Current Concepts

DYSLEXIA

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DEVELOPMENTAL dyslexia is characterized by an unexpected difficulty in reading in children and adults who otherwise possess the intelligence, motivation, and schooling considered necessary for accurate and fluent reading. Dyslexia (or specific reading disability) is the most common and most carefully studied of the learning disabilities, affecting 80 percent of all those identified as learning-disabled.[1] Although the diagnosis and implications of dyslexia were once quite uncertain, recent advances in our knowledge of its epidemiology, neurobiology, and genetics, as well as of the cognitive influences on this disorder, now allow physicians to approach dyslexia within the framework of a traditional medical model. This article reviews these advances and their implications for the approach to patients presenting with a possible reading disability.

EPIDEMIOLOGY OF DYSLEXIA

Recent epidemiologic data indicate that, like hypertension and obesity, dyslexia fits a dimensional model. In other words, within the population, reading ability and reading disability occur along a continuum, with reading disability representing the lower tail of a normal distribution of reading ability.[2,3] Dyslexia is perhaps the most common neurobehavioral disorder affecting children, with prevalence rates ranging from 5 to 10 percent to 17.5 percent.[4,5] Previously, it was believed that dyslexia affected boys primarily[6]; however, more recent data[7-9] indicate similar numbers of affected boys and girls. Longitudinal studies, both prospective[10,11] and retrospective,[12-14] indicate that dyslexia is a persistent, chronic condition; it does not represent a transient "developmental lag." Over time, poor readers and good readers tend to maintain their relative positions along the spectrum of reading ability.[10]

NEUROBIOLOGIC INFLUENCES

Heritability

Dyslexia is both familial and heritable.[15] Family history is one of the most important risk factors, with 23 percent to as much as 65 percent of children who have a parent with dyslexia reported to have the disorder.[16] A rate among siblings of affected persons of approximately 40 percent[15] and among parents ranging from 27 to 49 percent[15] provides opportunities for early identification of affected siblings and often for delayed but helpful identification of affected adults. Linkage studies implicate loci on chromosomes 6 and 15 in reading disability.[17-19]

Neurobiologic Studies

A range of neurobiologic investigations using postmortem brain specimens,[20] brain morphometry, functional brain imaging, and electrophysiology suggests that there are differences in the temporo–parieto–occipital brain regions between people with dyslexia and those who are not reading-impaired.[21,22] Some studies suggest differences in the striate or extrastriate cortex,[23] findings that coincide with those in a large body of literature describing anatomical lesions in posterior brain regions in acquired alexia, most prominently centered on the angular gyrus.[24]

COGNITIVE INFLUENCES

Reading — the process of extracting meaning from print — involves both visual–perceptual and linguistic processes. Theories of dyslexia have been proposed that are based on the visual system,[25] the language system,[26] and other factors, such as temporal processing of stimuli within these systems.[27,28] Whatever the contributions of other systems and processes, there is now a strong consensus among investigators in the field that the central difficulty in dyslexia reflects a deficiency within a specific component of the language system, the phonologic module, which is engaged in processing the sounds of speech.[29,30] According to the phonologic-deficit hypothesis, people with dyslexia have difficulty developing an awareness that words, both written and spoken, can be broken down into smaller units of sound and that, in fact, the letters constituting the printed word represent the sounds heard in the spoken word.

The Phonologic-Deficit Hypothesis

The language system can be conceptualized as a hierarchical series of components. At the higher levels are neural systems engaged in processing, for example, semantics, syntax, and discourse; at the lowest level is the phonologic module, dedicated to processing the distinctive sound elements that constitute language. The functional unit of the phonologic module is the phoneme, defined as the smallest discernible segment of speech; for example, the word "bat" consists of three phonemes: /b/ /ae/ /t/ (buh, aah, tuh). To speak a word, the speaker retrieves the word's phonemic constituents from the lexicon, assembles the phonemes, and then utters the word. Conversely, to read a word, the reader

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must first segment the word into its underlying phonologic elements. There is abundant evidence that a deficit in phonologic analysis is related to difficulties in learning to read: phonologic measures predict later reading achievement[31-33]; deficits in phonologic awareness (i.e., awareness that words can be broken down into smaller segments of sound) consistently distinguish children with dyslexia from those who are not reading-impaired[34,35]; phonologic deficits persist into adulthood[13,14]; and instruction in phonologic awareness promotes the acquisition of reading skills.[31,36-39] Additional findings of the strong heritability of phonologic awareness suggest "that it may be the main proximal cause of most genetically-based deficits in word recognition, and thus it may be the most appropriate focus for diagnosis and remediation."[40]

IMPLICATIONS OF THE PHONOLOGIC MODEL OF DYSLEXIA

Basically, reading comprises two main processes — decoding and comprehension. In dyslexia, a deficit at the level of the phonologic module impairs the ability to segment the written word into its underlying phonologic elements. As a result, the reader experiences difficulty, first in decoding the word and then in identifying it. The phonologic deficit is domain-specific; that is, it is independent of other, nonphonologic, abilities. In particular, the higher-order cognitive and linguistic functions involved in comprehension, such as general intelligence and reasoning, vocabulary,[30] and syntax,[41] are generally intact. This pattern — a deficit in phonologic analysis contrasted with intact higher-order cognitive abilities — offers an explanation for the paradox of otherwise intelligent people who experience great difficulty in reading.[42]

According to the model, a circumscribed deficit in a lower-order linguistic (phonologic) function blocks access to higher-order processes and to the ability to draw meaning from text. The problem is that the person cannot use his or her higher-order linguistic skills to access the meaning until the printed word has first been decoded and identified. Suppose, for example, that a man knows the precise meaning of the spoken word "apparition"; however, until he can decode and identify the printed word on the page, he will not be able to use his knowledge of the meaning of the word and it will appear that he does not know the word's meaning.

APPROACH TO THE DIAGNOSTIC EVALUATION

Guided by knowledge of the presumed underlying pathophysiology, the clinician seeks to determine through history, observation, and psychometric assessment, first, whether there are difficulties in reading that are unexpected, given the person's age, intelligence, or level of education, and second, whether there are associated linguistic problems at the level of phonologic processing[43] (Table 1). The way reading and language are assessed depends on the age and educational level of the patient.

EVALUATION OF SCHOOL-AGE CHILDREN

Presenting problems most commonly center on school performance ("she's not doing well in school"), and parents (and teachers) often do not realize that the cause is a difficulty in reading. Thus, an evaluation for dyslexia should be considered for all children presenting with learning problems, even if reading difficulty is not explicitly reported.

Assessment of Reading Ability

Reading ability is assessed by the measurement of decoding skills and comprehension. In the school-age child, the most important element of the psychometric evaluation is how accurately the child can decode words — that is, read single words in isolation. Reading passages allows bright children with dyslexia to use the context to guess the meaning of a word they might otherwise have trouble decoding. As a result, readers with dyslexia often perform better on measures of comprehension and worse on measures of the ability to decode isolated single words. In practice, the reliance on context makes such tests as multiple-choice examinations, which typically provide scanty context, especially burdensome for readers with dyslexia.

Assessment of Intelligence

The role of tests of intelligence in the diagnosis of dyslexia is controversial.[45,46] Traditionally, the concept of dyslexia as an unexpected difficulty in reading has been interpreted as underachievement in reading relative to ability (IQ) — that is, a discrepancy between the level of reading achievement predicted on the basis of IQ and the actual level of reading achievement. Consequently, IQ tests are generally used to assess dyslexia in school-age children, and in fact, eligibility for special-education programs in public schools is usually based on the demonstration of an ability–achievement discrepancy. More recently, many have questioned the requirement of such a discrepancy.[34,35] The issue is complex. In certain respects, children with dyslexia identified on the basis of an ability–achievement discrepancy do not seem different from those of average intelligence whose dyslexia is identified solely on the basis of low reading achievement for chronologic age; both have a deficit in phonologic processing[34,35] and follow the same developmental trajectory in reading.[11] At the same time, it should be recognized that the use of an approach based on such a discrepancy in the diagnosis of dyslexia is important for the identification of very bright children who have dyslexia.

Approximately 75 percent of children meeting the criteria for a discrepancy between ability and achievement also have low reading achievement[47]; however, the remaining 25 percent of children meeting discrepancy criteria — most of whom are extremely bright and also manifest a phonologic deficit — do not meet low-achievement criteria and would be excluded from special-education services if the low-achievement criterion were the only one employed. Thus, a consensus is developing that in school-age children the criterion of "unexpected" reading difficulties may be met by children of at least average intelligence who meet either discrepancy criteria relative to IQ or low-achievement criteria relative to chronologic age.[46,48]

SPECIAL CONSIDERATIONS IN YOUNGER AND OLDER AGE GROUPS

Assessment at School Entry

Currently, most children's reading disability is not diagnosed until they are in the third grade, or about nine years old.[49] The application of what has been learned about the acquisition of reading and the availability of tests of phonologic skills now make it possible, first, to identify children with dyslexia even before they fail in reading,[50] and then, to provide appropriate early interventions (see "Management," below). A history of language delay or of not attending to the sounds of words (trouble playing rhyming games with words or confusing words that sound alike), along with a family history, are important risk factors for dyslexia.[16] The most helpful measures in predicting difficulties are those often referred to as reading-readiness tests[16] (Table 2).

Assessment of Bright Young Adults

The developmental course of dyslexia has now been characterized. First, dyslexia is persistent; it does not go away.[10,11] On a practical level, this means that once a person is given a diagnosis of dyslexia there is no need for reexamination after high school to confirm the diagnosis. Second, over the course of development, the ability to decode words becomes more accurate and automatic in skilled readers; they do not need to rely on context for word identification. The skills of readers with dyslexia, too, become more accurate over time, but they do not become automatic. Residua of the phonologic deficit persist,[13,14] so that reading remains effortful, even for the brightest people with childhood histories of dyslexia.[51] The failure either to recognize or to measure the lack of automaticity in reading is, perhaps, the most common error in the diagnosis of dyslexia in accomplished young adults. It is often not understood that tests measuring word accuracy may be inadequate for the diagnosis of dyslexia in young adults at the level of college or graduate or professional school, and that for these people, timed meas-

TABLE 1. CLUES TO DYSLEXIA IN SCHOOL-AGE CHILDREN.*

History
Delayed language
Problems with the sounds of words (trouble rhyming words, confusion of words that sound alike)
Expressive language difficulties (mispronunciations, hesitations, word-finding difficulties)
Difficulty naming (difficulty learning the letters of the alphabet and the names of numbers)
Difficulty learning to associate sounds with letters
History of reading and spelling difficulties in parents and siblings
Reading
Difficulty decoding single words
Particular difficulty reading nonsense or unfamiliar words
Inaccurate and labored oral reading
Slow reading
Comprehension often superior to isolated decoding skills
Poor spelling
Language
Relatively poor performance on tests of word retrieval (name the pictured item)
Relatively superior performance on tests of word recognition (point to the pictured item)
Poor performance on tests of phonologic awareness
Clues most specific to young children at risk for dyslexia
Difficulty with tests assessing knowledge of the names of letters, the ability to associate sounds with letters, and phonologic awareness
Clues most specific to bright young adults with dyslexia
Childhood history of reading and spelling difficulties
Accurate but not automatic reading
Slow performance on timed reading tests (e.g., Nelson–Denny Reading Test[44])
Penalized by multiple-choice tests

*Clues are based on history, observations, testing, or a combination of the three.

TABLE 2. TYPES OF TESTS USEFUL IN IDENTIFYING CHILDREN AT RISK FOR DYSLEXIA AT THE TIME OF SCHOOL ENTRY.

Letter identification (naming letters of the alphabet)
Letter–sound association (e.g., identifying words that begin with the same letter from a list: doll, dog, boat)
Phonologic awareness (e.g., identifying the word that would remain if a particular sound was removed: if the /k/ sound was taken away from "cat")
Verbal memory (e.g., recalling a sentence or a story that was just told)
Rapid naming (rapidly naming a continuous series of familiar objects, digits, letters, or colors)
Expressive vocabulary, or word retrieval (e.g., naming single pictured objects)

ures of reading must be used to make the diagnosis. However, there are very few standardized tests for adult readers that are administered under timed and untimed conditions; the Nelson–Denny Reading Test[44] is an exception. The reading measures[52,53] commonly used for school-age children may provide misleading data on some adolescents and young adults, since they assess reading accuracy but not automaticity (speed).

DIAGNOSIS

For bright young adults especially, the history is perhaps the most sensitive and accurate indicator of dyslexia. A history of phonologically based language difficulties (e.g., mispronouncing words, speech punctuated by hesitations and dysfluencies), of trouble reading new or unfamiliar words, of spelling difficulties, and of requiring additional time for reading and taking tests relative to the level of education achieved represents the distinct diagnostic signature of dyslexia.

Tests of reading, spelling, language, and cognitive abilities (for school-age children) represent a core battery for the diagnosis of dyslexia; additional tests of academic achievement (in mathematics, for example), language, or memory may be administered as part of a more comprehensive evaluation of academic, linguistic, or cognitive function. There is no single test score that is pathognomonic of dyslexia. As with any other medical diagnosis, the diagnosis of dyslexia should reflect a thoughtful synthesis of all the clinical data available, including the history, observations, and testing data (Table 1). What the clinician is seeking is converging evidence of a phonologically based reading disability, as indicated by a disparity between the person's reading and phonologic skills and his or her intellectual capabilities, age, or level of education. Dyslexia is distinguished from other disorders that may feature reading difficulties prominently by the unique, circumscribed nature of the phonologic deficit, one that does not intrude into other linguistic or cognitive domains (Table 3). Primary sensory impairments should be ruled out, particularly in young children; other laboratory measures, such as imaging studies, electroencephalography, or genetic tests, are ordered only if there are specific clinical indications. Although there have been important advances in the use of imaging to study cognitive function, such technology is still reserved for investigational purposes. Attention deficit–hyperactivity disorder may also affect learning in both children and adults. It is an entirely different disorder from dyslexia; they differ in their proposed mechanisms, symptoms, assessments, and interventions.[57] A proportion of patients with dyslexia (12 to 24 percent)[5] will also have attention deficit–hyperactivity disorder, and if there is any suggestion that inattention may be a problem, the patient should be examined for specific symptoms that meet the criteria for attention deficit–hyperactivity disorder of the fourth edition of the *Diagnostic and Statistical Manual of Mental Disorders*.[58]

TABLE 3. SOME DISORDERS THAT MAY PRESENT WITH READING DIFFICULTIES.

Developmental dyslexia
Phonologic deficit primary
Reading impairment at the level of single-word decoding
Other components of language system intact (e.g., syntax, semantics)
Intelligence not affected and may be in superior or gifted range
Language-learning disability*: the primary deficit involves all aspects of language, both phonologic and semantic–syntactic
Reading difficulty at the level of both decoding and comprehension
Prominent language difficulties
Measures of verbal intelligence significantly affected by language deficit; may be in subaverage range
Acquired alexia†: the loss or diminution of reading ability
Result of trauma, tumor, or stroke (e.g., occlusion of posterior cerebral artery)
Several forms reflecting specific loci of neuroanatomical lesions (e.g., alexia with or without agraphia)
May be accompanied by other features reflecting locus and extent of the lesion
Hyperlexia‡
Word-recognition ability substantially better than reading comprehension
Early intense interest in words and letters
Exceptional word-recognition ability, apparent by the age of five years
Very poor reading comprehension
Disordered language development, especially affecting aural comprehension
Deficits in reasoning and abstract problem-solving
Behavioral atypicalities affecting interpersonal relationships

*Discussed in Catts and Kamhi.[54]

†Discussed in Damasio and Damasio.[55]

‡Discussed in Aram and Healy.[56]

MANAGEMENT

The management of dyslexia demands a life-span perspective; early on, the focus is on remediation. As a child matures and enters the more time-demanding setting of secondary school, the emphasis shifts to the important role of providing accommodations. Since physicians are frequently asked about various reading programs for dyslexia, they should understand the key elements of an effective training program. To learn to read, all children must discover that spoken words can be broken down into smaller units of sound, that letters on the page represent these sounds, and that written words have the same number and sequence of sounds heard in the spoken word. When children have made these associations, usually by the end of first grade, they have discovered the "alphabetic principle" and have essentially

broken the reading code. Children with dyslexia do not easily acquire the basic phonologic skills that serve as a prerequisite to reading; consequently, concepts such as phoneme awareness must be taught explicitly. Operationally, this is accomplished with systematic and highly structured training exercises, such as identifying rhyming and nonrhyming word pairs, blending isolated sounds to form a word, or conversely, segmenting a spoken word into its individual sounds. Furthermore, it is now recognized that although awareness of phonemes is necessary for learning to read, it is not sufficient.[59] In addition to learning that words can be segmented into smaller units of sound (phoneme awareness) and that these sounds are linked with specific letters and letter patterns (phonics), children with dyslexia require practice in reading stories, both to allow them to apply their newly acquired decoding skills to reading words in context and to experience reading for meaning.[60] A number of protocols differing in method, format, intensity, and duration of the reading intervention are now being tested in large-scale studies; data from these trials should provide important information to clarify further which specific programs are most effective for particular groups of children with dyslexia.[60] Large-scale studies to date have focused on younger children; as yet, there are few or no data available on the effect of these training programs on older children. People with dyslexia and their families frequently consult their physicians about unconventional approaches to the remediation of reading difficulties; in general, there are very few credible data to support the claims made for these treatments (e.g., optometric training, medication for vestibular dysfunction, chiropractic manipulation, and dietary supplementation).[61]

The management of dyslexia in students in secondary school, and especially college and graduate school, is based on accommodation rather than remediation. College students with a history of childhood dyslexia often present a paradoxical picture; they are similar to their unimpaired peers on measures of word recognition yet continue to suffer from the phonologic deficit that makes reading less automatic, more effortful, and slow.[13,14,51] For these readers with dyslexia the provision of extra time is an essential accommodation; it allows them the time to decode each word and to apply their unimpaired higher-order cognitive and linguistic skills to the surrounding context to get at the meaning of words that they cannot entirely or rapidly decode. Although providing extra time for reading is by far the most common accommodation for people with dyslexia, other helpful accommodations include allowing the use of lap-top computers with spelling checkers, tape recorders in the classroom, and recorded books (materials are available from Recording for the Blind and Dyslexic, telephone 800-221-4792) and providing access to syllabi and lecture notes, tutors to "talk through" and review the content of reading material, alternatives to multiple-choice tests (e.g., reports or orally administered tests), and a separate, quiet room for taking tests. With such accommodations, many students with dyslexia are now successfully completing studies in a range of disciplines, including medicine.

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