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Genetic and environmental etiology of speech and word reading in Chinese

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ABSTRACT

The present study examined the genetic and environmental etiology of the connection between speech and word reading in Chinese. A total of 371 pairs of Chinese twins (278 pairs of monozygotic twins and 93 pairs of same-sex dizygotic twins) were tested on speech discrimination and production, phonological skills, semantic skills, and Chinese word reading at the mean age 7.4 years. Results of univariate genetic analyses showed moderate genetic influences on speech, semantic skills, and Chinese word reading, while moderate shared environmental influences on phonological skills. The genetic correlations among all the variables were significant. Results of testing several models on the link between speech and word reading supported a common genetic factor underlying speech, phonological skills, semantic skills, and word reading in Chinese. The present findings suggest that around 50% to 60% of individual differences of speech, semantic skills, and word reading in Chinese are due to genetic factors. Individual differences of phonological skills appear to be relatively less heritable than those in English. This may be partly due to the fact that the Chinese script does not map directly on any segmental phonological information. A single common genetic etiology for speech, phonological skills, semantic skills, and word reading suggests that development of these skills is highly connected.

1. Introduction

The close connection between speech and reading development has been well established in typically developing children and children with disorders. Many research findings have reported overlaps among speech sound disorder, language impairment, and reading disability in English (e.g., Pennington & Bishop, 2009; Peterson, McGrath, Smith, & Pennington, 2007). Comorbidity among these disorders may suggest common underlying processes or common etiology. Deficits in the phonological pathway may be a common underlying cause for these speech-language related disorders in alphabetic languages. In some molecular genetic studies, genetic risk variants for speech sound disorder and reading disability were found to overlap (see review in Pennington & Bishop, 2009). Behavioural genetic studies with identical and fraternal twins learning to read English also show a significant genetic contribution of the early speech factor (based on articulation and nonword repetition) to later reading performance (Hayiou-Thomas, Harlaar, Dale, & Plomin, 2010). A strong genetic correlation between phonological awareness and word reading underscores the role of phonological skills in learning to read alphabetic languages (e.g., Gayán & Olson, 2001, 2003; Hayiou-Thomas, Harlaar, Dale, & Plomin, 2006; Hayiou-Thomas, Kovas, Harlaar, Plomin, Bishop, & Dale 2006). Most of these twin studies have been conducted in countries using alphabetic writing systems like US, UK, Australia, and Netherlands (see the meta-analysis of De Zeeuw, de Geus, & Boomsma, 2015), very little is known about nonalphabetic writing systems. In nonalphabetic writing systems like Chinese, semantics, rather than phonological skills, may be more important for learning to read. The present study examines whether there is a close connection, both behaviorally and genetically, between speech and word reading development in Chinese. If yes, the study further examines whether the connection is through a phonological pathway, a semantic pathway, or both. We introduce our study by reviewing evidence for the speech-reading connection in some behavioural and genetic studies. Then we give an overview of the unique characteristics of the Chinese writing system and previous research on speech and reading in Chinese.

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1.1. Continuities between the development of oral and written language

In learning an oral language, the first task of an infant is to master the perception and production of speech sounds. In the process of making sense of speech sounds, the child must determine which sound features are used to contrast different meanings and associated with which concepts. The speech sounds are thus represented phonologically and sequences of sounds are linked to semantic schemata in the mental lexicon. When a child's spoken vocabulary increases, phonological representation and its link to semantic representation may be refined (e.g., Metsala & Walley, 1998; Walley, Metsala, & Garlock, 2003). It is clear that development of written language skills is built upon earlier oral language skills. In the triangle word reading model, learning words implies making associations among orthography, phonology, and semantics (Seidenberg & McClelland, 1989). Two key components of word reading development, namely phonology and semantics, overlap with those of speech development. Therefore, both phonological and semantic skills are in principle important for speech and word reading development. Although basic speech skills develop quite early and reading is typically acquired later, the developmental restructuring of phonological representation and continuous reorganization of the mental lexicon which occur between ages one and eight (Fowler, 1991) may link the development of speech and reading in some way.

1.2. Roles of phonology and semantics in the development and difficulties of speech and reading

It would be helpful to first distinguish between two terms, “speech” and “phonological awareness”, as some people may find them confusing. Speech is the expression of or the ability to express thoughts and feelings by the articulation of words. Phonological awareness refers to an individual's awareness of the phonological structure, or sound structure, of words which may involve detection and manipulation of sounds. Simple speech perceptual skills are believed to develop naturally when infants have enough exposure to a rich language environment. Children gradually develop some abstract mappings from listening and talking and these form a phonological representation of the speech event. Development of speech perception and production may also arouse a child's sensitivity to some phonological units like rhymes, and this phonological skill helps learning of written words, especially in alphabetic languages where orthographic units map onto sound units like phonemes. So speech may contribute to reading through its route in phonological skill development.

On the other hand, speech inputs also help a child to develop a system of lexical representation that facilitates the representation and extraction of word meaning. For instance, discrimination of two simple vowels measured by a conditioned head-turn task at 6 months predicted vocabulary size and other language scores at 13 to 24 months of age (Tsao, Liu, & Kuhl, 2004). Vocabulary knowledge further facilitates the creation of mappings among orthographic, phonological, and semantic representations in a child's developing lexicon. So speech may contribute to later reading indirectly through its influences on vocabulary and semantic development.

There has been far more empirical support for the connection between speech and reading via a phonological route than a semantic route from studies with disabled readers or typically-developing readers. For instance, after reviewing about 20 studies on speech perception, McBride-Chang (1995) concluded that individuals with reading disability are weak at processing speech, particularly stop consonants. Bird, Bishop, and Freeman (1995) also reported that children with speech difficulties at age 5 went on to have deficits on phonological awareness at ages 6 and 7, and the majority displayed literacy problems at age 7. Presence of additional language difficulties did not significantly affect children's literacy outcomes. It appears that the root cause for reading disability is a poorly specified phonological representation. Similar support was also found in typically-developing readers. Significant correlations were reported between speech perception, and phonological awareness, and between speech and reading (e.g., Carroll, Snowling, Stevenson, & Hulme, 2003; Hurford, 1991; Roberts, 2005). Cheung (2007) has further reported that phonological awareness mediated significantly between speech processing and reading aloud. These findings suggest that the quality of phonological representation may be the underlying mechanism for the link between speech and reading development (e.g., Elbro, 1996). This may especially be the case in alphabetic languages.

1.3. Genetic contributions to the development and difficulties of speech and reading

Given support from some behavioural studies for the close link between speech and reading development, we would like to know whether the link is also a genetic one. In a review of nearly a hundred genetic studies of normal variations in language skills, Stromswold (2001) have concluded that almost all aspects of language acquisition and linguistic proficiency, including articulation, phonology, vocabulary, morphosyntax, and orthographic decoding, are influenced by genetic factors to some extent.

In general, both speech and language skills are found to be heritable and speech skills (heritability ranged from 37% to 95%) are somewhat more heritable than language skills (heritability ranged from 29% to 87%) (e.g., DeThorne et al., 2006; Hayiou-Thomas, Harlaar, et al., 2006; Kovas et al., 2005). This may especially be the case for individuals with specific language impairment (SLI). Bishop and Hayiou-Thomas (2008) reported that “Clinical SLI” was highly heritable (97%) while “Psychometric SLI” was entirely environmental. It was interesting that the “Clinical SLI” group had much lower speech scores than the “Psychometric SLI” group. It appears that speech deficits are the locus of genetic effects on language impairments and the speech deficits were more heritable than broader language deficits.

Given the high heritability of speech and language skills, how do the two skills relate to reading in both behavioural and genetic terms? In a UK longitudinal twin study, Hayiou-Thomas et al. (2010) reported that preschool speech and language skills were predictive of reading throughout the elementary school years. Both genetic and environmental factors played a role in the relationship between oral language skills and subsequent reading, but genetic factors played a much stronger role in the relationship between speech skills and later reading. However, it is noteworthy that nonword repetition, a measure of phonological short-term memory, was included in the speech factor of this study. This might have elevated the connection between speech and reading in the study.

Apart from speech and language skills, phonological skills have also been found in many behavioural studies to be salient for learning to read alphabetic orthographies. Would this relationship have any genetic foundation? Phonological awareness and phonological memory have been found to have moderate to strong heritability (ranged from 38% to 83%) (e.g., Bishop et al., 1999; Gayán & Olson, 2003; Kovas et al., 2005). The genetic correlation between the two phonological skills and reading was found to be moderate to strong (Gayán & Olson, 2003; Hayiou-Thomas, Kovas, et al., 2006). This suggests that phonological skills and reading may share some common genetic factors.

As for semantic skills, vocabulary is considered as a measure of individual differences in how well semantic and lexical knowledge is acquired. Unlike speech and phonological skills, the heritability of vocabulary is somewhat lower with a group heritability of 33% in Stromswold's (2001) meta-analysis of eight studies. However, Olson and his colleagues showed that the genetic contribution to vocabulary increased with age, from 0.29 in pre-kindergarten to 0.44 in Grade 2, to 0.57 in Grade 4 (Olson et al., 2011). The strong genetic role of the speech-reading link in alphabetic languages reviewed above raises the question of whether such a link would be found in a nonalphabetic script, Chinese. Although the genetic role of semantic skills has been