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Speech and Language Disorders Associated with Prader-Willi Syndrome

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The speech and language skills of individuals with Prader-Willi syndrome (PWS) differ greatly in the severity and type of deficits that they present, ranging from individuals who are nonverbal to those who acquire normal speech and language skills by adulthood. Due to the low incidence of the disorder—1 in 10,000 to 15,000 individuals[4,5]—professionals such as speech-language pathologists, physical therapists, and occupational therapists may encounter only a few individuals with Prader-Willi syndrome in their practice. An understanding of the characteristics of Prader-Willi syndrome that may impact speech and language abilities will allow the professional to evaluate each individual for potential contributing factors to communication deficits and to plan appropriate intervention strategies. The goals of this chapter are to summarize the existing literature on the speech and language skills of those with PWS, to describe features of PWS that may contribute to speech and language disorders, to chart a developmental course of the speech and language skills, and to suggest possible intervention strategies.

Speech and Language Characteristics

The speech and language skills of individuals with Prader-Willi syndrome are reported to be below expectations based on intellectual levels.[3,11,14,18] Although great variability exists in the speech and language skills of individuals with Prader-Willi syndrome, several common features have been noted. These include poor speech-sound development, reduced oral motor skills, and language deficits. Speech is often characterized by imprecise articulation, hypernasality, flat intonation patterns, an abnormal pitch, and a harsh voice quality. Prosody, or the melody of speech, may also be disrupted. A slow rate of speech, typical of a flaccid dysarthria, may be observed. In addition to these speech difficulties, the individual with Prader-Willi syndrome may have language problems. Language problems include deficits in vocabulary, grammar, morphology, narrative abilities, and pragmatics. Table 9.1 illustrates the clinical features of PWS and their potential

Table 9.1. Clinical Characteristics of Prader-Willi Syndrome and Potential Impact on Speech and Language

Clinical Feature	Impact on Speech/Language
Mouth	
• narrow overjet • narrow palatal arch • micrognathia	→ reduced articulatory skills
Larynx	
• altered growth due to endocrine dysfunction	→ pitch variations
Dentition	
• decay due to reduced saliva output and enamel hypoplasia	→ reduced articulatory skills
Hypotonia	
• poor velopharyngeal movement	→ hypernasality or hyponasality
• stretching of muscles of larynx	→ vocal pitch and quality variations
• slow movement of articulators	→ imprecise articulation and slow rate of speech
Cognitive	
• mental retardation • sequencing problems	→ delayed receptive/expressive language poor narrative skills
Behavioral Disturbances	
• temper tantrums • stubbornness • manipulative behavior • depression • emotional lability • compulsive behavior • skin picking • difficulty relating to peers • poor social relationships • difficulty detecting social cues • argumentative	→ poor pragmatic skills

Source: B. A. Lewis, L. Freebairn, S. Heeger, and S. B. Cassidy, "Speech and Language Skills of Individuals with Prader-Willi Syndrome," *American Journal of Speech-Language Pathology,* 2002;11:285–294.[16] Copyright by the American Speech-Language-Hearing Association. Reprinted with permission.

impact on speech and language abilities. The following section will summarize findings of each speech and language domain.

Speech-Sound Development

A distinguishing feature of PWS is poor speech-sound development.[10] Speech-sound disorders include both errors of articulation or phonetic structure (errors due to poor motor abilities associated with the production of speech-sounds) and phonological errors (errors in applying linguistic rules to combine sounds to form words). Individuals with PWS may exhibit deficits in either articulation or phonology or both. Several factors may account for the poor speech-sound development of individuals with PWS, including oral structure abnormalities, abnormal saliva,[12,23] hypotonia, poor phonological skills, and cognitive

deficits. Reduced breath support for speech may also be noted. Oral structures of the mouth and jaw that may impact articulation skills include a narrow overjet, a narrow palatal arch, and micrognathia. However, it is more likely that poor oral motor skills, especially reduced tongue elevation for speech and slower alternating movements of the articulators, account for poor speech-sound skills in PWS.[1,3,14] Speech characteristics are often similar to those reported for flaccid dysarthria.[14] Speech-sound errors that have been noted in individuals with PWS include sound distortions and omissions, vowel errors, simplification of consonant blends, and difficulty sequencing syllables. Phonemes that are motorically complex, such as /s/, /r/, /sh/, and blends are usually the most difficult.[14] As with most speech-sound disorders, single word utterances are often more intelligible than conversational speech, and the phonetic environment of the target sound can greatly influence speech intelligibility. For example, an individual might have difficulty producing the "r" sound in the word *crab* when it's preceded by the "qu" sound in the phrase "quiet crab's claws."

Other authors[10] have postulated that poor speech-sound development is the result of poor phonology skills, a component of a more general language deficit in PWS. Downey and Knutson[8] report that most individuals with PWS present with delayed speech-sound development characterized by phonological patterns typical of younger, normally developing children. However, some individuals demonstrate atypical patterns such as a phonological disorder or an apraxia of speech.[18]

Apraxia of speech is a severe speech-sound disorder that includes impairments in syllable sequencing, prosody, and speech-sound characteristics. Although the etiology of apraxia of speech is not well understood, it is presumed to result from impairment in the motor programming aspects of speech-sound production.[20] Rare cases of apraxia of speech have been reported in individuals with PWS. Children with apraxia of speech often do not develop intelligible speech until well into school age. Augmentative/alternative communication intervention (AAC—e.g., sign language and communication boards) may be employed to eliminate some of the frustration that the individual experiences in communication. AAC systems allow the individual to build vocabulary and pragmatic language skills while oral speech skills are developing.

Voice Characteristics

The voice of the individual with PWS may differ in pitch, quality, intensity, and resonance from that expected for his/her age and gender. Voice characteristics reported for individuals with PWS include a high-pitched voice, harsh/hoarse voice quality, inadequate vocal intensity, and hypernasality.[1,8] Hypotonia and altered growth of the larynx may result in a pitch that is too high or low. Nasal resonance may be disrupted by sluggish velopharyngeal movement and/or inadequate velopharyngeal closure potentially due to hypotonia. Poor velopharyngeal functioning may result in hypernasality, nasal emission, nasal

snorting, weak plosive consonant sounds, and unusual manner of sound productions. Although hypernasality is most frequently reported in individuals with PWS, hyponasality has also been noted.[16] Growth hormone therapy, sometimes utilized with PWS individuals, may also affect voice characteristics.[15] Surgical procedures such as those employed for children with cleft palates (e.g., a pharyngeal flap) may reduce hypernasality and improve speech intelligibility. It should be noted, however, that often speech-sound errors persist even though hypernasality has been reduced.

Fluency

Fluency disorders (stuttering and cluttering) are not frequently observed in individuals with PWS.[8] However, to date, a systematic study of the fluency and prosody characteristics of children with PWS has not been reported. Clinical observations of conversational speech suggest that interjections, revisions, and word repetitions may be related to cognitive and language deficits.[14] A slow rate due to poor oral motor skills and a monotone may disrupt the flow and melody of speech.

Language Skills

Individuals with PWS frequently demonstrate poor receptive and expressive language skills, with expressive language more impaired than receptive skills.[3,14,18] Analysis of conversational speech samples indicates that individuals with PWS employ a shorter mean length of utterance (MLU) than their peers. Several authors have described patterns of cognitive strengths and weaknesses frequently observed in PWS that might impact language abilities.[9] Specific deficits have been reported in auditory short-term memory,[9] linear or temporal order processing, and auditory verbal processing skills.[7] Poor speech-sound development may also affect language skills. For example, the acquisition of grammatical markers (morphology) may be delayed both because the child cannot produce the /s/ phoneme to form the plural and because the child does not understand the concept of plurals.[8]

Few studies have examined narrative skills of individuals with PWS. Narratives are accounts of events either real or imaginary. Narratives include storytelling, scripts, schema, and episodic memory. Narratives contain a chronological sequence of events and causal relationships. For an individual to use narratives successfully, he/she must form a topic-centered story, use specific vocabulary, sequence events within the story, describe relationships between people and events, and use correct story grammar. Story grammar includes a setting, a beginning, reaction, goal, attempt, outcome, and ending. Narrative skills are essential to social development as they promote conversational skills. In addition, good narrative skills are necessary for academic success as they promote reading and writing, develop organizational skills, and build linguistic abilities.

Clinical observation suggests that both children and adults with PWS have great difficulty with story retelling tasks. One study[17] examined the narrative abilities of 19 individuals with PWS, ages 3 to 30

years. A simple story entitled "The Fox and Bear" was read, and the individual was asked to retell the story. Participants showed deficits in recalling grammar elements and content items and had difficulty answering both factual and inferential questions about the story. While poor language skills may account for some difficulty with narratives, deficits in other cognitive skills such as temporal sequencing abilities, auditory short-term memory, and poor auditory processing skills may also contribute.[7,9] While narrative skills appear to develop into adulthood, the narrative abilities of the individual with PWS lag behind other language skills. Poor narrative skills may contribute to deficient conversational skills in adolescents and adults with PWS and thus impact social and job-related communication skills.

Pragmatic deficits, including problems with maintaining a topic, judging appropriate proximity to the conversational partner, and turn taking, have also been observed.[8] Pragmatic skills may be influenced by a number of behavioral disturbances frequently noted in individuals with PWS.[22] For example, temper tantrums, compulsive behavior, and skin picking may interfere with peer relationships. Children with PWS have difficulty with social relationships. Sullivan and Tager-Flusberg[21] demonstrated that children with PWS were less likely than IQ-matched children with Williams syndrome to show appropriate empathetic responses. A study of 30 adults with PWS reported perseverative speech.[6] Such pragmatic deficits may impede progress in therapy. Pragmatic language skills may vary by activity, routine, and environment. In adulthood, poor pragmatic language skills may create difficulties in the workplace and with interpersonal relationships.

Written Language Skills

Surprisingly, despite oral language deficits, individuals with PWS show relative strengths in written language skills. Strengths associated with PWS include vocabulary knowledge and reading decoding (i.e., sounding out words).[9] However, some individuals may present with poor reading comprehension skills possibly due to language deficits as described above. Visual spatial skills that have been reported as a relative strength[9] may also contribute to good reading decoding ability. However, variability has been noted in these patterns of strengths and weaknesses. Curfs, Wiegers, Sommers, Borghgraef, and Fryns[7] reported that 10 of 26 subjects with PWS had performance IQs at least 15 points higher than their verbal IQs, 3 had verbal IQs at least 15 points higher than their performance IQs, and 13 subjects did not show any discrepancy. In summary, individuals with PWS present with speech, language, and cognitive deficits that impede their communication skills.

Developmental Course of Speech and Language Skills in PWS

The speech-language pathologist may become involved with the child with PWS soon after birth. In infancy, children with PWS present with a weak cry and early feeding difficulties most likely due to

hypotonia. Reduced babbling and signs of early language delay are often observed.

All children begin acquiring word understanding essentially from birth (receptive language). Among typically developing children, expressive language follows soon after with cooing at 3 months, babbling at 6 months, and consonants in the form of "dada" and "mama" at around 8 months. For most, by 10 months of age, "dada" and "mama" are used discriminately, and there is evidence that the child understands the meaning of the word "no"; by 12 months of age, most children have acquired at least two words in addition to "dada" and "mama."

By contrast, children with PWS are 18 months of age before they begin to verbally evidence a vocabulary, combine words, and develop early syntax. A substantial number of affected children are much later in acquiring speech; some may be as late as 6 years of age. Oral motor skills remain poor and children exhibit many speech-sound errors that result in unintelligible speech. Pragmatic difficulties may be noted due to poor social skills and the emergence of behavioral disturbances. If the child's speech is highly unintelligible, AAC may be considered, including sign language or communication boards. AAC is usually transitional until oral speech abilities improve, and it alleviates some of the frustration that the child and caretaker may experience.

At school age (6 to 12 years), children with PWS are usually enrolled in speech and language therapy through the school. Articulation errors remain, with children less intelligible in connected speech than in single words. Receptive and expressive language skills lag behind those of their peers. Voice problems, as described above, may be observed as the child produces longer utterances. As noted previously, reading decoding emerges as a relative strength and children often become fluent readers. However, language difficulties may result in poor reading comprehension.

In adolescence and adulthood, the individual with PWS may continue to demonstrate communication difficulties including residual articulation errors, vocabulary deficits, poor conversational and pragmatic skills, and inappropriate pitch. Some individuals with PWS do achieve normal articulation skills. The individual with PWS may exhibit behavioral traits that are disruptive to good communication skills such as inappropriate laughter. Continued work on conversational speech is essential to adjustment and success in the workplace. Emphasis should be placed on functional language skills and life-skills training.

Summary of a Clinical Research Study

Previous research on the speech and language skills of individuals with PWS has been based on a small number of individuals and has not examined the developmental course of the speech and language disorder. Many studies have not distinguished between speech-sound errors due to poor oral motor skills and structural deviations and errors due to phonological deficits. Further, studies have not attempted to associate speech and language characteristics to a particular chromosome 15

abnormality (i.e., paternal deletion, uniparental disomy, or a translocation). A study of the speech and language abilities of a relatively large cohort of individuals with PWS representing three age groups (infant/preschool, school-age, and adolescent/adult) was undertaken. The details of this study are reported elsewhere (see Lewis, Freebairn, Heeger, and Cassidy[16]). The findings of this study are summarized below to illustrate the variability and range of speech and language skills found in individuals with PWS.

Participants

The participants were 32 individuals (16 males and 16 females), ages 6 months to 42 years. All met the diagnostic criteria for PWS,[13] and diagnoses were confirmed by chromosomal analysis.

Measures

Measures were selected to assess oral motor skills, articulation and phonology, receptive and expressive language skills, prosody/voice characteristics, reading, and narrative abilities. Assessments included standardized tests as well as a spontaneous speech sample analysis. Standardized tests varied according to the age and intellectual abilities of the individual.

A rating scale was adopted to summarize data across various age and skill levels. The speech and language skills of each subject were rated independently by two licensed and certified speech-language pathologists. Receptive and expressive language, articulation, oral motor skills, fluency, narrative ability, and reading skills were scored as normal, mildly impaired, moderately impaired, or severely impaired. Pitch was rated as normal, high, or low for a participant's age and gender, based on criteria proposed by Boone and McFarlane.[2] Voice quality was rated as normal, soft, harsh, hoarse, or strained. Resonance characteristics of speech were rated as normal, hypernasal, or hyponasal. The rate of speech was classified as slow, normal, or fast. A monotone quality to the spontaneous speech sample was also noted.

Results and Conclusions

All participants reported a history of communication difficulties and/or were enrolled in speech and language therapy. The majority of children (83%) received speech therapy prior to the age of 3 years, all had received therapy during the school-age years, and two (20%) continued to receive speech therapy into adolescence and adulthood. This suggests that therapy needs for the individual with PWS are identified early and required for most individuals across the life span.

Oral motor deficits and associated speech-sound disorders are prevalent in the PWS population, with 90.6% demonstrating mild to severe oral motor deficits including poor tongue mobility, shortness of palate, and incoordination of the articulators (see Table 9.2). Mild to severe articulation impairment was observed in 92% of the participants, with younger subjects more severely impaired. A variety of sound

Table 9.2. Speech Characteristics and Deficits in Individuals with Prader-Willi Syndrome

Speech Characteristic	Ratings of Speech Characteristics Percentage of Subjects (N)			
	Normal	Mild	Moderate	Severe
Oral Motor Skills* N = 29	9.4%	31.3%	31.3%	18.8%
Articulation Skills N = 25	8%	12%	36%	44%

* 9.2% of participants could not be tested but did have oral motor difficulties.

substitutions and distortions as well as phonological processing errors were noted. This supports previous suggestions that the speech-sound errors observed in PWS are the result of both poor oral motor skills and concomitant language deficits.

As predicted, receptive and expressive language deficits based on age normative data were observed in the majority of individuals, with 90.5% presenting with receptive language delays and 91.7% presenting with expressive language delays. In addition, vocabulary, pragmatic, and narrative deficits were observed. However, most of the participants who were school-age or older were able to read fluently (83.3%). Reading comprehension was not assessed. Future studies should examine reading comprehension abilities relative to reading decoding skills.

Ratings of pitch and nasality revealed great variability. Thirty-five percent of the participants presented with a high pitch, 30% with a low pitch, and 35% with a pitch appropriate for their age and gender (see Table 9.3). While hypernasality was frequently observed (70.6%), hyponasality was also noted (17.6%). It is not known whether or not the same factors that contributed to the hypernasality (hypotonia and oral structure) also contributed to the hyponasality that was observed. Further research is needed in this area.

Comparisons between age-matched individuals with uniparental disomy (UPD) and individuals with chromosome 15q deletions were inconclusive. On two of the speech-language measures UPD subjects received better ratings than did the deletion subjects; on two other measures the deletion subjects received better ratings than did the UPD

Table 9.3. Voice Characteristics in Individuals with Prader-Willi Syndrome

Voice Characteristic	Ratings of Voice Characteristics Percentage of Subjects (N)		
Pitch N = 20	Normal 35%	Low 30%	High 35%
Resonance N = 17	Normal 11.8%	Hyponasal 17.6%	Hypernasal 70.6%

subjects; and in one case the ratings for the UPD subjects were comparable. Further research is needed to draw associations between the type of chromosome abnormality and the clinical presentation.

Therapeutic Implications

The individual with PWS will require the services of a speech-language pathologist from infancy through adulthood. A team approach that includes an occupational therapist, physical therapist, dietitian, psychologist, physician, speech-language pathologist, genetic counselor, social worker, and educational specialist provides the optimal management strategy for the child or adult with PWS. Early intervention begins in infancy with a focus on improving oral motor skills for feeding. Continued monitoring of speech and language skills is important as the degree of hypotonia changes over time.[18] Assessment includes standardized and nonstandardized measures to assess oral structures and functions, speech-sounds, and receptive/expressive language skills. Later on, with the development of conversational speech, voice, fluency, and resonance characteristics may be assessed.

It is essential that the speech-language pathologist be aware of the unique characteristics of the syndrome that may impact on speech-language development. For example, in some children drooling is a sign of poor oral motor control. However, children with PWS seldom drool due to reduced saliva output. The speech-language pathologist may incorrectly assume that oral motor skills are intact since drooling is not observed. Further, reduced saliva output may cause dental decay, thus impairing articulation. Table 9.1 summarizes some of the characteristics of PWS that may contribute to speech and language impairment.

In addition to articulation and language therapy, intervention should emphasize social skills and the pragmatic use of language.[8] As shown in Table 9.1, many of the behavioral characteristics associated with PWS impede good pragmatic language ability. Early and ongoing training of social skills will assist the individual with PWS in maintaining appropriate social and interpersonal interactions.

Caretakers and professionals should also be aware of the wide range of communication deficits that are associated with PWS. Therapy should be tailored to address the specific speech and language deficits observed, rather than employing a cookbook approach. Therapy should include an emphasis on the development of oral motor skills. Imitation of movements of the tongue, lips, jaws, and palate may be incorporated into games (see Orr[19] for oral motor games for children). Oral motor skills may be trained in both speech-sound and nonspeech activities. Isolated movements may be mastered first, followed by sequential and motorically complex movements of the articulators.

Future Directions

The speech and language skills of individuals with PWS have not been as well described as those of syndromes with a higher prevalence, such as Down syndrome. While individual case studies have been

useful in outlining some of the characteristics of the speech and language disorders associated with PWS, such case studies have failed to describe the great variability of these skills found in PWS. Larger cohort studies are needed to understand the range of speech and language skills that individuals with PWS present. Specific cognitive strengths and weaknesses associated with PWS may impact the speech and language skills of an individual with PWS in a unique way. Comparison groups of children with similar IQ ranges may be employed to highlight the distinctive aspects of PWS. Therapists and other professionals should be acquainted with the features of PWS that may potentially influence communication development. Therapy programs designed for children without PWS may not be appropriate for the child with PWS.

As new medical treatments are employed with the PWS population, such as growth hormone treatment, continued research is needed to determine its impact on speech and language. Therapy strategies may be modified to augment these medical interventions.

Glossary

apraxia (*of speech*)—nonlinguistic sensorimotor disorder of articulation, characterized by impaired capacity to program position of speech musculature and sequence of muscle movements for the volitional production of phonemes.

articulators—the teeth, lips, and tongue, as they are involved in the production of meaningful sounds.

cluttering—speech characterized by overuse of fillers, rapid rate, and word and phrase repetitions. Unlike stuttering, the individual is usually unaware of the difficulty.

flaccid dysarthria—faulty speech production due to motor difficulties resulting from hypotonic (decreased) muscle tone, characterized by imprecise consonants and irregular articulation. Respiration, voice, fluency, and prosody (melody of speech) may be hindered as well. Both volitional and automatic actions, including chewing, swallowing, and other oral motor movements, may also be impaired. Anarthria, or the inability to articulate at all, is the result of severe neuromuscular involvement.

interjections—the insertion of extra sounds or words that do not add to or modify the meaning of the sentence, such as "you know" or "like."

larynx—the upper part of the trachea (windpipe); contains the vocal cords.

micrognathia—a small jaw.

morphology—the form and internal structure of words; the transformation of words in such ways as tense and number.

nasal emissions—airflow directed via the nasal cavity that passes out the nose rather than the more normal route, through the oral cavity.

nasal snorting—airflow directed into the nasal cavity producing a snorting sound.

pharyngeal flap—a surgical procedure designed to correct velopharyngeal insufficiency.

phoneme—the smallest unit of sound in any particular language; the English language designates approximately 44 different phonemes.

plosive consonants—p, b, t, d, k, and g.

velopharyngeal—of or relating to the structures of the soft palate and the pharynx.

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