Reintroducing Dyslexia: Early Identification and Implications for Pediatric Practice

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Dyslexia is a common learning disorder that renders children susceptible to poor health outcomes and many elements of socioeconomic difficulty. It is commonly undiagnosed until a child has repeatedly failed to learn to read in elementary school; this late diagnosis not only places the child at an academic disadvantage but also can be a precursor to psychiatric comorbidities such as anxiety and depression. Genetic and neuroimaging research have revealed that dyslexia is heritable and that it is undergirded by brain differences that are present even before reading instruction begins. Cognitive-behavioral research has revealed that there are early literacy skill deficits that represent red flags for dyslexia risk and can be measured at a preschool age. Altogether, this evidence points to dyslexia as a disorder that can be flagged by a pediatrician before school entry, during a period of heightened brain plasticity when interventions are more likely to be effective. In this review, we discuss the clinical implications of the most recent advances in dyslexia research, which converge to indicate that early identification and screening are crucial to the prevention or mitigation of adverse secondary consequences of dyslexia. We further highlight evidence-based and practical strategies for the implementation of early risk identification in pediatric practice so that physicians can be empowered in their ability to treat, educate, and advocate for their patients and families with dyslexia.

The development of reading proficiency in childhood is a public health issue: literacy is a widely recognized determinant of health outcomes and is associated with many indices of academic, social, vocational, and economic success.[1] In a recent National Academy of Medicine summary, the author highlights that duration of education, which is highly dependent on reading proficiency, is a better predictor of health and long life than cigarette smoking or obesity.[2] Children skilled in reading perform better in school, attain higher levels of education, experience lower rates of disease, are less likely to be incarcerated or experience poverty, are more likely to find employment, and achieve higher average incomes as adults compared with children who fail to achieve reading proficiency.[3] For many children with reading impairments, however, the process of learning to read is rife with struggle and frustration, and these children are left susceptible to adverse secondary outcomes, including anxiety and depression. A neurobiologically based specific learning disorder, dyslexia, affects 5% to 10% of children[4,5] and is a persistent barrier to reading acquisition.

Dyslexia (or word-level reading difficulty[6]) is predominantly

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characterized by a core deficit in phonological processing (the ability to recognize and manipulate speech sounds), which results in impairments in decoding ("sounding out" words), spelling, and word recognition.[7] These impairments almost always lead to difficulties in reading fluency and comprehension, reduced vocabulary, lower content knowledge,[8] and a decline in overall school performance.[9] Dyslexia cannot be explained by poor hearing or vision, low language enrichment, or lack of motivation or opportunity.[10] According to the *Diagnostic and Statistical Manual of Mental Disorders, Fifth Edition*, dyslexia falls under the umbrella of a "specific learning disorder" that "impedes the ability to learn or use specific academic skills (eg, reading, writing, or arithmetic)."[11]

Although there are many negative outcomes associated with dyslexia, particularly salient to the pediatrician is the association between dyslexia and poor mental health.[12,13] Children with dyslexia are more likely to suffer from generalized anxiety[14,15] and also exhibit higher rates of depression.[14,16] Because screening for dyslexia is not routinely performed, the direction of causation between dyslexia and comorbid mood disorders in each case is unclear, and this uncertainty can preclude effective early treatment. A mood disorder may be identified in a child with unidentified comorbid dyslexia when it is the dyslexia that is antecedent and causative, obscuring the primary target for intervention.

In addition to mood disorders, speech and language problems are frequently comorbid with dyslexia because both dyslexia and developmental language disorders can be characterized by poor phonological awareness[17–19] and other language deficits (eg, oral language comprehension).[20] Approximately one-half of children identified with dyslexia have language disorders, and approximately one-half of children with language disorders have dyslexia.[20] Dyslexia typically results from a core deficit in phonological processing; however, it is important to note that language deficits (eg, low vocabulary or low oral listening comprehension) can also lead to reading problems, especially problems with reading comprehension. Importantly, speech and language problems commonly precede problems in learning to read, so children with speech and language problems should be flagged as being at increased risk for dyslexia.[21]

There are many other developmental and psychiatric conditions that are frequently comorbid with dyslexia, further jeopardizing these children's health and academic outcomes. In total, 20% to 40% of children with attention-deficit/hyperactivity disorder have dyslexia,[22] and children with autism spectrum disorder are also at increased risk of having dyslexia.[23] Other behavioral disorders, such as conduct disorder and oppositional defiant disorder, are also associated with dyslexia.[24] As many as 85% of children with dyspraxia (developmental coordination disorder) have dyslexia,[25] and children with dyscalculia (math learning disorder)[26] and dysgraphia (writing learning disorder)[27] are more likely to have dyslexia than those without. Knowledge of dyslexia within pediatric practice is paramount in considering the most appropriate treatments for these many coexisting disorders.

Despite increasing collaboration among educators, physicians, neuroscientists, speech and language pathologists (SLPs), and psychologists, dyslexia is often overlooked in the field of general pediatrics, perhaps because the diagnostic label of dyslexia is not often used in practice, having been replaced largely by education language of strengths and weaknesses. The clinical implications of a reluctance to use dyslexia as a diagnostic label include children failing to receive an adequate response to early risk signs, appropriate interventions in school, and mental health support.

In this article, we provide an up-to-date overview of dyslexia, specifically addressing common knowledge gaps, neurobiological underpinnings of the disorder, and ways in which pediatricians can play an active role in the early identification of dyslexia risk.

THE ETIOLOGY OF DYSLEXIA

The etiology of dyslexia is multifaceted, including genetic, perceptual and cognitive, neurobiological, and environmental factors.[9] Dyslexia is strongly heritable, occurring in up to 68% of identical twins of individuals with dyslexia and up to 50% of individuals who have a first-degree relative with dyslexia.[19,28–30] Several genes[31–34] have been reported to be candidates for dyslexia susceptibility; it is thought that most of these genes play a role in early brain development.[31,34–38]

Furthermore, various studies have revealed atypical brain characteristics in individuals with dyslexia compared with their peers.[39] In functional MRI studies, researchers have indicated that reading for typical readers takes place predominantly in left-hemispheric sites of the brain, including the inferior frontal, superior temporal, temporoparietal, and occipitotemporal cortices.[40] As a group, individuals with dyslexia show hypoactivation in the left-hemisphere reading systems.[41] The structural and functional atypicalities in these brain regions include reduced gray matter volume,[42] hypoactivation in response to reading-related functional MRI tasks,[43] and weaker functional connectivity between key areas of the

reading network.[44] Importantly, differences in brain structure and function characteristic of dyslexia can be observed before the start of formal reading instruction, indicating that dyslexia does not result from a struggle to learn to read but, rather, represents a biological disposition present at the preschool age or perhaps as early as infancy.[45–47] Altogether, these neuroimaging findings suggest that children predisposed to dyslexia enter their first day of school with a brain that is less equipped to learn to read.

It is worth noting that reading proficiency is strongly associated with socioeconomic status[48–50]: 80% of fourth grade students from low socioeconomic backgrounds read below grade-level proficiency.[51] In particular, children with inadequate exposure to language are more likely to struggle with reading.[52] However, the diagnosis of dyslexia does not include socioeconomic disadvantage as a potential cause. Although these children do not necessarily meet a diagnosis of dyslexia, children who struggle with reading, regardless of etiology, have been shown to suffer the same adverse health and psychosocial consequences and benefit from interventions that have been primarily developed to address deficits associated with dyslexia.[53–55]

Although neuroimaging research has been invaluable in establishing the biological basis of dyslexia and reading impairments, neuroimaging technology (eg, brain MRI) does not have the ability to screen or diagnose dyslexia on an individual level, nor is it likely that this will be the case in the future. At this point, neuroimaging is not able to clearly disentangle differential neurobiological effects of dyslexia versus other reading impairments.[39,56] For these reasons and many others, cognitive-behavioral strategies are much more useful in screening.

The classic simple view of reading posits that skilled reading involves ≥2 major cognitive components: word recognition (including decoding and phonological awareness) and language comprehension (eg, knowledge of vocabulary and language structures); together, these strands coalesce to form what is classically known as the "reading rope."[57] Although the simple view of reading has been borne out by evidence,[58] its components are not single entities but are multifactorial, malleable, and context dependent (especially language comprehension) and cannot be captured in a single assessment.[59] Furthermore, recent research has revealed that skilled reading, especially in older children, is contingent on knowledge of academic language and the additional cognitive skills of perspective-taking and reasoning.[60]

In the past, dyslexia was diagnosed in the context of a discrepancy between reading ability and IQ, such that reading ability had to be 1 SD below cognitive abilities (IQ) for dyslexia to be diagnosed. However, this discrepancy model has been disproven, and dyslexia is no longer considered to be associated with IQ.[61]

Dyslexia is related to deficits in ≥1 strands of the reading rope and particularly to early struggles in phonological and/or phonemic awareness.[62] Other predictors include struggles in letter-sound correspondence, pseudoword repetition (the ability to pronounce spoken nonsense words), identifying rhyming sounds, rapid automatized naming (the ability to automatically retrieve the names of objects, letters, or colors), and deficits in oral language comprehension and receptive and expressive vocabulary.[63] These measures have been shown to be strong predictors of reading ability in the English language; in other languages, the precursors vary, and screening approaches should be tailored to a child's language environment. In this review, we focus on monolingual English speakers, however, and, among these children, these key linguistic and preliteracy measures can be assessed in children as young as 4 years old, and they can serve as crucial markers in identifying children at risk for dyslexia or other reading impairments.[64–66] Some of these literacy precursors measured in kindergarten have been shown to predict reading comprehension in the 10th grade.[67]

THE DYSLEXIA PARADOX AND THE RISKS OF DELAYED DIAGNOSIS

As children progress through the school system, reading becomes the expected vehicle for content learning; thus, it is imperative that children with dyslexia are identified early and receive intervention without delay. When at-risk beginning readers receive intensive early reading intervention, 56% to 92% of these children achieve average reading ability.[68] However, many children are diagnosed with dyslexia long after they first demonstrate recognizable struggles with preliteracy milestones.[69] Currently, children are typically diagnosed with dyslexia at the end of the second or beginning of third grade (and many much later), after they have already failed to learn to read over a long period of time and have fallen behind their peers academically.[70] This wait-to-fail approach fails to capitalize on the most effective window for intervention, which is during an earlier period of heightened brain plasticity in kindergarten and first grade.[70,71] Referred to as the "dyslexia paradox,"[63] the gap between the earliest time at which identification is possible and the time at which identification and treatment typically occur can preclude effective intervention and has profound academic and socioemotional implications for the developing child. Children at the 10th

percentile of reading ability may read as many words in 1 year as a child at the 90th percentile reads in a few days.[72]

In addition to the poor academic outcomes associated with untreated dyslexia, diagnosing children after a prolonged period of failure can have severe implications for children's mental health. Often perceived as lazy or labeled as "stupid," children with dyslexia may develop decreased self-esteem, which can progress to anxiety and depression.[16] Furthermore, children with learning disorders are less likely to complete high school,[73] less likely to attend programs of higher education,[74] and at increased risk of entering the juvenile justice system: 28% to 45% of incarcerated youth[75] and 20% to 30% of incarcerated adults[76] have a learning disorder. Additionally, adults with learning disorders are more likely to be unemployed and, on average, earn annual incomes well below the national average.[5] Given the prognostic benefit of early diagnosis and intervention and the many adverse consequences that can be avoided or mitigated, there is great value in identifying early risk for dyslexia in the pediatric clinic.

THE ROLE OF PEDIATRICIANS AND THEIR PROFESSIONAL ORGANIZATIONS IN DYSLEXIA RISK SCREENING

It is important to distinguish between screening for dyslexia risk and diagnosing dyslexia. Screening refers to a brief assessment that determines the risk of having or developing dyslexia, which can be undertaken at an early age before school entry.[75,77] Conversely, a formal diagnosis can only occur after reading instruction has begun and requires a more comprehensive neuropsychological evaluation, which can be motivated by a previous screening result.[76] Although attention to both screening and diagnosis is vital in ensuring that the appropriate interventions are implemented for the child, screening for risk of dyslexia is possible earlier in the developmental time course than is diagnosis; thus, it represents an opportunity for expeditious early intervention.

The consideration of any screening regimen requires that a valid and acceptable test be available, an effective and accessible means of treatment be available, and the potential benefits of screening outweigh the risks[78] without an undue burden to the practitioner or patient. In the case of dyslexia, screening children individually for risk can be accomplished quickly and inexpensively through a consideration of family history and through short behavioral assessments of early literacy abilities. Extensive evidence has revealed the benefit of an early evidence-based response to screening,[70,79,80] and the risks of implementing a screening process are minimal to negligible. A review of a child's family history of dyslexia is a worthwhile start to the process of early identification: a family history with positive results necessitates close monitoring, whereas a family history with negative results still requires a level of ongoing observation.

The risk of a false-positive result is present with any screening program, and, in the case of dyslexia screening, the risk is tantamount to further evaluation, monitoring, and educational supports. Although through these processes, demands are placed on the child and represent cost and effort on the part of practitioners, the burden of failing to identify these children early is ultimately greater than the burden of providing supplemental resources to a child needlessly. As discussed, although not all children who struggle with reading will meet the criteria for a dyslexia diagnosis, most children who struggle with reading will benefit from interventions designed to address dyslexia.

With the recognition that early literacy predictors of dyslexia can be identified before the start of kindergarten,[63,65] we can no longer afford to wait for screenings in children's first formal schooling experiences. In a 2009 position article negating visual deficiencies as the origin of dyslexia, the American Academy of Pediatrics stated that pediatricians should "be vigilant in looking for early signs of evolving learning disabilities."[81] The pediatrician's existing role in monitoring early child development and our understanding of the importance of early support for language and literacy development present pediatricians with the opportunity to implement dyslexia screening in well-child visits even before children are school-aged.

Pediatricians can contribute to a collaborative effort to screen for children at risk by capitalizing on their unique role in a child's early developmental trajectory and by taking advantage of their network of health and educational resources. It is important to note that many parents desire this proactive stance from their child's health care provider: more than one-third of surveyed parents indicated that they have not discussed reading with their pediatrician; nearly one-half of that group believed such conversations would be useful.[82] Pediatricians can also provide referrals to outside experts, such as neuropsychologists and SLPs, and communicate with patients' schools.[83]

Pediatricians typically rely on a developmental milestone checklist in evaluating a child's development in various domains; however, recent research reveals that there is great variability between the many available checklists, both in content and milestone age ranges.[84] Furthermore, although receptive and expressive language is accounted for in these checklists, a comprehensive inventory of key early literacy

measures that are crucial for assessing dyslexia risk is not included.

Early warning signs of dyslexia are visible before school entry[63,65]; thus, the pediatrician may be a child's first health or educational professional capable of identifying these signs and implementing a management plan. For example, pediatricians can document the extent to which a child can recognize rhyming sounds, repeat nonsense words, or report the sound that a letter makes. It is important to note that phonological deficits can present differently in different children, and children with dyslexia will vary in the specific tasks with which they show difficulty. Thus, screenings are used to pinpoint particular early literacy skills that may require remedial attention and also identify children who may eventually require a more detailed evaluation to come to a definitive diagnosis of dyslexia.

A MORE PROACTIVE APPROACH: SCREENING AND ADVOCACY

There are many methods by which pediatricians can work toward systematic early screening of dyslexia in their practices. Pediatricians should elicit a family history of dyslexia, recognize assessments done by schools that indicate a risk and/or diagnosis, and include dyslexia in the differential diagnosis for low self-esteem, depression, anxiety, or disruptive behaviors. The governing bodies of associations of pediatricians should provide training on dyslexia assessments and interventions as a part of ongoing continuing education so that pediatricians can become adept at implementing screening processes and at interpreting, monitoring, and responding efficiently to evaluations and interventions performed outside the clinic. The appropriate referral to outside consultants and interventionists should also be involved in this training when needed. Given the high overlap between dyslexia and deficits in speech and language, physicians should consider referring children who are at risk for dyslexia to an SLP who is trained in early literacy.[21] Furthermore, pediatric associations can partner with dyslexia researchers and education specialists to create educational resources and trainings that can assist pediatricians in providing education and support to their patients and families with or at risk for dyslexia.

Checklists, questionnaires, and interviews can be completed in conjunction with a child's parent to assess a child's key risk factors. Although these methods provide a quick account of a child's risk, they are often tools that are not scientifically validated or reliable[85] and are thus intended for a preliminary formative assessment only. Commonly used questionnaires like the Ages & Stages Questionnaires, for example, can be helpful as a starting point but do not provide a detailed assessment. Rigorously validated screeners composed of child-centered behavioral assessments (see ref 86 for a nonexhaustive evaluation of screening tools) are used to provide a reliable and unbiased testament of a child's risk status.

Pediatric clinics could consider hosting "screening days" with a literacy focus that can aim to simultaneously screen for early predictors of dyslexia while also facilitating a literacy-rich environment by making literacy materials available to families. Many pediatricians have already made great strides in promoting literacy within their clinics. The Reach Out and Read program has been effective in facilitating language and preliteracy skills in children through the distribution of books through primary care clinics.[87] A similar program with an additional screening component could be even more beneficial in supporting emerging literacy while also identifying children at risk for dyslexia.

A further possibility is the use of a standardized, brief 2- or 3-question first-step questionnaire, the likes of which have already been demonstrated to be successful in prescreening other conditions like depression[88] and attention-deficit/hyperactivity disorder.[89] Although no such standardized questionnaire yet exists, pediatricians can pose questions to a preschool-aged child's caregiver(s) pertaining to key risk factors for dyslexia. Affirmative answers to these first-step questions can be used to lead to a more detailed, validated screening tool that can be used to identify specific deficits present. There are storybooks and tablet- and smartphone-based gamified and self-administered screening tools currently being developed for the use in schools, clinics, or a child's home. These tools are being designed to be entertaining for the child and will be informative to the clinician in determining the appropriate next steps and referrals for further evaluation and intervention.

As new screening tools continue to become available in the coming years, it will be important for practitioners to be knowledgeable about the characteristics of an appropriate dyslexia screener and discerning in their selection. The ideal screener has been validated in a representative sample; has strong evidence for reliability, validity, and classification accuracy; has developmentally appropriate content given the age or grade level of the child; and has the capacity to measure both word recognition and linguistic comprehension.[77] A list of available dyslexia screening tools, along with an indication of their fulfillment of characteristics like those listed above, is presently available for practitioners to consult (see ref 86). An additional resource for practitioners is the What

Works Clearinghouse, which is used to provide evidence-based evaluations on literacy screening products as they become available.[90]

In addition to child-directed assessments, given the strong heritability of dyslexia, a crucial component of early identification is an assessment of the reading history of the child's parent(s) to determine the child's familial risk of dyslexia. Family history is both quick to elicit and informative in the global assessment of a child's risk. The Adult Reading History Questionnaire is an inventory[91] of an adult's literacy abilities and habits and can be used to indicate a reading impairment (see ref 92 for a digital version). Follow-up questions should be presented to a parent with a high-risk score to rule out an environmental explanation for reading impairment (eg, lack of formal reading instruction). This distinction is particularly important to consider in communities with considerable immigrant populations, who may be flagged by the Adult Reading History Questionnaire as "at risk" simply because they are adult learners of the dominant language. Regardless of a parent's dyslexia status, the quality of the home literacy environment is a strong predictor of reading outcome.[52,93,94] Thus, this parent inventory is useful not only because it can be used to indicate a child's possible familial risk but also because it can reveal less literacy-rich home environments that leave children with insufficient literacy materials and support, illuminating additional targets for intervention.

Beyond the clinic, the medical community can be vocal advocates in national conversations about dyslexia, many of which are currently happening in state legislatures; as of now, only a few US states lack state-level legislation focusing on early screening, teacher training, and/or instructional support (for an overview of state legislation, see ref 95). Despite such recent attention, there is much room for growth in pediatric neurocognitive research funding, which has lagged compared to adult neurocognitive disorders. Increased funding and research must be used to explore etiologic models, examine comorbid relationships, refine tools for the early identification of children at risk for dyslexia and other reading impairments, investigate additional tools for use in children for whom English is not their first language, and develop and evaluate intervention strategies and their effectiveness. A first-order goal should be the development of screening guidelines and tools for use during pediatric visits for 4- and 5-year-old children that can be used to identify children at risk before the optimal window for early interventions closes, while also refining guidelines to identify older children who were not screened earlier. Finally, with the help of policy makers, the current "failure" model of dyslexia must be replaced with a "support" model that enables school-, clinic-, and community-based early screenings and subsequent evidence-based response to screening through empowered and well-trained teachers within the general education framework. Physicians can be powerful agents of these positive changes, both at the level of their clinical practices and as advocates in their communities and beyond.

CONCLUSIONS

Our current knowledge of the neurobiological basis for dyslexia, its reliable developmental-behavioral predictors, the effectiveness of early intervention, and the myriad adverse effects of reading failure reveal a demand for a proactive, preventive approach (instead of a deficit-driven approach) to identify and treat children at risk. Early identification should start with an assessment of family history and should be followed with validated behavioral screening tools. After a positive screen, referrals to diagnosticians such as SLPs or neuropsychologists should be made. Diagnoses, when they occur, should be followed with letters to schools requesting the implementation of literacy intervention.

With time, new and innovative formats for screening will emerge. In acknowledging the significant effort devoted by pediatricians to screening various conditions, the future will require a consideration of novel approaches to office visits or increased community-based collaboration with preschools to accomplish screening for disorders that, like dyslexia, are of nontrivial prevalence and are associated with available and effective interventions. Pediatricians occupy a unique role in the lives of children such that they are well positioned to recognize and respond to risk factors for dyslexia even before children enter the education system; however, the delivery of dyslexia interventions is and will largely continue to be implemented outside the scope of the pediatrician's practice. Thus, the contributions of both pediatricians and other health and educational professionals are crucial to optimizing the process of identifying and treating dyslexia. Although the response to dyslexia screening and intervention is multifaceted and longitudinal, the trajectory of children's literacy outcomes has the potential to be improved through the implementation of early identification in pediatric practice.

ABBREVIATION

SLP: speech and language pathologist

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