Chapter 11

Lessons from atypical development

All parents have asked themselves why some children grow up to excel at mathematics, others to excel at languages and yet others to excel at sports. Occasionally, there are clear events that demarcate why one child has developed a strength in a particular area, but even without such obvious markers, individual children differ enormously in what they grow into.

Our aim in this chapter is to introduce the notion of developmental trajectory (e.g., Waddington, 1953; Thelen and Smith, 1994; Karmiloff-Smith, 1998). This is the idea that to fully understand a biological system such as the growing child, we need to consider each developmental step that the child has taken before. That is, we need to view the child as positioned on a continuous developmental trajectory rather than as simply passing through discrete stages of performance. An individual child's current position on this continuum is the outcome of a common developmental process that operates under slightly differing constraints.

Children whose development follows a trajectory that is very different from the typical trajectory expected of the majority of other children offer a unique opportunity to examine how these constraints operate. Developmental disorders, then, strike at the heart of the issues we are considering in this book. What are the constraints that shape development?

In this chapter, we will examine how development occurs in children with developmental disorders (see Box 11.1 What are developmental disorders?). The aim of this exercise is to explore how constraints at the genetic, neural, physical and social levels of description operate to guide cognitive development. We begin by asking what role, if any, development actually has in understanding children who follow atypical developmental trajectories. We then ask how the interactivity of brain systems constrains development, and the extent to which the timing of developmental events plays a significant role. Next, we ask what role differences in input encoding and motor abilities have on cognitive development. Finally, we ask how the child's social context can constrain development.

Box 11.1: What are developmental disorders?

Developmental disorders can be classified into four groups:

1. Genetic disorders caused by well understood genetic abnormalities (e.g., Fragile X syndrome, Down syndrome, Williams syndrome, Turner's syndrome);
2. Disorders defined by one or more behavioural deficits (e.g., developmental dyslexia, specific language impairment, autism);
3. Mental retardation of unknown aetiology; and
4. Disorders resulting from environmental factors (e.g., an impoverished environment, fetal alcohol syndrome).

The first and last of these groups situate the principal cause of the disorder at either end of the nature vs. nurture divide. The middle two groups tell us about the level of our current understanding of such disorders. For example, disorders like specific language impairment and autism appear to have a genetic component but the genes involved have not yet been identified (Bishop *et al.*, 1995; Pennington and Smith, 1997; Simonoff *et al.*, 1998).

The study of developmental disorders proceeds with two aims in mind. The first of these is to identify appropriate methods of remediation and, for behaviourally defined disorders, early diagnosis to maximize the impact of remediation programmes. The second aim is to use disorders to help our understanding of the normal processes of development. A successful research programme could use developmental disorders to throw into relief the form and potential variability of these constraints. Within this framework, 'normal' development would simply constitute a special case of the settings of the constraints that guide all processes of development, successful or otherwise.

In relation to the second aim, little progress has been made in understanding the cognitive basis of general learning disability (mental retardation) where performance is lowered across all cognitive domains, let alone the neural bases underpinning such lowered performance. Disorders that show an uneven cognitive profile in their end state offer the greatest promise of theoretical insights. A number of disorders demonstrate dissociations in behaviour across different cognitive domains in adulthood. For example, Williams syndrome (WS) is characterized by a behavioural profile of relative proficiency in language and face processing (i.e., relative

Box 11.1 *(continued)*

to overall mental age), but severe deficits in other skills such as visuospatial processing, number, and problem-solving (Karmiloff-Smith, 1998). In hydrocephalus with associated myelomeningocele (a protrusion of the membranes of the brain or spinal cord through a defect in the skull or spinal column), language can be the only area of proficiency (Karmiloff-Smith, 1998). Individuals suffering from specific language impairment (SLI) show the opposite pattern, performing within the normal range in all domains except language. In autism, even individuals with normal IQs are selectively impaired in tasks that require judging another's mental states (Baron-Cohen *et al.*, 1993). In Fragile X syndrome (FraX), the adult cognitive profile is characterized by relative strengths in vocabulary, long-term memory and holistic information processing but relative weaknesses in visuospatial cognition, attention, short-term memory and sequential information processing (Cornish *et al.*, 1999, 2001; Freund and Reiss, 1991).

Some genetic disorders are caused by fairly circumscribed genetic mutations (see Box 11.2 for a more detailed discussion of the relation between genes and behaviour). For instance, WS is caused by a microdeletion of approximately 25 genes from one copy of chromosome 7 (Frangiskakis *et al.*, 1996; Donnai and Karmiloff-Smith, 2000; Tassabehji *et al.*, 1996, 1999). FraX is caused by the duplication of genetic material (the CGG repeat) in the Fragile X mental retardation (FMR1) gene on the X chromosome, which prevents the reading of the DNA message that this gene encodes (O'Donnell and Warren, 2002). The absence of the gene's product is the sole genetic cause of the disorder. The combination of circumscribed genetic causes and uneven cognitive profiles means that these disorders have the potential to illuminate links between genotype and phenotype. However, the correct explanatory framework for this endeavour remains a matter of some debate.

A role for development in developmental disorders?

In Chapter 3, we discussed the maturational perspective of functional brain development, in which newly emerging sensory, motor and cognitive functions in the developing child are related to the independent maturation of areas of the brain (usually cerebral cortex) responsible for each function. We argued that this perspective is limited because in actual fact the emergence

of new behavioural skills is associated with widespread changes across many regions of the cortex, and functional brain development appears to involve both increasing specialization and localization. Nevertheless, the maturational viewpoint has been a popular one within which to conceive of developmental deficits. According to this view, selective cognitive deficits are caused by isolated failures of particular functional modules. For example, Baron-Cohen *et al.* (1993) have argued that in individuals with autism, an apparent deficit in reasoning about mental states can be explained by the impairment of an innate, dedicated module for such reasoning—the 'Theory of Mind' module (see also Baron-Cohen, 1999). Another example comes from the work of Van der Lely (1997) who maintains that behavioural deficits in the language performance of children with so-called 'grammatical' Specific Language Impairment (SLI) can be explained by damage to a genetically determined, specialized module for processing syntactic (rule-based) information. Or again, Clahsen and Almazan (1998) have proposed that in the language of individuals with Williams syndrome, syntactic skills develop normally but there is a deficit in a component of the modular language system involved in accessing information about words that form exceptions to syntactic rules.

In effect, this conception of developmental deficits seeks to extend the explanatory framework of adult cognitive neuropsychology to the developmental realm. Patterns of deficits in adults with brain damage are interpreted in terms of intact and impaired functional modules. This framework and its methods are certainly powerful tools for exploring cognitive deficits at a given point in time (Jackson and Coltheart, 2001). However, because the framework deals in static snapshots instead of a continuous process of development, its power to evaluate the multiple putative origins of deficits is limited. Moreover, in the case of *developmental* deficits, use of the framework leads to the curious emergence of explanations that actually exclude the process of development (Karmiloff-Smith, 1997, 1998; Thomas and Karmiloff-Smith, 2002a; Thomas and Karmiloff-Smith, 2005).

Au: Karmiloff-Smith 1997 - not in refs.

Somewhat more problematically, this static view sometimes makes assumptions about development that appear unlikely. For example, if as argued in 3, interactive specialization is the appropriate view of functional brain development, then it appears implausible that one emergent, specialized system in the brain could develop atypically while all those surrounding it develop normally (the assumption of 'residual normality') (Thomas and Karmiloff-Smith, 2002a). Atypicalities in one part of the system are likely to have ramifications on the development of other parts of an interactive system.

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There is another problem with a straight maturational account of developmental disorders. According to this view, uneven cognitive profiles observed

in adult would predict that the same uneven cognitive profiles should be found at earlier stages of development. Paterson *et al.* (1999) sought to test this hypothesis by comparing the disorders of Williams syndrome (WS) and Down syndrome (DS) (see Box 11.1 for definitions of these disorders). In the adult phenotype, WS demonstrates greater ability in language than DS, while DS demonstrates better ability in numerical cognition. Paterson *et al.*, using standardized receptive vocabulary tests and numeracy judgement tasks, replicated this pattern. However, when Paterson *et al.* explored the respective performance of toddlers with WS and DS using preferential looking measures to tap each domain, they found a *different* relative profile. While both groups were very delayed, toddlers with WS and DS exhibited equal performance on a language task while toddlers with WS demonstrated superior performance to the DS group on a numeracy task. To the extent that the infant and adult tasks assessed the same aspects of the respective cognitive systems, this study contradicts the notion that atypical cognitive profiles in infancy are miniature versions of those shown in adulthood. At the very least, the story involves differential delays and/or non-linear developmental profiles in the two disorders, and therefore requires a focus on the sequence of development rather than just static snapshots of deficits.

The idea that uneven cognitive profiles in genetic disorders can be explained by isolated, atypically developing functional brain systems does not fit well with what is currently known about how genes control brain development (see Box 1.3, p. 00). Consider, the case of the British KE family. In this family, certain members demonstrated what was initially reported as a language-specific developmental deficit. The pattern of individuals exhibiting the language problems pointed to an inherited cause, and indeed the deficit was subsequently linked to the mutation of a single gene called FOXP2. However, detailed research on the family has gone on to reveal widespread structural and functional brain differences in affected family members, beyond those areas of the brain typically associated with language function in normal adults (e.g., Watkins *et al.*, 2002). Moreover, other behaviour deficits, albeit of a subtler nature, have been found outside the domain of language, for example in performing less sophisticated oral-facial movements, and in non-verbal tasks involving rapid associative learning (e.g., Watkins, Dronkers, and Vargha-Khadem, 2002).

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In line with the idea that developmental disorders do not involve region-specific structural atypicalities, post-mortem studies of genetic developmental disorders, and subsequently a growing body of work in structural brain imaging, have revealed widespread anomalies in gross and fine anatomy of the brains of these individuals. Gross anatomical differences have been found in

Table 11.1 Neocortical cytoarchitectonic and dendritic abnormalities in genetic disorders associated with mental retardation*

Disorder	Laminar disturbance	Increased packing density	Reduced dendritic length	Spine dysgenesis
Down syndrome	Y	N	Y	Y
Fragile-X syndrome	N	N	N	Y
Neurofibromatosis-1	Y (focal)	N	?	?
Patau syndrome	N	N	Y	Y
Tuberous sclerosis	Y (focal)	Y (focal)	Y (focal)	Y (focal)
Williams syndrome	Y	Y	?	?
Phenylketonuria	N	Y	Y	Y
Rett syndrome	N	Y	Y	Y
Rubinstein–Taybi syndrome	end N	Y	?	?

* The conditions have been listed according to estimated incidence following Moser (1995).
Adapted from Kaufmann and Moser (2000).

Au: Table 11.1 Moser 1995 —not in refs.

disorders such as WS (Bellugi *et al.*, 1999), DS (Nadel, 1999), and Fragile-X syndrome (Reiss *et al.*, 1995). These appear both in the relative and absolute size of large-scale structures. Fine scale cytoarchitectonic and dendritic abnormalities have also been found across a range of developmental disorders. Table 11.1 illustrates a range of such observed abnormalities (Kaufmann and Moser, 2000).

Given first the presence of widespread brain differences in many developmental disorders and second that, as we have argued in Chapter 3, current evidence encourages the view that functional modules in the adult are not prespecified in the infant but emerge as a product of development, one thing is clear: explaining uneven cognitive profiles in the adult phenotype of developmental disorders will be a complex endeavour.

What would a final account of a developmental deficit look like? It appears likely that it would need to begin by identifying differences in low-level neurocomputational properties, perhaps in numbers of neurons and their thresholds, local or global connectivity, and activity-dependent changes in these parameters (Karmiloff-Smith, 1998; Oliver *et al.*, 2000). The perturbations that these initial differences cause on the subsequent developmental trajectories of emerging cognitive systems must then be mapped out, taking into account atypical interactions, both internally between developing components and externally with the environment.

However, in terms of specificity of cause and outcome, our understanding of the relationship between neurocomputational parameters and cognitive performance is at present limited (see Box 11.2 Computational approaches to developmental disorders). For example, it might be possible that a computational property is anomalous throughout the brain but only impacts on those cognitive domains that particularly rely upon it during development. It remains a possibility that the cytoarchitectonic properties that specify regions of cortex are disrupted by diffuse gene expression gradients in such a way that *computational* anomalies are more topologically restricted than *structural* differences.[1] This possibility might support a narrower scope for the cognitive domains impacted during development despite more widespread structural differences. Such issues remain to be worked out.

Box 11.2 Computational approaches to developmental disorders

Connectionist neural network models of cognitive development form an ideal framework within which to explore the view that developmental deficits are the outcome of atypical neurocomputational constraints. Such models throw a particular spotlight on the role played by initial computational constraints in influencing the nature and success of subsequent trajectories of learning and development (Karmiloff-Smith and Thomas, 2003b). The ability of a model to acquire information from a given domain is limited by its initial architecture, activation dynamics, learning algorithm, and the representations through which the domain is depicted. In connectionist models of *typical* development, such design decisions are justified as far as possible via empirical evidence. A model is then judged successful if it captures the end state competencies of the system as well as the developmental trajectory through which it passes. The opportunity here is to demonstrate that theoretically motivated alterations to the initial computational constraints of a normal model can then capture both the atypical trajectory and end state behavioural deficits found in a particular developmental disorder. Where the success of a developmental model depends upon *changes* in the computational constraints across development, as in constructivist systems, then manipulations to the way in which such changes occur can also be explored as a candidate cause of developmental deficits (Thomas and Karmiloff-Smith, 2002b; Westermann and Mareschal, 2003).

The transition of a model from normal functioning to the disordered state is often the result of modifying quantitative variables, such as learning

Box 11.2 *(continued)*

rate, levels of computational resources (processing units), or amount of noise. As such, connectionist models of developmental disorders lend themselves to an inherently continuous conception of pathology, with no absolute distinction between normality and disorder. However, alterations to models can also be more radical, for instance using a different network architecture or learning algorithm. These changes might be viewed as positing a qualitative distinction between normality and disorder. Referring back to our discussion on cognitive variability, the difference between these accounts will lie in the details of the developmental history of the processes that produced a computational system with these anomalies. Some parameters may alter quantitatively within the normal course of brain development, whilst others may require a genetic mutation to be altered.

Although this line of research is relatively new, connectionist models have already been used to explore the possible computational causes of deficits in several developmental disorders. Such investigations are contingent on the existence of valid models of typical development before parameter variations in the start state (or rates of parameter change during development) can be explored. In consequence, work on atypical cognitive modelling tends to lag behind that on typical development. Developmental dyslexia, autism, SLI, and WS have all been the subject of recent simulation work. Developmental dyslexia has been investigated by a number of researchers by manipulating the start state parameters of models of reading development (e.g., Harm and Seidenberg, 1999, and see also Chapter 9 by Joanisse in Volume 2). Autism has been investigated by manipulating the startstate of models of category formation (Cohen, 1994, 1998; see also Chapter 10 by Cohen in Volume 2). SLI and WS have been simulated by altering the start state of models of inflectional morphology (Hoeffner and McClelland, 1993; Joanisse, 2000 and Chapter 9 in Volume 2; Thomas and Karmiloff-Smith, 2003a).

Thomas and Karmiloff-Smith (2002a) used connectionist models to examine more general theoretical issues concerning the relation of developmental deficits to those found in cases of adult brain damage. This is another area to which connectionist models of cognition have been widely applied. Thomas and Karmiloff-Smith sought to assess whether disruptions occurring to the start state of a learning system tended to produce the same performance deficits as applying those same disruptions to the end state of a normally trained model. The results of the modelling indicated that start state damage to a system and endstate damage could in some circumstances cause similar behavioural impairments, but at other times the patterns

Box 11.2 *(continued)*

were very different. The relationship depended on whether the system was able to use the developmental process to compensate for damage applied in the start state, by attenuating or even overcoming the effects of early anomalies. In other cases, early deficits followed by development produced worse deficits than damage to the end state. Importantly, the simulations served to uncover the precise computational conditions under which each type of effect emerged. Moreover, the results convincingly demonstrated that in developmentally disordered systems, dissociations between impaired behaviour and range-in-the-normal-range cannot be unambiguously interpreted without an understanding of the developmental conditions that pertained in the underlying system. There is no inference from developmental deficit to underlying functional structure without stipulating an account of the developmental process.

Thomas (2003b) recently pursued this issue further in a computational consideration of the multiple causality of behavioural deficits. Simulations indicated that narrowly defined behavioural deficits can potentially have multiple underlying computational causes. The implication is that developmental disorders defined on behavioural grounds alone (such as SLI or dyslexia) may gather together individuals with differing underlying cognitive architectures. This would seem to limit the ability of behavioural experimentation using group studies to uncover any single 'cause' of the impairment defining the disorder. However, simulation work suggested that there may be behavioural markers that can be used to identify underlying causal heterogeneity, in the cross-measure variability within a disorder group. That is, the variability in performance across behavioural measures can indicate the extent to which the atypical behaviour of a disorder group has a single or multiple underlying cognitive causes.

In short, computational models can help to explore the contribution of the developmental process to developmental deficits. They can assess the viability of claims concerning the possible origins of developmental deficits, and so begin to trace back these deficits to their genesis in early brain development. In the field of developmental disorders, they serve to underline the crucial importance of formulating a precisely defined developmental account of a given cognitive ability before seeking to interpret behavioural deficits within a developmental disorder.

If one compares developmental disorders with cases of early acquired brain damage in healthy children, it becomes apparent that the appropriate way to conceive of the disorders is in terms of the *constraints that shape development* rather than in the loss or impairment of specific cognitive structures (Karmiloff-Smith and Thomas, 2003a; Pennington, 1999).

The most informative comparison here is not a direct one, but a triangular comparison that includes cases of adult-acquired brain damage. The exercise works as follows. For behavioural deficits of adults with a given developmental disorder (such as SLI), identify which area(s) of the brain of a healthy adult would have to undergo focal damage for the individual to show this deficit (e.g., for language, left temporal or frontal damage). Then examine the consequences of early focal brain damage in otherwise healthy children, occurring to the same area(s) of the brain. What is the behavioural deficit exhibited by these individuals once they have reached adulthood? Does it match up with the deficits shown in the adult with the developmental disorder? In almost every case, the answer is no. The otherwise healthy children with early brain damage can show recovery and no lasting behavioural deficits. This begs the question of why healthy children should show recovery after early focal brain damage while individuals with developmental disorders who sometimes show apparently specific behavioural deficits do not? The answer is that the two cases constitute different *limits on plasticity*, i.e., differences in the ways that the healthy and atypical brain can be modified by experience.

Thomas (2003a) argued that a comparison of developmental disorders and children with acquired brain damage actually suggests that the closest comparisons lie between individuals with developmental disorders and healthy children who have experienced *widespread* early brain damage. In the latter group of children, recovery is limited and development increasingly diverges from the normal pathway with age (Anderson *et al.*, 2001). This comparison fits more closely with the widespread structural anomalies found in the brains of individuals with genetic developmental disorders.

A first step in the consideration of atypical limits on plasticity is to consider how this may affect the emergence of specialized functional structures in the brain.

The role of embrainment in development

As described in Chapter 3, the interactive specialization account of functional brain development argues that processing becomes both more localized and more specialized with development. The evidence that we presented for this drew heavily on examples of typical development. However, several

developmental disorders suggest that this developmental process may be deflected by atypical constraints.

Take the example of face processing. Event-related potential (ERP) studies of face processing have indicated that upright and inverted faces elicit voltage wave form components that differ both in amplitude and location on the scalp. When adults who have followed typical developmental trajectories are presented with two matching faces vs. two non-matching faces, the ERP differences for upright faces in normal adults show a negativity around 320 ms that is largest over anterior regions of the right hemisphere. For inverted faces, however, the main difference between matched and mismatched stimuli is a symmetrical positive wave form component over parietal regions occurring between 400 and 1000 ms (Bellugi *et al.*, 1999). When the equivalent waveforms for adults with WS were examined, three differences emerged (Mills *et al.*, 2000): (1) the WS group exhibited the mismatch effect at 320 ms for *both* upright and inverted faces; (2) the 320 ms waveform component did not show the right-hemisphere asymmetry of normal adults but was bilateral; and (3) there was an abnormally large negative wave component at 200 ms both to upright and inverted faces. Bellugi *et al.* (1999) argued that this later effect is linked to increased attention to faces in adults with WS and appears specific to the disorder (see Grice *et al.*, 2001, for similar results and a comparison to face recognition in autism). In short, in WS, ERP activity patterns in adulthood suggested that the neural processing of faces is both *less localized* (bilateral instead of right lateralized) and *less specialized* (elicited by both upright and inverted faces instead of just upright faces, as well as by monkey faces and by other objects) than in adults who have followed a typical developmental trajectory.

Above, we suggested that developmental disorders represent atypical limits on plasticity, such that development cannot compensate for early functional brain damage in the way it appears able to do in typically developing individuals. However, this does not imply that no compensation occurs in developmental disorders. Indeed, each functional brain system develops in the context of other brain systems. If an anomaly emerges across development in one system, it may well have ramifications for other systems, perhaps ones that are recruited (atypically and potentially less efficiently) to drive the behaviour of importance to the individual. For example, fMRI studies have demonstrated that adults with (phonological) developmental dyslexia demonstrate less activity in left posterior temporal-parietal areas compared to controls during listening and reading tasks that are phonologically demanding (Brunswick *et al.*, 1999; Flowers *et al.*, 1999; Paulesu *et al.*, 1996; Rumsey *et al.*, 1992; Shaywitz *et al.*, 1998). However, several of these

studies also reported increased activity in occipital and/or frontal regions in dyslexia that may reflect efforts to compensate for developmental impairments in phonological abilities with the use of additional visual strategies (Casey *et al.*, 2001).

The role of timing in development

The timing and extent of basic neural developmental processes such as synaptogenesis, arborisation, and myelination have been found to show atypicalities in developmental disorders. These processes are inextricably linked to activity-dependent processes, that is, to the way that the brain alters itself in response to experience. For example, recent evidence from the PET imaging of neurotransmitter systems indicates that alterations in the plasticity of brain areas (as indexed by the numbers of particular types of synapses) may not follow the normal course in some developmental disorders (Huttenlocher, 2002). Thus, Chugani *et al.* (1999) found a difference when comparing children with autism and healthy controls. In the controls, serotonin synthesis capacity (which depends in part on the number of serotonergic synapses) in five-year-old children was twice the adult value, subsequently decreasing back to the adult value following synaptic pruning. This is consistent with greater brain plasticity in childhood. Children with autism, by contrast, had a lower serotonin synthesis capacity than controls at age 5, but the level steadily increased to 1.5 times the normal level by age 15, implying both delayed early synaptogenesis and then decreased synaptic pruning. Huttenlocher (2002) noted that this abnormal pattern has been found in the primary visual cortex of animals deprived of normally formed visual images during the system's early sensitive period, implicating activity-dependent processes in this abnormal marker of neuroplasticity.

In contrast to the preceding 'less-followed-by-more' pattern of development, Becker *et al.* (1986) found that dendritic arborizations in the visual cortex of children with Down syndrome (DS) were paradoxically greater than normal early in infancy but then considerably less than normal by the age of two years. Becker *et al.* speculated that the initial overabundance might be a consequence of a compensatory response to the absence of adequate synapse formation. In many cases, DS is also characterized by a postnatal delay in myelination (Wisniewski, 1990). The delay is initially global but then manifests primarily in those nerve tracts that are myelinated late in development, such as the fibres linking the frontal and temporal lobes (Nadel, 1999). This again suggests that the timing of neural events is essential in determining the unique developmental profile of children (in this case, children with DS).

The role for input encoding in development

Several developmental disorders have been characterized in terms of differences in the way that information arriving from the environment is encoded during initial processing. Atypical representations of the environment can facilitate or impair subsequent higher-level tasks. For example, in autism, individuals exhibit *superior performance* on visual search tasks compared to mental-age (MA) matched controls, where a participant must pick out a green T in a field of red Ts (O'Riordan, 2000; O'Riordan *et al.* 2001). O'Riordan and colleagues argued that the superior performance arises not through attentional biases in higher processes but because individuals with autism begin by encoding greater discriminability between the components parts of the visual scene, thus facilitating selection in an 'odd one out' task.

In SLI, it has been argued that information about word sounds is represented in such a way that higher cognitive processes like inflectional morphology and syntax cannot operate as efficiently on word forms, particularly under time pressure (see Leonard, 1998, and Chapter 9 Volume 2 for further discussions of this idea). Similarly, in developmental dyslexia, it has been argued that word sounds are represented in such a way that it becomes much harder to learn the association between the component sounds of words and their written forms.

In general, alterations in the level of abstraction achieved in forming internal representations, or in the dimensions of similarity that those representations encode, can play a material role in the ability of other brain systems to employ these representations to drive other processes. In the proposals on autism, SLI, and dyslexia, the consequence of atypical similarity structure in input representations may result in an apparent processing deficit higher up in a hierarchy of representational systems.

Our next step is to consider the role of the way in which the atypical individual co-specifies an atypical environment, either physically or socially. This provides an example of embrainment in that differential input to one region from another may shape its trajectory of functional specialization.

The role for embodiment in development

In Chapter 4, we argued that embodiment plays a critical role in typical development. In many developmental disorders, the physical body is normal and the individual can perform common physical activities (although sometimes fine motor performance is reduced, or gait and posture may be unusual). In some disorders, however, movement can be more seriously restricted. This provides

a potential window on the influence of the body on cognitive development. However, to date, there are few robust findings in this area. Although its findings are somewhat controversial and the conclusions speculative, one study serves to illustrate the directions such research may take, and the way in which atypical embodiment might impact on development.

Children with spinal muscular atrophy (SMA) show physical weakness. Their first six-month's progression is normal, such that these children can sit unaided. However, they never achieve the ability to stand and walk. Sieratzki and Woll (1998) examined the language development of a group of children with SMA. These children exhibited normal vocabulary and use of irregular inflectional forms (such as 'thought' and 'mice'). However, over-regularization ('thinked', 'mouses'), a marker for the acquisition of linguistic rules, was *accelerated*. Sieratzki and Woll speculated that the inability of these children to explore objects and forms in the environment might have advanced their analysis of patterning in language and the extraction of regularities. They speculated that at a neural level, the weakly used prefrontal motor areas of the brains of children with SMA were being exploited by grammatical processing to accelerate developmental processes.

Of course, as discussed in Chapter 4, an individual that has different physical abilities also has a different effective environment. In children with SMA, this was reflected in the following way. While their knowledge of many vocabulary items appeared to be developing normally, children with SMA nevertheless exhibited difficulties with certain vocabulary items, such as 'action' words, 'outside things', and 'places to go'.

Although the data here are as yet somewhat provisional, the hypotheses considered by Sieratzki and Woll illustrate the wider point that the constraints operating during development can have a profound effect on the subsequent trajectory followed by cognitive development.

The role of social context

The atypically developing child also has an atypical environment. This interactive effect may be straightforward: a child with dyslexia may spend less time reading because it is a struggle to read, resulting in reduced input to the relevant cognitive systems. However, the interactions may be subtler, operating on the effective social environment to which the individual is exposed. Two studies exploring language development in DS and WS illustrate this point.

The parents of children with developmental disorders that involve learning disabilities are understandably concerned about the developmental progress of

their offspring. Such anxiety may lead to changes in the effective social environment that the child experiences. For example, Cardoso-Martins, Mervis, and Mervis (1985) found differences between the parental language inputs of children with DS compared to that of typically developing controls in terms of the language parents used to label objects for the child. While 67 per cent of mothers of typically developing children used basic-level category terms to label objects in naming, only 31 per cent of mothers of children with DS used the basic-level category. Mothers of children with DS more frequently used precise object names (e.g., lion) than generic basic level terms (e.g., cat) during object labelling. This was possibly due to the parents' increased concern that their children might not come to learn the correct names for objects spontaneously. There is no evidence either way on whether this strategy was beneficial, but it serves to show that atypical development cannot be considered solely from the perspective of the atypical brain but must extend to consider interactions with an atypical environment.

The effective social environment may also be altered in more indirect ways by developmental deficits. For example, Laing and colleagues examined socio-interactive precursors to language development in toddlers with WS compared with mental age (MA) matched controls (Laing *et al.* 2002). Toddlers with WS were proficient at dyadic (two-way) interactions with a caregiver and indeed sometimes exceeded the scores of MA controls due to persistent fixation on the caregiver's face (see also Bertrand *et al.*, 1993; Jones *et al.*, 2000). However, there was a marked deficiency in *triadic* interactions that incorporated an object. Specifically, toddlers with WS had difficulty switching attention from the caregiver's face to an object that was being referred to in communication via pointing, looking and naming. Shared attention to newly named objects appears to be one of the main routes into vocabulary acquisition in normal development. The atypical nature of the social interaction found in children with WS may therefore have further ramifications for subsequent language development in this disorder. In fact, language development *is* delayed in this disorder (Karmiloff-Smith and Thomas, 2003a; Thomas and Karmiloff-Smith, 2003a).

Challenges from the study of atypical development

We have argued that one must view developmental disorders in terms of the operation atypical constraints deflecting the normal path of development. Thus, developmental disorders can be viewed as unfolding through the same general processes involved in typical development. However, this view also

throws up a number of immediate questions. We address two of these in turn below.

Question 1: How do variations in 'neurocomputational' constraints map to variations in behaviour?

An immediate issue is to work through the cognitive level implications of claims about atypical neurocomputational properties. For example, take the following speculation on how disorders that appear quite different at the behavioural level may in fact be related at the neurocomputational level: 'Subtle related initial deficits (e.g., firing thresholds which are either too high or too low) can give rise to huge differences in the end state which seem to bear no relation to one another' (Johnson *et al.*, 2000: 38). Here again, computational modeling can play an important role (see Box 11.2).

One immediate solution to this problem is to demonstrate in an implemented computational system that contrasting deficits in the endstate performance of a developing system can be produced by changes in a single initial computational parameter (see, e.g., Thomas and Karmiloff-Smith, 2003a, for a demonstration in the cognitive domain of language acquisition). But what allows us to call the initial parameter difference 'subtle' given that its ultimate impact on behaviour is so significant? It cannot simply be that the *magnitude* of the change is itself small, or that we have changed only *one* parameter, because we have no absolute scale of reference against which we can say that a change is 'small' or 'large'.

Again, we can think of two possible answers to this question. Both are of theoretical importance, but both ultimately require empirical support. First, 'subtle' can mean that there is a non-linear[2] relationship between changes in the start state parameter and ultimate developmental outcomes. Thus, perhaps initial neurocomputational parameter changes across wide ranges produce little variability in the end state behaviour, while much smaller changes in a sensitive range can produce great variability in the end state behaviour. An emphasis on non-linearity is an important aspect of the neuroconstructivist approach. It is central in helping us understand how differences in the genotype might be related to difference in the phenotype. Second, a 'subtle' effect can mean that the developmental process itself exaggerates the impact of the parameter. In this case, if contrasting developmental profiles were to be found in the end state of two disorders, one would expect much smaller behavioural differences in infancy. This points towards a particular empirical paradigm for comparing disorders across developmental trajectories, work that has begun to produce interesting results (e.g., Paterson *et al.*, 1999).

Question 2: How do atypical brain structures relate to atypical cognitive structures?

The final challenge for the neurconstructivist approach is to understand which differences in the apparent structure or function of the brain in a developmental disorder (as revealed, for instance, by brain imaging studies) actually have information-processing consequences for the development of cognition. The difficulty here is that while atypical functioning at the cognitive level seems to correlate with atypical activation patterns in the brain, atypical activation patterns in the brain do not guarantee atypical cognitive functioning. For example, 2–5 per cent of 'normal' individuals appear to have right-lateralized language systems (Bates and Roe, 2001). Yet these individuals are not marked out as having atypical cognitive-level language systems. Women can demonstrate more bilateral patterns of brain activation in language tasks than men (e.g., Shaywitz *et al.*, 1995). Indeed, sex steroid hormones have been shown to modulate a wide range of brain processes including neurogenesis, cell migration, growth of the neuronal soma, dendritic growth, differentiation and synapse formation, synapse elimination, neuronal atrophy and apoptosis, neuropeptide expression, the expression of neurotransmitter receptors and neuronal excitability (Cameron, 2001). Yet cognitive psychology does not (at present) posit qualitatively different functional structures for the language system in the two genders, let alone different overall cognitive architectures. Such differences in brain function are put down to the *multiple realizability* of cognitive architectures in neural structures, whereby the same cognitive level computations can be implemented in different ways in the wetware available. The negotiation between these two ideas—brain constraints that alter cognitive architecture versus multiple realisability of cognitive architectures—remains to be worked through.

Summary and discussion

In this chapter, we have asked what could be learned about the processes of development from studying children with atypical development. We have argued that developmental disorders are best conceived of as the outcome of atypical constraints operating on the normal developmental process. In this framework, one can view typical and atypical development as different trajectories in a continuum of developmental possibilities. Moreover, as development is a continuously ongoing process, one needs to consider the ontological history of the individual to understand what underlies the individual's current abilities and how those abilities came into being.

With this in mind, we reviewed possible sources of constraints on representation development that might be highlighted in developmental disorders. The first conclusion was that because brain systems were highly interactive, disorders are unlikely to be characterizable as a set of normally functioning vs. impaired cognitive components from the normal repertoire. We also found that differences in the timing of key neural developmental events could lead to atypical cognitive consequences. Changes in motor abilities could lead to changes in cognitive abilities such as generally improved language performance but decreased action-based vocabulary. Finally, an atypically developing child also co-specifies an atypical social context, both in terms of the way others interact with the child and the kind of experiences that he or she seeks out. All of these different constraints lead the organism (the developing child) to take a different trajectory in the space of developmental possibilities.

In this chapter, we have already drawn on many of the principles found to operate at the different levels of description reviewed in the four previous chapters. That is, principles that were found to operate at the cellular level, the functional brain systems and the whole body levels of description. Thus, perhaps one of the most important lessons to take from this chapter is that when considering the complex behaviours that are crucial to our cognitive level description, it becomes difficult if not impossible to disentangle the influences of the different levels of processing. In the next chapter we revisit these questions by focusing on the particular example of children with dyslexia.

Notes

1. For example, Karmiloff-Smith (1998) discusses the case of a *body-wide* genetic cellular difference, which nevertheless impacts only on hearing and results in the specific outcome of hereditary acquired deafness.
2. A linear process is one in which the output is some weighted combination of the input (e.g., $y = a.x + b$). In such systems, a change in the input (δx) leads to an *identical* change in output ($\delta y = a.\delta x$) no matter where it occurs in the range of input values (values of x). Processes for which this is not true are called non-linear processes.