Identifying Patterns of Developmental Delays Can Help Diagnose Neurodevelopmental Disorders

Raymond C. Tervo, MD, MSc

Introduction

Child development alludes to the enhancement and specialization of a child's functioning with the achievement of increasingly intricate abilities in the different functional domains. Failure to achieve developmental milestones reflects the complex interactions between the child and his environment.[1]

While most health professionals caring for children are committed to early identification of developmental delays and diagnosis, less than half use standardized tools, preferring instead to use a list of developmental milestones or to be prompted by parent concerns.[2-6] Yet clinical judgment misses more than 70% of children with mental retardation, language impairments, learning disabilities, and other developmental disabilities, and checklists on pediatric encounter forms are neither validated nor standardized.[7]

Neurodevelopmental Evaluations

The developmental disabilities in infancy and childhood have typical profiles.[1,8,9] Communication disorders, mental retardation, and cerebral palsy make up the "big three" of early childhood developmental disabilities.[10] So while there may be a variety of delays in development noted in a busy clinical practice, three patterns of delay are commonly seen: the speech and language variant, global delay, and the motor variant (Figures 1–3).

Developmental Domains

Although Gessell initially described six developmental domains,[1] for this article, we will discuss delay in the context of the following four main developmental streams:[1]

- Speech and language
- Motor
- Fine motor
- Personal and social

Global Developmental Delay

Children with a diagnosis of developmental cognitive disability or mental retardation often present with mixed or global developmental delays.[11] Two to three children of every 100 will have a mental handicap and those with IQs of 55 or less, those with more severe delays, will generally have an organic reason for their delay.[9,11]

Common reasons for global developmental delay include chromosomal anomalies[9] such as Down syndrome—the most common identifiable cause of mental retardation. Global delays also are common in children with fetal alcohol syndrome—the most common preventable reason for mental retardation—and in children with fragile X syndrome, the most common known cause of inherited mental retardation.

Speech and Language Delay

Disorders of speech and language development are the most prevalent among children with developmental disabilities.[10] Eleven percent of toddlers have a speech and language impairment. Clinical diagnoses to con-

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Department of Pediatrics, University of Minnesota, Minneapolis; and Pediatric Section, Gillette Children's Specialty Healthcare, St. Paul, MN.

Reprint requests and correspondence to: Raymond C. Tervo, MD, MSc, Gillette Children's Specialty Healthcare, 200 East University Avenue, St. Paul, MN 55101.

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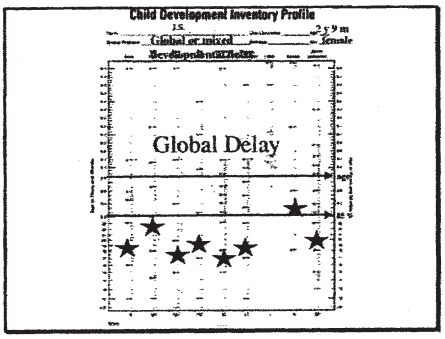

Figure 1. Global Developmental Delay. The profile on the Child Development Inventory (CDI) shows significant delays in all streams of development. A developmental cognitive disability ought to be considered in this 2-year 9-month-old girl.

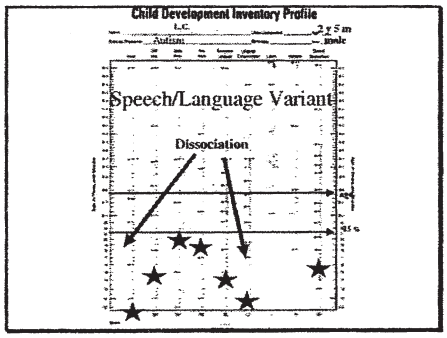

Figure 2. Speech/Language Variant. Note that this child's social, self-help, expressive language, and language comprehension skills lag behind his gross and fine motor skills. Expressive language is greater than language comprehension because of rote memory or echolalia. On more detailed evaluation, this 2-year 5-month-old boy was determined to have autism.

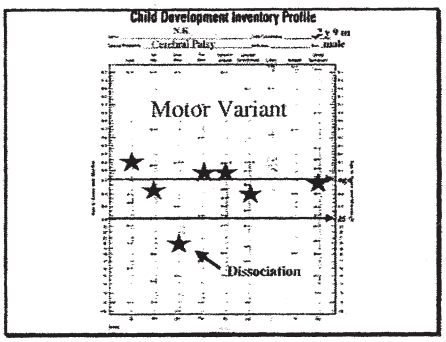

Figure 3. Motor Variant. Note that gross motor skills are substantially below expected levels. All other streams of development are at or close to age-appropriate expectations. This 2¼-year-old boy has cerebral palsy and spastic diplegia.

sider when a child presents with delayed speech and language include hearing loss, mental retardation, autism, dysarthria, a specific learning disability, and developmental language disorders (Table 1). A degeneration of previously acquired language skill is worrisome and requires more assessments.

Hearing Loss

A leading cause of acquired hearing impairment is otitis media with effusion (OME).[11-13] Approximately half of all preschool children have intermittent fluctuating hearing loss from serous otitis media.[13]

Expressive Language Delay

Expressive language delay is the most common developmental presentation in a primary-care

Table 1

SPEECH AND LANGUAGE DELAY

- Hearing loss
- Mental retardation
- Autism
- Dysarthria
- Specific learning disability
- Developmental language disorder
- Landau-Kleffner (LKS)
- Poor language environment

setting.[11] The social and educational development of children with delayed speech and language may be significantly disrupted—even in mild delays—so early identification and intervention is imperative.

Developmental Cognitive Disability

Delayed language is the most common presentation of mental retardation between 2 and 4 years, so children with delayed speech and language should be evaluated for cognitive disabilities.[14,15] The pattern of language acquisition in mental retardation is similar to typical development but at a slower rate. Since there is a close association among social and affective abilities, and cognitive, sensory, and language development, children who are unable to communicate effectively will have problems interacting verbally with their peers. Because social and pragmatic deficits are core characteristics of autism, it is important to look for a dissociation among language, social adaptive skills, and motor behavior (Figure 2).

Autism

Autism is a relatively common disorder, occurring approximately once in 500 children. It is one of the most complex neurodevelopmental disorders. Children with autism have significant impairment in communication; however, they have more difficulties with the social aspects of language than with grammar or speech production.[16] Children whose primary difficulty involves receptive language are more likely to be considered children with a developmental cognitive disability or with autism spectrum disorder. Deviancy in language development may suggest the diagnosis of autism.

There may be the impression that expressive language skills are better than they are; this is described as "cocktail chatter" and is the result of echolalia and rote memory skills rather than useful communication. True expressive skills cannot exceed receptive skills.[17] Yet, on assessment, the pattern seen is that expressive language is in advance of receptive language functioning (Figure 2). Inflated expressive language skills are seen in children with severe communication disorders, such as autism, hydrocephalus, or Williams' syndrome.

Dysarthria

Oral motor dysfunction of the speech-producing musculature (in which children have dysarthria or mechanical difficulties in speaking) is frequently seen in children with cerebral palsy and other conditions. It leads to uncoordinated oral musculature. During a physical examination, it is important to check the child's oral pharynx to rule out a cleft lip and palate—the most frequently seen anomaly of the oral facial complex—or related dysfunction or malformation of their velopharyngeal port.[15]

Learning Disability

A learning disability is often associated with speech and language problems in preschool children.[11,17,18] Children with a specific learning disability—similar to children with severe mental retardation or autism—may present with dissociation in developmental skills. For example, their language may be more delayed than their motor skills. Furthermore, not excelling academically at school reflects a disassociation between academic achievement and general intellectual abilities. Delays in language and cognitive domains may suggest a neurodevelopment diagnosis that presents as a nonverbal learning disability such as Williams' syndrome and Turner's syndrome. In such cases, a child may have impaired visual-spatial perceptual abilities.

Developmental Disorders of Language

Developmental disorders of language (language impairment) are behaviorally defined condi-

tions in which specific higher cortical functions of the brain are impaired. In a developmental language disorder, impaired language cannot be attributed to a neurological or general medical condition such as global cognitive disability, autism, hearing loss, primary or secondary social or emotional problems, or severe environmental deprivation.[18] It is characterized by a slow rate of language development in which speech begins late and advances slowly.

Children with a developmental disorder of language have a discrepancy between their cognitive functioning (nonverbal or performance measures) and their language skills.[19] Children with specific language impairment have more difficulty with grammar than with vocabulary and pragmatics. Different patterns of language impairment in developmental language disorder have distinct profiles of linguistic strengths and weaknesses. Developmental dysphasia is another term sometimes used to describe the language disorder in this group of children. There are many reasons for a developmental language disorder, which occurs in about 10% of the population.[15]

Landau-Kleffner Syndrome

An unusual cause of acquired language disorder is an epileptic syndrome called Landau-Kleffner syndrome. Children with Landau-Kleffner develop typical language skills up to 24 to 30 months of age at which age they then deteriorate or plateau. They may be confused with children with autism.[20]

Poor Language Environment

There is no evidence that parents cause language disorders except in very rare and extreme instances of neglect.[15]

Motor Delay

Referrals for motor delay are most common during the first 6 to 18 months of a child's life. By evaluating a child's developmental profile, a clinician may develop a differential diagnosis. Generalized hypotonia and motor developmental delay are the most common presenting manifestations of neuromuscular disease in infants and young children. These features may also be expressions of neurologic disease, endocrine and systemic metabolic diseases, and Down syndrome, or they may be nonspecific neuromuscular expressions of malnutrition or chronic systemic illness[21] (Table 2).

Motor Delays in Combination with Other Delays

Early motor delays are often a sign of neurological dysfunction. When a motor delay is combined with delays in other developmental domains, examining the child for visual impairment or a mental handicap should be considered.

Visual Impairment

An otherwise healthy visually impaired child is typically developmentally delayed. Early motor skills are delayed in blind children and this highlights the importance of vision as a crucial factor in the process of sensory and motor development and the need to provide alternative stimuli to reach those milestones.[22,23]

Developmental Cognitive Disability

Gross motor delay is the most common presenting symptom of mental retardation before 18 months of age.[14,21] A delay in motor milestones does not represent cognitive delay, but it is a symptom of aberrant neural development and should be regarded as a risk factor. Although motor delay is a common presenting symptom of mental retardation, most children with a developmental cognitive disability have normal ages of motor skill acquisition.[14]

Table 2

MOTOR DELAY

- In combination
  - Mental retardation
  - Visually impaired
- Alone
  - Cerebral palsy
  - Ataxia
  - Spina bifida
  - Spinal muscular atrophy
  - Myopathy

Primarily Motor Delays

When a child has only, or primarily, motor delays (Figure 3), conditions that should be considered include cerebral palsy, ataxia, spina bifida, spinal muscular atrophy, and myopathy.

Cerebral Palsy

Cerebral palsy is a nonprogressive disorder of motion and posture that results from a lesion to the developing brain,[24] and it is the leading cause of childhood disability.[25] If there is no motor delay, it is unlikely that a child has cerebral palsy.[26] The delay is caused by abnormal tone in some muscle groups and the lack of coordination between muscle groups to allow for directed movement.[27] Note the dissociation in the developmental profile where motor function and functional mobility is substantially behind other streams of development[26] (Figure 3).

Spina Bifida

Neural tube defects (NTDs) are congenital structural abnormalities of the brain and vertebral column that occur either as an isolated malformation, along with other malformations, or as part of a genetic syndrome.[28-30] Isolated (ie, nonsyndromic) NTDs are the second most common major congenital anomaly worldwide (cardiac malformations are first).[28] The most important NTD is spina bifida with meningomyelocele and most children with spina bifida will show some degree of motor paralysis.[30] The higher the lesion the greater the muscle weakness and the more ambulation will be impaired, so the more extreme the discrepancy between motor skills and other developmental domains (Figure 3). Even children with low-level lesions are likely to have significantly impaired mobility. Many infants with myelomeningocele have delayed rolling and sitting skills.[31]

Ataxia

Ataxia suggests a localized disorder of the cerebellum. Ataxia is a movement disorder that reduces motor skill activity. The differences in movement disorders, that is, ataxia, between adults and children are striking; their presentation is frequently insidious and may be characterized by mild hypotonia.[32] Hypotonia is often seen in children whose development is delayed.[33,34] The clinical symptoms of extrapyramidal disorders are profoundly influenced by the age of onset.[32]

Hypotonia

Hypotonia is the most common symptom of motor dysfunction in newborns and infants and central hypotonia is the most common cause.[33,34] The hypotonic child may present with global or isolated motor developmental delay. The developmental assessment will document delays.

The child's presentation in the neonatal period should be reviewed. Other evidence of abnormal brain function such as seizures or decreased level of consciousness is the strongest clue of central origin. Special attention must be paid to the family history to document the potential for a genetic disorder such as spinal muscular atrophy, muscular dystrophy, a congenital or metabolic myopathy, or Prader-Willi syndrome.[35] The key to diagnosing a hypotonic infant is a neurodevelopmental examination (Figure 3).[33,34]

Developmental Coordination Disorder

Older children with poor motor skills may have a development coordination disorder where their motor skills are substantially below their cognitive abilities and their clumsiness may be associated with a learning disability or attention-deficit hyperactivity disorder.[36] Parents are concerned about their child's ability to master complex motor tasks such as riding a tricycle, tying shoelaces, buttoning clothes, brushing teeth, and catching a ball. The school may report poor handwriting ro poor cutting skills. Clumsy children may have lifelong delays in achieving motor milestones. Symptoms such as weakness, ataxia, marked hypotonia or hypertonia, especially if progressive or asymmetric, suggest other diagnoses. Children with Asperger's disorder are frequently clumsy; however, their neuropsychological profiles display significantly stronger verbal skills than nonverbal abilities.[36]

Fine Motor-Adaptive Delay

A delay in fine motor-adaptive development, combined with delays in other developmental domains, should prompt concerns about a visual impairment or mental handicap (Table 3).

Visually Impaired

Visual impairment is a cause of apparent clumsiness.[22] It is imperative to assess the eyes and visual acuity of a child presenting with delayed fine motor-adaptive development.[23] It is never too early to examine a child's eyes and test their visual acuity.

Physically Handicapped—Cerebral Palsy or Brachial Plexus Injury

If the delay occurs primarily in one developmental domain, one

Table 3

FINE MOTOR-ADAPTIVE DELAY

- In combination
  - Visually impaired
  - Mentally retarded
- Alone
  - Hemiplegia
  - Erb's, Klumpke's
  - Fractured clavicle
  - Developmental coordination disorder
  - Disorder of written expression

should look closely to see whether the child has hemiplegia, a brachial plexus injury—such as Erb's or Klumpke's palsy—or a fractured clavicle. All asymmetries of movement in the first 2 to 3 years should be investigated.[37]

Disorder of Written Expression or Developmental Coordination Disorder

In older preschool or elementary school children with fine motor delays, a disorder of written expression or a developmental coordination disorder should be considered. A disorder of written expression is a learning disability in which a child displays a markedly reduced ability to organize and present information in writing compared to a stronger ability to organize and present information orally. Weaknesses in writing skills may arise for different reasons such as a deficit in visual-motor integration and motor coordination.[38]

If motor coordination alone is responsible for the writing weaknesses the child's symptoms are better regarded as a motor skills disorder such as developmental coordination disorder, which presents in about 6% of all children. Children with developmental coordination disorder generally have normal intelligence, but the condition is often associated with attention deficit hyperactivity disorder or a learning disability.[36,38]

Personal and Social Delay

When a child presents with personal and social delays, it is important to take into account whether the child has a developmental cognitive disability, autism, or is growing up in an environment of abuse, neglect, deprivation, or other less-than-ideal family dynamics (Table 4).

Developmental Cognitive Disability

More than half of the children with a developmental cognitive disability are impaired in everyday functioning by their symptoms compared with one third to one half of the nonintellectually disabled children, suggesting that psychopathology is more likely to cause impairment in everyday life in children with a developmental cognitive disability.[39]

Autism

A hallmark of autism is a serious impairment in the development of social skills.[16] To diagnose autism, it is important to observe whether the child shows restricted and repetitive behaviors (such as sustained odd play) and has social interaction disorders (such as difficulties playing with others in an age-appropriate way). Before 30 to 36 months, these behaviors—along with a severe communication disorder—may suggest a diagnosis of autism (Figure 2).[16,40] Although autism is a neurobiological disorder, there is no single cause, and the lack of a specific biological marker requires attention to the observable behavioral traits.

Table 4

PERSONAL SOCIAL DELAY

- Mental retardation
- Autism
- Abuse, neglect, deprivation
- Dysfunctional parenting, ergo, spoiled child

Reactive Attachment Disorder

A child who fails to discriminate between familiar and unfamiliar persons and respond differentially may have a reactive attachment disorder. Disorders of attachment may be relatively common in very-high-risk children of 12 to 48 months.[41-44] DSM-IV criteria for reactive attachment disorder require clinicians to document "abnormal social behavior" evident "across social contexts" and beginning before age 5 for diagnosis. Two subtypes of social behavior are possible: (1) indiscriminate sociability, in which the infant or young child readily engages with and seeks comfort from strangers, and (2) inhibited behavior, in which the infant or young child actively and fearfully disengages from caregivers, seeking little comfort in times of distress. Discriminating sociability emerges between 2 and 7 months of age. A dissociation between social skills and other milestones may raise suspicions of the diagnosis (Figure 2). Many children with reactive attachment disorder have language delays although the language delays are not uniformly severe.[41-44]

Abuse and Neglect

Child abuse can result in a development disability that may lead to further abuse. Any child in whom abuse occurs should be thoroughly evaluated for disabilities.[44,45] Neglect has extreme and long-term detrimental effects on a child's development. It can result in poor attachment, developmental delay, poor physical development, and antisocial behavior.[45]

Family Dysfunction

Social and self-help behaviors are influenced by environmental factors, level of parenting skills, education, and training.[1] Children's physical and emotional health and their cognitive and social functioning are influenced by how well their family functions. Children's needs, for which only a family can provide, include social support, socialization, and coping and life skills.[45]

Discussion

More than half of the children with delays, especially language-related delays, are not detected before starting school.[46] More than half of the primary care providers do not use a standardized instrument.[5] Using a validated screening tool would improve detection and aid in discerning patterns of delay that can help with the diagnosis of conditions that have a high probability of developmental delay.[47]

Screening Tools

Evaluating a child's development is essential in all clinical practices serving children, and all tests have their strengths and weaknesses.[2,5,47] A commonly used measure is the Denver II test.[6] Although pediatricians traditionally use the Denver II, cautious use is recommended because it may not detect cerebral palsy in a child's first 12 months and it consistently overrefers or underdetects. Some parent-report measures include the Parents' Evaluation of Developmental Status, the Ages and Stages Questionnaire, and the Child Development Inventories. Office checklists are commonly used,[47] but they are not recommended, because they are not standardized and validated and often miss developmental or behavioral concerns.

Neurodevelopmental Assessment

These instruments should never be used for a diagnosis or in place of a thorough neurodevelopmental history and physical examination.[1,5] Developmental delay is a symptom not a diagnosis. Listening to parents, paying special attention to their concerns, and doing a meticulous history makes the greatest contribution to a neurodevelopmental diagnosis.

Major malformations for syndrome identification should be obvious to the clinician on physical examination.[48-50] Careful examination of a child's head, extremities, and skin will allow the clinician to identify more than three fourths of minor anomalies in children suspected of having a neurodevelopmental disorder.[49]

The neurodevelopmental examination should include the classical neurological examination. An expanded neurological examination should include observations of minor neurological signs, associated movements, fine motor skills, and gait. Motor soft signs may demonstrate asymmetry of function, abnormal tone, or difficulty with motor control. This information is combined with a child's developmental profile to formulate a neurodevelopmental diagnosis.

Importance

Why is understanding the cause of a developmental delay important? Having an explanation for a child's delay helps the clinician design a treatment plan and can lessen a family's anxieties. By knowing the etiology of a child's delay, the clinician and family know what associated conditions to expect. A diagnosis guides the clinician in looking for rare but treatable diseases and helps predict the risk of a similar condition occurring in future pregnancies.[48-50]

Helping a family organize a plan to address a child's developmental delay is essential. Clinicians should seek resources in the patient's hometown and further away if necessary. Directing the child to early intervention services is a sound first start. Any child less than 3 years of age who has a physical or mental condition that has a high probability of resulting in developmental delay is eligible for services, so a neurodevelopmental diagnosis helps qualify a child for early intervention services.[51,52] Services are mandated for infants, toddlers and preschoolers. Although eligibility criteria vary by jurisdiction, common measures of delay are 25% and/or two standard deviations in one or more of the developmental domains.[52] Recognizing developmental patterns that guide a neurodevelopmental assessment is essential to help families get the support services they need.[53]

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