
Causal Models of Reading Disability: A Twin Study

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The genetic and environmental relationships among measures of phoneme awareness, naming speed, Intelligence Quotient (IQ), and reading performance were investigated in 623 identical and fraternal twin pairs tested in the Colorado Learning Disabilities Research Center. A Cholesky decomposition analysis of these measures provided evidence supporting the double deficit hypothesis that difficulties in phonological processing and naming speed both contribute to reading disability. Additionally, the model revealed marginally significant genetic and significant non-shared environmental relationships between IQ and reading independent of naming speed and phoneme awareness. Thus a more complete causal model of reading disability should include IQ as well as measures of phonological processing and naming speed.

The well-established relationship between phonological skills and reading performance (Gayan & Olson, 2001, 2003; Goswami & Bryant, 1990; Griffiths & Snowling, 2001; Manis et al., 2000; Pennington, 1991; Pennington et al., 2001; Share, 1995; Siegel, 1993; Stanovich & Siegel, 1994; Torgesen et al., 1997) has led to the formulation of a phonological core model, which argues that deficits in phonological processing are the primary cause of reading disability. Thus, if this model were veridical, most individuals with reading disability would be characterized by difficulties in phonological processing and deficits in any other cognitive processes would be less salient.

While the phonological core model is widely accepted, a competing double deficit hypothesis has also been proposed (Wolf & Bowers, 1999). The double deficit hypothesis postulates that reading disability may be caused by deficits in phonological processes, naming speed, or both. These two potential causes, while related, should be relatively independent of each other. One possible test of the double deficit hypothesis is whether naming speed accounts for phenotypic variance in reading independent of phonological processing (McBride-Chang & Manis, 1996; Pennington et al., 2001). Although several studies have found that naming speed explains unique variance in reading performance (Bowers & Swanson, 1991; Catts et al., 2002; Compton et al., 2001; Manis

et al., 2000; McBride-Chang & Manis, 1996; Neuhaus & Swank, 2002), others have failed to find a unique effect of naming speed (Ackerman et al., 2001; Pennington, et al., 2001; Wagner et al., 1994).

In a previous phenotypic analysis of a sample of twin pairs tested in the Colorado Learning Disabilities Research Center (CLDRC), Tiu et al. (in preparation) found support for the double deficit hypothesis. Using structural equation modeling, it was found that naming speed explained unique variance in reading in groups of normal readers, low achieving readers, IQ-discrepant readers, and readers meeting both low achieving and IQ-discrepancy criteria. This effect was independent of verbal IQ, performance IQ, and phoneme awareness. Although the double deficit hypothesis appeared to explain the data better than the phonological core model, the double deficit hypothesis was found to be incomplete. Both verbal and performance IQ scores explained significant variance in reading independent of both naming speed and phoneme awareness in all four groups. It was concluded that causal models of reading disability needed to incorporate measures of IQ.

While most previous research has been limited to the phenotypic relationships among reading-related measures and IQ, several studies have addressed the genetic and environmental origins of the relationships between these measures. For example, two studies found substantial genetic correlations (.53–.70) for group deficits in phoneme awareness and word reading (Gayan & Olson, 2001; Olson et al., 1994). Moreover, other studies have reported that 50–75% of the genetic influences in common between word reading and phoneme awareness are independent of IQ (Gayan & Olson, 2003; Hohnen & Stevenson, 1999).

The genetic and environmental etiologies of the relationship between naming speed and reading have also been investigated. Davis et al. (2001) analyzed twin pairs in which at least one member had a positive school history of reading problems. Using DeFries and

Received 21 January, 2004; accepted 6 April, 2004.

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Fulker's (1985) multiple regression models, they found significant bivariate heritabilities between naming speed and reading deficits as measured by a discriminant function score derived from subtests of the Peabody Individual Achievement Test (PIAT), indicating that reading deficits covary genetically with naming speed. Davis et al. (2001) analyzed data from both twins with a school history of reading difficulties in at least one member of each pair and unaffected control twins from the Colorado Learning Disabilities Research Center using structural equation modeling and found significant phenotypic and genetic correlations between naming speed and reading for both groups. However, both phenotypic and genetic correlations were higher in the sample with reading difficulties.

Several studies have examined the genetic and environmental basis of the relationship between reading and IQ. Three studies found that the phenotypic relationship between reading and IQ was largely genetic in origin (Brooks et al., 1990; Cardon et al., 1990; Wadsworth & DeFries, 2003). However, Alarcon and DeFries (1997) reported that the genetic correlation between IQ and reading was greater in a group of normally achieving readers than in a group with reading difficulties. They also found no shared environmental correlation between IQ and reading. Additionally, Wadsworth et al. (2000) showed that reading deficits were more heritable in twins with IQ scores above 100 than in those below 100.

There has been only one previous attempt to test the double deficit hypothesis from a genetic and environmental perspective. Compton et al. (2001) analyzed the genetic and environmental influences on reading and naming speed after removing the effects of phoneme awareness by fitting a Cholesky decomposition model to twin data. While not a test of group deficits, the analysis was conducted in a twin sample with a large proportion of individuals with school histories of reading difficulties. Results obtained from three separate analyses showed that, among this select sample of children with reading difficulties, individual differences in naming speed shared significant genetic influences with general word reading, non-word reading, and orthographic reading independent of phoneme awareness, thereby supporting the double deficit hypothesis.

In the present study, the analyses of Compton et al. (2001) will be extended to test the unique genetic relationship between IQ and reading. Additionally, a more general measure of reading performance will be used as opposed to the word reading measures used by Compton et al. (2001). It is hypothesized that there will be a significant relationship between naming speed and reading independent of phoneme awareness. Additionally, the present analysis will test the validity of the addition of IQ to the model. We hypothesize that there will be significant genetic and environmental influences of IQ on reading

performance independent of both naming speed and phoneme awareness.

Methods

Participants

The present study utilized data from twin pairs tested in the Colorado Learning Disabilities Research Center (CLDRC; DeFries et al., 1997). To reduce the possibility of ascertainment bias, participants were recruited from 27 school districts in the state of Colorado without regard to reading status. School records were used to identify every twin pair in a school. Parental permission was then sought to examine the school records for evidence of reading problems; for example, low reading achievement test scores or referral to a reading therapist because of poor reading performance. Those twin pairs in which at least one member had a positive school history of reading problems were invited to complete a battery of tests.

The sample analyzed in the present study included 342 (169 male, 173 female) monozygotic (MZ; identical) and 281 (154 male, 127 female) same-sex dizygotic (DZ; fraternal) twin pairs in which at least one member of the pair had a positive school history of reading disability. The mean age of the sample was 11.5 ± 2.61 (range 8.0 to 18.7). The zygosity of same-sex twin pairs was determined using selected items from the Nichols and Bilbro (1966) questionnaire. When zygosity remained in doubt, twin pairs were genotyped using DNA markers.

Measures

All participants were administered an age appropriate test battery of cognitive, academic achievement, and language measures. The test scores analyzed in this report include Full Scale IQ from the Wechsler Intelligence Scale for Children — Revised (WISC-R; Wechsler, 1974); a discriminant function score (DeFries, 1985) derived from the Reading Recognition, Reading Comprehension, and Spelling subtests of the Peabody Individual Achievement Test (PIAT; Dunn & Markwardt, 1970); a phoneme awareness composite; and a composite designed to test naming speed.

The phoneme awareness composite was comprised of a phoneme transposition task (Pig Latin), a phoneme deletion task, and the Lindamood auditory conceptualization test (LAC; Lindamood & Lindamood, 1979). The phoneme transposition task required the participant to move the first phoneme from the beginning of a word to the end of the word and then add the sound /ay/ to the end. For instance, the participant would change the word "can" into "an-cay". In the phoneme deletion task, the participant was asked to remove a specified phoneme from a spoken non-word. For example, the participant may be asked to say "cran" without the /r/ sound. The Lindamood auditory conceptualization test used colored blocks to represent phonemes. The participant

moved the blocks in accordance with changes in sequences of sounds spoken by the tester.

Naming speed was assessed using the numbers, letters, colors, and pictures subtests of the rapid automatized naming task (RAN; Denckla & Rudel, 1976). The administration differed from that typically used, however. Typically, the participant is asked to identify orally 50 items without any time limit and time to complete is the score of interest. In the alternate version used in the present study, each subtest had a time limit of 15 seconds and the score of interest was the number of correctly identified items. When the standard and alternate versions of the RAN task were compared in a subsample of twins, the alternate version accounted for significantly more variance in several reading measures (Compton et al., 2002).

All measures used to make the composites were standardized across all participants after controlling for age, age squared, gender, and age they began school. Composites were calculated as the average of the measures that comprised the composite. An examination of the four composite measures for normality revealed distributions reasonably close to normal. The largest skewness was for the distribution of phoneme awareness (-0.47 to -0.54). While this skewness was significant, possibly due to the large sample size, visual inspection showed that the phoneme awareness distribution was very close to normal.

Analysis

Structural equation modeling was used to analyze the covariance structure of the twin data. First, a phenotypic Cholesky decomposition analysis was used to test hierarchically the unique effect of naming speed on reading after controlling for phoneme awareness. This model also tested the unique effect of IQ on reading performance controlling for phoneme awareness and naming speed. Rather than analyze the covariance structure of the total sample collapsed across twin types, data from both members of the twin pairs divided by zygosity were analyzed separately and all paths were constrained to be equal across the two groups.

The phenotypic Cholesky (see Figure 1) is analogous to a hierarchical multiple regression analysis where phoneme awareness, naming speed, and intelligence are entered sequentially and reading is the dependent variable. Squaring the paths from the F1, F2, and F3 factors to reading estimates the change in the multiple R^2 due to the addition of the corresponding variable to the model. Squaring the path from F4 to reading estimates the amount of variance in reading that is not accounted for by the predictors.

A Cholesky decomposition was also employed to estimate the genetic, shared environmental, and non-shared environmental contributions to the variance of the measures and the covariance among the measures (Figure 2). Estimates of the proportion of variance in each measure due to genetic or environmental effects were equal to the sum of the squared standardized

path coefficients from all the specific and common factors. For example, the heritability of reading performance was estimated as: $a_{41}^2 + a_{42}^2 + a_{43}^2 + a_{44}^2$. Phenotypically standardized genetic and environmental correlations (e.g., $b_x \times r_{g(xy)} \times b_y$) were estimated from the sum of the products of the paths from each of the common genetic or environmental factors to the two measures. For instance, the phenotypically standardized genetic correlation between reading performance and naming speed was estimated from Figure 2 as $(a_{21} \times a_{41}) + (a_{22} \times a_{42})$. Genetic and environmental correlations between two measures may then be estimated by dividing their phenotypically standardized genetic or environmental correlation by the product of the square roots of the two heritabilities or environmentalities: for example, $[(a_{21} \times a_{41}) + (a_{22} \times a_{42})] / [h_{NS} \times h_{READ}]$.

Additionally, the Cholesky decomposition analysis facilitates genetic and environmental tests of the double deficit hypothesis. The unique relationship between naming speed and reading can be calculated as the correlation between naming speed and reading based only on the second common factor that does not load on phoneme awareness. For example, the unique genetic relationship was estimated as $a_{22} \times a_{42}$. Similarly, the unique relationship between IQ and reading can be calculated as the correlation between IQ and reading based only on the third common factor that does not load on phoneme awareness or naming speed. For instance, the unique non-shared environmental relationship between IQ and reading was estimated as $e_{33} \times e_{43}$. Finally, each of these unique effects may be divided by the total correlation to determine the proportion of the shared variance that is unique. For example, the proportion of the additive genetic relationship between naming speed and reading that is independent of phoneme awareness was calculated as $[a_{22} \times a_{42}] / [(a_{21} \times a_{41}) + (a_{22} \times a_{42})]$.

Results

Phenotypic Cholesky

Results of the phenotypic Cholesky are shown in Figure 1. Every path differed significantly from 0. Therefore, naming speed shared a significant amount of variance in common with reading performance independent of phoneme awareness. The proportion of the shared variance between naming speed and reading performance that is independent of phoneme awareness was: $[f_{22} \times f_{42}] / [(f_{21} \times f_{41}) + (f_{22} \times f_{42})] = 58\%$. The addition of naming speed accounts for a change in the multiple R^2 of 0.07 (f_{42}^2).

IQ also had a significant amount of variance in common with reading performance independent of both phoneme awareness and naming speed. The observed correlation between IQ and reading performance was: $(f_{31} \times f_{41}) + (f_{32} \times f_{42}) + (f_{33} \times f_{43}) = .42$. The proportion of the shared variance between reading performance and IQ independent of both naming speed and phoneme awareness was: $[f_{33} \times f_{43}] / [(f_{31} \times$

$\chi^2(54) = 58.826$

AIC = -1.174

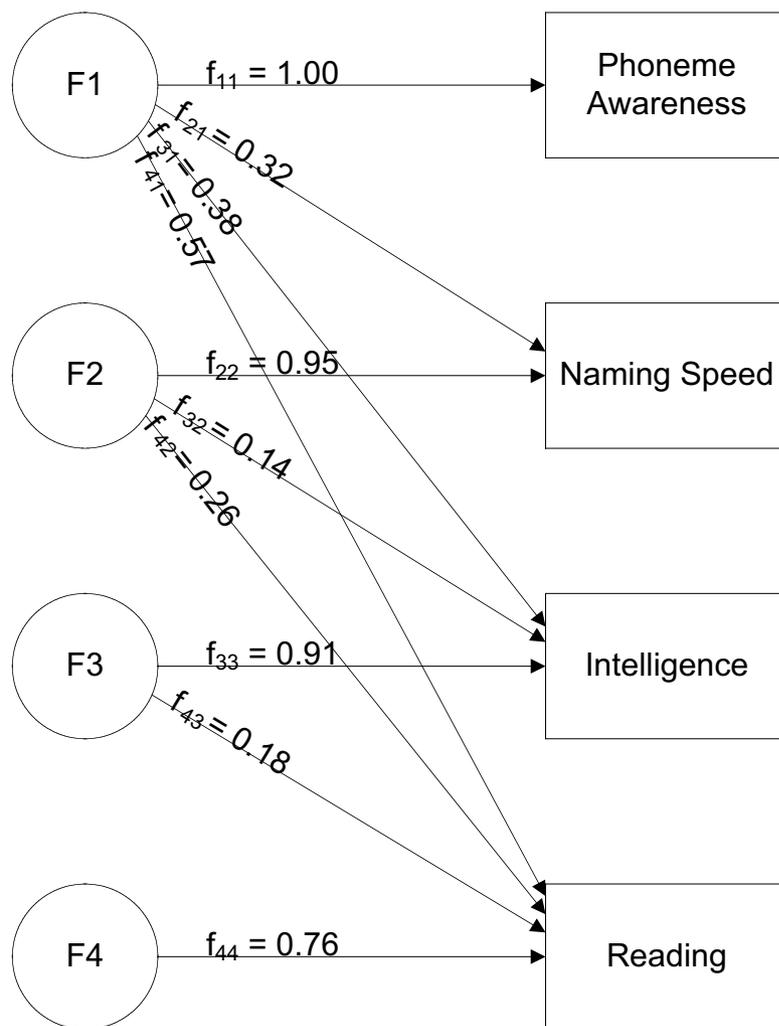


Figure 1
Results from the phenotypic Cholesky model.

$f_{41} + (f_{32} \times f_{42}) + (f_{33} \times f_{43}) = 39\%$. The addition of IQ accounts for a change in the multiple R^2 of 0.03 (f_{43}^2). However, squaring the path from F4 to reading indicates that less than half of the phenotypic variance in reading performance is accounted for by phoneme awareness, naming speed, and IQ in this sample.

Genetic and Environmental Influences

Results of the full genetic and environmental Cholesky analysis are shown in Figure 2. The C3 and C4 factors are omitted because paths c_{33} , c_{43} , and c_{44} were all estimated as 0. The full model shows strong genetic influences on the variance of each measure and the covariances between the measures. Shared environmental influences on the variance and covariance were rather small. Non-shared environmental influences on

the variance of each measure were also substantial, but less so on the covariance among the measures.

The full model fit the data well (Table 1). Models dropping the common E (Model 2) or all A factors (Model 4) fit significantly worse than the full unconstrained model. However, all the C paths could be constrained to 0 without significant worsening of fit (Model 3). The estimates of h^2 , c^2 , and e^2 for the full model are shown in Table 2. More than 50% of the variance in each of the four variables was attributable to genetic factors, whereas 10% or less of the variance was attributable to shared environmental factors. Table 2 also shows the phenotypically standardized genetic and environmental correlations (below the diagonal) and the unstandardized genetic and environmental correlations (above the diagonal) between the

Table 1

Model Comparisons for Genetic and Environmental Cholesky

Model	χ^2	df	AIC	$\Delta\chi^2$	df	p
1. Full	27.48	42	-56.52			
2. No E common factors	141.80	48	45.80	114.32	6	< .001
3. No C	30.15	52	-73.85	2.67	10	> .500
4. No A	227.38	52	123.38	199.90	10	< .001

Note: AIC = Akaike's Information Criterion

Models 2, 3, and 4 are compared to the Full Model

four measures. Moreover, as indicated by a comparison of the phenotypically standardized correlations presented in Table 2, 56%–100% of the phenotypic correlations were due to genetic influences.

As shown in Figure 2, more than half of the shared genetic covariance between naming speed and reading performance was independent of phoneme awareness: $[(.69 \times .29) / [(.31 \times .56) + (.69 \times .29)]] = 54\%$. The proportion of the genetic correlation between reading performance and IQ that is independent of both naming speed and phoneme awareness was $[(.68 \times .13) / [(.44 \times .56) + (.04 \times .29) + (.68 \times .13)]] = 26\%$. Most of the non-shared environmental relationship between reading performance and naming speed was independent of phoneme awareness: $[(.63 \times .06) / [(.12 \times .18) + (.63 \times .06)]] = 64\%$. Finally, the proportion of the non-shared environmental correlation between IQ and reading performance that is independent of phoneme awareness and naming speed was $[(.47 \times .12) / [(.09 \times .18) + (.09 \times .06) + (.47 \times .12)]] = 72\%$. Although the non-shared environmental correlations were small, most of the

non-shared environmental correlations between reading performance and naming speed as well as between reading performance and IQ were independent of phoneme awareness.

The path from the A2 factor to reading (a_{42}) represents the genetic relationship between naming speed and reading independent of phoneme awareness. Constraining this path to 0 led to a substantial deterioration of fit ($\Delta\chi^2 = +17.53$, $df = 1$, $p < .01$, Δ Akaike's Information Criterion [AIC] = +15.53) compared to the full model. Similarly, the path from A3 to reading represents the genetic relationship between IQ and reading independent of both phoneme awareness and naming speed. Constraining this path to zero led to an increase in χ^2 of 2.32 with 1 degree of freedom ($p = .13$) and an increase in AIC of 0.32 compared to the full model. While the χ^2 does not indicate a significant worsening of fit, the AIC increase suggests that including IQ in the model yields a slightly better fit.

Discussion

The present study assessed the phenotypic, genetic, and environmental etiologies of individual differences in reading performance in a sample of twin pairs ascertained for reading difficulties. Further, we examined whether measures of IQ would improve the prediction of reading performance based on a double deficit model which hypothesizes that reading disabilities are due to deficits in phoneme awareness and naming speed. The results of the phenotypic Cholesky analysis provided support for the double deficit hypothesis. More than half of the variance shared between naming speed and reading performance was independent of phoneme awareness. However, consistent with Tiu et al. (2004), the phenotypic results also supported the importance of IQ for the prediction of reading, even after taking phoneme awareness and naming speed into account. About 40% of the covariance between IQ and reading was independent of naming speed and phoneme awareness. However, less than half of the variance in reading performance was accounted for by phoneme awareness, naming speed and IQ in this model.

While the changes in the multiple R^2 for reading performance due to naming speed and IQ were

Table 2Genetic and Environmental Influences on IQ and Reading-related Measures^a

	PA	NS	IQ	Read
Genetic				
PA	0.71	0.41	0.54	0.67
NS	0.26	0.57	0.26	0.59
IQ	0.37	0.16	0.66	0.50
Read	0.47	0.38	0.34	0.72
Shared Environmental				
PA	0.01	-0.74	-0.99	0.93
NS	-0.01	0.01	0.61	-0.94
IQ	-0.02	0.02	0.10	-0.85
Read	0.01	-0.01	-0.03	0.01
Non-shared Environmental				
PA	0.29	0.19	0.18	0.37
NS	0.07	0.41	0.22	0.21
IQ	0.05	0.07	0.24	0.33
Read	0.10	0.06	0.08	0.26

Note: PA = Phoneme Awareness, NS = Naming Speed

^a h^2 , c^2 , and e^2 estimates based on the full model are listed on the main diagonal.Phenotypically standardized correlations are below the diagonal. Estimates of r_g , r_c , and r_e are above the diagonal.

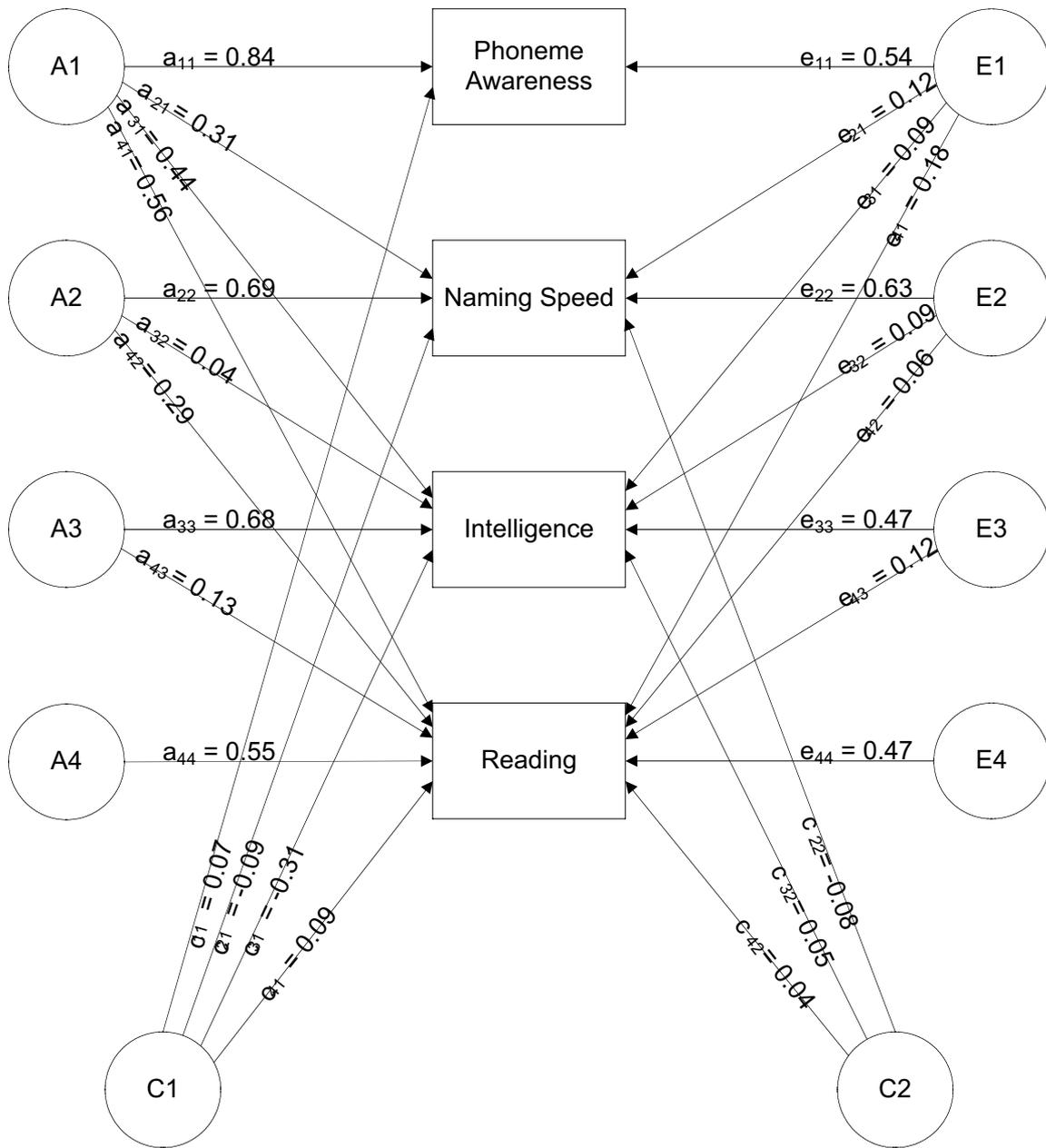


Figure 2
Results from the full genetic and environmental Cholesky model.

relatively small, these results do not necessarily imply that these two predictors are unimportant. As is typical with hierarchical regression analyses, the importance of a single predictor relative to the others is not best estimated by the change in the multiple R^2 , as shared predictive variance is attributed to the independent variable that comes first. Rather, the change in R^2 is a test of the significance of the addition of the variable to the model and is dependent on the order of entry (Tabachnick & Fidell, 2001).

As expected, the results obtained from the genetic analysis mirrored the phenotypic analysis in many

respects. Evidence of a genetic relationship between naming speed and reading independent of phoneme awareness provides further support for the double deficit hypothesis. However, these results differed slightly from those of Compton et al. (2001). In the present study, the genetic variance shared between naming speed and reading was more independent of phoneme awareness. The percentage of the genetic variance shared between naming speed and reading performance that was independent of phoneme awareness was nearly twice as high (54% vs. 30%) as that reported by Compton et al. (2001). This difference

may be due to the different reading measures used in the two studies. Compton et al. (2001) used word reading only (measured by PIAT word reading and a time-limited word reading task), whereas a composite of PIAT word reading, reading comprehension, and spelling was used in the current study. Thus, naming speed may be a more unique predictor of this composite of word reading, reading comprehension, and spelling than of word reading alone.

The results of the genetic analysis also support the importance of IQ scores in the prediction of reading performance. About 25% of the genetic correlation between IQ and reading was independent of phoneme awareness and naming speed. However, this unique effect was only marginally significant. Most of the large genetic correlation (.50) was mediated by the other two predictors, especially phoneme awareness. This may imply that the correlation between intelligence and reading performance is largely due to the relationship between intelligence and phoneme awareness. In fact, the genetic correlation between IQ and phoneme awareness (.54) was higher than the genetic correlation between IQ and reading (.50). Further, about 70% of the non-shared environmental correlation of .33 between IQ and reading performance is independent of phoneme awareness and naming speed. Overall, these results suggest that IQ scores should be incorporated into causal models of reading performance.

Estimates of h^2 , c^2 , and e^2 indicate that individual differences in reading-related measures are largely determined by genetic and non-shared environmental factors. Moreover, all estimates of shared environment could be fixed to 0 without significant worsening of fit. Although small in magnitude, fixing the common paths from the non-shared environmental factors to 0 led to a significant worsening of fit. These results indicate that not all the effects of phoneme awareness, naming speed, and IQ on reading performance were genetic in origin.

Examination of the genetic and environmental correlations reveals that all four measures are strongly related genetically. Phoneme awareness had the highest genetic correlations with reading performance (.67), followed by naming speed (.59), and then IQ (.50). There were also smaller, but significant, non-shared environmental correlations between the four measures. The largest non-shared environmental correlations were between reading performance and phoneme awareness (.37) and between reading performance and IQ (.33).

In conclusion, the results of the present study support the double deficit hypothesis as a model of reading disability. Naming speed accounts for additional phenotypic, genetic, and non-shared environmental variance in reading performance independent of phoneme awareness. Additionally, the results indicate the importance of IQ in the prediction of reading disability. Although much of

the covariance between IQ and reading performance was mediated by phoneme awareness and naming speed, about 40% of the phenotypic variance and 25% of the genetic variance between IQ and reading performance was independent of the other two measures. Additionally, more than 70% of the non-shared environmental correlation between reading performance and IQ was independent of the other two measures. Although phonological processing is highly related to reading, a more complete model of reading disability should also include measures of naming speed and IQ.

Acknowledgments

This work was supported in part by program project and center grants from the National Institute of Child Health and Human Development (HD-11681 and HD-27802), and Dr. Tiu was supported by a Training Grant from the National Institute of Mental Health (MH16880). The contributions of staff members of the Colorado school districts from which our sample was ascertained, and of the families who participated in the study, are gratefully acknowledged.

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