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# Heritability Across the Distribution: An Application of Quantile Regression

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**Abstract** We introduce a new method for analyzing twin data called quantile regression. Through the application presented here, quantile regression is able to assess the genetic and environmental etiology of any skill or ability, at multiple points in the distribution of that skill or ability. This method is compared to the Cherny et al. (Behav Genet 22:153–162, 1992) method in an application to four different reading-related outcomes in 304 pairs of first-grade same sex twins enrolled in the Western Reserve Reading

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W. Battelle Center for Mathematical Medicine, The Research Institute at Nationwide Children's Hospital, The Ohio State University, Columbus, OH, USA Project. Findings across the two methods were similar; both indicated some variation across the distribution of the genetic and shared environmental influences on non-word reading. However, quantile regression provides more details about the location and size of the measured effect. Applications of the technique are discussed.

**Keywords** Differential heritability · Differential shared environment · Quantile regression · Twins

#### Introduction

Estimates of genetic influences on continuously distributed abilities or skills are assumed to be consistent across the distribution of the measured skill or ability. However symptoms of a clinical disorder, although continuously distributed, may be more or less heritable at the extremes of the distribution (Deater-Deckard et al. 1997). In quantitative genetics, the most commonly used methodological options to examine whether heritability is constant across a continuous skill are regression-based techniques: the Defries-Fulker extremes analysis (DF analysis; DeFries and Fulker 1985), or the Cherny model (Cherny et al. 1992). Some other methods exist that are able to conduct similar analyses, such as the liability-threshold model (Gottesman and Shields 1973), or models that examine heritability across a second measured child- or family-level variable (e.g., Clifford et al. 1984; LaBuda and DeFries 1990; Purcell 2002). However, in the present study we focus on the regression-based techniques as they are the foundation of quantile regression, the technique that will be introduced later in this paper.

The Defries–Fulker extremes analysis (DF analysis) was developed for examining the heritability of a selected



Fig. 1 Heritability of a hypothetical trait (HT) across the distribution of ability, where a true group exists at the 20th percentile

group (i.e., proband group) at an extreme end of a continuous distribution (DeFries and Fulker 1985). The DF analysis method capitalizes on regression to the mean to measure the extent to which the differences between the proband group and the rest of the population are due to genetic effects (Plomin et al. 2008). Theoretically, if a proband scores extremely poorly on a continuously distributed skill, their cotwin should score relatively closer to the mean of the distribution. If the trait is heritable, then the dizygotic (DZ) cotwin score should show more regression to the mean than the monozygotic (MZ) cotwin twin score. The method was later extended to more explicitly test whether the genetic influences of the proband group differ from those of the unselected group (DeFries and Fulker 1988). For a more in-depth introduction to the conceptual underpinnings of DF analysis, see Plomin and Kovas (2005).

Because a proband group must be selected from an extreme end of the distribution, the DF analysis methods is very useful when a skill or ability has clear accepted criteria of what constitutes a disability (e.g., Attention Deficit/ Hyperactivity Disorder). However, DF analysis has also been frequently used when the skill in question is less welldefined, such as reading or math disability (see Plomin and Kovas 2005). Typically, the low-performing or proband group is identified by a cut point selected by the researcher or by convention, and all participants who score at or below the cutpoint are considered to be in the proband group. Even when guided by findings from the literature, the cutoff point may still be inaccurate for assessing the point or points at which underlying differences in etiology may emerge.

To illustrate this idea, suppose that a hypothetical trait (HT) shows differential heritability at or below the 20th percentile of a norm-referenced test. A graphical representation of what the true heritability of HT would look like is presented in Fig. 1. In Fig. 1, the x-axis represents percentile, or level of ability on HT, and the y-axis

represents the percentage of variance attributable to heritability at that point in the distribution of HT. It is clear from this graph that for the 20th percentile and below the heritability is about 80%, while the remainder of the distribution shows heritability of about 40%. Typical heritability estimates represent an average of the entire distribution, and so would report that HT was about 50% heritable. Because a true group exists, DF analysis would be an excellent choice for examining HT, but only if the correct cutoff is selected for the identification of the proband group. If the cutoff for HT is selected at the 30th percentile in a DF analysis, it would be unlikely to detect a significant group difference in heritability; even though a group is present.

Another group of regression-based methods does not have the same problems with selection. The extended DF analysis, developed by LaBuda and Defries (1990), uses the same basic regression equations but allows for the examination of heritability of one skill across the distribution of a second skill, ability, or other measured environmental variable. Cherny et al. (1992) took this method a step further. The regression model indentified by the authors included interaction effects, essentially producing a significance test of the interaction between the heritability of a skill or ability, and the score on that ability. The Cherny method can be used to measure linear (and quadratic) changes in heritability and shared environmental estimates across the distribution of a single measured skill or ability.

The one disadvantage of these methods is the imprecision of the finding's effect. The level of detail afforded regarding changes in  $h^2$  and  $c^2$  is imprecise. The researcher is only aware whether changes exist, and in what direction (i.e., whether heritability increases or decreases with increased skill). To illustrate, refer back to the same HT with a true differential group with higher heritability existing at the 20th percentile. The Cherny model would fit a line to the heritability function across that distribution. Thus, this method would report a negative parameter associated with the HT, but the researcher would have no indication that the 20th percentile group existed, only that heritability decreased as HT levels increased.

The technique introduced in this paper responds to the weaknesses of the Cherny method by providing not just a general trend of how heritability changes across a continuous distribution of scores, but also information about the location of that shift. In the present study, we present an application of quantile regression to familial data. Quantile regression was originally developed by Koenker and Bassett (1978). In the terminology of the technique, "quantile" refers to percentile, and references the fact that quantile regression is used to estimate relations between a dependent and independent variable and multiple given percentiles within a continuous distribution.

 Table 1 Means, standard deviations, minimums, and maximums for IQ, reading, and related abilities for all children with complete data

Variable	М	SD	Minimum	Maximum	п
Word Identification	104.22	18.04	76	174	589
Word Attack	103.24	12.21	81	141	586
Phonological Awareness <sup>a</sup>	13.24	6.5	0	30	573
Vocabulary	49.02	5.8	33	69	589

Letter Identification Woodcock Reading Mastery Test (WRMT) Letter Identification, Word Identification WRMT Word Identification, Word Attack WRMT Word Attack

<sup>a</sup> Figures represent composite raw scores as no norming information was available

Quantile regression was originally developed to examine how the relation of a dependent variable with an independent variable changes depending on the score of the dependent variable (Koenker and Bassett 1978). To illustrate a typical application of the quantile regression technique, a recent paper by Reeves and Lowe (2009) examined how demographic variables related to student's math achievement dependent on math achievement score. The authors demonstrated that gender and race were more strongly related to reading achievement at the lower end of the distribution of math achievement. In another example, Catts et al. (2009) used quantile regression to examine floor effects in several reading measures. The authors examined the predictive utility of different quantiles of pre-reading skills to later reading performance. These studies demonstrate that quantile regression typically focuses on the relatedness of predictors to an outcome, and examine how that relation changes depending on the score of the outcome.

In the present study, we build on the utility of quantile regression and extend it to estimates of heritability and shared environment. In the present application, quantile regression is able to simultaneously estimate heritability and shared environment across multiple points in the distribution of a measured variable. To demonstrate, quantile regression will be compared to the Cherny method (Cherny et al. 1992), and the techniques will be contrasted in an application to reading related skills.

## Method

# Participants

Participants were drawn from the Western Reserve Reading Project (WRRP), part of an ongoing longitudinal twin study in Ohio (Petrill et al. 2006). Children involved in the project are being assessed at seven waves of home visits each approximately 1 year apart. For the purposes of the demonstration of this new method, the present study utilized data collected during home visit 1, when children were approximately 6 years old (M = 6.07, SD = 0.67). Average standard scores for vocabulary, reading, and the related abilities measured in the present study are presented, along with the number of children with complete data for each variable, in Table 1. On average, the children enrolled in this study were slightly above average on the standardized measures.

Families were recruited through school nominations, Ohio State birth records, and media advertisements. A total of 304 pairs of monozygotic (MZ; n = 131) and same-sex dizygotic (DZ: n = 173) twins with known zygosity were used in the present study. Zygosity was determined with DNA analysis using a cheek swab. For cases where parents did not consent to genotyping (n = 76), zygosity was determined using a parent questionnaire on twins' physical similarity (Goldsmith 1991). Parent education levels varied widely and were similar for mothers and fathers, with a slight positive skew (0.04).

#### Measures

The goal of this study was to examine the heritability of reading and related skills across the distribution of ability scores. Thus several commonly-used reading-related tasks were selected for examination. Descriptive statistics were reported in standardized scores when available to allow for comparison to a normative sample (Table 1). In the analyses conducted in the present study, the primary concern was the relation between twins rather than the mean differences among twins. As such, prior to entry in analyses, all variables were regressed for age, age squared, and sex and were standardized to a mean of 0 and a standard deviation of 1 using a regression procedure.

Two subtests of the *Woodcock Reading Mastery Test* (*WRMT*; Woodcock 1987) were administered: The first is the WRMT *Word Identification*, an accuracy-based measure requiring children to read words aloud, and *WRMT Word Attack*, which requires children to pronounce presented non-words, and is scored based on accuracy.

*Phonological Awareness Test (PAT;* Robertson and Salter 1997). Phonological Awareness was assessed using the Phonological Awareness Test. It included three subtests that measured phonemic segmentation (whole word), and phonemic deletion (syllabic deletion, and phoneme deletion). The three subtests were summed to form a raw total.

*Stanford Binet: Vocabulary* (Thomdike et al. 1986). The Vocabulary subtest of the Stanford Binet requires students to describe a given picture, and as the items progress to give a definition of a presented word. Scores are based on accuracy.

#### Statistical technique: overview

The goal of the present study is to introduce and explain how quantile regression addresses the question of consistency of heritability across the distribution. To facilitate this explanation, the selected reading-related skills were first examined using the Cherny method and then with quantile regression. The techniques, while similar, approach the question in different ways. In the Cherny method, a single coefficient is produced indicating whether heritability significantly changes across the distribution. The Cherny method, therefore, estimates the shape of the cross-distribution change. By contrast, the application of quantile regression presented here uses all data to calculate the heritability at a specific point, for example at the 25th percentile/ quantile. Multiple analyses can be conducted simultaneously, such that estimates of heritability and shared environmental effects can be reported for multiple points in the distribution. Each technique is described in detail in the following sections.

#### Statistical technique: Cherny

Cherny et al. (1992) developed an extension of the DF model, which provides the ability to examine whether heritability changes linearly across the distribution of the outcome. Prior to explaining the equation presented in Cherny et al. (1992), we first introduce the equation it is based on, the Extended DF model (LaBuda et al. 1986). The extended DF model is estimated in a regression framework, and can be represented by:

$$T_2 = B_0 + B_1(T_1) + B_2(R) + B_3(T_1 * R) + e$$
(1)

where  $T_2$  is the one twin's score on the outcome,  $T_1$  is the Co-twin's score on the same outcome, R is the degree of genetic relatedness with MZ = 1 (sharing 100% of their genes) and DZ = 0.5 (sharing 50% of their genes),  $B_0$  is the intercept or mean of the Co-twin score,  $B_3$  is the estimate of heritability ( $h^2$ ), and  $B_1$  represents a direct estimate of the proportion of variance attributable to the shared environment, ( $c^2$ ; Rodgers et al. 2001).

The extension to this model proposed by Cherny et al. (1992) adds additional interactions to examine how heritability and environmental estimates change across the distribution. The regression equation can be represented by:

$$T_2 = B_0 + B_1(T_1) + B_2(R) + B_3(T_1 * R) + B_4(T_1 * T_1) + B_5(T_1 * T_1 * R) + e$$
(2)

where  $T_2$  is one twin's score on the outcome,  $T_1$  is the Co-twin's score on the same outcome, R is the degree of genetic relatedness,  $B_0$  is the intercept or mean of the

Co-twin score,  $B_3$  is the influence due to genetics, and  $B_1$  represents the effects independent of genetics. The linear trends of shared environment and heritability are estimated via the  $B_4$  and  $B_5$  parameters respectively. To illustrate, a significant  $B_4$  parameter estimate would indicate that significant linear change in the shared environmental estimate occurs across the distribution of the scores of  $T_1$ .

The model was also developed to be extended to look for quadratic changes across the distribution of skills. These can be estimated by adding two additional terms to the previous equation (from Cherny et al. 1992):

$$T_{2} = B_{0} + B_{1}(T_{1}) + B_{2}(R) + B_{3}(T_{1} * R) + B_{4}(T_{1} * T_{1}) + B_{5}(T_{1} * T_{1} * R) + B_{6}(T_{1} * T_{1} * T_{1}) + B_{7}(T_{1} * T_{1} * T_{1} * R) + e$$
(3)

where all interpretations are identical to Eq. 2, but now  $B_6$  provides an estimate of the quadratic trend in the shared environmental attribution of variance across the distribution, while  $B_7$  does the same for the estimate of heritability. In the present study, Eqs. 2 and 3 were fit to the data for each of the four reading outcomes examined.

Statistical technique: quantile regression

Quantile regression was developed by Koenker and Bassett (1978), and expands on the ordinary least squared (OLS) regression techniques. In OLS regression, equations are designed to estimate the mean of Y conditional on X. In quantile regression, equations are designed to estimate the relation of X with Y, conditional on quantiles (or percentiles) of Y. In other words, this technique examines how the relation of X with Y changes depending on the score of Y. As an example, OLS regression would examine how the mean of children's reading achievement changed conditional on parental socio-economic status, while quantile regression would examine the relation of socio-economic status with children's reading achievement at several points on the distribution of children's reading performance. In allowing this relation to change depending on the score of Y, quantile regression produces unique parameter estimates for each quantile it is asked to examine.

In OLS regression, the relation of X with Y is estimated by minimizing sums of squared residuals, the result being a single line through a scatterplot of points. Similarly, quantile regression can be used to estimate the relation of X with Y at a given quantile (percentile) within the distribution of Y by minimizing a sum of absolute residuals conditional upon the given quantile. The minimization function can be represented by:

$$\sum \theta |y_t - x_t \beta| + \sum (1 - \theta) |y_t - x_t \beta|$$
(4)

where  $y_t$  is the dependent variable,  $x_t$  is the independent variable or vector of independent variables,  $\beta$  is the coefficient (or vector of coefficients), and  $\theta$  is the quantile to be estimated. For median regression, a special case of quantile regression,  $\theta = 0.5$  (Koenker and Bassett 1978). At the median, the conditional result is reported when half the cases are less than or equal to y at 0.5, while the remaining cases are greater than 0.5. Quantile regression can also be extended to points beyond the median through asymmetric weighting of the values across the distribution. Referencing the formula above, positive residuals are given a weight of  $\theta$ , while negative residuals are given a weight of  $1 - \theta$ . For example, a quantile regression at the 75th percentile occurs when 75% of cases are less than or equal to y at 0.75. The positive residuals would be given a weight of 0.75, and the negative residuals would be given a weight of 0.25. There are several excellent resources available describing quantile regression and explaining the underlying equations (see Firpo 2007; Koenker and Hallock 2001; Koenker and Machado 1999).

It is important to note that, although it seems similar, quantile regression is not equivalent to dividing a sample into multiple subgroups based on percentiles of the dependent variable and subsequently fitting an OLS regression to each subgroup. OLS regression has many assumptions about the distributions of the variables being examined, and dividing the sample into multiple sub-samples would violate these assumptions. Quantile regression, on the other hand, makes no assumption about the distribution of error terms, and is thus better for estimating non-normal distributions. In addition, quantile regression includes all of the data when fitting each quantile, using bootstrapping, data re-sampling, and statistical inference to estimate the relations between variables at several points in the distribution.

#### New application

In the present study, quantile regression was adapted to examine heritability and shared environmental contributions to variance in a given outcome across a distribution of scores. This was accomplished by combining quantile regression with the regression-based methods of estimating heritability discussed previously (LaBuda and DeFries 1990; Cherny et al. 1992). In the regression-based methods, the scores of one twin are regressed onto the scores of the Co-twin, a variable coded for degree of genetic relatedness (MZ = 1, DZ = 0.5), and the interaction between the two variables. Similarly, in the present study, the score of one randomly selected twin was used to predict the score of the Co-twin, the degree of relatedness of the twins, and the interaction between the two, but in a quantile regression. Because quantile regression calculates estimates for the outcome conditional upon a given quantile, heritability and shared environment were estimated at several points along the distribution without the use of additional interaction terms.

The resulting representative quantile regression equation is identical to the model used in the extended DF analysis (Eq. 1). The solutions are estimated for each parameter separately for each quantile by inserting the Extended DF equation into the quantile regression minimization function (4). The  $\beta$  in that minimization function is always representative of a vector of coefficients. In this application, the vector of coefficients is represented by Eq. 1, such that the minimization of residuals at each quantile follows the representation:

$$\frac{\sum \theta |T_2 - (\beta_0 + \beta_1(T_1) + B_3(T_1 * R))|}{+ \sum (1 - \theta) |T_2 - (\beta_0 + \beta_1(T_1) + B_3(T_1 * R))|}$$
(5)

where  $\theta$  is the quantile to be estimated,  $T_2$  represents the outcome score for one twin (replacing  $y_t$  from representation 4 as the dependent variable),  $T_1$  represents the Co-twin,  $B_1$  represents the estimate of the shared environment, (c<sup>2</sup>),  $B_3$  is the estimate of heritability ( $h^2$ ), and R is the degree of genetic relatedness.

In the present study, this analysis was conducted at 17 different points in the distribution, resulting in 17 different estimates of  $h^2$  and  $c^2$  for each outcome, conditional on the quantile being examined. It should be noted that the only necessary data-preparation for quantile regression analyses are that any outcome should be z-scored prior to entry into the analyses for ease of interpretation (the code to do so is provided in Appendix 1).

It is important to note that the result of the quantile regression in the present application is several point estimates of the percentages of variance in the outcome attributable to heritability and shared environment. This is a natural extension of the multiple regression method (e.g., Cherny method) mentioned previously. However while the regression methods fit a linear or quadratic function to the shape of the change in heritability, the quantile regression does not. Rather, the estimates at each point in the distribution are dependent on all other points in the distribution. Thus, while the Cherny method estimates the shape of change across the distribution, and whether that slope is significantly different from zero, in quantile regression such a test is not appropriate because (1) no functional line is fit to the quantile regression estimates, and (2) all data is used to calculate each point estimate, and as such the estimates are not independent of each other. Though confidence intervals can be calculated to correspond to the point estimates, they test if the calculated estimate is significantly different from zero, and are not compared to one another.

However, it is still of interest to compare the estimates of heritability and shared environment obtained at different

 Table 2 Cherny results: linear change in heritability across the distribution

Word Attack 0.			inical C	lineai
	.47**	0.36** -	-0.02 (	).05
Word ID 0.	.26*	0.62**	0.03 -0	).03
PA 0.	.57**	0.26** -	-0.07 0	).08
Vocabulary 0.	.27	0.22 -	-0.13 0	).13

\* p < 0.05, \*\* p < 0.01

 Table 3 Cherny results: quadratic change in heritability across the distribution

	h <sup>2</sup>	c <sup>2</sup>	h <sup>2</sup> linear	c <sup>2</sup> linear	h <sup>2</sup> quad	c <sup>2</sup> quad
Word Attack	0.63**	0.25*	0.29	-0.17	-0.12**	0.08*
Word ID	0.15	0.72**	-0.10	0.09	0.06	-0.06
PA	0.31	0.47**	-0.03	0.04	0.09	-0.07
Vocabulary	0.18	0.27	-0.11	0.11	0.03	-0.02

\* p < 0.05, \*\* p < 0.01

points along the distribution of the outcome. To do so, we draw from the work of Petscher and Kim (2011), who used quantile regression to examine whether the percentage of variance explained in the outcome was meaningfully different between two different predictors at a single point in the distribution. A measure of effect size was calculated to compare the amount of variance in the outcome explained by one predictor with the amount explained by another. In the present study, we adapt this method to compare whether a meaningful difference exists in the amount of variance in the outcome whether a meaningful difference exists in the amount of variance in the outcome explained at three points of the 17 estimated quantiles.

To do so, measures of effect size (pseudo  $R^2$ ) were calculated for the point estimates at the 15th percentile to those at the 50th percentile, the 50th percentile to the 85th percentile, and the 15th to the 85th percentile. The squared difference between the two estimates was considered to be meaningful if the  $\Delta R^2 \ge 2\%$  (Cohen 1988). Changes in  $R^2$  between 2 and 13% of the variance were considered small, 14–26% medium, and greater than 26% large. To illustrate the effect size technique, we refer to the HT described previously and presented in Fig. 1. If these represented the results obtained in a quantile regression, the estimate obtained at the 15th percentile. This is considered to be a medium-sized meaningful change.

#### Data setup and statistical program

All models were fit with SAS 9.2. The syntax to import and prepare the data set is given with annotation in Appendix 1.

The code for analysis using the Cherny method is presented with annotations in Appendix 2. The quantile regression syntax is presented in Appendix 3. All quantile regression models were fit with the SAS simplex optimization algorithm. In the given syntax, the quantiles are selected for examination on the command "quantile = ". In the present study, 17 quantiles were examined, ranging from 0.10 to 0.90, in increments of 0.05. This number of quantiles was chosen to best estimate the dependence of heritability and shared environment on the outcome score.

## Results

#### Cherny method

As was previously indicated, the Cherny method uses regression based techniques to estimate linear change in heritability across a distribution of scores. The results of the Cherny model estimating the linear change effects are reported in Table 2. Each of the outcomes indicated significant overall influences of heritability and shared environment. For example, estimates for Phonological Awareness showed heritability and shared environmental proportions of variance were significantly different from zero (0.57 and 0.26 respectively). The unique part of this analysis is the test of the linear change in heritability and shared environment (represented in columns 3 and 4 of Table 2). However, none of the estimates of these effects were significantly different from zero, indicating that none of these outcomes show linear change in the estimates of heritability and shared environment across the distribution.

The Cherny method was also used to estimate whether quadratic changes in the estimates of heritability and shared environment exist. The results of this analysis are presented in Table 3. In this table, just as with Table 2, the first two columns again represent heritability (h<sup>2</sup>) and shared environment  $(c^2)$  for the entire distribution. Also like Table 2, the second two columns in Table 3 represent the linear change across the distribution for  $h^2$  and  $c^2$ . All of these linear coefficients remain non-significant after the inclusion of the quadratic effect. The columns labeled "h<sup>2</sup> quad" and " $c^2$  quad" represent the quadratic effect across the distribution of scores. Note that Word Attack shows small but significant effects for both quadratic estimates (-0.12 for heritability and 0.08 for shared environment). This indicates that a quadratic function is present in the estimates of heritability and shared environment across the distribution of Word Attack scores, such that contributions of variance to heritability decreases non-linearly, and shared environmental attribution increases non-linearly, as word attack scores increase.



Fig. 2 Quantile regression results for Word Attack, Word Identification, Phonological Awareness, and Vocabulary. Heritability estimates for each outcome are presented in the top panel, and shared environmental estimates are presented in the bottom panel

#### Quantile regression

Quantile regression results contain a great deal of information. There are 17 different estimates of heritability and shared environment for each outcome. To summarize the results, the estimates from each procedure have been graphed, and are presented in Fig. 2. In Fig. 2, the top graph for each outcome represents the heritability estimate, while the bottom graph represents the shared environmental estimate. As indicated previously, a statistical test comparing the significance of the difference between the results at multiple points in the distribution is inappropriate

**Table 4**  $R^2$  differences between given quantiles for each measure

Quantiles compared	Word Attack	Word ID	PA	Vocab
Heritability				
15-50	0.21 <sup>a</sup>	0.06 <sup>x</sup>	0.00	0.23 <sup>a</sup>
50-85	0.00	0.01	0.01	0.12 <sup>c</sup>
15-85	0.27 <sup>b</sup>	0.13 <sup>c</sup>	$0.02^{c}$	0.02 <sup>c</sup>
Shared environment				
15-50	0.41 <sup>b</sup>	0.22 <sup>a</sup>	0.00	0.04 <sup>c</sup>
50-85	0.01	0.02 <sup>c</sup>	0.00	0.05 <sup>c</sup>
15-85	0.58 <sup>b</sup>	0.37 <sup>b</sup>	0.00	0.00

Word ID WRMT Word Identification, Word Attack WRMT Word Attack, PA Phonological Awareness, Vocab Stanford Binet Vocabulary

<sup>a</sup> Medium difference, <sup>b</sup> large difference, <sup>c</sup> small difference

in quantile regression. As an alternative, effect sizes were calculated comparing the percentage of variance explained at the 15th quantile to the 50th quantile, the 50th to the 85th quantile, and the 15th to the 85th quantile. These quantiles were chosen as representative of the entire distribution of scores. Results of these comparisons are presented in Table 4.

Visual inspection of the graphed results in Fig. 2 reveals some distinct patterns. For example, the results for Word Attack suggest that when lower scores were weighted more heavily, heritability explained more variance in the outcome compared to when higher scores were weighted more heavily. The opposite pattern was observed for the shared environmental influences on Word Attack; these were comparatively stronger when higher scores were weighted more heavily. These can be followed up with effect size comparisons from Table 4. For heritability, medium effects were observed between the 15th and 50th quantile ( $\Delta R^2 = 0.21$ ), and large effects between the 15th and 85th quantile ( $\Delta R^2 = 0.27$ ). Large differences were also observed for the shared environmental estimates between the 15th and 50th quantiles  $(\Delta R^2 = 0.41)$  and between the 15th and 85th quantiles  $(\Delta R^2 = 0.58)$ . Examining Fig. 2, the pattern of results for Word Identification was similar to those of Word Attack: when lower scores were weighted heavily, heritability was stronger ( $\Delta R^2 = 0.13$ ; Table 4), and shared environment was weaker ( $\Delta R^2 = 0.37$ ).

Examining the graph for Phonological Awareness, no clear pattern is visible. The estimates of proportions of variance attributable to heritability and shared environment were relatively stable across the distribution. The comparisons presented in Table 4 suggest the same, with no meaningful differences observed. Finally, the graphed results of the Vocabulary assessment suggest that heritability is weakest at both tails of the distribution, but stronger when the scores in the middle of the distribution are weighted most heavily ( $\Delta R^2_{(15-50)} = 0.23$ ;  $\Delta R^2_{(50-85)} = 0.12$ ; Table 4). Shared environment showed the strongest influence on vocabulary when the highest or lowest scores were weighted most heavily, however this change was very small ( $\Delta R^2_{(50-85)} = 0.05$ ).

## Discussion

The present study introduced and demonstrated quantile regression and how it can be used to address whether differences in heritability exist across multiple points in the distribution of one continuous skill. Quantile regression was compared to the Cherny method of examining differential heritability across a distribution. To contrast the results, the Cherny results are discussed first, followed by the comparative quantile regression results for each outcome.

Beginning with the results of the Cherny method, the estimates of linear change in heritability or shared environment as a function of cotwin score were nonsignificant (Table 2). The results were consistent across all four examined outcomes. This indicates that there was no linear relation between estimates of heritability or shared environment and scores on the outcome. Thus, the findings of the linear portion of the Cherny method indicate no significant variation across the distribution of scores.

Switching to the results of the Cherny method of examining quadratic changes dependent on the score on the outcome, these results also indicated non-significant linear changes three of the four examined outcomes. However, significant quadratic changes in heritability and shared environment were observed for Word Attack performance. The significant negative quadratic effect for heritability estimates significantly negative (estimate = -0.12), indicating a gradual, nonlinear decrease in the proportion of variance in Word Attack attributable to heritability as scores increase. The reverse pattern was observed for the shared environmental estimates (estimate = 0.08).

The results of the quantile regression analysis showed some meaningful differences among the estimates of heritability and shared environment for Word Identification and Word Attack depending on the scores weighted most heavily. These followed the trend of higher contributions of variance to heritability at the lower end of the distribution. In addition, some meaningful, but small differences were observed for the results of the Vocabulary assessment, with lower heritability found at the lower and higher ends of the distribution. Finally, no meaningful differences were observed for Phonological Awareness.

## Comparison of the results

Comparing the results from the two analytic methods, the results were well aligned for the Phonological Awareness assessment. Neither technique suggested that any dependencies existed in heritability or shared environment based on the Phonological Awareness score. The results were also well aligned for Word Attack. The Cherny method suggested that some non-linear change in shared environmental attributions occurred, such that shared environmental influences increased as scores increased. The opposite effect was found for heritability; higher scores were related to lower estimates of heritability. Examining the results of the Quantile Regression, we were able to gain more information about the location of the shift and the size of the heritability and shared environmental effects. The lowest end of the distribution showed an estimated 0% of the variance due to shared environment, the middle of the distribution showed about 50% due to shared environment, and this increased to about 80% due to the shared environment at the highest end of the distribution. Heritability was high (80%) at the lowest end of the distribution, but decreased to about 20% heritable for the 60th percentile and above. Thus it is clear that with the Cherny method we are able to obtain information about the direction and the shape of the relation, but the quantile regression method provides more detailed information about the size and location of these changes.

The results for vocabulary did not align as well. The Cherny method showed no significant linear or quadratic effects, while some small to medium differences were observed dependent on the outcome score in the quantile results. The results were also not consistent for Word Identification. The Cherny method suggested that shared environmental estimates had no linear or quadratic dependencies on the score of the outcome. Yet the quantile regression method showed meaningful differences (ranging from medium to large effects) among the obtained estimates dependent on the percentile of the outcome. It is also important to note that though the quantile regression showed similar results for Word Identification and Word Attack in terms of effect size, the Cherny method showed significant quadratic trends for Word Attack but did not show significant linear or quadratic trends for Word Identification.

The conflicting results lead to the natural question of which technique is correct. It is possible that the meaningful differences observed in quantile regression are not truly present in the data. Alternatively, the Cherny method may be underpowered to detect an effect that truly is present in the data. An additional possibility is related to robustness of the two techniques to non-normality. The quantile regression technique was developed in part to identify heteroschedasticity present in relations among variables, while the sensitivity of the Cherny method to this phenomenon is unknown. Future studies should investigate the differences between the two methods further by simulating data with varied sample sizes, effect sizes of change across the distribution, directions of changes in heritability and shared environment, and distribution characteristics. Such a study would thoroughly illustrate the strengths and weaknesses of both techniques.

## Limitations

Other sources of variance that could impact estimates of heritability must be considered. First, heritability is, by nature, always a function of the population being examined. For example, reading was relatively normally distributed in the present study, with the distribution including few extremely poor readers. As a result, the findings of the present study could be different in a sample that was enriched with poor readers. Another concern to the findings of the present study, some studies have shown that estimates of heritability, especially how heritability changes across a continuous distribution of scores, are susceptible to issues of measurement error (Wicherts and Johnson 2009), and measurement invariance (Eaves 2006; Lubke et al. 2004). Thus, any meaningful differences in heritability or shared environment identified by the quantile regression technique could also be due to one or both of these issues. Questions of the role that measurement invariance plays in quantile regression would be an interesting future direction for research. In addition, differential heritability estimates could vary as a function of environmental or cognitive moderator variables. For example, the observed change in the environmental etiology of non-word ability across the distribution of scores could be related to phonological awareness skill or parental support of reading at home. The current application of quantile regression does not at present allow for investigation of potential moderators, but we are developing this aspect of the analysis as a direction for future studies.

#### Applications

Two possible uses of the quantile regression technique are discussed. First, the assumption of consistent heritability is central to the statistical assumptions underlying of most models used in behavior genetics. Because quantile regression is able to test this assumption in a way that is simple to run and easy to interpret, quantile regression would make an excellent initial descriptive step in preliminary analyses prior to more comprehensive quantitative genetic analyses. For example, the quantile regression results of Phonological Awareness in the present study showed no evidence of meaningful variation in heritability of shared environment across the distribution of skill. Thus, Phonological Awareness could be used in more complex modeling without concern of violating any statistical assumptions.

Second, quantile regression could be used as a second step in the extended DF or Cherny methods for examining heritability and shared environment across a distribution. If the results of an Extend DF model or Cherny model were to report a significant parameter representing linear or quadratic dependency of  $h^2$  or  $c^2$  on the score of the outcome, the data could then be put into a quantile regression for additional descriptive information about the effect. Just as was found for the results of Word Attack in the present study, the plots produced by quantile regression could provide the specific locations and magnitudes of any effects.

# Conclusion

The quantile regression method introduced in the present study provides an efficient and simple method of testing the assumptions of invariance in heritability and shared environmental influences across the distribution of one skill. We believe that this application of quantile regression to familial data will be useful for preliminary descriptive analyses, identifying or exploring group differences in heritability and shared environmental influences on any measurable skill or ability.

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## Appendix 1

SAS code: Data setup

\*\*\*\*\*\*\*\*\*\*\* This code requires a data set that has one variable (column) per twin. In this first data preparation step, the data is called in, and the scores on the desired variable for twin 1 and their cotwin (twin 2) are renamed as "Var1" and "Var2". libname df 'C:\yourpath\'; Data abc; set df.your\_dataset; Var1 = your\_twin1\_variable; Var2 = your\_twin2\_variable; **run**; \*\*\*\*\* For this next step, change the words "your\_zygosity" to the name of the zygosity variable in your dataset. Change "MZ" to read however MZ is coded in your dataset (i.e., 1). Do the same for DZ. The "proc standard" z-scores the variables (var1 & var2) prior to entry in the analysis. data abc2; set abc; if (your\_zygosity = MZ) then rel = 1; if (your\_zygosity =  $\mathbf{DZ}$ ) then rel = 0.5; run: **proc standard** data = abc2 m=0 std=1 out = z1;var var1 var2; run; 

#### Appendix 2

#### SAS code: Cherny method

The Cherny method can be conducted using the GLM procedure. This is done by using twin 1's score (var1) to predict twin 2's score (var2), along with an interaction of twin 1's score with degree of relatedness (established in Appendix 1).

#### **proc glm** data = abc2;

model var2 = var1 rel var1\*var1 var1\*rel var1\*rel;

#### run; quit;

\*\*\*\*\*

From these results, the parameter estimate associated with:

"var1" represents the proportion variance attributable to shared environment

"var1\*rel" represents the proportion of variance attributable to heritability

"var1\*var1" represents the linear change of shared environment across the distribution

"var1\*var1\*rel" represents the linear change of heritability across the distribution

SAS code: Quantile regression

\*\*\*\*\*\*\*\*\*\*\* After setting up the data, run the following code to obtain estimates of heritability and shared environmental effects. This is done using the variable "Rel," created in step 2 of Appendix 1 (indicating the degree of relatedness). The results are presented graphically via the ODS statements. Replace "quantile = all" with the quantiles desired. For the analyses in the present study, this syntax read: quantile = 0.10 0.15 0.20 0.25 0.30 0.35 0.40 0.45 0.50 0.55 0.60 0.65 0.70 0.75 0.80 0.85 0.90 ods html: ods graphics on; proc quantreg ci=sparsity; model var1 = var2 | rel/quantile= all plot = quantplot; run: ods graphics off; ods html close; \*\*\*\*\* The shaded areas on the graph represent 95% confidence intervals of differences from zero. The graph labeled "var2" represents shared environment.

The graph labeled "var2\*zyg" represents heritability.

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