

A Behavioral-Genetic Analysis of Reading Disabilities  
and Component Processes

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**I. Introduction**

The genetic and environmental etiology of reading disabilities and related cognitive deficits is being studied through the comparison of identical and fraternal twins in Colorado (DeFries, Filipek, Fulker, Olson, Pennington, Smith, & Wise, 1997). Earlier analyses based on a relatively small number of twins established the presence of a significant genetic etiology for the group deficit in a composite measure consisting of reading comprehension, word recognition, and spelling (DeFries, Fulker, & Labuda, 1987), and in related skills in phoneme awareness and phonological decoding (Olson, Wise, Conners, Rack, & Fulker, 1989). Now the twin sample has increased in size so we can begin to address more complex and interesting questions about bivariate and differential genetic etiology. We have begun to explore the genetic and environmental covariation between different reading and language skills, as well as their independent genetic and environmental influences. We have also begun to explore potentially important subtype differences in genetic etiology within the reading disabled population. Some results from these preliminary analyses are reviewed in this chapter.

We will focus primarily on deficits in printed word recognition, which is usually the core deficit in normally intelligent children with reading disability (Stanovich, 1986). We will also consider several related component skills in reading and language. In the following section (II), we review the results of recent behavioral-genetic analyses on group deficits in these skills. In section III, we review bivariate behavioral-genetic

analyses to assess both common and independent genetic influences on orthographic and phonological skills in word recognition. In section IV, we explore the question of differences in genetic etiology among disabled readers related to IQ, general processing speed, and "phonological-surface" subtype dimensions. In section V, the results of DNA linkage analyses will suggest that a major gene or genes on the short arm of chromosome 6 may be involved in the etiology of many reading disabilities. We end the chapter with a discussion of the implications of our behavior-genetic results for the prevention and remediation of reading disabilities.

## II. Behavioral-Genetic Analyses of Group Deficits

Twin pairs from the third through twelfth grades who speak English as their first language are identified from school records in 27 Colorado school districts. If at least one member of a pair has some indication of a reading problem in their school records (based on low test scores if available, or information from teachers), both twins are invited to participate in a weekend testing session at the University of Colorado. A smaller comparison sample of twins with no evidence of a reading problem in their records is also tested, although a small percentage of these twins actually show a significant reading deficit in our measures. Twins with IQ scores below 85 on both verbal and performance subscales of the Wechsler (1974) or neurological signs such as seizures are usually excluded from the analyses.

Our behavioral-genetic analyses are based on the comparison of identical and

fraternal twin-pair similarities: Identical or one-egg (Monozygotic)(MZ) twins share the same genes while fraternal or two-egg (Dizygotic)(DZ) twins share only half their segregating genes on average (Plomin, DeFries, & McClearn, 1990). With the assumption of equal shared environment for the MZ and DZ pairs, contrasts between within-pair similarities for MZ and same-sex DZ pairs provide evidence regarding the relative influence of genetic factors and shared-environment factors, while within-pair differences for MZ twins (who share the same genes and home environment) provide evidence for the influence of non-shared environment, including test error.

Behavioral-genetic analyses of the broad range of individual differences in the general population typically compare the correlations or covariance matrices for MZ and DZ twins, yielding estimates of the proportion of individual variation due to genetic factors ( $h^2$ ), shared environment ( $c^2$ ), and non-shared environment ( $e^2$ ). This approach focuses on individual differences across the general population, and not specifically on a deviant group. It is quite possible that the genetic influence on deviant group membership is different from the genetic influence on individual differences across the general population.

A different behavioral-genetic analysis is appropriate for assessing the heritability of deviant group ( $g$ ) membership (e.g., reading disabled) when twins are selected from the extreme low end of the normal distribution. The twin(s) of a pair who is (are) deviant enough on the reading dimension to be classified as reading disabled is (are) called the proband(s), and the other member of the pair is called the cotwin (who may or may not also be a proband). If both members of a twin pair meet

the classification criterion for deficit-group membership, they are entered twice in the analysis (see below), with twin members exchanging proband and cotwin status. Thus, within-pair twin differences below the criterion are canceled out. For a genetically influenced group deficit, this "double entry" procedure will happen more often for MZ pairs. The standard errors for the estimates are based on the actual number of twin pairs in the analyses.

The genetic ( $h^2_g$ ) and shared-environment ( $c^2_g$ ) proportional influence on the probands' group membership in the low tail (commonly  $< -1.5$  standard deviation in our analyses) of the reading dimension can be assessed by comparing the amount of regression toward the population mean for the MZ and DZ cotwins. For an extreme example, if the probands' group reading deficit was entirely due to genetic influence ( $h^2_g = 1$ ) and there was no test error, MZ cotwins would show no regression to the normal population mean while DZ cotwins would regress half way to the population mean on average, because they share half their segregating genes on average.

DeFries and Fulker (1985) developed a regression model for twin data to derive estimates and their standard errors for genetic  $h^2_g$  influences on deviant group membership:

$$C = B_1P + B_2R + A,$$

where the cotwin's (C) score is predicted from the proband's (P) score and the coefficient of relationship ( $R = 1$  for MZ twins and  $.5$  for DZ twins.). (A is the regression constant.)  $B_1$  provides an estimate of average MZ and DZ twin- pair similarity while  $B_2$  estimates twice the difference between the means of the MZ and DZ cotwins, i.e. a

test of the extent to which the deficit of probands is due to genetic influence. When the data are properly transformed,  $B_2$  yields a direct estimate of the proportion of genetic influence on deviant-group membership ( $h^2_g$ ) (DeFries and Fulker, 1988). As we will see in later sections of the chapter, this basic regression model has been extended to assess the degree to which the genetic influence on group membership may vary depending on within-group subtype, and to assess common genetic influences on group deficits in different but phenotypically correlated skills.

A broad range of reading and language skills are positively correlated in our twin sample, as they are in non-twin samples (Stanovich, 1986), but there is also significant independent variance for some measures (Olson, Forsberg, & Wise, 1994). Therefore, we separately select probands (affected twins) for each variable to be at least 1.5 standard deviation units below the normal mean (e.g., below the tenth percentile) on that variable. As a result, a proband for one measure of reading (e.g., word recognition) may not necessarily be a proband for another measure (e.g., reading comprehension). The less than perfect phenotypic correlations between different reading and related cognitive skills allows the possibility of significantly different estimates of genetic ( $h^2_g$ ) and shared environment ( $c^2_g$ ) influences on the group deficit across different measures.

For example, a behavioral-genetic analysis by Olson et al. (1994) yielded rather different estimates for the group deficits on the word recognition and reading comprehension subtests of the Peabody Individual Achievement Test (PIAT) (Dunn & Markwardt, 1970). For the group deficit in PIAT word recognition,  $h^2_g = .46(.10)$ ;  $c^2_g =$

.45(.11) (standard errors of the estimates are in parentheses). In contrast, the group deficit in PIAT reading comprehension appeared to be less heritable ( $h^2_g = .27(.12)$ ) and more due to shared-environment influences ( $c^2_g = .52(.11)$ ). We hypothesized that the world knowledge needed to understand the vocabulary and concepts in the more difficult PIAT comprehension questions may have depended largely on the twins' shared educational and home environment, while basic processes (phonological and orthographic coding) associated with the development of word recognition were more constrained by genetic factors.

Beginning with our initial phenotypic studies of reading disability (Olson, Kliegl, Davidson, & Foltz, 1985), we have examined two component processes in word recognition. The phonological decoding component is assessed primarily through the timed oral reading of nonwords, although similar results are found in a silent nonword reading task (see Olson, Forsberg, Wise, & Rack, 1994, for a detailed discussion of these and other measures mentioned below). The group deficit in phonological decoding was highly heritable ( $h^2_g = .59(.12)$ ;  $c^2_g = .27(.12)$ ). A similarly high genetic influence was found for the group deficit in phoneme awareness, as measured by an oral language game similar to pig latin ( $h^2_g = .60(.17)$ ;  $c^2_g = .20(.16)$ )(Olson, Forsberg, & Wise, 1994).

The focus on phonological decoding and phoneme awareness in reading disability has often been justified by the apparent deficit in these skills when groups of older disabled readers are matched to younger normal children whose level of word recognition is similar (the "reading-level-match" comparison). The results of our

reading-level-match comparisons (Conners & Olson, 1990; Olson, 1985; Olson, Wise, Conners, Rack, & Fulker, 1989) have been consistent with those of most other studies (see Rack, Snowling, & Olson, 1992, for a review). The results suggest that unique deficits in phonological decoding and/or phoneme awareness may play a causal role in most cases of reading disability, but we are mindful of the cautions raised by Jackson and Butterfield (1989) and the need for experimental confirmation of what essentially is a causal hypothesis based on correlational evidence (Berninger, 1995).

A second component skill in word recognition, orthographic coding, has gained more attention over the past decade. The measures of this construct have varied, as have the results in reading-level-match comparisons (see Olson et al. 1994, for a review). Our operational definition of subjects' skill in orthographic coding is their ability to access words' specific orthographic patterns accurately and quickly. We measure skill in orthographic coding by having subjects quickly choose between a word and a homophonic nonword (e.g., rain rane), or between two homonyms following a priming sentence (Which one lives in the woods? bear bare). Earlier genetic analyses on a small sample of twins found no significant genetic influence on the group deficit in orthographic coding (Olson et al., 1989), but it is now clear in a larger sample and different analyses that there is substantial genetic influence on the group deficit in orthographic coding ( $h^2_g = .56(.13)$ ;  $c^2_g = .29(.13)$ )(Olson et al., 1994). In the next section, we examine the common and independent genetic variance for orthographic and phonological coding skills.

### III. Bivariate Analyses of Shared and Independent Genetic Influence

The fact that two moderately correlated variables (e.g., orthographic and phonological coding) are both significantly heritable does not imply that their genetic influences are due to the same genes. Their moderate phenotypic correlation could be due to shared-environmental influences instead. Fortunately, the DeFries and Fulker (1985) regression procedure can be extended to the bivariate case, where probands are selected on one variable and cotwin regression is assessed on a different but phenotypically correlated variable. Olson et al. (1994) reported from this type of analysis that the phenotypic correlations between probands' group deficits in word recognition, phonological decoding, phoneme awareness, and orthographic coding were largely due to the same genes, with bivariate  $h^2$ s ranging from .58 to .85 after being divided by the phenotypic regressions between the variables. However, the phenotypic correlations between variables are substantially less than 1, and latent-trait modeling (confirmatory factor analysis) has shown that the independent variance is not simply due to test error. Are there significant independent genetic effects (different genes) for this independent phenotypic variance across different reading-related variables?

The above question has been addressed in a series of new behavioral-genetic analyses. The main focus has been on the independent variance in orthographic and phonological skills. The first set of analyses regressed subjects' orthographic scores

on their phonological decoding scores and vice versa. This yielded a phonological deficit score that was independent from the subject's orthographic score and an orthographic deficit score that was independent from the subject's phonological score. Subsequent analysis of these scores in the DeFries and Fulker (1985) basic model indicated that both were significantly heritable at about  $h^2_g = .4$ . However, we are still evaluating this regression procedure to assure ourselves of its validity.

Because the phonological and orthographic variables are normally distributed within the combined proband and cotwin sample (of course the mean of this distribution is below that of the normal-range comparison sample), it is possible to use the classic genetic model employing MZ and DZ covariance matrices to assess  $h^2$ ,  $c^2$ , and  $e^2$  for individual differences across the sample range, rather than for the probands' group deficit. Of particular interest is the heritability of the independent variance in phonological decoding and orthographic coding. Neale and Cardon (1992) described a multivariate "Cholesky decomposition" procedure which yields estimates of the genetic and environmental influences on a first variable, genetic and environmental influences that are shared with a second variable, and finally genetic and environmental influences on the second variable that are independent from those of the first variable. The underlying mechanism of this decomposition is essentially that of hierarchical regression.

We recently employed the Cholesky decomposition procedure to isolate the independent genetic and environmental effects on phonological decoding and orthographic coding. The results of these analyses of data from 243 MZ and 181 DZ

twin pairs are presented in Figure 1. The numbers next to the path arrows are path coefficients. The squares of the values on the A, C, and E paths are estimates of  $h^2$  (genetic),  $c^2$  (shared environment), and  $e^2$  (non-shared environment) respectively. Figure 1a shows the results of entering phonological decoding first and orthographic coding second. Figure 1b shows the results of entering orthographic coding first and phonological decoding second. The overall fit of both genetic models was very good ( $X^2(11) = 8.33, p = .68, AIC = -13.7$ ). Their fit was slightly improved by dropping from the model paths  $C_1$  (shared-environment influence that is the same for both variables) and  $C_2$  (shared-environment influence that is specific to the last-entered variable) ( $X^2(14) = 8.41, p = .87, AIC = -19.6$ ).

Note the significant independent heritability of orthographic coding in path  $A_2$  of Figure 1 a ( $h^2 = .6^2 = .36$ ), and the independent heritability of phonological decoding in path  $A_2$  of Figure 1 b ( $h^2 = .62^2 = .38$ ). A similar pattern of independent genetic influence for phonological and orthographic coding was found when these models were applied to data from the normal-range control twin sample.

Hohnen and Stevenson (1995) recently reported a Cholesky decomposition for a second "orthographic" reading factor in their unselected sample of 13 year old London twins. This factor also showed independent genetic variance after accounting for variance in a first "phonological" factor. The "orthographic" factor was mostly due to variance in the same-different matching of letter strings that were different in case, so it is not clear that this factor is tapping the same skills as our orthographic choice tasks.

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The significant independent genetic variance for both phonological decoding and orthographic coding adds new understanding to our prior knowledge of their partial phenotypic independence (Barker, Wagner, & Torgesen, 1992; Olson et al., 1994a). Stanovich and West (1989) and Olson et al. have linked part of the independent orthographic variance to differences in print exposure measured by the recognition of popular book titles in children's literature. Recent behavioral-genetic analyses of the group deficit in print exposure have revealed a strong and significant influence from shared environment ( $c_g^2 = .60 (.13)$ ), but no significant genetic influence ( $h_g^2 = .21 (.13)$ ). Therefore, the present twin analyses reveal a significant genetic basis for the independent variance in orthographic coding.

#### IV. Etiology of Reading-Disability Subtypes

Individual differences among disabled readers has been a long-term concern in the field of learning disabilities and it is a major focus of our research at both the phenotypic and genotypic levels of analysis. DeFries and Fulker (1985) noted that an extension of their basic model for determining  $h_g^2$  would allow a test for differential heritability in relation to subtype variables. The extension simply requires the inclusion

of the probands' subtype designation ( $S$ ) and the product of the coefficient of relationship and subtype designation ( $RS$ ):

$$C = B_1P + B_2R + B_3S + B_4PS + B_5RS$$

$B_5$  provides a test of the significance of differences in  $h^2_g$  as a function of the subtype variable. Subtype variables in the model can range from dichotomous groups, such as gender, to continuously distributed variables such as age, and IQ.

#### IQ and the Heritability of the Group Deficit in Word Recognition

Many have argued that a discrepancy between reading level and IQ is essential to the classification of dyslexia or specific reading disability, but this argument has recently been reconsidered. One reason for this reconsideration is that disabled readers with reading-discrepant and non reading-discrepant IQ scores tend to have similar phonological deficits (Siegel, 1989; Stanovich & Siegel, 1994). In addition, difficulties in reading may ultimately cause lower IQ (Stanovich, 1986; Shaywitz, Holford, Holahan, Fletcher, Steubing, Francis, & Shaywitz, 1995). Here we consider the possibility that disabled readers' relative level of IQ may be related to the genetic etiology of their deficit in word recognition.

In an earlier study we failed to find significant differences in genetic etiology for poor reading defined by ability or IQ-discrepancy levels, but we noted that these definitions were highly correlated ( $r = .93$ ) (Pennington, Gilger, Olson, & DeFries, 1992). In our most recent unpublished analyses, we increased the IQ range by

relaxing the minimum verbal or performance IQ criterion from 90 to 85 (the lowest full-scale IQ score was 77) and focused on group deficits in a highly reliable measure of word recognition. In addition, we increased the twin sample size to 279 MZ and 334 DZ pairs by relaxing the proband deficit severity criterion to -1 SD on word recognition and by adding opposite-sex DZ twins.

We defined a residual full-scale IQ-subtype variable that was not confounded with level of word recognition among the reading disabled probands.

The reason for using the residual IQ variable rather than an unadjusted IQ score is as follows. There was a broad range of proband deficits in word recognition below the -1 SD criterion, and a broad range of full-scale IQ scores (77 to 133). The word recognition and IQ variables are significantly correlated within the proband sample ( $r = .23$ ). Therefore, the use of unadjusted IQ scores as the subtype dimension would lead to a more severe proband deficit in word recognition in a low versus high IQ group. Any difference in the heritability of deficits in word recognition along the IQ subtype dimension could be due to differences in IQ and/or differences in the severity of probands' word-recognition deficit. Instead, we regress IQ on word recognition and then create an adjusted IQ subtype variable based on the deviation of subjects' IQ scores from the regression line for word recognition. This residual IQ variable is then uncorrelated with the severity of the probands' deficit in word recognition. The procedure is similar to approaches that distinguish "garden variety" from "dyslexic" readers based on differences in standard scores for reading and IQ (c.f., Stanovich, 1986). "Dyslexic" readers would be those whose IQ is relatively high compared to their

reading level, while "garden variety" poor readers would have IQ standard scores that are closer to or even below their reading level.

The residual IQ variable was tested for the significance of its interaction with level of heritability ( $h^2_g$ ) for the group deficit in word recognition. The interaction term (B5 in Table 1) has now reached an acceptable level of significance ( $p = .05$ , two tailed). Earlier analyses by Olson, Rack, Conners, DeFries, and Fulker (1991) and by DeFries and Light (1996) had reported similar but nonsignificant trends in the same direction with other reading measures. The direction of the IQ interaction with the heritability of probands' deficits in word recognition is indicated in Table 1 by dividing subjects along the residual IQ dimension into three nearly equal sized groups. The resulting groups had similar mean proband deficits in word recognition, but different mean full-scale IQ scores of 89 (range = 77-99), 99 (range = 92-106), and 110 (range = 96-133). (The IQ ranges for each group overlap because the groups were divided along the adjusted IQ dimension described above.) Heritability levels for the group deficit were then separately assessed within each of the subgroups. It is clear that with increasing levels of IQ, heritability level increases and shared-environment decreases. The low-IQ group has a very low level of genetic etiology and a high level of shared-environment influence on the group deficit in word recognition. The opposite pattern was found for the high-IQ group.

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We have examined the relations between the IQ-subtype dimension and many other variables to try to understand the related differences in genetic and shared-environment etiology. The percentage of female probands is higher (57%) in the low-IQ group than in the high-IQ group (39%), but males ( $h^2_g = .40$ ) and females ( $h^2_g = .36$ ) in this analysis were nearly the same in their group heritability-levels for word-recognition deficits. Not surprisingly, the low IQ group had significantly lower scores in reading comprehension (Conners & Olson, 1990). There were no IQ-subtype relations to a number of birth-problem indices that might have accounted for a strong shared-environment effect on both MZ and DZ twins. There were small but statistically significant differences in mothers' years of education and in number of books in the home (lower in the low IQ group) that might be linked to a poorer shared environment for reading development. The groups did not differ on parent reports regarding frequency of reading to their children.

#### General Processing Speed and $h^2_g$ for Word Recognition

A general processing-speed dimension was defined by subjects' z scores on four measures that loaded on a common factor, similar to a mental abilities factor called "perceptual speed" in John Carroll's (1993) review. The highest loading measure, Identical Pictures (IP) (French, Ekstrom, & Price, 1963), required subjects to find the match for a picture (e.g., a house) among similar foils (e.g., houses with slight variations in detail). WISC-R Coding (Wechsler, 1974) required the rapid mapping of symbols with numbers based on a key. Colorado Perceptual Speed (DeFries, Singer, Foch, & Lewiter, 1978) followed the Identical Pictures procedure but with patterns of

letters and numbers (e.g., bhsf). Finally, the Denkla and Rudel (1976) Rapid Automatic Naming (RAN) test for numbers and letters also loaded on the speed factor.

When the individual speed tasks (regressed on word recognition) were used as subtype variables, each showed a similar pattern for differential genetic influence on word recognition: The slowest third of the sample showed a relatively low level of genetic etiology for word recognition and high shared-environment influence. The opposite pattern was seen for the high-speed group. However, the subtype interaction was statistically significant only for the Identical Pictures task (see Table 1). Therefore, a composite speed dimension was created by adding the z scores for the four tasks. A similar pattern of differential  $h^2_g$  was found for the combined measure, and the subtype interaction was statistically significant ( $p = .05$ , two tailed) (see Table 1).

When comparing the speed-subtype groups on other variables, we found strong differences in the rapid naming of pictures and colors (not used to form the speed dimension), modest but significant differences in IQ (although no differences in reading comprehension), a significant deficit for the slow speed group in the orthographic coding tasks, and no significant differences in mothers' years of education or books in the home. Bowers and Wolf (1993) have speculated that naming speed for letters and numbers is related to precise timing mechanisms that have been proposed by Tallal (1980) as the primary basis for reading disability. Unfortunately, we have no measures in our battery that could be used to test this hypothesis.

In view of the low heritability for word-recognition deficits in the slow group, we wondered if genetic influence on the same subjects' speed deficits would be similarly low. In fact, the heritability for the group deficit ( $< -1.5$ ) in the composite speed measure was low and non significant ( $h^2_g = .18 (.13)$ ), while shared-environment influence was highly significant ( $c^2_g = .65 (.12)$ ). This result mirrors the low heritability and high shared environment for word recognition in the slow speed group. It suggests that same shared environment factor or factors may be causing this subgroup's slow speed and poor word recognition.

Further research is needed to understand the broad implications of the IQ and speed subtype dimensions for the etiology and remediation of different reading disabilities. At present there is limited evidence for shared-environmental deprivation (slightly lower print exposure and maternal education) that may jointly lead to depressed IQ and reading. However, these variables were not significantly related to deficits in the speed tasks. We may have to search more broadly for other shared-environmental effects such as maternal nutrition, illness, or lead exposure. In future analyses we plan to compare the behavioral-genetic subtype results with those from genetic linkage analyses of individual's patterns of DNA (see Section V) and from MRI studies of individual's brain morphology.

### Phonological-Orthographic Profiles

A third dimension of individual differences among disabled readers, the relative levels of phonological and orthographic skills, has a long history (e.g., Boder, 1973; Gjessing, 1953), and renewed attention in two recent papers (Castles & Coltheart,

1993; Manis, Seidenberg, Doi, McBride-Chang, & Peterson, 1996). Here we add to our understanding of this dimension by assessing its interaction with the heritability of probands' deficits in word recognition, by assessing its genetic basis, and by relating it to other dimensions of reading and language,

The subtraction of subjects'  $z$  scores for orthographic coding from their  $z$  scores for phonological coding yielded a normally distributed dimension that was not significantly correlated with their word recognition scores. Consistent with historical and current subtype terminology (Castles & Coltheart, 1993; Manis et al., 1996), we will refer to "phonological dyslexics" who were low on this dimension with relatively poor phonological coding, and "surface dyslexics" who were high on the dimension with relatively poor orthographic coding. The test-retest reliability for the subtype dimension was assessed in a longitudinal study (average test interval of 5 years) of 150 probands and cotwins. This correlation was .7, indicating a substantial reliability and longitudinal consistency for the measure.

Gayan, Forsberg, and Olson (1994) reported that the interaction between the phonological-surface subtype dimension and  $h^2_g$  for probands' group word-recognition deficit below -1.5 SD was not significant. More recently, we relaxed the minimum proband deficit criterion to -1 SD and found a slightly stronger but still non-significant interaction. The trend of this non-significant interaction suggested that the "phonological dyslexic" group word-recognition deficit ( $h^2_g = .53$ ) might be more heritable than that of the "surface dyslexic" group ( $h^2_g = .30$ ). However, the difference in heritability was much smaller when the subtype variable was based on percent

correct in the phonological and orthographic tasks, or when probands' deficits in word recognition were more severe (at least -2.0 SD).

Even though the interaction between  $h^2_g$  for word recognition and the "phonological-surface" subtype dimension was not significant, this does not imply that there is no significant genetic contribution to the subtype dimension itself. The possibility of at least partially different genetic mechanisms for "phonological" and "surface" subtypes is suggested by the previously discussed results showing significant genetic influence on the independent variance in phonological and orthographic coding. Gayan et al. (1994) assessed the group heritability of disabled readers' extreme positions on the normally distributed subtype dimension by selecting probands who were one standard deviation above the mean ("surface dyslexics") or below the mean ("phonological dyslexics") and then observing their cotwin regression to the sample mean. Group membership in both the "phonological" ( $h^2_g = .68 (.32)$ ) and "surface" ( $h^2_g = .65 (.34)$ ) subtypes proved to be significantly heritable.

After establishing the long-term stability and partial genetic basis for the "phonological-surface" subtype dimension, we have begun to explore its phenotypic relations with other variables. To understand how various speed measures are related to the dimension, we re-created the dimension based only on relative accuracy in the phonological decoding and orthographic coding tasks, rather than the combination of accuracy and speed used in the measures discussed earlier. We then looked at differences in performance for a number of variables depending on the

disabled readers' (N = 307) membership in the lower ("phonological dyslexic") or upper ("surface dyslexic") half of the dimension.

Although the "phonological dyslexics" were less accurate in nonword reading, they were significantly ( $p < .05$ ) faster on correct responses, and their errors were more frequently words. They were also faster in two measures of orthographic coding and in a timed word-recognition test. The "phonological dyslexics" were significantly lower in their accuracy on several oral language measures of phonological awareness, including phoneme deletion and a "pig-latin" game. They were also significantly lower on a rhyme generation task. However, the groups did not differ in the Rapid Automatic Naming (RAN) task or in Verbal IQ.

The groups did not differ significantly on the Identical Pictures (IP) test discussed earlier, although a trend favored the "phonological dyslexics". In contrast, the "phonological dyslexics" were significantly worse on a Perceptual Organization factor derived from the Wechsler subscales. This latter result suggested to us that there might be a significant gender difference between the subtypes: In fact, proportionally more females were on the "phonological dyslexic" side of the dimension. However, this was largely due to the females' superior performance on the orthographic tasks (and on a measure of timed word recognition). Males and females did not differ significantly on phonological decoding or on the language measures of phonological awareness, although the females were significantly faster in the RAN task. Thus, there are complex interactions between gender, subtype dimension, and related variables. Further research is needed to fully understand

these interactions.

While genetic factors seem to be playing a significant role in "phonological" and "surface" subtype membership and in related language skills, we should not overlook the possible role of environmental factors, particularly those related to methods of early reading instruction/remediation and print exposure (Manis et al., 1996; Wise and Olson, 1995). Unfortunately we have not been successful in obtaining reliable information on the twins' reading instruction, but we have collected parent reports on number of books in the home and we have used the Book Title Recognition Test (Stanovich & West, 1989) to assess individual differences in print exposure. Both of these measures indicate significantly higher levels of print exposure for the "phonological dyslexics", who had relatively high orthographic accuracy. Print exposure may play a significant role, along with genetic factors, in the establishment of rapidly assessable and accurate orthographic codes for words.

#### V. Implications for Specific Genetic and Environmental Effects on Individuals: Linkage and Training Studies.

Our behavioral-genetic data indicate that there is a significant genetic basis and significant shared environment for the group deficit in word recognition. However, behavioral-genetic analyses can not determine the relative balance of genetic and environmental influences on an individual's deficit. The deficit for some individuals in the group may be due entirely to shared and/or non shared environment, while for

other disabled readers it is likely that the genetic influence is quite strong. The foregoing subtype analyses indicated that the likely proportion of genetic and environmental influence may vary depending on an individual's profile of IQ, general processing speed, and possibly their "phonological-surface" subtype. These important results bring us a step closer to making more accurate predictions about the genetic and environmental etiology of an individual's reading deficit. Now, we turn to recent evidence from disabled readers' genes (DNA) that may eventually lead to a much more precise specification of individual etiology.

Linkage analyses are being used to search for genetic markers that could ultimately lead to the identification of specific genes associated with individual reading deficits. Markers indicating the presence of a specific DNA pattern have been shown to co-occur significantly more often in relatives that share a reading disorder. These markers are not the specific gene or genes that may contribute to a reading disability, but the markers are close enough to co-segregate with the genes that are responsible. (Adjacent regions on a chromosome tend to be inherited together.) Cardon, Smith, Fulker, Kimberling, Pennington, & DeFries (1994) reported evidence from two independent samples (ordinary sibling pairs and DZ twins) for linkage of a composite measure of reading within a 2-centimorgan region on the short arm of chromosome 6. Gayan, Olson, Cardon, Smith, Fulker, Kimberling, Pennington, & DeFries (1995) subsequently reported evidence in the same DZ twin sample that deficits in word recognition show the strongest linkage in this region, compared to deficits in reading comprehension and spelling.

A new sample of DZ twins in the Colorado study has recently been genotyped with new markers in the same region of chromosome 6 studied by Cardon et al. (1994) and Gayan et al. (1995). Preliminary analyses with this second replication sample have again indicated significant linkage for reading measures in the HLA region of chromosome 6. Moreover, linkage appears to be particularly strong for disabled readers' deficits in phonological decoding, phoneme awareness, and orthographic coding. An independent laboratory has also provided confirming evidence of linkage for deficits in reading and phoneme awareness in the same region of chromosome 6 (Grigorenko, Wood, Meyer, Hart, Speed, Shuster, & Pauls, 1997).

Much more work needs to be done to further verify the above linkage results, to find the specific gene or genes involved in this region as well as other possible regions of the genome, and to understand how the proteins coded by genes associated with reading disabilities ultimately influence brain development and behavior. The independent genetic influences on the phonological and orthographic components of word recognition add to the complexity of these goals. The specific genetic and environmental mechanisms will certainly vary across individuals.

In conclusion, behavioral-genetic analyses of MZ-DZ twin similarities and linkage analyses of data from DZ twins have both indicated a significant genetic influence on the group deficit in word recognition. Subtype analyses suggested that genetic influences were likely to be particularly strong for disabled readers with relatively high IQ and/or general processing speed. Nevertheless, we emphasize now

as strongly as before (Olson et al., 1989), that evidence for genetic influences on reading disabilities and the apparent longitudinal stability of phonological deficits (Wagner, Torgesen, & Rashotte, 1994), should not discourage our best efforts in remediation. In the medical field, environmental interventions for largely genetic disorders have been quite successful (e.g., insulin for diabetes, glasses for myopia, diet for PKU, etc.). Although reputable studies of different remedial programs for reading disabilities have not found such simple or complete cures, intense remedial training in reading and phonological skills can result in substantial gains for most disabled readers (c.f., Levy, this volume; Lovett, this volume; Olson & Wise, 1992; Wise & Olson, 1995). The evidence for genetic influence helps explain why additional instructional resources may be required for some children with reading disability. Until the specific genetic mechanisms are better understood, a main contribution of behavioral-genetic and linkage evidence may be the early identification of children at risk for reading disability followed by intensive remediation of their phonological and/or orthographic deficits during critical periods of language and reading development.

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Table 1: Genetic ( $h^2_g$ ) and shared-environment ( $c^2_g$ ) Influence on Group Deficits ( $< -1.0$  SD) in Word Recognition Depending on Subjects' Adjusted Full-Scale Wechsler IQ, Colorado Perceptual Speed (CPS), Identical Pictures (IP), Rapid Automatic Naming (RAN), the Coding subscale of the WISC-R, and a Composite of CPS, IP, RAN, and WISC-R Coding.

Subtype Level	$h^2_g$ (SE)	B5 signif.	$c^2_g$
Low IQ	.32 (.13) *		.63
Medium IQ	.39 (.12) *	$p = .05^*$	.56
High IQ	.54 (.16) *		.37
Low CPS	.28 (.14) *		.67
Medium CPS	.38 (.14) *	$p = .18$	.60
High CPS	.58 (.16) *		.31
Low IP	.10 (.16)		.84
Medium IP	.43 (.13) *	$p = .04^*$	.52
High IP	.61 (.16) *		.33
Low RAN	.26 (.16)		.69
Medium RAN	.38 (.13) *	$p = .30$	.56
High RAN	.52 (.17) *		.40
Low WISC Coding	.33 (.15) *		.62

## Genetics of Reading Disabilities

34

Medium WISC Coding	.22 (.14)	$\underline{p} = .19$	.73
High WISC Coding	.70 (.16)*		.22
Low Speed Composite	.20 (.15)		.76
Mid Speed Composite	.47 (.14)*	$\underline{p} = .05^*$	.48
High Speed Composite	.52 (.15)*		.40

Note: All subtype variables were adjusted for their relation to word recognition before being used to divide the groups. The significance of group differences in heritability depending on subtype (B5) was assessed by an extension of DeFries and Fulker's (1985) basic model (see text).

## Figure Note

For Figures 1a and 1b, The A1 paths indicate the proportional genetic influence on individual differences in first (left side) variable and shared genetic influences on the second (right side) variable. A2 indicates the proportional independent genetic influence on the second variable. The same relations hold for the shared-environment paths (C1, C2) and the non shared environment paths (E1, E2).