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Language in genetic syndromes and cognitive modularity

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Chapter 29: Language in Genetic Syndromes and Cognitive Modularity

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29.1 Introduction

In recent years, research into the impact of genetic abnormalities on cognitive development, including language, has become recognised for its potential to make valuable contributions to our understanding of the brain-behaviour relationships underlying language acquisition as well as understanding the cognitive architecture of the human mind. The publication of Fodor's (1983) book *The Modularity of Mind* has had a profound impact on the study of language and the cognitive architecture of the human mind. Its central claim is that many of the processes involved in comprehension are undertaken by special brain systems termed 'modules'. This domain specificity of language or *modularity* has become a fundamental feature that differentiates competing theories and accounts of language acquisition (Fodor 1983, 1985; Levy 1994; Karmiloff-Smith 1998).

However, although the fact that the adult brain is modularised is hardly disputed, there are different views of how brain regions become specialised for specific functions. A question of some interest to theorists is whether the human brain is modularised from the outset (nativist view) or whether these distinct brain regions develop as a result of biological maturation and environmental input (neuroconstructivist view). One source of insight into these issues has been the study of developmental disorders, and in particular genetic syndromes, such as Williams syndrome (WS) and Down's syndrome (DS). Because of their uneven profiles characterised by dissociations of different cognitive skills, these syndromes can help us address theoretically significant questions. Investigations into the linguistic and cognitive

profiles of individuals with these genetic abnormalities have been used as evidence to advance theoretical views about innate modularity and the cognitive architecture of the human mind.

The present chapter will be organised as follows. To begin, two different theoretical proposals in the modularity debate will be presented. Then studies of linguistic abilities in WS and in DS will be reviewed. Here, the emphasis will be mainly on WS due to the fact that theoretical debates have focused primarily on WS, there is a larger body of literature on WS, and DS subjects have typically been used for the purposes of comparison. Finally, the modularity debate will be revisited in light of the literature review of both WS and DS. Conclusions will be drawn regarding the contribution of these two genetic syndromes to the issue of cognitive modularity, and in particular innate modularity.

29.2 Nativist approach to language acquisition and cognitive architecture

The first half of the 20th century was dominated by a behaviourist view of language acquisition. According to this view, linguistic abilities are acquired in childhood as a result of training provided by the members of the child's immediate linguistic environment. Skinner (1957), the main proponent of behaviourism, proposed that language is just another behaviour which can be acquired through explicit teaching and reinforcement.

In an attempt to reject behaviourist explanations of language acquisition, Noam Chomsky launched in the late 1950s what has become known as a 'nativist' revolution. The adherents of the nativist tradition oppose the behaviourist's simplistic and rather mechanical way of explaining language acquisition. They propose instead that a generative grammar is to be taken as a starting point for explaining language acquisition. A generative grammar is a set of

rules or procedures which allow one to generate all and only the grammatical sentences in a language, characterise all the sentences which already exist in the language corpus and predict the existence and properties of new sentences (Chomsky 1965).

Another crucial aspect of nativist theories with regard to the organisation of human cognitive architecture is the assumption that the human mind is modular and that language is a separate, independent module. This idea has been clearly articulated by Jerry Fodor. Fodor (1983) argued that many of the processes involved in the comprehension of language are undertaken by specialized brain systems termed 'modules'. This domain specificity of language or modularity has become a fundamental feature that differentiates competing linguistic theories and accounts of language acquisition (Fodor 1983, 1985; Bates *et al.* 1988, 1995). Fodor argues for a distinction between a central system responsible for rational thought and the fixation of belief, and a number of modular input systems which deliver input into the central system. The theory posits that while cognitive processes, such as long-term memory and problem-solving are carried out by non-modular, relatively slow, central processes, input systems such as those involved in perception are modular in nature. The language faculty is viewed as an input system on a par with the senses, such as vision.

Fodor's main examples of modules are language comprehension and visual perception. Fodor (1983: 98) points out that 'the key to modularity is informational encapsulation'. Informational encapsulation describes the feature of modules whereby they only have access to information of a certain type. A visual perception module, for example, only has access to visual perceptual data. Other characteristics of modules, such as domain specificity, shallow output, neural localisation, etc. may be present, but they are not crucial. Given that modules are informationally encapsulated, one would expect there to be minimal interference in the

event of breakdown. So, for example, difficulties in the visuo-spatial domain should not affect comprehension of syntactic structures. In order to test this hypothesis, one inevitably needs to refer to atypical populations which present with breakdowns of different cognitive functions resulting in cognitive dissociations.

As mentioned above, a commonly assumed approach to language acquisition within the nativist view is represented by generative grammar. According to proponents of generative grammar, children's knowledge of morphosyntax and possibly phonology consists of knowledge of formal rules or operations (Ambridge and Lieven 2011). A large body of literature on language acquisition has emerged within the nativist and generativist frameworks (for a review of studies, see Ambridge and Lieven 2011). Discussion of this literature on typical language acquisition is beyond the scope of this chapter. However, it is worth pointing out that dissociations between language and other cognitive skills, such as is often found in atypical populations, have been used by nativist theorists as evidence in support of an innate language (or more often, 'syntax') module. Evidence of this type will be presented in a discussion of Williams syndrome in section 29.5 below.

In addition, there have been some theoretical developments within the nativist view since the publication of Fodor's landmark book in 1983. Evolutionary psychologists have proposed what is known as the massive modularity hypothesis (Machery 2011). According to this hypothesis, human cognitive architecture is built from a number of different systems and most of these systems are believed to be adaptations selected for specific purposes (Machery 2011). Unlike Fodorian modularity, this hypothesis does not propose informational encapsulation of the different systems. Importantly, it does not say anything about whether

the different systems are specified from birth and it proposes that development involves an interaction between the genome and the environment.

Evidence in support of the massive modularity hypothesis should come from developmental disorders. This is because developmental disorders often show dissociations between different cognitive skills, dissociations which are assumed to provide support for distinct components in human cognitive architecture (Machery 2011). The massive modularity hypothesis also proposes that the component parts of the human cognitive system are adaptations. The question of how the massive modularity hypothesis can provide evidence that these components are adaptations is still to be answered. However, the answer is to be sought in developmental disorders. This is because dissociations found in developmental disorders will need to show how specific systems fulfil specific functions and *only these functions*. For the time being, the evidence is not very strong (see discussion of Clahsen and Almazan's (1998) work on WS in section 29.5).

29.3 Neuroconstructivist approach

In the 1970s, a second stream of thought regarding language acquisition was derived largely from the work of Jean Piaget. Even though it seemed that this stream would be some kind of continuation of the empiricist thinking of the 17th and 18th centuries, this was not quite the case, as Piaget (1980: 23-4) explicitly acknowledged:

> 'The critique of empiricism is not tantamount to negating the role of experimentation, but the "empirical" study of the genesis of knowledge shows from the onset the insufficiency of an "empiricist" interpretation of experience. In fact, no knowledge is based on perceptions alone, for these are always directed,

> and accompanied by schemes of action. Knowledge, therefore, proceeds from action, and all action that is repeated or generalised through application to new objects engenders by this very fact a "scheme", that is, a kind of practical concept'.

This quotation from the Piaget-Chomsky debate suggests that although Piaget does not fully discard the role of experience in knowledge acquisition, he emphasises the necessity of the human subject having an important role in structuring activity. Thus, knowledge is supposed to proceed from action, or what Piaget terms 'assimilation' of objects to the schemes of the subject. What is the relevance of such a proposal to language acquisition?

According to Piaget, language is facilitated by the development of sensory-motor schemas that represent the joint outcomes of perception and action. Sensory-motor schemas undergo orderly changes which are nourished, but not shaped by, continuing experience in acting on the world. This means that the child will be able to separate thought from action in her schemas in due course, and her concepts of objects and events in the world will become independent of the actions to be performed on them. Thus, in order for the infant who is acquiring language to be able to make linguistic distinctions such as Object and Action (i.e. NP and VP), she needs to acquire the concept that Object is independent of Action.

For Piaget, language is a manifestation of intelligence and is, hence, not dissociable from it. It is also argued that certain cognitive prerequisites are necessary for the acquisition of language. The idea is that normal development of language is parasitic on the prior mental development of such abilities as 'seriation' and 'conservation'. In order to acquire linguistic structures such as correct word order or the passive, children are supposed to have reached a

level of cognitive development at which they could carry out tasks such as putting items in ascending order of size (seriation), or judging that the amount of liquid poured from a tall thin glass into a short one remained the same (conservation).

Thinking has moved on from the time when Piaget first proposed his view of language acquisition. Although some of Piaget's fundamental principles are still very much present, there have been a number of theoretical developments. For example, there is a large body of literature which addresses the issue of cognitive precursors to language acquisition. The very concept of cognitive precursors or prerequisites suggests that it is taken for granted that language acquisition is dependent upon the development of other cognitive skills. These skills include speech segmentation (Newman *et al.* 2006) and joint attention (Morales *et al.* 1998, 2000; Markus *et al.* 2000), amongst others.

Pertinent to the current chapter are challenges to the innateness of modularity, and the proposal that modularity is a product of development (Karmiloff-Smith 1994, 1998). This viewpoint recognises the existence of innate biological constraints on language acquisition but it considers these constraints to be not so detailed and less domain-specific as far as higher-level cognitive functions such as language are concerned. Development is seen as playing a crucial role in shaping phenotypical outcomes and the protracted period of post-natal growth is seen as essential in influencing the resulting domain specificity of the developing neocortex (Karmiloff-Smith 1998). As with the nativist view, it is difficult to test the idea that language acquisition is an inseparable part of general cognitive development, because in typically developing children different cognitive abilities develop in synchrony with each other. Assuming this theoretical approach, a child with developmental disorders would be expected to show language acquisition in line with their general cognitive abilities.

29.4 Genetic syndromes

Over the past four decades, language breakdown and atypical linguistic and cognitive development have played a very important role in the ongoing debate on modularity. In particular, genetic syndromes have played a major role in this debate. A 'syndrome' is defined as the presence of multiple anomalies in the same individual with all of those anomalies having a single cause. So far, over 300 different genetic syndromes have been identified (Shprintzen 1997). Populations affected by genetic syndromes have been studied for different reasons. One strong motivation is to gain knowledge about the behavioural manifestations of a specific genetic abnormality, with a view to having a better understanding of the condition and informing diagnosis and remediation. Another reason for studying genetic syndromes is the potential contribution that they can make to theoretical debates on the role of general cognitive mechanisms for language acquisition. Developmental disorders provide a naturalistic way of testing the relation between the biological (and psychological) basis of language and the biological (and psychological) basis of other cognitive or neural systems (Marcus and Rabagliati 2006).

Two syndromes which have attracted more interest than others are Williams syndrome (WS) and Down's syndrome (DS). There are a number of reasons for this. The foremost reason is the fact that affected individuals present with uneven, and qualitatively different, profiles of cognitive abilities within the context of similar IQ levels (~40 to 60; Udwin and Yule 1991; Roizen 2002). In DS, relatively good visuo-spatial abilities contrast with poor expressive language skills (Fowler *et al.* 1994; Rondal and Comblain 1996; Chapman *et al.* 1998; Klein and Mervis 1999; Jarrold *et al.* 2002; Laws 2002). Individuals with WS display relatively good expressive language abilities and poor visuo-spatial abilities (Bellugi *et al.* 1994; Grant

et al. 1997; Howlin *et al.* 1998). Within the context of language abilities, the two populations also show uneven profiles. Syntactic abilities are a relative weakness in DS but a relative strength in WS (Bellugi *et al.* 2000). Expressive prosodic skills, which are a relative weakness in DS (Pettinato and Verhoeven 2009; Stojanovik 2011), are relatively unimpaired in WS as they are in line with level of language comprehension (Stojanovik *et al.* 2007). Finally, pragmatic aspects of language are impaired in the context of WS (Laws and Bishop 2004; Stojanovik 2006), but are a relative strength for those with DS (Laws and Bishop 2004). Each of these syndromes will be reviewed separately below.

29.4.1 Williams syndrome

Williams syndrome is a relatively rare genetic condition which was first identified in 1961 by Williams and his colleagues in New Zealand (Williams *et al.* 1961). They labelled the syndrome following a clinical study of four patients with mental retardation and a peculiar facial appearance. WS occurs in one of 15,000-20,000 live births, although a study in Norway suggested an incidence rate of 1 in 7,500 (Strømme *et al.* 2002). WS results from a micro-deletion of one copy of about 20 contiguous genes in chromosome 7, affecting one of the alleles of the elastin gene (Frangistakis *et al.* 1996; Korenberg *et al.* 2000). Molecular genetic testing by means of fluorescence *in situ* hybridisation (or FISH) can now be used to confirm the deletion of one copy of the elastin gene and other surrounding genes in a small region of chromosome 7 at 7q11.23. WS is characterised by a range of moderate to severe physical abnormalities, including elevated blood calcium levels, high blood pressure, failure to thrive in infancy and abnormal sensitivity to certain classes of sounds (hyperacusis).

On the level of brain organisation, WS typically presents with no evidence of focal lesions. Given that approximately 22 out of the 28 genes within the WS critical region are thought to

be expressed in the brain, it is very likely that brain development in WS differs from typical development (Karmiloff-Smith 2011). There have been few studies of the WS brain during development (i.e. childhood) and most of what we know about brain structure in WS comes from adult studies. Brain volume in WS reaches about 80% of normal brain volume (Chiang *et al.* 2007) and there is increased cortical thickness (Thompson *et al.* 2005). The frontal lobes, superior temporal gyrus, amygdala, fusiform gyrus and cerebellum in WS adult brains are relatively preserved whereas the parietal and occipital lobes, the thalamus, the basal ganglia and the midbrain are smaller in volume (Karmiloff-Smith 2011). An interesting finding is that the amygdala and the cerebellum are larger than the rest of the WS brain and a large cerebellum has been shown to be present in early childhood (Jones *et al.* 2002).

Although WS was first identified in the 1960s, it was not until about two decades later that it started attracting increasing interest from researchers. The most striking phenomenon linked to the syndrome was the reported uneven cognitive-linguistic profile. This profile includes moderate to severe learning difficulties, impairments in planning, problem solving and spatial cognition alongside relative strengths in social cognition, linguistic abilities, face processing and auditory rote memory (Mervis and Klein-Tasman 2000). Despite the fact that performance IQ is around 50 on average for people with WS, the general view is that linguistic abilities are a relative strength. However, this view has been seriously challenged in recent years.

Early studies on Williams syndrome (1970s and 1980s):

Early studies of WS were typically multi-dimensional investigations which incorporated behavioural, medical, physiological, cognitive and linguistic aspects of the WS profile. These studies provided a rather broad picture of the WS phenotype. However, they suggested the

existence of possible dissociations in the WS profile. For example, Von Arnim and Engel (1964) described the profiles of four individuals with WS who were aged 5 to 15 years. The IQs of these children ranged from 43 to 56. They had physical growth problems, poor motor coordination, outgoing personalities, recurrent signs of unreasonable anxiety, and an unusual command of language. Von Arnim and Engel (1964: 375) stated that '[t]heir loquacity combined with friendliness and a great ability to make interpersonal contacts makes them appear brighter and more intelligent than in fact they are'. A decade later, Jones and Smith (1975) presented evaluation data on 14 children and adults with WS between the ages of 3 months to 23 years. The children were reported to have full IQ scores which ranged between 41 to 80 and a mean IQ of 56. This study also makes special mention of linguistic abilities in WS, but with not much elaboration. The personality of individuals with WS was described as 'friendly, loquacious, and cocktail party manner' (Jones and Smith 1975: 719).

It was not until 1978 that the first attempt was made to systematically quantify data on individuals with WS. Bennett *et al.* (1978) studied seven children with WS who were 4:06-8:05 years of age. The children had mild to moderate learning difficulties as revealed by scores (range 30-81, mean IQ 53.9) on the McCarthy Scales of Children's Abilities (McCarthy 1972). All seven children performed better on measures of verbal ability than on fine motor and gross motor measures. Bennett *et al.* concluded that verbal abilities in WS were superior in the face of impaired motor skills and cognitive deficits. Thus, a debate was begun which remains unresolved as yet.

A number of studies investigating linguistic abilities in WS followed throughout the 1980s. As the research base started to grow so too did the controversy regarding 'superior' language skills in WS. Kataria *et al.* (1984) did not find evidence of superior verbal skills over motor

abilities. Nor did they find evidence of an 'unusual command of language' (previously considered to be a marker of the syndrome). Arnold *et al.* (1985) found no difference in the performance of children with WS on the verbal and non-verbal subtests of the Wechsler's Intelligence Scale for Children (WISC; Wechsler 1976). Furthermore, on the Reynell Developmental Language Scale (Reynell 1977), the language skills of only three children exceeded the seven-year ceiling of the test, while those of the remainder ranged from three to seven years. Most children, though, were able to produce and respond to simple sentences.

Pagon *et al.* (1987) also administered the WISC to a group of individuals with WS. Seven of the group scored above the floor of the verbal scale, and had verbal IQs of between 47 and 85. Five of these individuals also scored above the floor on the performance scale and their performance IQs were between 45 and 69. These differences were non-significant. Only one individual with a high verbal IQ score showed a verbal advantage. Similarly, Crisco *et al.* (1988) found no significant verbal advantage on the Illinois Test of Psycholinguistic Abilities (Kirk *et al.* 1968) for a group of children with WS when matched on mental age with another group of children with non-specific developmental disabilities. However, they did observe significantly poorer performance for the WS group in comparison to the control group on visual reception, visual closure and visual memory. No verbal advantage on the WISC was reported by Dall'Oglio and Milani (1995) and Greer *et al.* (1997) on the Stanford-Binet Intelligence Scale.

Unlike the studies mentioned above, significant differences between verbal and performance IQ were reported in a series of papers by Udwin and colleagues (Udwin *et al.* 1986, 1987; Udwin and Yule 1990, 1991). These four papers give details from a single, large group of 44 individuals with infantile hypercalcaemia (former label for WS) aged between 6;0 and 15;09

years who were administered the WISC. A number of participants scored below floor on the verbal and performance scales. But verbal IQs for the remaining participants ranged between 45 and 109 (mean = 62.4), while performance IQs ranged between 45 and 73 (mean = 55.9). This suggests a verbal advantage for the majority of the participants albeit a fairly marginal one.

In summary, the majority of studies which have used IQ measures with the WS population have found no significant differences between their verbal and non-verbal profiles. The exceptions are Udwin and Yule (1990, 1991) and Udwin *et al.* (1986, 1987) who reported a marginal verbal advantage. The problem with using IQ measures for the purposes of investigating verbal and non-verbal skills is that the verbal part of both the WISC and the Stanford-Binet scales requires the use of metalinguistic skills and knowledge of social situations. Both scales examine knowledge of vocabulary by asking the participant to provide a definition (a metalinguistic skill) as opposed to, for example, only asking the participant to name a picture. Also, both scales have a comprehension sub-part which requires practical problem solving and social information. It has been widely documented across studies that individuals with WS have difficulties with problem solving tasks (Bellugi *et al.* 1988, 1989, 1994).

More recent studies on Williams syndrome (from 1990s to present day):

From the 1990s onwards, research into WS became more systematic. Researchers started investigating various levels of language including phonology, morphosyntax, lexical semantics and pragmatics. Each of these levels will be reviewed in this section. Some studies argued that each of these areas of linguistic functioning is somewhat 'spared' or is a relative strength in WS. A series of studies from the Bellugi group mainly involving older children

and young adults with WS suggested that individuals with WS have 'spared' or 'preserved' syntactic abilities (Bellugi *et al.* 1988, 1994, 1999). The evidence came from individuals with WS whose comprehension of complex syntactic structures such as reversible passives, negative clauses and conditionals was better than that of age- and IQ-matched individuals with DS. Also, individuals with WS were reported to be better than age- and IQ-matched individuals with DS at detecting syntactic anomalies and correcting ungrammatical sentences.

A study by Clahsen and Almazan (1998) also reported that individuals with WS are able to comprehend reversible passives, regular past-tense morphology and reflexive anaphors. A recent study by Musolino *et al.* (2010) argued that individuals with WS are able to understand core syntactic and semantic relations. These authors claimed that 'knowledge of core, abstract principles of grammar is present and engaged in WS' (Musolino *et al.* 2010: 53). Performance on regular as opposed to irregular morphology has been shown to be a relative strength for individuals with WS in different languages (Pleh *et al.* 2003; Clahsen *et al.* 2004). However, individuals with WS have never been shown to outperform mental age controls.

The study by Clahsen and Almazan (1998) has been referred to as an example of how a genetic disorder such as WS can support the massive modularity hypothesis. This is because the study revealed a dissociation between regular and irregular morphology, suggesting the existence of two systems: a computational system which involves the application of syntactic rules (such as 'add -ed to regular verbs'), and a lexical system responsible for lexical knowledge. Importantly, however, when Thomas *et al.* (2001) employed a developmental trajectory approach and controlled for mental age when analysing data from the performance on regular versus irregular morphology, they did not find an effect of regularity on

performance in the WS group. This throws into question the original claim that children with WS are better at regular than irregular inflectional morphology. This, in turn, raises doubts about the existence of two separate computational and lexical systems.

There is no evidence in the research corpus so far to show that individuals with WS perform better than expected for their level of non-verbal ability on morphosyntactic tasks. In fact, some studies have even shown that children with WS perform below levels expected for their non-verbal mental age. For example, Joffe and Varlocosta (2007) reported that participants with WS performed significantly worse than mental age matched controls on a task requiring them to repeat wh-questions and also on a standardised test of grammar, the Test of the Reception of Grammar (Bishop 2003).

With regard to lexical abilities in WS, one striking finding that has emerged from the existent literature is the relative strength in receptive vocabulary. Receptive vocabulary is one language domain in which individuals with WS tend to score better than might be predicted by their mental age (Bellugi *et al.* 1988; Rossen *et al.* 1996; Clahsen *et al.* 2004). This relative strength seems to hold for concrete rather than conceptual/relational vocabulary (Mervis and John 2008). There has been debate as to whether individuals with WS have atypical semantic organisation. Some studies have shown that naming, when accurate, is faster in individuals with WS than mental age controls (Temple *et al.* 2002). Ypsilanti *et al.* (2005) reported atypical responses by individuals with WS on a word definition task. However, Tyler *et al.* (1997) did not find any different priming effects of category structure and functional relations in an online task in individuals with WS compared to controls, suggesting that semantic organisation was not atypical.

Studies have also shown that individuals with WS may show delayed semantic development. For example, individuals with WS showed frequency and semantic category effects in a speeded picture-naming task which were in line with their receptive language skills (Thomas *et al.* 2006). Although some early research pointed out that individuals with WS may sometimes use rare and low-frequency vocabulary (Bellugi *et al.* 1992), which was taken as evidence that lexical semantics may be independent of general cognitive abilities (Bellugi *et al.* 2000), such findings have not been replicated. For example, Jarrold *et al.* (2000) reported that individuals with WS did not produce more novel items in a semantic fluency task than a control group of individuals with learning difficulties matched on receptive vocabulary. More recently, Stojanovik and van Ewijk (2008) showed that, when the conversational context was controlled for, children with WS did not produce more low-frequency words than controls matched for language age, non-verbal mental age and chronological age.

Phonological abilities in WS have perhaps attracted least attention. This is possibly due to the fact that individuals with WS, unlike those with DS, have intelligible speech by and large. Suprasegmental features have attracted more interest. A seminal study by Reilly *et al.* (1990), which included only four participants with WS, reported that individuals with WS have expressive prosody which is over-rich in affect intonation. Abnormally high pitch range, which results in individuals with WS being perceived as twice as emotionally involved as children of a similar language and chronological age, was confirmed in a larger study by Setter *et al.* (2007).

Research involving a developmental trajectory approach to data analysis showed that children and teenagers with Williams syndrome show a delayed onset (relative to chronological age) in the development of some prosodic skills, such as the ability to use prosody to signal which

is the most important word in a phrase (Stojanovik 2010). This same study demonstrated a delayed rate of development in the ability to use prosody to achieve the disambiguation of potentially ambiguous phrases and to produce questioning versus declarative intonation (Stojanovik 2010). In addition, children with WS have different prosodic profiles from children with DS despite having similar levels of receptive language and non-verbal abilities. In particular, children with WS were significantly better than children with DS at perceiving and producing affect and questioning versus declarative intonation (Stojanovik 2010). This cross-syndrome difference is interesting as it suggests some syndrome-specific characteristic and has implications for how we evaluate the evidence that genetic syndromes provide for innate modularity. This issue will be discussed in more detail in the conclusions of the chapter.

Last but not least, we need to mention pragmatic abilities in WS. Early studies of WS reported relative strengths in the domain of communication skills in WS. For example, Reilly *et al.* (1990) characterised individuals with WS as being 'highly social' and as 'having remarkable social understanding'. A series of studies by Jones *et al.* (2000) reported that individuals with WS used a greater number of descriptions of affective states and evaluative comments during an interview task. They also included more inferences about the affective state and motivation of story characters than children with DS or typically developing children.

However, other studies have shown that individuals with WS use more stereotyped conversation than children with DS or specific language impairment (Laws and Bishop 2004) and have difficulties with establishing social relationships and making friends (Davies *et al.* 1998). Social interaction deficits, and specifically difficulties with exchange structure,

responding appropriately to interlocutor's requests for information and clarification, interpreting meaning and providing enough information for the conversational partner were reported by Stojanovik (2006). Individuals with WS also have difficulties with the understanding of idiomatic expressions (Mervis *et al.* 2003) and figurative language, in particular lexicalised metaphor comprehension, which was reported to be lower than expected for receptive language abilities (Annaz *et al.* 2009).

29.4.2 Down's syndrome

DS is the most common genetic cause of developmental delay (Martin *et al.* 2009). It results from extra genetic material on chromosome 21 (Rondal and Edwards 1997). The majority of cases (about 95%) are caused by complete duplication of chromosome 21. The remainder of cases are caused either by translocation of material from chromosome 21 to another chromosome, typically 13 or 18 (4%), or mosaicism (i.e. a mixture of trisomy and unaffected cells) (1%) (Baum *et al.* 2008). A person of any race, socioeconomic status, or geographic location can have a child with DS, and the only aetiological factor definitely linked to DS is increasing maternal age (Hassold and Sherman 2002). DS has an incidence of 1 in 1,000 (Down's Syndrome Association 2012). The syndrome is characterised by a range of physiological and anatomical anomalies. Individuals with DS often have characteristic facial features due to midfacial hypoplasia. These include a flat broad face, flat nasal bridge, and a flat facial profile (Baum *et al.* 2008). Physical features such as short stature, hypotonia and hyperflexibility of the joints are also present (Baum *et al.* 2008).

At the level of brain organisation, some anomalies have been reported. For example, individuals with DS seem to present a right cerebral hemisphere lateralisation for receptive language and a left cerebral hemisphere lateralisation for production of simple and complex

movements (Heath *et al.* 2007). A study by Shoji *et al.* (2009) has also reported atypical linguistic lateralisation in a dichotic listening task in which individuals with DS showed a left ear advantage when presented with words with two consonant-vowel syllables. This is a different pattern to that seen in the neuro-typical brain, where language processing typically occurs in the left hemisphere. This has been supported by hand-preference studies (which are believed to be a marker of functional hemispheric specialisation). For instance, children with DS were found to show weaker hand preference than typically developing children (Groen *et al.* 2008).

The linguistic and cognitive profile of individuals with DS is often characterised as 'uneven'. There are weaknesses in auditory short-term memory relative to visual short-term memory and other aspects of cognition (Chapman 2003), and strengths in social functioning abilities (Kasari and Bauminger 1998). Children with DS are slow to acquire language. General language performance in individuals with DS tends to be lower than expected from their level of cognitive development (Fowler *et al.* 1994; Chapman and Hesketh 2000; Vicari *et al.* 2000; Perovic 2001, 2002). Exceptionally, language abilities may in some cases be higher than other cognitive abilities (Rondal 1994), but such cases are rare.

Vocabulary knowledge seems to be stronger than grammatical abilities (Chapman *et al.* 1991; Miller 1996). Many children with DS do not acquire their first words before the age of 2 years (Rondal 2001). However, early lexical development generally shows a positive linear relationship with mental age (Rondal and Edwards 1997). Receptive vocabulary has also been reported to be in line with mental age (Laws and Bishop 2003). Although vocabulary is a relative strength in the language profile of the DS population, there is individual variation. In a study of 43 children with DS, Miller (1999) reported that 65% scored below their mental

age on vocabulary and 35% had vocabulary scores consistent with their mental age on the MacArthur Communicative Development Inventories (Fenson *et al.* 1993). Jarrold *et al.* (2002) also reported that children and adults with DS often have receptive vocabulary deficits compared to typically developing individuals matched for mental age.

Morphosyntactic abilities in children with DS are a relative weakness. Eadie *et al.* (2002) compared the accuracy of marking finiteness in spontaneous speech in children with DS to children with specific language impairment. The study found that the profiles of the groups of children were similar, suggesting that grammatical abilities in children with DS are comparable to those of children with known language impairments. A study by Ring and Clahsen (2005) reported that adolescents with DS have marked difficulties with tense marking, plural marking and the marking of comparative and superlative on adjectives. Interestingly, the participants with DS had equal difficulties with tense and non-tense related morphemes, suggesting that the grammatical morpheme difficulties in DS extend beyond the finiteness cluster of morphemes.

Phonological abilities in DS have been investigated more frequently than those of individuals with WS, due to the fact that individuals with DS have difficulties with producing intelligible speech. A study by Cleland *et al.* (2010) reported that two thirds of a cohort of 15 individuals with DS (mean age 14;3 years) had severe speech disorders to the extent that they failed to meet the basal age-equivalent of 3;0 years in the Diagnostic Evaluation of Articulation and Phonology (Dodd *et al.* 2002). The most common phonological process evidenced was consonant cluster reduction followed by final consonant deletion and initial consonant deletion. Prosodic abilities are mainly in line with non-verbal mental age apart from production of affective intonation and pre-final narrow focus which seem lower than

expected for non-verbal mental age (Stojanovik 2011). In addition, and similar to morphosyntactic and lexical skills, prosodic abilities seem better with regard to comprehension as opposed to production.

With regard to pragmatic and social communication skills, it has been shown that people with DS are very keen to engage in conversation and to keep the conversation going but often lack the appropriate language skills to do so (Rondal 2001). Two studies described in Abbeduto and Murphy (2004) report relative strengths and weaknesses in the DS communication profile. For example, in a barrier task, individuals with DS were less likely than typically developing individuals matched for mental age and individuals with fragile X syndrome to provide listeners with referential frames which help the listener's comprehension. Also, individuals with DS were less likely than mental age matched controls to signal non-comprehension. This suggests that they may be unable to monitor their own comprehension which can seriously disrupt the flow of conversations. However, individuals with DS were found to appreciate shared knowledge in conversational exchanges. They were also able to make appropriate shifts from indefinite descriptions such as 'a house' to definite descriptions such as 'the house'.

29.5 Critical evaluation of the modularity debate

As already mentioned in the introduction, comparing linguistic abilities in children with WS and those with DS is relevant for addressing theoretical questions about the architecture of human cognition. This is because both genetic disorders present with similar levels of non-verbal IQ, but different linguistic profiles. The literature on linguistic abilities in WS is richer than the literature on DS and aspects of linguistic abilities in WS, such as morphosyntactic, lexical and pragmatic abilities, have been studied more systematically and in greater detail in

WS than in DS. Although it is obvious that there are areas of strengths and weaknesses in both genetic disorders, what is also striking is the fact that linguistic abilities in WS rarely, if ever, exceed non-verbal abilities. The literature review on WS in section 29.4.1 suggests that individuals with WS present with language deficits relative to chronological age, but in line with non-verbal mental age (e.g. Thomas *et al.* 2001; Stojanovik *et al.* 2004, 2007). Interestingly, language abilities in some studies have been shown to be even lower than non-verbal mental age (Joffe and Varlocosta 2007). This suggests that it is very unlikely that linguistic and non-verbal abilities are dissociable. It seems that linguistic abilities (with the exception of, perhaps, receptive vocabulary) develop in synchrony with non-verbal skills.

The innate modularity view and, in particular, the massive modularity hypothesis are based on the premise that typical cognitive architecture consists of a number of different systems. If this view is correct, it is expected that these systems will dissociate in developmental disorders. The lack of dissociations found in WS means that one cannot accept the nativist view and must accept the alternative neuroconstructivist view. This latter view posits that it is impossible for language abilities to exceed an individual's level of cognitive functioning as these abilities are not dissociable from general cognition. Studies which argue that language abilities in WS are superior to non-verbal abilities – a claim which supports the nativist view – do not have convincing evidence that this is really the case. Given that most individuals with WS reach a non-verbal mental age of between 5 and 7 years, it is not surprising that they are able to comprehend core syntactic and semantic relations (Musolino *et al.* 2010). These relations are expected to have been acquired by typically developing children by 5 years of age. Hence, there is no compelling evidence for clear dissociations between general cognitive abilities and language.

Although not as rich, evidence from DS is still very informative. This evidence leads to a different conclusion in the theoretical debate between innate modularity and neuroconstructivism. So far, research has shown that linguistic abilities in DS often lag behind non-verbal abilities. In particular, morphosyntactic skills and, in some individuals, vocabulary knowledge seem to be poorer than expected given non-verbal mental age. Such findings suggest that linguistic abilities may not be fully dependent upon non-verbal functioning, otherwise one would not expect to find language skills which are below a person's general level of cognitive functioning. It seems that in individuals with DS (although there are a few exceptions), non-verbal cognitive ability does not necessarily interact with language in the same way as it appears to in individuals with WS. This leads to the conclusion that perhaps language skills (or at least some language skills) do develop independently of other cognitive abilities, as the innate modularity hypothesis would suggest.

Interestingly, it has been research into WS rather than DS which has been referred to as the prototypical example of a dissociation between linguistic and non-verbal abilities in theoretical debates on innate modularity. However, the literature review above suggests that there may also be a dissociation between linguistic and non-verbal skills in DS. Unfortunately, linguistic abilities in DS have not received the detailed investigation, scrutiny and analyses needed in order to assess the magnitude of possible associations or dissociations between linguistic and non-verbal functioning. In addition, there have not been many studies which have adopted a developmental trajectory approach in order to explain the delay in language and other cognitive abilities often reported in individuals with DS. If it turns out that the developmental trajectory of language and other cognitive abilities is atypical in DS and that there are no clear dissociations between language and non-verbal cognitive abilities, one could argue that DS provides evidence for the neuroconstructivist view.

Despite the fact that research into developmental disorders over the past few decades has not resolved the theoretical debate between nativism and constructivism, the fields of language acquisition, developmental psychology and cognitive neuroscience have gained much from this research. For example, we now have a much more comprehensive understanding of the complexities of developmental disorders, in particular Williams syndrome and, to some extent, Down's syndrome. The evidence from these two disorders (at least what we have so far) does not exclusively support one or the other theoretical viewpoint. It would be fair to say that research into WS and DS has been driving the development of new thinking about how the theoretical frameworks we are currently working with may need to be modified. For example, based on a detailed overview of the language acquisition literature on inflectional morphology and with reference to the two competing theories (nativism and constructivism), Ambridge and Lieven (2011: 190) conclude that if nativist-generativist accounts of language acquisition could explain early errors reported in child language, and if constructivist accounts could explain how the adult state of language knowledge is reached, 'the field would be able to move closer to a complete understanding of the domain of inflectional morphology'.

In a similar manner, given that developmental disorders such as WS and DS provide evidence for both theoretical viewpoints, it seems that a more unified account is needed. This would bring the two views together so that instead of opposing each other, they would complement each other. For example, it is true that a number of cognitive skills in WS develop atypically and that there are no strong language and non-verbal dissociations in children with WS. Yet, there are dissociations found in the adult state. It seems that the neuroconstructivist view is focusing exclusively on development whereas the nativist view is focusing on the end state.

However, both these viewpoints are crucial for the ultimate understanding of human cognitive architecture, and they need to be viewed in synergy with each other in order to have the optimum explanatory power. In this respect, research into developmental disorders may lead the way to a more unified theoretical framework. Such a framework will have the power to explain developmental and end-state cognitive patterns reported not only for WS and DS but developmental disorders in general.

29.6 Directions for future research

What is particularly lacking in our present knowledge base are investigations of early linguistic and non-verbal skills in WS and DS. There have been only a small number of studies of infants with WS and DS. Mundy *et al.* (1995) identified non-verbal requesting as a possible predictor of expressive language in DS. However, although this study was longitudinal in nature, it did not go beyond the age of 36 months. This is the age at which many children with DS would still be in the holophrastic stage of language acquisition. Hence, it is impossible to know how language development proceeds beyond this age. Laing *et al.* (2002) investigated early language development in infants with WS, also in the context of how well early social communication skills predict language outcomes. The study does not provide a longitudinal examination of language skills *per se*, so we do not know how linguistic skills unfold over time in the first few years of life.

However, an interesting finding emerged from Laing *et al.*'s study. These investigators found that pointing may not necessarily precede the onset of first words in WS as it does in typically developing children. The question arises as to whether an atypical trajectory of development, as evidenced from behavioural studies, means by default that modules are not specified for certain cognitive functions from birth. Unless behavioural studies are

complemented by neurophysiological investigations, it will be very difficult to move forward the innate modularity debate. Our current state of knowledge would also benefit from carefully constructed, longitudinal studies including both behavioural and neurophysiological measures. These studies should examine in detail the acquisition of linguistic abilities as well as non-verbal skills in children with DS and WS from the onset of the two-word stage until about 10 years of age.

It goes without saying that the linguistic and general cognitive profiles of individuals with WS and DS are the result of brain development which occurs under certain genetic constraints. Given technological advances in cognitive neuroscience, and in particular the availability of a range of neurophysiological measures such as magnetic resonance imaging and electroencephalography, future research has the potential to explain the neurophysiological bases of resulting phenotypes. Possible neural localisation and impenetrability of different 'modules' from infancy would directly address the issue of innate modularity. Studies using event related potentials (ERPs) have shown that individuals with WS do not develop the hemispheric asymmetries associated with the processing of closed versus open class words by individuals in the general population (Neville *et al.* 1994). This suggests that the neural organisation of some aspects of language in WS may be different from that of the general population. However, we still do not know exactly what this means in terms of whether modules are innately specified or are a product of development. ERP studies of infants with WS would help address this issue.

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