mothers, fathers, brothers, and sisters of probands who were classified as LI is shown as a proportion of the total number of mothers, fathers, sisters, and brothers per group, rather than relative to the entire sample. Rates of impairment are shown as percentages. Totals (excluding probands) are given per group for parents, siblings, and all family members. As can be seen in Table 3, overall family impairment rates based on current testing were not significantly different from rates derived from

**Table 3.** Proportion of first-degree relatives (excluding probands) classified as LI, by group: current testing compared to family history questionnaire.

	# of probands	Group													
		Mothers		Fathers		Parents		Brothers		Sisters		All siblings		All family members	
		N	%	N	%	N	%	N	%	N	%	N	%	N	%
<b>Control</b>															
Testing	26	3/26	11.5	1/26	3.9	4/52	7.7	1/15	7.1	1/18	5.9	2/33	6.1	6/85	7.1
Questionnaire	26	2/26	7.7	3/26	11.5	5/52	9.6	2/15	13.3	0/18	0.0	2/33	6.1	7/85	8.2
<b>SLI</b>															
Testing	22	6/22	27.3	7/22	31.8	13/44	29.5	11/25	44.0	3/20	15.0	14/45	31.1	27/89	30.3
Questionnaire <sup>a</sup>	19	3/19	15.8	2/19	10.5	5/38	13.2	9/19	47.4	10/18	55.6	19/37	51.4	24/75	32.0

<sup>a</sup> Family history questionnaires were not completed by 3 proband families.

family-history questionnaire data for either the SLI or control group. Siblings in the SLI group have a significantly higher impairment rate than siblings in the control group based on current testing (31.1% vs. 6.1%,  $\chi^2_1 = 7.6, p = .006$ ) and family-history questionnaires (51.4% vs. 6.1%,  $\chi^2_1 = 21.2, p < .001$ ). Parents in the SLI group have a significantly higher impairment rate than parents in the control group based on current testing (29.5% vs. 7.7%,  $\chi^2_1 = 8.1, p = .004$ ). However, the questionnaire data showed that although the impairment rate of parents in the SLI group was higher than for parents in the control group, this difference did not reach statistical significance (13.2% vs. 9.6%,  $\chi^2_1 = .4, p = .54$ ).

When LI diagnoses derived from current testing were compared with LI diagnoses derived from family-history questionnaires there was 74% agreement. Of the 26% disagreement, family-history questionnaire data indicated a diagnosis of LI that was not confirmed by current testing more often than the reverse pattern, wherein current testing indicated an impairment that was not reported on the family-history questionnaire (16% false positives and 10% false negatives). Case-by-case comparisons indicated that of the 74% agreement, the majority (70%) was attributable to agreement that the family member did not have a language impairment (true negative), whereas only 4% was attributable to agreement that the family member did have a language impairment (true positive). A kappa statistic, which measures agreement based on true positives and true negatives, was found to be extremely low (.08).

### Offspring Impairment Related to Parental Impairment

Impairment rates were further examined by analyzing the proportion of children in each family identified as LI (based on current testing) relative to the

number of their parents who were also identified as LI based on current testing. The number of parents in each group who met the study criteria as LI is shown in Table 4. The distribution of offspring impairment (excluding probands) is shown in terms of the number of LI parents in the family (0 = neither, 1 = either, or 2 = both). Language-impairment rates for male offspring, female offspring, and total offspring are shown as percentages. In SLI proband families, LI occurred in 13.0% of offspring with neither parent affected, 40% of offspring with one parent affected, and 71.4% of offspring in families in which both parents were language-impaired.

The correlation between the number of parents identified as impaired and the proportion of their offspring identified as impaired (excluding probands) across both groups was found to be significant ( $r = .37, p = .010$ ), indicating that offspring impairment increases significantly with increased parental impairment.

## Discussion

### Family Aggregation Rates

This study is the first case-control family aggregation study in which all SLI probands, their matched controls, and all first-degree relatives received current neuropsychological testing in addition to providing family-history data. Results from the current case-controlled study agree with results from previous case-control studies (those based on questionnaire data alone) that language impairment aggregates in families. Parents and siblings of children with SLI are more likely to have a language impairment than are parents and siblings of matched control children with age-appropriate language abilities. Current testing with age-appropriate standardized language measures showed that over half of the SLI probands in this study had a positive family

**Table 4.** Distribution of LI offspring (excluding probands) by number of LI parents for families with SLI probands as compared to control families.

	Number of LI parents	Families N	Offspring N	Male offspring		Female offspring		All offspring	
				N	%	N	%	N	%
Controls	0	23	28	1/15	6.7	1/13	7.7	2/28	7.1
	1	2	4	0/0	0.0	0/4	0.0	0/4	0.0
	2	1	1	0/0	0.0	0/1	0.0	0/1	0.0
SLI	0	12	23	2/12	16.7	1/11	9.1	3/23	13.0
	1	7	15	5/8	62.5	1/7	14.3	6/15	40.0
	2	3	7	4/5	80.0	1/2	50.0	5/7	71.4

history of LI. That is, at least one of their nuclear family members (parents or siblings) also met the study criteria as LI. Results also showed a significant correlation between the number of parents in each family affected with LI (none, one, or both) and the number of offspring affected with LI. As the number of parents affected with LI increased, so did the proportion of their offspring affected with LI. The degree to which LI incidence in parents increases their risk of having offspring with LI should be interpreted cautiously, however, in light of the small number of families in this study with one or more affected parents.

### Power from Segregation Models

Familial aggregation data and specific patterns of affection within pedigrees can reveal some information about the underlying genetic models. However, most of the information that can be inferred from such data is exclusionary in nature. For example, the male-to-male transmission observed in our sample is inconsistent with an X-linked mode of inheritance. Furthermore, several probands do not have an affected parent, which eliminates a fully penetrant autosomal dominant mode of inheritance. Affection rates of around 30% in a given family may suggest an autosomal recessive model with high penetrance, an autosomal dominant model with reduced penetrance, or an oligogenetic model (a few genes with moderate effect). Previously, there has been one segregation analysis conducted on 45 pedigrees ascertained for low language abilities or articulation difficulty (stuttering excluded) in probands with nonverbal IQ in the normal range and absence of neurological impairments (Lewis et al., 1993). This segregation analysis failed to distinguish between major locus inheritance and oligogenetic models, which is consistent with the present study. The present sample is smaller than that of Lewis et al. (1993) and would not have sufficient power to distinguish between different modes of transmission by segregation analysis.

### Group Data vs. Individual Data: Issues for Genetic Studies of Developmental Disabilities

The overall rates of language impairment in the current study based on actual testing (30% for SLI families, 7% for control families) and family-history questionnaire data (32% for SLI families, 8% for control families) are relatively consistent with each other as well as rates that have been reported in the literature using either questionnaire data or current test data alone. Nonetheless, there was surprisingly low agreement between questionnaire data and current test classification as LI when family members were evaluated individually on a case-by-case basis. Questionnaire data based on a history of language problems identified 16% of family members as language impaired who scored within normal limits on current language tests. Conversely, 10% of cases who met criteria as LI based on current tests failed to report any history of speech or language problems. Thus, current testing and family-history questionnaire data resulted in approximately equal rates of family impairment. However, there is poor agreement between these methods when it comes to classifying individual cases as language impaired.

It is important to keep in mind that the questionnaire data are based on a history of a language impairment at any point in the individual's life, whereas the test data are based solely on a current demonstration of language impairment. Previous longitudinal studies following children with a specific language impairment throughout their early development have demonstrated that although many individuals continue to show language deficits, others improve into the normal range over time (Bishop & Adams, 1990). This normalization of language functions may contribute to differences observed between questionnaire and test data for individual subjects. These findings highlight the difficulties of making accurate phenotypic classifications of older family

members participating in genetic studies focused on linking genes for developmental disabilities such as language impairment.

Failure to show agreement between data pertaining to a history of speech or language problems and current language test data should not be interpreted as suggestive that case history data are necessarily unreliable. Rather, both of these types of data may be necessary to correctly classify phenotype in family genetic studies focusing on developmental disabilities. Developmental language disorders may normalize over time in some individuals, whereas they may change in form in others and present as reading, spelling, and/or perceptual problems later in life. In this article we report data pertaining solely to family aggregation rates for oral language impairment.

### **Sex Differences**

Sex differences based on questionnaire data (the finding that fathers and brothers are more likely to report a history of LI than are mothers and sisters) were reported in several previous family aggregation studies (Neils & Aram, 1986; Rice et al., 1998; Tallal et al., 1989). However, sex differences in the current study are not completely consistent with those found in previous studies. Specifically, in the current study, direct testing showed no significant differences between rates of impairment in mothers and fathers. However, there were significant differences in the impairment rates of siblings, with rates for males higher than for females. A similar pattern was observed when impairment was based on family-history questionnaire data.

In general, impairment rates of male siblings are approximately the same when direct testing data is compared with family-history questionnaire data. In contrast, impairment rates for female siblings were significantly lower based on direct testing as compared to parent report. This suggests that parents may be identifying more subtle language problems in their daughters, problems that did not reach the study criterion for language impairment based on direct testing. An exception to this was found in families where both parents are language impaired. In these families, impairment rates for female siblings based on direct testing are somewhat higher than reported on family-history questionnaires. This lack of consistency across methods and studies may reflect a reduction in statistical power when already relatively small sample sizes are divided to assess gender differences.

There has been considerable interest in sex differences in language impairments. Although studies based on clinically referred samples have consistently reported significantly more boys than girls affected, recent

epidemiologically based studies have found equal numbers of girls and boys reaching criteria as SLI (Rice, 1996; Tomblin et al., 1992). It has been suggested that sex differences reported in previous studies resulted from ascertainment bias, with the tendency for more boys to be referred for clinical services (Shaywitz, Shaywitz, Fletcher, & Escobar, 1990). Although ascertainment bias may explain the sex differences found in the SLI probands in the current study, in that they were ascertained from children receiving language services in public schools, this could not explain the sex difference (3:1 boys to girls found to be LI) found in the siblings of the SLI probands in the current study. Similarly, it has been suggested that parental bias may occur in reporting more language problems in the brothers than sisters of SLI probands. However, in the current study a greater sex-ratio difference was found based on current testing than was reported by parents on questionnaires for the same children. These data demonstrate that when SLI aggregates in families, there is a higher rate of impairment in males than in females, at least as determined by childhood test scores.

### **Ascertainment Issues**

When conducting family aggregation studies of the design employed in this case-controlled study, very strict ascertainment procedures are required to assure that very similar methods are employed in recruiting both the SLI and control probands. It is important to assure that the families ascertained are similar except that the probands differ with respect to LI status. As such, all elementary and middle school children who were receiving specialized language services in our target population were given the opportunity to participate in this family study, provided that they matched the family-size profile (both parents and at least one sibling willing to be tested) as well as our research criteria for SLI. Similarly, all case controls matched by age to the SLI probands who met the study criteria were included. This strict ascertainment procedure dictated that no other inclusionary or exclusionary criteria (such as sex, birth order, race, SES, parental age, IQ, or the many other variables that might affect language development) could be controlled, as they might introduce ascertainment bias. As such, the influence of these or other group differences on the current data set cannot be determined within the limitation of the current study. A similar caveat applies to the age range (4–14 years) of the probands in this study. Given this relatively small sample size (48 families with 222 individuals) it must be kept in mind that demographic variables, differences in exact profile of language disorder, and the range of ages of probands may affect the exact family aggregation rates found.

## Conclusions

The current study shows that the rates of family members affected with LI are significantly higher for children with SLI than for matched control children. Furthermore, language impairment rates for first-degree relatives of SLI probands, based on current testing, are relatively consistent with results reported previously from case-history questionnaire data. However, direct case-by-case comparison of individual testing versus questionnaire data showed a high proportion of disagreement in classification of LI using both methods with the same subjects. Therefore, for studies in which individual diagnostic classification of phenotype is essential, such as for gene linkage studies or for studies focusing on phenotypic subgroups, direct testing of each family member provides important information of current status that is not always consistent with case-history information derived from questionnaire data.

The case-controlled family aggregation data reported here derive from a larger family genetic study of SLI. Future papers will focus on other aspects of this large data set: rates of co-morbidity for related conditions such as reading, spelling, learning disorders and auditory rate processing disorders; differences in specific linguistic profiles across family members; correlation between family members for specific language and learning impairment; heritability estimates; and gene linkage analyses.

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