ORIGINAL RESEARCH



Differential Influences of Genes and Environment Across the Distribution of Reading Ability

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Abstract

We partitioned early childhood reading into genetic and environmental sources of variance and examined the full distribution of ability levels from low through normal to high as computed by quantile regression. The full sample comprised twin pairs measured at preschool (n=977), kindergarten (n=1028), grade 1 (n=999), and grade 2 (n=1000). Quantile regression analyses of the full distribution of literacy ability showed genetic influence in all grades from preschool to grade 2. At preschool, the low end of the distribution had higher genetic influence than the high end of the distribution and the shared environment influence was the opposite. These shared environment influences of preschool became insignificant with formal schooling. This suggests that higher scores in pre-literacy skills (preschool) are more influenced by shared environment factors, though these are short-lived. This study discusses the factors that may be influencing the results.

Keywords Reading ability · Twins · Early reading · Quantile regression · Shared environment

Introduction

Improving childhood literacy is a prime educational goal. This goal needs to be achieved against a background of substantial heritability for literacy. Previous research, carried out on separate samples, has shown that low-ability reading and high-ability reading are both significantly heritable, as is normal-range variation in reading (Olson et al. 2009). However, there has been little systematic research on individual differences in the balance of genetic and environmental influences depending on literacy levels within age or grade, or on how this within-grade balance might vary across grade in early literacy development. Quantile regression, an extension of OLS regression, examines variation in

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estimates across the continuum of ability rather than aggregate estimates at just the mean of a distribution (e.g. Koenker and Ng 2005; Logan et al. 2012). This statistical technique can provide more detailed knowledge of variation in the genetic and environmental influences on literacy skills, which has the potential to improve targeted interventions, and to examine heterogeneity in response to instruction. In this study, we use quantile regression to examine the genetic and environmental influences on individual differences in literacy across the full distribution of literacy scores using the same sample over the grade levels preschool to grade 2.

To date, only one study (Logan et al. 2012) has examined the differential contributions of heritability and shared environment across the distribution of literacy. In Logan et al., 304 pairs of same sex first grade students were assessed in four different reading-related outcomes. Results indicated heritability influences were higher at the lower ends of the continuum for decoding skills, and higher shared environmental influences were present at the higher ends of the continuum. Heritability influences were higher towards the middle of the distribution for vocabulary skills, and no clear patterns were present for phonological awareness. Critically, Logan et al. compared their novel method, quantile regression, against the Cherny (1992) method, and demonstrated that the new method provided more detail on the genetic and environmental influences across the distribution. The Cherny method extended on the DeFries-Fulker (DF) regression method (DeFries and Fulker 1985, 1988) that selects cotwins with a high or low score. Hence, the quantile regression, by using the full continuum, is more powerful and provides more fine-grained detail than DF analyses before it. The present study extends the findings of Logan et al. (2012) by using quantile regression to examine similar pre-reading and reading skills in a larger, multi-national sample. Further, we assess the influences across the distribution both prior to and during the early years of school to consider the impact of formal education. By comparing trends in the magnitude of heritability and shared environmental influences on literacy in preschool and after the start of formal schooling, we can examine whether the onset and duration of formal schooling is associated with differences among these trends. For example, a decrease in shared environmental influences after formal schooling begins provides evidence of a response to adequate and consistent instruction; however, any potential response to instruction may differ based on ability level, which will be evident by differences in the magnitude of influences across the quantiles.

Previous studies have devoted more attention to dichotomized reading groups such as low- or high- ability reading (e.g. Bishop 2001; Davis et al. 2001; Light and DeFries 1995; Spinath et al. 2004). By studying the full distribution, we gain more knowledge of the continuum and aetiology of reading. In addition, analysing the full spectrum of readers and pre-readers, as opposed to only the low ability readers and pre-readers, may add to our understanding of the aetiology of dyslexia. Examining heritability and shared environmental influences at multiple points along the continuum will provide more accurate identification of if and where differences in aetiology may emerge. We hypothesized, based on previous findings (Logan et al. 2012) that heritability influences will be larger at the lower ends of the distribution of reading ability than the higher ends, and shared environmental influences will be larger at the higher ends of the distribution.

Method

Participants

The data for this research comes from the International Longitudinal Twin Study (ILTS), a longitudinal study of twins from the U.S., Australia, and Scandinavia (Byrne et al. 2005, 2006, 2008, 2009, 2010; Livingstone et al. 2016; Samuelsson et al. 2005, 2007, 2008; Willcutt et al. 2007). The maximum sample comprised 1049 same-sex twin pairs, 487 recruited from the Colorado Twin Registry in the U.S., 267 from the National Health and Medical Research Council's

Australian Twin Registry, and 280 from the Medical Birth Registries in Norway and Sweden. There were 524 MZ pairs and 525 DZ pairs. Females constituted 49.6% of the sample. Participating twins were initially contacted in the last year of preschool, with ages ranging from 54 to 71 months (mean 58.8) in the U.S., 47-68 months (mean 57.8) in Australia and 58-68 months (mean 61.2) in Scandinavia. Zygosity was determined by DNA analyses of cheek swabs or from the Nichols and Bilbro (1966) questionnaire, which includes questions on hair colour and texture, eye colour, facial appearance and complexion, and birth weight. Compared to blood tests the questionnaire has a 95% accuracy rate. A detailed breakdown of the sample by country, age, zygosity and sex is presented in Samuelsson et al. (2005). ILTS has attrition levels close to zero, with participants leaving the study only if they move out of the geographical area covered by the study (Friend et al. 2009).

Measures

Print Knowledge was scored by testing preschool children on four measures; concepts about print (CAP), letter recognition from names, letter recognition from sounds, and environmental print (Samuelsson et al. 2005). The CAP measures the child's understanding of print conventions through 24 questions such as print direction, meaning of different writing symbols and difference between print and pictures. In the letter recognition from names and sound tests, the child had to identify the 26 letters of the alphabet by pointing to the letter in a string of four letters that matched the letter named or sounded by the tester. The environmental print test required the child to identify six words presented in environmental contexts such as a 'Stop' in a stop sign and 'Exit' in an exit sign. A composite of these measures has been found to have a high correlation with later reading ability (Samuelsson et al. 2005), as shown in Table 1. The four Print Knowledge measures were standardized and combined to form a composite measure.

Reading tests were administered in kindergarten, grade 1, and grade 2 using the Test of Word Reading Efficiency (*TOWRE*; Torgesen et al. 1999). In this test children read a list of words (*Sight Word Efficiency, SWE*) and a list of non-words (*Phonemic Decoding Efficiency, PDE*) as fast as possible in 45 s. They were scored on words or nonwords correct in the time given. Forms A and B, two equivalent forms of the test, both for SWE and PDE, were all administered and averaged to optimise the reliability (Byrne et al. 2010). This was justified by the high correlation of scores between the SWE and PDE tests (kindergarten, r = 0.85; grade 1, r = 0.89; grade 2, r = 0.83; Friend et al. 2009). The test–retest reliability for 6–9 years-old children for word and non-word standard scores is 0.97 and 0.90 respectively (Friend et al. 2009).

Variable	Mean	SD	Minimum	Maximum	Skew	n
Print Knowledge ^a	9.33	3.84	2.00	19.50	0.33	977
TOWRE K ^b	18.31	19.96	0.00	115.50	1.74	1028
TOWRE 1 ^b	54.06	27.56	0.00	142.00	0.38	999
TOWRE 2 ^b	101.53	14.78	63.5	143.75	0.10	1000
Correlations	Print Knowledge ^a		TOWRE K ^b	TOWRE 1 ^t)	TOWRE 2 ^b
Print Knowledge ^a	1.00					
TOWRE K ^b	0.56		1.00			
TOWRE 1 ^b	0.53		0.74	1.00		
TOWRE 2 ^b	0.46		0.63	0.86		1.00

 Table 1
 Means, standard deviations (SD), minimums, maximums, skew, and correlations for preschool- grade 2 Print Knowledge and TOWRE scores

All correlations significant at p < 0.001

n = number of twin pairs

^aPrint Knowledge composite

^bTOWRE composite

Procedure

Written informed consent was obtained from parents of all participants. Testing was carried out on each child individually in the home or in school with each test session lasting approximately 1 h (Byrne et al. 2010; Samuelsson et al. 2007). Testing was carried out by separate testers working with each twin at the same time in the U.S. and Australia. Only one tester was able to do the assessment of both twins in Scandinavia, where the testing of both twins in a pair was also carried out on the same day (Samuelsson et al. 2005). The children were initially tested in their preschool year. Follow-up testing of the children was carried out at approximately 12 month intervals in kindergarten, grade 1 and grade 2.

Analyses

Quantile regression was conducted to examine the univariate heritability and shared environmental contributions to variance across the distributions of Print Knowledge in preschool and TOWRE in kindergarten, grade 1, and grade 2. No predictors were included in these models. Data were pooled across countries to achieve an adequate sample size to conduct quantile regression. To control for mean differences between countries, scores were standardized within each country while controlling for age and sex prior to analysis (Byrne et al. 2009; Samuelsson et al. 2005; Willcutt et al. 2007). This application of quantile regression extends DF-based methods, which estimate heritability among subgroups, by utilizing all of the available data points to estimate the heritability at multiple quantiles (Logan et al. 2012), thereby increasing power to detect existing effects in the data. Additionally, quantile regressions can compare the estimates of heritability and shared environment obtained at different points (e.g. low and high) along the distribution of the outcome. While the estimates for each quantile are not independent and therefore the significance of changes in heritability and the shared environment across the range of scores cannot be calculated we can examine the change in pseudo- R^2 between sets of quantiles. Specifically, we investigate three separate sets of quantiles (0.1 and 0.9, 0.2 and 0.8, and the 0.3 and 0.7). These sets of quantiles were chosen to compare with literature that uses standard deviations (1.5 SD, 1 SD and 0.5 SD respectively). We then classed these differences as small ($\Delta R^2 \ge 2\%$ and $\le 13\%$), medium $(\Delta R^2 \ge 14\% \text{ and } \le 26\%)$, or large $(\Delta R^2 > 26\%)$ using the criteria applied by Logan et al. (2012). To check that the total variances were constant we computed the unique environmental estimates at each quantile by re-running the regression analyses with just MZ twins predicting Twin 1 from Twin 2 and thereafter using Falconer's formula (Falconer 1960). Analyses were conducted using SAS 9.4.

Results

Table 1 displays the descriptive statistics for each measure by grade level. It also presents intercorrelations among the measures; the correlations between Print Knowledge and the TOWRE results are reasonably substantial (0.46–0.56), supporting our comparison of preschool literacy with later reading.

The estimates of heritability and shared environmental influences at the 0.1 through the 0.9 quantile are presented for all measures in Figs. 1, 2, 3, 4. The results for Print

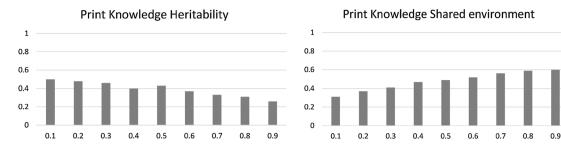


Fig. 1 Quantile regression results for the preschool Print Knowledge composite. Heritability estimates are presented on the left, and shared environmental estimates are presented on the right. Dark grey bars indicate estimates significant at p < 0.05

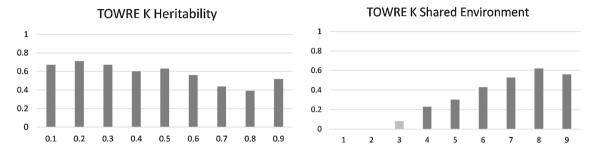


Fig.2 Quantile regression results for the kindergarten TOWRE composite. Heritability estimates are presented on the left, and shared environmental estimates are presented on the right. Dark grey bars indicate estimates significant at p < 0.05

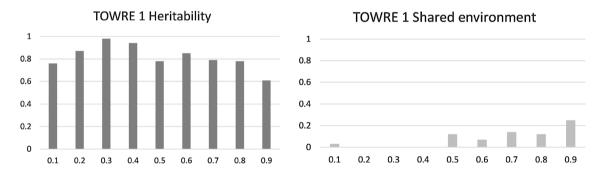


Fig. 3 Quantile regression results for the grade 1 TOWRE composite. Heritability estimates are presented on the left, and shared environmental estimates are presented on the right. Dark grey bars indicate estimates significant at p < 0.05

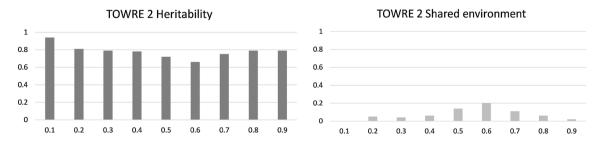


Fig. 4 Quantile regression results for the grade 2 TOWRE composite. Heritability estimates are presented on the left, and shared environmental estimates are presented on the right. Dark grey bars indicate estimates significant at p < 0.05

Knowledge (Fig. 1) indicated that heritability estimates were moderate to large and significant (0.26–0.50), and shared environmental influences were moderate to large and significant (0.31–0.60) across the distribution of scores. Heritability influences were larger when scores on reading ability were lower than when reading ability was higher, with heritability estimates decreasing at a relatively stable rate across the distribution from lower to higher scores. Results of the shared environmental estimates reflected the opposite pattern, with more variance explained by the shared environment when reading ability was higher than when ability was lower, and the estimates steadily increased across the distribution.

Results for kindergarten TOWRE (Fig. 2) indicated moderate to large and significant heritability estimates across the distribution (0.39–0.71), and small to large estimates of shared environmental influence (0.00–0.62), which were significant from the 0.4 to the 0.9 quantiles. The magnitudes of the estimates for both heritability and shared environmental influence for kindergarten TOWRE followed a similar pattern to preschool Print Knowledge, with heritability decreasing across the quantiles and shared environmental influences increasing across the quantiles.

In grade 1 (Fig. 3), the proportion of variance attributed to heritability was greater across the quantiles (0.61-0.98) than for preschool (0.26-0.50) and kindergarten (0.43-0.83), with the highest estimate at the 0.3 quantile and the lowest estimate at the 0.9 quantile. The proportion of variance attributed to shared environment influences was lower across the distribution of TOWRE in grade 1 (0.00-0.25) compared to preschool (0.31-0.60) and kindergarten (0.02-0.48), and non-significant.

Results for grade 2 TOWRE (Fig. 4) were similar to that of Grade 1 and indicated large and significant estimates of heritability (0.66–0.94) across the distribution with the largest estimate at the 0.1 quantile and the lowest estimate at the 0.6 quantile. Shared environmental estimates ranged from 0.0 to 0.14 with the lowest estimate at the 0.1 quantile and the largest at the 0.6 quantile, representing the inverse pattern to the heritability results.

Effect size comparisons are presented in Table 2 and indicated small differences in heritability between the 0.1 and 0.9, the 0.2 and 0.8, and the 0.3 and the 0.7 quantiles for all grade levels; with the exception of TOWRE 2, where there were no meaningful differences across the 0.2 and 0.8 and the 0.3 and 07 quantiles. Small differences in shared environmental estimates were present at all three sets of quantiles in preschool. In kindergarten, large differences were present between the 0.1 and 0.9 and the 0.2 and 0.8 quantiles, and medium differences were present between the 0.3 and 0.7 quantiles. Shared environmental differences were small or negligible in grade 1 and non-meaningful at all sets of quantiles in grade 2.

Table 2 Pseudo- R^2 differences between low and high quantiles for each measure

Quantiles compared	Print Knowledge composite	TOWRE K composite	TOWRE 1 composite	TOWRE 2 composite
Heritability				
0.1-0.9	0.06*	0.02*	0.02*	0.02*
0.2–0.8	0.03*	0.10*	0.01	0.00
0.3-0.7	0.02*	0.05*	0.04*	0.00
Shared envi- ronment				
0.1-0.9	0.08*	0.31***	0.05*	0.00
0.2–0.8	0.05*	0.38***	0.01	0.00
0.3–0.7	0.02*	0.20**	0.02*	0.00

*Small difference, **Medium difference, ***Large difference

Discussion

In previous studies, DF-methods have been used that examine the extreme groups of reading ability, although these analyses can compromise statistical power. The present study used quantile regression, which utilizes the full sample, maintaining adequate power, and importantly, providing thorough details on the changes in variance due to heritability and shared environmental influences at all levels across the distribution rather than just at the mean or at the extremes.

Results of effect size comparisons suggested that, generally, there were small but meaningful differences in the amount of variance explained by heritability and small to large and meaningful differences in variance explained by shared environmental influences on reading from lower to higher quantiles in preschool and kindergarten. By first grade, differences in variance explained by heritability and shared environmental influences decreased and they were negligible by grade 2. Additionally, results indicated that despite differences in magnitude, heritability influences were consistently present and significant across the continuum of literacy ability, with this influence strengthening at the start of formal education in kindergarten, and continuing to strengthen through second grade. In contrast, shared environment influences were strongest in the preschool year, yet became negligible with more time in formal school settings. We hypothesized that variance due to heritability at the lower end of the distribution would be higher than at the higher end of the distribution, and variance due to shared environment influences would be lower at the low end of the reading continuum than the high end. Our results were consistent with these hypotheses and with the results of decoding skills for first grade students from Logan et al. (2012), indicating heritability influences were higher at the lower end of the distribution than the higher end of the distribution (see Figs. 1, 2, 3, 4). Higher heritability at the lower end of the distribution may reflect the negligible influence of the shared environment, as these influences commonly counterbalance each other. In fact, the shared environmental influences were greater at the higher end of the distribution compared to the lower end of the distribution for preschool and kindergarten. Low shared environmental influences for lower ability readers may suggest that preschool and kindergarten children struggle with learning to read in spite of having adequate environmental support. In other words, differences in environmental resources and support could be suppressed by strong genetic influences on reading ability. Another possibility is that less environmental variation is present among struggling early-readers. In subsequent grades, these influences stabilised across the continuum.

Results for Print Knowledge suggested that heritability has a greater influence at the lower end of the distribution and gradually decreases across the distribution with an inverse pattern indicated for the shared environment influences. This suggested that those that scored higher on the Print Knowledge measures were more influenced by components of the shared environment such as exposure to print at home, education levels of the parents, preschool attendance itself, or aspects of the community and physical environment (e.g. learning centres). The presence of decreasing shared environmental influences across years of formal schooling, may suggest that the factors influencing pre-literacy skills are also present in kindergarten, but are short-lived. It is important to note that the measure of literacy is not consistent between preschool and formal school in the current sample, and the differences in patterns of heritability and shared environmental influences across grades may also be due to these measurement differences rather than true changes in environmental conditions. However, the similarity in the patterns of results between the different measures used in Pre-K and Kindergarten contrasted with the patterns found for first and second grades gives some support to changes in environmental influences.

The negligible influence of shared environment in grade 1 and 2 would suggest that irrespective of whether readers have a stimulating or impoverished early literacy environment, formal schooling has a stabilising influence on the environmental variance for literacy development. For example, some children in the lower end of the distribution (i.e. poor readers due to poor environment in preschool) may respond positively to a structured learning environment and intervention in school, improving closer to their genetic potential in the first and second grades. Those children who continue to fail to reach grade level outcomes in school may benefit from more intense and longer intervention programs than those that are sufficient for normal readers (Hindson et al. 2005). The nonsignificant influence of shared environment in grades 1 and 2 also suggests that the variation in teaching methods and teacher/classroom effects have relatively little influence on their reading ability scores (e.g. Byrne et al. 2009).

Researchers have tried to reconcile the presence of strong genetic influences on reading ability with known environmental influences (Olson et al. 2009). Previous research has indicated the strong relationship between pre-reading skills and environmental factors, such as shared book reading with parents and teaching of the alphabet impact on language skills important for reading (Hayiou-Thomas et al. 2006). Early intervention programs have demonstrated that improving these factors improves the pre-reading skills of those at risk (Samuelsson et al. 2005). Our findings are consistent with previous research; the influence of shared environment at an early age is seemingly overridden by the important stabilising influence of school, where teachers and classrooms have been shown to contribute rather less to student variability than previously thought and hence allow for genetic factors to assume a greater influence (Byrne et al. 2010).

These results, while informative, should be considered with several limitations in mind. Firstly, the measure used in K-2 (i.e. TOWRE) was different to the preschool measure (i.e. Print Knowledge) which may have been differentially influenced by shared environmental factors. Secondly, while the present study was under-powered to analyse each country separately, results from Samuelsson et al. (2008) suggested that the preschool and kindergarten shared environment was driven by the USA and Scandinavian samples. This could be a result of the differences in start age for formal school and/or the varied level of literacy instruction in kindergarten between countries. Had we not standardized within country, the shared environmental estimates would have been even higher due to country-level differences. By standardizing within country we want our shared environmental estimates to capture between-family variance instead of between country variance to the degree possible. Subsequent investigations can include environmental moderators to investigate whether variability in aspects such as school start age influences estimates across the quantiles. Lastly, the ILTS sample may not be representative because of a restricted environmental range, in particular the absence from the sample of the most impoverished communities with extremely negative educational environments (Olson et al. 2009). It would be instructive to see if the trends observed in the present study are maintained in a more representative sample, and, in fact, whether the influences detected in this study might be more pronounced in a group that was oversampled for social disadvantage.

This study makes a unique contribution to research by providing analyses of the full range of reading abilities using quantile regression. In addition, the data were analysed over several key early literacy years, from preschool through to grade 2. In summary, these results provide evidence that genetics influence reading ability across the full distribution from preschool through to grade 2, but the influence of shared environment factors at preschool are stabilised and re-set by the influence of the more consistent school environment, indicating the importance of formal literacy education that improves reading ability at the same time that it reduces environmental variance, leaving genes to account for more of the phenotypic variance in reading.

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Compliance with ethical standards

Conflict of interest Dipti McGowan, Callie W. Little, William L. Coventry, Robin Corley, Richard K. Olson, Stefan Samuelsson, and Brian Byrne declares no conflicts of interest.

Ethical Approval All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards.

Informed Consent Informed consent was obtained from all individual participants included in this study.

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