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Cognitive and familial risk evidence converged: A data-driven identification of distinct and homogeneous subtypes within the heterogeneous sample of reading disabled children

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ABSTRACT

The evident degree of heterogeneity observed in reading disabled children has puzzled reading researchers for decades. Recent advances in the genetic underpinnings of reading disability have indicated that the heritable, familial risk for dyslexia is a major risk factor. The present data-driven, classification attempt aims to revisit the possibility of identifying distinct cognitive deficit profiles in a large sample of second to fourth grade reading disabled children. In this sample, we investigated whether genetic and environmental risk factors are able to distinguish between poor reader subtypes. In this profile, we included reading-related measures of phonemic awareness, letter-speech sound processing and rapid naming, known as candidate vulnerability markers associated with dyslexia and familial risk for dyslexia, as well as general cognitive abilities (non-verbal IQ and vocabulary). Clustering was based on a 200 multi-start *K*-means approach. Results revealed four emerging subtypes of which the first subtype showed no cognitive deficits underlying their poor reading skills (*Reading-only impaired poor readers*). The other three subtypes shared a core phonological deficit (PA) with a variable and discriminative expression across the other underlying vulnerability markers. More specific, type 2 showed low to poor performance across all reading-related and general cognitive abilities (*general poor readers*), type 3 showed a specific letter-speech sound mapping deficit next to a PA deficit (*PA-LS specific poor readers*) and type 4 showed a specific rapid naming deficit complementing their phonological weakness (*PA-RAN specific poor readers*). The first three poor reader profiles were more characterized by variable environmental risk factor, while the fourth, *PA-RAN poor reader* subtype showed a significantly strong familial risk for dyslexia. Overall, when we zoom in on the heterogeneous phenomenon of reading disability, unique and distinct cognitive subtypes can be identified, distinguishing between those poor readers more influences by the role of genes and those more influenced by environmental risk factors. Taking into account this diversity of distinct cognitive subtypes, instead of looking at the reading disabled sample as a whole, will help tailor future diagnostic and intervention efforts more specifically to the needs of children with such a specific deficit and risk pattern, as well as providing a more promising way forward for genetic studies of dyslexia.

Abbreviations: PA, phonological/phonemic awareness; LS, letter-speech sound processing; RAN, rapid automatized naming.

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1. Introduction

While most children become fluent readers without much effort within a few years of reading instruction, a considerable number of children experience great difficulties in acquiring adequate literacy skills. Developmental dyslexia, a specific learning disability resulting in reading and spelling impairments despite normal intelligence and proper educational instruction (Lyon, Shaywitz, & Shaywitz, 2003; Peterson & Pennington, 2012), is only one possible cause of the reading failure experienced by the heterogeneous group of poor readers (Heim & Grande, 2012; Menghini et al., 2010; Pennington et al., 2012; van Bergen, van der Leij, & de Jong, 2014). Consequently, there have been numerous attempts to classify distinct and coherent cognitive profiles of reading failure. Traditionally, classification studies have divided poor readers into pre-defined categories, based on existing theoretical insights regarding the etiology of the different reading profiles, such as the 'IQ-reading performance discrepancy' hypothesis (Rutter & Yule, 1975), the 'phonological-core variable-difference' model (Stanovich, 1988) or according to a specific word decoding deficit or a broader decoding-comprehension deficit, i.e., the 'simple view of reading' (Gough & Tunmer, 1986). These earlier studies have significantly influenced the ongoing debate on what constitutes dyslexia and have at least supported the possibility to distinguish between a specific reading disabled, or dyslexic profile, and a more general poor, or garden-variety profile (Gough & Tunmer, 1986; Rutter & Yule, 1975; Stanovich, 1988). In more recent years however, new developments in the genetic underpinnings of reading disability added greatly to the current definition of dyslexia as a specific learning disability with a neurobiological origin and strong genetic disposition to develop reading difficulties (Byrne et al., 2006; Castles, Datta, Gayan, & Olson, 1999; Grigorenko, 2001; Pennington & Olson, 2008; Williams & O'Donovan, 2006). Reading problems experienced by dyslexic children are highly heritable and run in families (Pennington & Olson, 2008). Approximately 40–65% of children at familial risk are expected to develop dyslexia (Blomert & Willems, 2010; Pennington & Lefly, 2001; Pennington & Olson, 2008; Scarborough, 1990), indicating that the chances to develop dyslexia, given a dyslexic parent or sibling, amount to at least 10 times the population prevalence (i.e., 4–5%, Blomert, 2005). Although having a familial history of dyslexia is one of the strongest risk factors of the disorder (Thompson et al., 2015), it should not be considered as a pure genetic component (Plomin, Reiss, Hetherington, & Howe, 1994; Rutter & Silberg, 2002). Since at-risk children share both genes and home literacy environment with their close relatives, gene–environment interaction has been studied to understand reading deficits better. It has been shown that children with general impairments across various cognitive abilities are often more specially influenced by environmental risk factors and less by familial risk (Castles et al., 1999; Gayan & Olson, 2001; Grigorenko, 2001; Rack & Olson, 1993; Wadsworth, Olson, Pennington, & DeFries, 2000). A neuroimaging study supported two subtypes demonstrating the existence of two distinct brain activation profiles characterizing a primarily genetic poor reader type and an environmentally influenced more generally impaired type (Shaywitz & Shaywitz, 2005). One relevant question now is whether this environmental poor reader subtype can be differentiated from the poor reader subtype with a familial risk of dyslexia based on distinct, underlying cognitive deficit patterns. The present clustering study, for the first time, aims to investigate emerging cognitive subtypes of reading disability related to at-risk status based on history of dyslexia as well as environmental factors. We hypothesize that if the development of reading difficulties of a certain cognitive subtype of reading disability is influenced more strongly by a familial predisposition or alternatively more strongly by environmental risk factors, that this influence will also be reflected in their underlying cognitive deficit pattern. The possibility of identifying unique poor reading profiles will help tailor future diagnostic and intervention efforts more effectively than is currently possible.

Several important cognitive risk factors, or 'vulnerability markers', have been identified as important behavioral markers useful for the identification of reading disability. Traditional single deficit accounts of dyslexia point to weakness in phonological awareness (PA) as the core deficit in dyslexia (Adams, 1990; Goswami & Bryant, 1990; Ramus, 2003; Snowling, 2000; Vellutino, Fletcher, Snowling, & Scanlon, 2004). However, more recently a growing tendency emerged to view neurodevelopmental disorders like literacy impairment as the product of various interacting vulnerability markers with distinct, behavioral profiles as a result (Bishop, 2006, 2008; Pennington & Olson, 2008; Snowling, 2008, 2012). Family studies of dyslexia confirm that there is indeed a spectrum of reading disorders (Snowling & Hulme, 2012). Although a single PA deficit explanation of dyslexia is under discussion, it is undisputed that PA is a main characteristic of reading disability (e.g., Ramus et al., 2003; Sunseth & Bowers, 2002). Reading disabled children at increased familial risk for dyslexia show significantly weaker PA skills than non-risk controls (e.g., Carroll, Mundy, & Cunningham, 2014). In turn, both impaired and unimpaired at-risk children show PA problems (Puolakanaho et al., 2008; Snowling, Gallagher, & Frith, 2003), resulting in a parametric increase of reading problems (i.e., healthy < unimpaired at-risk < impaired at-risk: Boets, Wouters, Van Wieringen, & Ghesquiere, 2007; Elbro, Borstrøm, & Petersen, 1998; Moll, Loff, & Snowling, 2013; Pennington & Lefly, 2001; Snowling et al., 2003). Although it was always assumed that PA plays a causal role in reading acquisition (Snowling, 2000; Vellutino et al., 2004; Wagner & Torgesen, 1987), this has be opposed by others who indicated that a PA deficit develops in close relation with the developing reading deficit (Blomert & Willems, 2010; Castles & Coltheart, 2004; Castles, Wilson, & Coltheart, 2011; Morais, Cary, Alegria, & Bertelson, 1979; Perfetti, Beck, Bell, & Hughes, 1987). Genetic studies also indicated that a PA deficit is associated with a genetic predisposition of dyslexia which, although often differing in severity, can be present in both affected and unaffected family members (Berninger et al., 2006; Snowling, 2008).

Another candidate vulnerability marker of dyslexia is the ability to rapidly name familiar visual symbols such as objects, letters or colors, known as Rapid automatized naming (RAN) (Berninger, Abbott, Billingsley, & Nagy, 2001; Berninger et al., 2006; Bowers & Wolf, 1993). RAN is thought to reflect efficient and fast matching of visual and phonological codes and is