LANGUAGE DEVELOPMENT IN INFANTS,

CHILDREN AND ADULTS WITH WILLIAMS SYNDROME

Annette Karmiloff-Smith[1] and Debbie Mills[2]

[1]Institute of Child Health, London

[2]Emory University

Introduction

Chance genetic events sometimes give rise through development to interesting cognitive-level outcomes. Williams syndrome (henceforth WS), a neurodevelopmental disorder, is one such case. At first blush, individuals with WS seem to have intact language alongside IQs in the 50-60s range. Does this mean that language has developed independently of intelligence and that the existence of WS supports Nativist views concerning the innate pre-specification of language? In this entry we note that, despite the understandable initial excitement about the ostensibly intact linguistic capacities of this clinical group, in fact their language development follows an atypical pathway from infancy through to adulthood. Moreover, the Nativist literature frequently misrepresents the empirical findings about WS language by treating *relative* strengths as *absolute* strengths, i.e., intactness of function (see, also, Innate Knowledge, this volume). Here we present evidence to demonstrate how WS language processes actually display subtle impairments (Karmiloff-Smith, Brown, Grice & Paterson, 2003) and cannot serve to segregate the WS cognitive system into parts that develop normally and independently of parts that develop atypically (see, also, Modularity of mind and language, this volume).

It should be recalled that such adult patients had normal development prior to their brain insult. In the case of developmental disorders, by contrast, subtle initial impairments may affect numerous parts of the brain as it develops. One cannot simply assume that deficits in the phenotypic outcome are the same as those apparent in the infant start state (Paterson, Brown, Gsodl, Johnson, & Karmiloff-Smith, 1999). Indeed, even in the normal case, ontogenetic development plays an important role over time. Infants do not start life with the same brain structure as adults. Ontogenesis progressively gives rise to adult specialisation and localisation of function. Thus, higher-level specialisations such as morphology, syntax, the lexicon and pragmatics are likely to be the emergent product of development rather than its starting state (Karmiloff-Smith, 1992).

Williams syndrome

Williams syndrome occurs in approximately 1 in 20,000 live births and is caused by the deletion of some 26 genes on one copy of chromosome 7 (Donnai & Karmiloff-Smith, 2000). Clinical features include several physical abnormalities that are accompanied by mental retardation and a specific personality profile. The interest in WS to neuroscientists, linguists and psycholinguists stems from the syndrome's very uneven profile of cognitive abilities, with spatial and numerical cognition seriously impaired, while language, social interaction and face processing seem surprisingly proficient for a clinical population with a mean IQ of 56 (Bellugi, Wang & Jernigan, 1994; Udwin & Yule, 1991). Research by Bellugi and her collaborators first drew attention to the potential theoretical interest to psycholinguistics of these seeming dissociations in the

WS phenotype (Bellugi, Sabo & Vaid, 1988). Surprising proficiency with language was shown to co-exist with serious problems on non-verbal tasks, in particular those calling on visuo-spatial processing. However, as researchers subsequently examined in more depth the linguistic profile of individuals with WS, it became apparent that the static notions of "intactness" and of "selective dissociations" needed to be replaced with a developmental notion of "atypically developing trajectories" over time (Karmiloff-Smith, 1998; Karmiloff-Smith, Scerif & Ansari, 2003; Karmiloff-Smith, 2004). We focus here on how the linguistic trajectory in WS develops, arising initially from the unusual communicative and cognitive profile in the development of infants and toddlers with the syndrome.

Precursors to language in infants and toddlers with WS

Much of the initial research on WS language focused on older children and adults, with little known about the early stages. More recent studies from four different labs in the UK, USA, and Italy have pinpointed a serious delay in the onset of WS language (Mervis & Bertrand 1997; Nazzi, Gopnik & Karmiloff-Smith, in press; Paterson et al., 1999; Singer Harris, Bellugi, Bates, Jones, & Rossen, 1997; Vicari, Carlesimo, Brizzolara & Pezzini, 1996; Volterra, Capirci, Pezzini, Sabbadini & Vicari, 1996). It is not uncommon to find WS children of 4 or 5 years of age who produce levels of language typical of 18-24 month olds. Is the initial delay in getting WS language off the ground merely the late maturation of a set of language-specific genes, or are there more complex *developmental* reasons?

In our view, several cognitive and linguistic factors interact developmentally to cause the initial delay that affects the subsequent deviance in WS language development (see Developmental relationship between language and cognition, this volume). Our studies show that infants with WS are some 10-20 months behind their typical control infants in segmenting words out of the speech stream (Nazzi, Paterson, & Karmiloff-Smith, 2003). This early difficulty, solved by typically developing infants at around 8 months, contributes to the WS delay (see, also, Infancy: phonological and prosodic development, this volume). Furthermore, despite their abilities with dyadic interaction, infants and toddlers with WS are surprisingly atypical in triadic interaction and in their understanding of the referential function of pointing (Mervis & Bertrand, 1997; Laing, Butterworth, Ansari, Gsodl, Longhi, Panagiotaki, Paterson & Karmiloff-Smith, 2002), one of the ways in which children normally learn new words. In addition, while they behave like controls in mapping perceptual similarities between objects, toddlers with WS are significantly poorer than controls at using linguistic labels to map identity of object categories (Nazzi & Karmiloff-Smith, 2002).

Language in older children and adults with WS

It could be that the initial delay in language onset in WS is followed by a normal developmental trajectory, in other words that older children simply "catch up". This does not appear to be the case, however. Both behavioural and brain imaging studies of WS language processing make it empirically questionable as to whether any aspect of language - syntax, lexical-semantics, phonology, pragmatics, or discourse (see also Phonological, lexical, syntactic and semantic disorders in children, this volume) - is intact in WS, despite many claims to the contrary (e.g., Clahsen & Almazan, 1998; Pinker, 1999). For example, event-related potential (ERP, see also Evoked potentials, brain imaging of language processing, this volume) studies suggest that brain activity to semantic, syntactic, and sensory features of spoken language are abnormally organized in adolescents and adults with WS. In normal adults and typically developing children of 9-years and older, ERPs to grammatical function words, e.g., articles, prepositions, conjunctions, show marked differences in latency, amplitude and distribution, from those to semantic content words, e.g. nouns, verbs and adjectives, (Neville, Coffey, Holcomb, & Tallal, 1993). Grammatical function words elicit a left anterior negativity early in the waveform, whereas content words elicit ERPs that peak somewhat later, are largest over posterior regions, and are bilateral or larger over the right than the left hemisphere. ERP differences to function words and content words have even been observed in infants as young as 20-months of age (St. George & Mills, 2001). In contrast, adolescents and adults with WS do not show ERP differences to function and content words, nor do they show left greater than right ERP asymmetries to the grammatical function words (Bellugi, Lichtenberger, Mills, Galaburda & Korenberg, 1999; Bellugi, Mills, Jernigan, Hickok & Galaburda, 1999; Mills, 1998; Neville, Mills & Bellugi, 1993, 1995; St. George, Mills, & Bellugi, 2000). Additionally, WS brain activity to words appears to be abnormally organized even at the level of sensory processing of the words. In WS, ERPs within the first 200 ms of the onset of the word show a unique pattern of components, with abnormally large bilateral positivities at 100 and 200 ms, that are not observed in a similar configuration at any age in normal development (Bellugi, et al., 1999b; Korenberg, Bellugi, Mills & Reiss, 2003; Neville, Mills & Bellugi, 1995). These components are thought to be generated in primary auditory cortex and may be related to a disproportionally large superior temporal gyrus (STG) and abnormal cell packing density in this region in WS (Hickok, Neville, Mills, Jones, Rossen & Bellugi, 1995; Holinger, Mills, Bellugi, Korenberg, Reiss, Sherman & Galaburda, in press; Galaburda, Holinger, Mills, Reiss, Korenberg & Bellugi, 2003). Of particular interest is a report by Mills, Llamas and Doyle (2003) showing that although adults with WS show abnormally organized brain activity to language, infants with WS display a normal pattern of asymmetries until about 3 years of age. However, by 3-4 years ERPs to words begin to resemble the highly atypical pattern displayed by older children and adults with WS. Three years of age is also the time at which infants with WS show a marked increase in vocabulary development. This suggests that compensatory neural mechanisms linked to relative proficiencies in language processing may emerge at this point in development. Taken together, these studies provide neurobiological evidence against the notion of an intact, normally organized module for language processing in WS.

Despite growing evidence counteracting the claim for the intactness of language in WS, linguists of a Chomskyan persuasion have tried to identify at least one aspect of WS language that□s spared. For example, in a study comparing WS and Specific Language Impairment (SLI), Clahsen and Almazan (1998) argued for a double dissociation of innate mechanisms (see Specific Language Impairment, this volume). Their assertion was that lexical memory is impaired in WS whereas syntax is intact, and that the opposite holds for SLI. Specifically, they claimed that individuals with WS display a deficit in forming irregular past tenses (e.g., keep-kept) but intact performance in forming the regular past tense (e.g., talk-talked) (see Tense, this volume). But this failed to hold when a broader, in-depth study of WS past tense formation was carried out on a much larger population (Thomas, Grant, Barham, Gsodl, Laing, Lakusta, Tyler, Grice, Paterson & Karmiloff-Smith, 2001). Indeed, it is not sufficient to demonstrate in any group that irregular past tense formation is poorer than regular past tense formation, because this is also true of some stages of typical development. Rather, it is crucial to demonstrate that the level of past tense formation is poorer than would be expected in WS for their actual level of language development. Our study showed that when verbal mental age was controlled for, the WS group displayed no selective deficit in irregular past tense formation and that their performance could be placed on the typical developmental pathway found in younger subjects. The results were more consistent with the hypothesis that the WS language system is delayed because it has developed under different constraints. Mervis and collaborators (e.g., Klein & Mervis, 1999) have also concluded that the best way to characterize WS language, even in adulthood, is that it has not developed normally and reveals patterns typical of much younger children.

Not only is WS language delayed, but it is becoming increasingly clear that the WS language system develops along a different trajectory compared to controls, relying less on semantics than in the typically developing children (Mervis & Bertrand, 1997; Thomas & Karmiloff-Smith, 2003). Although local semantic organization looks normal in WS in terms of priming effects (Tyler, Karmiloff-Smith, Voice, Stevens, Grant, Udwin, Davies & Howlin, 1997) and category fluency (Scott, Mervis, Bertrand, Klein, Armonstrong & Ford, 1995), global semantic organization remains at the level of young children and never reaches the mature state, even in relatively high functioning adults with WS (Johnson & Carey, 1998). Further studies of oral and written language also point to a reduced contribution of semantics in WS language development. For example, Karmiloff-Smith and collaborators found that when participants with WS monitor sentences for a target word, semantic information becomes available too slowly to be integrated with the real-time processing of syntax (Karmiloff-Smith, Tyler, Voice, Sims, Udwin, Davies & Howlin, 1998; see, also, Sentence Processing, this volume). A recent study of reading also pointed to atypical processing, showing that whereas WS participants displayed equal levels of reading for both concrete and abstract words, the controls found concrete, imageable words much easier to read (Laing, Hulme, Grant & Karmiloff-Smith, 2001). Finally, in a non-word repetition study, participants with WS, despite a vocabulary test age of 8, behaved at the level of 4-5 year olds and were significantly less constrained by resemblance with meaningful words when repeating new words (Grant, Karmiloff-Smith, Gathercole, Paterson, Howlin, Davues & Udwin, 1997). In sum, like very young children, the participants with WS were less influenced by the semantics of the words that the nonce terms resembled and relied more on phonology. Taken together, these different studies suggest that, unlike typical development, semantics seems to place less of a constraint compared to phonology (Laing Grant, Thomas, Parmigiani, Ewing, & Karmiloff-Smith, in press) in the way in which WS language develops over time.

The above discussion concerns lexical-semantic development in WS, but it remains a theoretical possibility that WS syntax is intact (e.g., Clahsen & Almazan, 1998; Pinker 1999). There are, however, a number of lines of evidence to doubt this. First, vocabulary levels are usually better than syntactic levels, with both significantly below chronological age (Karmiloff-Smith et al., 1997). Second, even in simple imitation tasks, participants with WS show significant deficits with complex syntactic structures like embedded relative clauses (Grant, Valian & Karmiloff-Smith, 2000). Furthermore, even in simple grammatical concord in French, acquired easily by very young children, individuals with WS display serious deficits (Karmiloff-Smith, Grant, Berthoud, Davies, Howlin & Udwin, 1997). The same applied in a study of Italian grammatical gender in which the WS group produced errors encountered at no age in typical development (Volterra, et al., 1996). Several studies (e.g., Klein & Mervis, 1999) make it clear that the real problems that people with WS experience with semantics and syntax are often superficially camouflaged by their good verbal memory.

It is in our view theoretically misleading and empirically inaccurate to claim that grammar is spared in this clinical population. Although WS grammar is indeed relatively proficient compared to some other clinical groups and relatively good compared to WS spatial deficits, most studies show that it is in fact no better than their mental age would predict. This does not mean that the WS cognitive architecture is uninteresting. On the contrary. There is in WS a mixture of delay, deviance and asynchronies across the developing system. Researchers still need to understand why the language of people with WS language is initially so delayed and why, despite this, they develop surprising proficiency by adulthood compared to many other genetic syndromes with equal delay in early development. **Text = 2,367**

Acknowledgements

The writing of this paper was partially supported by NIH Grant No.

R21TW06761-01.

References

Bellugi, U.,Sabo, H., Vaid, J., (1988). Spatial deficits in children with Williams Syndrome. In J. Stiles-Davis, M. Kritchevsky, & U. Bellugi (Eds.) Spatial cognition: Brain bases and development. pp. 273-298. New Jersey: Lawrence Erlbaum.

Bellugi, U., Lichtenberger, L., Mills, D., Galaburda, A. & Korenberg, J. (1999a). Bridging cognition, brain and molecular genetics: Evidence from Williams Syndrome. Trends in Neuroscience,□22, 193-236.

Bellugi, U., Mills, D.L., Jernigan, T., Hickok, G., & Galaburda, A.□(1999b). Linking cognition, brain structure and brain function in Williams Syndrome. In H. Tager-Flusberg (Ed.) Neurodevelopmental Disorders: Contributions to a new framework from the cognitive neuroscience, pp.111-136, Cambridge, MA: MIT Press.

Bellugi, U., Wang, P. P.,Jernigan, T. L. (1994) Williams syndrome: An unusual neuropsychological profile. In S. Broman, S. & J. Grafman, (Eds). Atypical cognitive deficits in developmental disorders: Implications for brain function. pp. 23-56. New Jersey: Lawrence Erlbaum.

Clahsen, H., & Almazan, M. (1998). Syntax and morphology in Williams syndrome. Cognition, 68, 167-198.

Donnai, D., & Karmiloff-Smith, A. (2000) Williams syndrome: From genotype through to the cognitive phenotype. American Journal of Medical Genetics: Seminars in Medical Genetics, 97 (2), 164-171.

Galaburda, A., Holinger, D., Mills, D., Reiss, A., Korenberg, J. & Bellugi, U. (2003). El syndrome de Williams. Un resumen de hallagos cognitivos, delectrofisiologicos, anatomofunctionales, microanatomicos y geneticos. Rev. Neurol, 36(Sup 1): S132-137.

Grant, J., Karmiloff-Smith, A., Gathercole, S., Paterson, S., Howlin, P., Davies, M., & Udwin, O. (1997) Verbal Short-term Memory and its relation to Language Acquisition in Williams Syndrome. Cognitive Neuropsychiatry, 2, 2, 81-99.

Grant, J., Valian, V., & Karmiloff-Smith, A. (2002) A study of relative clauses in Williams syndrome. Journal of Child Language, 29, 403-416.

Hickok, G., Neville, H. J., Mills, D., Jones, W., Rossen, M.L., & Bellugi, U. (1995). Electrophysiological and quantitative MR analysis of the cortical auditory system in Williams syndrome. Cognitive Neuroscience Society, Annual Meeting Abstracts, Supplement to the Journal of Cognitive Neuroscience, 2, 66.

Holinger, D. P., Mills, D. L., Bellugi, U., Korenberg, J.R. Reiss, A. L., Sherman, G. F. Galaburda, A. M. (in press). Relative sparing of primary auditory cortex in Williams Syndrome. Brain Research.

Johnson, S. C.; Carey, S. (1998). Knowledge enrichment and conceptual change in folkbiology: Evidence from Williams syndrome. Cognitive Psychology, 37, 156-200.

Karmiloff-Smith, A. (1992) Beyond Modularity: A Developmental Perspective on Cognitive Science. Cambridge, Mass.: MIT Press/Bradford Books.

Karmiloff-Smith, A. (1998) Development itself is the key to understanding developmental disorders. Trends in Cognitive Sciences, 2, 10, 389-398.

Karmiloff-Smith, A., Brown, J.H., Grice, S., & Paterson, S. (2003) Dethroning the myth: Cognitive dissociations and innate modularity in Williams syndrome. Developmental Neuropsychology, 23 (1&2), 229-244.

Karmiloff-Smith, A., Grant, J., Berthoud, I., Davies, M., Howlin, P., & Udwin, O. (1997) Language and Williams Syndrome: How Intact is "Intact"? Child Development, 68, 2, 246-262.

Karmiloff-Smith, A. Scerif, G., & Ansari, D. (2003) Double dissociations in developmental disorders? Theoretically misconceived, empirically dubious. Cortex, 39, 161-163.

Karmiloff-Smith, A., Thomas, M., Annaz, D., Humphreys, K., Ewing, S., Brace, N., van Duuren, M., Pike, M., Grice, S., & Campbell, R. (2004) Exploring the Williams

Syndrome Face Processing Debate: The importance of building developmental trajectories. Journal of Child Psychology and Psychiatry, 45, 7, 1258-1274.

Karmiloff-Smith, A., Tyler, L.K., Voice, K., Sims, K., Udwin, O., Davies, M., & Howlin, P. (1998) Linguistic Dissociations in Williams Syndrome: Evaluating Receptive Syntax in on-line and off-line tasks. Neuropsychologia, 36, 4, 342-351.

Klein, B. P.; Mervis, C. B. (1999). Contrasting patterns of cognitive abilties of 9- and 10-year-olds with Williams Syndrome or Down Syndrome. Developmental Neuropsychology, 16, 177-196.

Korenberg, J,R., Bellugi, U, Mills, D L., & Reiss, A. L.(2003) Williams Syndrome: A Neurogenetic Model of Human Behavior. ., In D. N. Cooper (Ed.) Encyclopedia of the Human Genome , Vol.5 pp: 757-766. London: Nature Publishing Group.

Laing, E., Butterworth, G., Ansari, D., Gsödl, M., Longhi, E. Panagiotaki, G., Paterson, S., & Karmiloff-Smith, A. (2002)□Atypical development of language and social communication in toddlers with Williams syndrome. Developmental Science, 5: 233-246.

Laing, E., Grant, J., Thomas, M., Parmigiani, C., Ewing, S., & Karmiloff-Smith, A. (in press)□Love is...an abstract word: The influence of lexical semantics on verbal short-term memory in Williams syndrome.□Cortex.

Laing, E., Hulme, C., Grant, J., & Karmiloff-Smith, A. (2001) Learning to Read in Williams Syndrome: Looking Beneath the Surface of Atypical Reading Development. Journal of Child Psyhcology and Psychiatry, 42, 6, 729-739.

Mervis, C.B., & Bertrand, J. (1997). Developmental relations between cognition and language: Evidence from Williams Syndrome. In L.B. Adamson & M.A. Romski (Eds.) Research on Communication and Language Disorders: Contributions to Theories of Languge Development. pp 75-106. New York: Brooks,

Mills, D.L. (1998) Electrophysiological markers of abnormal brain function in Williams Syndrome. Cognitive Neuroscience Society, Annual Meeting Abstracts. Supplement of the Journal of Cognitive Neuroscience, (Abstract), 10.

Mills, D.L., Llamas, T. and Doyle, T. (2003) Brain and language development in infants and children with Williams Syndrome. Poster presented at: The Society for Research in Child Development, Biennial meeting, Tampa Florida, April 2003.

Mills, D.L., Llamas, T., St.George, M., Doyle, T., Neville, H., Bellugi, U. & Korenberg, J. (Tech report, 2003). Electrophysiological signatures of abnormal auditory language processing in infants, children and adults with Williams Syndrome.

Nazzi, T., Gopnik, A., & Karmiloff-Smith, A. (in press) Asynchrony in the cognitive and lexical development of young children with Williams syndrome. Journal of Child

Language.

Nazzi, T., Paterson, S., & Karmiloff-Smith, A. (2003) Early word segmentation by infants and toddlers with Williams syndrome. Infancy, 4, 2, 251-271.

Nazzi, T., & Karmiloff-Smith, A. (2002) Early categorization abilities in young children with Williams syndrome. NeuroReport, 13, 10, 1259-1262.

Neville, H.J., Coffey, S.A., Holcomb, P.J., & Tallal, P. (1993). The neurobiology of sensory and language processing in language impaired children. Journal of Cognitive Neuroscience, 5, 235-253.

Neville, H. J., Mills, D. L. & Bellugi, U. (1993). Effects of Altered Auditory Sensitivity and Age of Language Acquisition on the Development of Language-Relevant Neural Systems: Preliminary Studies of Williams Syndrome. In S. Broman (Ed.), Atypical Cognitive Deficits in Developmental Disorders: Implications for Brain Function. Hillsdale, NJ: Lawrence Erlbaum Associates, 67-83.

Neville, H., Mills, D., & Bellugi, U. (1995) Functional Brain Organization in Williams Syndrome. (Abstract), Special Issue, Genetic Counseling, 6, 141-142.

Paterson, S.J., Brown, J. H., Gsödl, M. K., Johnson, M. H., & Karmiloff-Smith, A.(1999) Cognitive Modularity and Genetic Disorders. Science, 286, 5448, 2355-

2358.

Pinker, S. (1999). Words and rules. London: Weidenfeld & Nicolson

Scott, P., Mervis, C., B., Bertrand, J., Klein, B.P., Armstrong, S.C. & Ford, A.L. (1995) Semantic organization and world fluency in 9- and 10-year-old children with Williams Syndrome. Genetic Counselling, 655, 172-173.

Singer Harris, N.G., Bellugi, U., Bates, E., Jones, & Rossen, M. L (1997). Contrasting profiles of language development in children with Williams and Down Sundromes. In D. J. Thal & Reilly (Eds.) Special Issue: Origins of Language Disorders, Developmental Neuropsychology, 13, 345-370.

St. George, M. & Mills, D.L. (2001). Electrophysiological studies of language development. In J. Weissenborn and B. Hoehle (Eds). Language Acquisition and Language Disorders. John Benjamins Publishing Co: Amsterdam: The Netherlands, 247-259.

St. George, M. Mills, D. and Bellugi, U. (2000). ERPs during auditory language comprehension in Williams Syndrome: The effects of word frequency, imageabilty and length on word class. NeuroImage, 11 (5), 357.

Thomas, M., Grant, J, Barham, Z., Gsödl, M., Laing, E., Lakusta, L, Tyler, L.K., Grice, S., Paterson, S., & Karmiloff-Smith, A. (2001) Past tense formation in Williams syndrome. Language and Cognitive Processes, 2, 16, 143-176

Thomas, M., & Karmiloff-Smith, A. (2003) Modelling language acquisition in atypical phenotypes. Psychological Review. 110(4):647-682.

Tyler, L.K., Karmiloff-Smith, A., Voice, K., Stevens, T., Grant, J., Udwin, O., Davies, M., & Howlin, P. (1997) Do individuals with Williams syndrome have bizarre semantics? Evidence for lexical organization using an on-line task. Cortex, 33, 515-527.

Udwin, O., & Yule, W. (1991). A cognitive and behavioral phenotype in Williams syndrome. Journal of Clinical and Experimental Neuropsychology, 13, 232-244.

Vicari, S., Carlesimo, G., Brizzolara, D., & Pezzini, G. (1996). Short-term memory in children with Williams syndrome: A reduced contribution of lexical-semantic knowledge to word span. Neuropsychologia, 34, 919-925.

Volterra, V., Capirci, O., Pezzini, G., Sabbadini, L., & Vicari, S. (1996). Linguistic abilities in Italian children with Williams syndrome. Cortex, 32, 663-677.