CHALLENGING CASE: DEVELOPMENTAL DELAYS AND REGRESSIONS

Expressive Language Delay in a Toddler*

CASE

Shelly is a 20-month-old white female whose parents thought she was in excellent general health when she came to her pediatrician for a health supervision visit. A developmental survey consisting of focused questions revealed that Shelly spoke only occasionally with a vocabulary limited to five words. Although motor and social skills were age-appropriate, verbal expressions typically consisted of nonintelligible utterances and frequent pointing to objects. She occasionally chatters “as if she had her own language.” Shelly reportedly responds to directions appropriately, and she appears to hear normal human voices, music, and a telephone ring.

Shelly has been in home day care since 10 months of age when her mother returned to work. With four other toddlers, she is cared for by a Spanish-speaking caretaker; her parents speak English at home. She is the only child in her family; her parents remarked that they each had a sibling whose early language acquisition was delayed but as adults did not seem language impaired.

Shelly’s prenatal course was complicated by premature contractions treated from 30 weeks gestation with terbutaline. She was delivered at term by a spontaneous vaginal delivery without complications. Apgar scores were 8 at 1 minute and 9 at 5 minutes.

On physical examination, Shelly appeared robust. Social and visual engagement occurred easily with her mother and the examiner. Growth parameters were at the 50th percentile. The examination was normal, including her tympanic membranes (normal appearance and compliance), palate, pharynx, facial structure, and neurological assessment. Gross and fine motor skills were documented at the 20- to 24-month level. She responded to commands given by her mother and the examiner. She was able to point to pictures of objects on request and correctly pointed to three body parts. When asked to “go get your shoes and sit down,” she completed the task after the second request. Throughout the interview and examination, Shelly did not say any specific words. However, she pointed to a toy and doll she wanted during a play situation.

Dr. Martin Stein

Developmental assessment constitutes the foundation of pediatric health supervision. Among the major domains of early childhood development, the recognition of language delay remains the most elusive for child care clinicians. Compared with motor and social-adaptive skills, language acquisition monitoring in pediatric practice is challenging for at least two reasons: (1) The clinician is often dependent on historical information from a parent or other caregiver, especially in the first 3 years of life when a toddler’s speech output in an office setting may be limited. In this age group, an assessment of walking skills, pincer grasp, and block building is relatively easy to accomplish compared with a successful survey of expressive and receptive language; and (2) the normal range of language skill acquisition between 1 and 3 years of age is broad and is affected by a variety of biological and environmental factors.

The case of Shelly represents a common diagnostic and therapeutic problem in primary care practice: a toddler with an isolated delay in expressive language development.

Pediatricians with different perspectives on the initial clinical approach to this case have been invited to comment. Using a similar database, their clinical construction of Shelly’s language delay varies in important ways that yield different diagnostic recommendations.

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Dr. James Coplan is an Associate Professor in the Department of Pediatrics at State University of New York in Syracuse. He is a developmental pediatrician with a specific interest in early childhood language development. He developed a widely used screening instrument known as the Early Language Milestone Scale.

Dr. Heidi Feldman is an Associate Professor of Pediatrics in the Department of Pediatrics at the University of Pittsburgh School of Medicine. She is a developmental pediatrician who also completed a doctorate degree in developmental psychology. Dr. Feldman directs the Division of General Academic Pediatrics at the Children’s Hospital of Pittsburgh.

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This case exemplifies the most common scenario of language delay facing the primary care clinician: the otherwise normal child with excellent receptive language but clear expressive language delay.

The most critical feature presented in the history is Shelly’s apparent normal receptive language. Adequate receptive language suggests that both the inputs for language (e.g., hearing) and the processing language are normal. If true, normal receptive language effectively rules out such important parts of the differential diagnosis as mental retardation, auditory processing disorder, developmental language disability, significant hearing loss, and other significant communication deficits (e.g., autism).

Shelly’s language difficulties are occurring on the output side of language development, which considerably narrows the potential etiologies. Apraxia or other fine motor problems of the oral motor system, although plausible, are less likely in association with normal gross and fine motor skills in other areas. Additionally, in such cases there are often clues, such as early feeding disorders or prolonged drooling. Bilingualism should be considered as a possible contributing factor. Although somewhat controversial, it is fairly well established that bilingual children achieve expressive language milestones at a slightly later date than their monolingual peers. In any event, bilingual children should catch up to monolingual children in the expressive realm by 3 years of age. Additionally, their receptive language should always be at age level. It is possible that Shelley’s task of simultaneously learning two languages is a factor in her delayed expressive language.

Finally, constitutional or maturational expressive language delay should be considered. This is a diagnosis of exclusion and is only made after one has ruled out all of the other treatable causes of expressive language delay. In Shelly’s case, this diagnosis is suggested by a positive family history. Against this diagnosis, however, is her female gender because it is well established that young boys are more commonly delayed in language skills than females.

The diagnostic dilemma facing the primary care clinician is: How much of a workup should be performed at this time, and should language treatment be initiated? Although the yield is probably low in this case, we recommend that all children with language delays receive a hearing test. Subtle (conductive or sensorineuro) hearing losses can contribute to expressive language delay, while largely sparing receptive language. Formal evaluation of language development is not essential in this child except to assure that language testing and parental history accurately reflect Shelly’s true language skills. Nevertheless, the decision about therapy at this stage is often a clinical quandary. Perhaps the best way to decide is by assessing the quality of the linguistic environment in which the child is functioning. If her day care and home contribute linguistically enriched environments in which language is used to communicate, to label everyday events, to express ideas and feelings, and to elicit these from Shelly, then language therapy is unlikely to provide appreciable supplemental benefits at this point. However, if Shelly is in a chaotic day care where there is very little language directed at her or expectation for her to use language to express herself and if the parents do not use language in a reciprocal, eliciting way with the child (which you may observe in the course of the office visit), language therapy can be very helpful, especially when the caregivers are involved.

Regardless of the decision made, Shelly should be followed up carefully by the primary care clinician. Language testing at 2 years of age may reveal marked improvement or continued deficits. If the delays continue, the same process of generating a differential diagnosis and potential treatment plan should be entertained.

There are no easy answers in this case, which is both common and subtle. The clinician must walk the fine line between delaying treatment in a child who will go on to have continued language deficits and for whom early treatment is beneficial and overtreating a child with a constitutional, maturational delay for whom treatment is unnecessary and for whom labeling may present a whole set of new problems for family and child.

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REFERENCES

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Shelly is a 20-month-old with delayed speech. She follows one-step commands, two-step commands (inconsistently), and points to body parts on command. Based on these observations, her receptive language skills appear to be normal.

We are told that Shelly’s growth parameters are at the 50th percentile. Does this include head circumference? Limited information regarding adaptive skills (“normal . . . fine motor skills”) is provided. Specific information regarding age at acquisition of spoon use, object permanence, means/ends behavior, and current play activities would be useful. Despite these gaps in our database, Shelly’s normal receptive language makes global cognitive delay (i.e., mental retardation) unlikely. She gives good eye contact, is socially engaging, and points appropriately to desired objects. These observations make infantile autism unlikely.

She responds to directions, “appears to hear normal human voices, music and a telephone ring,” and manifests “chatting as if she had her own language.” These observations rule out bilateral profound hearing loss. However, she could still have moderate, unilateral, or high frequency hearing loss.
Day care, bilingual upbringing, and the fact that she is an only child are irrelevant. Bilingual upbringing does not delay speech. First-born children and later-born children talk at the same time, and there is no evidence that parents who return to work prejudice their child's rate of development.

Shelly's developmental pattern may represent a variation of normal. However, this can only be determined in retrospect. Invoking "variation of normal" at this point as a justification for diagnostic inactivity is inappropriate. Partial hearing loss is still a possibility. Office screening for hearing loss at this age is impossible. Toddlers "cheat" by watching the examiner. The background noise level in a "quiet" office is 50 to 60 dB, far too loud for adequate hearing screening, and most of the stimuli available to the examiner, such as a hand-held bell, are far too loud (90 dB). For example, the bell in the Early Language Milestone Scale is a measure of auditory localization, a cognitive behavior, and is not intended as a measure of hearing. Shelly's history is most consistent with developmental language disorder (DLD).

DLD describes a heterogenous group of conditions with varying degrees of expressive and receptive language impairment, in the absence of hearing loss, emotional disturbance, global cognitive delay, autism, or physical disability. "Pure types," such as the receptive and expressive aphasias of adulthood, are rare during infancy. Expressive difficulties are noticed first, with delayed speech, delayed emergence of complex grammatical forms (phrases, sentences), and delayed acquisition of intelligibility. Typically, receptive language is initially regarded as "normal." As the child gets older, however, a variety of subtle receptive language abnormalities often surface ("auditory processing" disorders, impaired short-term auditory memory, etc.). These can be virtually impossible to detect in a 20-month-old child. The history of language delay in the parents' siblings may be relevant because DLD frequently migrates in families (although without a clear mendelian pattern). What is the parent's level of educational attainment and reading ability?

I would obtain an audiogram. If Shelly has not acquired a spoken vocabulary of a couple of dozen single words as well as the ability to produce some two-word phrases by her second birthday, I would refer her to a speech pathologist at that time. If significant language delay is documented, I would inquire about drug, alcohol, or other teratogenic exposure during pregnancy. I would consider G-banded chromosomes, Fragile X by DNA probe analysis, and HIV by ELISA because language delay can be the presenting feature of HIV encephalopathy.

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Dr. Heidi Feldman
Assessment and management of language development in toddlers, as exemplified in this case, may challenge even experienced child health care providers. First, typical toddlers rarely display their linguistic skills in the office. Second, although approximately 10 to 15% of children show substantial delays in language development at 2 years of age, only 4 to 5% remain delayed beyond age 3 years. The clinician must differentiate children who are "late bloomers," those who catch up with peers without special services, from children with language impairments, those with persistent delays who benefit from early intervention. Research in developmental psychology provides some assistance in this clinical area.

When direct observations of a child's language abilities are not possible, parents offer reliable and valid descriptions of their child's language skills. Their accuracy increases when they are asked to report on current functioning rather than historical milestones, emerging rather than well practiced skills, and when the question format requires recognition of the child's abilities rather than recall of specific vocabulary or sentences. A new parent inventory, the MacArthur Communicative Development Inventory, integrates these features and may be useful in a pediatric practice when parents have concerns about language. Based on the parent report in this case, the health care provider could reasonably conclude that 20-month-old Shelley is delayed in expressive language skills. Infants typically acquire their first words at about 1 year of age and slowly acquire a vocabulary of 50 words by 18 months of age. When the vocabulary reaches 50 to 75 words, most children rapidly accelerate the pace of language learning, acquiring, on average, four to six words per day. Simultaneously, the child begins to combine words into phrases and sentences. Shelley, who uses five words at 20 months of age, is functioning at the 12- to 15-month-old level, or 60 to 75% of age expectations.

The prognosis for the pace of future language development depends in part on a survey of risk factors. Many studies show increased family aggregation of language disorders, strongly suggesting a genetic contribution. Hurst and colleagues described a family pedigree in which 50% of members of three generations had speech and language difficulties, consistent with an autosomal dominant mode of transmission. Perinatal medical complications, including low birth weight or prematurity, are also associated with language delays, although some studies find no impact of prematurity on language development. The significance of prenatal complications such as preterm labor have not been extensively studied. In this case, the survey of risk factors offers limited insight into prognosis. Second-
degree relatives had apparent language delays, but no primary relatives (parents or siblings) suffer language disorders. Shelley’s mother experienced preterm labor, but Shelley was delivered without complications at term.

Developmental milestones in other domains have been shown to be very useful in determining prognosis for language development. Children between 18 and 24 months of age who function below the 10th percentile for receptive vocabulary but who are near the mean for expressive vocabulary have been shown to ultimately catch up with age-matched peers. Toddlers with expressive and receptive delays were found to fall farther behind over time and eventually to meet diagnostic criteria for specific language impairment. Mastery of the cognitive prerequisites for language development, such as imitation, intentionality, and symbolic thinking, are other good prognostic indicators. By history and observations, Shelley seemed to have age-appropriate receptive language skills, including recognition of pictures and body parts, and comprehension of two-part commands. Her interest in dolls suggests that she is capable of symbolic thinking. These developmental milestones put her in the good prognosis category. Moreover, language is primarily used for social communication, so this child’s age-appropriate social skills and nonverbal communication are additional signs of good prognosis.

The consensus in the field of psycholinguistics is that exposure to a bilingual environment offers no major disadvantages to children learning language, at least when studies control for the social class of the children. Children in bilingual environments proceed initially as if they have a single vocabulary, choosing one word from either language for each concept or referent they label. At the time of the language spurt, they begin to organize vocabulary words into separate language codes; early phrases and sentences usually contain words from only one language source. Language learning is facilitated when consistent environmental cues assist in this organization process, in particular when each caregiver consistently uses one language.Attributing the delay exclusively to bilingual input seems inappropriate in this case given the degree of delay and the relatively bilingual environment.

Assessment of a child’s language environment is appropriate when the child experiences delays in the absence of physical and neurological findings and risk factors. Though developmental psychologists debate the relative importance of the environment in language learning, all agree that language skills are not well learned from television, radio, or solitary play. Studies consistently show that children living in poverty acquire language more slowly than children from the middle class; the precise environmental factors that affect the rate of language development have not been clearly determined but may be either the quality or quantity of the input. Recent studies show that the rate of vocabulary growth correlates with the number of words parents use with their children. Children also seem to learn language more quickly when caregivers repeat and expand the child’s output rather than when adults initiate most of the conversation. In cases like this one, the clinician and parents together should assess the quality of the home environment in particular, (interactions of mother, father, and all other major care providers with the child) as another potential contributor to language delay.

Given that Shelley has no major risk factors and good prognosis for normal language functioning on the basis of the developmental evaluation, further workup can be deferred, at least briefly. However, close monitoring of development status and counseling about optimal environmental stimulation is essential. If Shelley fails to increase the rate of development over the next 4 months, then further workup is highly recommended. The most important diagnostic test would be a full audiological assessment, because hearing loss can account for about half of the cases of language delay in some series. General developmental assessment by qualified professionals would also be important because global developmental problems and mental retardation account for another large subset of cases. Clinicians should readily refer children with persistent language delays for early intervention. Early intervention for language delays and disorders has been shown to be quite effective. A child referred for early intervention who catches up to age mates can be discharged from services without harm. A child who could benefit from early intervention would be disadvantaged by delays in referral.

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REFERENCES

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Dr. Feldman reminds us of the clinical challenge language assessment in office practice when she states that “typical toddlers rarely display their linguistic skills in the office.” Relying on parental report
means that it is critical to learn the skills necessary to ask appropriate, open-ended, and focused questions. To gain an accurate assessment of a young child’s output and understanding of language, the Early Language Milestone Scale\(^1\) or the MacArthur Communicative Development Inventory,\(^2\) referred to by Drs. Coplan and Feldman, not only add a practical screening instrument to a pediatrician’s ability to screen for language disorders, but these instruments can also be used to learn to ask clinically appropriate questions to caregivers and children.

The differential diagnosis of a child with a language delay was addressed by Drs. Coplan and Parker. They excluded many specific and global causes of language delay by selective use of historical, family, and developmental data. The case presentation included information that made several diagnoses unlikely, including mental retardation, autism, and a severe hearing loss. Dr. Coplan noted that Shelley’s history and examination were consistent with the diagnosis of developmental language delay. He described this disorder as a heterogeneous group of conditions with a wide spectrum of expressive and receptive language impairment in the absence of other disorders that may affect language, including hearing loss, emotional disturbances, global cognitive delay, autism, and some physical disabilities. A developmental language disorder may start with delays only in expressive speech; with more precise diagnostic accuracy in preschool and school-age children, problems with receptive language, especially auditory processing and short-term memory impairment, may surface.

There is a consensus among the commentators that, at 20 months of age, Shelley’s isolated expressive language delay is significant and may either reflect a maturational delay (i.e., a benign and reversible form of developmental language delay) or may be the harbinger for a more protracted delay. Dr. Feldman’s reference to the fact that approximately 10 to 15% of children show substantial delays in language development at 2 years of age, whereas only 4 to 5% remain delayed beyond age 3 years, supports this observation.

The differences among the three clinicians in recommendations in light of a similar diagnostic formulation is striking. Dr. Coplan would perform an audiogram now and refer to a speech pathologist if Shelley’s language did not progress significantly by 24 months of age. Dr. Parker would also order an audiogram, while recognizing the low yield of a hearing loss in this clinical setting. He adds a caveat: a mild hearing loss can occasionally contribute to an isolated expressive language delay. Dr. Parker would refer to a language therapist at this point only if a child’s linguistic environment was inadequate.

Dr. Feldman provided some clues to assess language environment. Research has shown that the rate of vocabulary growth correlates with the number of words parents use with their children and that children learn language more quickly when caregivers repeat and expand the child’s output rather than when adults initiate conversation. Finally, Dr. Feldman would not perform either an audiogram or a full language assessment at this point. If the rate of language development did not increase by 24 months of age, she recommends both assessments.

That experienced clinicians have different approaches to clinical problems is found throughout medicine. The differences often are not based on available medical knowledge but rather on one’s practice experience. Dr. Coplan is a developmental pediatrician in an academic center where he evaluates children referred for a variety of developmental problems. Drs. Parker and Feldman, although both practice and teach in academic centers, have a primary care clinician’s perspective. Throughout medicine, the specialist is an expert in the numerator of disease frequency, and the generalist is the expert in the denominator! This difference, in part, is responsible for the different perspectives among the commentators. In addition, the primary care clinician typically works under the assumption that continuity of care is assured. The uncertain or equivocal diagnosis at one office visit can be followed up in a designated time interval with a degree of assurance. The specialist, who evaluates a child as a consultant, may not be comfortable withholding a diagnostic study or therapeutic intervention when continuity of care is less clear.

The case of Shelley exemplifies one of many challenges faced by pediatricians and other health care clinicians when monitoring for developmental variations and delays. The boundary between a normal variation and an irreversible delay is often not clear. The risk of inaccurate labeling must be balanced with the potential risk of delayed intervention. Both are real! They will not go away by either excessive use of “watchful waiting” or indiscriminate referrals for diagnostic studies and therapies.

Clinical experience, based on both research data and an effective synthesis of the medical history, physical examination, and developmental assessment, begins the process required for a reasoned decision.

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