Pathogenetic mechanisms of dyslexia: a review

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ABSTRACT: The authors of the present paper describe the probable pathogenetic mechanisms of specific reading disability, i.e. dyslexia. This analysis is grounded on the most widespread, yet hypothetical, cognitive deficit theories which account for the emergence and causation of dyslexia. These theories, coupled with the neurobiological underpinnings, imply that it is difficult to adopt a single approach in order to locate the causal relationships inherent to the disorder. Each one of the approaches provides critical insights to the mechanisms that underlie the development of literacy skills in normally developing children when exposed to literacy acquisition. Additionally, each theory attempts to explicate the factors that may be involved in the unexpected disruption of the learning process. The difficulty in establishing accurate phonological and orthographic representations in spite of adequate exposure to print, the transparency of mother tongue, brain architecture, and familial predisposition, all seem to contribute drastically to the deficient development of dyslexic children. Neuro-imaging studies along with molecular advances have been shedding even more light onto the backstage of dyslexic reading performance. Apparently, dyslexia could only be accounted for by an interactive approach within the framework of which the different influences from scientific trends may delineate the dyslexic phenotype.

Keywords: Dyslexia; reading skills; deficit theories.

INTRODUCTION

“Congenital word-blindness”, i.e. dyslexia, was first reported by general practitioners¹ who had identified this particular disorder over 100 years ago. Dyslexia, a Greek borrowing to literally mean ‘difficulties with words’, is a biologically based difficulty and presents with a twofold expression: a) acquired alexia, which is a brain lesion due to an insult (such as stroke or tumour) causing a disruption of a neural system related to reading skills²; and b) developmental dyslexia, which is usually associated with the non-acquisition of literacy skills². Dyslexia is an unexpected reading difficulty, which affects young children and adults, often co-occurring with other disorders. It is usually not predicted by lack of intelligence, lack of willingness to learn², poor education or abnormal developmental environment³. The effects of the disorder on literacy skills are persistent and chronic in nature²-⁴, placing impaired readers on the lower end of the normal distribution of reading ability².

Cognitive prerequisites for literacy acquisition

Language acquisition is inherently interwoven with alphabetic literacy development, i.e. learning how to read and spell. Distinct models of reading progress have depicted the constituents of this underlying processing system by providing different approaches to it. What has further attracted experimental interest is the specification of the factors that are responsible for the smooth functioning of the cognitive network, which paves the way towards normal literacy development. Abilities, such as reflecting upon and processing the sounds of language, or the transition from spoken to written form, have been strongly linked with the acquisition of linguistic and meta-linguistic skills.

Velluntino et al.⁵, in their extensive review of the

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acquired experience on dyslexia over a period of forty years, delineate the reciprocal and interactive relationships among cognitive processes and different types of knowledge required for the mastery of reading skills. According to the authors, at the core of learning to read, lies a three-dimensional schema, namely word identification, language comprehension and reading comprehension. The initial target of a new reader is to formulate a sight word vocabulary, that is, to establish and strengthen the relationship between the spoken and written forms of words. The second target is to master phonological decoding, that is, the ability to map the distinct graphic symbols onto the respective phonemic value units, i.e. graphemes to phonemes, letters to sounds. The attainment of these targets enhances meta-linguistic properties, particularly phonological awareness, orthographic awareness and syntactic awareness. It is the smooth functioning of these properties which will equip the reader with accuracy and fluency in order to tackle the alphabetic code. What will, ultimately, enable readers reach the threshold of reading abilities is the engagement of long-term and working memory, and the assimilation of written speech conventions.

The profile of dyslexia: Reading skills deficiencies

When a normal reader sets out to read a printed text, they move through three phases: in the pre-reading phase, judging from the topic or any accompanying visual aids, they deploy any previous knowledge, world knowledge or domain-specific knowledge, which enables them to adjust to the expected the content and context of the text, recognise its style, potential audience and publication origin, and guess the author’s intentions. In the actual reading phase, the reader aims to understand the content of the text, identify what is difficult to understand, and use appropriate strategies to resolve potential comprehension problems. Furthermore, they attempt to recognise the text structure, reconstruct it, guess the meaning of unknown words, pay attention to finer nuances accentuated by punctuation marks, seek specific information, justify their answers according to the text, extract underlying meanings, generate questions, comment on the author’s ideas, and compare and contrast it to other texts. In the post-reading phase, the reader summarises the content of the text, retains the essential information, contemplates the feelings triggered by the specific text, dares to answer more theoretical, abstract questions related to the text, and, occasionally, is inspired to produce a text of their own.

The transition from one to the following phase of reading comprehension may appear quite familiar, smooth, and manageable, only because the systematic teaching, constant practice, exposure to printed language, and the incentive to read have transformed the act of reading into an automatic practice. A mature reader would not struggle with how they read; the purpose of reading is what they read in order to understand it.

However, when a dyslexic reader is confronted with a printed text, they are forced to decipher strings of words, sequences of paragraphs, and pages of books, bearing one question in mind: how? They have difficulty in appreciating the qualitative traits of a given text, because the quantity of what they need to decode may be discouraging. The task to identify every component of written speech, from the smallest meaningful unit, the phoneme, to a whole word, then a sentence, and then a paragraph, is, under the dyslexic circumstances, cumbersome. By the time the dyslexic reader has reached the end of a text, they might have forgotten the beginning, meaning that they need to look back through the text, in order to retrieve it and understand it. The effects of reading decoding deficiencies are unavoidably reflected on the encoding skills, namely the orthographic skills and knowledge. Because the linkage between the spoken and printed words is not firmly established, the dyslexic person would resort to their visual memory skills in order to produce written speech, but would always question how the words are written instead of what is written.

The hidden profile of dyslexia:
Cognitive deficit theories

Visual deficits

There have been studies suggestive of the existence of more refined subtypes of dyslexia, the visual deficit being a very influential and strongly recurring one. According to the proponents of this theory, because words are initially visually encoded, reading difficulties should be the result of a deficient visual system or of visuo-spatial attention defying the causal role of phonological awareness in dyslexia. Obviously, these scholars would adopt visual deficit-oriented diagnostic tools and criteria when evaluating a dyslexic child and when designing the suitable reading intervention programmes.

The visual system is composed of two basic subsys-
tems, i.e. the sustained and the transient ones. The transient system inhibits the sustained one in order to help the visual system prepare for the next eye fixation\textsuperscript{9,10}. Lovegrove et al.\textsuperscript{10} suggested that there is a strong transient system deficit in children with specific reading difficulties. Longitudinal studies reported by Stein\textsuperscript{9} posited that early visual competence was predictive of later reading ability. Obviously, a deficit in the transient system could inflict both the visual and the phonological route of word processing\textsuperscript{9,10}. An impediment in formulating solid orthographic representations could be the cause and the result of visual memory deficits, considering that continuous feedback from the print enhances phonological awareness and processing\textsuperscript{11,12}.

However, Goulandris et al.\textsuperscript{11} suggested that neither perfect visual acuity nor fixed reference prevent both normally developing and dyslexic children from acquiring normal reading skills, even when the IQ had been controlled for. Shovman and Ahissar\textsuperscript{7} examined strictly visual aspects of reading abilities of dyslexics, having controlled for phonological, morphological and semantic factors. The visual deficit theory did not account for the performance of dyslexic participants, who performed equally well as the controls. Their results were in line with the findings of Ziegler et al.,\textsuperscript{13} corroborating dyslexics’ non deficient performance in symbol strings, yet displaying impaired results with letter and digit strings. More recently, Suárez Coalla and Cuetos Vega\textsuperscript{8} sought to determine whether developmental dyslexia may be characterised as a visual-perceptual disruption disorder. They concluded that visual perception and visual discrimination tools do not measure reading problems sufficiently, even though there might be few cases of dyslexic children presenting with perceptual problems as well.

**Language-based deficits**

**Phonological coding deficits**

The basic purpose of reading is the comprehension of the reading target. In order to achieve comprehension a complex, swift process of constant decoding of the written input needs to occur. The difficulty in developing highly specified phonological representations of words may be at the root of the literacy problems of dyslexic children\textsuperscript{2,14}, whose impairment lies in their inability to rapidly access the phonological lexicon\textsuperscript{2}, identify\textsuperscript{2}, decode, and recode the words.

The phonological deficit or ‘phonological representations’\textsuperscript{15} hypothesis denotes that there is a breakdown in the links that unify auditory/visual input with speech output\textsuperscript{16}. Mapping orthography onto phonology is completed with the involvement of semantic representations, which renders reading development an online, interactive process\textsuperscript{17}. Considering that the process of reading is disrupted due to erroneous or correct yet significantly delayed decoding, the ultimate goal of reading, i.e. comprehension, is unavoidably hampered\textsuperscript{2,15}. The reading intervention programmes which focus on phonics instruction in order to enhance phonological awareness in deficient readers and have beneficial results\textsuperscript{5} constitute the most manifest evidence of what underlies dyslexia.

Empirical evidence from the literature has focused on the poor performance of dyslexic children on variable tasks, which draw on phonological awareness, such as phoneme manipulation\textsuperscript{18}, and verbal short-term memory, as demonstrated in tasks such as digit span and non-word repetition\textsuperscript{19}. Dyslexic readers are also deficient at rapid automatized naming tasks. This is indicative of their difficulty in rapidly accessing not only lexical items but also their phonological representations\textsuperscript{20,21}. On the other hand, their rapid auditory processing skills seem to be adequately functioning\textsuperscript{22}. Moreover, dyslexics’ non-phonological language skills, such as grammatical abilities, are preserved, even though they score below the average\textsuperscript{23}.

Finally, it seems that failure in word boundaries discrimination along with persistent problems with syllable and phoneme discrimination are quite specific to English dyslexic children\textsuperscript{24}. Such an observation raises the important issue of the effect different orthographies exert on dyslexic readers. An orthography based on the consistent grapheme-to-phoneme correspondence renders word decoding rather easy, in which case visual and contextual features are employed as compensatory strategies\textsuperscript{14}. However, dyslexic readers of transparent orthographies are still compromised in terms of their reading speed\textsuperscript{25}, and phonological tasks\textsuperscript{14}, hence the universality of the phonological deficit in dyslexia regardless of the orthography.

**The double deficit hypothesis**

Quite early in the research, Denckla and Rudel\textsuperscript{20} validated the importance of speed deficits for distinguishing between dyslexia and other learning disabilities. They, thus, designed the Rapid ‘Automatized’ Naming tests which required serial or continuous decoding of colours,
objects, numbers and letters. It was objects and letters that resulted as the most critical predictors of dyslexia, signifying that dyslexic children may be deprived from becoming proficient in such retrieval processes. Interestingly, while rapid naming processing was correlated with dyslexia, the same was not true of rapid auditory processing, which did not prove to be predictive of reading abilities. Wolf and Obregón drew a speculative hypothesis to account for reading impairment. They suggested that, in conjunction with further deficits, it is failure in the function of a “specific timing mechanism” that explains decoding delay. This initial conceptualisation resulted in the postulation of the double-deficit hypothesis for developmental dyslexia. The hypothesis is structured upon the condition that phonological and processing speed deficits may contribute – independently or simultaneously – to the occurrence of reading difficulties. Three types of reading disability emerge: one caused by phonological skills deficits; a second caused by slow naming speed, affecting orthographic processing and reading fluency; and a third caused by a combination of both types of deficit, hence the ‘double deficit’. Interestingly, the double deficit theory seems to be influenced by the transparency of the language in question. In their longitudinal study, Papadopoulos et al. found that deficient reading skills in Greek, which is a fairly transparent orthography, depended on the contribution of the Rapid ‘Automatized’ Naming predictors, i.e. naming speed and phonological skills, in a different way and at different points of development.

Orthographic Depth Hypothesis:
Dyslexia in different languages

Alphabetic scripts differ in the degree of transparency. The notion of transparency, according to the alphabetic principle, reflects both the extent of consistent grapheme-phoneme correspondence, and the invariant grapheme-phoneme relationship. Visualising alphabetic scripts as extending over a continuum, at one end stand transparent, shallow, or regular languages, such as Serbo-Croatian, while, at the opposite end, lie opaque, deep, or irregular scripts, such as Hebrew. The way that different alphabetic systems reflect the phonemic organisation of their spoken form constituted the axis around which the ‘Orthographic Depth Hypothesis’ was built. The principal postulations of this hypothesis allude to the differential demands that orthographic systems place on their learners, even though “the underlying neuro-cognitive deficit” remains invariant. Moreover, according to this theory, transparent orthographies would necessitate phonological resources for decoding, while opaque scripts would depend on the morphology of the target lexical items. In shallow scripts, a combination of both decoding routes is also quite plausible.

The segmentation between transparent and opaque languages incited researchers towards investigating whether the effects of dyslexia could be conditioned by the nature of the script. Indeed, accumulating evidence from transparent languages, such as Brazilian Portuguese, German, Greek, French, and Turkish, advocates that phonological impairment is mitigated to a certain extent by the regularity of the orthography. Essentially, phonological awareness, auditory perception, and memory skills are spared. Nonetheless, occasional unexpected failures on phonemic segmentation tasks plausibly signify a flawed phoneme-grapheme storage and access, which might be smoothened, over time, by the very regularity of the language. Nonetheless, the interaction of transparency with phonological awareness does not necessarily improve word processing at later stages of reading acquisition.

The common denominator of transparent languages seems to be the deficit in “automatization skills”, i.e. in decoding speed. Nonwords, particularly morphophonologically irrelevant with real words, function or high-frequency words, and numerals, were among the most significant predictors of dyslexia in such languages. Apparently, deficient naming speed skills might hamper the rapid grapheme-phoneme conversion, while reading speed, in general, could be affected by the cognitive requirements for word recognition and naming. Despite potential speed defects, dyslexic young learners of a regular language do perform more quickly and accurately on such tasks than learners of an irregular orthography. Eventually, relatively high reading accuracy, indicative of the orthographic transparency, along with listening comprehension, which surfaced as a critical measure of reading fluency, compensate for the weaknesses of such learners.

Rapid naming proved to be the most deficient cognitive domain in Chinese dyslexic children. Ho et al., in their comprehensive study, attempted to determine which factors cause reading difficulties in a non-alphabetic language, such as Chinese. They concluded that dyslexia in learning to read Chinese could be due to multiple deficits (rapid naming, orthographic, visual,
and phonological processing), because of the peculiar script-sound and script-meaning mappings of Chinese, along with the chosen instruction of the language system at schools.

**The hidden profile of dyslexia: Neurobiological underpinnings**

**Neuroscience findings**

Learning to read is an acquired ability obtained through targeted instruction and systematic training. When children begin their formal tuition, what they actually gain is a change in the architecture of their brain. The more a child is exposed to print and trains in reading, the more the brain learns how to acquire this complex skill and becomes specialised for reading. And it is through systematic instruction that the reading neural system development is triggered.

According to neuroscience studies, there are three basic neural systems involved in the reading process, all of which are mainly located in the left hemisphere:

a) an anterior system in the left inferior frontal region;

b) a dorsal parieto-temporal system involving angular gyrus, supramarginal gyrus and posterior portions of the superior temporal gyrus;

c) a ventral occipito-temporal system involving portions of the middle temporal gyrus and middle occipital gyrus. Articulation, subvocal reading, and naming evoke another neural system in the inferior frontal gyrus, namely Broca’s area.

It seems that dyslexic reading process operates based on a mechanism different from that operating in normal reading, as shown in functional neuroimaging studies. In studies using a noninvasive method of functional brain imaging, namely magnetic source imaging techniques, dyslexic children showed not only increased activity, but also active involvement, located in the right posterior temporal and inferior parietal areas. These regions were analogous of the left activation areas of their non-dyslexic peers, which may have been activated as a compensatory strategy. Papanicolaou et al. delineated the neural dyslexic profile, aiming at shedding light onto the brain mechanisms involved in linguistic operations. The distinct brain activation profile of dyslexic children was particularly manifest in phonological decoding tasks. The neuropsychological activation in left tempo-parietal regions, which are essential for reading performance and reading skills acquisition, was not the proper one in the cases of dyslexia. However, the researchers posit that early and carefully designed intervention may alter the neural systems, which preserve their plasticity through childhood.

The neural bases of normal and dyslexic reading fluency in dyslexia is a rather understudied aspect of reading ability, which became the research object for Christodoulou et al. The adult participants of the study (typical readers and dyslexic ones) were asked to judge whether sentences, presented to them at slow, medium, and fast rates, were semantically meaningful or not. Evidently, it was at the fast rate that the disparity between the two groups grew most prominent, with the dyslexic group displaying disproportionately low performance. It was under this condition that semantic verbal knowledge was shown to differentiate the two groups, since activation in the left inferior frontal gyrus and left superior temporal gyrus of the dyslexic adults was significantly lower than the controls; hence, the diminished reading comprehension skills canonically inherent in dyslexia. Also, researchers pinpointed a defect in the functioning of the cerebellar component of the reading network.

Raschle et al., having examined pre-reading children with a familial risk for developmental dyslexia, demonstrated that the neural systems of two areas in the brain, i.e. left prefrontal region and posterior parieto-temporal and occipito-temporal areas, may be linked during rapid auditory processing and phonological processing tasks, respectively. These findings suggest a new neural phenotype of children at risk for developing dyslexia. Likewise, Clark and et al. examined pre-literate children at risk of dyslexia longitudinally, that is, a year before formal reading instruction began at school, a year after reading instruction, and a year after dyslexia had been diagnosed. The primary auditory cortex (Heschl’s gyrus) and the primary visual cortex (lingual gyrus, V2) were found to be thinner in children who later did develop dyslexia. This means that, before learning to read, these children displayed reduced neuro-anatomical capacity for processing auditory and low-level visual information. Notably, the thickness of the auditory cortex was not altered significantly over development, giving support to the postulation that the neural basis of dyslexia may originate in exactly that region.

**The genetics of dyslexia**

The first genetic linkage of a gene to reading disability partially accounting for the manifestation of dyslexia dates back to 1983. It was Smith et al. who suggested that a gene on chromosome 15 – later dubbed the
DYX1 locus\(^47\) – may causally underlie specific reading disability. Since then, molecular sciences and technologies have progressed to such an extent that researchers are in a position to identify more accurately possible genetic risk factors related to developmental disorders, such as dyslexia\(^48\). Moreover, scientists have been discovering candidate genes which are responsible for the attainment of various aspects of reading skills and sub-skills (see\(^39\) for an overview). Genetic linkage studies have uncovered dyslexia loci and genes; there are at least ten inter-related candidate genes, which may account – to a certain extent – for the causation of dyslexia\(^49\).

In order to understand the underlying mechanisms and associate the genes that are involved in genetically complex traits\(^50\), the endophenotype concept was introduced in psychiatry over a decade ago and has since been vastly employed in molecular sciences as well\(^51\). The psychometric measures administered for the assessment of the key cognitive skills, which are the basis of reading ability (that is, orthographic processing, phoneme awareness, rapid automatized naming, and phonological short-term memory), would constitute an example of endophenotypes in dyslexia\(^39\). Scientists have attempted to join genetic findings, i.e. dyslexia susceptibility genes, with neuroimaging techniques in order to identify the variability in reading ability\(^52\). Because they link genetic information and behavioural output, “neuroimaging endophenotypes”\(^39\) seem to promise a revolutionary opening towards the disclosure of the genetics of dyslexia.

CONCLUSION

Dyslexia remains a fascinating mystery, a multifaceted disorder, whose causality is still concealed, in spite of the massive research outcomes. The unexpectedly deficient acquisition of decoding and encoding skills constitutes a global trait for children who have received systematic teaching and who have not been diagnosed with any obvious sensory or neural deficits. Currently, new theories have emerged suggesting that dyslexia may be grounded on sensory deficiencies, visual and auditory sensory problems attracting most of the scientific attention. However, in order to approach the deeper truth of dyslexia carefully designed researches need to be initiated whereby the extent of reading experience should be controlled for, since it exerts changes on the brain. This is the reason why longitudinal studies with either pre-literate children or illiterate adults emerge as being crucial\(^53\). More accurate research findings would influence both screening and evaluation measures as well as intervention programmes, smoothing thus the academic experience and altering the future prospects of dyslexic people.

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Conflict of interest

None declared.
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