Chapter 8
Developmental Dyslexia:
Genes and Environment

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ABSTRACT

The aim of this chapter is to enhance the understanding of the complex and multifactorial etiology of developmental dyslexia, with particular emphasis on the interactions between genetic and environmental factors. Through the presentation of key single-deficit hypotheses related to the causes of the disorder, the analysis of the contributions of genetic and environmental factors, the presentation of the relevant research findings on the interactions between these factors, as well as the multiple deficit models for developmental dyslexia, the chapter seeks to advance the understanding of the underlying mechanisms of this heterogeneous and multifactorial learning disorder. Such multidimensional and synthetic approaches in this field are expected to provide new insights into the understanding of developmental dyslexia, while could contribute to the development of more targeted and effective educational interventions, making better use of the effects of environmental factors.

INTRODUCTION

Around 5% of school-age children have severe difficulties in learning how to read and/or spell, a condition known as dyslexia (Vlachos et al, 2013a). Developmental dyslexia (DD) is a mild form of neurodevelopmental disorder (Brimo et al., 2021; Peterson & Pennington, 2012) attributed to neurobiological (Kim, 2021), genetic (Gialluisi et al., 2021), and environmental factors (Mascheretti et al., 2018). It manifests with a variety of symptoms and encompasses a broad range of cognitive difficulties (Vlachos & Chalmpe, 2021). Individuals with dyslexia face challenges in acquiring written language skills, particularly in reading, despite having normal intellectual abilities and adequate educational experiences (Peterson & Pennington, 2012). The significant heterogeneity of the disorder's symptoms (Chalmpe & Vlachos, 2025) and the fact that individuals with dyslexia, beyond sharing common difficulties or

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strengths, also exhibit individual differences, make it difficult to establish a short, comprehensive and generally accepted definition (Vlachos, 2010).

One of the first definitions for DD was proposed by the World Federation of Neurology in 1968 (World Federation of Neurology, 1968). According to this definition, DD is described as a "*disorder characterized by difficulties in learning to read, despite appropriate education, adequate intelligence, and sociocultural opportunities*". For many years, dyslexia was considered as a result of a deficit in phonological processing which constitutes the core impairment for most individuals with dyslexia (Snowling, 1995). However, the progress of research revealed additional deficits beyond the phonological domain, such as difficulties in auditory and visual processing, sensory issues, and deficits in motor coordination (Ramus et al., 2003). This highlighted the need for broader definitions that would incorporate these findings.

Such a definition was formulated in 2010 by the British Dyslexia Association. According to this definition, DD is a learning disability that affects reading, spelling and phonological awareness and may be accompanied by problems with verbal memory and processing speed. It occurs across the whole range of intellectual abilities and is considered to be a continuous condition without clear boundaries. In addition, difficulties in areas such as language, motor coordination, concentration and organization, visual or auditory processing difficulties may co-exist. People with dyslexia often show strengths in areas such as creativity, problem solving and verbal skills (British Dyslexia Association, 2010).

Although the disorder is now systematically studied by various scientific fields and there is general agreement on its genetic, neurobiological, and environmental basis, a definitive consensus on a single definition has not been reached. Contemporary approaches argue that a single definition or a unified theory is insufficient to capture the complexity of DD (Ramus, 2004; Vlachos, 2010).

That is the reason, in recent years, researchers' attention has shifted to multidimensional approaches, recognizing that the etiology of dyslexia may be multiple and not one-dimensional (Peterson & Pennington, 2012). Current models attempt to explain the complex nature of the disorder based on multiple deficits (Pennington, 2006; van Bergen et al., 2014a), offering a more comprehensive understanding of the disorder.

The use of advanced genetic analysis methods, such as high-density genotyping chips, next-generation sequencing, specialized software, and the organization of databases, has been instrumental in identifying genes involved in the pathogenesis of dyslexia (Becker et al., 2017). Although the contribution of biological factors, particularly genetic ones, is important in the development of core deficits in cognitive processes leading to reading difficulties such as DD, the interaction between the brain and the environment appears to play a central role in the manifestation of the disorder (Vellutino et al., 2004). Environmental factors, such as the quality of teaching, seem to play a significant role either in enhancing or limiting reading difficulties.

The aim of this chapter is to advance understanding on the multifactorial and complex etiology of DD, with an emphasis on the interactions between genetic and environmental factors (G x E). Through the explanation of these interactions, it attempts to contribute on the understanding of how genes combine with environmental factors to contribute to the emergence and modulation of the symptoms of the disorder.

Initially, the chapter presents the most prominent and frequently cited single-deficit hypotheses regarding the causes of DD. Following this, the role of genetic factors and the environmental context in the manifestation of the disorder is examined. Finally, the chapter analyzes the contemporary models proposed for the etiology of the disorder and discusses recent research findings concerning G x E interactions in dyslexia.