BEHAVIORAL GENETICS:

THE POTENTIAL FOR EARLY DIAGNOSIS AND TREATMENT OF READING DISABILITY

By

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Abstract

This thesis examines reading disability, behavioral genetics, and the potential for methods in genetics to identify pupils for early reading intervention. A literature review was conducted to convey first the current understanding of reading disability and its aspects. This review was conducted through searching online databases and using published journal articles and literature. Then a summary of the evolution of the field of behavioral genetics, and the methods and the discoveries that have been found. Through an understanding of both, a relationship between the two was established, and methods used in behavioral genetics are suggested as ways of screening for reading disability so that early interventions may be applied for the greatest positive effect.

Behavioral Genetics: The Potential for Early Diagnosis and Treatment of Reading Disability

Reading Disability

Reading disability can be deconstructed by examining the different requisite functions needed to effectively read (Vellutino et al., 2004). The first of these two functions is the ability to decode, which is essentially the ability to recognize words in isolation. Decoding carries different meanings and interpretations, so a clearer understanding is essential before moving forward. While "sounding out" words might seem like a plausible description, this skill doesn't satisfy enough of the ability to be sufficient. There are many words that are irregular and for which this skill doesn't apply, take *knight* for example. A better metric and definition for decoding concerns pseudowords, and the ability to pronounce them (Gough and Tunmer, 1986). Pseudowords are fake words that follow grapheme-phoneme conversion rules, such as *phun* or *cigtop.* A test of pronunciation of these pseudowords demonstrates an ability to apply the grapheme-phoneme rules, which is beyond the level of sounding out as it requires recognizing groups of letters and segmenting them to process the proper pronunciation. The second function is called comprehension, which is more straightforward, and is the ability to understand what is being decoded. Reading comprehension can be measured by listening comprehension, as it is the ability to understand the words and sentences that are being conveyed (Vellutino et al., 2004). It is clear that comprehension is necessary for reading ability by the simple analogy that I can decode French, but I cannot understand any of it.

The Simple View of reading follows that reading ability is the product of comprehension and decoding. This can quite literally be demonstrated as $R = C \times D$ where R is reading ability, C is reading comprehension, and D is decoding ability (Gough and Tunmer, 1986). All variables range on a scale from 0-1, with 0 being completely unable, and 1 being perfect. It makes sense then, that reading ability can be predicted by measuring ability to decode and listening

comprehension, by the Simple View. This breakdown of reading ability provides a lens to understand why individuals may suffer from a reading disability.

Three possibilities are left open by the Simple View and its equation that explains reading disability. The first is that an individual lacks the ability to decode words (or psuedowords as well). In the Simple View equation, this can be represented as *D* being close to zero, leading to the product of *D* and *C* to be lower. This has come to be known as dyslexia, and the cause remains unknown. What is common to many individuals with dyslexia is that they have otherwise normal faculties, including intelligence and sensory function, and that their dyslexia arises independent of neurological issues, physical disabilities, or socioeconomic disadvantages (Gough and Tunmer, 1986).

The second possible explanation of reading disability is a lack of reading comprehension skills. In the equation, this would be demonstrated as *C* is close to zero, and when multiplied by *D*, the resulting product *R* is lower. This condition has come to be called hyperlexia (Healy et al., 1982), and though it is not as common as dyslexia, it has been shown in multiple studies. Further, its discovery was important in providing support for the Simple View of reading, as it showed that reading ability is not just equated to decoding ability.

The third possible instance of reading disability by the Simple View is that both decoding and comprehension score low. This combination is compelling, especially when looked at in the opposite light. It has been shown that for most people *C* and *D* are positively correlated, those good at decoding tend to be good at comprehension, leading to a good reader. This instance, of both abilities being deficient, is the most general form or reading disability (Gough and Tunmer, 1986). As a point of interest, this positive relationship is in stark contrast to the other two options above, where both are cases of a negative correlation.

Behavioral Genetics

Behavioral Genetics is a field of research that emerged in the 20th century and attempted to begin disentangling the effects of nature from the effects of nurture. It has blended theories from Psychology and Genetics to push the science forward. At the time that research in Behavioral Genetics began, it was the prevailing theory that the environment, nurture, is responsible for almost all of the differences between individuals (Pinker, 2003). This theory has been challenged and largely disproven in the last half-century through the use of two primary research methods, the adoption study and the twin study (Plomin et al., 2016). What is critical to understand about this research and the statistical analysis involved, is that they deal with differences between individuals, not differences between groups (Jensen, 1972). Both of these methods examine the differences between twins, whether they were reared together or apart, or the differences between adopted siblings, and take these differences on aggregate to determine a degree to which a trait is heritable (Rhee and Waldman, 2002).

Heritability is the principal concern of Behavioral Genetics. A common confusion is that heritability can be used to predict traits, but heritability is only a measure of what is. Specifically, heritability is the extent to which the variation of traits for a given population can be attributable to genetic differences (Plomin, 2019). So if someone says that obesity is 70% heritable, they are not saying that 70% of obesity is caused from genetics, but what they are saying is that the differences between individuals is 70% attributable to genetics. The quintessential example of this stems from Mark Twain's children in barrels thought experiment. He imagined four kids all raised in barrels until the age of twelve, such that their environment was 100% controlled and the same for each. Once tested at age twelve for intelligence, because they have had the same environment, the differences between them could only be attributable to genetics. This would be their heritability for intelligence is 100% (Jensen, 1972). Now, the same four children, if all

raised in different environments and tested at age twelve, the differences in their intelligence scores would be less attributable to genetics and more to the environment, so heritability would decrease. But the amount attributable to genetics and the environment in this second case would be almost impossible to discern, and here is where the twin and adoption studies become useful.

There are a couple of different instances of both of these types of studies. First, I will discuss adoption studies. These experimental designs compare children with their genetic and adoptive parents, and the concept is relatively intuitive, if adopted children are more similar to their genetic parents for a trait, then nature (genetics) is the cause. If the adopted child is more similar to their adoptive parents, then nurture (environment) is the driving source (Rhee and Waldman, 2002). A further test of this, is with environmental siblings, who have different biological parents but grow up in the same household. The concept remains the same.

Genetic influence can be tested more dramatically using twin studies. Two types of twins exist, monozygotic (identical) and dizygotic (fraternal), and both are used to evaluate heritability. Monozygotic twins share 100% of the same DNA, while dizygotic share 50%. To clarify, all humans share greater than 99% of the same DNA (National Human Genome Research Institute, 2018). The differences exist in portions of DNA that make up our genes, and the differences here are the ones of importance. As for the 100% vs 50%, monozygotic twins are from the same sperm and egg, and inherit the same set of chromosomes. Dizygotic twins are from different egg and sperm cells and share half of the same chromosomes (Kaminsky et al., 2009). In the case of twins, if differences between individuals were caused entirely by genetics, then identical twins would correlate 1.0 for any trait, while fraternal twins would correlate 0.5 for the same trait. If genetics played no role, then fraternal twins should be just as similar as identical twins. The clearest test of genetic influence uses monozygotic twins that were separated early in life by

adoption, as this directly measures the degree to which the similarity between twins is genetic (Silventoinen et al., 2009).

Utilizing these two experimental designs, twin and adoption studies, researchers in Behavioral Genetics have made great strides in understanding the relationship between genes and the environment. The finding that genetics have significant influence on psychological traits has become ubiquitous (Plomin et al., 2016). In a study on intelligence from the world's literature, 10,000 pairs of twins were analyzed. Identical twins had a correlation of 0.85, while fraternal twins had a correlation of 0.60 (Bouchard and McGue, 1981). Results yielding genetic influence extend to studies of schizophrenia, breast cancer, depression, addiction, obesity, reading disability, and more (Plomin, 2019).

Behavioral Genetics and Reading Disability

Of the many traits that exhibit heritability, reading disability is one of them. Family history was first shown as an indicator for risk of reading disability (Vogler et al., 1985), but more importantly since twin-studies and sibling-pair linkage techniques have been used to demonstrate significant heritability for reading disability (Gayán and Olson, 1999). It is critical to be mindful that heritability does not predict what could be, and only evaluates what is. While parents can serve as indicators of a child's likeness to suffer from reading disability, more accurate markers are needed. Specifically, genomic indicators that will be able to more reliably predict an individual's likeness to develop a reading disability. With modern technology, this is now possible. Genome-Wide polygenic scores are now capable of predicting reading performance in schoolchildren (Selzam et al., 2017). By using polygenic scores as a screening method for reading disability, young pupils that are afflicted can be identified at a younger age and be supported with early reading interventions that been shown to have strong positive

influences. The earlier the interventions, the greater impact and success they have in creating better outcomes for students (Foorman et al., 1997). Moreover, heritability has been shown to increase from childhood to young adulthood in cognitive ability (Haworth et al., 2010), further demonstrating the need and importance of identifying and treating children from a young age. Behavioral genetics has revolutionized our understanding of nature and nurture, but it is now time for the research to be applied to the benefit of society. Early reading intervention is one avenue that must be pursued.

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