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Children's History of Speech-Language Difficulties: Genetic Influences and Associations With Reading-Related Measures

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Abstract

Purpose—This study examined (a) the extent of genetic and environmental influences on children's articulation and language difficulties and (b) the phenotypic associations between such difficulties and direct assessments of reading-related skills during early school-age years.

Method—Behavioral genetic analyses focused on parent-report data regarding the speech-language skills of 248 twin pairs ($M = 6.08$ years) from the Western Reserve Reading Project. In addition, phenotypic associations between children's speech-language status and direct assessments of early reading-related abilities were examined through hierarchical linear modeling (HLM).

Results—Probandwise concordance rates and intraclass tetrachoric correlations indicated high heritability for children's difficulties in expressive language and articulation, with estimates of .54 and .97 accordingly. HLM results indicated that children with histories of speech-language difficulties scored significantly lower than unaffected children on various measures of early reading-related abilities.

Conclusions—Results from the parent-report survey provided converging evidence of genetic effects on children's speech and language difficulties and suggest that children with a history of speech-language difficulties are at risk for lower performance on early reading-related measures. The extent of risk differed across measures and appeared greatest for children who demonstrated a history of difficulties across articulation, expressive language, and receptive language. Implications for future genetic research and clinical practice are discussed.

Keywords

elementary school pupils; articulation disorders; language disorders; written comprehension disorders

The purpose of the present study was to examine the etiology of speech-language difficulties and the phenotypic associations between speech-language difficulties and measures of early reading-related skills. In the past decade, clinicians' understanding of the genetic influences on speech and language difficulties has grown substantially from familial aggregation and behavioral genetic studies. Familial aggregation studies, which compare the prevalence rates of a communication disorder in families of affected individuals to the population rate at large, have revealed the tendency for articulation and language difficulties to run in families (Choudhury & Benasich, 2003; Felsenfeld, McGue, & Broen, 1995; Lewis, 1992; Lewis, Ekelman, & Aram, 1989; Rice, Haney, & Wexler, 1998; Tallal, Ross, & Curtiss, 1989; Tomblin, 1996). A summary of seven familial aggregation studies provided by Stromswold (1998) revealed a median incidence rate of 35% for language difficulties in the families of children with language impairment, compared with a median incidence rate of 11% in control families (p. 665).

One primary limitation of familial aggregation studies is that they confound genetic and environmental factors. In other words, genes "run in families" but so do many environmental factors, such as wealth, parental interaction style, and so forth. In contrast to familial aggregation studies, behavioral genetic designs are able to partial out genetic versus environmental effects by comparing the phenotypic similarity of family members with different levels of genetic relatedness. A summary of twin studies conducted by Stromswold (2001) concluded that there were substantial genetic effects on language difficulties, with 25%–100% of the variance due to genetic factors (p. 665; see also Plomin & Kovas, 2005). Converging evidence for genetic influences on speech-language disorders has also emerged from adoption studies (see Felsenfeld, 2002; Stromswold, 2001).

Fewer behavioral genetic studies have examined more than one type of communication difficulty within the same group of children. Bishop (2002) reported the extent of genetic effects on low language and articulation scores using a 10th percentile cutoff in a total of 22 dizygotic (DZ) and 57 monozygotic (MZ) twin pairs, age 7 to 13;11 years. Group heritability estimates for individual measures were .65 and .80 for expressive language, .48 and 1.23 for receptive language, and .97 for articulation (see also Bishop, North, & Donlan, 1995). A study by Kovas et al. (2005) examined the extent of genetic and environmental effects on different aspects of speech-language development and disability using 787 pairs of 4-year-old twins from the Twins Early Development Study. Probands were identified with scores that fell more than one standard deviation below the sample's mean. Results indicated substantial genetic effects on all expressive language measures ($a^2 = .51-.69$) except the one measuring expressive grammar, in which only environmental factors emerged as significant ($c^2 = .48$). Shared environment was also significant for low scores on a measure of receptive grammar (.56) and of articulation (.41), paired with no significant genetic effects (see also Byrne et al., 2005).

The present study adds to past behavioral genetic investigations of communication disability in at least three ways. First, this sizeable twin sample is independent from previously reported twin data on language disability, which have come from a limited number of large-scale twin projects (see Plomin & Kovas, 2005; Stromswold, 2001). Because estimates of genetic and environmental effects are influenced by the characteristics of a sample, in terms of severity of disability, socioeconomic status, and so forth, replication across independent samples is particularly important to determine the extent to which findings can be generalized. A second

strength of our study is the relatively restricted age range of the twins. Because estimates of heritability can differ based on age (see Plomin, Fulker, Corley, & DeFries, 1997), having a relatively narrow age group is important in deriving meaningful estimates for cross-study comparisons. The third strength of the present study is its ability to examine phenotypic associations between spoken communication disabilities and early reading-related measures. This latter point is critical in that previous studies have indicated that children with communication difficulties, particularly in the areas of language and articulation/phonology, are at risk for difficulties in reading (Bishop & Adams, 1990; Nathan, Stackhouse, Goulondris, & Snowling, 2004; Scarborough & Dobrich, 1990). Similarly, studies have found that children with reading difficulties are more apt to have spoken language difficulties (e.g., Larrivee & Catts, 1999; Nation, Clarke, Marshall, & Durand, 2004; Scarborough, 1990).

The literature on the relation between spoken language and reading disability has recently turned toward attempts to pinpoint the specific cognitive deficits, such as phonological processing, that may be contributing to the overlap and independence of these two disorders. A two-dimensional model of the relation between language and reading disability proposed by Bishop and Snowling (2004) suggests that phonological skills may be responsible for the overlap between disabilities in some cases, whereas in other cases the overlap is due to deficits in nonphonological language skills, such as difficulties in syntax and vocabulary skills.

A study by Catts, Adlof, Hogan, and Weismer (2005) examined the phenotypic overlap between reading and language disabilities, as well as the relation of these disabilities to phonological awareness, in kindergarten, second, fourth, and eighth grades. The authors confirmed previous findings that the prevalence of reading disability among children with specific language impairment was significantly higher than the expected base rate. However, the authors found that children with specific language impairment did not differ from same-age peers in terms of phonological awareness skills at later ages, whereas children with reading disabilities demonstrated longitudinal difficulties in this area. On the basis of this latter finding, Catts et al. concluded that phonological awareness is central to reading disability, but that specific language impairment is best characterized by deficits in other cognitive domains. Despite their conclusion, it is worth noting that children with specific language impairment did score lower in phonological awareness than did same-age peers at the kindergarten time point.

One of the many issues that makes studying the relation between speech-language and reading disabilities particularly challenging is the possibility that it changes over time (Chall, 1983; Snow, Burns, & Griffin, 1998). For example, phonological skills may contribute most to the overlap between reading and spoken language difficulties when children are first learning to read and rely most heavily on sounding out words. During later years, overlap between disabilities may be due largely to children's use of semantic and syntactic knowledge to facilitate reading comprehension, and vice versa. The present study examines the phenotypic associations between children's history of speech-language difficulties at an age when early reading skills are emerging and utilizes a variety of reading-related measures. If phonological skills are responsible in part for the overlap between speech-language disability and early reading difficulties, clinicians would expect children with a history of speech-language difficulties to perform more poorly on reading-related measures that tap phonological skills. Such phenotypic findings will help guide future multivariate genetic analyses, which provide the most definitive test of causal overlap but require a larger sampling of children with speech-language difficulties than our study is currently able to provide.

In addition to examining the role of phonological abilities in the phenotypic overlap between reading and language disability, the present study examines the extent to which children with histories of different types of speech-language disabilities are at risk for early reading-related

difficulties. A longitudinal study by Nathan et al. (2004) contrasted literacy outcomes for three groups of 19 school-age children: (a) children with speech difficulties, (b) children with speech and language difficulties, and (c) children with typical speech-language development. Results suggested that at age 6;9, children with speech and language difficulties were at increased risk of difficulties in both reading and spelling, more so than were children with speech difficulties only (see also Bishop & Adams, 1990). Similarly, Catts (1993) administered reading-related measures to 56 children with speech-language impairments and found that children with language impairment scored lower than controls on measures of word identification and word attack in first and second grades. In contrast, the children with articulation as their primary impairment did not score lower than the control group. From a genetic standpoint, familial aggregation studies (Flax et al., 2003; Lewis, 1992; Lewis, Ekelman, & Aram, 1989) and twin studies (Bishop, 2001; Hohnen & Stevenson, 1999; Samuelsson et al., 2005) have found evidence of shared genetic influences on individual differences and disabilities in articulation, spoken language, and reading. In summary, prior literature suggests that evidence of articulation or language disability may be a risk factor for early reading-related skills, with language disability and comorbid communication disabilities seeming to pose the greatest risk.

The present study uses a genetically sensitive design to examine the genetic and environmental influences on spoken communication disabilities and examines phenotypic associations between reported difficulties in spoken communication and children's early reading-related skills. The specific research questions are as follows:

1. What is the magnitude of genetic and environmental effects on parents' report of children's speech-language difficulties?
2. What is the phenotypic relation between children's history of communication difficulties, as reported by parents, and examiner-administered measures of reading-related skills during early school age?

On the basis of prior research, we predicted that a history of articulation and language difficulties would be highly heritable, and that difficulties in speech language would be associated with lower reading abilities during early school-age years. We also predicted that children with a history of speech-language difficulties would be more apt to have difficulty with reading-related tasks that relied heavily on phonological skills. Understanding the extent of genetic influences on communication disabilities and how such influences might overlap with reading is critical in shaping intervention. In addition, because of the potential complexity of genetic influences on speech-language disabilities, behavioral genetic designs, such as this one, become increasingly important in honing the efficiency of molecular research (Felsenfeld, 2002; Martin, Boomsma, & Machin, 1997).

Method

Participants

The present study includes 248 same-sex twin pairs (107 MZ pairs, 40% boys and 141 DZ pairs, 43% boys) from the Western Reserve Reading Project (WRRP), a longitudinal population-based twin project of reading, math, and associated cognitive abilities. Twins were recruited from throughout the state of Ohio, with the majority of participants living in the greater Cleveland, Columbus, and Cincinnati metropolitan areas. The WRRP is a longitudinal project with four home visits conducted within a 3-year period. The present study focused on data obtained at the initiation of the study when children had entered kindergarten, but not yet completed first grade. Of the 352 twin pairs recruited to the WRRP at the time of this study, 248 pairs had complete data to be used in the present analyses. The resulting sample of 248 pairs had a mean age of 6.08 years ($SD = 0.69$), with a mean general cognitive score of 101.42 ($SD = 12.94$) as assessed by the Stanford-Binet Intelligence Test (Thorndike, Hagen, & Sattler,

1986). All participating families were fluent in English. Zygosity of the twins was determined by DNA testing via buccal swabs. The handful of parents who did not consent to this procedure were asked to complete a measure of twin physical similarity, which is 95% accurate in relation to zygosity as determined by DNA markers (Goldsmith, 1991; Price et al., 2000). Two families were excluded from the study: one because the twins had disabilities that prevented them from completing the assessment protocol and one because the primary caregiver was not fluent enough in English to participate fully.

Families were recruited to be representative of the larger U.S. population in terms of gender, socioeconomic status, and ethnic composition. Based on caregiver self-report of race/ethnicity, the present sample is approximately 92% White/European, 5% African American, 2% Asian American, 1% Hispanic, and 1% other. In terms of highest level of education attained by the child's primary caregiver, less than 1% of the sample reported no high school diploma, 8% reported having attained a high school diploma or equivalent, 16% some college, 11% completion of a 2-year college degree, 33% 4 years of college, 7% some graduate or professional school, 19% completion of graduate or professional school, and 6% other. In comparison to Census 2000 (U.S. Census Bureau, 2000) data for the state of Ohio, our twin sample underrepresents African Americans and overrepresents White/European families, but is fairly consistent with Ohio census data for Latinos and Asians. Although educational information from WRRP is not directly comparable to census data, our sample appears to overrepresent more highly educated individuals.

Procedures

The initial recruitment of participating families was coordinated through the Pennsylvania State University and included school nominations, Ohio state birth records, Mothers of Twins Clubs, and media advertisement. In regard to school nominations, schools were asked to send a packet of information to parents in their school systems with twins who had entered kindergarten, but not yet completed first grade. After participating families were identified, each family was mailed (a) a brief questionnaire assessing demographic and pre- and perinatal information, (b) a questionnaire of home environment and parental attitudes, and (c) the Speech-Language Survey that serves as the focus of this study.

Once the questionnaires were returned, families of participants were contacted through a subcontract site at the Case Western Reserve University to schedule an initial home visit. Each home visit was conducted by two examiners that had been trained extensively on the assessment protocol and had significant experience with children in the target age range. The home visit included assessments was used to assess reading-related cognitive skills, reading outcomes, and more general cognitive skills in both children and parents. Additionally, the home visit assessed parent-child and sibling-sibling interaction as well as indices of the home environment. Also at the initial home visit, examiners obtained DNA for each twin using cheek swabs (see Price et al., 2000). All DNA was sent to the Center for Developmental and Health Genetics at Pennsylvania State University for analysis.

Measures

Speech-Language Survey—The Speech-Language Survey, designed specifically for the WRRP based on case history forms used in a number of different clinical settings, consists of a series of yes–no questions related to different aspects of communication development. The four questions used for the present study are listed in Table 1. Primary caregivers responded to questionnaire items by selecting from the following three options: (a) Yes, the first born; (b) Yes, the second born; and (c) No, neither twin has had this difficulty. A flowchart of the follow-up questions is provided in Figure 1. If the caregiver selected yes for either twin, he or she was asked to provide additional follow-up information for that particular item. Taking the

question regarding expressive language as an example, caregivers were asked to provide (a) the age of the child when they first became concerned, (b) the age at which the twin's difficulties seemed to resolve, and (c) whether that twin still had difficulties in this area. The question regarding hearing was asked separately for each twin. All of the survey items were coded in a trimodal fashion with 0 (*representing a negative response*), 1 (*representing a history of resolved difficulties*), and 2 (*representing unresolved difficulties at the time the survey was completed*).

Although identification of children's communication difficulties through individual parental survey items should be viewed as a limited form of assessment, we included this measure based on three forms of support. First, parent-report measures of child development have demonstrated high agreement with professional assessments (e.g., Dinnebeil & Rule, 1994), including parent-report measures of child language (e.g., Oliver et al., 2002). Second, previously published twin studies have used various forms of parent and teacher reports (Spinath, Price, Dale, & Plomin, 2004; Tomblin & Buckwalter, 1994), which have demonstrated convergence with studies using other forms of assessment (Plomin & Kovas, 2005; Stromswold, 2001). Last, by capitalizing on parental perceptions and focusing on children's history of difficulties, the speech-language questionnaire offers inherent social validity and is likely to provide a relatively stable construct. Parents know their children well and have observed them across time and multiple contexts. To directly examine the validity of our survey data, we grouped children according to their history of expressive language difficulties and examined their performance on direct assessments of expressive language.

Reading-related measures—Given the age of our participants, measures focused on the development of early literacy skills, rather than on fluency or comprehension of reading connected text. Three subtests were administered from the Woodcock Reading Mastery Test (Woodcock, 1987) and considered independently in analyses: (a) Letter Identification, in which children were asked to identify, either by name or sound, printed letters; (b) Word Identification, in which children were asked to identify English words; and (c) Word Attack, in which children were asked to read nonsense words or words with very low frequency in the English language. The construct of rapid automatized naming was assessed through the average of the letter-naming and number-naming tasks from the Comprehensive Test of Phonological Processing (Torgeson, Wagner, & Rashotte, 1999), which required rapid naming of letters and digits accordingly. Because of the age of the sample and a need to remain consistent with published studies using the WRRP data (Petrill, Deater-Deckard, Thompson, DeThorne, & Schatschneider, in press), the final composite for rapid automatized naming was corrected for letter identification abilities. Skills in phonological awareness were assessed using the Phonological Awareness Test (Robertson & Salter, 1997), which included the following tasks: rhyming, initial phoneme isolation, whole-word phonemic segmentation, and phonemic deletion (both at the phoneme and syllable levels).

Additional measures—The Stanford–Binet Intelligence Test (Thorndike et al., 1986) was used as a covariate in the reading analyses. It included the following subtests: Vocabulary, Pattern Analysis, Memory for Sentences, Memory for Digits, and Quantitative. Two formalized assessments of vocabulary were included in the present study as a check on the validity of the Speech-Language Survey: the Boston Naming Test (Goodglass & Kaplan, 2001), which required children to name a series of pictured items, and the Vocabulary subtest from the Stanford–Binet Intelligence Scale, which required children to define words.

Analyses

Descriptive information and group differences on the formalized vocabulary measures are presented to address the validity of our measures. In addition, we addressed the initial study

question regarding the extent of genetic and environmental influences on children's speech-language history through probandwise concordance rates, intraclass correlations, and structural equation model fitting procedures (Neale, Boker, Xie, & Maes, 2003). This latter approach estimated genetic (h^2), shared environmental (c^2), and nonshared environmental (e^2) sources of variance in language difficulties and provided inferential statistics based on differences in genetic similarity between MZ and DZ twins. Heritability is significant when MZ twins appear more similar to each other than do DZ twins. The similarity between MZ twins that cannot be accounted for by genetics provides an estimate of shared environment ($MZr - h^2$). Nonshared environment, in conjunction with error, and is estimated by the extent to which MZ twins appear different from one another ($1 - MZr$).

The second research question regarding phenotypic associations between speech-language and reading was addressed through a series of HLMs in which the mean reading-related scores from five groups were compared: (a) children with a history of articulation difficulties only; (b) children with a history of expressive language difficulties only; (c) children with a history of both articulation and expressive language difficulties; (d) children with a history of difficulties in articulation, expressive language, and receptive language; and (e) peers with no history of articulation or language difficulties (i.e., unaffected). HLM was used rather than analysis of variance because of the nested issues inherent in twin designs. Bivariate genetic analyses of spoken language and reading were not conducted because of insufficient power associated with the relatively limited number of children with histories of speech-language difficulties.

Results

Descriptives

A summary of the prevalence and comorbidity rates for a history of difficulties in articulation, expressive language, and receptive language abilities based on the Speech-Language Survey is presented in Figure 2. Of the 141 children with histories of articulation difficulties, 64 (45%) were considered resolved and 77 (55%) were considered persistent at the age the survey was completed. Similarly, of the 94 children with expressive language difficulty, 35 (37%) were considered resolved and 59 (63%) were considered persistent. In regard to the 19 children with histories of receptive language difficulties, 5 (26%) were considered recovered and 14 (74%) were considered persistent. Note in Figure 2, and throughout the remainder of the analyses, both the resolved and persistent difficulties were combined and considered as affected. The primary rationale for collapsing these two groups was our interest in examining causal influences on children's history of speech-language difficulties, regardless of whether the difficulties were perceived as resolved or persistent at the time the survey was completed.

Age-corrected raw scores were derived for all dependent reading-related variables and saved as standardized residuals. A principal-factor analysis on tasks from the Phonological Awareness Test (Robertson & Salter, 1997) revealed one primary factor (eigenvalue = 3.45) that accounted for 57.47% of the variance. Consequently, the phonological awareness tasks were collapsed into a single composite (see also Schatschneider, Francis, Foorman, Fletcher, & Mehta, 1999). A distribution of the phonological awareness composite and all other dependent reading-related variables is presented in Figure 3. Note that all variables are unimodal, although distributions are somewhat skewed for Word Identification and Word Attack. We expected these tasks to be relatively difficult for children in kindergarten as they represent early reading skills, such as recognizing whole words and sounding out unfamiliar words, as opposed to more precursor skills, such as phonological awareness and letter identification.

The Boston Naming Test (Goodglass & Kaplan, 2001) and the Vocabulary subtest from the Stanford–Binet (Thorndike et al., 1986) were not utilized in the present study as reading-related skills, per se, but as formalized measures of language in order to assess the validity of our Speech-Language Survey. As such, two HLMs were run for the Boston Naming Test and the Vocabulary subtest, respectively, using history of expressive language difficulties as the independent variable (two levels: affected vs. unaffected). History of expressive language was chosen from the survey items as the most relevant independent variable because of the nature of both the Boston Naming Test and the Vocabulary subtest as expressive language measures. Significant mean differences emerged for the Boston Naming Test, $t(212) = -3.81, p = .0002$, with means of 0.10 ($SD = 0.97$) and -0.53 ($SD = 0.98$) favoring the group with no history of expressive language difficulties. Similarly, group means from the Stanford–Binet Vocabulary subtest differed as well, $t(213) = -2.21, p = .03$, with a mean of 0.06 ($SD = 1.01$) for children with no history of expressive language difficulties and a mean of -0.33 ($SD = 0.86$) for children with a history of expressive language difficulties.

Behavioral Genetic Analyses

Probandwise concordance rates and twin tetrachoric correlations were calculated for MZ and DZ twins, respectively, for relevant items from the Speech-Language Survey. Probandwise concordance consisted of the total number of probands in which both twins demonstrated a history of speech-language difficulty divided by the total number of affected probands with the same type of speech-language difficulty. In addition, ACE modeling was used on the entire twin sample to estimate heritability (h^2), shared environmental effects (c^2), and non-shared environmental (e^2) sources of variance. Note that for all analyses probands were identified as children who either had past or present difficulties in the area of interest. Resulting proband concordances for both MZ and DZ groups are presented in Table 2 for each type of speech-language difficulty. Note that MZ concordance rates appear higher than DZ concordance rates for all items, a finding suggestive of genetic influence.

Tetrachoric correlations and ACE modeling were also used to estimate genetic and environmental influences on reported histories of communication difficulties. Tetrachoric correlations were used to accommodate the categorical nature of the variables and to account for population incidence rates (Neale et al., 2003). Based on the assumption of an underlying normal distribution, tetrachoric correlations utilize a particular threshold to define an affected group. When one twin is affected, the probability of a cotwin being affected is a function of the population risk and the tetrachoric correlations between twins (Sham et al., 1994). Correlation and modeling results are presented in Table 3. Heritability estimates emerged as statistically significant for articulation and expressive language. Shared environmental influences did not emerge as statistically significant for any item. Because the DZ correlation for articulation was slightly less than half of the MZ correlation, we examined the possibility of genetic dominance by using an average derivative estimation (ADE) model. The ADE model did not yield a significant effect for dominance ($d^2 = .38, p > .05$).

Phenotypic Associations Between Communication Difficulties and Reading

The second research question regarding the phenotypic relation between children's speech-language skills and reading-related measures was addressed through two HLMs with five groups each (Raudenbush & Bryk, 2002): (a) children with histories of articulation difficulties only; (b) children with histories of expressive language difficulties only; (c) children with histories of both articulation and expressive language difficulties; (d) children with histories of articulation, expressive language, and receptive language difficulties; and (e) peers without a history of articulation or language difficulties. Individual a priori contrasts were formed comparing each of the affected groups to the unaffected comparison group. The rationale for forming these groups was to tease apart the potentially independent effects of isolated speech-

language difficulties, to the extent sample sizes allowed (see Bishop et al., 1995). For example, the grouping allowed us to address whether single impairments such as articulation or expressive language difficulties were associated with group differences in reading-related measures. There were not enough children with a history of isolated receptive language difficulties in our sample to view this group independently. Table 4 provides group means for the reading-related measures, with significant differences based on contrasts from the HLMs. Note that rapid automatized naming differs from other measures in Table 4, in that higher scores are indicative of slower, and consequently “poorer,” performances. Also, IQ is presented in terms of standardized scores, rather than as standardized residuals, so that groups can be additionally compared with the test’s normative mean of 100 and standard deviation of 15 (Thorndike et al., 1986).

HLM results indicated that children with histories of articulation difficulties differed significantly from the unaffected group in phonological awareness, word attack, and word identification. Children with histories of both articulation and expressive language difficulties differed on the same three reading-related measures, plus IQ. Children with histories of isolated expressive language difficulties differed from the unaffected group on letter identification and word identification. Children with histories of all three speech-language difficulties scored significantly lower than the unaffected group on phonological awareness, letter identification, rapid automatized naming, and IQ. It is important to note here that statistical power differed across comparisons, depending on the number of children in any particular group. There were 65 children with a history of isolated articulation difficulties, 57 with histories of both articulation and expressive language difficulties, 21 children with histories of isolated expressive language difficulties, and 13 children with histories of all three speech-language difficulties. Given differences in group size, the same mean difference could emerge as statistically significant in one group, but not in another. For example, note that children with histories of all three communication difficulties scored nearly half a standard deviation lower than the unaffected group on word attack and word identification, but such differences did not emerge as statistically significant even though lesser mean differences emerged as significant for other larger groups.

Because some significant group differences in IQ emerged, all HLMs were rerun with IQ regressed out of the reading-related measures in order to address the possibility that group differences in reading-related measures were related to general cognition rather than speech-language status. Results were largely similar, with the only changes being that the group means on word identification were no longer significantly different between the unaffected group and the group with expressive language difficulties or between the unaffected group and the children with both articulation and expressive language difficulties.

Discussion

Two primary findings emerged from the present study. First, although confidence intervals were large, reported difficulties in children’s expressive language and articulation abilities were heritable, without significant evidence of shared environmental effects. Second, early school-age twins with histories of speech-language difficulties tended to score lower on reading-related measures. Before considering the details and implications of these findings, we first address validity evidence for the Speech-Language Survey and potential concerns regarding the generalization of findings from our twin sample to the nontwin population.

Validity Evidence

One potential limitation of the present study is the identification of children’s speech-language difficulties based on single items from a parental survey. Evidence for the validity of our survey data comes from congruence between the survey data and results of examiner-administered

measures of language. Specifically, children identified with a history of expressive language difficulties based on the survey scored significantly lower on both the Boston Naming Test (Goodglass & Kaplan, 2001) and the Vocabulary subtest of the Stanford–Binet (Thorndike et al., 1986) than did the children identified without such a history. Differences between group means equaled two thirds of a standard deviation in the case of the Boston Naming Test and one third of a standard deviation for the Vocabulary subtest. Group differences are striking given the different forms of assessment and the fact that the survey item was more global, referring to both vocabulary and grammatical skills, than the formalized measures that focused on expressive vocabulary only.

Generalization

A second potential concern surrounding the present study is the extent to which results from twins can be generalized to singletons. In the general population, the reported prevalence rates, based predominantly on direct assessment, have ranged from 2% to 11% for articulation difficulties (Shriberg, Tomblin, & McSweeny, 1999) and from 7% to 10% for language disability (Leonard, 1998; Reed, 1986; Tomblin et al., 1997). Prevalence rates vary based on age and specificity of the impairment. In the present study, the prevalence of persisting difficulties was 16% for articulation, 12% for expressive language, and 3% for receptive language. Consequently, in comparison to the singleton literature, rates of speech-language difficulties in the present twin sample appeared somewhat higher.

Higher rates of speech-language difficulties in twins are likely due to higher rates of the same risk factors that contribute to speech-language difficulties in singletons, specifically biological factors, such as prematurity, lower birth weight, and so forth (Field, Dempsey, & Shuman, 1981; Luke & Keith, 1992), and social factors, such as less individualized parental interaction (e.g., Conway, Lytton, & Pysh, 1980; Thorpe, Rutter, & Greenwood, 2003). In part because of these risk factors, twins often receive more frequent professional vigilance, which may in turn make communication difficulties more likely to be identified than in singleton children. In summary, there is evidence to suggest that twins are at increased risk for communication difficulties, but little evidence suggesting that the communication skills of twins is impacted by different factors than the communication skills of singletons. Consequently, generalizing our findings to singleton communication development seems appropriate.

Genetic Effects

Given support for internal and external validity, we now turn to how our estimates of genetic and environmental effects compared with other twin studies of speech-language difficulties. Reported estimates of group heritability for language difficulty have ranged from .25 to 1.00 (Stromswold, 2001; see also Plomin & Kovas, 2005), but most studies report moderate genetic effects that are relatively comparable to the estimate of .54 obtained here. For example, Bishop (2002) reported group heritability estimates of .65 and .80 for low scores in word finding and recalling sentences, whereas Kovas et al. (2005) reported estimates of .51 and .69 for low scores on expressive semantic measures. Of interest, Kovas et al. found no significant genetic effect on a measure of expressive grammar. Spinath et al. (2004) found heritability estimates ranging from .47 to .49 for a verbal aggregate of expressive vocabulary and grammar derived from parent report across 3 years. Like Spinath et al., we did not tease apart expressive difficulties in grammar versus vocabulary. Consequently, it is possible that the obtained heritability estimate is driven largely by difficulties in one domain or the other.

In regard to receptive language, the present study resulted in a high group heritability estimate of .87 that did not reach statistical significance, perhaps because of power limitations. Note that only 19 twins from our sample were identified with receptive language difficulties, as opposed to 144 and 94 children with articulation and expressive language difficulties,

respectively. An additional complication, highlighted by the present study, is the high overlap between difficulty in receptive language and other domains, such as articulation and expressive language. In our sample, only 2 out of 19 children with receptive language difficulties did not have reported difficulties in one of these other two domains. Consequently, it is particularly difficult to estimate genetic influences on receptive language difficulties independent of comorbid speech-language difficulties.

The heritability estimate of .95 for articulation difficulties in the present study is highly consistent with the estimate of .97 reported by Bishop (2002), who used a direct articulation measure of percent consonants correct in children age 7–14 years. In contrast, Kovas et al. (2005) revealed insignificant heritability for articulation difficulties paired with significant shared environmental effects, using a standardized articulation test with 4-year-old twins. Differences across studies may be driven in part by the different measures or differences in child age. Although speculative at this point, there is some evidence to suggest that heritability measures increase as children get older because of increased influence over their own environments (Plomin, DeFries, & Loehlin, 1977; Plomin et al., 1997; Scarr & McCartney, 1983). In addition to age, our estimates of substantial heritability for both articulation and expressive language difficulties may be influenced in part by two relevant factors: severity and form of measurement.

Severity—In regard to severity, previous research has suggested that the magnitude of genetic effects on language disability may increase as a function of the severity of the difficulties (e.g., DeThorne, Petrill, Hayiou-Thomas, & Plomin, 2005; Spinath et al., 2004; Viding, Spinath, Price, Bishop, Dale, & Plomin, 2003). Although the present study does not offer a direct measure of severity, one might expect that the more severe the children's difficulties, the more likely they are to elicit and sustain parental concern. Consequently, the children in the present study who have a positive history of parental concern in the areas of articulation or language may be the children with more severe or more consistent difficulties, and consequently demonstrate higher heritability and lower shared environmental effects. It is important to note that although shared environmental effects did not reach statistical significance in the present sample, the relatively high DZ correlations paired with relatively high estimates of shared environmental effects (c^2) for expressive language are indicative of such influences.

Form of measurement—The second potential factor that may have influenced our estimates of genetic and environmental effects is form of measurement. Specifically, one concern with parent-report measures is the possibility that parents may overestimate MZ similarity and underestimate DZ similarity, thereby leading to inflated estimates of heritability. This effect is suspected when the DZ intraclass correlation is close to zero and substantially less than the MZ intraclass correlation. Although we cannot determine the exact extent to which such contrast effects may be influencing the present results (Bishop, Laws, Adams, & Norbury, in press), evidence from other studies that used direct examination is generally supportive of our results. For example, intraclass correlations for articulation difficulties in our sample appeared most indicative of possible contrast effects, yet the group heritability estimate is highly consistent with results from Bishop (2002), who used direct assessment.

Associations With Reading

Also consistent with prior literature (e.g., Nathan et al., 2004; Scarborough & Dobrich, 1990), the present study indicated that children with histories of speech-language difficulties tended to score lower on early reading-related measures. Three findings in regard to the nature of this relation are worth highlighting. First, group differences emerged most consistently for phonological awareness and least consistently for rapid automatized naming, thereby suggesting that phonological skills may be responsible in part for the overlap between spoken

language and reading disabilities, at least at the stage of emergent literacy (see Bishop & Snowling, 2004). All groups of children with a history of speech-language difficulties, except expressive language only, scored significantly lower than the unaffected group on phonological awareness, with mean differences ranging from one third to more than one and one half standard deviations. The mean difference for the expressive language group exceeded one third of a standard deviation, but did not reach statistical significance.

Significant group differences in word attack, letter identification, and word identification were variable, reflecting perhaps the emergent nature of these skills. In other words, low scores on these measures at this age may not be indicative of present or future difficulties in reading but rather developmental variation. In addition, the variability may be influenced by the fact that these measures are less directly tied to phonological skills. For example, letter identification focuses on naming letters presented orthographically, although the letter's most common sound association is an acceptable response. Of interest, group differences in rapid automatized naming emerged only for children with histories of difficulty in all three speech-language domains. Rapid automatized naming performance is likely influenced by cognitive processes other than phonological skill, such as visual skill, speed of processing, and so forth. It is possible that overlap between rapid automatized naming and the presence of multiple communication difficulties is indicative of a more generalized learning deficit. Supportive of this idea is the fact that children with multiple speech-language difficulties also scored close to one standard deviation below the mean on the Stanford–Binet composite (Thorndike et al., 1986).

The second finding in relation to history of speech-language difficulties and early reading-related skills is that a history of communication difficulties across all three domains served as a particularly notable risk factor for difficulties with early reading-related measures. Specifically, the extent of mean differences in the reading-related measures appeared larger for children in this group, reaching more than one and one half standard deviations in the case of phonological awareness and letter identification. This degree of group difference suggests a clinical level of difficulty in these areas. The context of the present study does not allow us to tease apart whether such increased risk was due to group differences in severity of speech-language difficulties or the added presence of receptive language difficulties. It is possible that comorbid cases are also the cases with the most severe communication difficulties, or that it is the addition of receptive language difficulties that creates the greatest risk. Regardless of the reason, our results are relatively consistent with past literature suggesting that children with concomitant speech and language impairment are at greatest risk for reading difficulties (Bishop & Adams, 1990; Nathan et al., 2004). However, unlike Catts (1993), our findings do suggest that children with histories of isolated articulation difficulties are at an elevated risk of lower scores on early reading-related measures. It may be that differences in the severity of the articulation difficulties or the underlying cause of the articulation difficulties are responsible for such cross-study differences.

The last point in regard to the relation between speech-language difficulties and early reading-related tasks is that the observed group differences were less robust but largely unchanged after the effects of IQ were regressed out of the reading-related measures. This finding is particularly striking given that the IQ composite was not limited to nonverbal subtests. Consequently, it appears that children with histories of speech-language difficulties are not performing lower on early reading-related tasks due solely to lower general cognitive abilities.

Implications

Findings from this study offer implications both for future research and clinical practice. In regard to future genetic investigations, findings support the use of children's history of speech-language difficulty as a phenotype of interest for behavioral and molecular analyses. It is easier to identify specific genetic effects and loci if the phenotype of interest is highly heritable, which

was the case in the present study, particularly for history of articulation difficulties. In addition, reports of children's history are relatively easy to elicit and allow for larger numbers of participants than other more extensive assessments. Ease of assessment is critical given the large number of participants needed to identify the relatively small effect of any one particular gene that may contribute to complex behavioral traits such as speech and language (see Martin, Boomsma, & Machin, 1997, p. 389).

A second implication of present findings for future genetic research is to focus on multivariate genetic analyses across disabilities in articulation, spoken language, and reading-related measures. Multivariate genetic analyses provide estimates of genetic correlation, which represents the extent of genetic overlap between traits regardless of their respective heritabilities. In other words, two traits with high heritabilities could demonstrate low genetic overlap, or two traits with relatively low heritability estimates could demonstrate high genetic overlap. Limitations in sample size prevented us from conducting multivariate analyses in the present study, but the resulting evidence of phenotypic overlap across speech-language disabilities and between speech-language disabilities and reading-related measures warrants such analyses. In addition to the genetic analyses presented here on data from the Speech-Language Survey, previous studies from WRRP have found significant additive genetic effects for our reading-related measures (Petrill, Deater-Deckard, Thompson, DeThorne, & Schatschneider, 2006; see also Petrill et al., in press), thereby setting the stage for multivariate work in direct assessments of language and reading that are being collected in the second wave of the longitudinal project.

In terms of clinical practice, we offer three direct implications. First, evidence of genetic effects on articulation and expressive language difficulties highlights the importance of collecting family history information to help guide recommendations. For example, imagine a child whose assessment results are borderline. In such case, a family history of speech-language difficulties could tip the scale toward more immediate and intensive intervention. Similarly, caregivers might be advised to monitor more closely the speech-language development of children whose family histories put them at risk of difficulties in this area. Second, evidence that children with a history of speech-language difficulties are at risk in early reading suggests that caregivers and clinicians should make a point to facilitate and monitor preliteracy and literacy development in such children, particularly if their speech-language difficulties are more severe or include receptive deficits. Facilitation of phonological abilities may be particularly important (e.g., McFadden, 1998). Third, it is appropriate to counsel caregivers about genetic influences on speech-language difficulties in a way that may help reduce the parental guilt that can be associated with the diagnosis of childhood speech-language difficulties. Although we are not yet at a point where clinicians can state with certainty the presence of genetic effects in any particular case, informing parents of the potential of genetic influence is warranted. On that note, it is important to highlight that evidence of significant heritability does not relegate the environment to a secondary role. In other words, traits influenced by genetics are not unchangeable. On the contrary, identifying and understanding the mechanism through which genes influence behavior will allow clinicians to fashion the most effective therapeutic and preventative efforts.

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Figure 1.
Flowchart depicting the follow-up questions for Item 1 on the Speech-Language Survey.

Figure 2.

Diagram of prevalence rates and comorbidity for difficulties in articulation, expressive (Exp.) language, and receptive (Recept.) language. Numbers in the diagram represent number of twins. Note that the number of cases within the diagram for articulation and expressive language difficulties do not add up to their respective totals because of missing data in one or more of the comorbid areas.

Figure 3.

The distribution of age-corrected raw scores for all dependent variables used in the hierarchical linear modeling: (a) Letter Identification, (b) Word Identification, (c) Word Attack, (d) Phonological Awareness, (e) Rapid Automatized Naming, (f) Boston Naming Test, and (g) the Vocabulary subtest from the Stanford-Binet Intelligence Scale. Each variable has been graphed using the same scale. The x-axis represents standardized residuals ranging from -4 to 4, and the y-axis represents frequency of individuals, with increments of 20.

Table 1

Items from the Speech-Language Survey.

Survey question	Aspect of communication development
1. Has either of your twins ever had difficulty learning words (vocabulary), forming sentences (using correct grammar), or both?	Expressive language
2. Has either of your twins ever had a hard time understanding what is said to him or her (e.g., understanding age-appropriate directions)?	Receptive language
3. Has it ever seemed that either of your twins pronounces words poorly for his or her age (has trouble making particular speech sounds)?	Articulation
4. Has your first/second born twin ever had hearing problems (hearing loss)?	Hearing

Table 2

Probandwise concordance rates for MZ/DZ twins for parent report of difficulty on individual Speech-Language Survey items.

Parent report of children's difficulty	MZ	MZ	DZ	DZ
Expressive language	40/45	89%	26/49	53%
Receptive language	6/9	67%	2/10	20%
Articulation	54/63	86%	34/77	44%

Note. MZ = monozygotic; DZ = dizygotic.

Table 3
Intraclass tetrachoric correlations and estimates of heritability (h^2), shared environment (c^2), and nonshared environmental (e^2) effects based on average derivative estimation modeling.

Parent report of children's difficulty	MZr	DZr	h^2	c^2	e^2	$\chi^2(3, N = 494)$	ak
Expressive language	.98*	.69*	.58* (.23-.99)	.40 (.00-.74)	.02* (.003-.09)	4.79	-1.22
Receptive language	.92*	.48	.87 (.00-.99)	.05 (.00-.86)	.08* (.01-.41)	4.25	-1.75
Articulation	.95*	.38*	.95* (.62-.99)	.00 (.00-.31)	.05* (.01-.15)	3.84	-2.16

Note. Estimates are presented with their 95% confidence interval in parentheses; *ak* = Akaike's information criterion.

* $p = .05$.

Table 4

Group means and standard deviations on measures of reading-related skills based on history of articulation difficulties, expressive language difficulties, and comorbid speech-language difficulties.

Measure	Unaffected ^e	Articulation ^e	Expressive language ^e	Articulation and expressive language ^e	Articulation, expressive, and receptive language ^e
PA ^a	0.17(0.95)	-0.13* (1.01)	-0.26(0.85)	-0.38* (0.96)	-1.45* (0.86)
WA ^b	0.11 (1.07)	-0.22* (0.74)	-0.08 (0.89)	-0.27* (0.88)	-0.46 (0.64)
UD ^b	0.12(0.90)	-0.08(1.18)	-0.67* (1.28)	-0.02 (.76)	-1.68* (1.31)
WID ^b	0.07(1.06)	-0.14* (0.84)	-0.12* (0.78)	-0.09* (0.95)	-0.40 (0.48)
RAN ^c	-.03 (0.94)	-.08(1.07)	-.15(0.67)	.21 (1.12)	1.22* (2.30)
IQ ^d	102.99(12.95)	101.74(12.42)	94.82(12.34)	96.73* (11.53)	88.60* (7.56)

^aPhonological awareness (PA) assessed via the Phonological Awareness Test (Robertson & Salter, 1997).

^bWord Attack (WA), Letter Identification (LID), and Word Identification (WID) subtests from the Woodcock Reading Mastery Test (Woodcock, 1987).

^cRapid automatized naming (RAN) assessed via the Comprehensive Test of Phonological Processing (Torgesen, Wagner, & Rashotte, 1999).

^dComposite score from the Stanford-Binet Intelligence Test (Thorndike, Hagen, & Sattler, 1986); normative $M = 100$, normative $SD = 15$.

^eHistory of speech-language difficulties based on the Speech-Language Survey.

* $p = .05$ (significant difference compared with the unaffected group).