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Williams Syndrome and language development

Williams syndrome (WS) is a rare genetic disorder (around 1 in 20 000 live births) characterised by learning disability and an uneven cognitive profile. It is relevant to theories of language development because language acquisition is a relative strength in the disorder.

In the late 1980s, it was thought that it might demonstrate the independent genetic underpinnings of language and general cognition, i.e., that language could indeed develop normally in the face of impaired general cognition. Subsequently, twenty-five years of research suggest that this is probably not the case. While language in WS does look impressive compared to some other developmental disorders that have particular language problems perhaps relating to poor phonology (such as Down syndrome [DS] and Specific Language Impairment [SLI]), it is best characterised by delay, with most linguistic abilities not markedly different from overall mental age. There may be subtle patterns of atypicality, some of which may be connected with other aspects of the WS cognitive profile, including a 'hypersociable' personality and weaknesses in visuospatial cognition. Research seeking to understand how the genetic mutation that causes WS results in the characteristic cognitive profile (via effects on brain development and cognitive development) is still ongoing.

WS is caused by a spontaneous deletion of around 28 genes from one copy of chromosome 7, resulting in a range of physical anomalies, including a characteristic 'elfin' facial dysmorphology, as well as a characteristic uneven cognitive profile, with relative strengths in language, social skills, and face processing, and relative weaknesses in visuospatial cognition, motor skills, and problem solving, where 'relative' means 'compared to overall mental age as assessed by a standardised test battery'. WS was first described in the early 1960s but did not receive significant attention from developmental psychologists and linguists until the late 1980s. Initial studies by Ursula Bellugi and colleagues indicated that, while individuals exhibited mild to moderate learning difficulties, they had no problems with understanding passive sentences, negation or conditionals, suggesting that syntactic abilities might be normal in WS. An analysis of spontaneous speech showed that the language in WS included well-formed and complex structures despite low levels of IQ. These initial findings led Steven Pinker to contrast WS with SLI – a familial developmental disorder with impaired language development but nonverbal cognitive skills in the normal range – and conclude that the two demonstrated a 'genetic double dissociation': while WS demonstrates normal development of language but not cognition, SLI shows the opposite pattern with normal development of cognition but not language. He concluded that together the disorders provided evidence supporting the thesis that there was a specific genetic basis for human language development separate from general cognition.

Since then, two decades of research on language and cognitive development in WS has not supported these claims (nor indeed the specificity of language impairment in SLI). Language in WS represents a complex developmental pattern, with fractionation (unevenness of development) throughout, and marked variability between individuals with the disorder. Receptive vocabulary, as measured by tests such as the Peabody Vocabulary Test and the British Picture Vocabulary test, does represent a particular strength of individuals with the disorder, but even this ability is rarely at a level predicted by chronological age. The high scores may in part reflect the nature of these tests of language ability (e.g., pick one of four pictures that goes with the target word). Apart from receptive vocabulary, most language abilities are more in line with overall mental age, and thus represent a pattern of delayed development.

Nevertheless, some anomalies have been identified. The earliest stages of language acquisition in WS show a delay in onset of perhaps two years. When language begins to emerge in toddlers with WS, some of the associated precursors appear atypical. For example, in contrast to typical development pointing does not precede naming in WS. Vocabulary development does not appear to have the same relationship with markers of developing semantic knowledge (such as the ability to sort objects) or with gesturing. Toddlers with WS also show differences in shared attention, in particular the way that they attend to caregivers during labelling situations (e.g., when the caregiver says 'look at the doggie', instead of switching her attention back and forth from the caregiver to the dog, the child with WS tends to continuously fixate the caregiver's face). Together, these point to possible differences in the early developmental pathways for WS language acquisition.

Language development in WS accelerates in later childhood and early adolescence, where the sometimes-impressive facility with vocabulary can be observed (compared to mental age). However, it is not always clear that the individual fully understands the meaning of the figurative expressions and certain words he or she is using, rather relying on a good memory for phrases and an approximate understanding of meaning in their usage.

One particular research focus has been on the development of grammar in WS, with initial claims that it develops normally (albeit delayed). Several studies investigated morphology, contrasting performance on regular versus irregular inflectional paradigms in a quest to identify stronger performance on rule-based rather than irregular inflections, compared to children matched on overall language ability or on overall mental age. The evidence here proved mixed, with no clear demonstration of selective deficits. Studies of syntax also yielded mixed results, with for example, one study showing the presence of core syntactic relations in the comprehension of conditionals (scope and c-command), while other studies uncovered deficits in the processing of relative clauses and passive constructions. It is worth pointing out that in the study examining conditionals, the group of individuals with WS had a mean age of 16 years and were scoring slightly more poorly than typically developing 6-year-olds. This type of comparison undermines the initial claims of Pinker that syntax might be

developing *independently* of general cognition in WS – that is, at a level predicted by chronological age.

Researchers have engaged in a number of other debates in the field. There is disagreement about whether the cognitive system in WS (and the language system in particular) should be explained in terms of the normal language system (advocated by researchers such as Harold Clahsen, Helen Tager-Flusberg, and Barbara Landau), or whether such a theoretical framework does not provide sufficient scope to depict the subtle developmental anomalies revealed by some studies (advocated by researchers such as Annette Karmiloff-Smith and Michael Thomas). In other words, there is still debate on *how atypical the WS language system is*.

In another debate, two competing hypotheses have been evaluated for the best explanation of language in WS. The *Semantics-Phonology Imbalance hypothesis* argues that while relatively strong, language functions in a subtly atypical way: there might be greater emphasis on the sounds of words (phonology) and less emphasis on their precise meaning (semantics). The *Conservative hypothesis* argues that language ability is in line with mental age and any observed anomalies arise indirectly from other characteristics of the disorder, such as the spatial processing deficit that causes problems in, for example, learning certain spatial prepositions (*in*, *on*, *under*), and the hypersociable personality profile that may lead these individuals to use language strategically to capture and maintain attention in social interactions. A review by Jon Brock in 2007 suggested that research was favouring the Conservative over the Imbalance hypothesis, but this question has yet to be definitively resolved.

Clinically, since language acquisition is a relative strength in WS, maximising language ability would enable it to serve as a scaffold for intervening on weaker skills, and to develop compensatory strategies (e.g., verbal strategies to address difficulties in, for example, navigation skills).

Overall, WS is notable for the relative strength of language development compared to other skills, but since this strength is relative to mental age rather than chronological age, WS cannot provide support for the independence of language development from cognition. Moreover, language acquisition in WS has to be viewed in the context of other strengths and weaknesses in the cognitive profile, and reveals many subtle anomalies when examined in detail.

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See also

Assessment of language abilities; Conceptual foundations of early word learning, Domain specificity in language development; Genetic basis of language development and impairment; Genetic syndromes and language development, Grammatical development in children with language impairments; Joint attention and language development; Metaphor (development of); Non-literal language use (development of); Phonological development in children with language impairments; Semantic development in children with language impairments

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