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# Phenotypic Profiles of Language-Impaired Children Based on Genetic/Family History

PAULA TALLAL

Center for Molecular and Behavioral Neuroscience, Rutgers University

JEANNE TOWNSEND

Language Research Center, Children's Hospital, San Diego; Joint Ph.D. Program, Psychiatry Department, University of California, San Diego and Psychology Department, San Diego State University

SUSAN CURTISS

Department of Linguistics, University of California, Los Angeles

AND

## **BEVERLY WULFECK**

Department of Pediatrics, University of California, San Diego

Although etiological influences in developmental language impairment (dysphasia) are not well defined, a significant increase of family aggregation for the disorder has been reported. We report data from a large cohort of language-impaired (LI) children participating in the San Diego longitudinal study in which we examined whether children with or without positive family histories show different phenotypic profiles. Due to the longitudinal design of the study, questions pertaining to change over time are also addressed. Second, a subgroup of the most impaired children were reevaluated to obtain additional information pertaining to family history and phenotypic outcome. Approximately 70% of the LI children met criteria for inclusion as family history positive, with fathers reporting a history of language or learning problems one and a half to two times as frequently as mothers. LI children with or without a positive family history were not sig-

Send all correspondence and reprint requests to Paula Tallal, Rutgers University, Center for Molecular and Behavioral Neuroscience, 197 University Ave., Newark, NJ 07102. This work was funded by NINCDS Contract No. NS92332 and Center Grant P50-NS22343.

nificantly different on language skills or IQ. However, subjects having a positive family history for developmental language/learning problems were significantly lower in socioeconomic status and were rated by parents and teachers as having more attention-related behavior problems than their family history negative counterparts. Similarly, family history positive LI children performed more poorly on standardized academic tests as well as on tests of auditory processing and attention. © 1991 Academic Press, Inc.

It has been estimated that 8–15% of preschool children have some form of speech or language disorder (Beitchman, Nair, Ferguson, & Patel, 1986; Silva, 1980). Such disorders can be secondary to sensory impairment, motor dysfunction, neurological disease, or more global developmental disorders such as mental retardation or autism. Language disabilities may also result from postnatal brain injury (acquired childhood aphasia). Language disorders occurring in the absence of such primary impairments are known as developmental dysphasia or specific language impairment (Benton, 1965). Diagnosis of such disorders is largely by exclusion (DSM-III-R, 1987), and current research focuses on etiology, symptomology, subtypes, and course. A number of questions pertaining to etiology and subtypes were addressed in the recently completed San Diego Longitudinal Study; a large, multidisciplinary, longitudinal study designed to evaluate the outcomes of preschool receptive and expressive language impairments.

Findings from the San Diego Longitudinal Study related to familial etiology of developmental language disorders demonstrated that there was a significantly higher incidence of childhood language/learning disabilities among first degree relatives (mothers, fathers, siblings) of developmental dysphasics than among first degree relatives of age-, IQ-, and Socioeconomic status (SES)-matched control children who were acquiring language normally (Tallal, Ross, & Curtiss, 1989a). An unexpected result from this study was finding a significant sex ratio difference for language-impaired (LI) children (3:1 male to female) that obtained only for LI probands with a familial history of language disorder. Further analyses of these data revealed that the expected increase of boys to girls with language impairment occurred only in families with an affected mother. These mothers gave birth to three times as many males as females altogether, and half of their offspring were affected, accounting for the observed sex ratio difference in this population. Importantly, sex ratios were not significantly different from the expected 1:1 in families with only an affected father. or in which neither parent was affected (Tallal, Ross, & Curtiss, 1989b).

The findings of familial aggregation in language disorders and aberrant sex ratios among families with affected parents may reflect the influence of individual or interacting genetic, hormonal, or environmental factors. Although the specifics of etiological influences are yet to be determined, it is clear that some subsets of LI children have a family history of developmental language impairment while others do not. It is possible that such differences in familial history may define subgroups of developmental

dysphasia displaying different behavioral (phenotypic) profiles. This study used data from the San Diego Longitudinal Study and from the Center for the Neurological Basis of Language, in which a subset of the more severely impaired dysphasic children from the original study were followed to examine whether specifically language-impaired children with and without positive family histories show different phenotypic profiles.

## STUDY 1

#### **Subjects**

Subjects for this study came from a pool of 89 well-selected, specifically language-impaired children who were chosen at age 4 to participate in the San Diego Longitudinal Study evaluating the outcomes of preschool language impairment (for a detailed description of the subject groups see Tallal et al., 1989a). Inclusion criteria were: (1) age 4;0-4;11 at induction; (2) a Leiter performance IQ of 85 or better (Leiter, 1940); (3) a language age computed from standardized expressive and receptive scores which was at least 1 year below performance mental age and chronological age; (4) normal hearing acuity; (5) no motor handicaps or oral structural impairments affecting nonspeech movement of the articulators; (6) an English language background only; (7) not autistic (as defined by DSM-III, 1980); and (8) no known neurological disorders. Over the course of the 5-year period of this longitudinal study, subjects were evaluated annually on measures of speech, language, neuropsychology, academic achievement, intellectual, emotional, and social development. In addition, information pertaining to medical and family history, as well as current status and educational history, was obtained from questionnaire data.

## Procedures

Family history of language disorder was determined from questionnaire and interview data. If an LI proband's mother or father reported (1) having been held back a year in elementary school, (2) having been below average in reading or writing, or (3) having had a speech or language deficit or speech therapy, that subject was coded as family history positive for language/learning impairment. Data were collected from each parent directly, and information supplied by one parent about the other was not used. Information about primary and secondary affected relatives (e.g., siblings, aunts, uncles, grandparents) was collected but, in this study, only information from the biological mother and father was used for coding of family history of language impairment.

## Results

In this study, family history was determined for 65 of the languageimpaired subjects on whom there were complete data for the full 5 years of the longitudinal study. Approximately two-thirds (42) of these probands

 
 TABLE 1

 Study 1 Demographic Data for Language-Impaired Subjects Having a Positive or Negative Family History for Language Disorder

	Family history		
	Negative	Positive	
N	23	42	
Affected parent			
Father only	0%	52%	
Mother only	0%	24%	
Both	0%	24%	
SES			
Upper	35%	10%	
Middle	65%	82%	p < .03
Lower	0%	8%	
Sex			
Male	65%	71%	
Female	35%	29%	
	Mean (SD)	Mean (SD)	
Age at Year 1 of Study	4;4 (0;4)	4;5 (0;4)	
Receptive language age equivalent	3;1 (0;5)	3;1 (0;5)	
Expressive language age equivalent	3;0 (0;3)	3;0 (0;2)	
Leiter performance IQ at Year 1 of study	109.5 (11)	107.5 (13)	
Leiter performance IQ at Year 5 of study	102.2 (14)	99.0 (13)	

*Note.* Receptive language age was computed using the following standardized tests: The Sequenced Inventory of Communicative Development (SICD) receptive scales (Hedrick, Prather, & Tobin, 1979), Northwestern Syntax Screening Test (NSST) receptive scales (Lee, 1969), and The Token Test (De Renzi & Vignolo, 1962). Expressive language age was computed using: SICD-Expressive Scales, NSST expressive scales, and The Carrow Elicited Language Inventory (Carrow, 1974).

met criteria for a positive history, with fathers reporting developmental language or reading difficulty about one and one-half times as often as mothers. Twenty-three LI probands (approximately one-third) came from families in which neither parent reported childhood speech, language, or academic difficulties. These were classified as family history negative. Proportions of affected mothers and fathers and demographic data for this sample are presented in Table 1. This is the same proportion of affected to nonaffected parents reported for the full cohort in the first years of the study (Tallal et al., 1989a). There were no statistically significant differences between the family history negative and positive groups in age, IQ, receptive or expressive language age equivalents, or distri-

bution of gender. There were, however, more family history negative children living in families with higher socioeconomic status,  $\chi^2(2) = 7.01$ , p < .03. Because of this association between family history of language/learning impairment and SES, all statistically significant effects were also analyzed with SES as a covariate. All statistically significant effects remained statistically significant with SES covaried.

There were also differences between the family history negative and positive groups in reported behavior problems. In Year 1 of the study when the children were age 4, on the Achenbach Child Behavior Checklist, parents reported more problems overall for the positive history subjects, t(61) = 3.13, p < .005. In Year 5 of the study the trend was in the same direction with generally more problems being reported for the positive history group, t(59) = 1.69, p < .098.

There were no statistically significant differences between the groups in prereading skills as assessed with the Comprehensive Tests of Basic Skills (McGraw-Hill, 1973). However, by Year 3 of the study when subjects were 6 years old and standardized tests of academic skills were first administered, the children with no family history of language impairment consistently performed better on tests of reading and math than did children with a family history of language impairment (see Fig. 1). A repeated measures Multivariate Analysis of Variance (MANOVA) showed that a linear combination of the four academic tests across the 2 years was statistically different for the family history negative and positive groups with the children having no family history of language impairment performing significantly better, F(1, 63) = 4.46, p < .04. A MANCOVA with SES as the covariate produced a similar result (F(1, 60) = 4.32, p < .05) and showed no statistically significant relationship between SES and test performance. It should be noted that the means for both groups are below the 35th centile for all academic tests, reflecting well below average academic performance for the language-impaired children compared to a norming sample of their age and school grade. Details pertaining to academic achievement outcomes for this group and preschool factors which predict academic achievement outcomes in LI and normal children have been reported previously (see Tallal, 1988).

There were no other consistent differences between the family history positive and negative groups for clusters of measures representing language (see Table 1 note) or neuropsychological assessment including perception, speech, and motor skills (Tallal Repetition Tests, Tallal & Piercy, 1973; Goldman–Fristoe–Woodcock Test of Auditory Discrimination, 1970; Seriation Task adapted from Piaget & Inhelder, 1959; Simultagnosia adapted from Fink & Bender, 1953; Finger Identification, adapted from Benton, 1959; Finger Opposition, Touwen & Prechtl, 1970; Coins in Box, Doll, 1946; Rapid Automatized Naming, Denckla & Rudel, 1976; Manual Rapid Automatized Naming, adapted from Denckla & Rudel, 1976).

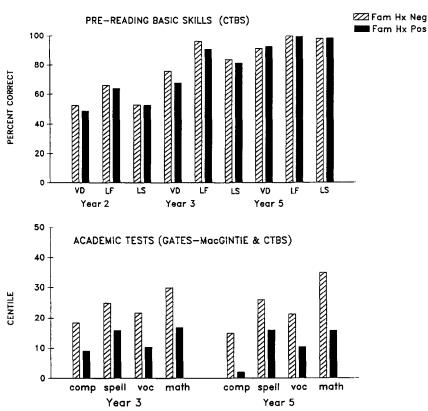


Fig. 1. Performance of language-impaired children with a positive or negative family history of language impairment on Comprehensive Tests of Basic Skills (McGraw-Hill, 1973) prereading tasks (Visual Discrimination, Letter Form, and Letter Sound) from Year 2 to Year 5 of the study, and on Gates-MacGintie tests of Vocabulary and Reading Comprehension (Gates MacGintie, 1972) and Comprehensive Tests of Basic Skills in Math and Spelling for Years 3 and 5. Subjects were 4 years old in Year 1 of the study and 8 years old in Year 5.

## Discussion

Major effects from this study are that children with a positive family history of language/learning problems show more behavior problems and come from families with a lower SES. Additionally, although they show similar neuropsychological language abilities, LI children with a positive history perform more poorly on standardized academic tests than do LI children with no family history of language/learning disorder.

It is not surprising that LI children having a positive history of language/learning impairment have a significantly lower SES than those with a negative family history. Language problems are often associated with poor academic achievement and hence lower levels of education. As education level is one of two factors used to determine SES (Hollingshead

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and Redlich, 1958), it follows that a difference would be found on this variable.

That the family history positive and negative subgroups would be similar on all of the neuropsychological and lingusitic measures, but differ in academic achievement and behavior, was an unexpected finding. However, several factors mitigate against a clear interpretation of these data. First, due to the longitudinal design of the study, subjects who were selected at age 4 years as language impaired differed considerably in their course of development, with some remaining severely language impaired and others performing within the normal range by the conclusion of the study. Second, despite differences occurring between family history positive and negative subgroups of LI children in academic achievement, the overall performance of both groups was well below average, raising concerns about possible floor effects in data interpretation. Third, relatively better academic performance by the family history negative group could have been affected by assistance to these children in academic endeavors from parents who had no history of academic problems. Finally, the data pertaining to increased behavior problems in the LI subgroup with positive family history are difficult to interpret as those data were derived only from parents. An interaction between parents who were themselves affected and their assessment of their affected offspring must be considered a potential source of bias. It is, therefore, important to have some outside validation of rated behavior problems to aid in the interpretation of these results.

## STUDY 2

In an attempt to enhance our ability to interpret the results of the first study, we examined a subset of the LI subjects who participated in a second study. These subjects represented those who were the most severely language impaired at the conclusion of the longitudinal study. This assured that all of the subjects participating in the second family history study were actually still language impaired, thus representing children with both serious and persistent language disorders. Although selection was based on test performance in the final year of the longitudinal study when subjects were 8 years old, testing for Study 2 commenced approximately a year later to ensure that the LI children had had a better opportunity to develop more adequate academic skills. A more detailed family history questionnaire was also introduced into Study 2. Finally, two forms of the Achenbach Child Behavior Checklist were included and compared; one completed by parents and one by teachers. These subjects also participated in other studies as part of the Center for the Study of the Neurological Basis of Language, which included electrophysiological and magnetic resonance brain imaging studies.

## **Subjects**

Following the fifth year of testing in the San Diego Longitudinal Study, when the subjects were between 8 and 9 years old, 28 of the most severely language-impaired subjects were selected to participate in Study 2. In addition to meeting original inclusion criteria described in Study 1, these subjects demonstrated age equivalence scores on standardized language tests that were at least 1 year below their chronologic age, and WISC-R Performance IQ or Leiter Performance IQ scores greater than 85. Approximately 1 year later subjects received a large battery of standardized and experimental language, neuropsychological, speech, and academic tests, as well as measures of behavioral, social, and emotional development.

## Procedures

Family history of language disorder was determined using a method similar to that of Study 1. Questionnaires were modified to elicit more specific information regarding family background, and results from these instruments were compared to earlier data. Where there were discrepancies parents were contacted to resolve questions. Cases for whom discrepancies could not be resolved were not used in this sample. Criteria for inclusion as family history positive were that a proband's father or mother or both reported having had one or more of the following problems: (1) below average or impaired school achievement in reading or writing; (2) placement in a remedial class for reading or writing; (3) kept back a grade or having failed a class; (4) below average or impaired language development as a child; or (5) speech therapy. As in the previous study, we accepted information about a parent only from that parent. Probands were coded as family history negative only if both mother and father reported having had none of the above listed problems.

# Results

The final cohort for Study 2 consisted of 23 language-impaired subjects for whom direct data from both mother and father were available to determine family history. Approximately 70% of these probands met criteria for a positive history of language impairment, with fathers reporting being affected about twice as often as mothers. Proportions of affected parents and demographic data are presented in Table 2. Note that age, language age equivalents, and Leiter IQ presented in this table were computed at selection for Study 2. All other results reported in this section are from testing done approximately 1 year later (mean age at testing 9;2, range 8;2–10;10, with no significant difference in age between positive and negative history groups). There are no statistically significant differences between family history positive and negative groups in age at

NEGATIVE FAMILY HISTORY FOR LANGUAGE DISORDER			
	Family history		
	Negative	Positive	
N	7	16	
Affected parent			
Father only	0%	62%	
Mother only	0%	19%	
Both	0%	19%	
SES			
Upper	43%	12.5%	
Middle	57%	75.0%	
Lower	0%	12.5%	
Sex			
Male	86%	62%	
Female	14%	38%	
	Mean (SD)	Mean (SD)	
Age at Selection	8;1 (0;2)	8;4 (0;5)	
Receptive language age equivalent	6;8 (1;0)	6;7 (1;0)	
Expressive language age equivalent	7;5 (0;6)	7;8 (1;0)	
Leiter performance IQ at Selection	107.1 (12)	94.2 (10)	p < .02
WISC-R performance IQ at center induction	105.6 (8)	101.5 (14)	

 
 TABLE 2

 Study 2 Demographic Data for Language-Impaired Subjects Having a Positive or Negative Family History for Language Disorder

Note. Receptive language age was computed using the following standardized tests: The Token Test (De Renzi, & Vignolo, 1962), and The Peabody Picture Vocabulary Test— Revised (Dunn & Dunn, 1981). Expressive language age was computed using: Grammatic Closure Subtest from the Illinois Test of Psycholinguistic abilities—Revised (Kirk, McCarthy, & Kirk, 1968), The Clinical evaluation of Language Function—expressive subtests (Semel & Wiig, 1980), and Expressive One-Word Picture Vocabulary Test (Gardner, 1979).

selection for Study 2, receptive or expressive age equivalents (determined at selection), WISC-R Performance IQ, or distribution of gender. The family history positive group, however, showed a lower Leiter performance IQ at selection for the Center, t(21) = 2.68, p < .02 (with SES covaried, F(1, 19) = 12.85, p < .002). Although the difference is not statistically significant in this sample, there was a trend for more family history negative children to be from families with higher socioeconomic status (see Table 2), a result also found in Study 1.

Significant differences between the family history positive and negative groups in behavior problems reported by parents in Study 1 were replicated in Study 2. A similar result was also found based on teacher reports.



On the parent report of the Achenbach Child Behavior Checklist, the overall total behavior score showed a trend for more behavior problems to be reported (t(20) = 1.79, p < .09) for the family history positive language-impaired children. Scales developed from factor analysis of the parent (Achenbach & Edelbrock, 1983) and teacher (Achenbach & Edelbrock, 1986) rating scales showed that family history positive children were rated by parents as being more "Hyperactive" (t(18.5) = 3.82, p)< .001, with SES covaried, F(1, 19) = 6.69, p < .02), and by teachers as being more "Inattentive" (t(19) = 2.17, p < .05, with SES covaried,F(1, 17) = 3.84, p < .07) than language-impaired children with a negative family history. There is considerable overlap in items comprising the scale labeled "Hyperactive" developed from parent ratings and the scale labeled "Inattentive" developed from teacher ratings. Items with the highest loadings that are common to both scales relate to attention and concentration (e.g., can't concentrate, daydreams, inattentive, confused), and academic performance (e.g., poor school work). Interestingly, no other scales from the Achenbach (e.g., aggression, anxiety, social withdrawal, etc.) were significantly different for the two groups, as rated by either parents or teachers. These data confirm the results of Study 1 while further specifying the nature of the behavior problems to be primarily in the area of attention deficit and poor academic performance.

A series of experimental measures of nonverbal auditory processing and attention consistently showed better performance by language-impaired children with negative family history (Hotelling's  $T^2 = 38.9$ , F(7, 15) = 3.97, p < .015). This analysis included all of the auditory processing tests administered in Study 2: the Tallal Repetition Test (Tallal & Piercy, 1973); Computerized Tests of Selective Attention (Townsend & Tallal, 1989); the Digit Span subtest of the Wechsler Intelligence Scale for Children-Revised (Wechsler, 1974); and the Quiet and Noise subtests of the Goldman-Fristoe-Woodcock Test of Auditory Discrimination (1970). Mcan standardized scores from these tests are presented in Fig. 2, along with scores from an age- and IQ-matched normal control group. Note that standardized scores were not used in the analysis, but have been computed for the convenience of displaying this series of tests on a common grid.

There were no further significant differences between groups of family history positive and negative children for performance on clusters of measures representing academic performance (Wide Range Achievement Test, Jastak & Wilkinson, 1984; Decoding Skills Test, Richardson & Dibenette, 1985), language abilities (see note Table 2), visual-spatial skills (Developmental Test of Visual Motor Integration, Beery, 1967; Benton Visual Retention, 1974; Benton Facial Recognition, 1983; Motor-Free Visual Perception Test, Colorusso & Hammill, 1972), audiological competence (audiological exam, brainstem auditory evoked responses), or

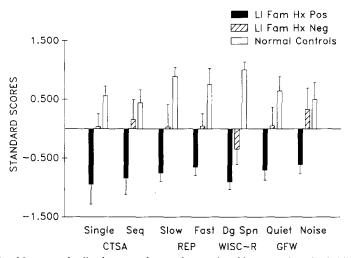


FIG. 2. Mean standardized scores of normal control and language-impaired children with a positive or negative family history of language impairment from tests of auditory processing/attention: Single and sequential auditory target accuracy, Computerized Tests of Selective Attention (Townsend & Tallal, 1989); Tallal Repetition Test (Tallal & Piercy, 1973) correct recall of sequences of high and low tones presented at long (slow) and short (fast) intervals; WISC-R Digit Span subtest (Wechsler, 1974) scaled score; and Goldman– Fristoe–Woodcock Test of Auditory Discrimination (1970) percentile ranks for discrimination of words in context of quiet or background noise.

speech/oral motor abilities (Oral Mechanism Exam, adapted from Dworkin & Culatta, 1980; Goldman–Fristoe Test of Articulation, 1986).

## FINAL DISCUSSION

Although the sample size in this study prevents development of statistical models of behavioral profiles, examination of this group is useful in a number of ways. Comparison of profiles from the smaller subset to those of the original sample allows us to see whether the major effects noted in Study 1 are observable with a group of children who clearly have persistent language impairments and have had the opportunity to progress further in academic achievement. Similarly, the comparison between parent and teacher reports of behavior can be made. It is important to note that a similar proportion of children in both Study 1 and 2 met criteria for a positive family history (approximately 70%). Thus, it is not the case that those with affected parents are more likely to be either more severely or more persistently language impaired.

In the light of current concerns about social-emotional disorders and language impairment, it should be noted that children from both parts of this study were from a cohort chosen more than 10 years ago and followed longitudinally. At that time the issue of coexistence of Attention Deficit

Hyperactivity Disorder (ADHD) with language disorders was not as central as it is today, and so children who were diagnosed as hyperactive or medicated were excluded from the original sample. Additionally, information that could be used to make a diagnosis based on DSM-III-R (1987) criteria was not collected for either probands or family history. The family history positive children in Study 2 do show elevated scores on the Achenbach hyperactivity scale, indicating that this is an issue which should be carefully examined in future studies.

Another issue to be considered is that of the relationship among SES, family history, and behavior. Although covariance analyses were used with these data to demonstrate that statistically significant effects in both studies were independent of SES, this is not the optimal approach to this problem. It is not possible from these data to determine causal links among these variables, but with much larger samples and replication, causal models can be constructed to define and clarify these associations.

In Study 1, subjects having a positive family history of language impairment showed lower SES, poorer academic performance, and were rated by their parents as having more behavior problems. In Study 2, subjects having a positive family history again showed a trend to lower SES and were rated by both their parents and their teachers as having more behavior problems, specifically in the areas of attention and academic achievement. Interestingly, empirical studies of nonverbal auditory attention, perception, and memory also demonstrated significant group differences between language-impaired children with and without positive family history. The positive group performed more poorly on these measures than the negative family group. Despite both parent and teacher reports of significantly more academic problems in the family history positive LI group, based on the Achenbach Child Behavior Checklist, empirical tests of academic achievement failed to show significant differences between groups.

One of the purposes of Study 2 was to assess phenotypic differences in older language-impaired children who clearly demonstrated a serious and persistent language impairment and who also have had more of an opportunity to acquire academic skills. The results of Study 2 demonstrate that when a group of language-impaired subjects who are more homogeneous in terms of the degree and duration of language deficit are assessed, attention/perceptual deficits consistently identify phenotypic differences between those with or without a positve family history. This was observed on empirical tests of both nonverbal auditory attention and perception, as well as on parent and teacher behavioral checklists. In both cases language-impaired children with positive family histories were found to perform more poorly than those with negative family histories in the area of perception and attention. The finding that behavior, particularly attentional, was rated to be more problematic for children with a positive

family history not only by parents but also by teachers demonstrates that these ratings are not the result of a biased perspective from parents who were themselves language/learning impaired.

The results from Study 2 further specify the nature of the behavioral differences observed in Study 1. These results indicate that only those aspects of social and emotional behavior which pertain to attention and perception are significantly different. As such, they fit closely with previous reports demonstrating specific nonverbal temporal processing deficits in language-impaired children (Tallal & Piercy, 1973; Tallal et al., 1985a,b). They also support our hypothesis which suggests that previously reported social and emotional problems in language-impaired children may reflect primarily neurodevelopmental (attention, perception, motor) disturbances (Tallal, Dukette & Curtiss, 1989). They further suggest that the profile of attentional and perceptual deficits, which has been reported consistently to characterize language-impaired children, occurs significantly more often in language-impaired children with a positive family history than without, and thus may have a familial or genetic basis.

Although a significant difference in academic achievement skills was found in Study 1 between LI children with or without a positive family history, academic achievement scores were not found to be significantly different in the language-impaired groups included in Study 2. This difference between studies may have occurred for several reasons. Although one of the intentions of Study 2 was to attempt to assess language-impaired children after they had had more of an opportunity to acquire academic skills, in fact, test performance of subjects included in Study 2 continued to demonstrate exceptionally low scores (below the 30th centile in all academic domains assessed). These data demonstrate that language-impaired children who continue to have serious and persistent deficits throughout their 9th and 10th year of life are also characterized by extremely severe deficits in academic achievement across domains. Because uniformly low scores were found to characterize this group of LI children, it is possible that there was not enough variance in the data to further differentiate between groups based on family history. It is of interest that although academic achievement scores did not prove significantly different between groups in Study 2, teachers rated the family history positive children as being significantly more inattentive and as doing poorer school work.

In sum, the results of these studies demonstrate significant phenotypic differences between language-impaired children with a positive or a negative family history of language disorder. Language-impaired children with a positive family history showed a profile consistent with attentional and perceptual deficits. Thus, LI children who cannot properly process basic auditory information, or who have inadequate auditory attentional capacity, appeared to their parents and teachers to be "confused," "inat-

tentive," or "unable to concentrate." As family history did not differentiate significantly between degree or pattern of language deficit, general intelligence, or other neuropsychological or cognitive profiles, these results suggest that genetic and/or familial factors might play a specific role in determining the degree of auditory, perceptual, and attentional deficits which have been suggested to underlie the linguistic disorder of LI children.

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