Family Histories of Children With SLI Who Show Extended Optional Infinitives

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Previous family history studies have demonstrated that there are elevated rates of language and language-related impairments in families identified through probands with language impairments. This study examines family histories of children with specific language impairment (SLI) known to have particular grammatical limitations in a core feature of grammatical acquisition, a stage known as Extended Optional Infinitives (EOI). Family affectedness rates are reported for 31 families identified through preschool probands with this clearly defined language impairment and 67 control families, identified through nonaffected preschool children developmentally similar to the probands.

It was found that significantly more speech and language difficulties, as well as language-related difficulties, such as reading, were reported for proband families than control families. The elevated rates were obtained for nuclear family members and extended family members as well. Fathers of probands were more often reported as having difficulties (29% for speech/language impairments) than mothers of probands (7%), but there was no difference between brothers (26%) and sisters (29%). No differences were evident between proband families based on proband gender. The findings are relevant for theoretical models of sources of unexplained variations in grammatical competence in young children. In addition, the findings contribute new information about expected rates of affectedness, means of identification of affected family members, and comorbidity of symptomatology.

KEY WORDS: specific language impairment, genetics of language, family history, morphosyntactic deficits, extended optional infinitives

For more than 30 years there has been interest in familial patterns of language impairments, as a necessary, but not definitive, source of evidence for establishing inherited contributions to language aptitude (cf. Lenneberg, 1964). In the context of contemporary advances in human genetics, this long-standing question has new momentum, with renewed and enlivened relevance. It is now entirely feasible to entertain the possibility that language impairments are likely to be among the growing list of inherited conditions (cf. M. L. Rice, 1996).

At the same time, there are some daunting challenges to be overcome on the way to achieving definitive evidence. Of paramount importance is the definition of the phenotype (i.e., the behavioral manifestation of the effect of the underlying genetic influence). Briefly put, the better the specification of the phenotype, the less likely are errors in detection of affected individuals and the more accurate are methods of determining genetic influence. The point is that it is very important to
know as precisely as possible what is meant by “language impairment.” This study contributes to what is known about the familiality of language impairment by investigating the families of children with SLI who are known to have particular grammatical limitations in a core feature of language acquisition, a stage known as Extended Optional Infinitive (EOI; Rice & Wexler, 1986a, 1986b; Rice, Wexler, & Cleave, 1995).

Family studies begin with a proband, who is the affected individual whose family is studied. Among contemporary advances in empirical studies of families is the differentiation of studies that begin with probands who have significant speech disorders from those that begin with probands who have been identified as having language impairments. The distinction between these two phenotypes is still somewhat fuzzy, but becoming more clear. The best current estimate is that about 26% of the kindergarten children who score low on language tests also have significant speech problems (Tomblin, 1996). Conversely, in a current study, Lewis (1996) reports that in a sample of probands selected for preschool “moderate-to-severe” phonological disorders, 50% of the children showed clinical levels of performance on a battery of language tests. In this study, we focus on child probands defined as having language impairment.

Because our interest here is on familiality and available space is limited, we do not address the full range of evidence relevant for an examination of possible genetic effects on language impairment. Much of the evidence is summarized, and critiqued, in chapters that appear in M. L. Rice (1996). In particular, we wish to note that twin studies (cf. Tomblin, 1996; and Bishop, North, & Donlan, 1994, 1996) are very important new sources of evidence that fall outside the scope of this review.

Evidence of Familiality of Language Impairment

Early studies, based on case history data (Arnold, 1961; Eustis, 1947; Lenneberg, 1984), indicated that general language disabilities seemed to run in families. More recently, a number of studies have appeared with children defined as having language impairment, in which elevated rates of affectedness in family members are reported (Ellis Weismer, Murray-Branch, & Miller, 1994; Lahey & Edwards, 1995; Niels & Aram, 1986; Paul, 1989; Rescorla & Schwartz, 1990; Tallal, Ross, & Curtiss, 1989; Tomblin, 1989; Van Der Laly & Stollwerck, 1996). During this same period, positive family histories were reported for phonological disorders (Lewis, 1990, 1996), and for stuttering (Ambrose, Yairi, & Cox, 1993; Kidd, Heimburg, & Records, 1981; Poulos & Webster, 1991). One study (Whitehurst et al., 1991), on the other hand, did not find a strong familial aggregation of language delay for young probands, ages 24 to 38 months, who showed a language deficit in expressive language only. Ellis Weismer et al. (1994, p. 864) suggest that the unexpected null finding is attributable to methodological differences across studies.

Four of the studies have important similarities to the study reported here (i.e., Lahey & Edwards, 1995; Niels & Aram, 1986; Tallal et al., 1989; Tomblin, 1989). In these studies, the age range of the probands was 4 to 9 years. The children were identified as having specific language impairment (SLI), either on the basis of clinical judgment (Lahey & Edwards, 1995; Tomblin, 1989) or clinical judgment plus test performance below normative levels (Niels & Aram, 1986), or discrepancy between language age and performance mental age scores (Tallal et al., 1989). The proband groups included children with mixed profiles of language impairments (receptive + expressive) as well as children with expressive impairment only. In these studies, familiality was measured by asking a family informant to report who in the family had language impairments (and, usually, impairments of speech, stuttering, and reading or school achievement, as well). The major finding, replicated across studies, is that the rate of affected nuclear family members (i.e., mother/father/siblings) is elevated in families with an affected proband as compared to control families. The percentages run between 20%-40%, depending on the details of how the impairments of the family members were defined. With the exception of Tomblin (1989), these studies relied on a relatively broad definition of affectedness, described as “language-related disorder,” which could include any kind of academic achievement problem (cf. Tallal et al., 1989). In these studies, the rate of affected family members in the control families was around 5%, which corresponds closely to the best current estimate of rate of language impairments in the general population (7% for kindergarten children, reported by Tomblin, 1996).

A crucial limitation of these studies is that little is known about the grammatical competencies of the affected children. The children are identified by means of conventional clinical methods, which are heavily reliant on standardized omnibus language tests. In the tests used, it is simply not possible to interpret low levels of test performance in terms of known grammatical structures. Thus, the grammatical competencies of the probands are not known. At the same time, children with SLI (as a group) are known to have difficulty with several aspects of grammar (Bishop, 1992; Leonard, 1998; M. L. Rice, 1996; Watkins & Rice, 1984). Therefore, we do not know whether the higher familial concentration reported in these studies applies to the children who demonstrate the known grammatical problems.

A further and highly relevant complication is that
language impairments, as noted above, are to some degree conflated with significant speech impairments. In the Tallal et al. (1989) sample, for example, more than 60% of the probands had significant articulation deficits. The significance of this observation is that it is not possible to know if what is moving through families is a limited capacity for motor speech production or phonological representations (i.e., if the findings are more in keeping with the speech proband studies) or if the familial component is associated with grammatical impairments of young children. We now know that 75% of kindergarten children with language impairments, in an epidemiologically ascertained sample (Tomblin, 1996), do not show significant speech problems. Because speech problems are more readily ascertained, it is probable that children with only language impairment may be underrepresented in earlier studies that drew from clinical caseloads where speech problems are more heavily represented. The import is that an inherited grammatical deficit could be masked by the imprecision of conventional language tests and the comorbidity of highly salient speech disorders.

In contrast to the clinical sampling approach of the previous studies, Van Der Lely & Stollwerck (1996) selected a group of children with SLI who showed particular grammatical impairments over a protracted period of time, a group they called “grammatical specific language-impaired children.” Their sample of 12 children, aged 9;3 to 12;10, was screened for speech impairments, and was known to omit third person singular -s from lexical verbs and make tense errors involving dropping of BE forms or past tense -ed. They also performed poorly on a task assessing passive sentences. For the 9 children for whom data are available, 39% of the nuclear family members were reported as affected, as compared to 9% of a control group of 49 children. This sample of children was drawn from a more broadly defined sample of children with SLI studied by Haynes and Naidoo (1991) who reported that 41% of the children had positive histories. Van Der Lely and Stollwerck (1996) report that the rate of affectedness for the grammatical SLI group is higher than that of the globally defined group from which the subsample is drawn.

Considered as a collection of findings, the available evidence for familiarity of SLI points strongly in the direction of a higher rate of risk in affected children, but the extent to which this is tied to particular symptoms, such as grammatical limitations, is not well documented, particularly in a group of children drawn from a general clinical sampling. Another important limitation is that the available evidence is drawn from nuclear family members (i.e., mothers, fathers, and siblings). Full family information, from extended family members as well (i.e., aunts, uncles, and cousins), would offer more complete information about possible transmission mechanisms.

A Grammatical Marker of SLI: Evidence of an Extended Optional Infinitive Stage

For some time, it has been reported that children with SLI are likely to show delayed acquisition of verbal morphology. Various explanations for the observed grammatical differences have been proposed and are under active investigation (cf. Leonard, 1998; Leonard, Eyer, Bedore, & Grela, 1997; M. L. Rice, 1996; Watkins & Rice, 1994). Of interest to this study is that a grammatical marker has been found in a group of clinically sampled children with SLI, that is characterized as an EOI stage of development (Rice et al., 1995; Rice & Wexler, 1996a, 1996b). In this account, we extend what is known about morphosyntax in the early period of acquisition in normally developing children to capture important generalizations about grammatical competence in affected children. Wexler (1994) proposed an Optional Infinitive (OI) model of early morphosyntax, in which young children are relatively late in marking grammatical tense in root clauses. This grammar, he argued, is evident in English in the following contexts:

1. Patay walk(s) home.
2. Yesterday Patsy walk(ed) home.
3. She (is) walking.
4. She (is) happy.
5. (Does) she walk every day?

In these examples, the parentheses indicate parts of the surface structures that may not appear in an OI stage (i.e., the child sometimes uses these structures and sometimes [optionally] drops them). It is essential to emphasize that the dropping of surface structures is not the crucial part of the grammatical profile, because it is well known that the OI stage in languages such as German does not show dropping but instead shows optional use of infinitival verbal affixes (cf. Ingram & Thompson, 1996; Poeppel & Wexler, 1993; Verrills & Weissenborn, 1992). What is important is that children in an OI stage optionally project grammatical tense, which may appear as either a bare stem or an inflected form, depending upon a given language’s properties. Among other contributions, this model is the first to highlight that DO in English-speaking children with SLI should also be affected because it serves as a finiteness marker. Furthermore, it predicts that the set of affected forms should cohere as children progress toward an adult grammar.

What is now known about English-speaking children with SLI is that the forms affected by an EOI period are very late in first appearance (Hadley & Rice, 1996), remain at low levels of accuracy (i.e., below that of unaffected language-equivalent control children 2 years younger) throughout the preschool period even though the unaffected children show virtually no variance in their adult-like command of these forms by age 4 years.
(Rice & Wexler, 1996b), and these deficits can persist to age 7 years, well beyond the normative expectations (Rice & Wexler, 1997). The predicted patterns appear for each of the target morphemes in spontaneous samples as well as elicited productions, and in grammaticality judgments (Rice & Wexler, 1997). In short, these symptoms are remarkably coherent across different surface forms, consistent in differentiating affected from control groups, stable over time, and evident in multiple forms of measurement. Furthermore, growth curve analyses show that maternal education does not predict growth of grammatical tense-marking morphemes (illustrated in 1–5 above) for the SLI group, nor for the control children; nonverbal intelligence and receptive vocabulary scores are also nonpredictors (Rice & Wexler, 1997).

Detailed studies show that surface phonological properties of the affected morphemes do not account for the patterns of surface dropping (cf. Cleave & Rice, 1997; Oetting & Rice, 1983; Rice & Oetting, 1993). This conclusion is further supported by the finding that German-speaking children with SLI show a pattern of EOI, which appears in the form of overt affixation on verb stems (Rice, Noll, & Grimm, 1996). Altogether, the evidence points away from surface properties that lead to the dropping of grammatical forms in affected children. Instead, an EOI stage seems to reflect differences in the underlying grammatical representations that control where tense must be marked.

A further defining characteristic of the EOI period is that much of the scaffolding for syntax is present. This is evident in the fact that even though the English-speaking children are likely to drop tense-carrying morphemes from surface structures, they are not likely to drop non-tense-carrying control morphemes, such as regular plurals (e.g., the "s" in "cup"), and they consistently show correct form choice. For example, they are likely to say "he is running" and very unlikely to say "he are running."

The conclusion is that the EOI grammar represents a protracted period of time in which a very selective grammatical deficit is operative (although this is not likely to be the only deficit). In this way, it is a very useful grammatical marker of the condition of SLI. Most of the evidence comes from preschool children with receptive and expressive language deficits, although there is no reason to believe that the EOI marker is necessarily restricted to that clinical subgroup of children with SLI. At this early stage, the pervasiveness of the marker is unknown. On the other hand, because the OI grammar is resolved by almost all nonaffected children during the preschool years, an EOI stage is potentially a clear indicator of affectedness for language impairment of some sort in preschool and elementary-aged English-speaking children. For theoretical reasons, evidence of an EOI stage in children who meet the exclusionary criteria of SLI is especially important, because it suggests that core grammatical deficiencies can appear in children with cognitive, social, and perceptual mechanisms apparently adequate for the support of grammatical development. At the same time, as Rice and Wexler (1996b) explicitly note, evidence in support of an EOI marker is not evidence that an EOI is the only possible clinical marker, nor the only possible one with theoretical ties to genetic contributions (cf. Bishop et al., 1990).

Criteria for a Phenotype of Language Impairment

The full interpretive import of the EOI phenotype for the condition of SLI is that it meets five of the criteria for a desirable phenotype for language impairment posited by M. L. Rice (1996). Those criteria are as follows:

1. **Consistent with Universal Grammar (UG).** A preferred definition of language impairment would have clear linkage to theoretical models of the endstate adult grammar, could be evaluated across different languages, and would be theoretically indexed to the fundamental human capacity for grammar.

2. **Yields to reliable measurement.** For obvious reasons, indices of language impairment to be used in genetic studies should show reliable properties of measurement.

3. **Differentiates affected from unaffected individuals.** Measures of the phenotype should correctly identify true cases (i.e., show sensitivity) and correctly identify true non-cases (i.e., show specificity).

4. **Shows variation where none is expected.** One long-standing challenge to the determination of language impairments is the fact that children show considerable individual variation in the progress from a child grammar to an adult level of competence. This is especially true in the domain of semantics, where vocabulary tests show wide variation from one individual to another. In fact, the adult state of knowledge about vocabulary also shows great individual variation. In contrast, certain properties of the grammar are obligate and show minimal variation from one adult speaker to another. Furthermore, fundamental grammatical properties are known in an adult-like way by unaffected children during the preschool years. In these areas of the grammar, then, there lies the possibility of identifying children who show variation that is not expected because almost all young children know these properties of the grammar.

5. **Relatively resistant to environmental effects.** A preferred language phenotype is one that shows minimal effects from environmental input. Consider
physical attributes, such as eye color. Because environmental factors are minimal for these attributes, it is relatively straightforward to deduce genetic effects. It has long been observed that young children's language acquisition can be enhanced by environmental "enrichment." Recent findings show, however, that certain properties of the grammar do not show this effect. In addition to the evidence that shows that maternal education is a nonpredictor of the growth of grammatical tense-marking (Rice & Wexler, 1997), a longitudinal study of children with perinatal risk (i.e., neonatal intensive care) carried out by Rice, Spitz, and O'Brien (1997), found no correlation between maternal education and welfare status on children's mean length of utterance (MLU) or children's growth of grammatical tense at 4 years of age, although maternal education/welfare status did predict children's Verbal IQ scores on the Wechsler Preschool and Primary Scale of Intelligence—Revised (WPPSI–R; Wechsler, 1989), and vocabulary acquisition (scores on the Peabody Picture Vocabulary Test–Revised [PPVT–R]; Dunn & Dunn, 1981). What these findings suggest is that children who show unexpectedly low levels of MLU may not be expressing a variation attributable to differences in home environment as much as variations attributable to child-intrinsic factors.

Of interest here is an additional criterion identified by M. L. Rice (1996): The phenotype applies across the age span. In order to identify affected individuals in families, measurement must be available across the age span. The challenge is that some phenotypes detectable in young probands tend to disappear with age. Stuttering and phonological deficits are two conditions for which this problem is well known. In these areas, investigators rely on positive history as a phenotype for affected family members (cf. Ambrose et al., 1993; Lewis, 1990, 1996). Grammatical impairments such as EOI that affect core properties of the grammar may be another condition that goes "underground" with age; on the other hand, the symptoms may be more subtle in older individuals but still amenable to detection. At this time, it is an open question. Other investigators have used conventional language testing for older individuals (cf. Schuele & Rice, 1996; Tomblin, Freese, & Records, 1992), or tasks thought to be related to the underlying grammatical problem, such as the nonword repetition task of Bishop et al. (1996).

Aims of the Current Study

Our aims were twofold: First, we set out to investigate family history data collected from families identified through probands with SLI who are also known to demonstrate an EOI grammar. The probands are further specified as having minimal speech impairment, if any, and as having no history of perinatal risk. In this way, the study adds to what is known about the clinical symptomatology of probands with SLI who show familiarity of language impairment. Second, we aimed to include extended family members as well as nuclear family members. Previous studies reported on nuclear family members only. Inclusion of information about the extended family members broadens the available database with regard to familiality of language impairments.

Method

Participants

Participants for the study were drawn from the families of children who participated in a study of the morphosyntax of preschool children with SLI (Rice & Wexler, 1996b). A total of 98 families agreed to participate, 31 proband families and 67 control families. In terms of total family members, there are 555 proband family members, 110 nuclear and 445 extended members. In the control families, there are 1283 members, 197 nuclear and 1086 extended members. As noted in Rice and Wexler (1996b), the sample of children comprises two separate samplings, one for the cross-sectional study reported by Rice et al. (1995), and a second sample recruited for the longitudinal study. As shown by Rice and Wexler (1996b), the two samplings do not differ with regard to patterns of language impairment. In this study, 19 of the 31 probands are from the longitudinal study and 12 are from the earlier cross-sectional study. In her thesis study, K. J. Rice (1996) examined whether the two samplings differed on the variables of interest here. No differences between the two samples appeared, and in fact the patterns are strikingly similar. For this reason, the groups are collapsed into one proband group, although occasional descriptive information available only for the longitudinal group is provided where relevant.

Criteria for Inclusion and Related Medical History

The children with SLI were recruited for participation during their preschool year prior to kindergarten. They were all between 4 1/2 and 5 1/2 years of age. They were recruited from the caseloads of area speech-language pathologists and were thus all receiving speech and language services at the time. The control children were recruited from area preschools and daycare centers.

For the probands, inclusionary and exclusionary criteria were as follows. The children with SLI were required to (a) score below a standard score (SS) of 85 on the receptive language measure, the PPVT–R; (b) fall
below age expectations in terms of expressive language, as measured by MLU, which was compared to the normative sample of Leadholm and Miller (1993); (c) score within normal limits, above a SS of 85, on a nonverbal performance measure, the Columbia Mental Maturity Scale (CMMS; Burgemeister, Blum, & Lorge, 1972); and (d) have no major articulation problems that would hinder the intelligibility of the grammatical morphemes of interest. The sounds-in-words portion of the Goldman-Fristoe Test of Articulation (GFTA; Goldman & Fristoe, 1986) was administered, and participants were also required to pass a phonological screening, which assured the accurate production or consistent approximations of final /d/, /v/, /s/, and /l/. Children with consistent mispronunciations, most of which were interdental productions of sibilants, and w/l or w/r distortions, were included. At this age level, such phonological patterns can be regarded as within the range of developmental expectations. Children with limited intelligibility were excluded. Furthermore, the children with SLI were required to pass hearing screenings and to have no known neurological, social/emotional, or behavioral disorders as reported by the speech-language pathologists.

Of the 31 proband children, further information is available for 19 children who are participating in the ongoing longitudinal study. According to parent report, these children's perinatal status and subsequent medical histories are uneventful, in comparison with available data on 41 control children. Children with a history of NICU placement comprise 11% of the SLI group and 12% of the controls; the proportion of children of low birthweight (3 to 5.1/2 lbs) is actually higher in the controls (7%) than in the SLI group (0%). There is no history of seizures in either group. A high proportion of children in each group has experienced at least one episode of ear infection (85% of the control group and 79% of the SLI group). In terms of frequency, 79% of the SLI group and 87% of the control group were reported to have ear infections once or twice a year; 21% of the SLI group and 28% of the control group were reported to have ear infections once a month. Because these children were recruited in the same way as the children in the proband group of the cross-sectional study, and the two samplings are similar in many other ways (cf. Rice & Waxler, 1996b), it is presumed that their medical histories will be similar, although because such evidence for the earlier sample of children was unavailable, this assumption cannot be confirmed.

Control children were selected in two ways. One group consisted of children at equivalent language levels, who were 2 years younger than the children with SLI. The second group were children matched to the SLI group on the basis of chronological age. For the purpose of this study, the two groups are collapsed. Because their selection criteria differed somewhat, they will be described separately here. The language-matched group was matched on the expressive language measure of MLU, for which there was a child-to-child match within 0.1 MLU based on a 200-utterance sample of spontaneous speech. These younger children were required to score within normal limits, above a SS of 85, on the PPVT-R and to have no clinically significant articulation problems. As with the children with SLI, these children were required to pass hearing screenings and to have no known neurological, social/emotional, or behavioral disorders. The language-matched children were too young to be given the CMMS at the time of recruitment. However, the CMMS was administered 6 months later, and all of them scored well within normal limits, above a SS of 85.

The children in the second control group were matched by chronological age to the children with SLI and were in the preschool year prior to kindergarten, aged 4½ - 5½ years, at the time of recruitment. They were required to score within normal limits, above a SS of 85, on the PPVT-R, to score within normal limits, above a SS of 85, on the CMMS, and to have no clinically significant articulation problems. As with the other children, these children were required to pass hearing screenings and to have no known neurological, social/emotional, or behavioral disorders.

**Evidence of Affectedness for EOI**

The proband group differed from each of the control groups in their performance on the set of morphemes that mark grammatical tense. A composite tense score was calculated, consisting of the mean percentage correct in obligatory contexts on probe and spontaneous measures of third person present tense singular -s, regular past tense -ed, copula and auxiliary BE, and auxiliary DO. Detailed information about the measures is provided in Rice and Waxler (1996b). The group means and standard deviations were as follows: SLI group, .36 (.20); language-matched controls, .55 (.23); and age-matched controls, .93 (.07). These group differences are significant, as determined by an overall ANOVA, F(2, 95) = 94.24, p < .001, and pairwise comparisons show the SLI group below the language-matched control group (p < .001). Subsequent analyses of the longitudinal sample outcomes show that the measures yield the same pattern of group differences through six times of measurement, to age 7 years (Rice & Waxler, 1997).

Another way to examine affectedness is to consider how many children in the SLI group are truly affected and how many children in the age-control group are truly not affected. This can be determined by setting a criterion level of 70% accuracy for the composite tense measure. At that level, 28 of the 31 (90%) children with SLI are affected and none of the 37 (0%) age-matched control children are affected. If the criterion is raised to
30, 30 (97%) of the SLI children are affected and one (3%) of the age-matched control group is affected. It is important to note that two of the SLI children who performed in the .70-.79 range are children who were added to the longitudinal study at the second round of testing and are actually 6 months older than the children in the SLI group tested at the first round. Thus, the exceptional status of these two children is probably attributable to their greater age, and the more accurate estimate of selectivity is probably 97% (30/31).

Procedures

Following the methodology of previous studies, most of the data were collected through phone interviews, although some personal interviews were carried out as well. The limitations of this method include the following: (a) we are dependent upon the family informant's judgment of a clinical condition that may have only subtle clinical symptomatology in individuals beyond the early childhood period; (b) the informant may not know the individual's earlier clinical status; (c) there are probably cohort effects for clinical identification, such that clinical services may have been less available to older individuals; (d) individuals with an earlier history of language impairments may have learned to compensate in such a way that surface symptoms may no longer be evident; (e) the actual nature of the impairment cannot be determined, and (f) an unknown level of uncertainty on the part of the respondent. These limitations appear to apply equally across groups. In most cases, the respondent was the child's mother, but there were a few fathers and one maternal grandmother, the child's legal guardian, who were the respondents. The format for data gathering was a family listing, adapted from Lewis and Freebairn (1993). In her thesis study, K. J. Rice (1996) found that a listing method was preferred to a general questionnaire method (cf. Lahey & Edwards, 1995) because the listing seemed to help the informant remember the status of each individual. Beginning with the nuclear family members (mothers, fathers, and siblings), respondents were asked to provide name, birthdate, handedness, age, whether or not there was history of speech difficulties, history of language difficulties, history of reading difficulties, history of spelling difficulties, and history of learning disabilities (e.g., mental retardation) for each member of the family. For the extended family, this was asked about mothers' parents, siblings, nieces, and nephews, and fathers' parents, siblings, nieces, and nephews. Maternal and paternal grandparents and cousins were not included in the information. Also note that only blood-related family members were included in the listings. Relatives who married into the families were not included.

Consistent with previous studies, we included as affected for speech/language impairment any individual identified as previously enrolled in speech or language treatment. At the outset, we had hoped to elicit judgments from the informant about the grammatical status of the family members. Two problems arise. One is that it is not reasonable to assume that all cases of affectedness for grammatical impairments are likely to have been enrolled in language treatment. The other is that surface symptomatology is likely to change with age, and the family informant may not be aware of the individual's childhood status. For these reasons, we sought descriptors that were more sensitive than enrollment in treatment and that could meaningfully describe adults with a positive history. In early pilot testing, we learned that informants were uncertain what was meant by "grammatical impairment" or, for that matter, "speech impairment" or "language impairment." For this reason, we provided examples of surface symptoms for each category of impairment. In order to clarify what is meant by a "speech difficulty," the following examples were provided: mispronounces long words; is hard to follow when (s)he tells personal experiences or long stories; is hard to understand or mispronounces words; stutters. For "language difficulty," the following examples were provided: slow in learning to talk; has awkward sentence structures when writing or talking; has difficulty making sentences or finding the best words to express ideas; has a hard time carrying on a conversation; has difficulty thinking of words (s)he wants to say when talking; is hard to follow when (s)he tells personal experiences or long stories; has difficulty explaining things; has a more limited vocabulary than other family members. For "reading difficulty," the following examples were provided: doesn't like to read; doesn't read well; has had reading therapy and/or been identified for special reading help at school. Informants considered the symptoms when determining whether or not individuals should be classified as speech/language impaired. Likewise, for reading/spelling/learning impairments, surface symptoms were considered when it was not known if the individual had received special services in school or if there had been a history of diagnosis such as mental retardation.

Results

Percentage of Families Who Reported Affectedness

A positive family history of affectedness was calculated as the percentage of families who reported affectedness for at least one nuclear family member other than the proband or control child. This percentage, for each group, resulted from dividing the number of families that reported at least one nuclear member as affected by the total number of families. The two major symptoms of
affectedness are the dichotomous variables of (a) speech/language disorders, which is defined as a positive report for either speech or language disorder according to the examples provided and/or a history of treatment; and (b) reading/spelling/learning disorders, which is a positive report according to the examples provided or, most frequently, academic difficulties in these subjects, receiving special services, or grade retention. A third, and more inclusive, dichotomous category of affectedness is also computed, (c) overall affectedness, which is a composite of the first two categories. The positive history findings are as follows for the 31 proband families: (a) 58.1%, (b) 35.5%, (c) 64.5%, and for the 67 control families, (a) 19.4%, (b) 22.4%, (c) 37.3%.

Further information is available for the longitudinal sample only, in the form of informant responses to a series of questions beginning with “Does anyone in your family have a history of...?” The symptoms queried were as follows: slow in learning to talk, uses awkward sentence structure, has a hard time carrying on a conversation, has difficulty thinking of words (s)he wants to say when talking, has a poorer vocabulary than other family members, is hard to follow when (s)he tells you something, has difficulty explaining things, is less talkative, stutters, mispronounces long words; is a poor speller, doesn’t read well, doesn’t like to read. The findings for the proband and control groups are reported in Table 1. Chi square ($\chi^2$) analyses yield significant differences between groups for all symptoms with the exception of the following: has difficulty choosing words, is hard to follow, is less talkative, stutters, and is a poor speller.

**Affectedness Rates of Family Members**

Next, affectedness rates of family members within proband and control families were calculated for the areas of affectedness. These are broken out according to whole families (nuclear and extended), nuclear family members (mothers, fathers, brothers, and sisters), and extended family members (aunts, uncles, cousins, and grandparents). This rate is calculated, for example, by dividing the number of nuclear proband family members reported as affected by the total number of nuclear proband family members. Probands and control children are not included in the counts, nor are adopted family members or half-siblings. Chi-square analyses were used to indicate whether the differences in proportions between groups were statistically significant.

**Whole Families**

For the proband group ($N = 555$), about 15% of proband family members, as compared to about 6% of control family members ($N = 1285$), are reported to have speech and language difficulties, a significant difference, $\chi^2 (1, N = 1838) = 41.4, p < .001$. In terms of reading/spelling/learning difficulties, however, such differences do not appear. About 7% of proband family members and about 5% of control family members are reported to have reading/spelling/learning difficulties, $\chi^2 (1, N = 1838) = 2.32, n.s.$ If one takes into account overall affectedness, that is, being affected in either area, the overall affectedness rate for proband families is about 18%, whereas for control family members, it is about 9%. This difference is significant, $\chi^2 (1, N = 1838) = 31.6, p < .001$. Therefore, speech and language difficulties, as well as overall affectedness rate, differentiate proband and control families as wholes.

**Nuclear Families**

Although speech and language difficulties and overall affectedness seem to differentiate the families as wholes, it is important to determine whether this pattern holds for nuclear and extended families, considered separately. A higher rate for probands is evident in these counts, as well. About 22% of nuclear proband family members ($N = 110$) are reported to have speech and language difficulties, as compared to 7% of nuclear control family members ($N = 197$). This difference is significant, $\chi^2 (1, N = 307) = 14.1, p < .001$.

As seen in the whole family data, there is no significant difference between families in terms of reading/spelling/learning difficulties for nuclear members. About 13% of nuclear proband family members and about 9% of nuclear control family members are reported to have such difficulties, not a significant difference, $\chi^2 (1, N =$
307) = .81, n.s. With regard to overall affectedness, the rate is about 26% for nuclear proband family members and about 18% for nuclear control members, a significant difference, $\chi^2 (1, N = 307) = 9.10, p < .01$. Overall, the proband and control nuclear families are differentiated by difficulty with speech and language, as well as overall affectedness.

Extended Families

For extended family members, a pattern similar to that for nuclear family members is evident, in which there are higher rates for speech and language symptoms, and overall affectedness, but not for reading/spelling/learning difficulties. About 14% of extended proband family members ($N = 445$) are reported to have speech and language difficulties, as compared to about 6% of extended control members ($N = 1066$), a significant difference, $\chi^2 (1, N = 1531) = 26.0, p < .001$. In contrast, about 5% of extended proband members and about 4% of extended control members are reported to have reading/spelling/learning difficulties, not a significant difference, $\chi^2 (1, N = 1531) = .97, n.s.$ With regard to overall affectedness, the overall rate of affected extended proband family members is about 16% whereas for extended control family members, it is about 9%. This difference is significant, $\chi^2 (1, N = 1531) = 20.7, p < .001$.

To determine if the rate of affectedness in nuclear family members is higher than that of extended families, a series of chi square analyses were carried out. Recall that the rate of affectedness for speech/language symptomatology is about 22% for the probands' nuclear family members and about 14% for extended family members. This difference is significant, $\chi^2 (1, N = 555) = 4.77, p < .05$. Likewise, there is a significant difference between nuclear and extended proband family members in terms of reading/spelling/learning difficulties. About 12% of nuclear proband family members are reported to have these sorts of difficulties whereas about 5% of extended proband family members are reported to have such difficulties, $\chi^2 (1, N = 555) = 5.85, p < .05$. Finally, a significant difference is found in terms of overall affectedness between nuclear and extended proband family members. The rate of overall affectedness for nuclear family members is about 26%, compared to about 16% for extended members, a significant difference, $\chi^2 (1, N = 555) = 5.83, p < .05$.

To summarize, there are significantly higher rates of reported speech and language difficulties, as well as overall affectedness, in whole proband families than in whole control families, a pattern that holds when these larger families are broken down into nuclear and extended levels. Furthermore, there is a significant difference between the two levels of families in proband families. That is, proband nuclear family members have significantly higher rates of reported speech and language difficulties, reading/spelling/learning difficulties, and overall affectedness rates than proband extended family members.

Affectedness in Nuclear Proband Families

The rates of affectedness within the nuclear proband family, broken out by fathers, mothers, brothers, and sisters are examined next. These analyses are reported in Table 2. Focusing on speech and language difficulties, about 16% of proband parents and about 27% of proband siblings are reported to have such difficulties, although this difference is not statistically significant, $\chi^2 (1, N = 110) = 1.38, n.s.$ In terms of which parent is more often reported to have speech and language difficulties, however, there is a significant difference between proband mothers and proband fathers: 29% of fathers are reported to have such difficulties compared to 7% of mothers, $\chi^2 (1, N = 62) = 5.42, p < .05$. Between siblings though, there is not a significant difference in terms of speech and language difficulties. About 26% of brothers and about 29% of sisters are reported to have such difficulties, $\chi^2 (1, N = 48) = .04, n.s.$

In terms of reading/spelling/learning difficulties, there are no significant differences between parents and siblings, $\chi^2 (1, N = 110) = 2.53, n.s.$, mothers and fathers, $\chi^2 (1, N = 62) = .48, n.s.$, or brothers and sisters, $\chi^2 (1, N = 48) = .14, n.s.$ Similarly, there are no significant differences between parents and siblings, $\chi^2 (1, N = 110) = .02, n.s.$, mothers and fathers, $\chi^2 (1, N = 62) = 3.08, n.s.$, or brothers and sisters: $\chi^2 (1, N = 48) = .04, n.s.$, in terms of overall affectedness.

Gender Differences

Do affectedness ratings differ between families of probands based on proband gender? The families are split into two groups for these analyses according to the

**Table 2.** Percentage of nuclear proband family members reported to have positive histories of affectedness.

<table>
<thead>
<tr>
<th>Nuclear family members</th>
<th>Speech/ language difficulties</th>
<th>Reading/Spelling/ learning difficulties</th>
<th>Overall affectedness</th>
</tr>
</thead>
<tbody>
<tr>
<td>Parents, $N = 62$</td>
<td>17.7</td>
<td>16.1</td>
<td>25.8</td>
</tr>
<tr>
<td>Fathers, $N = 31$</td>
<td>29.0</td>
<td>19.4</td>
<td>35.5</td>
</tr>
<tr>
<td>Mothers, $N = 31$</td>
<td>6.5</td>
<td>12.9</td>
<td>16.1</td>
</tr>
<tr>
<td>Siblings, $N = 48$</td>
<td>27.1</td>
<td>6.3</td>
<td>27.1</td>
</tr>
<tr>
<td>Brothers, $N = 27$</td>
<td>25.9</td>
<td>7.4</td>
<td>25.9</td>
</tr>
<tr>
<td>Sisters, $N = 21$</td>
<td>28.6</td>
<td>4.8</td>
<td>28.6</td>
</tr>
</tbody>
</table>

Proband families: $N = 31$
gender of the proband: male \(N = 20\) and female \(N = 11\). The percentage of affected nuclear family members for the male probands \(N = 72\) are as follows: speech/language, 25%; reading/spelling/learning, 12.5%; and overall affectedness, 29.2%. For the female probands \(N = 38\), speech/language, 15.8%; reading/spelling/learning, 10.5%; and overall affectedness, 21.1%. There is almost no difference between groups of families based on proband gender. Although more members of male proband families than female proband families seem to be reported as having speech and language difficulties, these differences are not significant, \(\chi^2(1, N = 110) = 1.24\), n.s. Likewise, there are no significant differences between families of male and female probands for reading/spelling/learning difficulties or for overall affectedness. K. J. Rice (1966) examined whole and extended families for gender effects in a parallel way, and reports very similar findings.

**Composite Family Pedigree**

The findings for the speech/language affectedness rates for the nuclear and extended family members of the families of proband children are summarized in Figure 1. In this figure, the shaded hexagon represents the group of male and female probands through whom the families were identified. The percentage inside the square to the left of the hexagon indicates that about 26% of proband brothers are reported as having speech and language difficulties. Likewise, the 0 inside the uppermost right hand circle indicates that none of the proband maternal grandmothers are reported as having speech and language difficulties. It can readily be seen that the rate for fathers (29%) is higher than that of mothers (7%), and the rates for brothers and sisters are nearly equivalent (26%, 29%), and, in general, the rate for nuclear family members is higher than that of extended family members (although the rates for aunts and uncles are relatively high). A comparable figure for the families of children in the control group would show that the rates of speech/language affectedness approximate the reported prevalence rates for the general population (i.e., in the range of 3–11%). Nowhere in the nuclear and extended pedigrees do the rates exceed 11%; mothers' and fathers' rates are each 9%; brothers', 3%; and sisters', 4%.

**Discussion**

What this study establishes is that children with SLI known to demonstrate an EOI grammar, a form of language impairment characterized by specific grammatical affectedness, also show elevated rates of reported speech/language impairments in their families, relative to control children who have unaffected grammars. This points in two directions. One is to establish that children with SLI (at least of the expressive/receptive kind demonstrated by the sample here) who, as a group, are likely to have certain kinds of grammatical deficits also show a strong pattern of familial speech/language impairment. The second direction is to add further support to the conclusion that grammatical impairments could be inherited. The findings here are congruent with the nine older children studied in Van Der Lely and Stollwerck (1966) who were selected on the basis of persistent grammatical impairments.

Collapsing across nuclear and extended family members, 15% of the family members of the probands are reported to have speech/language difficulties. If the definition of affectedness is widened to include difficulties
of reading, spelling, and learning, the rate rises to 18%. Furthermore, the rates of affectedness conform to expected properties of a condition transmitted through families, in that reported rates of affectedness follow the degree of relatedness: Higher rates appear within nuclear families (22% for speech/language difficulties) than among extended family members (14% for speech/language difficulties). What must be acknowledged, however, is that the reported rates of affectedness could be influenced by how well the family informant knows the other members of the family. Because the informant is likely to be more familiar with nuclear family members, and therefore more likely to detect their impairments, a familiarity effect cannot be disentangled from the degree of relatedness effect for informant-report data.

The actual rates of affectedness found in this study are strongly congruent with previous findings from studies following similar methods. Because previous studies report findings for nuclear families only, the comparisons here are limited to nuclear families. The finding here of 26% for overall affectedness is similar to the average overall affectedness rate of about 20% reported by Niels and Aram (1986), and is identical to the 26% reported by Lahey and Edwards (1995). Here we report 22% for speech and language difficulties, which is very similar to the 23% rate reported by Tomblin (1989). It seems safe to conclude that rates of 20–25% are to be expected for the number of nuclear family members affected with speech/language difficulties, with an additional 2–5 percentage points if the definition of affectedness is expanded to include language-related difficulties, described here as “overall affectedness.” From the perspective of these findings, estimates in the range of 40%, as reported by Tallal et al. (1989), are likely to be too high. The higher rate of Tallal et al. may be attributable to an unknown confound with speech impairments. An alternative possibility is inadvertent selective sampling. This is suggested by the 39% rate reported by Van Der Lely and Stollwerck (1966) in their selectively sampled group of children with grammatical SLI.

When individual nuclear family members are considered, the present findings indicate that 29% of proband fathers, about 26% of proband brothers, about 29% of proband sisters, but only about 7% of proband mothers are reported to have speech and language difficulties. The relatively high rate of father affectedness is very informative because it rules out an X-linked dominant trait in which affected men never transmit the disease to their sons but always transmit it to their daughters (Bruzustowicz, 1996). In the family pedigrees of the probands there are 10 families in which father-to-son patterns are reported. With regard to other studies, these findings differ from those of Tallal et al. (1989), who found a higher rate for mothers than for fathers for a history of language problems (about 33% of mothers and about 18% of fathers). They also differ somewhat from the findings of Tomblin (1989), who found speech and language affectedness reported for about 40% of brothers, 17% of sisters, about 10% of fathers, and about 20% of mothers. These discrepancies suggest that estimates of risk for mothers versus fathers, and brothers versus sisters, show some instability from one study to another, and therefore must be viewed with caution.

At least part of these differences may be attributable to differences in the definition of affectedness. Recall that Lahey and Edwards (1995) identified a mixed group, that is, a group of children with both expressive and receptive difficulties, which could also characterize the probands in this study. Their findings for proband siblings is 27% (not reported for brothers and sisters separately), the same as the finding here.

A final replication was the finding that it did not seem to matter whether the families were identified through a male or female proband. This finding parallels that of Tomblin (1989), who also found no difference in the rates of affectedness within families based on proband gender.

**Matters of Identification: Probands and Family Members**

This study adds important details to what is known about proband identification. As argued elsewhere (cf. Rice & Wexler, 1996a), there are theoretical reasons to expect that an EOI grammar could be an inherited form of grammatical limitation. If such a grammatical marker exists undetected beneath low performance on conventional language tests, we would want to know about it in order to better understand possible inherited deficits, to better predict which subgroups of children with SLI are likely to carry inherited deficiencies, to develop appropriate measures for older individuals, and to provide the precision necessary for carrying out molecular linkage studies. For the foreseeable future, such specific grammatical markers are not likely to replace conventional language assessments as a standard part of the clinical assessment battery (cf. Tomblin, Records, & Zhang, 1996), but they may become highly significant assessment methods to be used in tandem with more comprehensive tests, especially for the purpose of genetic studies.

The study also sheds some light on the important problem of how to elicit judgments from family informants about the current and previous language competencies of family members. When asked about specific symptoms, the informants’ responses differentiated the proband and control groups across a range of symptoms. It was encouraging that questions that probed more abstract grammatical properties, such as “awkward sentence structures,” as well as more obvious symptoms.
such as word pronunciation, were meaningful and seemed to differentiate the groups.

**Comorbidity of Symptoms in Affected Family Members**

Although the probands studied here were selected because their language impairment is not comorbid with extensive speech problems or stuttering, these symptoms were reported at higher rates in the families of probands than of controls. These findings have parallels in a recent study of family aggregation by Lewis (1996), who recruited two groups of probands, one for moderate to severe phonology disorders and another for reading disorders. Each of these groups were reported to have a higher prevalence of speech/language impairments, reading, and spelling disorders than is expected in the general population. Interestingly, in families of the phonology probands, a higher prevalence of speech/language disorders was evident than for the families of reading probands. The converse was true for the prevalence of reading, spelling, and learning disabilities. The rates were higher in the reading proband families than in the phonology proband families. At this level of analysis, then, reported symptomatology of family members paralleled the symptomatology of the probands.

In this study, the highest rates of family affectedness for individual symptoms appeared for the item, “doesn't like to read.” Another symptom, “doesn't read well” also showed relatively high rates. This is interesting because preliminary data from the longitudinal sample, collected when the children were in first and second grade, show that, as a group, they are encountering great difficulty in the transition to reading. It is highly likely that this group of children with known grammatical limitations will go on to become a group of children with known reading limitations. This raises the possibility that at least some of the evidence supporting the known genetic basis of reading disability (cf. Smith, Pennington, & DeFries, 1986) includes children with previously undiagnosed preschool grammatical impairments. Clearly, much more needs to be learned about the ways in which different phenotypes (i.e., those of grammatical limitations, more general language impairments, phonological deficits, and reading deficits), are running through the same families, and whether these phenotypes arise from the same sources. In future studies it will be important to establish, as precisely as possible, the symptomatology of affected adult and child family members.

**Similar Symptoms, Different Etiologies**

Another possible complication is that, out of the population of children identified as language impaired, different etiologies are at work, such that some cases of language impairment are familial in nature whereas others are not. This possibility is clearly demonstrated in a recent study by Rice et al. (1997), who report on family history data from parents of children who, as newborns, received care in a neonatal intensive care unit (NICU). This sample of 34 infants constituted a group of high-risk children based on gestational and/or perinatal factors. Out of this sample, 18 children (53%) showed language impairment at 4 years of age (a number of these children also showed speech impairments). The family history data, however, show that the affected group did not differ from the unaffected group in rate of affectedness among nuclear family members. The rate of speech/language problems in the affected group was .05, for the control group, .06. Both of these rates are far below the 20–40% estimates that appear in other studies, and in fact are highly similar to the population estimate of 7%. The conclusion is that it is likely that the language impairments evident in this sample of NICU babies are not of a familial nature. In a similar vein, investigators have entertained the possibility that an expressive-only language impairment (as with the children studied by Whitehurst et al., 1991) may arise from a different etiology than a mixed profile of receptive + expressive impairment, although, as noted earlier, the available evidence is contradictory (cf. Lahey & Edwards, 1985).

The study reported here shows that 5-year-old probands with known grammatical limitations, a history of delayed acquisition of language, and a history of minimal perinatal risk demonstrate a strong pattern of familiality, in contrast to the findings from the NICU sample. Recall also that Rice et al. (1997) clearly show that environmental factors, such as mother's education or the family's welfare status, do not predict the children's grammatical development. Comparing the findings across these two studies, it is quite possible that grammatical limitations, such as EOI, can conceivably arise from different etiologies, one attributable to perinatal risk during infancy and the other attributable to familial factors, such as genetic risk. Of course, the same underlying neurocognitive mechanisms may be implicated if the biological contributor is the same whether caused by conditions at conception, during gestation, or at birth. It is well known that this can occur with other biological conditions, such as breast cancer, that may or may not be caused by inherited factors. The lesson to be drawn from other genetic research, and the reason to highlight the issue here, is that if grammatical impairments can arise from familial and nonfamilial sources of risk, it will be crucial that probands are well described and relatively homogenous. Otherwise, the estimates of familiality can be spuriously raised or lowered.
Conclusions

This study shows that a group of child probands known to have carefully specified grammatical deficits (i.e., an EOI stage) come from families more likely to have individuals with speech/language impairments than is the case for a control group of unaffected children. This is a significant step in furthering what is known about possible etiologies for this unexplained and unexpected variation in a fundamental property of human linguistic competence. At the same time, the study of Rice et al. (1997) suggests that conditions such as EOI may not necessarily be familial but can also result from other forms of risk to an infant. This is not contradictory because both inherited conditions and non-inherited conditions can conceivably affect the biological structures that underpin language acquisition in similar ways. Grammatical competencies can vary significantly across children who are highly similar in other developmental domains, and these individual variations can be linked to familiality. These findings join other results pointing in the direction of increased familial risk for language impairment in the families of children diagnosed with SLI.

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