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Language development in genetic disorders

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27.1 Introduction

The study of language in developmental disorders is an important endeavour for several reasons. First, it is essential to identify areas of relative strength and weakness in order to gain a profile of the disorder, so that we may best support and facilitate the development of language and communication skills in these individuals. Second, such research provides an insight into questions about the process of typical language development. For example: to what extent do biological factors influence language development? Does language learning rely on general cognitive processes, or processes that are specific to language? In this chapter, we focus on the process of language development in two contrasting developmental disorders: (i) Williams syndrome and (ii) Down syndrome. We provide an overview of communicative development in Williams syndrome (a more detailed description of the language profile of individuals with Williams syndrome can be found in Chapter 28), and a description of the developmental profile of communicative skills in Down syndrome. We will then discuss what has been learned about typical language development through the study of these disorders, as well as introducing new methodologies and techniques for studying cognitive development. Specifically, we will explain how developmental trajectories can be used to characterize both typical and atypical development as a process, and how population modelling techniques can be used to investigate the causal mechanisms of atypical development and behaviour. We conclude with a discussion of future challenges in the study of developmental disorders, and issues that are as yet unresolved.

Both Williams syndrome (WS) and Down syndrome (DS) are genetically defined disorders. WS is caused through the deletion of approximately twenty-eight genes from one copy of chromosome 7.[1] The incidence of WS

[1] The length of missing DNA is well understood, while the functional role of the relevant base pairs is a topic of active research.

is rare, occurring in approximately 1 in 20,000 live births (Morris, Demsey, Leonard, Dilts & Blackburn 1998). DS is more common by comparison, affecting approximately 14 in 10,000 live births (Roberts, Price & Malkin 2007) and is the result of three copies (referred to as *trisomy*) of chromosome 21 (Tassabehji 2003). In typical individuals, there are only two copies of chromosome 21 – one from each parent. Both disorders result in some degree of learning disability or learning difficulties, with IQ in WS typically falling between 51 and 70 (Donnai & Karmiloff-Smith 2000, Mervis & Becerra 2007), and in DS ranging from 35 to 70 (Chapman & Hesketh 2000). Both disorders are also accompanied by a series of clearly distinguishable physical characteristics. For example, individuals with WS and DS frequently suffer from co-occurring heart problems and growth deficiency; individuals with DS may also suffer from respiratory problems. A prominent feature of both disorders is that they are accompanied by a distinctive facial appearance (Morris 2006, Roizen & Patterson 2003).

The profile of verbal and nonverbal skills differs between the disorders. In WS, overall IQ measures mask areas of relative strength and weakness in mental abilities, such as language, problem-solving ability, and visuospatial processing, resulting in an uneven *cognitive* profile (see Chapter 28 for additional discussion on the cognitive profile of WS). Language in WS is frequently hailed as being a particularly strong skill (Thomas & van Herwegen 2014). Notably, compared to overall mental age, individuals with WS tend to develop extensive vocabularies that exceed expectations when compared to typically developing children with the same mental age (Bellugi, Marks, Bihrle & Sabo 1988). Children with WS also have a relatively good auditory rote memory processing, having a longer forward and backward digit span than typically developing individuals matched for mental age, as well as individuals with DS (Klein & Mervis 1999). By contrast, visuospatial skills in WS are particularly poor, for example as measured through the use of drawing and pattern construction tasks (Mervis *et al.* 2000, Udwin & Yule 1991, Wang & Bellugi 1994). WS is also characterized by a distinctive personality profile, which is described as hypersociable or 'over-friendly'. However, individuals with WS are also prone to suffering from anxiety, particularly when in unfamiliar surroundings, or faced with a new set of circumstances (Klein-Tasman & Mervis 2003).

In terms of the profile of strengths and weaknesses in DS, visuospatial and visuomotor skills are considered to be relatively good compared to overall mental age (Klein & Mervis 1999). However, in DS spoken language can be problematic. This is, in part, due to differences in facial musculature and oral structure (such as a larger tongue and smaller palette) that can limit the speed and range of motion in mouth movements, making articulation more difficult, and resulting in poor clarity of speech (Dodd & Thompson 2001, Miller & Leddy 1998). Moreover, cranial facial differences and narrow auditory canals, in conjunction with a slight deficiency of the

immune system, results in a susceptibility to *Otitis media* – inflammation or infection of the middle ear. This can lead to fluctuations in clarity of hearing or even hearing loss, which occurs in approximately two-thirds of children with DS (American Academy of Paediatrics 2004, Roberts *et al.* 2004, Roizen 2002). Early loss of hearing in DS has a profound impact on subsequent language development which cannot be explained solely by learning difficulties or linguistic factors (Laws & Hall 2014). Therefore, a combination of issues makes it more difficult for individuals with DS to develop clear well-articulated spoken language. In general, individuals with DS have an outgoing nature, which is consistent with the perspective that socio-cognitive skills in DS are a relative strength, and that children with DS are socially motivated communicators. However, infants and young children with DS do exhibit subtle differences in their socio-cognitive development, which affect early communicative and subsequent linguistic development (Cebula, Moore & Wishart 2010, Fidler, Hepburn & Rogers 2006).

In the following section, we consider the similarities and differences in the process of early communicative development in WS and DS in comparison to typically developing children, indicating how the initial characteristics of these disorders shape language learning from the very beginning of the acquisition process.

27.2 Early communicative development

The initial development of communication skills begins in infancy (as discussed in Chapter 5) with the use of nonverbal elements, such as gestures and eye gaze. This communication takes place between the infant and caregiver (*dyadic interactions*), and subsequently between the infant, caregiver and referents such as objects or toys (*triadic interactions*). It is these initial patterns of interaction that underpin the development of *conceptual knowledge* – learning how objects are used and in what context; and *vocabulary* – learning what objects are called. In this section, we consider these early stages of communicative development in WS and DS, where initial differences from typically developing infants may be clearly identified.

27.2.1 Early communicative development in WS

The strong desire for social interaction that characterizes individuals with WS is apparent in infancy through a keen interest in faces. However, this results in infants with WS preferring to look at the face of their caregiver, as opposed to engaging in gaze-following behaviour, which is usually seen in typically developing infants (Bellugi, Bihrle, Neville, Jernigan & Doherty 1992). This initial reluctance makes more complex interactions between the infant, caregiver and an object or toy problematic, because toddlers

with WS have difficulty in switching their attention from the caregiver to an object being referred to during communication (via pointing, looking and naming).

During the early stages of communication development, typically developing children use deictic gestures such as pointing as well as eye gaze to direct the attention of their caregiver to objects. This behaviour facilitates the child in learning the terms of reference for objects and events. Since shared attention to newly named objects is one of the main routes into the development of vocabulary knowledge (see Chapters 5 and 29; Carpenter, Nagell & Tomasello 1998), difficulty in triadic interactions places toddlers with WS at a disadvantage in vocabulary development. Indeed, difficulty in triadic interactions is considered to be a major source of the delay in the development of vocabulary knowledge in WS (Laing *et al.* 2002, Mervis *et al.* 2003) (for further discussion see Chapter 28). Mervis and Bertrand (1997) found that in WS the use of pointing behaviour emerges after these children start to use verbal labels. This finding was confirmed by Laing *et al.* (2002), and could not be attributed to any *deficit* (difficulty in performance below typical level) in fine motor skills that could potentially impede the development of pointing behaviour. Thus the development of early nonverbal communication skills in WS deviates from that found in typical development.

The age at which children with WS produce their first words is delayed by approximately 24 months. The use of two-word combinations is also delayed and occurs at a slower pace in comparison to typical development. However, both delay in onset, and the slower pace of acquisition in grammatical development is less than that seen in children with DS (Levy & Eilam 2013). The development of productive vocabulary in WS often matches, or sometimes even extends beyond mental age expectations. This rapid growth has been attributed to the high attentional value placed on verbal input and increased auditory memory for words found in WS, rather than the early use of *semantic* knowledge to support vocabulary growth (Mervis & Bertrand 1997, Singer-Harris, Bellugi, Bates, Jones & Rossen 1997, Thomas & Karmiloff-Smith 2003). Indeed, there is preliminary evidence that compared to typically developing children, those with WS show a reduced comprehension vocabulary in comparison to their production vocabulary (Paterson 2000), implying that these children have a poorer understanding of word knowledge than their use of vocabulary suggests.

27.2.2 Early communicative development in DS

Like infants with WS, infants with DS also encounter difficulties that result in a delay in establishing early nonverbal communication skills. Specifically, infants with DS usually have difficulty in establishing mutual eye contact with the caregiver, which makes the development of patterns

of interaction more challenging (Berger & Cunningham 1981, Jansow *et al.* 1988). Moreover, once this initial problem is resolved, infants with DS prefer to continue to focus on the eyes of their caregiver, rather than the facial features, as young infants typically do (Berger & Cunningham 1981). This can subsequently lead to further difficulties in establishing more complex nonverbal exchanges. For instance, triadic interactions which involve joint engagement – whereby an object or event is shared with a caregiver, are delayed in children with DS. Also, children with DS initiate fewer communication exchanges than seen in typically developing children (Adamson, Bakeman, Deckner & Nelson 2012). These early difficulties in the development of requesting behaviours, such as the use of gaze following, gesture and vocalization to communicate with a caregiver can be moderated through intervention. However, the long-term effect of such intervention on subsequent language development is not yet known (Feeley, Jones, Blackburn & Bauer 2011, Fidler *et al.* 2006).

Although the development of nonverbal communication skills is delayed in DS, socio-communicative skills are a relative strength – particularly in comparison to children with WS. Children with DS are much better at understanding the meaning (*communicative intent*) behind pointing and gazing gestures, and at 36 to 60 months perform at a similar level to typically developing children at 18 months of age (Behne, Carpenter & Tomasello 2005, John & Mervis 2010, Thurman & Mervis 2013). This result demonstrates the stronger pragmatic skills in children with DS relative to WS, and supports the perspective that children with DS are socially motivated communicators.

Once nonverbal communication skills are established children with DS have a strong preference for the use of communicative gestures over and above vocal production (Chan & Iacono 2001), and produce more gestures than seen in typical development (Caselli *et al.* 1998) and more than those observed in WS (Singer-Harris *et al.* 1997). This additional use of gesture is considered to be a method of compensating for the delay in the onset of developing spoken language due to frequently occurring articulatory difficulties (Singer-Harris *et al.* 1997).

In addition to a difference in the number of gestures produced by children with DS, there are also some qualitative differences in the types of gestures produced. A comparison of the types of gestures used by children with DS versus typically developing children has found that children with DS produce more deictic gestures (pointing, giving, showing) and iconic gestures (depicting the use of an object – such as gesturing the use of a spoon), and use a particularly large number of iconic gestures in their communication (Stefanini, Caselli & Volterra 2007). The use of iconic gestures, in particular, implies that these children have conceptual knowledge and are extracting meaning from their environment, which is not necessarily evident through their expressive language skills. Whilst the quantity and type of gestures used by children varies subtly from

typical development, the role of gesture in communicative development in children with DS appears to be similar to that of typical development, in that gesture provides a bridge between comprehension and language production. A study of typically developing children and children with DS found that the number of gestures produced at 36 months of age was positively correlated with vocabulary production at 42 months (Zampini & D'Odorico 2009).

Early vocal development through babbling occurs at a similar time during development in children with DS and typically developing children, but can be delayed and possess fewer speech-like sounds. This is in part attributed to the articulatory difficulties that can occur in conjunction with this disorder (Miller, Sedley, Miolo, Murray-Branch & Rosin 1992). This period of delay, however, has been found to be as small as two months (Lynch *et al.* 1995), and is small in comparison to the delay observed for other developmental milestones in motor skills and vocal development (Kent & Vorperian 2013). The age at which children with DS produce their first words can vary. For instance, parental report measures indicate that 80 per cent of children with DS have produced their first words by the age of 3 years, although some children can take longer (Berglund, Eriksson & Johansson 2001). However, a recent study of developmental milestones, which included children with DS, placed the production of first words at ~13 months, which is within the range of 8 to18 months of age during which typically developing children produce their first word (Horovitz & Matson 2011). The pace at which MLU increases in children with DS is slower than that of typically developing children, who reach a MLU of 3.8–4.4 within 16 months of producing their first words, whereas children with DS take ~74 months to reach this stage. Also, onsets for these phases of language development appear to have a larger variability than that seen for typically developing children, where although there is a degree of variance in ability, the variance in onset is limited. This suggests that the timing of developmental milestones may be critical in the process of language development (Levy & Eilam 2013).

27.3 Patterns of language development

The development of language is heavily dependent upon the extent of the learning difficulties of the individual. Although there are cases of children who show exceptional language proficiency despite low IQ (Cromer 1994), it is usually expected that language ability in a child with learning difficulties will not surpass that of their mental age (Miller, Chapman & Bedrosian 1978). For example, children with DS with a low IQ (below 50) may never develop complex structured language (Miller 1988). Furthermore, in order to succeed in acquiring language, children must also be socially motivated with the desire to communicate,

and have some ability to understand the thoughts and intentions of others. In conjunction, these factors are crucial to the level of overall proficiency attained.

In the following section, we consider different aspects of language development in WS and DS, and discuss how these children differ from the typically developing population.

27.3.1 Later language learning in WS

Although the main feature of language development in WS is delay (Brock 2007, Thomas *et al.* 2006, Thomas *et al.* 2001, Thomas & van Herwegen 2014), the eventual outcome is relatively successful in that in most cases children with WS become proficient users of language. However, this is not to say that language in these individuals is typical; indeed, there is a disparity between different aspects of language that results in an *atypical* profile (differing from typical development). Moreover, there is also variability in terms of the relative strengths and weaknesses found in both linguistic and cognitive skills (Porter & Coltheart 2005). Children with WS usually develop an extensive vocabulary and complex syntax – though their vocabulary skills generally exceed their syntactic ability in terms of mental age (Karmiloff-Smith *et al.* 1997). However, experimental evidence indicates that children with WS develop knowledge of the core elements of syntax without formal instruction despite their cognitive impairment, although this development is not typical (for a detailed discussion see Chapter 28). This profile of language skills contrasts with that found in children with Specific Language Impairment (SLI), who have a particular difficulty in processing grammatical constructions (see Chapters 24–26). In typically developing children there is a relationship between the length of utterance and the complexity of grammatical structure – this relationship also holds in WS (Mervis, Morris, Bertrand & Robinson 1999).

Children with WS are known to make more errors in morphology than in syntax, that is, in verb tense and agreement and personal pronouns (Karmiloff-Smith *et al.* 1997, Volterra, Capirci, Pezzini, Sabbadini & Vicari 1996). Also, French-speaking children with WS find grammatical gender particularly difficult (Karmiloff-Smith *et al.* 1997) (a detailed description of this study and further discussion on morphology in WS can be found in Chapter 28). Whilst the cognitive profiles of children with WS have been studied across many different languages, current work reflects a similar profile to that found in English-speaking children with WS (Levy & Bechar 2003). Although, syntactic complexity is higher than expected upon the basis of nonlinguistic skills, such as visuospatial construction or reasoning, these children nevertheless fall below expectations based upon levels of receptive vocabulary ability or auditory short-term memory (Mervis *et al.* 1999). A close inspection of grammatical abilities was carried out by Mervis *et al.* (1999) using the Test of Receptive Grammar (TROG: Bishop 1983),

which assesses the ability to understand different types of sentence constructions of varying levels of complexity. This study was carried out with a large sample of 77 individuals, between the ages of 5 and 52 years. Only 18 per cent of the participants (22 per cent of the adults) passed the test block that assessed right-branching relative clauses (e.g. *The girl chases the dog that is jumping*) and only 5 per cent (9 per cent of the adults) passed the block assessing centre-embedded relative clauses (e.g. *The duck the ball is on is yellow*).

In terms of the development of semantic knowledge, children with WS exhibit a relative strength in category concepts (e.g. the distinction between animals, tools, clothing, furniture). This contrasts with problems understanding semantic relational concepts. For example, children with WS have difficulty in understanding sentences containing spatial terms of reference (such as *The bottle is in the boat*; Phillips, Jarrold, Baddeley, Grant & Karmiloff-Smith 2004). Within category concepts, evidence has indicated differential naming problems across categories, such as animals being named better than foods (Robinson & Temple 2009, Thomas *et al.* 2006). On the basis of such evidence it has been argued that the *lexicon* is an area of specific anomalies in WS (Clahsen & Almazan 1998, Rossen, Klima, Bellugi, Birhle & Jones 1996, Temple, Almazan & Sherwood 2002). Fractionation such as this also appears in other areas of the WS language system (Thomas 2006). For example, in the area of pragmatics children with WS have relatively good social sensitivity (e.g. making dyadic eye contact, sensitivity to nonverbal cues) but exhibit problems in areas such as greeting behaviours, topic maintenance and answering questions (Semel & Rosner 2003). The results concerning semantic knowledge in WS must be interpreted with some caution since they are somewhat dependent on the task used to assess it. Categorization tasks have been shown to produce better performance in participants with WS compared to giving definitions, since the latter task requires metacognitive knowledge of what is required to give a full definition of a semantic category such as 'elephant' (Purser, Thomas, Snoxall, Mareschal & Karmiloff-Smith 2011).

Thomas and Karmiloff-Smith (2003) characterize two types of hypotheses regarding the developmental profile of WS: (i) a series of *imbalance* hypotheses, which account for the profile shown in WS in terms of an imbalance in the integration of phonological and semantic processing, and (ii) a *conservative hypothesis*, which proposes that language development in WS is delayed but not fundamentally altered. In the latter case, any anomalies in the language profile of children with WS would be accounted for by nonlinguistic characteristics of the disorder – such as a strong desire for social interaction and poor visuospatial skills (Brock 2007). Research continues in an attempt to distinguish these respective hypotheses (see, e.g., Lee & Binder 2014 for recent evidence in favour of the *imbalance* hypothesis).

A debate continues with respect to whether syntactic knowledge in WS is typical or atypical, to which we return in Section 27.4 (for additional discussion, see also Chapter 28). Claims that syntactic knowledge is typical in WS are often associated with modular interpretations of cognitive development, whereby it is proposed that components such as syntactic knowledge can develop independently and normally in the face of atypical or delayed development in other cognitive domains (such as those underpinning nonverbal skills). For example, Musolino, Chunyo and Landau (2010) investigated logico-syntactic properties of negation and disjunction (for a detailed description see Chapter 28). The authors concluded that individuals with WS acquire language no differently and develop grammars indistinguishable from those of typically developing individuals, and thereby with respect to syntax, language acquisition is not fundamentally altered in WS.

Given Musolino *et al.*'s (2010) conclusion, it is worth noting that the performance of the individuals with WS in this study was nevertheless fairly poor on the experimental tasks (Thomas, Karaminis & Knowland 2010). The individuals with WS, who had a mean chronological age of 16, were given a language comprehension task in which the task demands were greatly simplified: the individuals only had to decide, under no time pressure, whether a spoken sentence was a correct depiction of a pictorially represented scenario (yes or no). The individuals with WS performed much worse on this task than would be expected for their age (estimated from the performance of undergraduate students). The individuals with WS performed so poorly that they were worse than 6-year-old children, some ten years younger. The task also included control conditions, which were used to predict performance on the key experimental conditions. The individuals with WS showed a different relationship between control and experimental conditions compared to the 6-year-olds. They did, however, show a similar relationship as that observed in 4-year-old children (some twelve years younger), while performing at a higher overall level than the 4-year-olds. That such task performance somehow constitutes evidence of 'typical' language acquisition within the modular framework evidences the role of the theoretical assumptions that researchers bring to bear in interpreting the theoretical significance of language skills in WS.

27.3.2 Later language learning in DS

There are limitations in the overall level of complexity of language attained in DS. Moreover, the production of intelligible speech remains a persistent difficulty. Disturbances in voice, articulation, fluency and prosody are assumed to contribute to poor intelligibility; although the relative contribution of each of these factors is unclear (Kent & Vorperian 2013). Phonological development is delayed, proceeding at a slower rate in terms

of mental age expectations and is associated with more error-prone production (Barnes *et al.* 2009, Bleile & Schwartz 1984, Roberts *et al.* 2005, Stoel-Gammon 1980), as well as atypical errors not usually seen in typically developing children (Cleland, Wood, Hardcastle, Wishart & Timmins 2010). Children with DS also make a higher number of systematic sound changes to lower the phonological complexity of words than seen in typically developing children (Barnes *et al.* 2009).

The development of vocabulary knowledge in DS is also slow, and has been characterized as showing poorer expressive vocabulary relative to receptive vocabulary (Ypsilanti, Grouiod, Alevriadou & Tsapkini 2005). Some children with DS experience a vocabulary spurt (Berglund, Eriksson & Johansson 2001, Klein & Mervis 1999); but this tends to occur at a more advanced age than seen in typically developing children. A recent study of vocabulary knowledge found that the difference between receptive and expressive vocabulary knowledge for children with DS was similar to that of typically developing children matched for vocabulary size (Polišenská & Kapalková 2014). However, a study by Roberts, Price, Barnes *et al.* (2007) indicated that 8-year-old children with DS have a weaker receptive vocabulary than typically developing children matched on the basis of nonverbal cognitive ability. However, this pattern was not present in a study of 12-year-old children with DS, suggesting that individuals with DS continue to acquire vocabulary, catching up during adolescence and early adulthood (Finestack, Sterling & Abbeduto 2013). A qualitative analysis looking at the types of words present in the vocabularies of 16-year-olds with DS also found that the types of words present were very similar to those of typically developing children matched for developmental level (Facon, Nuchadee & Bollengier 2012). Therefore, studies to date suggest that the process of vocabulary acquisition in DS is delayed, and possesses similar characteristics to typical development rather than being atypical or deviant.

Grammatical development varies widely in DS, and is impaired more than would be expected on the basis of intellectual disability (Martin, Losh, Estigarribia, Sideris & Roberts 2013, Roberts, Price & Malkin 2007). For individuals with DS whose language development does progress beyond the use of two-word utterances, utterances tend to be shorter in comparison to typically developing children (as measured by mean length of utterance), and consist of less complex noun phrases and verb phrases in comparison to typically developing children matched for mental age (Price *et al.* 2008). Finestack *et al.* (2013) found that expressive grammar is particularly weak in children with DS in comparison to typically developing children, even after controlling for mental age. Also, like children with SLI there are difficulties in grammatical morphology (Eadie, Fey, Douglas & Parsons 2002). Overall, the development of syntax in DS is particularly challenging, and has a prolonged developmental span, with increases in syntactic complexity and utterance length being known to continue throughout adolescence and into early adulthood (Chapman, Hesketh & Kistler 2002).

The development of pragmatics abilities in children with DS is delayed relative to mental age expectations. However, pragmatic language skills appear to present less of a challenge in comparison to the acquisition of vocabulary and syntax (Martin *et al.* 2013). Also, children with DS are socially motivated in their use of language, and display the same range of communicative interests and interactions as typically developing children. However, children with DS differ from typically developing children in their quality of topic maintenance – using less elaborate forms of topic maintenance – although the frequency of initiating a topic is similar to that of typically developing children (Roberts, Martin, Moskowitz *et al.* 2007). Although children with DS do not perform at the same level as typically developing children, it is clear that individuals are capable of holding and maintaining conversations in a similar way to typically developing children.

27.3.3 Summary

Overall, the development of language skills in both WS and DS is delayed, as well as featuring differences in the rate and overall level of acquisition. Early nonverbal communicative skills (such as eye gaze, gesture and pointing) are delayed, but also possess distinct differences across the two disorders. For instance, children with WS experience more difficulties with interpreting communicative intent than would be expected given their cognitive and linguistic abilities. By contrast, children with DS are more effective at interpreting nonverbal gestures than children with WS, and develop a preference for the use of gestures over vocalisation in order to communicate; which is in part driven by articulation difficulties that make the development of intelligible speech challenging. In children with WS, the development of productive vocabulary is rapid and indeed exceeds expectations based on mental age. However, the rapid development of productive vocabulary in WS is not necessarily met with a corresponding level of understanding. Language in children with DS asymptotes at a lower level of complexity, and is hindered by articulation difficulties. However, children with DS possess a relative strength in the use of pragmatic language skills, but find grammar particularly difficult. Thus, both disorders exhibit an uneven profile of strengths and weaknesses both with and across different aspects of language (such as phonology, vocabulary, grammar and pragmatics).

27.4 What can WS and DS tell us about language and cognitive development?

The differing profiles of linguistic and nonlinguistic skills in WS and DS illustrate the ways in which the typical developmental process may be

deflected. The comparison of language skills across syndromes is particularly informative in terms of establishing what skills are crucial to successful language acquisition. In general, the contrasting language skills of those with WS and DS indicate that general cognitive ability cannot be considered to be a reliable indicator of all aspects of language function in children with learning difficulties. Comparisons carried out both in early development (Tager-Flusberg & Sullivan 1998) and later childhood (Fowler 1998), suggest that pragmatics and semantics are more closely linked to overall mental age across different disorders, while phonology and syntax can dissociate. McDonald (1997) compared language acquisition across different disorder groups (including WS and DS) who exhibited varying degrees of success and concluded that good representations of speech sounds (phonology) are a critical requirement to the successful development of language. However, Morton (2004) argued that successful language acquisition is dependent upon multiple cognitive components, and that impairment in any one of these can potentially result in the system failing to develop typically. Therefore, under Morton's view, good phonological skills may be a necessary but not sufficient requirement for successful language acquisition. Consideration of what components of language are critical to successful acquisition raises key questions about how these components (and other nonverbal cognitive components) emerge as a product of development and what happens when something goes wrong.

27.4.1 Models of cognitive development

In broad terms there are two contrasting perspectives as to how the underlying structure of the human cognitive system develops. The first of these, the *nativist* view draws heavily upon the field of adult cognitive neuropsychology. Within this framework, separate components of the cognitive system are identified through the use of dissociations between specific aspects of cognitive abilities. For example, within the context of language processing, patients with acquired deep dyslexia typically struggle with the pronunciation of abstract versus concrete word types (Plaut & Shallice 1993). The observation of an opposite pattern, known as a *double dissociation* is taken as evidence that these two cognitive components (abstract and concrete word processing) are represented separately in the brain. Specific processing components or *modules* are labelled as being *spared* when performance for a given cognitive task falls within the normal range, or *impaired* when performance is comparatively poor (for discussion of this cognitive profile in relation to WS see Chapter 28). Within the context of developmental disorders, a specific difficulty or strength in a given aspect of language processing is viewed as reflecting the underdevelopment or overdevelopment of that specific component of the language system (Clahsen & Temple 2003).

This framework provides a comfortable fit between the results of standardized language tests and atypical functional structure. Assuming we have tests that give an indication of the integrity of individual modules (e.g. tests of vocabulary, tests of grammar, tests of phonological processing, and so on), scores in the normal range of performance on a given test can be interpreted as reflecting a typically developed component. By contrast, scores above or below the normal range can be read as reflecting an (atypically) over- or underdeveloped component. This correspondence between test scores and underlying modular structure within the context of developmental disorders rests upon one of two underlying assumptions. First, that the structure of the modular system identified in adults is also present in the infant – so that language development can commence with an initially selective anomaly in one or more components; or that modular structure emerges in such a way that when things go wrong, some parts emerge with atypical functionality while the rest nevertheless manage to emerge displaying their normal functionality. Together, these alternatives constitute the assumption of *residual normality*, whereby the rest of the system can develop normally irrespective of a selective difficulty in one processing component (Thomas & Karmiloff-Smith 2002).

The second and contrasting explanatory model of cognitive development argues that the process of development itself must play some role in shaping the underlying structure of the cognitive system and the profile of any given developmental disorder. This framework, known as *neuroconstructivism,* is based on the premise that components of the adult cognitive system are a *product* of the process of development and not initially present in infancy (Mareschal, Johnson, Sirios, Spratling & Thomas 2007). This view is strongly motivated by data from developmental cognitive neuroscience (Elman *et al.* 1996, Karmiloff-Smith 1998), and calls into question a key assumption of the modular view, which is that performance within the normal range on a given test of cognitive ability is an indicator of an *intact* or *spared* cognitive module. Instead, neuroconstructivism argues that performance within the normal range may be achieved through atypical means, and that the underlying mechanisms that give rise to the same level of performance may be fundamentally different. Therefore, experimental tasks that are sensitive to the on-line operation of cognitive processes are necessary before claims of typicality can be substantiated. Standardised cognitive and language tests frequently do not have this characteristic. (For discussion of these two opposing theoretical views in relation to WS see Chapter 28). Deficits within this framework are therefore unlikely to be module-specific, affecting lower-level cognitive processes that have different degrees of relevance for different cognitive skills. This is particularly relevant in the study of genetically defined developmental disorders, where cross syndrome comparisons have revealed many common neural and socio-cognitive deficits in infancy, but have differing phenotypic outcomes. In this context, it is important

to track the developmental trajectories of disorders over time in order to understand how phenotypic outcomes originate in infancy (Karmiloff-Smith *et al.* 2012).

The debate between these two explanations of uneven linguistic profiles has at times become polarized. On the one hand, there are strong claims that for given developmental disorders, certain cognitive structures must have developed normally given behaviour in the normal range (sometimes these are referred to as *intact* or *spared* systems) or even, as we saw in the previous group, behaviour that resembles some much younger typically developing control group. On the other hand, there are counterclaims that since the developmental processes we know about could not have produced such an uneven modular outcome, the relevant behaviour must be produced by structures that are at least quantitatively, and perhaps qualitatively different. In the following section, we explore the relationship between genetically determined deviations in the structural development of the brain and the cognitive phenotypes of WS and DS.

27.4.1 From genotype to phenotype

The process of development is shaped by combined genetic and environmental influences. Within the context of developmental disorders, the nativist perspective predicts that the effects of a deletion or duplication of genetic material will result in specific impairments, whereas within the neuroconstructivist framework the effects of genes operate at a more basic level during brain development, subtly altering developmental pathways and resulting in widespread differences that are profound in some cognitive domains but weaker in others (Karmiloff-Smith 1998, 2009). As both WS and DS are clearly defined genetic disorders, they provide a unique opportunity to investigate the influence of genes upon the developing cognitive system. Specifically, research has focused upon the genetic regulation of brain development in order to establish links between genes, deviations from typical brain structure, and the behavioural phenotype of the disorder in question. Deviations in brain structure may include differences in overall or regional brain volumes, cortical thickness, patterns of cortical folding, differences in patterns of brain symmetry (between left and right hemispheres) or anatomical (white matter) connectivity.

Neuroanatomical research into WS has identified a plethora of structural differences. Firstly, a consistent feature of the brains of those with WS is an overall reduced cerebral volume relative to typical individuals, which is at least, in part, attributed to a reduction in cerebral white matter (Martens, Wilson & Reutens 2008). (See Chapter 24 for a discussion of white matter in relation to SLI, and Chapter 33 for a discussion of white matter in relation to dyslexia.) As deletions on chromosome 7 include genes involved in the regulation of white matter development, alterations in the white matter tract pathways are likely to be due to this deletion

(Hoeft *et al.* 2007, Jackowski *et al.* 2009, Marenco *et al.* 2007). As white matter pathways (see Chapter 4) are essential for effective and efficient communication between different brain regions, disruption of these pathways is likely to affect higher-level cognitive processes that are characterized by the interaction of multiple regions distributed throughout the brain.

In addition, grey matter volume is reduced in the superior parietal and occipital cortex (Boddaert *et al.* 2006, Martens *et al.* 2008) and contrasts with the proportionately higher volume of the frontal lobes (Reiss *et al.* 2000, Reiss *et al.* 2004). Reduced grey matter volume has also been found in the intraparietal sulcus (Meyer-Lindenberg, Mervis & Berman 2006). Both parietal (precuneus) and occipital (cuneus) regions also show a pattern of increased gyrification. This atypical cortical folding is a common feature in the WS brain (Fahim *et al.* 2012, Gaser *et al.* 2006, Kippenhan *et al.* 2005, Van Essen *et al.* 2006). Genes on chromosome 7 have been implicated in *morphogenesis,* the developmental process of cortical folding, consistent with the view that the atypical patterns of folding are a consequence of the deletion. Interestingly, as the timing of this developmental process differs according to brain region, some regions are more affected than others (Fahim *et al.* 2012). Structural differences in the parietal and occipital lobes of those with WS are linked to the visuospatial processing and numerical deficits associated with the WS phenotype (Boddaert *et al.* 2006). Although structural differences in temporal brain regions, such as reduced asymmetry, may be linked with the language skills in WS (Dennis & Thompson 2013, Eckert *et al.* 2006) no specific links between any aspect of language ability and the deleted genes on chromosome 7 have been made.

Whilst WS provides an opportunity to study the effects of a deletion of genetic material, the effects of additional genetic material upon brain structure are studied in DS. Research into the relationship between brain structure and the DS phenotype has included the use of mouse models in order to understand the relationship between trisomy and brain development. These studies have found that cellular defects during brain development that alter the quantity and types of neurons produced, particularly within the mouse forebrain, hippocampus, and cerebellum, as well as a delay in the development of white matter pathways (Haydar & Reeves 2012). Whilst a mouse with trisomy on chromosome 21 cannot be said to have DS, mouse models do exhibit deficits in learning and memory, thus suggesting that altered gene expression levels as a result of trisomy on chromosome 21 are responsible for the learning difficulties associated with DS (Wiseman, Alford, Tybulewicz & Fisher 2009). Structural imaging studies in individuals with DS have identified a decrease in total brain volume; in particular the volumes of the frontal and temporal lobes and the hippocampus are reduced (Dennis & Thompson 2013, Gardiner *et al.* 2010, Nadel 2003, White, Alkire & Haier 2003). These data draw many

parallels with mouse models (Haydar & Reeves 2012). However, the precise relationship between genotype and structural brain differences, and how these differences shape the cognitive phenotype of DS remain the topic of ongoing research.

Current studies investigating the relationship between genotype and brain development suggest that low-level broad-spectrum alterations may form the basis of the cognitive differences observed in WS and DS. However, whilst the underlying neural substrate of the cognitive system has a different structural organisation, it is not clear how these differences alter brain function. In addition, as the developmental process involves an interaction between genetic and environmental factors, the environment also plays a role in shaping the course of developmental disorders. In terms of environmental influences, there are two main points for consideration. The first of these is the extent to which differences in the internal cognitive system result in differences in the way in which the environment is experienced. We know that the neurology of the cognitive system in children with WS and DS differs from that of typically developing children, which may in turn result in differences in which the external environment is perceived. Autism is a particularly good example of this (Happé & Frith 1996; see also Chapter 29). Thus, although the external environment may not have been altered in any explicit manner, it may be subtly different from that of a typically developing child. In addition, children with developmental disorders display different initial preferences as to what they find interesting in their external environment (such as a keen interest in faces in WS). This means that the way they manipulate their external environment in order to participate in exchanges they perceive as rewarding may result in a subtly altered role for the environment in shaping the course of development.

The second environmental influence and one of the most important motivating factors for the study of developmental disorders, is how we might support and facilitate effective development through a process of intervention. The degree and type of intervention appropriate may depend upon a number of factors. These include the profile of the individual child and the level of intervention services that may be accessed. Interventional methods seek to manipulate the environment and in doing so attempt to influence the course of development for those with disorders in a positive way. Somewhat counter-intuitively, it may often be useful to interfere and further bolster an area of relative strength (such as language in WS) so that this ability can be used strategically to aid areas of weakness (such as visuospatial skills: Semel & Rosner 2003). For example, a series of memorized verbal cues may be one way to improve picture drawing or tying of shoe laces.

As the different constraints placed on the system in developmental disorders put these children at a disadvantage, it is likely that their cognitive systems will attempt to overcome the challenges they face through the

process of *compensation* (Thomas 2005a, 2005b). The process of compensation within the context of developmental disorders is frequently incomplete, as evidenced by children failing to deliver levels of performance within the range of typically developing children. This may be because the process of compensation has resulted in the atypical system utilizing a secondary, less efficient route to task success, or the system has a reduced capacity or less efficient processing resources (i.e. Bishop 1994b). However, children are renowned for the adaptive capacity, which is generally attributed to a property known as *plasticity* – the flexibility of the learning system to adapt and alter in order to incorporate new information from the environment. Researchers have recently speculated that for some disorders, the major features of the observed cognitive profile may represent adaptive responses to the atypical conditions present in early development, rather than direct outcomes of genetic anomalies (e.g. 2015). Notably, however, the plasticity of the learning system is generally considered to decrease with age (Uylings 2006), and may offer only a short window of opportunity for optimal adaptive change (known as a *sensitive period*; Johnson 2005). In conjunction with genetic and environmental influences, these factors play an important role in shaping the language systems of children with developmental disorders (Fowler 1988).

In sum, developmental disorders such as WS and DS occur as a result of changes in the balance of gene expression due to chromosomal abnormalities, which affect the developmental process of the biological substrate of the human cognitive system at a cellular level. The extent and number of structural differences calls into question whether it is appropriate to label any aspects of cognitive function developing within the normal range as being *spared*. In addition to genetic influences, environmental factors may also influence phenotypic outcomes, which may be due to changes in the cognitive system altering the way in which the environment is perceived. In this respect, the environment should not be viewed as a static influence; the environment can be changed externally, and may also undergo internal changes as the child's ability to interpret the environment or gain knowledge from it alters over time. In the following section we turn to discuss a characteristic feature in the trajectories of cognitive development in children developmental disorders such as WS and DS – developmental delay.

27.4.2 Developmental delay

In Sections 27.2 and 27.3 we used the term *delay* mainly to describe aspects of communicative and cognitive development in WS and DS falling below that of chronological age (CA) matched typically developing controls. In such studies, the disorder group may (also) be matched on the basis of mental age (MA) – typically developing younger controls – in order to establish whether the cognitive ability of the disorder group (as measured

by performance on a specific cognitive task) is *delayed* (below CA group but similar to MA group) or *atypical* (differs from both CA and MA groups). The convention of *matching* essentially controls for (factors out) age in order to characterize the pattern of behaviour for the cognitive skill of interest at that point in time. Whilst this is informative, there are restrictions as to what we can learn using this methodology. For instance, matching does not provide a clear profile of how cognitive ability changes over developmental time (age) – a function also known as a developmental *trajectory* (Thomas *et al.* 2009, Knowland & Thomas 2011). This approach is a departure from traditional methodologies that involves matching typically developing children and disorder groups on the basis of CA or MA. The use of descriptors such as 'spared', 'intact' or 'impaired', adopted from the neuropsychological model used for studying acquired disorders in adults – frequently adopted in the context of developmental disorders, is also replaced with a richer terminology for characterizing deviations from typical development. In this section, we will explain how adopting the trajectories approach provides a more detailed descriptive framework for characterizing developmental delay, and interpreting the role of adaptive and compensatory mechanisms in the process of development.

The aim of the trajectories approach is to build a function linking performance with age on a given cognitive task in order to determine how this function differs from typical development. The methodology for this approach when implemented as a cross-sectional study design involves a disorder group that spans a reasonable chronological age range (e.g. from childhood to adulthood) and a typically developing (TD) comparison group that spans from the youngest mental age of the disorder group to the oldest chronological age (the trajectories method can also be implemented via longitudinal tasks; indeed this would be the ideal method for studying developmental trajectories). Looking at if and where the performance of the disorder group (for a specific task) fits on the TD group trajectory can help us to determine whether the trajectory for the disorder group is similar to that of the typically developing group at some point in developmental time, or whether the trajectory for the disorder group deviates from that of typical development. Crucially, use of the trajectories method provides a more detailed description of *delay*.

The term *delayed* is frequently used to describe the performance of a disorder group in comparison to typical development. In the majority of instances this term simply serves the purpose of re-describing the data, with no explanation as to the causal mechanisms behind the delay. Furthermore, use of the term delay is often insufficient to provide a precise description of performance, because the term actually covers multiple patterns of performance. For instance, delay may refer to a later onset of development, or a lower rate of development, or a combination of both a later onset and slower rate. Therefore, *delay* can actually be described in three different ways: (i) delayed onset, (ii) slowed rate, (iii) delayed

onset + slowed rate. Additional descriptors for differences in trajectory include: (iv) nonlinear trajectory, (v) premature asymptote, (vi) flat trajectory and (vii) no systematic relationship with age. These classifications describe differences in timing and rate of developmental change (i–iii), and trajectories that do not follow a linear pattern (iv, e.g. an s-shaped function). In accord with this approach, Rice (2004, 2012) suggests that separate trajectories should be established for each of the subcomponents of the language system.

The use of the developmental trajectories approach allows for at least three different types of comparison to be made between the trajectory for typical development and the trajectory of a disorder group. A first comparison would be to determine whether the performance of each participant in the disorder group fits anywhere on the TD trajectory. This process effectively standardizes an experimental task or measure of interest, and may not necessarily be of particular interest on its own, but can also be used with two or more measures. In this instance an MA measure for each individual in the disorder group can be used to determine whether any disparity in the disorder group is also present anywhere on the TD trajectory. If it is not, then this would indicate atypical performance of the disorder group. The second comparison involves constructing a trajectory for the disorder group linking task performance with chronological age. The trajectory for the disorder group can then be compared to the TD trajectory to identify any differences in developmental performance. This type of comparison can be particularly useful when exploring performance for cognitive domains outside the primary area of interest (such as nonverbal task performance in children with SLI; see Conti-Ramsden, St. Clair, Pickles, & Durkin 2012). A third type of comparison involves exploring the developmental relations in the disorder group. For this type of comparison, separate trajectories are constructed for the disorder group on a given standardized measure linking performance with mental age. These MA trajectories for the disorder group are then compared against the TD trajectories. This process can be used to determine whether task performance is consistent with the standardised measure. If it is, then the trajectories for the disorder group and the typically developing group rest on top of each other.

There are a number advantages to using the trajectories approach: (i) the process of development itself is central to any explanation of the data. This is in contrast to the matching method, whereby age is essentially controlled-for (factored out), and therefore looks a development as a series of discrete snapshots in developmental time; (ii) it provides an opportunity for multiple analyses to be conducted, increasing the informative value of a dataset. It also allows for more flexible matching, because this approach involves explaining age-related factors rather than controlling for them; (iii) there is a richer descriptive vocabulary available for characterizing different patterns of performance; and (iv) cross-sectional trajectories can

be validated with longitudinal follow-up for efficient design. Although the trajectories methodology requires a wide age range of participants to be tested, it does offer additional flexibility in terms of study design, and a new approach to data analysis.

Therefore, in sum, development is a process of change – in developmental disorders it is vital that we understand what changes are occurring and when, in order to establish the similarities and differences in children with disorders and typically developing children. Exploring the nature of these differences using the trajectories approach provides a richer descriptive vocabulary for characterizing developmental delay. The use of this methodology not only enhances our understanding of developmental disorders but also provides us with an insight into cognitive processes in general, in terms of the emergence of modularity and expertise, and the scale and flexibility of cognitive processes during learning. In the following section, we describe how the developmental process may be simulated in large populations of individuals to explore how different combinations of genetic and environmental influence result in different developmental trajectories.

27.4.3 Population modelling techniques

Even if developmental delay can be better characterized in a descriptive sense, it remains poorly understood as a cause of developmental falling outside the normal range (Thomas *et al.* 2010). Does it merely constitute lower plasticity? If so, performance should eventually reach typical adult levels, but at a later age. This is rarely the case. What other neurocomputational properties could cause delay? When would suboptimal setting of these properties cause trajectories resembling those found in the typical population, but with later onset or slower growth, and when would suboptimal settings produce qualitatively atypical trajectories?

One way to investigate causal mechanisms affecting development is through the construction of computational models. In the current context, the challenge is to explain three phenomena: first, there is variability present in language development in the typical population; second, developmental disorders represent different or greater variability to that observed in the typical population – something has happened to put development on a different course in the disorder group; third, variability is often observed *within* disorder groups, in the severity of the symptoms individuals exhibit. Explanatory models need to be able to reconcile these three forms of variability in terms of modulation of developmental mechanisms.

One recent computational approach has been to simulate large populations of children acquiring a particular language skill (such as verb morphology), and to include intrinsic (neurocomputational) and extrinsic (environmental) factors that interact to produce variability in

developmental trajectories across the whole population. This approach to the computational modelling of language development provides a framework for considering the causes of population-wide individual differences whereby, for example, the bottom tail of a normal distribution of performance can be defined as 'delayed'. Genetic mutations can be considered as a new manipulation to the learning properties of the system occurring in a subset of individuals. The population-wide individual differences in development provide a background against which genetic disorders occur, so that such individual differences translate into protective and risk factors that attenuate or exaggerate the effects of genetic mutations on developmental trajectories, thereby producing variability within the disorder group.

So far, this approach has been applied to consider the causes of delay in typical populations (Thomas & Knowland 2014), as well as variability in developmental disorders (Thomas, Knowland & Karmiloff-Smith 2011a, 2011b), and environmental effects on language acquisition (Thomas, Forrester & Ronald 2013). For example, Thomas and Knowland (2014) simulated an empirical phenomenon whereby in many cases of early-diagnosed language delay (e.g. at 4 years of age), the delay resolves so that the children's language skills later fall in the normal range (e.g. at age 6). Nevertheless, in some children, poor language skills persist, and the children subsequently gain a diagnosis such as SLI. Theoretically, there is a debate about whether resolving and persisting language delay constitute qualitatively different groups, such that, for example, resolving delay is the bottom end of a distribution of variation in typical development, while persisting delay corresponds to a qualitatively distinct deficit in language acquisition.

Thomas and Knowland (2014) simulated a large population of learners in a language domain using an artificial neural network to simulate each child's development. Developmental trajectories were modulated by two factors: first, by variations in the learning ability of each network (produced by variations in a number of underlying processing properties that influenced the *capacity* of a network in terms of how much knowledge it could acquire, or the *plasticity* of a network in terms of how much input it needed to learn); and second, by variations in the quality of the language environment in which each network acquired its language abilities. The authors found that profiles of resolving and persisting delay could be found among the simulated population, even though there were only quantitative differences in underlying mechanisms. Resolving delay tended to be caused by variations in processing properties that impacted on network plasticity, while persisting delay tended to be caused by variations in processing properties that impacted on capacity. In addition, the model generated a novel prediction: the final level of performance for children with resolving delay would depend to some extent on the richness of the language environment in which the child was raised, while for

children with persisting delay, a rich environment could not overcome capacity limitations. This novel prediction was supported by empirical data on children's acquisition of English morphology.

In another example, Thomas *et al.* (2011a, 2011b) used population modelling to simulate developmental regression in autism. This is a pattern observed in a minority of children with autism, where early language acquisition apparently proceeds normally, but in the second year of life, skills then go backwards. Early-acquired vocabulary may be lost, and other cognitive and social skills also can regress (see Pickles *et al.* 2009). In the model, this was captured by the exaggeration of an otherwise normal phase of brain development: the pruning of unused brain connectivity. In the model, exaggerated pruning led to the loss of early-acquired skills. Of particular interest here is that regression only forms one subgroup of children with autistic spectrum disorders (ASDs). Landa, Gross, Stuart and Faherty (2013) also characterize early-onset and late-onset subtypes. In recent work, Thomas and colleagues have shown how interactions between atypical processing properties (in this case, the severity of connectivity pruning once it starts) with population-wide individual differences in other properties (in this case, the onset and speed of pruning) could produce the three different subgroups observed in ASD. That is, in the normal population, there are variations in pruning severity, pruning onset and pruning speed that cause only minor deflections to developmental trajectories (so-called typical individual differences). When one of these properties adopts atypical levels (either by a rare genetic mutation, or accumulation of risk variants common in the population), it can then interact with normal variation in other properties to produce exaggerated variation within the disorder group. In the model, severe pruning with early onset and fast speed produced the *early-onset ASD* trajectory. Severe pruning with early onset and slow speed produced the *late-onset ASD* trajectory. Severe pruning with late onset and fast speed produced the *regressive subtype of ASD*.

Together, these examples demonstrate the important of mechanistic accounts of development, which can account for the varieties of developmental trajectories observed both in typically developing children and in children with developmental disorders. The complexity of the developmental process means that computational modelling can be a useful tool for investigating theoretical hypothesis. Developmental models scaled to the population level allow us to begin to investigate the causes of different types of variability, such as the relationship between variation in typical development and atypical development, and whether we should distinguish different subgroups within developmental disorders. Mechanistic frameworks are necessary to move the concept of developmental delay beyond a description of observed behavioural trajectories to an explanation of their origins.

27.5 Unanswered questions and future challenges

In this chapter, we have explored the profiles of language development in WS and DS. We did so first to gain an insight into the relative patterns of strengths and weaknesses that characterize these disorders, and second to understand how the course of typical language development may be altered. However, a range of unanswered questions remain, which pose challenges for future research. Specifically, how does the functional organization of the language system emerge, and to what extent is this constrained by the processing properties of our neurology? These key questions are important to modular theorists and neuroconstructivists alike. Within the context of developmental disorders, we need to be able to answer questions such as: does deficiency in one component (say, phonology) affect the development of another (say, syntax)? And, what level of disruption is necessary to produce a developmental disorder? Moreover, understanding the processing capabilities of different neural substrates in both typical development and developmental disorders is an important step towards understanding what kinds of differences result in a disorder. In short, we need to understand the parameters that affect the course of development and the different paths that development may take when faced with adverse circumstances. In this respect methods such as brain-imaging and computational modelling of language development may help in characterizing typical and atypical developmental processes (Thomas & Karmiloff-Smith 2003).

Suggestions for further reading

The following reviews provide a useful summary of language and communication skills in DS and WS:

- Mervis, C., & Becerra, A. (2007). Language and communication development in Williams Syndrome. *Mental Retardation and Developmental Disabilities Research Reviews*, 13, 3–15.
- Kent, R., & Vorperian, H. (2013). Speech impairment in Down syndrome: A review. *Journal of Speech Language and Hearing Research*, 56, 178–210.

For further reading on the study of brain structure in WS and DS, see:

- Haydar, T., & Reeves, R. (2012). Trisomy 21 and early brain development. *Trends in Neurosciences*, 35, 81–91.
- Jackowski, A., Rando, K., Maria de Araujo, C., Del Cole, C., Silva, I., & Tavares de Lacerdaa, A. (2009). Brain abnormalities in Williams syndrome: A review of structural and functional magnetic resonance imaging findings. *European Journal of Paediatric Neurology*, 13, 305–16.

For a detailed description of the developmental trajectories methodology:

- Thomas, M., & Annaz, D., Ansari, D., Scerif, G., Jarrold, C., & Karmiloff-Smith, A. (2009). Using developmental trajectories to understand genetic disorders. *Journal of Speech Language and Hearing Research*, 52, 336–58.