

ARTICLE

Cognitive and Behavioral Characteristics of Children With Williams Syndrome: Implications for Intervention Approaches

CAROLYN B. MERVIS* AND ANGELA E. JOHN

Portrayals of individuals with Williams syndrome (WS), a genetic disorder caused by a microdeletion of ~25 genes on chromosome 7q11.23, have reached the general public through a variety of media formats. These descriptions are often paradoxical in nature with individuals with WS repeatedly described as demonstrating near-normal language despite the presence of significant intellectual disability and as being extremely sociable and friendly in spite of their seemingly limited understanding of basic social norms. While this depiction of WS served to attract the interest of basic-science researchers, the results of subsequent studies have provided a more nuanced view. For example, rather than across-the-board “near-normal” language, children with WS demonstrate relative strengths in concrete vocabulary and verbal short-term memory, grammatical abilities at the level expected for general intellectual ability, and considerable weakness in relational/conceptual language and pragmatics (social use of language). To provide a more thorough characterization of the WS behavioral phenotype, we summarize recent findings related to intellectual ability, language development, memory development, executive function development, adaptive behavior skills, and behavior as it relates to learning by children with WS. Finally, we briefly discuss intervention approaches that may help children with WS to achieve their full potential. © 2010 Wiley-Liss, Inc.

KEY WORDS: Williams syndrome; cognitive development; language development; behavior; intervention

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INTRODUCTION

Williams syndrome (WS) is a complex neurodevelopmental disorder caused by a deletion of ~25 genes on one copy of chromosome 7q11.23 [Hillier et al., 2003; Osborne, 2006] with an estimated prevalence rate of 1 in 7,500 live births [Strømme et al., 2002]. WS is associated with specific physical and medical characteristics including a characteristic

facial appearance, congenital heart disease (especially supravalvular aortic stenosis), connective tissue abnormalities such as hernias or diverticuli of the bladder or colon, and failure to thrive or growth deficiency [Morris, 2006]. Infants and young children with WS have developmental delay and older children typically have intellectual or learning disabilities, although intellectual level ranges from average for the

general population to severe intellectual disability. In addition, WS is associated with specific cognitive [Mervis et al., 2000] and personality [Klein-Tasman and Mervis, 2003] profiles.

Portrayals of people with WS have reached the general public through a variety of media formats. Over the last two decades, a major source of information has been articles in lay publications. The first article featuring WS in a lay magazine was published in *Discover* [Finn, 1991]. In this article, Finn stated, “People with Williams syndrome are smart and mentally retarded, gifted and inept at the same time [p. 55].” He went on to say that “People with Williams syndrome can show lots of intelligence in certain areas—language, music, and interpersonal relations, for example—and yet their IQ is typically between 50 and 70, low enough to qualify them as moderately to mildly retarded [p. 56].”

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Cognitive Children With and Williams Behavioral Syndrome: Characteristics of Implications for Intervention Approaches

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Blakeslee [1994], a contributor to the New York Times, wrote that in the case of WS, “There are severe

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malformations throughout the brain and heart, yet the capacity for language is remarkably unaffected. If anything, language and sociability are enriched [p. 1 of online article].” Fourteen years later, an article published in the New York Times Magazine offered a very similar description. Dobbs [2007] wrote that “Many with Williams have so vague a concept of space, for instance, that even as adults they will fail at six-piece jigsaw puzzles, easily get lost, draw like a preschooler and struggle to replicate a simple T or X shape built with a half- dozen building blocks....The low I.Q., however, ignores two traits that define Williams more distinctly than do its deficits: an exuberant gregariousness and near-normal language skills. Williams people talk a lot, and they talk with pretty much anyone [p. 1 of online article].”

Increased public awareness of autism also led to increased interest in WS. In an article in Newsweek presenting a new theory on autism, Cowley [2003] included the following description of WS highlighting the seemingly opposite social phenotype to that associated with autism: “As fate would have it, some of the best natural readers of feelings and faces are themselves profoundly disabled. People with a rare genetic disorder called Williams syndrome are often severely retarded. Yet they’re hypersocial, highly verbal and often deeply empathic [p. 50].”

A comparison of these depictions yields a recurrent theme: a fascination with how an individual could have significant intellectual disability but still have near-normal language, and could be extremely sociable and friendly yet have seemingly little understanding of basic social norms. It is this very depiction of WS that attracted the attention of basic-science researchers and encouraged them to attempt to characterize the WS behavioral phenotype systematically. Bellugi et al. [1988] argued that despite demonstrating severe intellectual disability and functioning in Piaget’s preoperational period, the adolescents with WS they studied nevertheless had excellent language abilities. In particular, Bellugi et al. reported that

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these adolescents were unable to conserve either number or quantity yet could comprehend and produce complex linguistic constructions (e.g., reversible passives, conditionals, and tag questions). They further argued that given this pattern of language strengths and cognitive limitations, WS provided strong evidence of the independence of language from cognition. This characterization of WS quickly attracted the attention of researchers interested in the relation between language and cognition and launched WS to the forefront of the debate on the modularity of language. As additional researchers began to study children with WS, experts concerned with modularity began to write about the syndrome, taking a considerably more strident position than did Bellugi and colleagues. Piattelli-Palmarini [2001] offered a particularly provocative statement: “For instance, children with WS have barely measurable intelligence and require constant parental care, yet they have an exquisite mastery of syntax and vocabulary [p. 887].”

Over the last two decades researchers have not only empirically evaluated these characterizations of WS but also have looked beyond these depictions, providing a more nuanced view of the overall behavioral phenotype associated with WS. Our aim in this manuscript is to provide a summary of recent findings related to intellectual ability, language development, memory development, executive function development, adaptive behavior skills, and behavior as it relates to learning in children with WS, with the goal of providing a more thorough characterization of the WS behavioral phenotype. In addition, we briefly discuss intervention approaches that may help children with WS to achieve their full potential.

OVERALL INTELLECTUAL ABILITY

An important purpose of intellectual ability assessment is to determine IQ. This purpose is well known throughout the medical, educational, and lay communities. An equally important, but much less often considered, goal is to identify an individual’s relative strengths and weaknesses as a function of type of intellectual ability. For example, as we describe below, WS is associated with relative strengths in (concrete) language, (concrete) nonverbal reasoning, and verbal short-term memory and severe weakness in visuospatial construction. WS also is associated with a range of intellectual ability, from the rare individual in the average range for the general population through

the equally rare individual in the severe intellectual disability range. Most individuals have

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overall IQs in the borderline to moderate intellectual disability range. An assessment that is able to detect the pattern of cognitive strengths and weaknesses associated with WS across the full range of levels of intelligence associated with the syndrome would be particularly useful.

There is no mandatory set of abilities or higher level organization of these abilities required for tests of intellectual ability. Similarly, there is no mandatory range of intellectual ability levels that must be covered. Thus, different tests measure somewhat different abilities and group these abilities in different manners. Furthermore, the subtests are normed to three standard deviations below the mean for some

assessments and to four standard deviations below the mean for others. The manner in which subtests are grouped and the number of standard deviations below the mean for which subtests are normed determine how likely a particular assessment is to identify the pattern of strengths and weaknesses associated with a particular syndrome.

The most commonly used assessments of intellectual ability for individuals with intellectual disability are the Wechsler tests [e.g., Wechsler Intelligence Scale for Children, e.g., Wechsler, 2003; Wechsler Adult Intelligence Scale, e.g., Wechsler, 1981]. These assessments are normed to three standard deviations below the mean and group subtests measuring nonverbal reasoning ability and subtests measuring spatial ability together in a single composite (Performance or Perceptual Reasoning). Thus, although on average Wechsler Verbal composite standard score is 5 points higher than Performance composite standard score for individuals with WS [Howlin et al., 1998; Searcy et al., 2004], Searcy et al. found that only 24% of individuals with WS scored significantly higher on the Verbal composite than on the Performance composite (1% scored significantly higher on the Performance composite than on the Verbal composite). Thus, for most individuals with WS, pattern of performance on the Wechsler composites does not mirror the cognitive profile associated with the syndrome. Furthermore, on some of the subtests many individuals earned the lowest possible standard score, indicating that the test is not normed low enough to accurately capture the ability levels of many individuals with WS.

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standard score, indicating that the test is not normed low enough to accurately capture the ability levels of many individuals with WS.

The Differential Ability Scales assessment [DAS; Elliott, 1990 and DAS-II; Elliott, 2007] was designed to identify individuals' patterns of strengths and weaknesses. This measure provides separate composite (cluster) standard scores for Verbal, Nonverbal Reasoning, and Spatial abilities. Verbal short-term memory is assessed by a supplemental subtest that is not included in any of the clusters. The DAS-II subtests are normed to four standard deviations below the mean. Examination of the pattern of mean standard scores (Table I) indicates that at the group level, the DAS-II accurately captures the pattern of strengths and weaknesses previously reported for individuals with WS, with performance on the Verbal, Nonverbal Reasoning, and verbal short-term memory measures at about the same level and performance 20 points lower on the Spatial measure. The pattern of significant weakness in spatial abilities is also captured at the individual level; 86% of children performed significantly better on either the Verbal or Nonverbal Reasoning cluster (or on both) than on the Spatial cluster. Two children (2%) scored significantly higher on the Spatial cluster than on the Verbal cluster. Examination of the standard deviations indicates that the DAS-II is normed low enough to capture the abilities of even low functioning children with WS.

The Mullen Scales of Early Learning [Mullen, 1995] may be used to assess the intellectual abilities of very young children with WS. This measure also includes separate measures of nonverbal reasoning (referred to as Visual Reception) and spatial ability (referred to as Fine Motor). As indicated in Table I, the same pattern of relative strengths in nonverbal reasoning and verbal abilities and severe weakness in visuospatial construction abilities is apparent even for 2-year-olds with WS.

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Unfortunately, the Mullen is only normed to 3 standard deviations below the mean, so this assessment does not accurately capture the abilities of lower functioning children with WS.

In collaboration with Karen Berman's research group at the National Institute of Mental Health, our research group has conducted neuroimaging studies comparing adults with WS to groups of individuals in the general population matched for gender, age, and IQ. We have identified an area of reduced gray matter and sulcal depth in the intraparietal sulcus [Meyer-Lindenberg et al., 2004, 2006; Kippenhan et al., 2005]. Results of functional neuroimaging (fMRI) studies indicated that this area served as a roadblock to dorsal stream information flow in a two-dimensional analog to the DAS Pattern Construction subtest (the hallmark visuospatial construction weakness in WS). The convergence of behavioral and neuroimaging results strongly suggests that one or more genes in the WS region, in transaction with other genes and the environment, contributes to the development of visuospatial construction skills.

LANGUAGE ABILITIES

For the past 20 years, the modal topic for behavioral research studies of individuals with WS has been language development. This pattern reflects the fact that behavioral researchers initially were drawn to the study of WS to address

questions of modularity, in particular, whether language ability was independent of cognitive ability. Until recently, most studies of language ability focused on language content (vocabulary) and structure (grammar). Although studies of these aspects of language development have continued, as the question of similarities and contrasts between WS and autism or autism spectrum disorders (ASDs) has become a major focus, studies of the socio-communicative aspects of language ability (pragmatics) have become much more common. Below we briefly review results of studies focused on early language development followed by findings from studies of vocabulary, grammar, literacy, and pragmatic development.

Early Language Development

The onset of language acquisition by children with WS is almost always

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TABLE I. Descriptive Statistics for Standardized Assessment Performance of Children and Adolescents With WS

Measure N

CA range (in years)

Mean standard

score (SS) SD SS range

Differential Ability Scales-II

GCA (similar to IQ) 120 4.01–17.71 64.56 12.33 31–96 Verbal Cluster SS 120 4.01–17.71 74.06 16.41 30a–111 Nonverbal Reasoning Cluster SS 120 4.01–17.71 78.89 15.44 37–118 Spatial Cluster SS 120 4.01–17.71 54.82 11.27 32a–81 Recall of Digits—Forward SS 120 4.01–17.71 72.06 15.71 40a–102 Mullen Scales of Early Learning

Early Learning Composite 144 2.01–4.96 61.45 11.31 49a–96 Visual Reception T 144 2.01–4.96 29.51 3.21 20a–58 Fine Motor T 144 2.01–4.96 21.18 9.68 20a–41 Receptive Language T 144 2.01–4.96 29.45 9.58 20a–55 Expressive Language T 144 2.01–4.96 32.60 11.31 20a–56 Scales of Independent Behavior-Revised

Broad Independence 122 4.02–17.77 55.11 15.45 24a–95 Motor Skills 122 4.02–17.77 57.82 15.13 24a–88 Social Interaction and Communication Skills 122 4.02–17.77 73.16 14.72 30–110 Personal Living Skills 122 4.02–17.77 61.22 14.53 24a–98 Community Living Skills 122 4.02–17.77 57.35 17.20 24a–96 Peabody Picture Vocabulary Test-4 129 4.01–17.71 81.84 15.04 20a–124 Expressive Vocabulary Test-2 129 4.01–17.71 79.43 14.83 20a–120 Test of Relational Concepts 92 5.00–7.95 55.79 21.37 25a–104 Test for Reception of Grammar-2 170 5.02–17.71 74.55 17.73 55a–116

For the general population, mean 1/4100 (SD 1/415) for SS and mean 1/450 (SD 1/410) for T scores. aLowest possible SS or T for the relevant assessment.

delayed. Masataka [2001] has argued that this delay is due to specific motor delays. In particular, he has argued that rhythmic hand banging provides the motor substrate for canonical babble and that without canonical babble, word production is for the most part impossible. In a longitudinal study of eight children with WS, Masataka [2001] found that the onset of rhythmic hand banging was considerably delayed relative to expectations for TD children. Nevertheless, the pattern of correlations among abilities shown by children with WS was consistent with that for TD children, with the onset of rhythmic hand banging strongly correlated with the onset of canonical babble, which in turn was strongly correlated with the attainment of a 25-word expressive vocabulary. Mervis and Bertrand [1997] reported similar findings for the two children in their longitudinal study who were not producing canonical babble at the start of

the study. Velleman et al. [2006; see also Mervis and Becerra, 2007] analyzed the phonological repertoires of six 18-month-olds with WS and found that their babble was considerably delayed relative to that of the age-matched TD comparison group. Consistent with Masataka's [2001] argument that the production of canonical babble is critical for word production, Velleman et al. found that the children whose language was the most advanced had the most normal babble histories and that the child whose language was the most delayed had not met the criterion for canonical babble even at the age of 36 months.

Although speech perception skills are critical for spoken language development, only two studies of these abilities in young children with WS have been reported. Nazzi et al. [2003], in a study of 17 children with WS aged 17–47 months (mean age 33 months), found

that the children were able to segment words with a strong–weak stress pattern (the predominant pattern in English) from ongoing speech but could not reliably segment words with a weak– strong stress pattern. Nazzi et al. argued that this combination of results suggested that young children with WS were using prosodic cues (which are adequate to identify words with a strong-weak stress pattern) rather than distributional information (which is needed to identify words with a weak – strong stress pattern) to identify words in ongoing speech. Cashon et al. [2009] studied the speech segmentation skills of ten 9- to 20- month-olds (mean age of 14 months) with WS using an artificial language in which all syllables were equally stressed, so that “words” could only be identified based on distributional properties. Results indicated that the children were able to use distributional properties to segment words from continuous speech in the absence of prosodic cues. In both studies, the youngest children with WS were older than the ages at which these abilities are shown by TD infants, so data are not available to address the question of whether speech perception development is delayed for children with WS.

As expected given delays in the onset of canonical babble, the onset of word production is also delayed. Mervis et al. [2003] followed 13 children with WS longitudinally and found that age at acquisition of a 10-word expressive vocabulary was below the 5th centile (the lowest centile included in the norms) for the MacArthur-Bates Communicative Development Inventory (CDI) [Fenson et al., 1993] for all of the children. Age at acquisition of 50- and 100-word expressive vocabularies was also below the 5th centile for 12 of the 13 children. Despite these delays, the underlying categories for the early object labels comprehended and produced by children with WS were similar to those for both TD children and children with Down syndrome (DS), indicating that in contrast to prior claims that for individuals with WS, language was independent of cognition [e.g., Bellugi et al., 1988], the early cognitive development and early language devel-

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opment of children with WS in fact were closely linked [Mervis and Bertrand, 1997; Mervis and Becerra, 2007]. For example, similar to TD children and children with DS, the extension of the early object labels of children with WS corresponded to their play patterns with the objects (e.g., the children rolled a wide variety of approximately spherical objects whether or not they were balls; they also comprehended and produced the word “ball” in relation to the same set of objects) [Mervis and Bertrand, 1997; Mervis and Becerra, 2007].

The expressive vocabularies of young children with WS and young children with DS have been compared in several studies. Results indicated that the mean expressive vocabulary size of young children with WS was significantly larger than that of children with DS when the children were matched for chronological age (CA) [Mervis and Robinson, 2000] but when the two groups were matched for developmental level, the mean expressive vocabulary size of the DS group was almost the same as that of the WS group

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[Vicari et al., 2002]. At the same time, the grammatical abilities and verbal memory abilities of the WS group were considerably more advanced than those of the matched DS group [Vicari et al., 2002; see also Volterra et al., 2003].

Vocabulary Development

Receptive concrete vocabulary (comprehension of labels for objects, actions, and descriptors) has consistently been identified as an area of relative strength for individuals with WS. As indicated in Table I, the highest mean standard

score for individuals with WS is on the Peabody Picture Vocabulary Test (in this case, the 4th edition) [PPVT-4; Dunn and Dunn, 2007]. Furthermore, 83% earned a standard score of at least 70 (the bottom of the “normal” range) and 8% earned a standard score of at least 100 (the 50th percentile for the general population). Similar results have been reported for previous editions of the PPVT as well [e.g., Bellugi et al., 1988; Brock et al., 2007; Mervis and Baccerra, 2007]. The finding that receptive concrete vocabulary is a relative strength is not unique to WS, however. Glenn and Cunningham [2005] reported that pattern for individuals with DS and Facon et al. [1993] reported similar findings from a meta-analysis, for studies in which mean IQ was <70. Participants in these studies had a wide range of etiologies.

Expressive concrete vocabulary as measured by the Expressive Vocabulary Test—2nd edition [EVT-2; Williams, 2007] also is a relative strength for individuals with WS. As indicated in Table I, mean EVT-2 standard score was 2 points lower than mean PPVT-4 standard score, and similar to the findings for the PPVT-4, 83% of individuals with WS earned a standard score of at least 70 on the EVT-2 and 6% earned a standard score of at least 100.

In sharp contrast to their relative strength in concrete vocabulary, individuals with WS have a great deal of difficulty with relational/conceptual vocabulary. Basic relational vocabulary includes terms for spatial, temporal, quantitative, and dimensional concepts;

more advanced relational vocabulary includes conjunctions (e.g., and, or) and disjunctions (e.g., although, however, nevertheless, neither... nor); all of these concepts are very difficult for individuals with WS. A comparison

In sharp contrast to their relative strength in concrete vocabulary, individuals with WS have a great deal of difficulty with relational/ conceptual vocabulary. Basic relational vocabulary includes terms for spatial, temporal, quantitative, and dimensional concepts; more advanced relational vocabulary includes conjunctions (e.g., and, or) and disjunctions (e.g., although, however, nevertheless, neither... nor); all of these concepts are very difficult for individuals with WS.

of the performance of 5- to 7-year-olds with WS on the PPVT-III [Dunn and Dunn, 1997] and the Test of Relational Concepts [TRC; Edmonston and Litchfield Thane, 1988] indicated that mean standard score on the concrete vocabulary measure was 30 points higher than on the relational vocabulary measure [Mervis and John, 2008]. In fact, the children's performance on the TRC was similar to their performance on the DAS Pattern Construction subtest [Elliott, 1990], the signature weakness of individuals with WS. The pattern of errors indicated that children with WS had difficulty with all types of relational concepts, not just with spatial concepts. This finding is consistent with Walsh's [2003] argument that spatial, temporal, and quantitative processing are all controlled by a common magnitude system that is located in the inferior parietal

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cortex—the area in which Meyer-Lindenberg et al. [2004, 2006] identified a region of reduced gray matter that served as a roadblock to dorsal stream information flow, suggesting a possible common basis for the findings of extreme difficulty in both visuospatial construction and relational language for individuals with WS.

Although most individuals with WS eventually acquire basic relational concepts, they continue to have difficulty with many of the more advanced relational concepts. For example, twelve of twenty-nine 9- to 11-year-olds with WS tested on the Formulated Sentences subtest of the Clinical Evaluation of Language Fundamentals-IV [CELF-IV; Semel et al., 2003], a measure that includes both simple and advanced relational concepts, earned the lowest possible scaled score [Mervis and John, 2008]. A comparison of the performance of the children who participated in both the TRC study and, an average of 4 years later, the CELF-IV Formulated Sentences study indicated very strong continuity in relational language ability over the age range of the two studies [Mervis and John, 2008].

Grammatical Development

Bellugi et al.'s [1988] initial report that the grammatical abilities of individuals with WS were well above those expected for their cognitive abilities was based on a comparison between CA- and IQ-matched adolescents with WS and DS. The finding that the grammatical abilities of individuals with WS are more advanced than those of matched individuals with DS has been replicated for both children whose native language is English [Mervis et al., 2003; Joffe and Varlokosta, 2007a,b] and children whose native language is Italian [Vicari et al., 2002, 2004]. However, these results most likely reflect the inordinate difficulty that individuals with DS have with grammatical development, rather than indicating that individuals with WS have better-than-expected grammatical abilities. In fact, when the group compared to WS is composed of either CA- and

IQ-matched children with other etiologies of intellectual disability (ID) or MA-matched TD children, the grammatical abilities of the WS group are consistently at or below that of the contrast group across a variety of languages: English [e.g., Udwin and Yule, 1990; Grant et al., 1997; Zukowski, 2004; Mervis and Becerra, 2007; Perovic and Wexler, 2007], Hungarian [Lukács, 2005], and Italian [Volterra et al., 1996; 2003]. For a discussion of potential difficulties in interpreting results of studies in which individuals with ID are compared to much-younger

TD children—a problem for a subset of the studies just referenced—see Mervis and Klein-Tasman [2004] and Mervis and Robinson [2005].

The most common standardized assessment of receptive grammatical ability used in studies of individuals with WS is the Test for Reception of Grammar [TROG; Bishop, 1989 or TROG-2; Bishop, 2003]; this measure has been translated into several languages. The TROG measures a variety of sentence structures, ranging from simple subject–verb constructions to center-embedded relative clauses. As indicated in Table I, the mean level of performance for children and adolescents with WS is in the borderline range; 28% earned the lowest possible standard score. Karmiloff-Smith et al. [1997] reported similar findings for a smaller sample. The results of studies using the Italian and Hungarian versions of the TROG have indicated that the order of difficulty of grammatical constructions for individuals with WS is highly similar to that for children in the general population acquiring the same native language [for Hungarian: Lukács, 2005; for Italian: Volterra et al., 1996]. Receptive grammatical ability as measured by the TROG is strongly related to verbal working memory ability (as measured by digits backward recall ability) for individuals with WS. Furthermore, this relation is stronger for individuals with WS than for TD children with similar levels of receptive grammar ability, suggesting that individuals with WS may rely more heavily on verbal working memory when parsing com-

plex grammatical constructions than do TD children [Robinson et al., 2003].

Literacy

Three of the first five behavioral research articles published on children with WS included information about reading abilities. The results of these studies indicated a wide range of reading abilities, with a small proportion of children decoding (reading single words) and comprehending at grade level and a small proportion not able to read at all [Pagon et al., 1987; Udwin et al., 1987; MacDonald and Roy, 1988]. Udwin et al. reported that the median reading level for a group of individuals with WS aged 10–20 years was 2nd grade. Presaging later findings, MacDonald and Roy noted that children who were being taught to read with phonics seemed to benefit more than children being taught to read with the “look–say” (sight-word or whole-word) method.

For TD children, phonological awareness is strongly related both to decoding and to reading pseudowords based on the phonics rules of the relevant language [see reviews in Ehri, 2004 and McCardle et al., 2008]. The relation of phonological awareness to decoding has been addressed in several studies involving individuals with WS. Levy et al. [2003], studying individuals learning to read English, found that elision (the ability to delete specified syllables or sounds from a word) was strongly related to both decoding and pseudoword reading. This same finding has been obtained for individuals learning to read Italian [Menghini et al., 2004] and Hebrew [Levy and Antebi, 2004]. Based on their findings, Levy and colleagues recommended that children with WS be taught to read using phonics [Levy et al., 2003; Levy and Antebi, 2004].

Becerra et al. [2008; see also Mervis, 2009] analyzed the reading performance of forty-four 9- to 17-year-olds with WS. Initial analyses considered the sample as a single group to provide information about the reading abilities of individuals with WS relative to general-population norms for children

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of the same CA. Mean standard scores on the Reading section of the Wechsler Individual Achievement Test-II [WIAT-II; Wechsler, 2005] were 73.00 [range: 40 (floor)–112] for Word Reading (decoding), 78.75 [range: 0 correct–113 (standard score)] for Pseudoword Decoding, and 64.61 [range: 40 (floor)–102] for Reading Comprehension. Mean standard score for Reading Comprehension was significantly lower than for either Word Reading or Pseudoword Decoding. For all three subtests, the standard deviations were >15, indicating more variability than in the general population. All children could read at least a few words, but eight (18%) could not read any pseudowords. Several participants, including 1 in 11th grade, read and comprehended at grade level.

In a second set of analyses, the participants were divided according to their primary method of reading instruction: phonics (n1/424) or whole word/sight word/whole language (n1/420). The children’s reading standard scores were then compared to those predicted based on their DAS-II GCA, using the tables in the DAS-II manual [Elliott, 2007]. Results indicated large and significant differences as a function of reading method for all three reading subtests. Most children in the Phonics group read at or above the level expected for their GCA. In strong contrast, most children in the Whole Word group read below the level expected for their GCA. These findings are consistent with those of the meta-analyses conducted by the National Reading Panel [see summaries in Ehri, 2004; McCardle et al., 2008], which stressed the importance of early, explicit, and systematic instruction in phonemic awareness and phonics for all children.

Pragmatics

The combination of a relative strength in the structural and concrete content aspects of language and increased sociability paired with consistent problems in making friends and sustaining friendships led researchers to hypothesize that children with WS likely have difficulty with the pragmatic aspects of language.

The results of the studies examining the pragmatic abilities of individuals with WS are consistent with this position, documenting pragmatic difficulties across developmental stages. The emergence of joint attention in children with WS is delayed relative to both CA and language ability [Mervis and Bertrand, 1993, 1997; Mervis et al., 2003]. Furthermore, although both TD children and children with DS begin to comprehend and produce

pointing gestures prior to the onset of referential expressive language, children with WS do not comprehend and produce pointing gestures until well after the onset of referential word production [Mervis et al., 2003; Mervis and Becerra, 2007]. Children with WS are significantly less likely to engage in joint attention and to comprehend and produce gestures than are either mental-age-matched TD children or children with DS individually matched on CA, developmental quotient, and expressive vocabulary size [e.g., Laing et al., 2002; Rowe et al., 2005]. John and Mervis [in press] examined the ability of preschoolers with WS and CA-matched preschoolers with DS to comprehend communicative intent expressed by pointing gestures and eye gaze. Despite having significantly lower mean developmental quotient scores, the children with DS were significantly better at inferring communicative intent than were the children with WS; 60% of the children with DS but only 27% of the children with WS found the hidden toy at a rate significantly above that expected by chance.

These findings that the pragmatic abilities of even young children with WS are more limited than expected for their developmental level provided the initial basis for questioning the accuracy of the early characterizations of WS as the “opposite” of autism.

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The overlap between the phenotypes associated with WS and ASDs has been addressed in three studies of the performance of toddlers and preschoolers [Klein-Tasman et al., 2007, 2009; Lincoln et al., 2007] on the Autism Diagnostic Observation Schedule- Generic [ADOS-G; Lord et al., 1999], a semi-structured play-based interaction designed to press for behaviors central to a diagnosis of ASD. Participants in these studies had very limited to no expressive language.

Results of these studies indicated that a large portion of the participants with WS demonstrated behaviors typically thought to be characteristic of children with ASDs. For example, approximately half of the participants reported by Klein-Tasman et al. [2007, 2009] and Lincoln et al. [2007] did not clearly integrate eye contact with their communicative partner in order to reference a desired object that was out of reach. Almost three quarters of the children did not integrate eye contact or vocalization with acts of showing objects, and nearly all of the children with WS in these studies did not spontaneously use a doll or other object as an independent agent or use objects to represent other objects. These difficulties were such that many of the children in these studies were classified on the ADOS algorithm as “autism-spectrum disorder” (38% in the Klein-Tasman et al. studies, and 5% in Lincoln et al.) and some children were classified as “autism” (10% in the Klein-Tasman et al. studies and 5% in Lincoln et al.). However, differences between the behavioral phenotypes associated with WS and ASDs also were found. Few children with WS evidenced difficulty directing vocalizations or facial expressions to other people or sharing affect, and the quality of social overtures was generally good [Klein-

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Tasman et al., 2007, 2009; Lincoln et al., 2007].

Studies examining pragmatic abilities in older individuals with WS have shown that these difficulties continue into the school-age and adult years and are of considerable concern to parents. These studies have focused on parental responses to questionnaires addressing communicative competence, analyses of children’s conversations with a researcher, and children’s responses in a task measuring comprehension monitoring skills. In addition, researchers have examined children’s theory of mind (ToM) ability, as ToM is considered to play a key role in pragmatics.

Four studies have addressed the general pragmatic abilities of individuals with WS using a version of the Children’s Communication Checklist [CCC; Bishop, 1998; or CCC-2; Bishop, 2002], a parent-report measure. Laws and Bishop [2004] studied 19 individuals with WS (mean CA/414.83 years) and found that 15 (79%) met the CCC cut-off for pragmatic language impairment. The WS group evidenced significant difficulties in all areas of pragmatics measured by the CCC. Relative to a DS group (mean CA/415.92 years) and a group of children with Specific Language Impairment (mean CA/46.00 years), the WS group evidenced particular difficulty in the use of stereotyped conversations, inappropriate initiation of conversations, and overdependence on context to interpret what was said to them. These findings have been well replicated by other research groups [Peregrine et al., 2005; Philofsky et al., 2007; Harmon et al., 2009]. Many of these pragmatic problems are evident even when the comparison group is children with autism; Philofsky et al. [2007] reported that although children with WS earned significantly better scaled scores than CA-matched children with autism on the CCC-2 Stereotyped Language and Nonverbal Communication scales, the two groups evidenced similar impairments on the Inappropriate Initiation and Use of Context scales.

Several studies directly examining the conversational abilities of children with WS have been conducted. In the first such study, Udwin and Yule [1990] reported the results of analyses of 30-min conversations between a child with WS and a researcher. Of the 43 children who participated (mean CA/411.1 years), 16 (37%) met the authors’ criteria for hyperverbal speech (fluent speech including an excessive number of stereotyped phrases or idioms, over-familiarity, introduction of irrelevant personal experiences, and perseverative responding). More

recently, Jones et al. [2000] found that while adolescents and adults with WS (CA1/415.8 years) answered the same number of questions within a biographical interview as did CA- and IQ-matched adolescents and adults with DS and MA-matched TD children, the WS group was significantly more likely to describe affective states, make evaluative comments, and use character speech and emphatic markers than was either comparison group. Stojanovik [2006] found that, within a semi-structured conversation, regardless of whether the researcher asked for information or clarification, the responses of the WS group (mean CA 1/49.17 years) were less likely to be adequate than were the responses of either children with specific language impairment (mean CA1/410.58) matched for receptive vocabulary and grammatical ability or slightly younger TD children (mean CA1/48.67). In particular, the WS group was more likely to provide too little information or to misinterpret what the researcher had meant and was considerably less likely to produce a response that continued the conversation.

A person's conversational success depends, in part, on the ability to monitor whether he/she understands what the speaker has said and to request clarification when needed. John et al. [2009] examined comprehension monitoring and verbalizations of message inadequacy by 57 children with WS (mean CA 1/49.24 years) using a listener-role referential communication task modeled after Abbeduto et al. [2008] in which the child and an adult were separated from one another by a barrier.

The child's task was to place the picture requested by the adult into a picture of a larger scene. Although children performed very well when they understood the instructions and the required picture was available, they had considerable difficulty when the researcher's instructions were inadequate (the requested picture was not one of the referents available, the researcher's instruction was ambiguous, or the researcher's instruction contained vocabulary that the child did not understand). Children verbally indicated that there was a problem less than 50% of the time on average and most of their verbalizations were either too vague for the researcher to understand the nature of the problem or indicated the wrong problem. Performance was related to CA and first-order ToM.

Successful communication between two people involves not only a mastery of the language but also taking into account basic information about the communicative partner (e.g., his or her status, knowledge, feelings, focus of attention) and using this information to help formulate an effective message. Thus, successful communication depends at least in part on the ability to understand another person's perspective (i.e., ToM). A deficit in ToM would likely contribute to pragmatic difficulties. Studies of the development of false belief (one of the first types of ToM demonstrated by TD children) have indicated that acquisition of this concept by children with WS is considerably delayed. Tager-Flusberg and her colleagues have compared the performance of children with WS aged 4–10 years on false belief tasks to the performance of CA-, IQ-, and language-matched children with Prader–Willi syndrome (PWS) or nonspecific ID [Tager-Flusberg and Sullivan, 1994, 2000; Joseph and Tager-Flusberg, 1999; Tager-Flusberg and Plesa Skwerer, 2007]. Findings indicated that the WS group did not perform better than the contrast groups, and none of the groups performed well. John and Mervis [2009] used the Unexpected Contents task (a false-belief task routinely passed by TD 4-year-olds) to study the development of ToM in children with WS aged 6–

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14 years. Only the 13- and 14-year-olds reliably passed this task; of the younger children, only three of twenty-eight 6- to 10-year-olds and three of six 11- to 12-year-olds were successful, providing further confirmation that acquisition of even basic ToM ability is greatly delayed.

MEMORY

The pattern of relative strengths and weaknesses for individuals with WS within the memory domain is consistent with the overall phenotypic pattern. In particular, individuals with WS evidence significantly better verbal memory than spatial memory [e.g., Wang and

The pattern of relative strengths and weaknesses for individuals with WS within the memory domain is consistent with the overall phenotypic pattern. In particular, individuals with WS evidence significantly better verbal memory than spatial memory.

Bellugi, 1994; Jarrold et al., 1999]. This pattern also holds in comparisons with CA- and IQ-matched individuals with other forms of ID, if the memory tasks used do not involve mental manipulation (e.g., for “rote” or “short-term” memory tasks). In particular, individuals with WS perform significantly better on measures of forward digit recall than do CA- and IQ/MA-matched groups with DS [Wang and Bellugi, 1994; Jarrold et al., 1999; Klein and Mervis, 1999; Edgin et al., in press] or ID of unknown or mixed etiology [Udwin and Yule, 1991; Devenny et al., 2004]. This same pattern is observed for the first trial of word list recall [Nichols et al., 2004]. In contrast, children and adults with WS perform significantly worse than CA- and IQ/MA-matched individuals with DS on spatial memory tasks such as forward Corsi recall [Wang and Bellugi,

1994; Jarrold et al., 1999; Edgin et al., in press]. No comparisons of spatial memory with contrast groups with ID of mixed or unknown etiology have been reported.

When verbal memory tasks require mental manipulation (“working memory”), however, differences between individuals with WS and those in the contrast group(s) are considerably reduced. On backward digit recall (verbal working memory) tasks, while groups of individuals with WS consistently demonstrate longer spans than CA-

and IQ/MA-matched groups with other forms of ID [e.g., Wang and Bellugi, 1994; Edgin et al., in press; Devenny et al., 2004], these differences are not significant. In the only study that compared performance on backward Corsi recall (spatial working memory), mean span for the WS group and the DS group was almost identical [Edgin et al., in press]. Finally, in contrast to initial characterizations of WS that stressed the independence of language and cognition, there is now mounting evidence that verbal memory abilities are strongly related to both grammatical and vocabulary abilities for individuals with WS [Grant et al., 1997; Plich et al., 2002; Robinson et al., 2003; Mervis, 2006; Mervis and Becerra, 2007].

EXECUTIVE FUNCTION

Executive functioning is a blanket term referring to a set of higher order cognitive processes associated with planning and regulatory control [e.g., Welsh and Pennington, 1988; Hughes and Graham, 2002]. These processes include working memory, inhibition, set shifting/cognitive flexibility, self-monitoring, and generativity. To date four studies of executive functioning abilities in individuals with WS have been published. Tager-Flusberg et al. [1997] compared the performance of children with PWS and children with WS aged 5–8 years on two executive functioning tasks, one examining the ability to verbally inhibit a prepotent response (Day–Night Stroop Task) and one examining the ability to motorically inhibit a prepotent response (Tapping

Task). These tasks were quite difficult for both groups, with 56% of the PWS group and 25% of the WS group passing the verbal inhibition task and 22% of the PWS group and 17% of the WS group passing the motoric inhibition task. Atkinson et al. [2003] also tested individuals with WS aged 4–15 years on tasks measuring inhibition, comparing their performance to norms for TD children whose CA matched the children with WS's vocabulary age on the British Picture Vocabulary Scale (BPVS; the British version of the PPVT). Performance was considerably better on the verbal inhibition task (Day–Night Stroop task), than on the two motor inhibition tasks (Detour Box and Pointing/Counterpointing). For many children with WS, performance on the verbal inhibition task was at or above the level expected for vocabulary age, while for most children with WS, performance on the spatial inhibition tasks was well below the level expected for vocabulary age. There was a significant correlation between performance on the Detour Box spatial inhibition task and performance on the verbal inhibition task. Rhodes et al. [in press] investigated executive functioning in teenagers and young adults with WS (mean CA/4 18.08 years) relative to TD individuals matched for vocabulary age on the BPVS (mean CA/49.25). Individuals with WS evidenced difficulty on the CANTAB (www.camcog.com) tasks of attention set-shifting (Intra-Dimensional/Extra-Dimensional), working memory (Spatial Working Memory), and planning (Stockings of Cambridge) relative to the contrast group. The validity of these comparisons depends on the assumption of similar rates of development for both the control variable (receptive vocabulary) and the target variable (executive functioning). No data are available to address this assumption for executive function, but Mervis and Klein-Tasman [2004] and Mervis and Robinson [2005] have shown that it does not hold for a variety of other control and target variables. If the assumption is not valid, then the prediction that two groups with identical raw scores on the BPVS but very different CAs should be expected to have similar scores on tasks of executive functioning is incorrect.

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John and Mervis [2010] used parent-report measures to consider the relation between sensory modulation impairments and executive functioning. Cluster analysis identified two clusters of children with WS varying in terms of sensory symptom severity (mild abnormalities versus more severe abnormalities), with parent-reported executive functioning ability [based on the Behavior Rating Inventory of Executive Functioning (BRIEF); Gioia et al., 2000] accounting for the largest proportion of between-cluster differences (46%). Children in the severe sensory modulation impairment group were reported to have significantly more difficulty than children in the mild sensory modulation impairment group on transitioning between activities, appropriate modulation of emotional responses, initiating a task or activity, staying on task, using working memory, anticipating future events/setting goals, and monitoring their own behavior/performance. On all these activities, mean level of performance was in the clinical range for the severe sensory impairment group. Mean level of performance for the mild sensory impairment group (although significantly better than for the severe-sensory impairment group) also was in the abnormal range for the metacognitive skills (working memory, planning, monitoring).

ADAPTIVE BEHAVIOR

An important aspect of development that brings together contributions from both cognition and personality is adaptive behavior. As defined by the American Association on Intellectual and Developmental Disabilities (AAIDD), adaptive behavior refers to “the conceptual, social, and practical skills that people have learned to be able to function in their everyday lives [AAIDD website, 2010].”

In addition, adaptive ability can be considered to be the extent to which a person functions, maintains independence, and demonstrates the social responsibility expected of individuals in his or her age and cultural group [Cicchetti and Pogge-Hesse, 1982; AAMR, 1992]. As such, examination of adaptive behavior performance in individuals with WS provides insight into the impact of WS on real-world functioning [Mervis et al., 2001].

Most studies of the adaptive behavior of children with WS have used the parent-interview form of the Vineland Adaptive Behavior Scales [VABS; Sparrow et al., 1984]. Gosch and Pankau [1997] compared the performance of children with WS to a CA- and IQ- matched group with nonspecific ID and found that overall the

nonspecific ID group performed significantly better than the WS group. The authors hypothesized that the difference was primarily due to the large number of items that required fine motor skills, an area of particular weakness for children with WS. Greer et al. [1997] examined the performance of children with WS aged 4–18 years and found that standard scores on the Socialization and Communication scales were significantly higher than standard scores on the Daily Living Skills or Motor Skills scales. Using a larger sample of participants with WS over a narrower age range (4–8 years), Mervis et al. [2001] found significant differences in standard scores between all pairs of scales. Performance was ordered as follows: (1) Socialization, (2) Communication, (3) Daily Living Skills, and (4) Motor Skills. In this study, VABS composite standard score was not related to CA. However, Fisch [2010], in a study of 34 children with WS aged 4–15 years, found a significant negative correlation ($r = -0.65$) between CA and VABS composite standard score, with the regression line suggesting a standard score decline of 25 points between ages 4 and 15 years.

In Table I, we report adaptive behavior performance for 122 children with WS aged 4–17 years, using a different parent-interview measure, the Scales of Independent Behavior—Revised [Bruininks et al., 1996]. This measure includes four scales: Motor Skills, Social Interaction and Commu-