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Genetic and environmental bases of reading and spelling: A unified genetic dual route model

TIMOTHY C. BATES[1], ANNE CASTLES[2], MICHELLE LUCIANO[3], MARGARET J. WRIGHT[3], MAX COLTHEART[1], and NICHOLAS G. MARTIN[3]
[1]*Macquarie Centre for Cognitive Science, Macquarie University, Sydney, Australia;* [2]*Department of Psychology, University of Melbourne, Melbourne, Victoria, Australia;* [3]*Genetic Epidemiology Laboratory, Queensland Institute of Medical Research, Brisbane, Australia*

Abstract. We develop and test a dual-route model of genetic effects on reading aloud and spelling, based on irregular and non-word reading and spelling performance assessed in 1382 monozygotic and dizygotic twins. As in earlier research, most of the variance in reading was due to genetic effects. However, there were three more specific conclusions: the first was that most of the genetic effect is common to both regular and irregular reading. In addition to this common variance evidence was found for distinct genes influencing the acquisition of a lexicon of stored words, and additional genetic effects influencing the acquisition of grapheme–phoneme correspondence rules. The third conclusion, from a combined model of reading and spelling, is that reading and spelling have a common genetic basis. Models that did not distinguish lexical and non-lexical performance fit significantly worse than dual route genetic models. An implication of the research is that models of reading, whether connectionist or dual-route, must allow for the genetic independence of neurological processes underlying the decoding of non-words and irregular words.

Key words: Dyslexia, Genetics, Reading, Spelling, Twins

In this paper we describe and test a model of the genetics of reading that suggests that reading of irregular words and of non-words is influenced by different genes: a genetic extension of the dual route model of reading (Coltheart, Rastle, Perry, Langdon, & Ziegler, 2001). This model is extended to spelling, and, finally, to a joint genetic model of reading and spelling. The predictions of these models are tested using a large adolescent twin sample, and the fit of the genetic dual route model is examined for both reading and spelling. It appears that both the correlation and the differences between lexical and non-lexical reading measures are highly genetic. Subsequently, we test genetic models of reading that do not make a basic distinction between lexical and non-lexical processing and consider the implications of the genetic

data for connectionist triangle models of reading (Plaut, McClelland, Seidenberg, & Patterson, 1996).

We begin by briefly describing the dual route model of reading before a brief introduction to genetic modeling in a cognitive context, followed by constructing genetic dual route models for reading and spelling.

The dual route behavioral and computational model of reading

Dual route models of reading postulate a system of modules which support two simultaneous routes to reading aloud: a lexical route, and a non-lexical route as shown in Figure 1 (Coltheart, 1978; Coltheart, Curtis, Atkins, & Haller, 1993; Coltheart et al., 2001; Morton & Patterson, 1987). Both routes begin with (shared) systems for using visual stimuli to

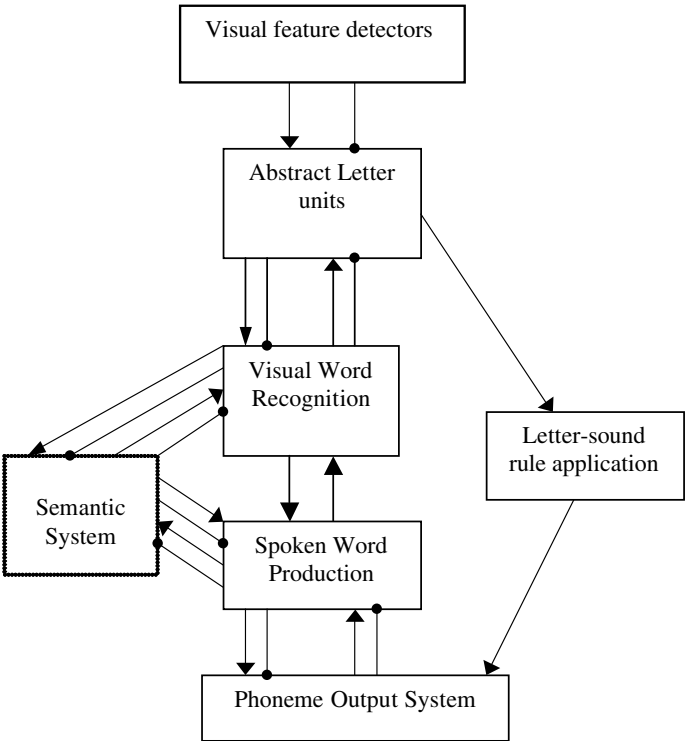

Figure 1. Dual route Cascaded Model of reading aloud (Coltheart et al., 2001). *Note*: Arrowed connections represent excitatory links and ball-ends show inhibitory connections. In this context, excitatory connections act to increase activation in their target, while inhibitory connections act to decrease target activity.

construct letter representations. In the lexical route, this letter array is used to access a lexicon or memory store of local representations of previously seen written words. In the case of the non-lexical route, no previous experience of the word string is required; instead, a rule-based process is used to convert the input graphemes into phonemes. Both routes generate a phonemic output that is directed to common systems for generating phonology and speech. The quite different processes used by these two routes allow an easy assessment of their functioning by asking subjects to read out-loud irregular words and non-words. While both routes are obligatorily activated by any written input, the non-lexical route alone can correctly derive the phonology of novel strings and non-words such as "slint". Non-word skill is thus diagnostic of the non-lexical route as the lexical route can only read words that are present in the lexicon. Similarly irregular words such as "colonel" can only be read correctly via the lexical route, as grapheme–phoneme correspondence rules will regularize the phonology to KOLONEL.

In the next section we briefly outline the concepts of behavior genetic modeling and then describe a genetic model in which the two routes for reading are modeled in terms of a source of shared genetic variance as well as two separate gene effects, one for each route.

How may genetic effects on behavior be modeled and tested?

To turn behavioral research into behavior genetic research, the behavior (or phenotype as it is known in genetics) must be assessed in a genetically informative sample. That is, subjects must differ in their genetic and environmental similarity to each other in ways known to the researcher. Such a design allows researchers to move beyond simply observing that a trait appears to be "familial" – to run in families, and to model this familial effect in terms of distinct components due to the genes which family members share, the environment which they share, and, finally, to unshared non-genetic effects, including measurement error.

Several genetically informative designs are possible, usually taking advantage of natural manipulations of environment (as found in adoption studies) or genetic similarity within a family (as in the twins of the present study), or both (for instance twins adopted out). The power of twin designs arises from the fact that twins may be either monozygotic (MZ: developing from a single fertilized egg and therefore sharing all of their genes) – or dizygotic (DZ: developing from two fertilized eggs and therefore sharing on average 50% of their genes, the same level of genetic similarity as found in non-twin siblings). These known differences in

genetic similarity, together with a testable assumption of equal environments for MZ and DZ twins (Bouchard & Propping, 1993) creates the basis for the twin design for exploring the effects of genetic and environmental variance on a phenotype (Neale & Cardon, 1992). The assumption of equal environments has been tested: for instance in cases where parents believe their twins to be non-identical when in fact they are genetically MZ, studies of a range of psychological traits indicate that these children remain as concordant as MZs raised by parents who "treated them as identical" (Kendler, Neale, Kessler, Heath, & Eaves, 1993).

Because twins raised in their biological families share the family environment of their co-twin, and have only a random similarity with the environment of other twin pairs, and because the genetic similarity of MZ and DZ twins differs in a known quantitative fashion (100% in MZ, 50% in DZ), the variance in any trait measured in a population of MZ and DZ twins can be analyzed in terms of genetic effects, effects of the environment which are shared by twins, and residual unshared environmental effects, including measurement error (Neale & Cardon, 1992) as shown in Figure 2. These variance components are commonly abbreviated as "A" for additive genetic effects, "C" for common or shared environmental effects (effects common to siblings within a family, and varying between families), and "E" for the residual variance not otherwise explained, and comprised of unique environmental effects and measurement error. This includes environmental events experienced by only one twin: for instance accidents affecting one twin, and differences between the peer-groups experienced by the twins.

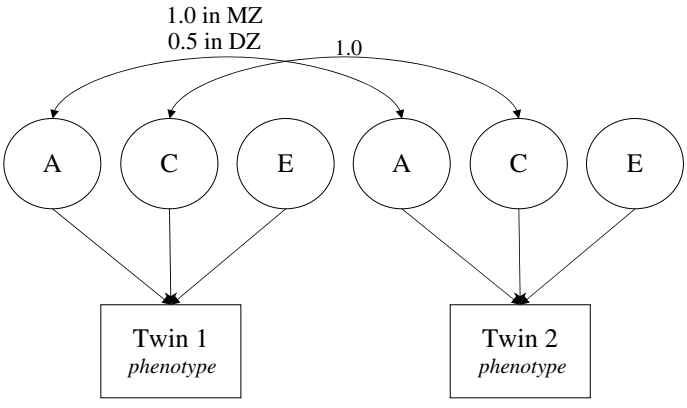

Figure 2. ACE Structural Model for calculating the trait variance due to Additive genetic (A), shared environmental (C), and unique environment (E).

Additive genetic effects represent the effects of genes whose joint effect on the trait is simply the sum of their individual effects. Since MZ twins are genetically identical, the correlation for additive genetic effects between MZ twins is 1. For DZ twins, the correlation for additive genetic effects is .5. This additive genetic variance is distinguished from a fourth variance component (''D'') not shown in Figure 2, representing non-additive genetic effects including dominance (where some alleles or versions of a gene only exert their effect when present on both chromosomal copies in a cell) and epistasis (interaction effects between genes). Adding data from separated twin pairs, half-siblings, or similar pairs of relatives would un-confound non-additive and shared environmental effects but as our study only involved twins raised together, only one of D or C can appear in a given model (Neale & Cardon, 1992), and we preferred C in all cases as the raw correlations were not suggestive of dominance effects (which would be indicated by MZ correlations more than twice the magnitude of the DZ correlations). Example variables often argued to explain ''C'' environmental effects include home environment factors such as family reading habits, SES, and schools. It is worth noting that just as dominance can decrease the similarity of sibs in a family, other effects exist which make DZ twins more genetically similar to each other than the .5 value which is assumed in the model. The most common case of this effect occurs when parents select each other on the basis of similarity to each other. This ''assortative mating'' bias causes parents to be more genetically similar than would occur by chance. As a consequence, assortative mating causes children to be more genetically similar to their siblings than is implied by a simple .5 gene sharing coefficient. This excess genetic similarity in DZ twins will appear in models as a common environment effect.

Given the ACE model shown in Figure 2, researchers can determine what proportion of variance in a trait is heritable, versus the proportions which are due to shared environment or unshared environment. So long as dominance is not present, simple formulae for calculating these values for standardized variables are as follows:

$$\mathrm{A} = 2 * (r_{\mathrm{mz}} - r_{\mathrm{dz}})$$

$$\mathrm{C} = (r_{\mathrm{mz}} - \mathrm{A})$$

$$\mathrm{E} = 1 - r_{\mathrm{mz}}$$

where r_{mz} and r_{dz} are simply the correlations of the trait in MZ and DZ twins respectively (Jinks & Fulker, 1970; Plomin, DeFries , McClearn, & McGuffin, 2001).

Beginning in the 1970s, research transitioned to explicitly modeling the values of A, C, and E within a maximum likelihood framework (Martin & Eaves, 1977). While computationally much more complex, benefits of this approach are manifold, and modeling tools such as Mx (Neale, Boker, Xie, & Maes, 2002) have made the new techniques relatively accessible. Critically, for studies such as the present analyses of reading and spelling, modeling approaches generalize to the multivariate case where more than one phenotype is under study. Phenotypic correlations between traits can be explicitly decomposed into shared and independent sources of genetic and environmental variance. Simultaneous equations, established by the relationships among MZ and DZ co-twins predicted from genetic theory are applied to the raw data. Models are fitted in which the total variances (and covariances between variables) are parameterised in terms of A, C and E, with the expected covariance for MZ twin pairs expressed as Variance (A) + Variance (C), while for DZ twin pairs it is .5 * Variance (A) + Variance (C).

In the present case, we use multivariate modeling to test for separate genetic bases for lexical and non-lexical phenotypes, and, finally, in explaining both reading and spelling within a single model. By modeling the data, confidence intervals can be calculated for the values of each of A, C, and E, and the significance of each variance component can be tested by dropping it from the model to test for the change in fit. The modeling approach also allows researchers to evaluate *a priori* theoretical models by empirically examining the goodness of fit of the theoretical model (such as those described below for reading) by comparing them to an a-theoretical ''saturated'' solution.

The saturated solution for any set of variables simply refers to a model of the standardized test scores that has a χ^2 goodness of fit value of zero, i.e., a system of paths that fit the data perfectly. Theories of the traits in question will usually not predict that nature follows this saturated case, but rather will hypothesize a more meaningful and usually more simple system in which certain connections are dropped, combined, or held equal. Such models may fit less well, but will also release some degrees of freedom and they can thus be tested against the saturated model by evaluating the loss of fit (if any) in the proposed model versus the saturated solution in a χ^2 test against the degrees of freedom gained. This saturated model against which nested, more theoretically interesting models can be evaluated is the Cholesky genetic factor decomposition (Neale & Cardon, 1992).

The nature and function of the Cholesky is most easily understood graphically and a decomposition for three variables is shown in Figure 3. As can be seen, the Cholesky contains a latent variable for each of the

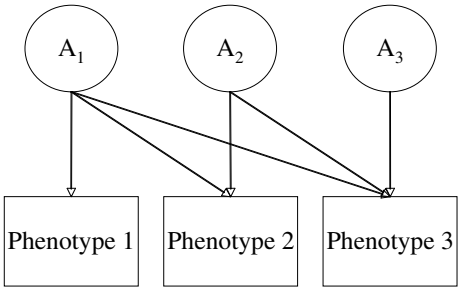

Figure 3. A Cholesky Decomposition for 3-traits producing a saturated solution, i.e., a model that in standard terms explains all the variance in the model, and therefore has a χ^2 of zero. *Note*: "A" is additive genetic effect. For the sake of clarity, only additive genetic effects are shown here, but the model would typically contain the same structure for C (common environment) and E (unique environment) variables for each trait.

measured traits in the model. Each latent variable is connected not only to the trait beneath it, but to all traits to its right. In this way, an attempt is made to model the variance of each new trait using the latent variables to its left, and the residual variance of the trait is explained using the latent variable above the trait. This solution is readily specified and is easily extended to as many traits or phenotypes as have been measured. Once the Cholesky has been calculated, it can itself be simplified in an exploratory manner by dropping latent variables or paths to see if these significantly alter the goodness of fit, or, as in the present case where a definite theoretical model is proposed, this model can be directly tested.

In summary, genetically informative samples and modeling are powerful tools to understand the environmental and genetic causes of behaviors. Much of the evidence for hypothesized causal agents in development such as exposure to print are based on observed familial similarities. As experimental interventions which could potentially test such hypotheses are often impossible, samples in which family environment and genetic similarity are disentangled are often the only method for testing such hypotheses. In addition to establishing the heritability of traits, advanced genetic modeling can test whether cognitive tasks such as reading contain discrete genetic components, allowing researchers to test and compare different cognitive models. It is to such a model that we now turn.

Genes and reading: A genetic dual route model of reading and spelling

In this section, we develop a genetic dual route model for reading, which is then extended to spelling, and finally, a combined dual route architecture

of reading and spelling is proposed. As discussed above, there is strong support for a cognitive dual route model of reading. There is also strong support for a genetic basis to reading. Therefore it is possible that the cognitive dual route model will map on to a genetic dual route model. However this is not a given. First, there might be no individual differences in the genetics of the components to reveal the different systems for the behaviour. Second, it is quite possible for two cognitive processes to be behaviourally distinct, but to have only a single genetic cause. Therefore the model we develop here makes novel predictions that can be falsified independently of behavioural support for the dual route model of reading.

A genetic dual route model of reading

As can be seen in Figure 1, within the computational DRC (Dual-route Cascaded) model of reading (Coltheart et al., 2001) irregular and non-word reading performance share all stages from initial visual feature detection through to abstract letter unit extraction, and then also share a final common pathway through the phoneme output system. Any heritable variance in the performance of these components will impact on both irregular and non-word reading. In between these stages, the two routes to reading diverge. Heritable variance on lexical storage or access, or on the development and execution of grapheme–phoneme conversion will affect only irregular word or non-word reading skill, respectively. Given the very high heritability of reading outlined in the introductory passage (Bates et al., 2004; Gayan & Olson, 2003; Tiu, Wadsworth, Olson, & DeFries, 2004) we predicted that the observed dissociations of non-word and irregular word performance would be due in part to genetic effects, and that the dual route architecture itself would have a substantial genetic basis. Our prediction of a genetic dual-route architecture was supported to some degree by the findings of Gayan and Olson (2003) of independent genetic variance accounted for by measures of lexical (orthographic) and non-lexical (phonological) reading, respectively, although in this research orthographic processing was assessed using visual choice measures (speeded word-choice tasks using either printed homophones (“bear|bare”) or pseudohomopone pairs (“rain|rane”). The genetic dual route model is shown graphically in Figure 4.

Behavior genetic studies of young readers suggest that shared environment exerts a significant influence on early reading (Byrne et al., 2005). However, by age 8–9 and above the effects of shared family environment reduce to zero, and are replaced with increases in genetic effects and unshared environment (Bates et al., 2004; Gayan & Olson,

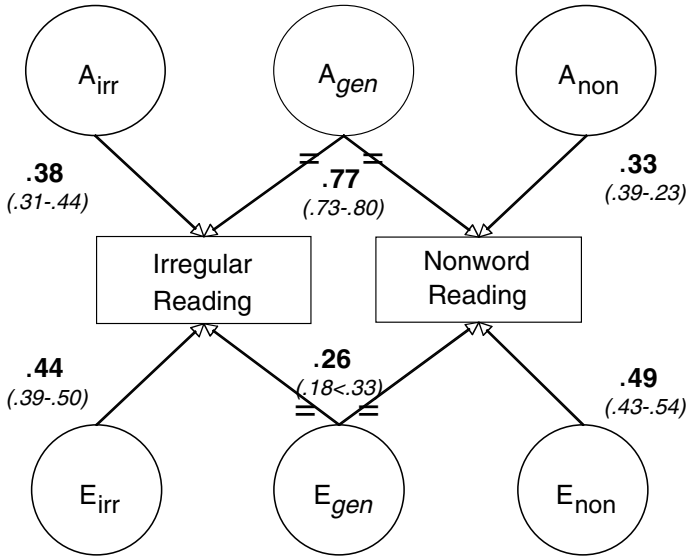

Figure 4. A Dual Route Genetic Model of Reading. *Note*: A_{irr} is Additive genetic effects for lexical processing; A_{gen} is general Additive genetic effects for reading; A_{non} is Additive genetic effects for non-lexical processing. E variables are the non-shared environmental effects on reading. C (common or family environment) is not shown as it was dropped without significantly affecting the fit of the model. Path coefficients are the standardised path loadings. The values in brackets beside each path loading show the 95% confidence intervals for the effect-size. An "=" sign on a path indicates that that it has been constrained to be equal to the adjacent path for model identification.

2003; Tiu et al., 2004). For this reason, while it is possible to implement a dual-route architecture for the influences of shared and for non-shared environment, we did not predict significant effects of shared environment in our adolescent sample on any of our measures.

Dual route genetic model of spelling

While spelling has received considerably less attention than has reading (both in terms of cognitive modeling, and, more especially, at a genetic level), there is a historical precedent for dual route models of spelling. The first such approach was suggested soon after the dual route model of reading was developed (Coltheart, 1978; Ellis, 1982), and more recently dual route models of spelling have begun to be implemented computationally (Houghton & Zorzi, 2003). Like Ellis (1982) and Houghton and Zorzi (2003), we suggest that spelling occurs in dual routes that mirror the processes of reading (see Figure 5).

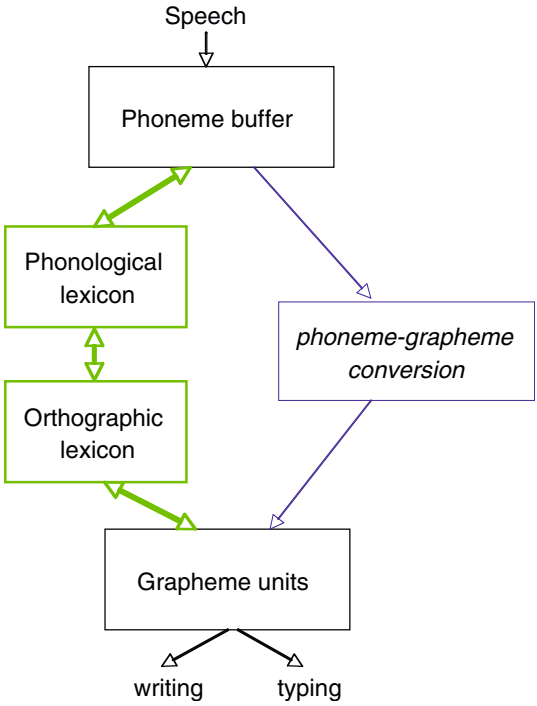

Figure 5. Dual route cognitive model of spelling.

The genetics of spelling have been addressed only in a few studies to our knowledge, and non-word spelling has not been studied, nor have spelling and reading been analysed jointly before. The first report on the heritability (Stevenson, Graham, Fredman, & McLoughlin, 1987) supported a higher heritability for spelling than for reading (.75 when intelligence was controlled). Subsequently DeFries, Stevenson, Gillis, and Wadsworth (1991) reported modest heritability for a test of spelling, again not differentiating lexical and non-lexical processes. Beyond these heritability studies, spelling has also been a phenotype in molecular genetic studies, again often showing more substantial relationship than reading (Grigorenko et al., 2001; Nothen et al., 1999; Petryshen et al., 2001; Schulte-Körne et al., 1998). Our model will therefore add to our confidence in estimating the heritability of spelling, and move beyond preceding studies to examine potential of separate genetic effects on lexical and non-lexical spelling.

In building a testable genetic model of spelling, it is important to note that the spelling of any word type involves common processes for converting speech into a phoneme sequence, and, again, a shared stage of translation into letter strings for output. Thus a general genetic factor will

be required, if only to explain these modules external to the separate routes postulated to implement lexical lookup or phoneme–grapheme conversion. As in reading, we predict the existence of distinct genes underpinning lexical activity and phoneme–grapheme conversion, as shown in Figure 6.

A unified model for the genetic basis of reading and spelling

We have suggested a dual route genetic architecture underpins both reading and spelling. A further question is how these models are related. Is reading merely similar to spelling, but implemented under independent genetic control, or does the correlation of reading and spelling in both cross-sectional and developmental studies reflect a deeper biological connection? In producing a joint model of reading and spelling we have adopted the extreme but parsimonious approach and suggest that spelling relies on exactly the same genetic basis that underlies reading, and that spelling is essentially reading "driven in

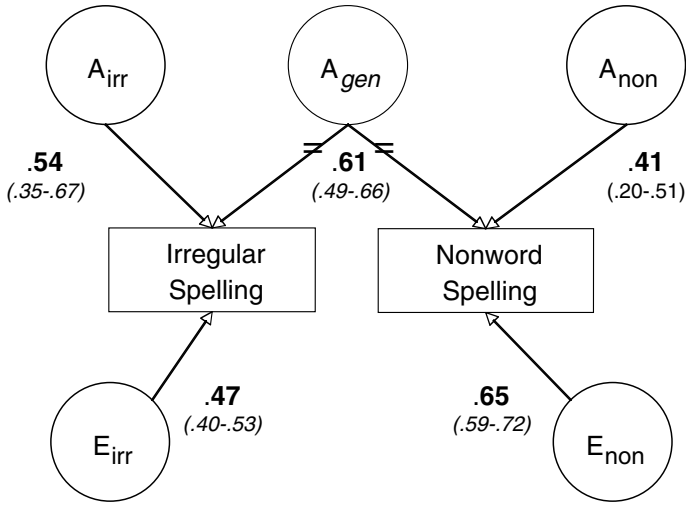

Figure 6. A Dual Route Genetic Model of Spelling. *Note*: A_{irr} is additive genetic effects for lexical processing; A_{gen} is general additive genetic effects for reading; A_{non} is additive genetic effects for non-lexical processing. E variables are the non-shared environmental effects on reading. There was no significant evidence for a general E effect in spelling. C (common or family environment) is not shown as it was dropped without significantly affecting the fit of the model. Path coefficients are the standardised path loadings. The values in bracketed range beneath each path loading shows the 95% confidence interval for the effect-size.

reverse'' with no need for additional genetic effects. This genetic model is shown in Figure 7.

Environmental effects on reading and spelling

Just as we have developed a genetic dual route model, so too, environmental effects on the dual route architecture can be postulated and tested. As described above, Behavior Genetics distinguishes two kinds of environmental effect: non-genetic familial effects such as sharing the same family background, upbringing, SES, and, probably, schools and even teachers, and non-shared effects, which includes all remaining variance: both causal but unshared effects such as accidents affecting one twin, and also measurement error.

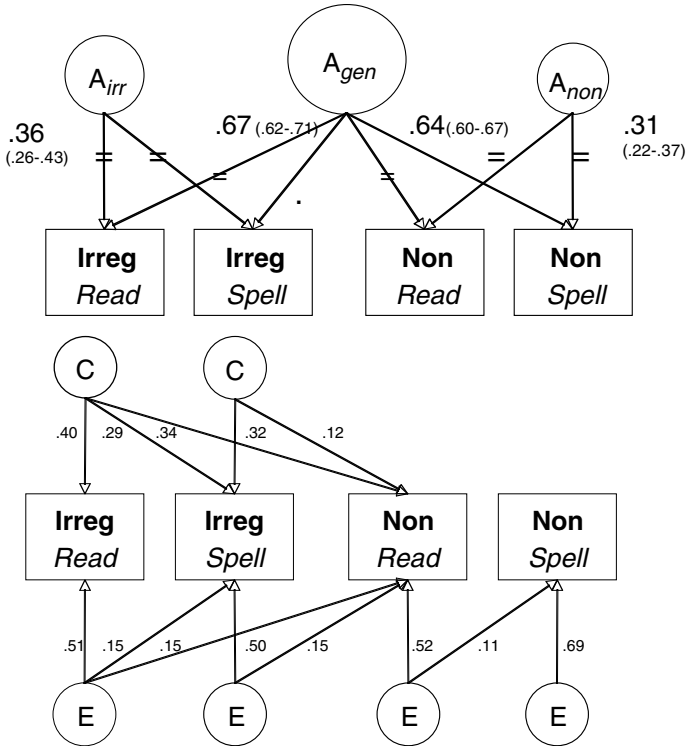

Figure 7. Unified Genetic Model of Reading and Spelling. *Note*: A_{gen} is the general additive genetic effect on reading, A_{irr} and A_{non} are specific effects for irregular and non-word reading respectively. C is Common environment effect; E is Unique environment. Numbers on each path are path coefficients. 95% confidence intervals are shown beside each path estimate for A effects. E and C variance was modelled using a Cholesky genetic factor structure.

Just as in the case of building the genetic dual route architecture, shared and non-shared environmental influences can be postulated which would exert a general effect on reading of whatever kind. For instance, an effective teaching programme may well improve both sight vocabulary and provide practice with grapheme–phoneme correspondence rules via phonics. Individual differences in the exposure to such effective teaching methods will create environmental variance reflected in performance on both non-word and irregular word reading. In addition to such general effects of the environment, specific practice with phonics should improve non-word reading without enhancing irregular word reading, which is dependent on exposure to specific non-regular lexical items and vice versa.

Summary

Each of the models proposed has the virtue of being easily falsified if any of the following strong predictions fail: In both reading and spelling, a genetic dual route architecture will be required to achieve a good fit to the data, supporting three sources of genetic variance: shared elements common to both routes, and two distinct sets of genes active in producing the computationally distinct processes of lexicalised storage and access, versus rule-based grapheme–phoneme conversion and its inverse, phoneme–grapheme conversion for spelling. It is predicted that, while this model will fit the data as well as the perfect or saturated model, dropping any genetic element of this dual route structure will significantly decrease fit. While not an essential prediction from dual route models, as most of the sample is at an age where their skilled cognitive performance no longer reflects different family environments, we predict that it will be possible to drop common environment without significant loss of fit. Unique environmental variance includes measurement error as well as opportunistic exposure to events helping or hindering one or more elements critical to reading. We therefore expect to retain much of the unique environment structure.

Testing the models

Method

Subjects

Reading and spelling data were collected from 1382 Australian twins (mean age 17.6 yrs, SD = 3). Informed consent was obtained from all participants and parents prior to testing. More than 95% of

great-grandparents were identified as being of northern European ancestry, mainly from Britain and Ireland. Parental education measures were in line with Australian norms, with 18.4% reporting up to 10 years of schooling, 24.4% with 8–10 yrs schooling and apprenticeship or diploma, 33.4% with 11–12 yrs school (with or without apprenticeship or diploma), 23.8% with Technical/teachers college or university graduate/ postgraduate degree. Parental occupational classifications were 16.5% Managers & Administrators/Professionals, 11.7% Associate professionals, 33% (tradespersons & related workers/ advanced clerical & service workers), 29.2% (intermediate clerical, sales & service workers/intermediate production & transport workers), 9.5% (elementary clerical, sales & service workers/labourers & related workers). Further details are reported elsewhere (McGregor et al., 1999; Wright & Martin, 2003; Wright et al., 2001). Participants received a thank you letter and two complimentary movie tickets for their participation.

Measures & Procedure

Regular word, irregular word and non-word reading were assessed using the CORE (Bates et al., 2004), a 120-word extended version of the Castles and Coltheart (1993) test with additional items added to increase the difficulty of this test for an older sample (all test materials available in Bates et al., 2004). All items were presented in mixed order to avoid blocking effects.

Regular and irregular-word spelling was tested by verbally presenting a subset of 18 regular words and 18 irregular words from the CORE. Items were presented in mixed order to avoid blocking effects and the subjects' oral spelling of the words was recorded. The items were one-to-one matched on frequency, number of phonemes and grammatical class. Because it can be difficult for subjects to unambiguously identify the desired phonetic target from verbally presented non-words, non-lexical spelling skills were further assessed by having subjects spell the words given in the irregular spelling test under a different set of instructions requiring them to "regularize" the spellings. Subjects were told:

> "Next I am going to read 18 of those words back to you again. This time, I would like you to give me a spelling for each word so that someone who had never seen the word could still pronounce it properly. For example, if I gave you the word "yacht" you could say "Y O T". Do you understand?"

Each word was then presented verbally, and the letter string used for spelling was recorded. Words were repeated on request. Regular reading

and regular spelling were not used in the present analyses which focussed on modelling the difference between irregular and non-word processing.

Procedure

An approach pack was mailed to each participant, and, within 2 weeks of receipt of this pack, a trained researcher interviewed each participant over the telephone. After the rationale for the study was explained and consent obtained for testing, including a re-test if required, subjects completed the reading test or arranged a subsequent time to take the test. Each subject was asked if they had opened the envelope prior to testing (none reported opening the envelope before testing). Four sib-pairs (3 Male DZs and 1 male MZ pair) required re-testing (data from these pairs were anomalous in that one twin was discordant from the sample mean or from their co-twin by 4–5 *z*-score units on a single test, against a background of otherwise normal performance. These eight subjects were followed up for retest to exclude the possibility that their initial results were due to errors of data-transcription or misunderstanding of test instructions) and their retest data, in each case much closer to their co-twin, are included in the present analyses. The reading tests, mixed regular and irregular word spelling test, and regularization-spelling tests were then given, in that order. Testing took less than 15 min, with each test given in accordance with the instructions outlined above, and responses were recorded and entered into a database. Telephone testing for reading ability has not been used commonly, but when used has been found to be effective: for instance it has been used extensively in the Twins Early Development Study (TEDS; Trouton, Spinath, & Plomin, 2002). Test scores on each of the three reading subtests and three spelling tests were calculated as a simple sum of correct items. Prior to analysis, all raw data were log-odds transformed and normalized.

Analyses

Multivariate genetic models were fitted to the individual observations (i.e., raw data method) by the method of maximum likelihood in Mx (Neale et al., 2002) to decompose the total variance in the observed measures into the most parsimonious values of A, C and E to explain the observed pattern of MZ and DZ twin correlations and estimate the size of the genetic and environmental parameters (Neale & Cardon, 1992). It is computationally more demanding to model the raw observational data

(rather than summary variance covariance matrices). The benefit of this approach is that fixed effects such as age and sex can be modeled in the means (as might occur in a regression analysis), while simultaneously the A, C and E effects are modeled in the variances and covariances. Viewing the model in this way is didactically valuable: over time as the causal gene alleles and environmental effects are discovered, they too will be modeled in the means until the variance/covariance element of the model contains only measurement error and all the causal elements are specified as fixed effects.

Results

Descriptive Statistics and Assumption tests: Numbers of twins in each group, and the age- and sex-corrected maximum likelihood twin pair correlations (and 95% confidence intervals) are shown Table 1. It was clear from the much larger MZ than DZ correlations that all of the tests were highly heritable (model testing suggested that around 75% of the variance in each measure was due to additive genetic effects), and these univariate analyses have been reported previously (Bates et al., 2004). No differences in the means for the reading and spelling measures were observed between first and second born twins (testing for effects of birth order) or across zygosity or sex for any of the variables. A significant sex difference ($P < .01$) in the variance of irregular word reading was found with males showing larger variance (.93) than females (.82). This variance difference was accommodated in the genetic modelling by estimating separate male and female variances. Table 2 shows the summary heritability and

Table 1. Table of sex and age-corrected maximum likelihood twin pair correlations with 95% confidence intervals.

	Reading		Spelling	
Twin type	Irregular words	Non-words	Irregular words	Non-words
MZF (116)	.70 (.60–.77)	.69 (.60–.76)	.76 (.68–.82)	.61 (.49–.69)
MZM (97)	.76 (.66–.82)	.70 (.60–.78)	.74 (.64–.80)	.51 (.34–.64)
DZF (122)	.38 (.21–.52)	.28 (.08–.44)	.33 (.14–.48)	.21 (−.01–.39)
DZM (127)	.57 (.45–.67)	.45 (.31–.56)	.49 (.36–.60)	.12 (−.04–.27)
DZFM (107)	.37 (.20–.51)	.36 (.18–.51)	.44 (.29–.57)	.12 (−.12–.33)
DZMF (122)	.44 (.28–.57)	.32 (.13–.48)	.36 (.19–.50)	.36 (.20–.49)

Note: MZF is monozygotic female twins; MZM is monozygotic male twins; DZF is dizygotic female twins; DZM is dizygotic male twins; DZFM is dizygotic opposite-sex twins, female born first; DZFM is dizygotic opposite-sex twins, male born first. The number of pairs of each twin type are shown in brackets after the twin type label.

Table 2. Heritability estimates for reading spelling.

	Trait			
	Irregular read	Irregular spelling	Non-word reading	Non-word spelling
A^2	.58	.54	.55	.51
C^2	.16	.19	.13	.00
E^2	.26	.27	.32	.49

common and unshared environment effects for each trait, derived from Figure 7.

Genetic Analyses

Reading: Our theorized dual route genetic model of reading is shown in Figure 4, together with the path loadings following reductions of the full ACE dual-route model. This model is equivalent to a Cholesky and therefore has an optimal fit. For our purposes the empirical question is whether either or both of the specific genetic effects can be dropped without loss of fit. Attempts to drop any of the additive genetic factors significantly reduced model fit, confirming the likelihood that distinct genes exert control over the lexical and non-lexical systems. As can be seen in Figure 4, the best fitting dual-route ACE model could be simplified by dropping the common environmental parameter to form an AE model without any significant change in fit when compared to the saturated model ($\chi^2(3) = 4.02$, $P = .26$). This suggests that all of the familial aggregation for reading ability is explained by additive genetic effects of a little over 70% of the variance in reading (heritability or h^2 can be calculated for each variable by adding the squared additive genetic path loadings falling on that variable). The remaining sources of variance were attributable to unique environmental effects including measurement error.

Spelling: Testing of the spelling model proceeded identically to the analyses described above for reading, and the final path model for spelling is shown in Figure 6. The preferred model for spelling mirrored that found for reading. As in the case of the reading model, common environmental parameters could be dropped without any significant change in the log-likelihood ($\chi^2(3) = 1.37$, $P = .71$). The path coefficients indicated strong genetic control for both lexical and non-lexical spelling with heritabilities of 66% and 54% respectively.

Joint reading and spelling genetic model

The joint or unified model of reading and spelling proposed is shown in Figure 7. This model has a number of constraints which assume that reading and spelling share a common genetic basis: a single general genetic factor is used to model both spelling and reading; and the genetic specific variable for lexical and non-lexical skill are used to explain performance on both reading and spelling. Comparison of the hypothesized joint dual route model of reading and spelling to the Cholesky indicated that this highly reduced model was a good fit to the data, showing no significant reduction in fit from the perfectly fitting saturated model ($\chi^2(13) = 1.65$ $P < .64$). For this model, analysis suggested that some common and unique environmental influences could be dropped without significant change in the log-likelihood from the saturated ACE model ($\chi^2(10) = 7.76$ $P < .65$). The significant C & E pathways are shown in Figure 7. The retained C effects appear to model shared environment effects on reading, especially for irregular words, however no hypotheses were made as to the basis of these family effects. An alternative genetic model with a common factor for all four variables, and two separate factors for the two reading tasks (Irreg and Non) and the two spelling tasks was tried, but did not fit as well as the preffered model shown in Figure 7 ($\chi^2(13) = 19.79$ $P < .05$). This further constrains the genetic architecture of reading and spelling. As well as emphasising the similarity of reading and spelling, it supports the need for genetics effects specific for irregular and non-word processing, rather than for other possible divisions of the variance.

Single-route model of reading

A benefit of model testing is that simpler models, which might be thought of as more parsimonious, can also be tested, to see if their parsimony comes at the cost of significant loss of fit. The simplest genetic model of reading that can be proposed is shown in Figure 9. In this model, irregular and non-words are seen as representing two outcomes of a single reading process. Because the paths to each type of word are not constrained to be equal, the irregular and non-word tests can be viewed as potentially testing a continuum of word ''difficulty'', as reflected in different path loadings from the single genetic source. This model is nested with the saturated model above, and thus the fit can be tested empirically. In fact, the model shown in Figure 9 fits extremely poorly, with a highly significant loss of fit from that shown by the hypothesised dual route model ($\chi^2(1) = 43.3$ $P < .0001$).

Discussion

The results supported the dual route genetic models proposed here, both for reading and spelling separately, and for both systems jointly. The heritability values obtained and the lack of effect for shared environment are broadly in line with other recent research. However the main impact of the new modeling lies in the support for distinct genes as a basis for much of the observed dissociability of the lexical and non-lexical systems. The unified genetic model suggests, furthermore that at a genetic level, there is no distinction between reading and spelling: in dual-route terms, the genetic basis for lexical or storage-based information processing systems and for rule-based assembly systems is shared across reading and spelling processes. It would be invaluable to extend this data set to include language processing to begin to identify the points of contact between variance in language impairment and variance in reading disorder or ability, and we are undertaking this extension.

It is notable that distinct influences were found for non-word and irregular word reading. The only other behavior genetic study of individual differences examined word and non-word reading differences (Gayan & Olson, 2003) reported a genetic correlation of .97 for word and non-word latent traits. They found no significant genetic influence on word reading, independent from non-word reading, a finding also supported for group (rather than individual) differences (Gayan & Olson, 2001). Gayan and Olson point out that as their word-reading measure contains both regular and exception words, a significant effect of lexical access ought to have been found, should this have independent genetic basis, as we predict. Gayan and Olsson assessed orthographic processing as well as word recognition, and this trait showed a lower genetic correlation with phonological decoding (.82). This lower genetic correlation and significant independent genetic effects follow the same pattern as found in the current study. Further work is required to understand the differences between these reports and the present model, if in fact they point to different conclusions.

It is possible that the result reported here is not due to the lexical status of the irregular words, but to their low frequency, relationship to IQ, or dependency on semantic processes. This brings us neatly to a comparison of the present results with single-route models of orthographic-to-phonological processing, as these too depend on semantics to form a "second route" to reading in the absence of successful O $\rightarrow$ P processing. To further explore this role for genetics in informing cognitive modeling, in the second part of this paper we outline alternative cognitive architectures for reading which do not assume the dual route distinction of

lexical and non-lexical processes, and attempt to model these systems within the twin behavior genetic paradigm.

Connectionist models of reading

In the section above, evidence was reported for a genetic basis of the cognitive distinction between lexical and non-lexical processing, both in reading and in spelling. As noted at the beginning of the paper, however, there is a class of single-route models which suggest that all reading (and perhaps all spelling), whether of previously learned words or novel words is conducted within a single distributed processing system. In this section these models are described in detail and attempt is made to model this approach with the genetically informative twin sample.

One of the principle distinctions between connectionist and dual-route models of reading is that the former do not contain separate modules supporting lexical and non-lexical reading nor do they predict the existence of distinct processing methods (lexical access versus application of rules) as being involved in reading (Bates et al., 2004; DeFries, Fulker, & LaBuda, 1987; Gayan & Olson, 2003). However, the main impact of the new modeling lies in the support provided for distinct genes as a basis for much of the observed dissociability of the lexical and non-lexical systems. The unified genetic model suggests, furthermore, that, at a genetic level, there is no distinction between reading and spelling: in dual route terms, the genetic basis for lexical or storage-based information processing systems and for rule-based assembly systems is shared across reading and spelling processes.

The analyses to date shed light on the ways in which genetics can inform cognitive modeling. For example, simpler models of reading in which non-word and irregular word reading are performed by a single system, with these word types simply placing more or less stress on this single system, are falsified by the data. The genetic evidence suggests that non-words and irregular words, while they do share common influences, also reflect two additional sources of genetic influence: genetic effects which influence only non-word reading or only irregular word reading.

As noted earlier, in contrast to dual route models, there exists a class of models of reading aloud which do not distinguish between lexical and non-lexical processes, broadly referred to as connectionist models (Plaut et al., 1996; Seidenberg & McClelland, 1989). Proponents of these models argue that all reading occurs via the activation of a set of weighted connections between sets of units representing words in a distributed fashion. Neither a lexicon nor rules are explicitly built into connectionist models, and both

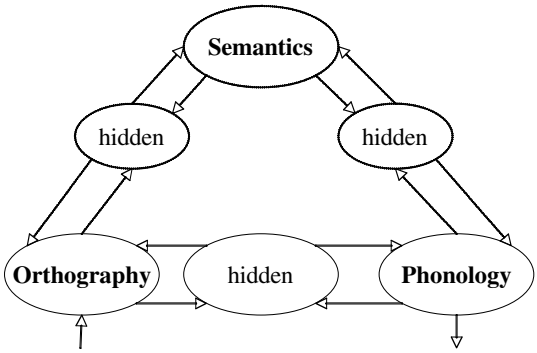

Figure 8. The standard (Seidenberg & McClelland, 1989) connectionist "triangle" model of reading aloud.

non-words and words can be read via a single orthographic-to-phonological translation system. A set of semantic units is also included, making up the typical "triangle" architecture presented in Figure 8.

What implications, then, do the present results have for this class of model? As shown by the very poor fit of the single route model (see Figure 9), it seems that, based on the genetic data, it is insufficient to propose that the reading aloud of words and non-words can be accounted for entirely in terms of a single O $\rightarrow$ P translation system. So it seems that the present data require that these models implicate additional nodes in the network to explain the independent genetic influences that are found on irregular and non-word reading. This requirement does not pose a particular difficulty for these models: additional nodes have indeed already been recruited in explaining the behavioral dissociations of non-word and irregular word reading that are found in acquired dyslexia (Plaut et al., 1996). Such dissociations have been accounted for by referring to the differential effects of, for instance, semantic processing in supporting irregular word reading compared to non-word reading (Plaut et al., 1996).

Explaining dissociations between non-word and irregular reading in this way makes certain auxiliary predictions about performance, in particular very poor semantics in acquired dyslexics (Plaut et al., 1996). However, more pertinently for the present purposes, this pattern of co-morbid deficits can be tested very acutely in genetic models. The hypothesis that two genetic routes are required to fit the present data because, for instance, semantic variables influence the reading of irregular words but not non-words suggests that what we have called a genetic factor for lexical processing is in fact a genetic factor for semantics. This prediction can be explicitly tested within a twin sample by separately

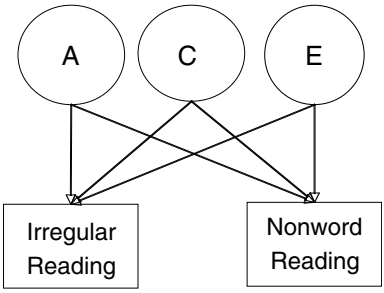

Figure 9. Single-route connectionist model of reading: Approach 1. In this model, there is a single source of each of Genetic (A), Common environment (C), and Unique (E). Each affects both types of word stimulus, though the paths are not constrained equal, and thus effects could be much stronger for one class of words than the other, allowing for a differences in difficulty or demand of the irregular and nonword items, as has been suggested.

assessing semantic performance in addition to irregular word and nonword reading (for instance by verbally asking subjects to define words). If the connectionist account is correct, this measured semantic performance should load on the same genetic factor that explains irregular word reading. If on the other hand, semantics is a cognitive module independent of lexical processing, then a good fit to these three variables should require a new, additional genetic factor.

Similar predictions can be made for the proposed effects of phonological skill on non-word reading (Harm & Seidenberg, 2001). On a connectionist account, scores on measures of phonological language skill and on measures of non-word reading both represent the influence of a single, phonological factor. If this is the case, measures of spoken language phonology should load on the same genetic factor that we have found to load independently on non-word reading. If the genetic influence on nonword reading we have identified is not attributable to phonology, an additional genetic factor will be required to obtain a good fit for these variables.

These differing predictions can be tested, and confirmed or disconfirmed. It should be noted, however, that the processes involved in all of these tasks are complex, and the verbal distinctions between phonological and orthographic processing often are used for diverse tasks, distinguishing tasks which probably share much in common, while masking dissimilarities in some tasks which often fall into the same category, be it phonological awareness or orthographic processing. That said, it should be noted here that some progress in this area has been made. Gayan and Olson (2001; 2003) have reported finding a higher genetic correlation between phonological awareness (awareness of units smaller than a

syllable demonstrated by correct performance on tasks such phoneme deletion or manipulation) and phonological decoding (ability to translate graphemes into phonemes according to regular rules, assessed by reading aloud of non-words) than to orthographic coding (speeded choice between printed homophones or word–homophone pairs). They also report a role for IQ in reading task performance, though this was not present after phonological awareness was taken into account. This suggests that, while a broad range of abilities, including general cognitive ability or *g*, working memory, and auditory processing impact on reading test performance, orthographic coding as assessed by speeded choice measures appears to contain considerable variance which is independent of phonological awareness/decoding. We propose to undertake extensions of the current data set of precisely this kind in order to explore the rolls of semantics, lexical processing, phonological decoding, and phonological awareness to begin to identify the points of contact between variance in language ability and variance in reading ability.

In summary, in this paper we have presented a genetic extension of the dual route model of reading, providing evidence that the genetics of reading is also dual route in nature. This model is then extended to spelling, and, finally, to a joint genetic model of reading and spelling. These data illustrate the power of genetically informative samples and modelling for understanding the environmental and genetic causes of behaviours. We would further argue that genetic modelling of this kind provides researchers with a valuable means of constraining and elaborating upon cognitive models of complex processes such as reading.

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Address for correspondence: Timothy C. Bates, Department of Psychology, University of Edinburgh, 7 George Square, Edinburgh, EH8 9JZ, UK,
Fax: +44-131-650-3461; E-mail: tim.bates@ed.ac.uk