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A Case Study of Early Development in Williams Syndrome: Implications for Early Intervention

Susan Hepburn, PhD, Amy Philofsky, MA, CCL-SLP, Angela John, BS, and Deborah J. Fidler, PhD

Department of Psychiatry, University of Colorado Health Science Center, Denver (Dr Hepburn and Mss Philofsky and John); and the Human Development & Family Studies, Colorado State University, Fort Collins (Dr Fidler).

Abstract

The aim of this article is to provide an in-depth description of the behavioral phenotype of Williams syndrome in a preschool-aged child. Williams syndrome is a neurodevelopmental, multisystem genetic disorder associated with mental retardation that predisposes individuals to a characteristic pattern of strengths and weaknesses in neuropsychological functioning. While much is known about functioning in adults, very few descriptions of early development are available in the literature. Implications for designing early intervention programs for children with this debilitating disorder are discussed.

Keywords

case study; early intervention; Williams syndrome

Williams syndrome (also called Williams-Beuren syndrome) is a neurodevelopmental, multisystem disorder associated with mental retardation and linked with an abnormality on chromosome 7 (Ewart et al., 1993). Prevalence of Williams syndrome is approximately 1 in every 25,000 live births (Bellugi, Lichtenberger, Jones, Lai, & St. George, 2000). Abnormalities are noted in several aspects of development: medical, cognitive, language, social, and emotional-behavioral and will be briefly outlined below.

MEDICAL

During early development, individuals with Williams syndrome are at particular risk for certain serious physical conditions. Infants are often small for gestational age and show intrauterine growth retardation (Huang, Sadler, O'Riordan, & Robin, 2002). Williams syndrome is also termed Infantile Hypercalcaemia and many infants are treated with low calcium and vitamin D diets (Arnold, Yule, & Martin, 1985). Vomiting, gastroesophageal reflux, and excessive irritability are common in infancy as well (Francke, 1999). Beyond infancy, physical characteristics associated with Williams syndrome include cardiovascular complications, a unique craniofacial appearance, and hyperacusis (Bellugi et al., 2000).

COGNITION

Many individuals with Williams syndrome demonstrate cognitive impairments, usually within the range of moderate mental retardation; however, substantial within-group variability can be observed. Studies have reported IQ ranges between 40 and 90 on the WISC-R (Udwin, Yule, & Martin, 1987) and the Leiter International Performance Scale (Volterra, Capirci, Pezzini, Sbbadini, & Vicari, 1996). Rather than showing a flat profile of generally impaired functioning, individuals with Williams syndrome show a mixed profile of relative strengths and weaknesses in cognition. Verbal processing is an area of strength relative to nonverbal processing, although verbal IQ scores generally lag behind chronological age (CA) (Bellugi, Wang, & Jernigan, 1994; Don, Schellenberg, & Rourke, 1999; Morris & Mervis, 1999). Visuospatial processing tends to be an area of relative weakness in Williams syndrome (Mervis et al., 2000; Pezzini, Vicari, Volterra, Milani, & Ossella, 1999; Udwin & Yule, 1991). In particular, tasks that demand visuoconstructive abilities have been shown to be particularly impaired. On the test of Visual-Motor Integration (VMI), children with Williams syndrome perform significantly worse than CA- and IQ-matched adolescents and young adults, and mental age (MA)-matched typically developing children (Bertrand, Mervis, & Eisenberg, 1997; Wang, Doherty, Rourke, & Bellugi, 1995). According to Mervis (1999), performance in the 20th percentile or below on pattern construction tasks may be a useful diagnostic feature in Williams syndrome.

Amidst these deficits, there are islands of relatively spared visuospatial functioning in individuals with Williams syndrome that relate to sociability. While many studies have shown poor visuoconstructive abilities in Williams syndrome, others have found that children with Williams syndrome show relatively higher Draw-A-Person scores than VMI scores. In fact, Draw-A-Person scores have been found to be comparable to those of children with Down syndrome, who tend to show significantly better visuospatial skills than children with Williams syndrome (Dykens, Rosner, & Ly, 2001). These findings may tie into relative strengths in sociability in Williams syndrome, which will be discussed below. In addition, while visuospatial memory may be weak, individuals with Williams syndrome have strong memory for familiar and unfamiliar faces (Rossen, Klima, Bellugi, Bihle, Jones, 1996). Indeed, individuals with Williams syndrome have shown near-CA levels of performance on the Benton Test of Facial Recognition, even while unable to even pass the pretest of the Benton Test of Line Orientation (Bellugi et al., 2000). Contrary to initial reports, recent findings suggest that children with Williams syndrome use neurocognitive mechanisms for perceiving faces that are similar to those used by typically developing individuals (Tager-Flusberg, Plesa-Skwerer, Faja, & Joseph, 2003).

LANGUAGE

Initially thought to be a “spared” area of functioning, language in Williams syndrome is unevenly developed. Strengths include good recall of previously heard words and phrases (Udwin & Yule, 1990), an unusual, sophisticated diction compared to other children (Bellugi et al., 2000), socially engaging use of prosody, discourse, and narrative skills (Bellugi, Lichtenberger, Mills, Galaburda, & Korenberg, 1999), and stronger expressive morphosyntactic skills, compared to other syndromes of mental retardation (Bellugi et al.,

1994; Karmiloff-Smith, 1998; Volterra et al., 1996). Weaknesses in language usually emerge as tasks become more complex (Don et al., 1999; Klein & Mervis, 1999; Rossen et al., 1996). It has also been suggested that children with Williams syndrome may not understand everything that they express, as evidenced by use of words or phrases that may be slightly unconventional (Bellugi et al., 2000).

Closer examination of linguistic abilities in school-age and adolescent children with Williams syndrome has revealed a general pattern of peaks and valleys within the verbal domain. Among the peaks, children are reported to have a strong verbal working memory compared to other syndromes of mental retardation (Jarrold, Baddeley, & Hewes, 1998; Klein & Mervis, 1999; Wang & Bellugi, 1994). Researchers have found conflicting results with regard to receptive vocabulary, with some arguing its relative strength (Don et al., 1999; Jarrold et al., 1998) and others arguing the contrary (Klein & Mervis, 1999). Some studies have revealed unusual semantic organization in individuals with Williams syndrome (Rossen et al., 1996). Individuals with Williams syndrome are said to do well on simple linguistic tasks (eg, auditory closure, receptive vocabulary, and word association to category), demonstrating increasing difficulty with tasks of increasing linguistic complexity (eg, sentence memory and comprehension on the WISC-III; Don et al., 1999).

Children with Williams syndrome also show a great proclivity for using language in social ways. Individuals with Williams syndrome are described as loquacious, articulate, facile communicators who enjoy telling dramatic stories with a penchant for capturing their listener's ear. During narratives, individuals with Williams syndrome have been shown to use a great deal of "social evaluation," so as to grade a listener's attention (Reilly, Klima, & Bellugi, 1990). Individuals with Williams syndrome frequently elaborate a story to make inferences about the mental states of the characters of the story, and they also use attentioneliciting devices whose sole purpose is to engage the listener (Reilly et al., 1990).

SOCIAL

Excessive interest in others and a distinct lack of inhibition with regard to approaching others in social contexts are behavioral hallmarks in this population (Cones et al., 2000). Therefore, these children are often described as "hypersocial." Individuals with Williams syndrome also seem to be especially drawn to social stimuli, as researchers have noted high rates of looking at caregivers' or playmates' faces in school-aged population (Mervis, 1999). Strengths in interpreting social signals have been found on eye gaze direction interpretation tasks (Karmiloff-Smith, Klima, Bellugi, Grant, & Baron-Cohen, 1995). Stranger anxiety is frequently absent during early development (Mervis, 1999), with children showing less frequent negative facial expression, and lower intensity of facial and vocal distress during laboratory situations (Bellugi et al., 2000). In addition, individuals with Williams syndrome give abnormally positive approachability ratings to unfamiliar people compared to CA- and MA-matched controls (Jones et al., 2000), and some studies have described increased empathic responses (Gosch & Pankau, 1997). It has been hypothesized that the tendency toward hypersociability (ie, increased social approach, low disinhibition, increased empathy) may predispose these individuals to be socially vulnerable (Semel & Rosner, 2003).

Researchers have attempted a number of social cognition tasks (eg, theory of mind) with individuals with Williams syndrome and have found some interesting results. These tasks measure one's ability to understand what another person is thinking (Karmiloff-Smith et al., 1995; Mervis & Klein-Tasman, 2000; Tager-Flusberg, Boshart, & Baron-Cohen, 1998; Sullivan & Tager-Flusberg, 1999). Early in the pursuit of research on theory of mind in Williams syndrome, researchers suggested a relative sparing of this ability (Karmiloff-Smith et al., 1995; Sullivan & Tager-Flusberg, 1996). More research, however, illuminated that individuals with Williams syndrome, in fact, have difficulty with higher-level tasks to consider theory of mind (Sullivan & Tager-Flusberg, 1999). Sullivan and Tager-Flusberg (1999) argued that theory of mind in Williams syndrome is complex. They concluded that in individuals with Williams syndrome, the social-perceptual components of theory of mind (eg, ability to make quick judgments of others' mental states on the basis of their expressions and affect) remain relatively preserved, while the social-cognitive components (eg, conceptual understanding of others' minds as containing potentially different thoughts about matters) are not (as reviewed in Mervis & Klein-Tasman, 2000).

EMOTIONAL-BEHAVIORAL

A small but growing literature is emerging on psychopathology in Williams syndrome. Studies have suggested difficulties with anxiety, concentration problems, and obsessions and preoccupations in this population (Davies, Udwin, & Howlin, 1998; Dykens & Rosner, 1999; Einfeld, Tonge, Rees, 2001; Sarimski, 1997; Udwin, Howlin, Davies, & Mannion, 1998). Children and adolescents with Williams syndrome are more likely to have difficulties with anxiety compared to children with other mental retardation syndromes (Einfeld et al., 2001; Sarimski, 1997). In addition, children and adolescents with Williams syndrome show significantly more fears than MA-matched children with mental retardation of mixed etiology (Einfeld, Tonge, & Florio, 1997), and a matched group of individuals with Prader-Willi syndrome (Dykens & Rosner, 1999). In addition, Udwin et al. (1998) report that two thirds of their sample of individuals with Williams syndrome were described as restless, distractible, irritable, anxious, and fearful.

DEVELOPMENTAL CONSIDERATIONS

Although there is a wealth of new research describing the behavioral phenotype associated with Williams syndrome, most of this research is based on older children, adolescents, and young adults. However, researchers have begun to recognize the developmental process that leads to these pronounced outcomes in older individuals with genetic disorders. Karmiloff-Smith (1998) argues that genetic disorders predispose children to "tiny variations in initial state" that become magnified throughout development into domains of relative strength and weakness. Therefore, early development may be a crucial window of opportunity for intervention, as these "tiny variations" have not yet snowballed into impairments in whole domains of processing.

At this point, very little is known about how and when the Williams syndrome behavioral phenotype emerges in the first few years of life. We present the case study of Jenny, a 3-year-old girl with Williams syndrome, so as to shed light on how the Williams syndrome

behavioral phenotype may present in some young children. We also present ideas for how early manifestations of the Williams syndrome behavioral phenotype may be ameliorated through time-sensitive and targeted interventions.

CASE STUDY: JENNY

Jenny (fictitious name), a 38-month-old girl, was referred to a research study on early development in Williams syndrome through a local pediatrician. Diagnosis was made via FISH (Fluorescent In Situ Hybridization) testing 2 months prior to referral. She is the first-born child of a married couple, lives at home, and is receiving home-based and center-based intervention. Prior to her involvement in this research study, Jenny had not participated in any standardized tests of her development.

Described by her parents as a healthy child, Jenny was born at 40 weeks' gestation, following an uncomplicated pregnancy and delivery. She was a small infant (weight and height both within the 5th percentile). A heart murmur was detected, requiring monitoring, but no treatment. She breast-fed very well, although she was described as "finicky" with regard to prepared formula. She had no significant history of illnesses and was on no medications.

Her parents first became concerned about Jenny's development around the age of 15 months, when they noticed that her physical development was "slow." She rolled over at 7 months, sat alone without support at approximately 8 months, crawled at 11 months, and walked at 22 months. Jenny also seemed withdrawn from interactions with other children and would watch them play instead of joining. Despite this social reticence with other children, they also observed hypersociability with adults (eg, overly friendly style, lack of discrimination of familiar vs unfamiliar).

Language development was reported to be somewhat delayed. First words were reported to be emerging at 18 months, however; while she was babbling and directing her vocalizations to adults frequently, she was sometimes difficult to understand and tended to intermingle babble with true words. Subsequent to her use of first words, language development was reported to happen slowly. An observational evaluation at the age of 34 months suggested a profile of stronger expressive (approximately 27 months) than receptive language (scattered abilities between 18 and 27 months). At that time, echolalia predominated Jenny's expression; however, spontaneous phrase speech was newly emerging.

Jenny's parents described her as curious and happy most of the time. Disciplining Jenny is difficult for them, partly because she laughs when she is being reprimanded.

Results of developmental evaluation

Standardized testing was conducted in Jenny's home over a period of 3 days by 2 experienced examiners (clinical psychologist and a speech-language pathologist [SH and AP, respectively]). Testing sessions were up to 2 hours in length and were videotaped. Jenny was responsive to the adults and eager to participate in all testing activities. She was highly distractible during testing (particularly during visual-spatial and receptive language

activities). Frequent reinforcement (eg, play with an adult) was applied to maintain her attention and effort throughout the battery.

Overall developmental profile

Two general tests of overall development were administered in a standardized manner. The Mullen Scales of Early Learning (MSEL; Mullen, 1995) provides standard scores and age equivalent estimations on 4 domains of early development: visual reception, fine motor, receptive language, and expressive language. Please see Table 1 for a summary of her results.

Jenny obtained scores within the low end of the normal limits for expressive language, with relative weaknesses in receptive language, visual-spatial skills, and, most notably, fine motor development.

The Vineland Adaptive Behavior Scales (VABS; Sparrow, Balla, & Cicchetti, 1984), a parent interview measuring social, communication, independence in daily living, and motor development was also administered in a standardized manner by an educational psychologist (DF) (Table 2). Of note, Jenny obtained scores within the low end of the normal limits for socialization with slightly lower scores in the area of communication. She presented with relative weaknesses in daily living skills and motor skills. Overall, parent report was consistent with direct assessment of Jenny's abilities, as she presents with a moderate developmental delay across skills, with a strength in expressive (but not receptive) language and significant impairments in motor development and nonverbal problem solving.

Language

In addition to the MSEL, the Preschool Language Scale-4 (PLS-4; Zimmerman, Steiner, & Pond, 1992) was also administered by a speech-language pathologist. The PLS-4 provides a more precise, domain-specific assessment of language development than the MSEL (Table 3). Consistent with the MSEL, Jenny demonstrated significantly stronger expressive than receptive skills on the PLS-4. Interestingly, Jenny's receptive language skills are more closely linked to her nonverbal problem-solving skills than her expressive skills. This finding is clinically very significant, as most adults may assume that a child's language understanding is at the same developmental level as his or her expressive ability and, clearly, this is not the case with this particular child.

Play

According to Jenny's parents, her play with toys is limited and it has been difficult for her parents to engage her in functional play activities. The Fewell Play Scales (Fewell, 1992) were administered by a trained examiner in 2 sessions. This assessment involves providing free access to several developmentally sequenced play sets and observing the child's spontaneous play acts with toys. After ample time for eliciting spontaneous play, the examiner then provides a hierarchy of prompts (eg, verbal, modeling, verbal and modeling) so as to elicit more play. An age equivalent range is derived from the spontaneous acts of the child and the prompted acts.

During the assessment, Jenny moved quickly from toy to toy, with a limited focus on any one activity. She obtained an overall developmental age of 11 to 14 months, which is consistent with her overall scores on the MSEL and the VABS. Given clear modeling prompts, Jenny was able to imitate the examiner's actions with toys and obtained a prompted play score in the 27- to 30-month range. While she was only demonstrating one-step pretend play sequences spontaneously (eg, stirring in a cup, feeding a doll), she was able to sequence 2-step pretend play sequences following a model (eg, stir in cup, then feed baby; feed baby bottle, then burp her). This finding suggests that adult modeling and teaching through imitation is a potentially fruitful intervention for Jenny.

Attention

Jenny presented as easily distractible, particularly to social stimuli. She delighted in being clapped for and joined by an audience of adults; however, such situations proved so distracting that Jenny readily went off-task by joining into the clapping and directing smiles and pleasure to every face in the room. Furthermore, when difficult tasks were presented to Jenny, she often sought social interactions in an effort to escape. Even without social distractions, her focus to tasks was generally brief; however, she was also easily redirected back to the task.

Social

Jenny participated in several experimental tasks that tap sharing affect, sharing attention, demonstrating empathy, and social relatedness (responsiveness, initiations, reciprocity). Tasks were taken from the literature on social development in typical and atypical children and are described elsewhere. These tasks were as follows: the Early Social and Communication Scales (Seibert, Mundy, & Hogan, 1982), Yummy-Yucky Task (Repacholi & Gopnik, 1997), Empathy battery (adapted from Sigman, Kasari, Kwon, & Yirmiya, 1992), Surprise Boxes (Repacholi, 1998), and the Autism Diagnostic Observation Schedule (Lord, Rutter, DiLavore, & Risi, 1999) (Note: we chose to do this assessment not as a diagnostic tool for autism, but for the social and communicative presses provided by the instrument).

Several consistent observations of Jenny's social style were observed across these tasks and activities. First, Jenny was unusually responsive to changes in facial expression and mood, demonstrating extreme emotional contagion. For example, if an adult demonstrated excitement, Jenny mirrored the affect in an exaggerated way and became overaroused and difficult to calm. This emotional contagion was also observed in negative affects. If the adult frowned or mimicked sadness in the presence of ambiguous stimuli, Jenny adopted the sad expression and became extremely upset (eg, crying inconsolably), even in the absence of any salient stimuli. If the adult seemed to be injured (as in the empathy tasks), Jenny expressed concern in an exaggerated manner. Remarkably, her affect shifted almost immediately when the adult's affect changed. Thus, a pattern of extreme mood lability in response to modeled affects was noted across tasks.

Jenny's social initiations were usually comprised of well-integrated verbal and nonverbal behaviors. For example, she frequently looked directly at an adult, verbalized her requests (eg, "I want doll now"), and pointed to the desired toy. She alternated gaze effectively with

her requests and frequently directed the adult's attention to toys and events in the room. She frequently showed objects to others and shared her enjoyment by directing her facial expressions and her vocalizations to adults. Although sometimes it was difficult to get her attention, she was able to follow an adult's redirection of attention fairly well.

In terms of prosody and intonation (which are socially learned aspects of verbal communication), Jenny applied affective meaning to her statements quite effectively. Even when her words were unclear, the affective meaning was communicated via natural changes in tone and rhythm of speech.

Temperament

Both of Jenny's parents contributed to the completion of the Carey Temperament Scales (Toddler Temperament Edition; Carey, 1998). Overall, Jenny was reported to present with an easy-going behavioral style, characterized by a primarily positive mood, high interest in novelty, adaptability with changes, moderate emotional intensity, and relatively low activity level. Most notable in her profile were extremely high scores (ie, beyond 2 standard deviations from mean) indicating nonpersistence, or a tendency to avoid challenges. This behavior was evident throughout her testing sessions, as well. She also was reported to be somewhat indiscriminate in her approach behaviors—as she is drawn to novelty and social engagement much more often than most of her peers. Jenny was also reported to be extremely unpredictable, or arrhythmic, in her daily habits and routines (such as sleeping, eating, eliminating).

IMPLICATIONS FOR INTERVENTION

Given the findings of this comprehensive developmental assessment, several implications for early intervention for this particular child seem relevant. Considering that any case study lacks generalizability, we are not proposing that every child with Williams syndrome will present with this profile or benefit from these intervention approaches; however, Jenny's profile does seem consistent with an early manifestation of the later phenotype that has been so well documented in the literature (Mervis, 1999). The overall philosophy of designing her early intervention program involves simultaneously targeting her weak skills (eg, receptive language, fine motor, daily living skills), while utilizing and encouraging her strengths (eg, imitation, emotional contagion, social interest, expressive language).

Thus, our first consideration for intervention involves consideration of Jenny's split in language abilities. She tends to mask her difficulties in language understanding and overall cognition through good social interaction and expressive language skills. Adults often base their interaction style on the child's expressive language ability and one of the first steps in Jenny's educational programming is to encourage all teachers and therapists to interact with her at the level of her receptive and nonverbal problem-solving (eg, 26 months). Cognitive goals should also be at this age level.

Accommodations should be made in Jenny's educational environment to address her difficulties in receptive language. While our assessment does not definitively lead to the diagnosis of an auditory processing disorder, Jenny is showing a language and attentional

profile similar to a child with an auditory processing disorder, and interventions designed for this disability may prove to be beneficial. Such accommodations include the following: (1) presenting information to Jenny verbally and visually (ie, this could include gestures, facial expressions, simple pictures, etc); (2) ensuring that Jenny is paying attention to directions by calling her name and touching her; (3) breaking directions into smaller parts; (4) chunking together similar bits of information; (5) using hands-on learning and modeling to teach new skills; and (6) choosing classroom seating that minimizes sound distractions such as fans, heating vents, or the door.

Jenny's social skills with adults are a relative strength; however, her ability to interact well with other children is limited. There are several implications to this profile. First, it will be helpful to use social interactions with adults as motivators for completing difficult tasks (ie, puzzles, toy play). Adults should be careful, however, of not allowing Jenny to avoid difficult tasks by retreating into social interactions. A work-play routine (ie, finish this motor task, then we will play), with limited verbal interactions during work and lots of rich social interaction following the completion of work, may be most helpful in supporting attention to difficult tasks.

Second, adult facilitation in play activities with other children will be essential for teaching her how to appropriately initiate and maintain a spontaneous interaction with other children. Relying upon her strengths in learning through imitation, 2 possibilities emerge: (1) encouraging her to model the acts of an adult; or—and this is probably the most developmentally appropriate and effective strategy—(2) providing a peer model and encouraging her to imitate the other child. These strategies are well elucidated in the work of Odom (2000), Goldstein and Kaczmarek (2001), Shuler and Wolfberg (2000), and Strain (2002). In addition, pretend play schemes can also be taught through social imitation. Developing play schemas will be helpful for creating a context for social interaction with peers.

Another area of potential focus for early intervention is building attentional skills and persistence, which are critical for succeeding in a classroom environment. Structured teaching strategies developed by the TEACCH program (Schopler, Mesibov, & Hersey, 1995) may be very helpful in encouraging persistence. Ideas include the following: (1) using a visual and verbal schedule to outline the order of activities and expectations; (2) using visual and verbal reminders of how much work is expected (eg, saying “1 more minute” or coloring in a circle to indicate that she is expected to work until the circle is completed); (3) using a finished box and visually structured tasks such that when all of the task pieces are gone, the task is over; (4) starting with brief work periods, followed by rewarding social interactions and gradually increasing the duration of work; and (5) employing errorless learning strategies in difficult activities (eg, providing enough prompting and structure so that the child practices completing the task correctly).

In summary, this case study provides an example of how a young child with Williams syndrome may already be demonstrating the characteristic pattern of strengths and weaknesses observed and reported in older individuals with the disorder. It is important to emphasize that any case study has limited generalizability and future studies will clarify

whether the profile of strengths and weaknesses observed in Jenny are representative of those presented by a large proportion of children with Williams syndrome. Of interest to early interventionists is the notion that perhaps early intervention strategies that target this complex mix of abilities and deficits can alter the developmental trajectory of the disorder. Only longitudinal studies that clearly describe intervention histories and initial child characteristics can address this issue scientifically, but case studies such as the one described herein provide a descriptive starting point for early interventionists committed to employing child-specific strategies designed to improve child functioning.

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Table 1

Results from the Mullens Scales of Early Learning at the age of 38 months

Domain	<i>T</i> score *	Age equivalent, mo	Descriptive category
Visual reception	29	27	Very low
Fine motor	20	24	Very low
Receptive language	34	28	Below average
Expressive language	40	32	Average
Overall learning composite	65 [†]	28	Very low

* Mean = 50 ± 10.

[†] Mean = 100 ± 15.

Table 2

Results on the Vineland Adaptive Behavior composite

Domain	Standard score *	Age equivalent, mo
Communication	83	27
Daily living skills	60	18
Socialization	94	34
Motor skills	59	18
Adaptive behaviour composite	59	Not applicable

* Mean = 100 ± 15.

Table 3

Results on the Preschool Language Scales—Version 4

Domain	Standard score [*]	Age equivalent
Auditory comprehension	61	2 y, 2 mo
Expressive communication	99	2 y, 11 mo
Total language score	78	2 y, 5 mo

* Mean = 100 ± 15.