Williams Syndrome: Dissociation and Mental Structure

Mitch Parsell (mitch.parsell@mq.edu.au)
Human Sciences, Macquarie University
Sydney NSW 2109 Australia

Abstract

Williams syndrome (WS) is a genetic disorder that, because of its unique cognitive profile, has been marshalled as evidence for the modularity of both language and social skills. But emerging evidence suggests the claims of modularity based on WS have been premature. This paper offers an examination of the recent literature on WS. It argues the literature gives little (if any) support for mental modularity. Rather than being rigidly modular, the WS brain is an extremely flexible organ that that co-opts available neural resource in a highly dynamic manner to cope in the world.

Keywords: Williams syndrome; modularity; social cognition.

Introduction

If you aim to understand the structure of the human mind you face a substantial methodological hurdle at the very outset: namely, mental processes are not directly observable. This brings difficulties at the most fundamental level. It means, for example, that even individuating mental processes is extremely demanding. One way to attack the problem of individuation is with the logic of dissociation. If you can show that some intervention impacts cognitive ability A1, but not A2, you seem to have reason for supposing A1 and A2 are independent abilities. Different domains of cognitive science favour different interventions. Neuropsychologists tend to favour impairments. They look to atypical brains to provide information about the associations between cognitive deficits and the dissociations between cognitive skills.

For neuropsychological impairment, the basic notion of dissociation is straightforward: if ability A1 is impaired while other abilities (A2,N) remain intact, A1 is said to dissociate for A2,N. For example, if some lesion is found to impact face recognition only, face recognition is said to dissociate from other cognitive abilities. Notice it is essential that A1 is selectively impaired. If all (or a substantial range of) abilities are impaired, then we have no dissociation. Thus dissociative evidence relies not merely on the presence of deficits, but the co-existence of deficits with preserved abilities. It relies on patterns of spared and impaired abilities. When two patterns mirror each other, we have a double dissociation. Specifically, if A1 can be impaired independent of A2 and A2 can be impaired independent of A1, then A1 and A2 form a double dissociation. If, for example, face recognition can be impaired without impacting object recognition and object recognition can be impaired independent of a deficit to face recognition, then face recognition and object recognition form a double dissociation.

Double dissociations can hold between individuals or between entire atypical populations. The literature on semantic processing is rich with individuals who form double dissociation pairs for particular concepts. See, for example, Samson and Pillon (2003) who examine individual patients that appear to demonstrate double dissociations for the concept “animal” versus the concept “fruit and vegetable.” The literature on atypical populations also has abundant claims of double dissociations. The population of individuals with Williams syndrome (WS) has attracted increasing attention in this regard.

WS is a genetic disorder that results from a disruption to a contiguous section of approximately 26 genes in the 7q11.23 region on chromosome 7 (see Francke, 1999; Peoples et al. 2000). It is extremely rare, reported to occur in between 1 per 5 500 and 1 per 50 000 live births (see Wang et al. 1997; Strømme, Bjørnstad & Ramstad, 2002). The disorder has well-documented behavioural and cognitive effects. People with WS have general intelligence in the range of 50-65 IQ points, specific difficulties with arithmetic, spatial reasoning, planning and problem solving, while auditory and music skills, language, face recognition and social abilities appear to remain relatively spared. This unique pattern of apparently spared and impaired abilities has seen WS recruited as evidence for mental modularity. Apparently preserved language coupled with general intellectual impairment seems to many especially significant (e.g. Bellugi et al. 2000). This has seen WS recruited as evidence for the modularity of language and especially syntactic processing (e.g. Pinker, 1994; 1999). Pinker relies heavily on the apparent double dissociation with individuals with Specific Language Impairment (SLI): ‘the genetic double dissociation is striking . . . . The genes of one group of children [SLI individuals] impair their grammar while sparing their intelligence; the genes of another group of children [WS] impair their intelligence while sparing their grammar ‘(Pinker, 1999, p. 262). For others, the pattern of impaired versus “spared” abilities recommends WS as evidence for a social reasoning module (e.g. Tager-Flusberg, Boshart & Baron-Cohen, 1998; Baron-Cohen, 1998). Still others have argued that preserved musical
abilities in WS individuals support the existence of an independent music module (Levitin & Bellugi, 1998).

But recent research has questioned whether it is legitimate to characterise language as preserved in WS. Language abilities once thought to be intact are now thought to be either significantly delayed or to follow atypical developmental trajectories (see Brock, 2007). On the one hand, comprehension, phrase repetition, mean utterance length and object categorisation appear to be delayed. On the other hand, phonological processing and morphology appear to follow an atypical developmental trajectory (see Martens et al. 2008, pp. 580-582). More complex grammatical skills (such as gender agreement, pragmatics and semantic fluency) seem to be both delayed and developmentally atypical (see Martens et al. 2008, p. 582). And if language is delayed or developmentally atypical (or both), then the argument from the WS population to mental modularity is jeopardised. And it is not just skills in the language domain that seem to have been previously overstated for the WS population. In all four domains once thought preserved, the latest research is suggesting the real story is more nuanced and complex than simple skill preservation.

In this paper, I look at the recent literature on WS. I argue the literature gives little (if any) support for mental modularity. The argument takes place over four sections that examine each of the cognitive abilities often claimed to be preserved in the WS population: namely, music, face recognition, language and social skills. I argue in each case there are significant challenges to using the WS cognitive profile to argue for independent cognitive modules. There is strong empirical evidence that the preserved music and face recognition abilities of the WS population are served by different cognitive resources than in the typically developmental (TD) brain. This means the necessary contrast with the TD brain cannot be made and the WS population cannot be used as one side of a double dissociation. Thus WS does not provide evidence of mental modularity. Recent evidence for language and social skills demonstrates that these abilities are not actually preserved. Obviously, if these skills are not preserved, then they cannot be used to ground dissociative claims and the arguments to mental modularity are again jeopardised. While my main purpose is negative I will offer a very broad-brush sketch of a positive account of mental architecture in WS.

---

2 It is important at the outset to be clear about the argument structure. My purpose is primarily negative: specifically, to counter arguments from WS to mental modularity. This is achieved by demonstrating that the assumptions required to licence arguments to modularity do not hold in the case of WS. More specifically, that in all four cases to be discussed the cognitive skills are either impaired or atypical. Hence the WS population does not provide evidence for mental modularity. Nevertheless, the mind may still be modular; the claim is merely that WS does not provide any support for modularity. In sum, my primary aim is neither to develop an alternative to nor arguments against modularity, but merely to remove one specific line of evidence for modularity.

3 Of course, even extreme modularists accept there is interaction between modules, but the interaction must remain shallow. Although I do not have time to rehearse the argument in detail, I have previously argued that modules are characterised by information encapsulation (see Parsell 2009; also see Fodor 1983, especially p. 122). Critically, encapsulation entails that any processing within a module is unavailable to the rest of the mind. For example, face processing cannot access earlier stages of the representation(s) delivered to it. Thus interaction between modules is shallow in that modules can trade outputs, but cannot penetrate each other to impact processing. The type of dynamic interaction I will point to in this paper is suggestive of deeper interaction then the shallow communication allowed between modules.

---

Music

Individuals with WS have often claimed to have preserved musical abilities (see Don, Schellenberg & Rourke, 1999; Levitin & Bellugi, 1998; 1999). Further, they have a particularly high musical drive: they tend to spend more time listening to music and demonstrate an elevated emotional response to music (Don et al. 1999; Levitin & Bellugi, 1999). Indeed, individuals with WS are often reported to have hyperacusis: a heightened sensitivity to all or some specific sounds. Udwin (1990), Klein et al. (1990) and Don et al. (1999) all report over 90% of WS individuals surveyed had a history of hyperacusis. More recently, Levitin et al. (2005) have looked closely at the response of 118 WS individuals to sound. They distinguish 4 types of abnormal reactions to sound:

(i) **Hyperacusis**: lowering of hearing threshold;
(ii) **Odynacusis**: lowering of pain threshold;
(iii) **Phobias** (auditory allodynia): aversion to or fear of certain sounds; and,
(iv) **Fascinations**: abnormal attraction to certain sounds.

True hyperacusis remains low at only 4.7% of the sample, whereas both odynacus and phobias are remarkable high in the WS sample: 79.8% and 90.6% respectively. The types of sounds reported to cause aversions in WS were similar to the other surveyed groups (namely, Down syndrome, autism and TDs). The most commonly cited sounds were fireworks,
engines, thunder and vacuum cleaners. Several unusual sounds were also reported to cause phobias in WS individuals including cicadas, cows, spraying of shaving cream and the saxophonist Kenny G. While fascination levels in WS individuals are low (9%), they are much higher than the levels reported by other surveyed groups: only one non-WS participant reported a fascination. Fascinations tended to be for broadband noises such as buzzing, humming and motor noises. Interestingly, the attraction to sounds felt by people with WS extends to fascinations with the objects themselves. Levitin et al. (2005) note many reports of collecting either the objects or depictions of the objects. One WS individual is reported to own 18 vacuum cleaners and can name the types from their sound alone.

What is the cause of these unusual sensitivities, phobias and, to a lesser extent, fascinations? The problem does not appear to reside in the peripheral auditory system. Levitin et al. (2005) note that there is no evidence of systematic auditory abnormalities in WS. They suggest rather an explanation based on heightened awareness and central auditory abnormalities. Bellugi et al. (1990) have provided direct empirical evidence of hyper-excitability at a cortical level in WS. Further, they suggest that different neurological pathways may well handle auditory processing in WS individuals. The WS brain appears to deal with auditory information differently than the TD brain. The auditory cell-packing density and neuronal size is abnormal in WS (Galaburda & Bellugi, 2000; Levitin et al. 2005). These abnormalities are consistent with hyper-connectivity (Galaburda & Bellugi, 2000). For Levitin et al. (2005) the evidence points towards organizational differences at both the micro and macro levels of the brain that may explain not merely the relative sparing of music, but the increased music drive and unusual reactions to sounds. Musical fascinations—the special emotional meaning associated with particular sounds and their associated objects—can be explained by supposing the auditory neurology co-opts or otherwise significantly interacts with emotional centres.

Levitin & Bellugi (1998) have argued that preserved musical abilities in WS individuals support the existence of an independent music module. Such an argument depends on the musical/auditory abilities of WS individuals being served by same cognitive resources as in the TD brain. If this does not hold the argument to modularity breaks down for it depends on selective impairment/preservation. Preservation of a skill (behaviour) due to compensation (alternative cognitive route) undermines selectivity. And this seems to be the case with the preserved musical abilities of the WS population. More generally, as Gerrans (2003) has emphasised, alternative routes are an significant open possibility for genetic deletions as the cognitive/neurological impact of the deletion can be compensated for in different ways over a protracted developmental history in which the brain is at its most plastic. As a consequence, the end state or behavioural phenotype of someone with a genetic defect may be very similar to that of a normal subject, but the developmental route, cognitively and neurally, may be very different (Gerrans 2003, p. 46). If the WS brain is developmentally atypical then it is illegitimate to characterise the WS brain as a TD brain with a pattern of specific preservation and deficits. But without selectivity the argument from preserved behavioural skills in the WS population to specific cognitive modules fails. This will become clearer in the next section where I discuss face recognition which also provides support for the existence of alternative routes.

**Face Recognition**

WS individuals have preserved face recognition abilities, despite having deficits in spatial cognition (see Bellugi et al. 2000). They are relatively good at interpreting socially significant facial cues. They can judge emotions from facial expression (Tager-Flusberg et al. 1998) and can infer intentions from eye-gaze data (Karmiloff-Smith et al. 1995). In a parallel to auditory input, faces seem to hold a particular attraction to people with WS. In infancy, individuals with WS show abnormal intense looking towards strangers (Mervis et al. 2003) and throughout life demonstrate abnormally high ratings of approachability for unfamiliar faces (Bellugi et al. 1999; Frigerio et al. 2006). Again paralleling auditory input, although face recognition abilities are preserved in WS, there is substantive evidence to suggest that the neurological route that delivers these abilities is different from the route in the TD brain (e.g. Deruelle et al. 1999; Grice et al. 2003). There are two broad ways that faces can be recognised: feature by feature (featureally) or in virtue of their overall pattern of features (configurally). TDs rely heavily on configural information to recognise faces. But individuals with WS are known to have a relative deficit in processing configural information generally (see Farran, Jarrold & Gathercole 2002) and their processing of visual information appears to rely heavily on featural analysis (Donnai & Karmiloff-Smith, 2000). This compensation strategy applies equally to face recognition: individuals with WS predominantly employ featural information to recognise faces (see Donnai & Karmiloff-Smith, 2000). Thus WS individuals have spared face recognition, but the cognitive-level skills used to support that ability are different to TD subjects.

How is this compensation achieved? The early brain is known to be extremely flexible. Thus it is plausible to suppose that the early brain compensates for a lack of neural resources responsible for configural processing by redeploying available resources to significant tasks. We have already seen that faces hold particular attraction for WS individuals. Thus we should expect available neural resources to be co-opted for face recognition. Thus neural plasticity supports cognitive reorganisation in early development. There is direct empirical evidence for face recognition being served by different neural pathways in individuals with WS. Mills et al. (2000) tested ERP functioning of WS subjects in tasks requiring the matching of upright face to inverted face. WS displayed different pattern of activation to TD subjects in the right hemisphere.
that is responsible for configural processing. Mills et al. (2000) suggest this is due to abnormal cerebral specialisation of spared cognitive functions in WS (also see Grice et al. 2001). Thus we have a case of spared abilities being served by an alternative neural route that supports different cognitive-level skills (featural as opposed to configural). The behavioural output is the same, but the cognitive level realisation and the neurological instantiation are substantially different. This makes face recognition an inappropriate domain to ground modular claims: the ‘data refutes the idea of an intact module and instead suggests that people WS may use a general object processes to recognize faces’ (Donnai & Karmiloff-Smith, 2000).

**Language**

Individuals with WS are often claimed to have preserved language abilities (e.g. Bellugi, Lai & Wang, 1997). Grammatical and syntactic skills (e.g. Pinker 1994; 1999), receptive vocabulary (e.g. Bellugi et al. 1990) and affective language (e.g. Reilly, Klima & Bellugi, 1990) have been cited as particular strengths. But recently the view that language is preserved has come under escalating assault. The most prominent attack is perhaps due to Stojanovik.

Stojanovik and colleagues have closely examined three areas of the language claimed to be preserved in the WS population: syntactic skills, social communicative abilities and vocabularies. They argue that it is misleading to describe WS skills across these areas as intact and, by extension, that the WS profile does not provide evidence of a modular basis for language (see, for example, Stojanovik, Perkins & Howard, 2001; Stojanovik, Perkins & Howard, 2004; Stojanovik, 2006; Stojanovik & van Ewijk, 2008). The most explicit argument for mental modularity from the basis of the WS cognitive profile is found in the area of grammatical and syntactic processing. The argument proceeds via a posited double dissociation with SLI. Individuals with SLI have specific problems with language comprehension and/or production, while all other cognitive areas appear to be preserved. According to Pinker (1994, 1999) this is in clear contrast to the WS population: ‘the genetic double dissociation is striking, suggesting that language is both a specialisation of the brain and that it depends on generative rules that are visible in the ability to compute regular forms. The genes of one group of children impair their grammar while sparing their intelligence; the genes of another group of children impair their intelligence while sparing their syntax’ (Pinker 1999, p. 262). Stojanovik and colleagues question whether this double dissociation actually holds. Most directly, Stojanovik et al. (2004) found no significant difference between the WS and SLI populations on standardised language measures. Furthermore, that the two populations often overlap in abilities and on some measures their SLI participants out performed their WS participants. This tells against the purported double dissociation and offers a direct challenge to those who argue for an independently language module on the basis of the contrast between the WS and SLI profiles. The challenge is further strengthened by later work on the social communicative language skills.

Individuals with WS have a number of documented social communicative problems. They tend to chatter incessantly, ask socially inappropriate questions and talk to themselves (Stojanovik, 2006) and are not sensitive to the needs of their conversational partner (Udwin & Yule, 1991). Stojanovik (2006, p. 166) reports they are actually ‘conversationally less able and less mature’ than both the TD and SLI populations. They produce significantly fewer of the continuations that demonstrate a willingness to maintain a conversation rather then merely respond in conversational turn. Their linguistic problems tend to be pragmatic (Stojanovik, 2006; Laws & Bishop, 2004). They provide insufficient information and fail to interpret either the literal or inferential meaning of interlocutors’ utterances more than the TD individuals (Stojanovik, 2006; pp. 166-167). Thus the linguistic behaviour of individuals with WS either tends to be parasitic on their partner or relies on their specialised knowledge in their domains of fascination.

In sum, despite earlier claims to the contrary, it appears language skills in WS are either significantly delayed or follow atypical developmental trajectories. The work of Stojanovik and colleagues suggests there is no evidence of a double dissociation between the WS and SLI. Rather the language trajectory in WS is atypical and the WS phenotype is not a profile of ‘clearly identifiable patterns of strengths and weaknesses’ (Stojanovik, 2006; p. 167-168). Further, interaction between language and other preserved domains is indicative of a highly interactive mind in which ‘language abilities cannot develop separately from other cognitive skills’ (Stojanovik, 2006; p. 168). I now move to consider social cognitive skills that show significant interaction with the face recognition and language domains.

**Social Cognition**

Individuals with WS are often described as having preserved social cognitive abilities. This is an extreme simplification. They are in fact inappropriately sociable, hyperactive and socially anxious, overly friendly and demonstrate abnormal social approach behaviour (see, for example, Gosch & Pankau, 1994; Bellugi et al. 1999; Frigerio et al. 2006). These behaviours appear to be the result of increased social drive. Individuals with WS have been characterised as having a strong compulsion towards social engagement (Frigerio et al. 2006). Importantly, this hyper-sociability is not restricted to familiar people. Individuals with WS show abnormally high ratings of approachability for unfamiliar faces (Bellugi et al. 1999; Frigerio et al. 2006) and are far more likely then non-WS individuals to approach strangers (Doyle et al. 2004). This is regardless of the perceived trustworthiness of faces (see Porter et al. 2007 for more details). That is, whether a face is rated as trustworthy or not, individuals with WS will rate the face as more approachable than non-WS individuals. Thus it is surely illegitimate to characterise the social skills of individuals with WS as preserved. Thus the WS profile
does not provide dissociative evidence for the modularity of social cognition. Moreover, social skills interact in complex ways with other cognitive domains. Such interaction tells against rigid modularity.

Recently, Riby, Doherty-Sneddon and Bruce (2008) have argued that face perception abilities may be especially significant to the social cognitive abilities displayed by WS individuals. A rather unusual dissociation effect for face recognition vividly demonstrates the complex interaction between these domains. TD subjects rely more on internal than external cues when recognising familiar faces. Thus masking internal cues (eyes, nose and mouth) disrupts recognition abilities for familiar faces more than masking external cues (hair, chin and ears) (see Young et al. 1985). But this is not the case for unfamiliar faces (see Bonner & Burton, 2004). Thus normal subjects show a familiarity effect: they demonstrate a relative advantage for internal cues for familiar, but not for unfamiliar faces. Individuals with WS do not show this familiarity effect. Moreover, WS individuals can more accurately match unfamiliar faces using internal cues than TDs (Riby et al. 2008). In fact, WS subjects match unfamiliar faces to an accuracy only ever reported in any population for familiar faces (Riby et al. 2008). Riby et al. (2008) suggest this unusual ability to process unfamiliar faces, results from the atypical social phenotype of WS: specifically, their hyper-sociability and drive to gaze at face regardless of familiarity.

TDs are only willing to attend to internal cues for familiar faces. That is, normal subjects will not attend to eyes, nose and mouth of people they do not know. Thus the superiority for internal cues for familiar faces is due to attentional bias. Individuals with WS, on the other hand, will attend to internal cues for unfamiliar faces. It is their increased social drive—hyper-sociability irrespective of familiarity and decreased levels of physiological arousal when gazing at unfamiliar faces—that “enables” this attention. Thus their reliance on external cues is decreased for all faces. Thus there is no superiority effect for internal cues for familiar faces, as internal cues are available for processing faces whether or not they are familiar. This is indicative of a complex interaction between face processing and social abilities. It is not that face processing provides the building blocks or part of the input for social cognition. We have rather a dynamic system in which social drives and face processing abilities show a complex two-way interaction. Such dynamic interaction is indicative of a holistic “problem solving” system rather than a rigidly specialised modular mind. Thus we have not merely a counter to the argument from WS to modularity as evidence by the impaired social abilities of individuals with WS, but also positive evidence that at least points towards a cognitive profile that is non-modular. Such complex interaction can be found in other areas of social reason. Consider, for example, an otherwise surprising result on drawing tasks.

Dykens, Rosner and Ly (2001) report no global-local dissociation on person drawing tasks: individuals with WS can draw both global and local features of human figures. This is in distinction to the drawing of geometrical shapes where there is a clear for bias to local features. Dykens et al. (2001) argue this may results from the social phenotype of WS. Thus due to their high sociability and interest in human faces, global features of humans can be more easily drawn than those of geometric figures. This interaction overcomes a deficit in the WS phenotype by driving attention to particular stimulus. In the above suggestion by Riby et al. (2008) a similar attentional drive explains the lack of familiarity effect for face recognition. In both cases, increased drive forces attention to a particular stimulus that in turn explains increased performance in the target domain. In the drawing task, the drive-to-attention corrects performance: increased attention leads to performance over the WS baseline such that the usual global processing deficit is overcome. In the lack of familiarity effect, increased attention actually places WS individuals at a relative advantage over TD individuals in the processing of unfamiliar faces. This appears to be unique, but the drive-to-attention correction of otherwise usual deficits appear to be widespread. Consider just two cases involving interaction between language and social cognition. First, the relatively preserved ability of adolescents to convey affect using both lexical expressions and vocal prosody appears to result from increased social drive (Reilly et al. 1990; Losh et al. 2000). Individuals with WS use affective modifiers more than TDs (Losh et al. 2000). Both Reilly et al. (1990) and Losh et al. (2000) suggest that WS individuals use these devices to keep listeners engaged in the conversation. They argue this represents a linguistic manifestation of the hyper-sociability. Second, consider Theory of Mind (ToM) reasoning. A number of studies suggest ToM abilities are spared (e.g. Karmiloff-Smith et al. 1995; Tager-Flusberg et al. 1998). But, more recently, Porter, Coltheart and Langdon (2008) have found ToM reasoning to be impaired. The difference between this and earlier studies was the use of a spatial (i.e. sequencing) false belief task. On these non-verbal false belief tasks the WS group performed significantly below TD controls. This suggests ToM skills have previously been overestimated because of a verbal biasing: that the apparent preservation of ToM abilities in previous studies is actually an artefact of the verbal nature of standard false belief and other ToM tests. That is, WS individuals are able to rely on their relatively spared language skills to pass ToM tasks even though their ToM abilities are impaired.

Before concluding it is worth making two more speculative points in support of a thoroughly anti-modular, dynamic account of WS. First, there is emerging evidence to challenge the syndrome-specific understanding of WS. Porter and Coltheart (2005) demonstrate that only some WS individuals display the typical cognitive peaks and valleys, while others actually display relative strengths in nonverbal skills and weakness in verbal abilities. Indeed, WS seems to be a particularly heterogeneous disorder (Porter & Coltheart, 2005; 2006). If this is the case, then the argument from particular spared and impaired abilities in the WS profile to modularity fails at the very outset. In fact,
rather than presenting a population that can form one side of a double dissociation, the most recent evidence is pointing to double dissociations within the WS population between two large sub-groups (Porter & Coltheart, 2005). A rigidly modular account of cognitive architecture would seem to lack the resources to explain such within population variation. Second, the present account has the resources to naturally explain the late onset of some “preserved” abilities in WS individuals. For example, consider the surprising results that younger children with WS perform lower than expected for their chorological age on vocabularies tasks (Thal, Bates & Bellugi, 1989; Volterra et al.2003) and WS individuals typically have delayed first words acquisition (Mervis et al. 2003) despite having essentially preserved vocabularies in later life. The drive-to-attention correction has a straightforward explanation of such delayed onset. Extra time is required for an atypical route to develop due to the drive-to-attention correction forcing the co- opting of alternative neural resources. Hence WS individuals should show slow starts followed by increased performance leading to “preserved” abilities.

Conclusion
The WS brain is developmentally atypical. As such, it is wrong to characterise the WS brain as a TD brain with a pattern of specific preservation and deficits. Thus arguments from WS to specific modules fail. Rather than speaking to a system of functionally independent modules, the WS literature suggests a highly dynamic cognitive architecture. The story is one of complex interaction over the developmental trajectory of the WS cognitive profile, a story far too nuanced and intricate to be accounted for by positing functionally independent modules.

Acknowledgments
Thanks to Jay Garfield, Cynthia Townley, Kim Sterelny and members of the audience at ACS09, especially Phil Gerrans.

References


**Citation details for this article:**
