

Preschool Children Who Have Atypical Patterns of Development

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OBJECTIVES

After completing this article, readers should be able to:

1. Describe the probability of identifying a specific etiology for mental retardation.
2. Delineate the strongest influences on young children's language development.
3. Explain how autism can be distinguished from other disorders.
4. Describe the evaluation of preschool children who have atypical patterns of development.

Case Presentation

At 42 months of age, CG, a sturdy appearing Caucasian male, was unable to communicate clearly because of speech and language difficulties. His parents reported that he spoke "90% gibberish and 10% words" and often answered questions irrelevantly by reciting the alphabet or a nursery rhyme. Although very affectionate, he did not interact much with peers. His behavior was mostly self-directed, and he had difficulty maintaining attention to therapist-directed tasks. He was easily overstimulated and often flapped his arms with excitement. He also was observed to shake his head repetitively, stare at objects, and arrange his toys in straight lines. He became very upset if his toys were moved by someone else. His father, a computer software engineer, was reported to have started talking "late" and had had difficulty with reading in elementary school. Nonverbal cognitive testing found CG's intelligence to be within the average range, but his language abilities were at the level of a 26-month-old. His articulation errors varied, depending on the length of his utterance, and he had a disorder of syntax (grammar) and semantic (word meaning) language.

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Introduction

Children who have atypical patterns of development are a diagnostic challenge in the preschool years. Delayed or inappropriate use of language often is the most prominent symptom, and it frequently is accompanied by unusual social interaction patterns, odd behaviors, or limited play skills. Atypical behaviors include those considered to be uncommon, such as perseveration on specific activities, adherence to strict daily rituals, aloofness, and echolalia (repeating words, phrases, or sentences). Atypical patterns are characterized by deviations in the order of skill acquisition, discrepancies among areas of development, or regression and loss of previously established abilities.

Some "atypical" patterns of development represent normal variation; others reflect disorders of cognition, language development, or social functioning. Three major disorders that must be considered in a diagnosis include mental retardation, language disorders, and autism. Contributing to the diagnostic challenge is that many early signs and symptoms of developmental dysfunction are nonspecific, and key features of these disorders overlap substantially (Figure). The long-term prognosis for affected children varies considerably, and accurate determination of the child's strengths and weaknesses will assist parents, teachers, and therapists to support the child's development.

Mental Retardation

Mental retardation refers to a wide range of nonprogressive brain dysfunction syndromes that affect the development of both intellectual and social-adaptive abilities. Historically, definitions of retardation were based on the severity (mild, moderate, severe, and profound) of cognitive impairment as measured by standardized intelligence tests and the individual's level of social-adaptive functioning. In 1992, the American Association of Mental Retardation changed the definition of mental retardation by shifting the emphasis from the severity of cognitive impairment to the intensity level (intermittent, limited, extensive, and pervasive) of support services required by the individual for daily functioning. This change in classification reflects the capacities and interactions of the affected individual with his or her environment.

PREVALENCE

Mental retardation occurs in 2% to 3% of the population. Mildly affected individuals comprise approximately 85% to 89% of this group; moderately affected, approximately 7% to 10%; and severely and profoundly affected combined, 1% to 2%. Nearly twice as many males as females are affected. The prevalence of mild retardation varies inversely with socioeconomic status of the family, but moderate-to-severe retardation occurs with equal frequency across all social classes.

CLINICAL FEATURES

Clinically, mental retardation is characterized by a delayed pattern of development. In the majority of cases, the child progresses sequentially through the typical stages but develops at a slower pace than normal. The age at which parents first become concerned about their child correlates with the degree of retardation. Children who have major central nervous system anomalies and severe retardation (intelligence quo-

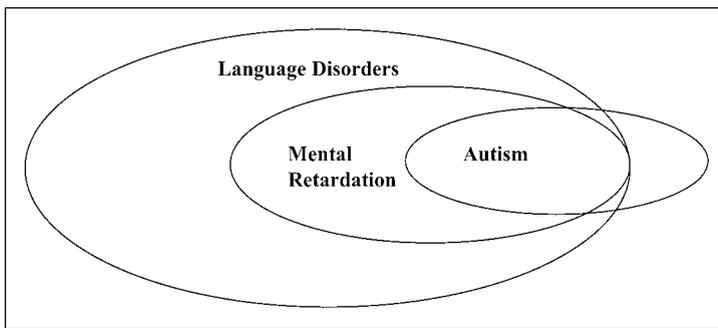


FIGURE. Relationship of autism, language disorders, and mental retardation.

tient [IQ] <50) usually are identified in the first year of life because of motor delays. Mild-to-moderate retardation is identified later when delays in language and play behaviors become more noticeable.

Children who are mentally retarded exhibit a range of deficits in language and communication ability compared with their normally developing peers. However, the language development of mildly affected children resembles that of younger age children matched for mental age, suggesting that mental retardation does not include a linguistic deficit above and beyond that attributed to the cognitive deficit. However, nearly 50% of children who have severe-to-profound mental retardation fail to acquire any symbolic communication skills, such as use of words or signs. Mental retardation per se does not have a primary impact on interest in social interaction. Although the most severely affected infants may be slow to learn typical social games, they are otherwise responsive and affectionate toward others. On the other hand, approximately 40% to 60% of mentally retarded children have significant emotional and behavioral disorders due, in large part, to their immature communication skills. For example, older preschoolers who have cognitive delays are more likely than their normally developing peers to respond to frustration with tantrums or physically aggressive behaviors (eg, biting, grabbing objects from peers) than by expressing themselves verbally or seeking assistance from adults. Even minor changes in routines or environment may lead to adverse behavioral reactions and stereotypies, such as self-injury, aggression, or rocking.

ETIOLOGY

Mental retardation is not a disease, but rather the developmental consequence of a pathogenic process affecting the central nervous system. Although often an isolated disability, mental retardation commonly is associated with other developmental disorders, including cerebral palsy, epilepsy, vision and hearing impairments, communication disorders, pervasive developmental disorders, and attention deficit disorder.

A definite or probable cause can be identified in approximately 25% of mentally retarded individuals, but this varies widely with the degree of cognitive impairment. Approximately 80% of those who have severe-to-profound retardation have an identifiable etiology; this decreases to less than 50% among those who have mild impairment. Approximately 60% to 75% of mental retardation can be attributed to prenatal factors, including genetic causes such as chromosomal disorders (eg, trisomy 21 and fragile X syndrome), inborn errors of metabolism (eg, phenylketonuria), neurocutaneous disorders (eg, tuberous sclerosis and neurofibromatosis-type 1), and many nonchromosomal dysmorphic syndromes (eg, Angleman, Joubert, and Williams syndromes). Other prenatal causes include abnormalities of brain development (eg, cerebral dysgenesis and congenital hydrocephalus) and central nervous system insults from toxins (eg, fetal alcohol syndrome), teratogens (eg, chemotherapy, anticonvulsants), infections (cytomegalovirus, *Toxoplasma*), or maternal illness. Perinatal causes such as prematurity, asphyxia, and birth injury are less common, accounting for perhaps 10% of cases. Postnatal factors (eg,

meningitis, encephalitis, trauma) contribute fewer than 10% of all cases.

Language Disorders

The terms speech and language disorder, developmental language disorder, developmental dysphasia, or language impairment are used commonly to describe children whose communication skills are significantly delayed compared with their cognitive development. The term specific language impairment is applied to children who experience difficulty acquiring language skills despite normal nonverbal intellectual ability. However, a language disorder also can be present in a child who has mental retardation if the delay in language development is greater than that expected for the child's intellectual impairment.

PREVALENCE

Speech and language problems are very common among young children. Nearly 20% of 2-year-olds are thought to have delayed onset of speech. By age 5 years, the prevalence of speech and language disorders is about 19%, of which 6.4% is speech impairment, 4.6% is both speech and language impairment, and 8% is language impairment.

CLINICAL FEATURES

To communicate effectively with others, children must learn the structure, content, and functional use of speech and language. The structure of language consists of rules about word sounds (phonology); modifications of words to form plurals, possessives, and tenses (morphology); and sentence formation and grammar (syntax). The content of language consists of a store of known words (lexicon) and their meanings (semantics). Functionally effective and appropriate communication depends on mastery of language pragmatic skills—the use of language in the context of social discourse. Pragmatic behaviors include topic maintenance, reciprocity between speaker and listener, eye contact, and providing the partner with optimum information to facilitate conversation.

If these were truly independent functions, these structural components of language might be expected to exist singly or in a wide range of combinations. In clinical expression, however, problems with phonology, morphology, and syntax frequently cluster together and may affect only expressive abilities or both expression and comprehension of language.

The fourth edition of the American Psychiatric Association's *Diagnostic and Statistical Manual of Mental Disorders* (DSM-IV) identifies five types of communication disorders, including expressive language disorder, mixed receptive-expressive language disorder, phonological disorder, stuttering, and communication disorder (not otherwise specified). Many experts also recognize "higher order" language processing deficits that involve inappropriate use of semantic and pragmatic aspects of language (Table 1). A pure form of receptive language

deficit that involves intact expressive ability does not occur in children because children are unable to speak in the absence of verbal comprehension.

Verbal dyspraxia deserves special mention as a cause of language delay because it can result in completely unintelligible speech. As infants, affected children may have engaged in little or no babbling. Onset of speech often is delayed significantly, and once they begin to talk, articulation errors may be inconsistent, varying from sound to sound, word to word, sentence to sentence, or day to day. The deficit in motor praxis (the ability to plan and execute coordinated movements) appears to be at the level of motor planning, and the disorder often responds poorly to intensive speech therapy. Verbal dyspraxia frequently accompanies other problems with language development and may be part of a generalized coordination

disorder that affects feeding and fine motor and gross motor development.

Behavioral problems are common among children who have disorders of language development. As might be expected, they often experience a high degree of frustration, especially when they have difficulty understanding social situations or using language to regulate their own behavior or interactions with others. Children who have limited communication skills frequently withdraw from social situations or become very anxious in unfamiliar settings. Young children may rely heavily on schedules and routines as means of ordering or controlling their environment. As communication skills improve, many of these behaviors normalize. However, some degree of social impairment continues in older individuals whose language disorders persist long after they have acquired functional conversational language skills. Emerging evidence indicates that these children have difficulties correctly interpreting a variety of visual and auditory stimuli, including the emotional intentions and expression of others.

TABLE 1. Rapin and Allen Clinically Defined Subtypes of Developmental Language Disorders

Mixed Receptive and Expressive Disorders

- *Verbal auditory agnosia (word deafness)*: Profoundly impaired comprehension deficit because of very deficient phonologic decoding with resultant severe expressive deficit. Children are nonverbal or have very limited expression with defective phonology
- *Phonologic/syntactic deficit disorder*: Comprehension equal to or better than production, which is impoverished, with short, often ungrammatical utterances, impaired phonology, and a limited vocabulary

Expressive Disorders

- *Verbal dyspraxia*: Extremely dysfluent with sparse output and very poor phonology
- *Speech programming deficit disorder*: Fluent with jargon

Higher Order Processing Disorders

- *Lexical deficit disorder*: Severe word-finding deficits and comprehension difficulty for connected speech. Spontaneous language superior to language on demand. Inadequate formulation of discourse. Early on, jargon, pseudo-stuttering because of word-finding deficit, and simplified syntax are frequent. Later, phonology and syntax are unimpaired.
- *Semantic pragmatic deficit disorder*: Verbosity with comprehension deficits for connected speech. Word-finding deficits and atypical word choices frequent. Phonology and syntax unimpaired. Inadequate conversational skills: speaking aloud to no one in particular, poor maintenance of topic, answering beside the point of a question.

Reprinted with permission from Rapin I. Practitioner review: developmental language disorders: a clinical update. J Child Psychol Psychiatry. 1996;37:643-655.

ETIOLOGY

Gender and genetics exert the strongest influences on language development. The overwhelming majority of children who have language disorders are boys (85%), and specific language impairments tend to cluster in certain families at a much higher rate than in the general population. A careful family history may identify current or past speech or language problems in up to 30% of first-degree relatives of proband children. This compares with a base rate of only 3% in the general population. The degree of familial clustering depends on the type of language disorder. For example, there is no increased prevalence of speech, language, or learning disorders in families of children who have isolated expressive disorders compared with a three to four times higher prevalence among children who have mixed expressive and receptive disorders. Further evidence of a genetic basis for some of these disorders emerges from studies of

twins in which monozygotic pairs show concordance rates of 70% to 90%, with the concordance rate dropping to 30% to 60% in dizygotic pairs.

Social and environmental factors also play a role in influencing young children's language development. Children who are economically disadvantaged and later-born children from large families in which siblings are closely spaced are more likely to exhibit delayed language development. However, handedness and pre- and perinatal factors do not correlate with language development. Frequent episodes of otitis media may influence early articulation, but well-controlled studies have not shown it to be a cause of language delays, particularly for delays persisting beyond age 4 years.

NEUROANATOMIC STUDIES

Abnormalities of language development may be associated with identi-

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fied brain lesions, but because language function has not been identified with specific sites in the brain, specific functional deficits cannot be predicted according to the site of injury. In children, bilateral brain lesions are more likely to disrupt language development than are unilateral or focal injuries. Furthermore, in contrast to traditional concepts of regional anatomic specialization, recent findings from studies of adults who had acquired aphasia suggest that complex functions such as language and memory are the result of vast interconnected neural networks that are synchronized for specific activities across the cerebral cortex and subcortical regions. Although fairly consistent anomalies of brain development have been found in the limited number of autopsy studies of adults who had lifelong disorders of language and learning, further research is needed

to confirm that these abnormalities play a role in the disorders and that they represent physical manifestations of a genetically transmitted trait in familial cases of language and learning disorders.

NEUROPHYSIOLOGIC STUDIES AND LANDAU-KLEFFNER SYNDROME

Electroencephalographic (EEG) abnormalities in the absence of clinical seizures have been identified in prolonged sleep studies in nearly two thirds of children who have normal nonverbal IQ and language comprehension disorders and in 12% to 20% of those who have disorders of expressive language. Paroxysmal electrical abnormalities are more likely to be markers of cerebral dysfunction than causes of language disorder because treatment with anti-epileptic agents has been largely unsuccessful in improving the language abilities of affected children. One exception is Landau-Kleffner syndrome (LKS), a rare condition

(prevalence estimate, 0.2% all childhood epilepsy) in which children who have normally established skills gradually or suddenly develop a progressive aphasia, most commonly between 3 and 7 years of age. Initially, there is loss of verbal understanding, followed by deterioration in expressive communication ability. Affected children may become completely mute and fail to respond even to nonverbal sounds, such as the ring of the telephone, a knock on the door, or a dog barking.

LKS is associated with an EEG pattern that shows electrical status epilepticus during slow-wave sleep. Such abnormalities may be unilateral or bilateral and may fluctuate from the right to the left hemisphere, although characteristically they are located over the temporal and parietal areas. When seizures occur, they commonly are associated with eye blinking or brief ocular

deviation, head dropping, and minor automatisms with occasional secondary generalization. However, 20% to 30% of children who have LKS do not exhibit behavioral seizures.

Functional imaging in several cases of LKS has shown decreased perfusion in the posterior temporal cortical regions. LKS is a prototype for considering a connection between EEG abnormalities and language disorders in the absence of clinical epilepsy.

Autistic Spectrum Disorders

In 1943, Kanner described 11 children whom he believed suffered from "autistic disturbances of affective contact." They resembled one another in their inability to "relate themselves in the ordinary way to people and situations from the beginning of life" and in their preference to be alone. Kanner noted that these children exhibited absent or delayed language development, inability to use language to convey meaning to others, and delayed echolalia, although their rote memory was excellent. Their spontaneous activities were limited in variety, and their behavior was "governed by an anxiously obsessive desire for the maintenance of sameness that nobody but the child himself may disrupt on rare occasions." All of the children appeared to be physically healthy, although nearly 50% had relatively large heads, and several were somewhat clumsy in gait and gross motor performance. These keen observations aptly describe the principal features of the syndrome now known as "classical" autism. Symptoms of autism begin before 3 years of age, and affected individuals have qualitative impairments in the areas of social interaction and communication and demonstrate restricted, repetitive, and stereotyped patterns of behavior, interests, and activities.

In the intervening 46 years since this initial report, a number of similar disorders, referred to broadly as the pervasive developmental disorders (PDDs), have been described. These encompass a spectrum of related autistic symptoms that vary in severity, age of onset, and spe-

TABLE 2. Autistic Spectrum Disorders

<p>Autistic Disorder (classic autism)</p> <ul style="list-style-type: none"> • Severe qualitative deficits in social interaction and communication skills • Restricted and stereotyped patterns of behavior, interests, and activities • Onset before the age of 3 years
<p>Pervasive Developmental Disorder-Not Otherwise Specified (PDD-NOS)</p> <ul style="list-style-type: none"> • Severe and pervasive deficits in social and communication skills or • Restricted and stereotyped patterns of behavior, interests, and activities, but • Symptoms do not meet the “threshold” for autistic disorder
<p>Asperger Syndrome</p> <ul style="list-style-type: none"> • Deficits in sociability and a narrow range of interests • No speech or language delays • Normal intellectual abilities • Motor clumsiness
<p>Childhood Disintegrative Disorder</p> <ul style="list-style-type: none"> • Previously completely normal children who undergo massive regression between 2 and 10 years of age • Mental retardation

cific clinical features (Table 2). Childhood disintegrative disorder (CDD) shares many behavioral features with autism, but it always is characterized by severe developmental regression, including loss of bowel and bladder continence, and has a distinctly later onset than “classic” autism. The DSM-IV category of “subthreshold” autism, pervasive developmental disorder-not otherwise specified (PDD-NOS), refers to a heterogeneous disorder in which there are deficits in peer and family relationships and usual sensitivities to the environment, but the degree of social impairment is less than that in strictly defined autism.

Individuals who have Asperger syndrome share many of the social deficits found in autism, but their overall language and cognitive development is not impaired. This disorder is not recognized in most cases until after 3 years of age, and many children actually appear precocious in their ability to talk. Also in contrast to autism, children who have Asperger syndrome often are interested in making friends but have great difficulty because they lack sensitivity to others’ feelings, engage in one-sided conversations, and tend to have very eccentric interests. Their preoccupation with unusual topics, which may change over time, often becomes intense and dominates their social interactions.

PREVALENCE

Autistic spectrum disorders are relatively uncommon, affecting 0.05% to 0.2% of the population; when broader definitions of the milder forms of autism (PDD-NOS) are included, the estimates have been as high as 0.5%. Asperger syndrome is much less common than autism, with estimates of its prevalence varying from 0.01% to 0.03%.

CLINICAL FEATURES

A primary, unexplained inability to relate socially to others is the hallmark of autistic spectrum disorders. Even when cognitive and language deficits are taken into account, autistic individuals lack interest in reciprocal social relationships.

Approximately 75% to 80% of individuals who have “classic” autism also are mentally retarded. However, in contrast to most people who have mental retardation, those who have autism usually have a wide scatter in cognitive abilities, with language skills more impaired than visual-motor skills. Some autistic individuals have highly specialized, but isolated, “savant” skills, such as calendar calculations and hyperlexia (the precocious ability to recognize written words beyond expectation based on general intellectual ability). Language abilities in autism may range from absent (verbal auditory agnosia) to intact but odd. Children who have autism typi-

cally are limited in their ability to develop functional, symbolic, or pretend play, although they usually demonstrate adequate sensorimotor play. An impaired ability to imitate the actions of others (dyspraxia) also is characteristic. In approximately one third of children whose parents may believe that development is progressing normally, unexplained autistic regression in cognitive, language, and social development occurs between the ages of 15 and 24 months.

ETIOLOGY

The specific cause of autism and other PDDs cannot be determined in the majority of cases, even with extensive clinical laboratory evaluations. However, a large number of neuroanatomic and imaging studies of the central nervous system in children who have autism and PDDs have documented developmental anomalies in the cerebellum and the limbic networks of the temporal lobe, particularly the cingulate gyrus, amygdala, dentate gyrus, and hippocampus. Once thought to be restricted to the control of motor functions, the cerebellum now is known to play a large role in sensory processing, including attention to novelty, modulation of pain and fear, and social and emotional responses. In some individuals who have autism, the posterior cerebellum is reduced in size, and anatomic studies have shown a loss in the number of Purkinje cells, suggesting prenatal dysgenesis of brain structures.

In approximately 10% to 30% of cases, identifiable disorders may result in autism or autistic-like syndromes, including untreated phenylketonuria, congenital rubella infection, tuberous sclerosis, hydrocephalus, fragile X syndrome, Angelman syndrome, hypomelanosis of Ito, incontinentia pigmenti, and destruction of the temporal lobe due to congenital or acquired herpes simplex encephalitis or tumor. In addition, EEG abnormalities, in the absence of clinical seizures, are common, usually involving unilateral or bilateral centrotemporal paroxysmal activity. Clinical seizures of all types develop in up to 25% of

individuals who have autism, and the incidence increases during adolescence and with the severity of mental retardation.

A genetic basis for autism is suggested by a markedly increased risk of recurrence in siblings and monozygotic twins compared with the general population and a higher-than-normal incidence of other cognitive deficits, learning problems, and personal-social deficits in first-degree relatives.

Atypical Behaviors Seen in Normally Developing Children

Some of the most troubling behaviors reported by parents and teachers of children who have developmental difficulty actually may be variants of "normal" and should be considered as neurobehavioral "soft signs" (ie, of no specific pathologic or

routines and "rituals" of otherwise normally developing children usually help the child make transitions from one activity or situation to another during the day, are less pervasive and intense, and fade over time.

Although frequently reported in children who have neurologic and developmental disorders, many otherwise normal children have great difficulty tolerating a variety of sensory stimulation. These so-called "sensory integration" disorders involve abnormalities of sensory reactivity (sound, sight, touch, taste, smell, and movement) and regulation. Sensory integration disorders are descriptive symptoms and do not suggest a particular etiology or diagnosis. During infancy, children who have extreme difficulty in regulating emotional states may appear distant and unresponsive to social interactions with caretakers.

Children who have major central nervous system anomalies . . . usually are identified in the first year of life . . . Mild-to-moderate retardation is identified later when delays in language and play behaviors become more noticeable.

diagnostic significance). For example, hand-flapping, head-banging, head-rolling, body-rocking, and toe-walking are seen in both developmentally delayed and normal children younger than 3 years of age. These stereotyped, rhythmic motor movement patterns may arise in response to emotional excitation involving happiness, fear, anxiety, frustration, overstimulation, or boredom. They are intentional movement patterns that appear to provide the child with both pleasure and relief from tension. In some cases, familial predisposition to rhythmic patterns can be identified, an observation noted by Charles Darwin and others. Repetitive and compulsive behaviors are also common among preschool-age children. In a recent survey of nearly 1,500 healthy infants and children, approximately 60% to 70% of parents reported the presence of compulsive behaviors and strong adherence to routines in their 1- to 5-year-old children. In contrast to the behavior of autistic children, the

Comprehensive Evaluation

A comprehensive evaluation of a child who has atypical developmental patterns should include assessment of physical, cognitive, and language development; social interactions; and play skills. Because many children find it difficult to adapt to testing situations, detailed descriptions of the child's behaviors and abilities gleaned from standardized questionnaires and structured interviews should be combined with results of formal assessment tools and careful observation by a multidisciplinary team of professionals. In the Child Development Center at the Children's Hospital of Wisconsin, a core team consists of a developmental pediatrician, psychologist, speech and language pathologist, and social worker. Laboratory studies and consultations from additional medical subspecialists and members of health-related disciplines are included as necessary. Once the evaluations are completed, the team meets with the family to review its

findings and make specific recommendations for treatment and follow-up.

HISTORY

Review of the pregnancy and birth history should include risk factors for poor fetal development. Poor fetal growth patterns may reflect effects of chromosomal abnormalities, major structural malformations, intrauterine infection, or prenatal exposure to toxic substances. However, many neurobiologic dysfunctions that result in atypical developmental patterns are not associated with overt pregnancy or birth complications.

Family history may be helpful in the differential diagnosis because, as noted previously, children who have developmental language disorders are much more likely to have first-degree relatives who exhibit speech, language, and learning disorders than are those who have autism or isolated moderate-to-severe mental retardation. Affective disorders, including bipolar depression and obsessive-compulsive disorder, are common among families of children who have autistic spectrum disorders. Moderate-to-severe mental retardation, while more common than autistic disorder, usually is an isolated phenomenon and has a very low recurrence risk in extended family members.

The early developmental history may suggest the presence of biologically based deficits of sensory awareness and regulation. Parents may recall that their infant was excessively irritable or very placid. Although motor milestones may have progressed normally, social interactive games, such as "waving bye-bye," "so-big," or "pat-a-cake," may have been delayed or never achieved. Delayed acquisition and loss or regression of skills should be distinguished carefully.

PHYSICAL EXAMINATION

Abnormalities of growth, motor or sensory problems, and physical anomalies or dysmorphic features may point to a specific etiology for the developmental delay. Inspection of the skin surfaces for signs of neu-

rocuteaneous disorders and a Wood lamp examination for the depigmented macules of tuberous sclerosis can be conducted easily in an office, even with an uncooperative child. Micro- or macrocephaly can be associated with malformation of the brain, and somatic growth deficiency suggests the possibility of an endocrine or metabolic disorder or prenatal alcohol exposure. Children who have atypical developmental patterns frequently have disorders of motor coordination or hypotonia. Sensory problems may include vision or hearing impairments. Many children who have severe visual impairments exhibit delays in motor, communication, and social development. Repetitive and self-stimulatory behaviors (so-called "blindisms") are common and may be confused with autistic rituals. Hearing difficulties may result from peripheral sensorineural or conductive loss or from central auditory processing problems and can interfere with language and social development.

PLAY AND SOCIAL SKILL ASSESSMENT

In a primary care pediatric setting, standardized measures, such as the Denver-II Developmental Screening Test, may be more helpful in providing a framework for observation and interaction than for their specific value as tests of development. Observing play skills in the office is an informative, informal method of screening for cognitive level. It is useful to know that infants and toddlers through about 15 months of mental age typically engage in sensorimotor and manipulative play (eg, placing objects in containers and dumping them out) as well as cause-and-effect play (eg, activating toys that make noises, light up, and open doors). At about 16 to 18 months, children start showing forms of domestic mimicry and early pretend play. Mimicry includes pretending to drink from a cup, pretending to activate a household appliance such as a vacuum cleaner, and placing a brush or comb to the head. More advanced symbolic and representational play, which emerges at about 24 months of age, includes

pretending to feed dolls or human figures and combining two toys in pretend play, such as placing a figure in a toy car or a toy bed.

The child's social skills can be assessed by direct observation and parental report. By 4 to 5 months of age, infants are attracted to people and respond to smiles or being picked up. They imitate facial expressions and vocal intonation early in life. Toward the end of the first year, infants look to adults to draw attention to what they are doing, and by 15 to 18 months of age, they bring toys to adults to "share" their experiences. They delight in surprise and enjoy chase-

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and-catch games, tickle games, or vigorous wrestling with family members. Although some may resist their parent's overtures during the day, night-time routines typically include close, affectionate interactions, such as initiating contact when tired or lying down with the parents in the bed.

Table 3 shows an interview outline we use to assess the various domains of behavior associated with atypical development.

SPECIFIC EVALUATION INSTRUMENTS FOR AUTISM

A number of autism rating scales are available to assist in diagnosis by supplementing clinical evaluation with quantitative measures of specific symptoms and their severity. These scales provide normative data and classify the child along a spectrum from "not autistic" to "classically autistic." Some of the more common measures include the Rimland E2, the Autism Diagnostic Inventory, the Childhood Autism Rating Scale, and the Wing Schedule of Handicaps, Behavior and Skills. These scales are complex, and their general use is limited by the amount of training required to

interpret them and the time necessary for administration. However, rating scales provide a method of charting changes in a child's clinical course over time. The Checklist for Autism in Toddlers is a brief screening tool that appears to be very effective in identifying children at high risk for autism during routine 18-month-old health supervision visits.

COGNITIVE ASSESSMENT

When assessing children who have atypical developmental patterns, particularly communication deficits, it is important to differentiate between

verbally based cognitive abilities and nonverbal abilities. Psychologists employ special techniques and measures. Traditional intelligence tests are language-dependent, with both the content of the assessment instrument and the method of assessment requiring the child to comprehend and respond to spoken directions and questions. For example, the content of an instrument may include an expressive or receptive vocabulary recognition task. The format also can be language-dependent, such as giving verbal directions to a child, even if the child's response is nonverbal. However, it is important to assess children who have communication impairments with standardized instruments to compare their performance with that of typically developing children of their own age and determine the status of their intellectual functioning.

NONVERBAL COGNITIVE ASSESSMENT

Certain intellectual functions depend minimally on verbal understanding (what the examiner says to the child) or verbal expression (what the child says to the examiner); these can be assessed reliably in children who have atypical development

TABLE 3. Interview Outline for Atypical Behaviors

<p>Instructions: Circle affirmative answers.</p>
<p>Socialization</p> <ul style="list-style-type: none"> • Uses a transitional object: blanket doll pacifier thumb bottle • Cuddles, hugs, kisses: initiates accepts from parents • Kissing games, allows parents to blow on: cheek belly • Enjoys wrestling and tickling games; tries to keep game going • Calms down after wrestling game terminated (within 5 min) • Seeks comfort when hurt, shows small sores or scrapes to parent, asks for a bandage • Hyposensitive to pain? • Plays chase-and-catch with another person • Plays imitation games: peek-a-boo so-big wave bye-bye pat-a-cake Itsy-Bitsy-Spider Ring-Around-the-Rosey
<p>Sensation</p> <ul style="list-style-type: none"> • Likes bath play • Likes sand play • Strongly protects or panics with hair washing, hair cutting, nail clipping • Resists textures: in mouth on hands • Reacts to feel of clothing on skin (eg, shirt tags, coarse denim, elastic cuffs) • Hyperacusis (vacuum cleaner, hair dryer, hand driers in public restrooms, other appliances, fireworks, crowd sounds) • Resistant to movement: tipping sideways holding upside down spinning
<p>Communication</p> <ul style="list-style-type: none"> • Echolalia: delayed immediate • Loss of speech skills: age of loss greater than 10 words acquired before loss? • Points gestures • Pushes parents to object Places parent's hand on object • Draws parent's attention to other objects (joint attention) • Uses formal sign language Uses a picture communication system • Babbles Uses inflected jargon • Sings parts of songs • Dances to music • Recites: ABCs counts • Reading: pages through books allows parent to read (what level of material?) • Television: what types of programs? length of attention shows emotional responses to movies (eg, laughs, becomes apprehensive)
<p>Play</p> <ul style="list-style-type: none"> • Brings objects to mouth • Sniffs; tastes objects • Manipulates objects • Uses representational play: domestic routines vehicles • Uses pretend play: dolls human figures pretends to be an animal

through the use of nonverbal tests of intelligence. Useful nonverbal tests include the Merrill-Palmer Scale of Mental Tests, the Preschool Performance Scale, the Leiter International Performance Scale-Revised, the Test of Nonverbal Intelligence-Third Edition, the Columbia Mental Maturity Scale, and the Raven's matrices and Raven's Colored Progressive matrices. Several other tests have specialized nonverbal sections that are helpful in assessment, including the Bayley Scales of Infant Development-Mental Scale, the Wechsler Intelli-

gence Scale for Children-Third Edition, the Kaufman Assessment Battery for Children, and the Differential Ability Scales.

Speed of information processing is an excellent index of intellectual ability and can be evaluated by a variety of methods, such as a formboard task. Short-term memory traditionally has been measured by number recall and sentence repetition. However, nonverbal methods can be used to assess this function, including tasks of visual memory for sequential information, such as a

hand gestures task, and spatial memory for position of pictures on a page or shapes on a formboard. Inductive reasoning can be measured by classification or picture similarities tasks as well as by use of interlocking puzzles. A matrix analogy is an excellent measure of nonverbal reasoning. In this task, the child is required to complete patterns and sequences or match picture items that are analogous to a model (eg, a carrot matches with a rabbit as a bone matches with a dog). Matrix analogy items can be found on

many child assessment instruments. Copying two- and three-dimensional models is a perceptual-motor skill that correlates well with general measures of nonverbal intellectual functioning. Typical tasks include copying constructions made from wooden blocks, copying two-colored patterns with blocks or forms, and copying line drawings. Both inductive and deductive reasoning can be assessed with various puzzle tasks, such as use of interlocking puzzles to measure inductive reasoning and inset puzzles, in which pieces are placed in defined spaces in a picture to measure deductive reasoning.

Nonverbal measures also provide examiners with opportunities to assess the quality of the child's self-control and monitoring capability. This is obtained by observing behaviors such as self-correction, in which the child demonstrates the ability to analyze and check a work product and make appropriate corrections.

Some nonverbal tests combine a variety of tasks. It is important not to rely on a single IQ score for the overall test as an index of the child's nonverbal intellectual functioning. Rather, examination of variability of performance across different types of tasks can offer important diagnostic information for the differential diagnosis. For example, autistic children typically tend to do well on nonimitative, visual-perceptual tasks, such as placing shapes in a formboard. In contrast, they are deficient in performing imitative tasks, such as block constructions, limb movements, and paper folding.

In choosing specific nonverbal tests, examiners should be alert to particular nuances of tasks and administration. Some nonverbal tests involve extensive manipulation of materials, which requires hand dexterity. This is advantageous in testing restless children who have short attention spans, but presents liabilities in testing children who have coordination disorders (eg, construction apraxia). Other tests minimize manual manipulation by requiring simple pointing or placing responses, and one test even is designed to accept eye-gaze

responses if arm or hand movement is impossible (Leiter-R).

SPEECH AND LANGUAGE ASSESSMENT

Comprehensive speech and language evaluation should include assessment of receptive and expressive language, phonologic processes (speech sounds), oral motor functioning (speech and feeding), and hearing. In addition, examiners should note the child's social and pragmatic use of language. This evaluation can be made with infants and young children. Many standardized assessment scales measure children's expressive and receptive language functions, including the Rossetti Infant-Toddler Language Scale, the Preschool Language Scale, and the Test of Language Development.

Many children who have language disorders still do not produce or pronounce words clearly beyond 3 years of age. They may rely on gestures, facial expressions, or vocal inflections to communicate meaning because they cannot express themselves with words. In this situation, it is important to attend to nonverbal communication patterns. Does the child show toys to the parent (joint attention)? Does the child point to objects either to call attention to them or to request the object? Does the child imitate gestures prompted by an adult (waving bye-bye)? If the child's speech is unintelligible, does the child speak in jargon with inflected patterns that sound like speech? These nonverbal and paraverbal attempts to communicate can help differentiate types of developmental problems.

As words emerge, the child may not use complex phrases and sentences and may speak in choppy or telegraphic utterances. Repetition of questions or statements (immediate echolalia) or memorization of dialogs or scripts from videotapes (delayed echolalia) usually indicates that the child hears but does not understand what has been said.

Although echolalia often is identified as a key feature of autism, it is not unique to this disorder. Rather, it reflects a disorder of language processing, often specific to syntac-

tic (grammar) understanding. Children who have semantic and pragmatic difficulties may begin to speak on schedule and even may seem precocious, using clear speech and good grammar. Although their comprehension of concrete language is intact, their understanding of abstract language concepts is poor. As a result, their verbal responses are inappropriate or bizarre. Such children may give tangential or perseverative responses to questions, cannot maintain a topic of conversation (so-called "cocktail party speech"), or use overly formal or odd speech patterns.

ADDITIONAL CONSULTATIONS AND LABORATORY TESTING

Hearing and vision examinations should be undertaken routinely in all children who have developmental disability. Findings on the history and clinical examination should determine whether additional consultations and laboratory examinations are necessary. There are no specific laboratory examinations to diagnose autism or language disorders, but all children whose mental retardation is unexplained should have chromosome karyotype analysis with fragile X testing. A magnetic resonance imaging scan of the brain may be indicated if there is a history of perinatal brain injury, encephalitis, or developmental regression. Many specific genetic and metabolic disorders can be screened by using specific gene probes or biochemical tests. Dysmorphic features should prompt a referral to a geneticist for further evaluation. If a seizure disorder is suggested by the history, an EEG is indicated. A prolonged sleep EEG should be considered if LKS is suspected. Further consultation with a pediatric neurologist may be necessary. Occupational and physical therapy consultations may be helpful when sensory-integration difficulties are suspected and when fine and gross motor coordination difficulties are present.

Differential Diagnosis

In the general population, developmental language impairment is approximately 42 times more com-

mon than moderate-to-severe mental retardation and from 25 to approximately 250 times more common than autistic spectrum disorders (PDD, autism, or both). Similarly, uncomplicated mental retardation is more common than autism (Table 4).

It is unlikely that the results of any one evaluation will establish a diagnosis clearly. Language disorders and autism share the characteristics of linguistic impairment, although several subtypes (verbal auditory agnosia and semantic-pragmatic disorder) are found with greater frequency among autistic children. In addition, both language disorders and autism may occur in the context of normal intelligence or mental retardation. Stereotypic behaviors are nonspecific and may be seen in normal children as well as those who have mental retardation, language disorders, or autism. A symptom pattern profile that considers a variety of factors, such as shown in Table 5, may help in the differential diagnosis.

Recent research suggests that specific aspects of a child's social behavior may provide useful clues to distinguish among these disorders. For example, Lord, Rutter, and Le Couteur noted that children who had mental retardation and language disorders displayed more spontaneous imitation, imaginative play, and imitative social play than children who had autism. Also, separation anxiety was seen less often in autistic children. Wetherby, Prizant, and Hutchinson reported that joint attention, symbolic play, and social/affective signaling allowed examiners to discriminate reliably among children who had autism and children who had developmental language disorders. A study by Robertson et al also showed that three clusters of social behaviors—joint attention, affective reciprocity, and pretend

play—represented critical areas of deficiency in autistic children.

Although the idea of an "autistic spectrum" is conceptually simple, milder forms of the disorder are hard to define clearly. As currently described in DSM-IV, the entity of PDD-NOS encompasses many, but not all, features of autism. Difficulties arise in attempting to separate milder autism from language disorders or mental retardation. The changing development of young children introduces another variable that makes early distinction among these entities difficult. In a recent study by Fein et al, approximately 25% of children who were thought to have PDD as preschoolers were reclassified as not having PDD when older, primarily because of improvements in language abilities, and a smaller number of children were reclassified in the opposite direction. Thus, for milder forms of autism (eg, PDD-NOS), the lack of sharp boundaries and overlap of symptoms with specific language impairment should raise caution in diagnosing young children.

In some situations, a diagnostic approach that focuses on the process as well as the outcome of the child's development may be preferable to one that results in a categorical classification. Atypical behaviors should be interpreted both for information provided about the child's developmental difficulty and for what the behaviors reflect about attempts to compensate. For example, children who rely too heavily on memorized scripts or stylized phrases may have difficulty formulating their own ideas into words and sentences (syntactic construction). However, such formulaic speech patterns also provide a scaffold-like framework into which the child may insert functional phrases to accomplish effective communication with others until

more "natural" language skills emerge. As noted by Tanguay et al, comprehending the meaning of a child's disabilities sometimes may be more important than making a firm diagnosis.

Treatment

Regardless of the specific nature of a preschool-age child's developmental difficulty, early intervention services such as Birth to Three and Early Childhood Programs are universally available in the United States through public school systems. These programs provide a variety of individual and group educational and therapeutic services, including special education, speech, and occupational and physical therapy up to three to four times per week. For many children, these services can be supplemented with private therapies.

Occupational therapists trained in "sensory integration" techniques can assist in the management of children who have aversive behaviors. Children who have oral-motor deficits, especially speech apraxia, require intensive speech and language therapy. Appropriate supports for vision- and hearing-impaired children are also available through public channels.

Intensive speech and language therapy by a qualified and experienced professional should be a cornerstone of treatment for children who have any type of communication disorder. A number of approaches seem to be effective. Use of signing by nonverbal children has proven to be an effective bridge to spoken language, and there is no evidence that it slows or prevents this transition. A newer system involves the use of icons or Picture Exchange Communication System. Both methods allow the child to use cognitive and visual processing abilities to compensate for expressive language difficulties.

Parents should work closely with teachers and therapists in the treatment, reinforcing newly emerging skills. Several excellent books are available to guide parents in stimulating their children's language development. There is an emerging role for medications in managing

TABLE 4. Population Prevalence Rates in Preschool Children

Language Disorders	120/1,000
PDD-NOS	5/1,000
Moderate-to-severe Mental Retardation	3/1,000
Autism	0.5 to 2/1,000

TABLE 5. Differential Diagnosis of Atypical Patterns of Development

	MENTAL RETAR- DATION	DEVELOPMENTAL LANGUAGE DISORDER	SPECIFIC LANGUAGE IMPAIRMENT	AUTISM/PERVASIVE DEVELOPMENTAL DISORDER	ASPERGER SYNDROME
Cognitive Ability	Delayed	Normal/delayed	Normal	Normal/delayed	Normal
Language Ability	Delayed	Disordered	Disordered	Disordered	Normal
Social Ability	Normal	Normal	Normal	Abnormal	Abnormal
Family History	Negative	Speech/language	Speech/language	Affective disorder	Social deficits

certain symptoms common to atypically developing children. Although there is no specific pharmacotherapy for mental retardation, language disorders, or autism, several of the newer selective serotonin reuptake inhibitors (especially fluoxetine, fluvoxamine, and paroxetine) and atypical antipsychotics (risperidone) have been shown to improve fluctuations of mood, irritability, and obsessive-compulsive symptoms, with fewer long-term adverse effects in autistic disorder and PDD than older medications (eg, haloperidol, thioridazine, clomipramine). DeLong and associates noted significant improvement in the language functioning of children who had PDD and received fluoxetine for treatment of behavioral symptoms. These effects were noted more often among children who had a family history of affective disorders. In many cases of LKS, marked improvement in language ability follows treatment with steroids, anti-epileptic drugs (such as valproic acid and carbamazepine), intravenous immune globulin, or even surgery.

Nonstandard Treatments

Physicians treating children who have these conditions will become aware of a vast array of controversial treatments promoted through the Internet and public media. Although some eventually may be shown to be efficacious, most have not been tested in a sound scientific manner. National advocacy groups, such as the Autism Society of America, the Unicorn Foundation (for speech and language disorders), the Association for Retarded Citizens, and other government and university-based organizations and Web sites, are

good resources for parents and professionals who desire to learn more about these newer treatments.

Prognosis

As with most developmental disorders, intellectual level is a major determinant of long-term outcome. In addition, the prognosis for young children who have atypical developmental patterns varies widely according to the nature of the underlying disorder.

In mental retardation, functional outcome depends on the overall cognitive level and degree of social adaptive impairment. For example, many mildly retarded individuals can live productive lives, be gainfully employed, and raise families; moderately and severely retarded individuals generally require some degree of supervision and cannot live independently. The language development of mentally retarded people may improve slowly with age, even for those who exhibit minimal verbal skills as young children.

The prognosis for the vast majority of young children who have speech disorders or problems with expressive language is excellent. Most will have normal language skills by the time they enter primary school. However, those whose speech and language problems persist to age 5 years, despite intervention therapies, are at high risk (70% to 80%) of continuing communication difficulty and reading disorder into their adult years. Autism has a very poor long-term prognosis. Although many autistic individuals who have normal IQs may show an increase in language ability and decrease in autistic behaviors over time, most continue to have signifi-

cant autistic social deficits. In one recent follow-up study of adolescents and adults diagnosed with autism during childhood, more than 50% of the adults lived in residential placements, and only 11% were employed, all in menial jobs. The prognosis for milder forms of autism (PDD-NOS) is believed to be considerably better, but good long-term data are lacking. The outcome of individuals who have CDD is extremely poor.

The long-term prognosis for resolution of the EEG abnormality seen in children diagnosed with LKS is good, but language recovery is unpredictable. In this condition, younger children usually have the poorest prognosis. This is in stark contrast to most other acquired lesions that affect children, in which younger children often do better than older ones.

Summary

Despite considerable overlap of clinical features in preschool children who have atypical developmental patterns, it often is possible to determine the nature of the underlying difficulties with careful assessment. Multidisciplinary evaluation is essential and can help to identify the child's strengths and weaknesses. Parents, teachers, and therapists can support the child's continued development through a variety of effective educational, behavioral, and therapeutic interventions. Newer pharmacotherapeutic agents play an increasing role in ameliorating some of the more troublesome features of these disorders. Because prognoses vary considerably, it is important to be certain of the diagnosis. In many instances, longitudinal follow-up and

a trial of therapeutic interventions may be required before an accurate diagnosis can be provided.

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PIR QUIZ

Quiz also available online at www.pedsinreview.org.

1. The hallmark feature of autistic spectrum disorders is:
 - A. Delayed expressive language.
 - B. Echolalia
 - C. Functional intelligence quotient in the superior range.
 - D. Inability to relate socially.
 - E. Stereotypy.

2. A true statement about the etiology of mental retardation is that:
 - A. It is identifiable in 50% of mentally retarded children overall.
 - B. It is identified most easily among children who have mild-to-moderate impairment.
 - C. Perinatal causes account for about 25% of all cases.
 - D. Prenatal causes are most common.
 - E. Sex-linked causes have not yet been identified.

3. Although a 2½-year-old girl usually points when she wants something, she has a 50-word vocabulary to identify objects (car, baby) or desires (go bye-bye). She does not otherwise combine words. About 50% of her utterances are gibberish. She points to her face but cannot identify other body parts. She walks up and down stairs and kicks a ball forward, but she cannot throw a ball overhand. She uses a fork and spoon. She pulls off her socks to play with her feet, but she does not dress herself. When presented with blocks, she stacks two and then loses interest. She has two favorite dolls that she cuddles constantly. New situations or frustrations are likely to result in tantrums. In child care, the other children treat her like a "mascot." The most likely diagnosis for this child is:
 - A. Asperger syndrome.
 - B. Autism.
 - C. Mental retardation.
 - D. Pervasive developmental disorder.
 - E. Specific language impairment.