CHALLENGING CASE: DEVELOPMENTAL DELAYS AND RERESSIONS

A Two-Year-Old Boy with Language Regression and Unusual Social Interactions*

CASE

Jimmy, a 2½-year-old boy, was seen for the first time by a new pediatrician after a recent family move. His mother made the appointment for a health supervision visit although she had concerns about his language and social skills. She stated that he spoke primarily with unintelligible sounds and often communicated by pointing with his finger. He spoke only 10 words that were clear enough to be understood. Jimmy’s mother said that he could hear, but she was not sure whether he understood everything she said. Although he played at home with his 4-year-old brother, he typically played by himself when he was in the presence of other children. Jimmy’s mother was asked if he had had a 2-year-old visit to a pediatrician and what the assessment was at that time. She said that his delayed development was discussed with the pediatrician, but she was reassured that he would progress during the following year.

An uncomplicated full-term gestation was followed by a vaginal delivery without perinatal problems. Jimmy was a “calm” baby who was breastfed for the first 6 months of life. He sat at 7 months, pulled himself up to stand with support at approximately 9 months, and walked at 13 months. Transitions were always difficult for Jimmy; he screamed and was difficult to settle whenever cared for by someone other than his parents. He typically resists physical contact when children or adults approach him. His mother recalled that language emerged early. He acquired a significant number of words between 12 and 15 months of age. Jimmy apparently recognized letters when his parents were teaching the older sibling. At 15 months, Jimmy’s language output regressed dramatically, and by 18 months, he no longer used words to communicate. Since then, he has spoken fewer than 10 single words. He mostly babbles and uses repetitions of the same sounds.

The pediatrician inquired into family structure and life events at the time Jimmy lost language milestones. He was told that, at this time, the father, an engineer, changed his position in the company and began to travel extensively. Jimmy’s mother thought the absence of his father might be related to the language regression. She also noted that Jimmy seemed to have a stronger attachment for his father: “Jimmy has always been attracted to his father, and his brother seems to prefer me.” The parents’ marriage was strong and free of any major disharmony.

During the interview, the pediatrician noted that Jimmy played persistently with his set of small trains, repetitively lining them up in order. He was not interested in other toys that were on the floor next to him. He ran around the trains, mostly on his toes, while making unintelligible sounds. He looked away when the pediatrician called his name and became agitated when his mother attempted to redirect his attention to the examination.

The pediatrician, 4 years in practice after his residency, had never seen a child with Jimmy’s pattern of development. That Jimmy’s development was unusual in two domains was apparent to his pediatrician from the preceding information and brief observations. He asked himself what the next steps should be.

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Dr. Martin T. Stein

At the end of the second year of life, Jimmy experienced a loss of previously acquired language skills associated with several atypical behaviors during play and social interactions. Regression of language skills persisted and was temporally associated with periodic separations from his father because of his father’s employment demands. Jimmy’s attention was sustained when he played repetitively with trains, which he preferred to line up in a specific order; however, he could not be engaged in either conversation or play with his pediatrician.

The monitoring and assessment of language milestones are, arguably, the most challenging components of developmental surveillance for most pediatricians. The range of predicted acquisition of many language skills is broad, and pediatricians often must rely on a parent’s report rather than on clinical observation. What makes this case different from the toddler who presents isolated language delay is the presence of unusual social skills and sensitivities. It should immediately raise the possibility of a pervasive developmental disorder (PDD) and other diagnostic categories.

As a result of an expanded perspective on the initial presentation, diagnostic criteria, interventions, and prognosis of children with PDD (also known as “autistic spectrum disorder”), expectations for early recognition, referral for evaluation, and treatment have changed dramatically. Ten to 15 years ago, a general pediatrician with a busy office practice may
not have recognized a toddler with mild to moderate developmental delays consistent with an autistic spectrum disorder. In fact, the average age of diagnosis of PDD in the United States is 3 to 4 years. Currently, however, there is sufficient empirical research to demonstrate the effectiveness of early and intensive interventions in optimal educational settings when they occur for at least 2 years during the preschool period.1,2 Recent studies demonstrate improved outcomes in most young children with autism, including speech in 75% or more and significant improvement in social skills and intellectual performance.3,4 The challenge now for primary care pediatricians is to design and implement an office-based screening program that is sufficiently sensitive to identify those young children who might benefit from further assessment.

Drs. Charles Cowan and Suzanne Dixon comment on a diagnostic approach to Jimmy’s presentation with the limited information available in the case summary. The history obtained from Jimmy’s mother and the brief office observations reflect time limitations in most primary care settings. Dr. Cowan is a Clinical Professor of Pediatrics at the University of Washington. Before joining the Neurodevelopmental Program at the Children’s Hospital and Regional Medical Center in Seattle, he was a primary care pediatrician. He brings the perspective of a generalist into the discussion. Dr. Dixon is a developmental-behavioral pediatrician and Emeritus Professor of Pediatrics at the University of California San Diego. Currently she is at the Great Falls Clinic in Great Falls, Montana, where she evaluates and treats children who have a wide range of developmental and behavioral problems.

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Jimmy presents an atypical pattern of behavior and development, encompassing language, social-relatedness play, altered sensitivities, and poor adaptability. This cluster suggests the autistic spectrum of disorders, severe language disorders, or a difficult temperamental profile.

Regression in language was seen at 15 months. Although this was coincidental with a life stressor, we cannot now explain this lasting loss of skills by an adjustment reaction that usually lasts less than 6 months. It is now more than a year since the stressful event, and the language and behavior are unusual, as well as delayed.

Language regression with autistic behavior suggests the Landau-Kleffner (L-K) syndrome, epileptiform aphasia. This disorder presents with loss of language and seizures, often subclinical, and sometimes is only apparent on an overnight sleep electroencephalogram (EEG) or other special studies. Temporal sharp waves are typical. Recent reports suggest that EEG irregularities are as common in autism as in L-K syndrome, blurring the distinction. Treatment is high-dose steroids for 6 to 12 weeks and analeptic medications. Sustained, dramatic improvements can be seen with treatment, especially in the youngest children.2,3

Fragile X syndrome should be considered, especially if this boy has long ears and face and hyperextensible joints. At 2 years of age, his testes would be of normal size and the phenotype subtle. CATCH 22, another chromosomal disorder, is also a possibility and can present the behavioral profile without the cardiac or other features. Other considerations are the neurocutaneous syndromes, especially the tuberous sclerosis complex. A careful skin examination should be performed for depigmented macules, often the first visible sign of the disease and only seen with an ultraviolet light (Wood’s lamp). A complete family history is in order; family studies have shown a 50- to 100-fold increase in the rate of autism in first-degree relatives. Metabolic disorders and congenital infections may result in an autistic-like profile, but they would usually present with a generalized delayed pattern of early development and microcephaly. Most children with autistic disorders have large head sizes compared with typically developing children. If there have been regressions in other areas of development, the differential diagnosis widens to include several neurodegenerative disorders.

A temperamental profile of social withdrawal, poor adaptability, and high persistence could explain some of Jimmy’s unusual behaviors. However, the atypical language and the restricted and repetitive play patterns are not explained by temperamental differences alone. A severe language disorder can present with some of the atypical behaviors described here. I am skeptical that this is the case because of the language regression and the highly unusual behavior.

The Diagnostic and Statistical Manual for Primary Care: Child and Adolescent Version describes social withdrawal problems for youngsters who do not fully meet the criteria for autistic disorder.4 The term “pervasive developmental disorder, not otherwise specified” is used to describe youngsters who have some, but not all, of the features of autism. As we learn more about Jimmy, we may find that these terms best describe his behavior. However, it will not change what needs to be done, medically or educationally.

The physician needs more family history, because language disorders and autism run in families, and a
detailed early developmental history, including other developmental regressions. A physical examination to assess skin, joint mobility, cardiac status, facial features, and neurological parameters is the next step. A hearing test is needed with any language problem. The chromosomal studies should be sent. A referral to a behavioral pediatrician and/or pediatric neurologist would help in this workup, which should include the overnight EEG.

A complete speech and language appraisal will set the baseline and aid in educational planning. This child is a candidate for early intervention/educational programs for the communicatively handicapped. This family will need support throughout this evaluation process as they learn that there is a serious concern. The observations of the speech pathologist, the neurologist and/or the behavioral pediatrician, the early intervention intake staff, and others would support or refute the presence of the autistic features in this child.

The young pediatrician’s instinct that this is an unusual child is a good insight. He will probably see many more in his practice life, and early identification and treatment is vital.

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Jimmy had normal early development, including a “significant number” of single words by 15 months of age. His language progression then halted and even “regressed.” By 2 years of age, typically developing children have vocabularies in excess of 20 words and should be using 2-word sentences. However, at 2½ years of age, Jimmy had only infrequent use of 10 words. Primarily, he babbled or used unintelligible repetitions of sounds; his mother reported that he did communicate with some gestures. His receptive language seemed delayed, because his mother was not sure he understood her and he actively turned away when his name was called. Behaviorally, he showed early difficulty with transitions and avoided physical contact; later, he played with toys in a repetitious fashion, danced around on his tiptoes, and played by himself in the presence of other children.

Although this developmental pattern is suggestive of an autism spectrum disorder (ASD), other conditions need to be considered. The most striking component of this child’s history is that of abnormal language development. The differential diagnosis of abnormal early language development falls into three major categories: specific speech-language disorders (e.g., articulation disorders, developmental language disorders, etc.), cognitive deficits, and the social and communication deficits that characterize ASDs. In addition, the presence of language regression after the acquisition of normal language should suggest the possibility of the L-K syndrome or acquired epileptic aphasia.1 There are associated convulsive seizures in 70% to 80% of cases. Hearing loss must also be considered and systematically ruled out by a complete audiometric evaluation. A history of “he hears well” should never be accepted.

When autism or one of its variants is considered, a careful history of early language and social development is critical. Attention to early nonverbal communication, such as pointing to show or share (protodeclarative pointing), eye contact, orienting to name, and other communicative gestures (“bye-bye” and “hi”), will assess important areas of social development. Pretend play should be established by 18 months in typically developing children. It can be demonstrated with a plastic cup and spoon or similar objects during a routine well-child examination. A history of repetitive mannerisms (rocking, twirling, hand flapping), sensory oddities (aversion to common household noises such as vacuum cleaners or blenders), and persistent preoccupation with parts of objects (wheels of toy cars) also suggests a diagnosis of ASD. Finally, a standardized autism screening questionnaire, such as the Checklist for Autism in Toddlers (CHAT)2 or the Pervasive Developmental Disorders Screening Test (PDDST),3 may be used. These are tools that pediatricians should add to their screening armamentarium, in addition to the standard Denver II.

Distinguishing ASDs from specific speech-language disorders and global cognitive deficits is never an easy task. Referral to a child development team, skilled speech-language therapist, child psychologist, child neurologist, or a developmental-behavioral pediatrician is in order if any of these diagnoses are contemplated. Initial evaluation and therapy can also be initiated by referral to an early intervention program or public school program available in all communities. ASDs are currently diagnosed with much greater frequency, and all primary care physicians should have office protocols for screening of these disorders (Fig. 1).4

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Routine Developmental Surveillance

- Absolute indications for immediate evaluation
  - No babbling by 12 months
  - No gesture (pointing, waving bye-bye, etc.) by 12 months
  - No single words by 16 months
  - No 2-word spontaneous (not echolalia) phrases by 24 months
  - Any loss of language or social skills at any age
- Always screen for hearing loss

Fig 1. Screening for autism spectrum disorders. Modified from Filipek et al. 4 *Checklist for Autism in Toddlers*; **Pervasive Developmental Disorders Screening Test–Stage 1.3

Web Site Discussion

The case summary for this Challenging Case was posted on the Developmental and Behavioral Pediatrics web site* (http://www.dbpeds.org) and the Journal’s web site (http://www.lww.com/DBP).

Comments were solicited.

Keith Goulden, M.D., from Edmonton, Alberta, Canada, wrote: “For the child with autism, early intensive intervention is required, although the specific program available locally may follow one of a number of ‘schools’ (how intensive is ‘intensive’?). There is evidence for discrete trials therapy (e.g., Lovaas), but there is also evidence for education programs and communication enhancement programs; a mixture of the three is probably best, if available. The pediatrician may be very helpful to a family trying to negotiate the shoals of competing ‘cures,’ as long as their own biases don’t lead them to encourage a family to ignore available resources. The autism societies are also very helpful in general.

‘Depending on local circumstances, this case might be handled most efficiently by referral to a tertiary team assessment program, or might require some local individual referrals as well. The pediatrician has a role in ongoing management even if he is not required as ‘care coordinator.’ There is a need for information to be available to the family, including grandparents, siblings, etc.; there are genetic counseling implications for both the parents and extended family.’

W. Douglas Tynan, M.D., wrote: “From the data presented, this child certainly fits in the autistic spectrum. As Kanner originally described these children in the 1940s, there is often regression in language skills at 15 to 18 months, but what may be happening is that they never move from single words into appropriate syntax and complex language.

‘Some parents look to events that occurred around 16 months of age to account for the regression at 18 months. In this case, the parents are concerned about the father’s change in work as a possible impact to
explain their child’s development. Recently, we see parents who attribute the cause of the developmental regression to immunizations the child received at 14 to 16 months of age. They need to be given a clear explanation about the developmental course of this disorder.”

Morris Wessel, M.D., from New Haven, Connecticut, wrote: “I am intrigued by the case of Jimmy. In our practice the first thing we would want to do is to be certain that his hearing is adequate. I recently saw a child who became increasingly deaf at 11 years of age. There is a strong family history of this syndrome. If it could happen at 11 years, maybe in some instances it could happen at 2. In addition, the regression suggests to me that something in his receptive skills changed. I also would be concerned about a processing deficit; possibly he doesn’t understand what he hears, or maybe he is unable to process his thoughts into language.”

Dr. Martin T. Stein

The recognition of a unique pattern of behaviors described as “autistic disturbances” was reported in 1943 by the child psychiatrist Leo Kanner. In 11 children who presented with symptoms between 2 and 8 years of age, he described delayed acquisition of language characterized as noncommunicative and echolalic; social remoteness; a preference for repetitive, stereotyped, nonimaginative play; and a “need for sameness.”

Although Kanner’s original description continues to describe these children, the diagnostic criteria for autistic disorder have been modified in response to a recognition of a continuum of cognitive and neurobehavioral manifestations. The three core features include impairments in socialization, impairments in verbal and nonverbal communication, and restricted and repetitive patterns of behavior. The DSM-IV emphasizes the qualitative attributes of symptoms by defining a range of impairments, rather than the absolute presence or absence of a particular behavior (Table 1).

That the language patterns and atypical behaviors seen in young children with ASDs evolve gradually in the first 2 years of life is perhaps the most important guide to an early diagnostic strategy for primary care pediatricians. The development of most of these children seems normal to parents and clinicians in the first year of life. However, when home videotapes of first birthday parties in infants with autism were compared with those of infants showing typical development, four behaviors correctly identify more than 90% of the autistic infants. They included diminished eye contact, an inability to orient to name when called, point with a finger to indicate interest in something, and show an object by bringing it to a person. Delays in pointing and showing are early signs of atypical development in “joint attention behaviors.” Recent studies demonstrate that these characteristics of autism are measurable by 18 months of age and stable through the preschool period. From these observations, 18 months is considered to be an optimal time for early assessment of children with ASD.

Figure 1 in Dr. Cowan’s commentary describes a plan for developmental surveillance of all toddlers beginning with the first birthday. It is consistent with the algorithm recently proposed by a multidisciplinary consensus panel convened by the Child Neurology Society and the American Academy of Neurology. In a comprehensive review of the literature on ASD, the consensus panel emphasized the importance of developmental surveillance for children seen in primary care medical offices. Any of the five absolute indications of early language and social delay or regression should trigger an evaluation with a specific screening test for autism. Screening instruments with good validity and high sensitivity and specificity include the CHAT, designed to screen for ASD at 18 months, and the PDDST-Stage I, a parent questionnaire designed for use in primary care settings for infants and toddlers up to 36 months. The CHAT includes nine questions to be answered by a

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<th>TABLE 1. Diagnostic Criteria for Autistic Disorder</th>
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<td>A total of six (or more) items from (1), (2), and (3), with two from (1), and at least one each from (2) and (3):</td>
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<td>(1) qualitative impairment in social interaction, as manifested by at least two of the following:</td>
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<td>(a) marked impairment in the use of multiple nonverbal behaviors such as eye-to-eye gaze, facial expression, body postures, and gestures to regulate social interaction</td>
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<td>(b) failure to develop peer relationships appropriate to developmental level</td>
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<td>(c) a lack of spontaneous seeking to share enjoyment, interests, or achievements with other people (e.g., by lack of showing, bringing, or pointing out objects of interest)</td>
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<td>(d) lack of social or emotional reciprocity</td>
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<td>(2) qualitative impairment in communication as manifested by at least one of the following:</td>
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<td>(a) delay in, or total lack of, the development of spoken language (not accompanied by an attempt to compensate through alternative modes of communication such as gesture or mime)</td>
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<td>(b) In individuals with adequate speech, marked impairment in the ability to initiate or sustain a conversation with others</td>
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<td>(c) stereotyped and repetitive use of language or idiosynthetic language</td>
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<td>(d) lack of varied, spontaneous make-believe play or social imitative play appropriate to developmental level</td>
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<td>(3) restricted repetitive and stereotyped patterns of behavior, interests, and activities, as manifested by at least one of the following:</td>
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<td>(a) encompassing preoccupation with one or more stereotyped and restricted patterns of interest that is abnormal either in intensity or focus</td>
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<td>(b) apparently inflexible adherence to specific, nonfunctional routines or rituals</td>
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<td>(c) stereotyped and repetitive motor mannerisms (e.g., hand or finger flapping or twisting, or complex whole-body movements)</td>
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<td>(d) persistent preoccupation with parts of objects</td>
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<td>Delays or abnormal functioning in at least one of the following areas, with onset prior to age 3 years: (1) social interaction, (2) language as used in social communication, or (3) symbolic or imaginative play.</td>
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<td>The disturbance is not better accounted for by Rett’s Disorder or Childhood Disintegrative Disorder.</td>
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parent and five specific observations by a clinician (Table 2). This model of early screening for ASD is consistent with the principles of developmental surveillance.7

ASDs are not rare. As the clinical phenotype expanded and clinical recognition improved, recent prevalence studies have indicated that 1 to 2 of 1000 children have the disorder, with a male-to-female ratio of approximately 3:1 to 4:1.4 This translates to between 60,000 and 115,000 children younger than 15 years of age in the United States who meet diagnostic criteria for ASD.8 These estimates include children with milder forms of the disorder. The pediatrician’s challenge for early diagnosis is heightened by studies of children with a delayed diagnosis. In one study, 15% of children who were in special education classes, but without a diagnosis of autism, met diagnostic criteria for ASD.9 In another study, 74% of children with high-functioning autism had been misdiagnosed with attention-deficit hyperactivity disorder (ADHD).10 This is a reminder to pediatricians that ADHD-specific parent and teacher questionnaires do not identify developmental domains of social competence, cognitive skills, and the limited range of activities seen in children with milder forms of ASD who may appear to have ADHD.

TABLE 2. Checklist for Autism in Toddlers (CHAT)—A Screen for Autism at 18 Months of Age in a Primary Care Office

Section A: To be completed by parent

1. Does your child enjoy being swung, bounced on your knee, etc.? 
2. Does your child take interest in other children? 
3. Does your child like climbing on things, such as up stairs? 
4. Does your child enjoy playing peek-a-boo/hide-and-seek? 
5. Does your child ever PRETEND, for example, feeding or diapering a doll, or pretend other things (e.g., a block represents a cookie)? 
6. Does your child ever use his/her index finger to point to ASK for something? 
7. Does your child ever use his/her index finger to point to indicate INTEREST in something? 
8. Can your child play appropriately with small toys without mouthing, fiddling, or dropping them? 
9. Does your child ever bring objects over to you (parent) to SHOW you something?

Section B: To be completed by clinician

1. During the appointment, has the child made eye contact with you? 
2. Get the child’s attention, then point across the room at an interesting object and say, “Oh, look, there’s (a name of toy)! Watch the child’s face. Does the child look across to see what you are pointing at? 
3. Get the child’s attention and then give the child a doll and miniature cup and say, “Can you give the baby some juice?” The person asking the question should hold the doll and cup in front of the child with no further cues. The child should “feed” the baby from the cup. 
4. Say to the child, “Where’s the light?” or “Show me the light.” Does the child POINT with his/her index finger at the light? 
5. Can the child build a tower of 3 blocks?

If “NO” is checked on more than 3 of the following items: A5, A7, B2, B3, and B4, the child is at risk of having autism/PDD because he has not responded to referential pointing, gaze monitoring, or pretend play.

Modified from Baron-Cohen et al.5

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