



## A twin study of the etiology of high reading ability

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**Abstract.** The present study examined the etiology of high reading ability in an overall sample of 350 twin pairs in which at least one member of 100 pairs (54 MZ, 46 DZ) had a reading composite score one standard deviation above the sample mean. These high readers also had significantly higher scores than the rest of the sample on Full Scale, Verbal and Performance IQ scores, as well as on measures of phoneme awareness, orthographic coding, phonological decoding, and verbal short-term memory. The MZ proband-wise concordance rate for high group membership was significantly higher than the DZ proband-wise concordance rate and further behavioral genetic analyses corroborated that high reading ability is partly due to genetic influence ( $h_g^2 = 0.55 \pm 0.22$ ). Bivariate multiple regression analyses demonstrated that high phonological awareness, orthographic coding, phonological decoding, and short-term verbal memory skills all share significant common genetic influence with high reading ability. These results suggest that reading ability and its cognitive correlates are on a continuous distribution, with both extremes of the distribution being similarly heritable. They also support the hypothesis that the same cognitive processes that are associated with dyslexia are important for the development of high reading ability.

**Key words:** Behavioral genetics, Orthographic coding, Phonological processing, Skilled reading

### Introduction

Previous research has clearly established that there is a significant genetic influence on low reading ability, or dyslexia. The present study examined the etiology of high reading ability in a twin sample obtained as part of the Colorado Learning Disability Research Center twin project (DeFries, Filipek, Fulker, Olson, Pennington, Smith & Wise 1997). To date, no findings have been published regarding the etiology of high reading ability, although such data would provide a useful test of the universality of the cognitive and genetic influences on reading skill that have already been found in individuals with either low or normal reading ability. More specifically, the issue of whether dyslexia (specific reading disability) and high reading skill are part of the same distribution can be partly addressed by ascertaining the etiology of high reading ability, and comparing these results with those

already established for low reading ability. In addition, the case for one continuous distribution of reading ability would be strengthened if the same cognitive processes found to covary genetically with poor reading exhibit similar relations with good reading. In what follows, a brief overview of what is known about the genetic and cognitive underpinnings of specific reading disability (dyslexia) is provided.

Specific reading disability (dyslexia) has been the focus of intense investigation over the past few decades, during which time there have been important advances in our understanding, especially in the area of behavioral genetics. Researchers have known for some time that dyslexia is familial (Hallgren 1950); however, more recent twin studies have also yielded significant low-group heritability estimates ( $h_g^2$ ) indicating that approximately half the proband deficit is attributable to genetic influences (see Defries & Alarcón, 1996, for a review). Results from the Twin Family Reading Study indicated low-group heritability estimates of 0.44 for poor reading, 0.62 for poor spelling ability, and 0.75 for nonword reading (DeFries, Stevenson, Gillis & Wadsworth 1991). Similar heritability results were obtained for spelling deficits in the London twin study, indicating that a significant proportion of the variance in poor reading and spelling is due to genetic influences (DeFries, Olson, Pennington & Smith 1991; Olson, Gillis, Rack & DeFries 1991).

As researchers have studied the cognitive underpinnings of developmental dyslexia, there has been mounting evidence that phoneme awareness skill (the ability to segment spoken speech into phonemes) is a pre-requisite for learning to read an alphabetic language like English (Lieberman, Shankweiler, Fischer & Carter 1974; Wagner & Torgesen 1987; Pennington, Van Orden, Smith, Green & Haith 1990). Phoneme awareness, as well as phonological decoding (the ability to read regular non-words) and orthographic coding (the ability to distinguish words from pseudo-homophones as well as the ability to read irregular words), have also been shown to be significantly heritable (Olson, Forsberg & Wise 1994). Olson et al. (1994) obtained a group-deficit heritability ( $h_g^2$ ) estimate of 0.56 (SE 0.13) for orthographic coding and 0.59 (SE 0.12) for phonological decoding. What is more, both of these skills were influenced by common genes, as was shown using a bivariate regression analysis. Both these skills were also related to phoneme awareness, with a bivariate  $h_g^2 = 0.60$  (0.17). The latter is the cognitive phenotype that is most strongly related to reading outcome. Thus, not only are these different cognitive processes related to one another and to reading outcome, but they also share, in part, a common genetic etiology.

At the molecular level, segregation and sib-pair linkage analyses indicated the existence of a major gene locus for reading disability with

reduced penetrance (Pennington, Gilger & Pauls 1991; Smith, Kimberling & Pennington 1991; Cardon, Smith, Fulker, Kimberling, Pennington & DeFries 1994). Using sib-pair linkage methods, Cardon et al. (1994) identified a QTL on the short arm of chromosome 6 in two separate samples that influences extreme scores on a continuous measure of reading ability (a reading composite). Grigorenko, Wood, Meyer, Hart, Speed, Shuster, and Pauls (1997) replicated this result by finding significant linkage to the same region of chromosome 6 for a phoneme awareness phenotype. In addition, the Grigorenko study found separate linkage for another phenotype, word recognition, near the centromere of chromosome 15. Most recently, Fisher et al. (1999) and Gayan et al. (1999) have each demonstrated significant linkage to chromosome 6 (6p21.3) for the component skills of phonological coding and orthographic coding. With respect to the various QTL findings on chromosome 6, it is possible, although not as likely, that there is more than one loci in this region associated with dyslexia. However, we await actual gene identification in order to know for certain whether it is the same gene or various different genes that are affecting the various reading phenotypes used in these studies.

As the behavioral genetic underpinnings of dyslexia have been studied, researchers have also been interested in the relation between dyslexia and normal reading ability. A number of different methods have been used to assess whether reading disability is qualitatively distinct from normal reading skill. For example, Rutter and Yule (1975) found that those children with specific reading disability formed a "hump" at the bottom of the normal distribution. They argued that the apparent bi-modality in the Isle of Wight sample indicated that dyslexia is a specific and distinct entity, separate from normal reading skill. On the other hand, various other studies investigating this same question did not find bimodality in the distribution (Stevenson 1988; Shaywitz, Escobar, Shaywitz, Fletcher & Makuch 1992).

More sensitive tools from the area of genetic epidemiology were subsequently used to test for multimodality in a continuous trait such as reading. A commingling and segregation analysis by Gilger, Borecki, DeFries, and Pennington (1994) found that the familiarity, heritability and transmission results for normal variations in reading skill were similar to those for dyslexia. The finding of a major gene locus effect on normal reading skill suggested that the same loci may be involved in the transmission of normal reading skill and dyslexia. More definitive evidence, of course, would come from a linkage analysis using a large sample of normal readers that yielded associations to the same known (or yet to be discovered) QTLs observed in analyses of dyslexic samples.

Another approach that has addressed the question of whether dyslexia is a separate disorder with a distinct genetic etiology is to compare a group heritability estimate for a reading deficit to the heritability estimate of individual differences along the whole distribution. Equal heritability estimates would support a single distribution hypothesis of reading ability/disability, as would finding a similar group heritability estimate for an extreme group of good readers. In contrast, if heritability estimates differed significantly in these three situations, the opposite interpretation would be supported, namely that dyslexia is a unique entity with its own etiology separate from that of normal variations in reading.

Previous studies using this approach support a single distribution hypothesis. In six studies of normal reading skill, reviewed by Harris (1986), the estimate of broad heritability ( $h^2$ ) was 0.56, which is comparable to group heritability estimates ( $h_g^2$ ) of poor reading of 0.50 reported by DeFries and Gillis (1993) and DeFries, Olson, Pennington, and Smith (1991). In a Twin Family Reading Study (Olson, Gillis, Rack & DeFries 1991), there was also no significant difference between  $h_g^2$  of 0.47, the heritability of a group deficit in reading, and  $h^2$  of 0.73, the heritability of normal reading. Even if the latter comparison had reached significance, it would not necessarily prove that different genetic influences were operating in the two cases, although such a result would be consistent with that alternative hypothesis.

In summary, there is ample evidence that reading deficits are significantly due to genetic influence. There is also evidence that poor reading is associated with deficits in phoneme awareness, phonological decoding and orthographic coding. These associations are due, in part, to common genetic influences. Furthermore, there is evidence from a segregation analysis in a sample of normal readers that the etiology of normal reading skill and dyslexia may be one in the same. The hypothesis that variation in reading skill lies on a single distribution, with individuals with dyslexia comprising the lower tail due to a disproportionate representation in those individuals of alleles imparting liability, can be tested further. Ascertaining the etiology of reading and reading component processes in groups selected to be at the ends of the proposed continuum and comparing them to each other as well as to the heritability estimate for individual differences can help answer this question. Results from twin studies have shown that these heritability estimates are not significantly different when comparing dyslexia and normal variation, supporting the "one-distribution" hypothesis. This study proposes to extend this work by testing the "one-distribution" hypothesis at the opposite extreme, by ascertaining the etiology of high reading ability. Furthermore, if it can be shown that the same reading component processes that are associated with dyslexia are also bivariate heritable with high reading skill, the univer-

salinity of the underlying cognitive phenotype (a core phonological processing capacity) would be also be supported.

## **Method**

### *Participants*

Participants in this study were part of the Colorado Learning Disabilities Research Center twin project (CLDRC; DeFries et al. 1997), an ongoing study of the etiology of learning disabilities. Subjects were ascertained via a two-stage procedure. First, with the collaboration of school administrators and personnel, all twin pairs from 27 school districts within a 150-mile radius of the Denver/Boulder area were contacted and parental permission was requested to review each child's academic records for evidence of reading problems. If either member of a twin pair had a positive history of reading difficulty (e.g. low reading achievement test scores, referral to a school reading specialist or tutor, reports by classroom teachers or school psychologists etc.), both members of the twin pair were invited to complete an extensive battery of tests in the laboratories of the CLDRC. If neither member of a twin pair had a positive history of reading difficulty, then both twins in the pair were invited to participate in the study as part of a normal range comparison group. These pairs were selected to match the RD twin pairs on various characteristics such as age, gender and school district. For the purposes of this study, only those twins originally selected for the normal range comparison group were used. The school history of reading difficulty was only used as a screening; a definitive reading diagnosis (RD or not) was subsequently derived from the reading achievement tests administered in the battery (described below). Thus, a small proportion of twins originally selected to be in the normal range comparison group met criteria for reading disability, although they were only mildly impaired. These dyslexic readers comprised 7.1% of the total normal comparison group, a figure that is somewhat higher but not significantly different from prevalence rates of reading disability in the population (Shaywitz, Shaywitz, Fletcher & Escobar 1990). Twin pairs were all between the ages of 8–18, and did not have any neurological disorder, congenital syndrome, history of hearing loss or head injury. Subjects with Full Scale IQ below 80 were also excluded from the analyses.

Zygoty of the twins was determined using a modified version of the Nichols and Bilbro zygoty questionnaire (1966), which has been shown to reliably classify 96% of twins. If zygoty classification was ambiguous based on the questionnaire, DNA polymorphisms obtained from a blood plasma

or buccal swab were analyzed to ensure accurate categorization. Based on these criteria, a total of 215 monozygotic (MZ) twin pairs and 135 same-sex dizygotic (DZ) twin pairs were selected for the current sample (N = 700 children).

### *Measures*

*Reading achievement.* The Reading Recognition, Reading Comprehension and Spelling subtests of the Peabody Individual Achievement Test (PIAT; Dunn & Markwardt 1970) were administered to all twins in this sample. A normally distributed reading composite score was created based on a previous discriminant function analysis of these three reading subtests conducted in separate samples of non-twin individuals with and without a history of reading problems (DeFries 1985). As the purpose of the initial discriminant function was to establish a formula to identify individuals with reading disability (RD), a 1.65 standard deviation discrepancy (from the mean of the sample of control twins without a history of reading problems) was used to define reading disability. This cutoff, recommended by Reynolds (1984), selects approximately five percent of the entire control sample (which includes opposite sex twin pairs), a prevalence that is consistent with estimates from epidemiological studies (e.g. Shaywitz, Shaywitz, Fletcher & Escobar 1990).

The sample in this study is a subset of the entire control group described above, as it excludes opposite sex twin pairs. The subjects in this study did not have a positive history of reading difficulties in school records; however, 7.1% of them did have a discriminant function score that fell below the RD cutoff. This same reading composite score was used to identify those children with high reading (HR) ability, as will be explained further in the data analyses section.

*Cognitive ability.* Each child's cognitive ability was assessed using the revised version of the Wechsler Intelligence Scale for Children (WISC-R; Wechsler, 1974). The digit span subtest of the WISC-R was used as a specific measure of short-term verbal memory, while the three composite scales were used for exclusionary purposes and to provide demographic information.

*Phoneme awareness (PA).* The first measure is a modified version of the *Lindamood Auditory Conceptualization Test* (LAC; Lindamood & Lindamood 1971). On the first part of the test, the child is asked to listen to a series of three or four phonemes and to represent the order of said phonemes using different colored blocks. On the second part of the test, the child is required to add, remove or transpose blocks to reflect changes in non-words

spoken by the examiner. For example, the participant might be shown three different colored blocks in a row and told: "if this says *aps*, show me *asp*". The correct response would then be to exchange the position of the second and third blocks. This test requires the child to parse a sound string into individual phonemes and also identify how the order of these sounds changes. The original items of the LAC were used in addition to 6 more items that were constructed for part II of the test, in order to prevent a ceiling effect from older children. The total number of items correct was utilized as the dependent variable.

The second test used is the *Pig Latin task* (Olson, Wise, Conners, Rack & Fulker 1989). This task requires the participant to transform words into their Pig-Latin equivalent. The participant is told the rules for transforming words (e.g. move the initial sound to the end of the word, and add the long sound *a*), and completes nine practice words read by the examiner. The examiner is permitted to provide assistance during the practice trials in order to ensure that the participant understands the task. The test trial then requires the participant to transform 48 words read by the examiner into their Pig-Latin equivalent. The total number correct was utilized as the dependent variable.

The third phoneme awareness measure, the *Phoneme Deletion Task* (Olson, Forsberg & Wise 1994), is based on the Bruce (1964) phoneme deletion task and the Rosner and Simon (1971) auditory analysis task. The examiner reads aloud a pronounceable non-word (e.g. *plift*) and then asks the participant to say the word after removing one phoneme (say 'plift' without the 'p'). Total correct responses were utilized as the dependent variable.

Previous exploratory and confirmatory factor analysis in the CLDRC sample indicated that the three PA measures loaded on a single factor (Olson et al. 1994). Exploratory factor analysis in the present sample also indicated that the three PA measures all loaded above 0.84 on a single factor. Thus, the standardized and age regressed scores from each task were averaged to create a PA composite score which was then utilized in all analyses. The Lindamood and Phoneme Deletion tasks were only administered to a subset of subjects in the entire sample, thus, in cases where one of these was not administered, the PA variable was computed by averaging scores from the two remaining tasks.

*Orthographic coding.* This test measures the participant's ability to directly access an orthographic representation in the lexicon in order to distinguish words from non-word letter strings that would be identical in sound if pronounced (pseudo-homophones). Thus, a participant is visually presented a pair such as 'cain' and 'cane', and s/he has to press a button to designate which of the two letter strings is a word. Use of the phonological code

will not assist the participant as both words sound identical. A correct decision is exclusively based on the word's orthographic code. The current task has 80 trials presented via computer, with pairs balanced to ensure the distractor item is a plausible letter string ('certain – sertain' is balanced by the trial 'serpent – cerpent'). In this manner, participants must make use of word-specific knowledge rather than general information about orthographic structure. Total number correct is the dependent variable.

*Phonological decoding.* This test is a silent lexical decision task for pseudo-homophone nonwords. It requires the child to pick from three pronounceable letter strings presented side by side on a computer screen (e.g. "coam – baim – goam"), the one that would sound like a common word if read aloud ("coam"). The child presses one of three buttons as quickly as possible denoting his or her selection. The response is immediately underlined for one second, then the correct choice is highlighted for one second, and then the next trial is presented two seconds later. The targets and distracters are balanced in terms of their visual similarity to real words. Thus, subjects could not respond by simply picking the most visually familiar letter-string. The task requires first that the subject generate the internal sound codes for the nonwords (because there is no oral response, variability in overt articulatory skills should not have any direct influence on performance on this task). The child then has to match the phonetic code for the nonword (e.g., coam) and match it to a word (e.g., comb) in his/her lexicon. Total number correct is the dependent variable. The age-adjusted correlation of this task with oral non-word reading is 0.80.

#### *Data analyses*

*Phenotypic analyses.* High reading ability (HR) was defined as a reading composite score that fell at least 1.0 standard deviation above the sample mean. Although this is not as extreme a cutoff as could have been selected, it was chosen for two reasons. First, as the mean reading scores on the PIAT subtests for this sample were already above the national norm, a one standard deviation cutoff selected individuals with reading scores above the 87–90th percentile relative to national standards. Second, when more extreme cutoffs were attempted, they yielded smaller sample sizes, which when divided by zygosity status led to insufficient power for the behavioral genetic analyses. A one standard deviation cutoff provided a reasonably high reading ability group with a sufficient number of subjects. This cutoff yielded a total of 131 subjects with high reading (74 MZ twins and 57 DZ twins), that corresponds to 18% of the total sample (N = 700). This is just higher than the percentage predicted from a one standard deviation cutoff, due to a slightly negatively



skewed sample. In terms of twin pairs, 54 MZ twin pairs and 46 DZ twin pairs (out of a total of 350 pairs) had at least one twin who had high reading (HR) ability based on the composite reading score that is further explained below.

The utilization of twins for phenotypic analyses in which each subject is considered as an individual data point presents a methodological dilemma, as the scores of the twins in each pair do not represent independent observations. Therefore, for each twin pair in which both twins met criteria for HR ability (concordant pairs) or both qualified for the control sample, one twin was selected at random for inclusion in the initial phenotypic analyses. For those pairs in which one twin met criteria for HR and one twin did not (discordant pairs), the twin with HR ability was included in the HR sample. Cotwins of HR probands who did not meet criteria for HR (e.g., cotwins from discordant pairs) were not included in either sample to avoid biasing the non-HR comparison sample.

Mean reading, phoneme awareness, short term verbal memory, phonological decoding, orthographic coding, and IQ scores of participants with and without high reading scores were compared using simple *t*-tests. In order to facilitate the comparison of the relative magnitude of the differences between the two groups on each measure, the effect size for each comparison was computed using the following equation (Cohen 1977):

$$d = (M_{\text{Control}} - M_{\text{RD}}) / [(SD_{\text{Control}} + SD_{\text{RD}}) / 2] \quad (1)$$

where *d* is the effect size,  $M_{\text{Control}}$  and  $M_{\text{RD}}$  are the means of the control and HR sample, respectively, and  $SD_{\text{Control}}$  and  $SD_{\text{RD}}$  are the standard deviations of the two groups.

In order to understand the relationship between reading and the component processes further, a correlational matrix was computed that included all four component process variables and the reading composite score. In addition, a logistic regression model was conducted in which HR status was predicted from scores on the PA, phonological decoding, orthographic coding, and short-term verbal memory tests. In this analysis, all four variables were entered into the regression equation at the same time. A series of follow up logistic regressions were also performed excluding one variable, in turn, from the overall model in order to further refine the interpretation of the results.

*Concordance estimates.* A comparison of concordance rates in monozygotic twin pairs and dizygotic twin pairs can be used as a test for genetic etiology. A pair is concordant if both members are affected with the same disorder, but discordant if only one member is affected. Since monozygotic twin pairs share all of their genes, whereas DZ twins share half of their segregating genes, on average, the MZ concordance rate is expected to exceed the DZ

concordance rate if a disorder is at least partly heritable. Therefore, a significantly higher rate of concordance for HR between pairs of MZ twins than DZ twins would suggest that HR ability is attributable to the influence of genes. For the present analysis, proband-wise concordance rates were computed, whereby all twins who met criteria for HR were selected as probands. For those cases in which both twins from a pair met criteria for HR, the pair was double entered into the file (i.e., each member of the pair was entered once as a proband and once as a cotwin). This procedure provides the most valid estimate of twin concordance when a sample has been ascertained using truncate selection (e.g. DeFries & Alarcon 1996; McGue 1992).

*Univariate behavior genetic analyses.* Although a comparison of concordance rates in MZ and DZ twin pairs provides a conceptually simple appraisal of the etiology of a discrete disorder, the multiple regression method proposed by DeFries and Fulker (DF method; 1985, 1988) provides a more powerful and versatile test of the etiology of extreme scores (high reading ability in this case). This method takes advantage of the continuous dimension of reading ability rather than using a dichotomized variable (e.g. whether the proband meets high reading cutoff or not), as is the case when calculating concordance rates. Although reading disability is diagnosed primarily on the basis of quantitative measures with arbitrary cutoff points (Stevenson, Graham, Fredman & McLoughlin 1987), transformation of scores from a continuous measure to a categorical variable necessarily results in a loss of information regarding the variation present in reading performance along the entire ability continuum. In contrast, multiple regression analyses of twin data allow for this variation to be used when studying the etiology of extreme scores as well as individual differences within an affected group (DeFries & Fulker 1985). Thus, the same methodology can be applied to determine the etiology of extreme scores at the high end of the reading ability spectrum.

The multiple regression model proposed by DeFries and Fulker (DF method; 1985, 1988) is based on the regression of monozygotic (MZ) and dizygotic (DZ) cotwin scores toward the population mean when probands are selected due to extreme scores on a phenotype. In this particular case, MZ and DZ probands are selected for high reading ability and their cotwins' reading scores would be predicted to regress toward the population mean. Although scores of both MZ and DZ cotwins would be expected to regress toward the population mean, it is the differential regression by zygosity that allows inferences regarding the etiology of an extreme score. To the extent that extreme scores on the trait are influenced by genes, scores of DZ cotwins should regress further toward the mean of the unselected population than scores of MZ cotwins. This is true because MZ twins share all of their genes,

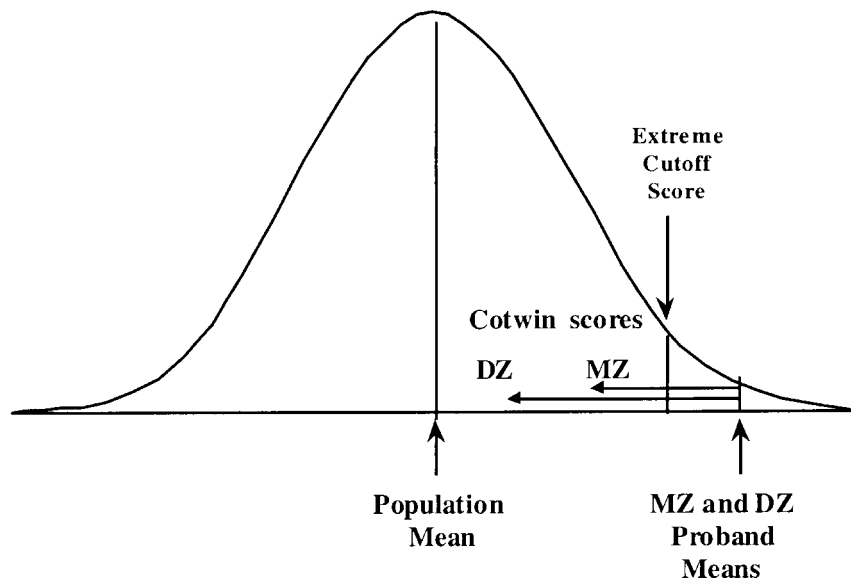


Figure 1. The DeFries-Fulker multiple regression model (DeFries & Fulker 1985, 1988). This model assesses the differential regression to the mean of MZ and DZ cotwins when probands are selected due to an extreme score.

whereas DZ twins share half of their segregating genes on average. Therefore, the greater genetic similarity between MZ twins would cause the MZ cotwin to remain further out on the continuum of reading ability (closer to his high reading sibling who was picked as the proband), as compared to a DZ cotwin (see Figure 1). The magnitude of this differential regression by zygosity provides an estimate of the heritability of extreme scores on the trait under consideration ( $h_g^2$ ).

Because the double entry of concordant pairs artificially inflates the sample size, the standard errors of the regression coefficients and resulting  $t$ -values were corrected for double entry in order to obtain unbiased tests of significance (e.g., Stevenson, Pennington, Gilger, DeFries & Gillis 1993). Prior to multiple regression analysis, standardized scores were created based on the mean and standard deviation of the control sample in order to facilitate the estimation of  $h_g^2$ . The standardized scores for the selected sample of MZ probands and cotwins were then divided by the MZ proband mean, and the DZ proband and cotwin scores were divided by the DZ proband mean. This procedure ensures that the MZ and DZ probands are equally divergent from the mean score of the controls prior to the regression analysis.

The basic regression model for the univariate case is as follows:

$$C = B_1P + B_2R + K \quad (2)$$

where  $C$  is the expected cotwin score,  $P$  is the proband score,  $R$  is the coefficient of relationship (1 for MZ pairs, 0.5 for DZ pairs), and  $K$  is the regression constant. The  $B_1$  coefficient represents the partial regression of the cotwin's score on the proband score, and provides a measure of twin resemblance irrespective of zygosity. The  $B_2$  parameter represents the partial regression of the cotwin's score on the coefficient of relationship, and after appropriate transformation of the data provides a direct estimate of  $h_g^2$ . After adjustment of the standard errors of the regression coefficients to correct for the double entry of concordant pairs, the significance of the  $B_2$  parameter provides a statistical test of the extent to which extreme scores are attributable to genetic influences.

*The bivariate case.* A simple generalization of the univariate model facilitates the application of the DF method to bivariate data (e.g., Light & DeFries 1995). Instead of comparing the relative similarity of MZ and DZ twins for the same trait, bivariate analyses examine the relation between the proband's score on the selected trait and the cotwin's score on a second, unselected trait. Therefore, if common genetic influences contribute to the association between HR and PA, the mean PA score of cotwins of DZ probands with HR would be expected to regress more toward the population mean than the mean PA score of cotwins of MZ probands.

The bivariate regression equation is expressed as follows:

$$C_x = B_1P_{HR} + B_2R + K \quad (3)$$

where  $C_x$  is the expected cotwin score on the unselected measure (PA, Digit Span or Orthographic Coding),  $P_{HR}$  is the proband score on the selected measure (HR). In the bivariate model, the  $B_2$  coefficient provides a direct estimate of the extent to which high reading ability is attributable to genetic influences that are also associated with high PA (or Digit Span, Phonological Coding, or Orthographic Coding).

## Results

### *Demographic characteristics*

The overall sample was, on average, from the middle SES range, and of high-average intelligence (Table 1). The latter is consistent with control samples in previous studies published by the CLDRC (DeFries et al. 1997) and may reflect a combination of general demographic characteristics in Colorado and self-selection bias. The ethnic composition of the sample as a whole was 84.8% White, 7.9% Hispanic, 4.0% African American, 1.7% Asian, and

Table 1. Descriptive statistics for individuals in whole sample (same-sex controls,  $N = 700$ )

	Mean	SD
Age	11.53	2.38
SES	3.45	0.91
Gender	47.4% male	
<i>Achievement</i>		
PIAT reading recognition	106.86	8.22
PIAT reading comprehension	109.65	9.16
PIAT spelling	105.03	10.42
Reading composite	1.21	0.83
<i>WISC-R</i>		
FSIQ	113.05	11.00
VIQ	112.78	11.30
PIQ	110.76	12.26
Freedom from distractibility	11.24	2.28
Perceptual organization	11.75	1.83
Verbal comprehension	12.20	1.96
ACID	11.23	1.72
AVID	11.48	1.77
Digit Span	10.90	2.86

Reading composite is a z-score. All other test scores are standardized scores with population means of 100 or 10. SES is given as the level score, according to Hollingshead Index.

1.1% American Indian, and was not significantly different in the samples of individuals used in this study. The mean reading composite score for the whole sample, which is based on the discriminant function analyses described earlier, is 1.21. Since a score of 0 marks the reading disability cutoff, most of this sample is reading well into the average range. The reading scores on each of the PIAT reading subtests corroborates this as the sample means are 5–10 points higher than the norm mean of 100. As explained previously, one twin from each concordant HR pair, as well as cotwins of HR discordant probands were removed from the phenotypic analyses.

High reading (HR) probands did not differ from other normal-range comparison subjects (hereby “controls”) in age or in familial SES (Hollingshead 2-factor Inventory 1975). Because of the way they were selected, the two groups differed on reading measures (the PIAT reading subtests and the

Table 2. Descriptive statistics in control sample by HR status

Measure	Controls ( <i>N</i> = 500)		High reading ( <i>N</i> = 100)		' <i>d</i> '	<i>t</i>
	Mean	SD	Mean	SD		
Age	11.50	2.39	11.60	2.39	0.04	0.55
SES	3.42	0.92	3.51	0.89	0.10	0.64
Gender	53% female		48%female		–	–
<i>Achievement</i>						
PIAT reading recognition	104.62	6.57	116.65	6.17	1.89	19.9***
PIAT reading comprehension	107.74	8.30	118.25	6.10	1.46	13.7***
PIAT spelling	102.32	8.55	116.08	8.02	1.66	18.4***
Reading composite	0.98	0.66	2.53	0.46	2.77	26.7***
<i>WISC-R</i>						
FSIQ	111.29	10.23	120.37	11.16	0.85	9.4***
VIQ	110.51	10.07	121.75	11.54	1.04	12.2***
PIQ	104.04	11.87	114.25	13.45	0.33	3.3**
Freedom from distractibility	10.92	2.16	12.61	2.17	0.78	8.2***
Perceptual organization	11.64	1.78	12.27	2.00	0.34	3.1**
Verbal comprehension	11.84	1.81	13.60	1.86	0.96	11.0***
ACID	10.94	1.61	12.39	1.56	0.92	9.8***
AVID	11.11	1.58	13.00	1.65	1.17	12.8***
<i>Cognitive processes</i>						
Digit Span (STVM)	10.57	2.74	12.31	2.80	0.63	6.6***
Phoneme awareness composite #	1.71	0.25	1.49	0.17	1.05	7.2**
Orthographic coding	–0.17	0.89	0.64	0.79	0.96	9.8***
Phonological decoding	–0.17	0.98	0.79	0.84	1.06	8.3***

\*\* $p < 0.01$ ; \*\*\* $p < 0.001$ . One twin from each concordant twin pair was excluded from these analyses (see data analysis section). Due to transformations to reduce skewness, lower means are indicative of better phonological awareness.

reading composite) as well as on variables correlated with reading skill. The latter included the IQ scales from the WISC-R, and measures of phonemic awareness, short-term verbal memory, phonological decoding, and orthographic coding (Table 2). A comparison of the effect sizes suggests that high reading ability may be associated more strongly with some variables than others. For example, the effect sizes for phoneme awareness, phonological decoding, and orthographic coding are at least a third larger than the Digit

Table 3. Correlational matrix of component processes and reading composite variables

	Phoneme awareness	Orthographic coding	Phonological decoding	Digit Span
Phoneme awareness				
Orthographic coding	0.25			
Phonological decoding	0.63	0.41		
Digit Span	0.39	0.17	0.31	
Reading composite	0.53	0.52	0.57	0.29

All Pearson  $r$  correlations are significant at  $p < 0.001$ .  $N$  for all correlations = 350.

Span effect size, although the latter is still more strongly associated with HR than non-verbal measures such as Perceptual Organization or PIQ.

Correlations between the four component process variables and the reading composite were all highly significant, as was expected given the results of prior research at the low end of the reading skill continuum (Table 3). The correlations among the four component processes showed that phoneme awareness and phonological decoding were the most highly correlated ( $r = 0.63$ ,  $p < 0.001$ ). Orthographic coding, on the other hand, only shared approximately six percent of the variance with phoneme awareness ( $r = 0.25$ ,  $p < 0.001$ ).

The logistic regression analyses indicated that both orthographic coding and phoneme awareness contributed a significant amount of unique explanatory variance in high reading ability. The amount of unique variance accounted for by phonological decoding and by Digit Span did not reach significance in the full model (Model 1 in Table 4). Due to significant shared variance between phoneme awareness and phonological decoding, as well as between phonological decoding and orthographic coding ( $r = 0.41$ ,  $p < 0.001$ ), follow-up logistic regressions were performed that excluded one of the two significant variables, in turn, from the full regression model. The results of these subsequent analyses are listed as Models 2 and 3 in Table 4. When the phoneme awareness variable is removed, all three remaining processes significantly predict reading status. In addition, when orthographic coding is removed from the equation, phonological decoding and phoneme awareness are then significant predictors of reading status, with Digit Span showing a trend ( $p = 0.06$ ). The other two potential combinations (removal of Digit Span or Phonological Decoding) yielded the same results as the full model, and thus are not shown in Table 4.

Table 4. Logistic regression analyses predicting HR status

Model	Variable	B coefficient	Wald statistic	<i>p</i>
1.	Phoneme awareness	-1.03	19.14	0.001
1.	Orthographic coding	1.28	29.52	0.001
1.	Phonological decoding	0.30	1.49	0.22
1.	Digit Span	0.28	2.50	0.11
2.	Orthographic coding	1.15	22.97	0.001
2.	Phonological decoding	0.74	12.12	0.001
2.	Digit Span	0.40	5.78	0.016
3.	Phonological decoding	0.76	10.77	0.001
3.	Phoneme awareness	0.80	10.30	0.001
3.	Digit Span	0.31	3.47	0.062

In Model 1, all four component process variables were entered into the regression equation. In Model 2, phoneme awareness was removed from the initial model. In Model 3, orthographic coding was removed from the initial model.

#### *Concordance estimates for HR in MZ and DZ twins*

The proband-wise concordance rate for HR was higher for MZ twins (54%) than DZ twins (39%); a chi-square test yielded a trend toward significance ( $\chi^2 = 3.12$ ,  $p < 0.08$ ), suggesting that HR status may be partly attributable to genetic influences. The lack of statistical significance may have been due to the high DZ concordance rate. The latter was most likely a result of the relatively lenient cutoff score on the reading composite variable (i.e. +1.0 standard deviations above the mean). This cutoff, as compared to ones at 1.5 or 2.0 standard deviations above the mean, would have increased the proportion of DZ twin pairs where both twins met the threshold for high reading.

#### *Univariate heritability analyses*

Concordance analyses provide only a rough index of the extent to which there are genetic influences that contribute to HR status. The DF multiple regression method provides a more powerful test of the etiology of these associations, and facilitates the comparison of the relative influence of genetic and environmental factors. The mean reading composite scores of the MZ and DZ high reading probands each fell more than one and one-third standard deviations above the mean for the whole sample (MZ  $M = 2.68$   $SD = 0.49$ ; DZ  $M = 2.52$ ,  $SD = 0.45$ ). However, as shown in Table 5, their cotwins had different means, with MZ cotwins regressing back less to the population mean



Table 5. Univariate heritability estimates of reading variables in twin pairs selected for HR

Cotwin measure	MZ cotwins	DZ cotwins	Univariate	
	mean (SD)	mean (SD)	$h_g^2$ (SE)	$t$
Reading composite	1.08 (0.92)	0.57 (0.99)	0.55 (0.22)	2.25*
PIAT reading recognition	0.94 (0.91)	0.57 (0.87)	0.47 (0.20)	2.08*
PIAT reading composition	0.77 (0.89)	0.53 (0.88)	0.40 (0.23)	1.55
PIAT spelling	0.96 (0.82)	0.46 (0.86)	0.64 (0.22)	2.64*

\* $p < 0.05$ ; Means are for standardized scores on each variable. Reported standard errors and  $t$ -values reflect correction of standard errors for double entry.

(1.08 standard deviations above the mean) than DZ cotwins (0.57 standard deviations above the mean). This differential regression is consistent with what would be expected if genetic influences contribute to HR. Consequently, results of the univariate analysis of the heritability of HR in this sample indicated that high reading ability was significantly heritable,  $h_g^2 = 0.55$  (0.22),  $t = 2.252$ ,  $p < 0.05$  (see Table 5).

In order to determine if the three subtests that make up the composite reading score are each contributing to the overall finding that high reading is genetically mediated, three additional univariate multiple regressions were conducted with the extreme group re-selected using each of the three PIAT subtests. As with the reading composite variable, the extreme group was selected using a 1.0 standard deviation above the sample mean cutoff. Table 5 also lists the MZ and DZ standardized cotwin means for each of these analyses, heritability estimates and significance levels. Significant heritability estimates were obtained for the PIAT Reading Recognition subtest,  $h_g^2 = 0.47$  (0.20),  $t = 2.08$ ,  $p < 0.05$ , and the spelling subtest,  $h_g^2 = 0.64$  (0.22),  $t = 2.64$ ,  $p < 0.05$ ; however, the Reading Comprehension subtest heritability estimate, albeit not much lower than reading recognition, did not reach significance ( $h_g^2 = 0.40$  (0.23),  $t = 1.55$ ,  $p < 0.14$ ). Thus, in this sample, high spelling ability and high reading recognition may be influenced by genetic factors to a somewhat greater degree than high reading comprehension. Although this pattern of results could be due to the different reliabilities of the three subtests, a similar pattern of results was obtained by Olson et al. (1994) at the low end of reading ability.

It should be noted that an extension of the DF method also allows for the calculation of the proportion of variance in extreme scores that is due to shared environment effects, rather than genetic effects. However, shared environment effects, which are commonly smaller than genetic effects, could not be reliably computed in this study due to limitations in sample size.

Table 6. Bivariate heritability estimates of HR and component processes in twin pairs selected for HR

Cotwin measure	MZ cotwin	DZ cotwin	Bivariate	
	mean (SD)	mean (SD)	$h_g^2$ (SE)	$t$
Phoneme awareness	0.69 (0.90)	0.30 (0.99)	0.59 (0.28)	1.85*
Orthographic coding	0.56 (0.95)	0.10 (1.11)	0.56 (0.25)	2.02*
Digit Span	0.55 (1.02)	0.15 (1.00)	0.46 (0.24)	1.73*
Phonological decoding	0.54 (0.48)	0.25 (0.71)	0.56 (0.29)	1.75*

\* $p < 0.05$ ; Single tailed  $t$ -test. Means are for standardized scores on each variable. In the case of phoneme awareness, the composite variable was transformed first to reduce negative skew, then standardized. Reported  $t$ -values and standard errors reflect correction of standard errors for double entry.

### Bivariate heritability analyses

The DF method can also be used to determine if the same genetic influences contribute to two traits that overlap phenotypically. The logistic regression analyses suggested that phonemic awareness, phonological decoding and orthographic coding were associated with high reading ability and predicted HR status. The short-term verbal memory measure (Digit Span) contributed less specific variance. In the first analysis, MZ and DZ probands were selected for HR using the same reading composite score cutoff as before. As shown in Table 6, the mean standardized MZ cotwin score for phoneme awareness was more extreme (0.69 standard deviations above the sample mean) than the mean standardized DZ cotwin score (0.30 standard deviations above the sample mean), consistent with what would be expected if the same genetic influences contributed to both high PA and HR. The multiple regression analysis revealed a significant bivariate heritability for high PA and HR,  $h_g^2 = 0.59 (0.28)$ ,  $t = 1.850$ ,  $p < 0.05$ .

Table 6 also lists the standardized mean MZ and DZ cotwin scores for orthographic coding, phonological decoding, and Digit Span when the probands are selected for high reading ability. The multiple regression analyses yielded a significant bivariate heritability estimate for orthographic coding and reading,  $h_g^2 = 0.56 (0.25)$ ,  $t = 2.020$ ,  $p < 0.05$ , a significant bivariate heritability estimate for Digit Span and reading,  $h_g^2 = 0.46 (0.24)$ ,  $t = 1.734$ ,  $p < 0.05$ , and a significant bivariate heritability estimate for phonological decoding and reading  $h_g^2 = 0.56 (0.29)$ ,  $t = 1.75$ ,  $p < 0.05$ . Thus, not only are these component processes able to predict high reading status, but they all share a significant proportion of common genetic etiology with high reading ability.

## Discussion

This study examined the etiology of high reading ability (HR) and its relation to three cognitive components that have been associated with poor reading in dyslexic populations. Individuals who met criteria for HR did not differ significantly from the Control group on age or SES variables, but did demonstrate higher IQ scores on all three subscales of the WISC-R, as well as on phoneme awareness, orthographic coding, and short term verbal memory. Effect sizes suggest that HR is associated more strongly with certain cognitive-linguistic variables, such as phoneme awareness and orthographic coding, than with others (e.g. verbal short-term memory). As would be expected, HR is also more strongly associated with various verbal components of IQ than with non-verbal components of IQ.

In the logistic regression analyses used to predict HR status, phoneme awareness and orthographic coding contributed significant unique explanatory variance when all the variables were entered into the model simultaneously. This is consistent with findings at the low end of the reading ability continuum, where PA and orthographic coding predicted poor reading status and outcome (Olson et al. 1994; Rack, Snowling & Olson 1992). In the full logistic regression model, short-term verbal memory (Digit Span) and phonological decoding did not reach significance. However, when phoneme awareness was removed as a predictor variable, all three of the remaining component processes become significant predictors of HR status. Similarly, when orthographic coding was removed from the equation, phonological decoding and phoneme awareness significantly predicted HR status (with Digit Span exhibiting only a trend). This pattern of results is consistent with the correlations among the variables in question, especially the high correlations between PA, phonological decoding and Digit Span. Although each contributes some unique variance to HR prediction, their shared variance constrains phonological decoding and Digit Span when all are included in the regression. In addition, performance on Digit Span may be influenced by a number of other cognitive factors (e.g. attention, strategy use), reducing its direct relationship with the reading and core reading processes. The overall classification rate of Model 1 is 84.5%.

### *Genetic analyses*

Proband-wise concordance rates were calculated, as a preliminary test of genetic etiology for high reading ability. The concordance rate was significantly higher in MZ twins than in DZ twins, indicating that HR status is significantly attributable to genetic influences. As the DF regression analysis method is a more powerful and versatile method to ascertain the heritability

of high reading and its genetic overlap with other variables, it was used in subsequent analyses. A univariate analysis of the reading composite score indicated that HR is significantly heritable ( $h_g^2 = 0.55$ ). This estimate is almost exactly equal to that for the group heritability estimate in dyslexia ( $h_g^2 = 0.56$ ), recently reported by DeFries and Alarcon (1996), as well as with the heritability estimates of individual differences in reading ( $h^2$  estimates ranging between 0.35 and 0.81). Ancillary univariate analyses examined each component of the reading composite score and found that the PIAT Reading Recognition and Spelling Subtests were each significantly heritable (0.47 and 0.64, respectively) but the heritability estimate of Reading Comprehension (0.40) did not reach significance. Again, the Reading Recognition and Spelling skills on the high end have almost identical heritability estimates as those obtained in the dyslexic samples of the Twin Family Reading Study (DeFries, Stevenson, Gillis & Wadsworth 1991; Olson et al. 1994). These results are consistent with an interpretation that dyslexia and normal variation in reading (both in the middle range as well as on the high end) are on a single distribution of reading ability, and are not due to distinct etiological factors. Although an argument could be made that different sets of genes are acting at each end of the distribution, the bivariate analyses presented next do not support such a conclusion.

The DF bivariate multiple regression model was also fit to the data to assess the genetic overlap between reading and each of the component processes under investigation. The results indicated that HR shares a common genetic etiology with high scores in phoneme awareness, phonological decoding, orthographic coding, and Digit Span. This evidence for a significant amount of shared genetic influence among these underlying cognitive components and high reading ability suggests that the same cognitive phenotype is associated with reading ability at both ends of the reading skill distribution. This weakens further the case for distinct etiologies acting at each end of the reading distribution, as it is more parsimonious to suggest that common genetic influences are acting across the distribution when the same cognitive phenotypes are co-heritable with reading throughout. This account would also be more consistent with a QTL hypothesis that states that reading is a continuous complex behavioral trait that is influenced by a number of genetic loci in the human genome.

It is important to note that heritability does not equal gene identification, and although we have made a strong case for the possibility that the same genetic influences are acting on both ends of the reading continuum, only molecular genetic studies can conclusively prove that this is so. Linkage analyses showing that the same QTLs are associated with poor and skilled reading would be a logical next step, as well as establishing linkage to the

same QTLs with the phonological and orthographic processing variables that underlie reading ability and disability. Investigation of the cognitive phenotypes at the molecular level may be more fruitful in the end, as these variables are undoubtedly more proximal to brain mechanisms than is a complex trait such as reading. Refining the behavioral phenotype by looking at the different potential cognitive mechanisms that lead to reading difficulty can also be beneficial in that it can suggest important relationships among “discrete” behavioral phenotypes in the language learning disability arena.

Although the present study shows that the positive genetic effects that influence reading also positively effect various phonological and orthographic skills, these influences may not be as specific as first assumed. The phenotypic association between high reading and various indices of cognitive ability leaves open the possibility that these genetic influences may have a broader effect. Bivariate genetic analyses like those used in the present study could be implemented to corroborate whether these phenotypic relationships are indeed a product of shared genetic etiology.

Lastly, it has been recently shown that the heritability of dyslexia, or reading disability, varies as a function of IQ (Wadsworth, Olson, Pennington & DeFries 2000). Wadsworth and colleagues showed that the heritability of reading disability was higher in those subjects with higher Full Scale IQ scores than in those subjects with lower IQ scores. In the case of low reading ability, this result makes intuitive sense. As Wadsworth et al. (2000) explain, children with higher IQ scores conceivably benefit from a more enriched environment that promotes reading ability. Furthermore, if the positive environmental effects are more homogeneous in families of higher IQ children, then the variability in poor reading scores among these children is likely to be disproportionately due to heritable influences, as compared to children with lower IQ whose shared environment may be more variable. Preliminary analyses conducted with the current data set to ascertain the heritability of high reading ability as a function of IQ showed a similar pattern of results. Although the overall sample size was not large enough to create higher and lower IQ groups, an extension of the DF model was used to test the differential heritability of high reading ability as a linear function of Full Scale IQ (residualized for the influence of reading). Consistent with the findings of Wadsworth et al. (2000) with the reading impaired population, the results of these analyses indicated that high reading ability is also more heritable in higher IQ subjects.

This result can seem counterintuitive, as one might have predicted a higher heritability estimate for high reading ability in those subjects whose extremely high reading score was more discrepant from their IQ (i.e. lower IQ subjects with very high reading scores). In fact, these “hyperlexic” readers

(they are reading at a higher level than would be expected given their cognitive level) have a lower heritability estimate, indicating that environmental factors help explain a greater proportion of the variance in high reading scores. Conversely, as Wadsworth et al. (2000) argued in their paper, higher IQ individuals may be more homogeneous with respect to environmental factors, leading to proportionately greater genetic influence on the variance in high reading ability. Although these results are suggestive, it should be noted that the current sample did not have as wide an IQ range as Wadsworth et al. (2000). Therefore, these results should be considered preliminary until they are replicated in a larger sample of twins with high reading ability who have a wider range of IQ scores.

*Limitations of the current study and implications for future research*

There are various limitations of the current study that should be taken into account when interpreting these results. First, the relatively small sample size of the present study reduces the reliability of the computed heritability estimates, due to larger standard errors. The current findings should be replicated in larger samples in order to increase the confidence and generalizability of these results. This is especially necessary given the preliminary evidence for differential heritability of high reading as a function of IQ, which questions the generalizability of these results to those with high reading ability and lower IQ. The limitation in sample size also precluded using an extension of the DF method to estimate the influence of shared environment on high reading ability. Given that shared environmental effects are generally smaller than heritable effects, the power in this study was insufficient to obtain reliable estimates of shared environment influences on the variability in high reading scores. Comparing the relative proportion of genetic and shared environmental effects in reading disabled and high reading ability samples would be of interest, in order to begin to understand how genetic and environmental interactions might differ in different segments of the population. In the same vein, analysis of gender and age effects on the heritability of high reading ability would also be important. Given the possible difference in prevalence of reading disability in males and females, it is important to look for gender differences in the obtained results as well. Lastly, it would be interesting to investigate reading ability in a sample of truly hyperlexic readers (e.g. as is sometimes found in high functioning autism or Asperger's syndrome), as this might reveal different etiological and cognitive mechanisms by which good reading is achieved.

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