Language cannot be reduced to biology: Perspectives from neuro-developmental disorders affecting language learning

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The study of language knowledge guided by a purely biological perspective prioritizes the study of syntax. The essential process of syntax is recursion – the ability to generate an infinite array of expressions from a limited set of elements. Researchers working within the biological perspective argue that this ability is possible only because of an innately specified genetic makeup that is specific to human beings. Such a view of language knowledge may be fully justified in discussions on biolinguistics, and in evolutionary biology. However, it is grossly inadequate in understanding language-learning problems, particularly those experienced by children with neurodevelopmental disorders such as developmental dyslexia, Williams syndrome, specific language impairment and autism spectrum disorders. Specifically, syntax-centered definitions of language knowledge completely ignore certain crucial aspects of language learning and use, namely, that language is embedded in a social context; that the role of envrironmental triggering as a learning mechanism is grossly underestimated; that a considerable extent of visuo-spatial information accompanies speech in day-to-day communication; that the developmental process itself lies at the heart of knowledge acquisition; and that there is a tremendous variation in the orthographic systems associated with different languages. All these (socio-cultural) factors can influence the rate and quality of spoken and written language acquisition resulting in much variation in phenotypes associated with disorders known to have a genetic component. Delineation of such phenotypic variability requires inputs from varied disciplines such as neurobiology, neuropsychology, linguistics and communication disorders. In this paper, I discuss published research that questions cognitive modularity and emphasises the role of the environment for understanding linguistic capabilities of children with neuro-developmental disorders. The discussion pertains to two specific disorders, developmental dyslexia and Williams syndrome.

[Vasanta D 2005 Language cannot be reduced to biology: Perspectives from neuro-developmental disorders affecting language learning; *J. Biosci.* **30** 129–137]

1. Overview

The main argument of this paper is that an approach based on genetic determinism leads to problems when it comes to understanding the complexity associated with certain neuro-developmental disorders such as developmental dyslexia and Williams syndrome. The problems are not just philosophical, and pertain to one's concept of what language is (see Rice 1996 for a fuller discussion on the definitional issue). In addition, there are attendant social and ethical issues with regard to assessment and intervention of children with these disorders. This is particularly true in India, where the resources for conducting certain kinds of genetic research and using pharmacological interventions are limited. This paper is organized under three sections: The first part provides some definitions and brief descriptions of key terms used in the paper; the second part summarizes published evidence that provides grounds to challenge a strictly biological perspective on language; the third part deals with the problems of char-

Keywords. Cognitive modularity; developmental dyslexia; orthography; specific language impairment; Williams syndrome

Abbreviations used: FG, Fusiform gyrus; LH, left hemisphere; QTL, quantitative trait loci; SLI, specific language impairment; STG, superior temporal gyrus; SVAS, supra valvar aortic stenosis; VWFA, visual word form area.

acterizing genotype-phenotype relations in neuro-developmental disorders. In the final part of the paper, I discuss static versus dynamic characterizations of these disorders with particular reference to dyslexia and Williams syndrome. I conclude that adequate characterization and management of neuro-developmental disorders of language learning calls for dynamic descriptions and a 'whole-brain approach' to understanding brain-behaviour relationships (see Lem 1992 for a detailed discussion on this point). This position is in no way intended to undermine research programmes designed to investigate the origin of the syntactic component of language, a component that is widely believed to differentiate human communication from animal communication. Instead, the arguments put forth in this paper must be seen as part of the resistance with which the modular conceptions of language are increasingly being viewed in recent years. It is instructive to cite Piatelli-Palmarini (2001: 19):

> "From SLI (Specific Language Impairment) to prosopagnosia, from domain-specific semantic deficits to Williams syndrome, the standard (and in my opinion still correct) modular interpretations are being challenged by insiders and outsiders as well. "Much more goes berserk in those cases" is the recurring critical punch line. Cognitive deficits produced by restricted brain lesions are alleged to be, after all, not so specific and circumscribed. The case is pending, and modularists now face a fight."

I offer the arguments in this paper as an outsider interested in furthering the debate on the genetic basis for language development and language disorders.

2. Key terms used in the paper

2.1 *Developmental dyslexia*

This term refers to a condition in which a child experiences significant reading difficulties in the absence of any demonstrable brain pathology and stands in contrast with acquired dyslexia in adults following brain injury. Unlike mental retardation in which all domains are uniformly affected, in developmental dyslexia, the behavioural deficit is limited for the most part to reading and spelling. A detailed discussion on brain basis for this condition in addition to the genetic aspects are discussed in latter parts of this paper. Readers may also consult Plomin and DeFries (1998) and Padakannaya (2003) for more information.

2.2 *Williams syndrome*

Williams syndrome is a rare neurodevelopmental disorder characterized by a microdeletion on one copy of chromosome 7 and results in specific physical, cognitive and behavioural abnormalities (Bellugi *et al* 1999a). Individuals with Williams syndrome exhibit uneven cognitive-linguistic profiles together with mild to moderate mental retardation. Specifically, they have considerable difficulty in numerical cognition, problem solving and planning, but are largely successful in language acquisition.

2.3 *Specific language impairment*

Specific language impairment (SLI) is a heterogeneous disorder of language acquisition in children who do not have any other apparent cognitive, social or neurological deficits that can account for their linguistic problems. There is considerable controversy over whether an input processing deficit or a grammar specific deficit causes SLI. One reason for this controversy is the variation in linguistic and cognitive characteristics found across different groups of children diagnoses to have SLI (see Van der Lely and Ullman 2001).

2.4 *Modular conceptions of language*

In his influential publication, *The Modularity of Mind*, Fodor (1983) made a distinction between input systems which he argued are modular as opposed to 'central' systems which are not modular. The latter according to him deal with the fixation of beliefs about the world. He offered the following list of properties to define what a module is: domain specificity; mandatoriness and speech of operations, informational encapsulation, autonomy of computation, lack of access by other systems to intermediate levels of representations, shallow output, neural localization and susceptibility to characteristic breakdown. Fodor cited language comprehension and visual perception as two prime examples of modules. They can influence language acquisition as well as disorders (see Levy 1996; Levy and Kave 1999).

More recently, Sperber (2001) elaborated the concept of modularity not just of mind but of any biological system as operating at five different levels:

(i) At the developmental level, modules are approached as phenotypic expressions of genes in an environment. Cognitive modules are hypothesized to explain why and how children develop competencies in specific domains in ways that could not be predicted on the basis of environmental inputs and general learning mechanisms alone.
(ii) At the morphological or architectural level, what is investigated is the structure and function of specific modules, and more generally, the extent to which, and the manner in which the organism and its subparts in particular the mind/brain are in articulation of autonomous mechanisms.

(iii) At the neurological level, modules are typically seen as dedicated brain devices that subserve domain-specific cognitive functions and that can be selectively activated or impaired.
(iv) At the genetic level, what is at stake are the pleiotropic effects among genes such that relatively autonomous 'gene nets' get expressed as distinct phenotypic modules.
(v) At the evolutionary level, hypotheses are being developed about the causes of the evolution of specific modules, and of genetic modularity in general.

2.5 *Socio-cultural perspective on language*

This perspective locates the source of learning in the pursuit of action in our social worlds. The focus is not on linguistic units and representations themselves, but on the processes whereby these forms interrelate with each other and with the circumstances of their use. In other words, the fundamental concern is the study of the use of language in real world circumstances (Hall 2002). Thus learning language does not depend exclusively on the activation of internal innately specified language acquisition device that seeks to assimilate new forms of knowledge. Instead, it involves process of being socialized into the communicative and other social activities of sociocultural importance to the group(s) or communities one aspires to be a member of. The inherited biological characteristics of language are only the necessary preconditions for the ability to learn language. They have to merge with and are shaped by sociocultural processes in the course of our interaction with the others [see Widdowson 2000 for a critique of Chomskyan conception of Internalized (I) language].

3. Limitations of a biological perspective on language

The biological approach, discussed by Shukla (2005), rests on the premises that language is a rule based symbolic computational device; that language knowledge entails ability to create limitless representations by the creative combination of a finite vocabulary; and that the logical problem of language acquisition is to find out how a child who is exposed to a limited set of sentences is able to create infinitely many possible sentences. Linguistics working within this paradigm (generative framework), posited that infants have an innate predisposition to learn language(s) in that certain aspects of language (the Universal Grammar) are encoded in their genes. These ideas, systematized by Chomsky in his influential theory of principles and parameters, basically argue for a core syntactic module, the operations of which are constrained by both the semantic system and an output (phonetic) system (see Hauser *et al* 2002). In terms of methodology, generative linguists have studied sentence structure independently of situational meaning, whereas psycholinguists paid attention to the processing of linguistic units rather than the units themselves.

Within the largely computational perspective adopted by generative linguists, language is viewed as a purely cognitive, non-communicative event involving the speaker's beliefs and intensions. The goal is to explain the relationship between sentences and something in the world about which the sentences state something. Thus, this perspective emphasizes one and only one relationship between language and reality. In the context of a discussion on 'reconstruction and interpretation in the social sciences', Habermas (trans. Lenhardt and Nicholsen 1990) argued that in another (communicative) view of language use (which comes under the purview of hermeneutics), at least three different relations are involved: one, giving expression to one's beliefs, intentions, etc.; two, establishment of an interpersonal relationship between speaker and hearer, and three, expression about something in the world. Thus, according to Habermas, language use serves three functions, that of reproducing culture and keeping traditions alive; that of social integration or the coordination of the plans of different actors in social interaction; and that of socialization or the cultural interpretation of needs. Such a multi-functional view of language has also been upheld by psycholinguists and researchers investigating sign languages (see Johnston 1992 for elaboration of this point in relation to American Sign Language).

If we accept the multi-functional, communicative aspect of language use, the questions of innateness and modularity of cognitive operations take on a different meaning. Specifically, a purely biological (or biolinguistic) approach to language that rests on modular conceptions (see Elman *et al* 1996) cannot support recent observations on infants, children and adults who are bilingual or multilingual; literate, or illiterate. Some examples of such research are cited below. My hope is that readers interested in gathering evidence for a socio-cultural aspects of language will be able to begin their search with these examples since explicit theories accounting for this (social interactive) perspective are still in the making.

(i) Lesion studies of infants and children have questioned the notion that the left hemisphere comes into this world with hard-wired linguistic knowledge (Bates *et al* 1999). In a recent study Bates (2002) concludes that the path from genes to language is indirect, and many different genes can affect language even though none of them is specific to language.
(ii) Studies of monolingual and bilingual babies under the age of six months demonstrated that bilingual infants

do not treat language input the way monolinguals do in that they build their language systems differently at different rates (Sebastian-Galles and Bosch 2001).
(iii) Age of language acquisition may be a significant factor in determining the functional organization of cortical areas known to have influence over language processing. Specifically, it has been shown that second languages acquired in adulthood are spatially separated from native languages as evident in fMRI studies (Kim *et al* 1997).
(iv) Cultural learning, including learning to read and write in early childhood, has been shown to modify brain structure as well as function. Related to this, in brain imaging studies literates do better than ex-literates or illiterates on a whole lot of metaphonological tasks such as phoneme monitoring under difficult listening conditions (Castro-Caldas *et al* 1998; Frith 1998).
(v) Recent proposals about autonomous modules for spoken and written language have also been challenged in studies involving literate and illiterate participants (Morais and Kolinsky 2001 for details).
(vi) Both hearing and hearing impaired children make use of extensive coverbal gestures (gestures that accompany speech) during communication. Three-dimensional motion analysis studies directed at understanding neural control of language and movement (e.g. Poizner and Kegel 1992; Hadar and Yadlin-Gedassy 1994; Hadar *et al* 1998) have shown that gestures facilitate cognitive processes that underlie speech by allowing motor cognition to process some of the material required for message construction. Thus it has been shown that action verbs are particularly likely to be accompanied by gestures suggesting that motor-semantic priming has a role to play in message construction and delivery. The fact that most people prefer a face-to-face conversation to a telephone conversation when important matters have to be discussed suggests that co-verbal gestures play an important role in communication. This evidence may not be sufficient to challenge existing modular conceptions of language, but they certainly beg radical rethinking of the relationship between brain and language in which semantics takes precedence over syntax (Vasanta 2000).
(vii) A strictly biological perspective on language is incapable of accounting for sudden loss of speech in so-called hysterical aphonia cases. It cannot explain why we experience writer's cramp or slips of tongue, or why deaf children denied instruction in sign language invent their own sign language (home sign) which eventually (from one generation to another) becomes a regular rule-governed system, whereas hearing siblings of deaf children fail to acquire and use stable lexicon of gestures (see Singleton *et al* 1993).
(viii) Connotation, appreciation of verbal humor, metaphor and prosody are all part of language mediated by the right hemisphere. These features along with the tone of voice and facial expressions constitute the context of communication. Interpretation of contextual clues is intimately linked to visual perception. How is one justified then to exclude these features from the definition of language? [See Laver (1999) for a discussion on 'an integrated theory of non-verbal communication' which takes into account a whole lot of paralinguistic behaviours into consideration].

I would like to argue that an adequate characterization, assessment and treatment of neuro-developmental disorders affecting language learning call for a broad definition of language that is not simply syntax centered – a definition that underscores the communicative aspects of language use. In the next section, I will discuss the problems in relating genotype-phenotype relations in complex disorders of language learning and then present the static versus dynamic characterization of two disorders, viz developmental dyslexia and Williams syndrome.

4. Genotype-phenotype relations in neuro-developmental disorders

The construct 'behavioural phenotype' implies that for every identifiable genetic syndrome, a specific behavioural pattern linked to that genetic abnormality must exist. In recent years, a less restrictive definition has been offered by Dykens (1995). A behavioural phenotype according to him is best described as the heightened probability or likelihood that people with a given syndrome will exhibit certain behavioural and developmental sequelae relative to those without the syndrome. There is also a recommendation (Turk and Hill 1995 cited in McMahon 1999) that the observed traits be subjected to assessment using measurable criteria in at least five different domains: the domains of intellectual functioning, speech and language, attention deficits, social impairment and other behavioural disturbances such as for instance self injury. It must be noted that there are very few tests for assessing the domain of social impairment compared to tests which assess structural aspects of language.

The neuropsychological tests typically used to assess linguistic function tap on knowledge of linguistic representations (grammatical or morphological rules) along with the working memory needed for implementing those representations in either production or comprehension. Therefore, poor performance on such tests cannot distinguish between deficits in linguistic knowledge from deficits in working memory. Most of these tests, standardized on monolingual English-speaking adults, are designed primarily to localize brain dysfunction in specific domains (motor, visuo-spatial and cognitive-linguistic). As such, they are not suited for use with children with neuro-developmental disorders associated with language-learning

difficulties such as Williams syndrome or SLI or developmental dyslexia.

When a normal adult brain gets lesioned, these tests reveal that certain functions are intact, while others are impaired, resulting in 'double dissociation' across different patients. Thus, in some forms of acquired aphasias, syntax is impaired and semantics spared and vice versa in other forms; or face processing is impaired but not other visuo-spatial functions in conditions such as prosopagnosia. Lesion studies based on such adults led to the claim that the brain has a modular structure; that a number of domain-specific cortical structures are responsible for different cogntive-linguistic functions. When such dissociations are found in children with genetic disorders, the deletion, reduplication or mis-positioning of genes has a one-to-one correspondence with specific impairments in the end state. It is assumed further that these children are born with some parts of the brain intact and other parts impaired (see Baron-Cohen 1998; Temple 1997), instead of postulating that such children use language processing mechanisms differently compared to typically developing children.

Pinker (1999) proposed that SLI and Williams syndrome, both of which have been noted to have genetic etiology, provide evidence for a double dissociation between the rule-based mechanism and associative language mechanism. Specifically, according to Pinker, SLI children are impaired in the rule-based syntactic mechanism and unimpaired in their ability to memorize words; on the other hand, children with Williams syndrome show a selective sparing of syntax in the presence of an impaired associative memory. Since the two mechanisms can fail independently in two distinct developmental disorders, he feels that we have evidence for genetic double dissociation. Aspects of the genetics of Williams syndrome are discussed later in this paper. For details about molecular genetic findings concerning SLI, see Williams and Stevenson (2001).

Karmiloff-Smith (1997, 1998) has been consistently arguing against such unquestioned application of adult neuropsychological models in explaining neuro-developmental disorders, especially SLI and Williams syndrome. Her argument is that the brains of genetically impaired children develop differently at multiple levels – brain anatomy, brain biochemistry, temporal patterns of brain activity through embryogenesis as well as in the postnatal period. She and her colleagues (e.g. Paterson *et al* 1999: 2357) conducted a series of studies and concluded that "cognitive scientists, neuroscientists and philosophers of mind cannot use the purported sparing and impairment of a cognitive function in middle childhood or adulthood to support the claim that cognitive modules are innately specified in infancy".

There are other reasons why genetic determinism and the implied cognitive modularity are problematic in characterizing neuro-developmental disorders affecting language learning. These disorders are often associated with diffuse cortical damage rather than focal lesions; sometimes there is involvement of sub-cortical structures. The question of brain plasticity or equipotentiality (that is, absence of left hemispheric dominance for language functions) is also a serious issue in this population. It is only recently that systematic studies of the effects of unilateral lesions in infants and children have begun to be reported. Bates *et al* (1999) conducted a review of such studies, all of which presented a paradox. In the case of unilateral lesions in infants and children, there were surprisingly few differences between left hemisphere (LH) and right hemisphere (RH) cases whereas with adults, in 95% to 98% of all the cases, whether of speaking or signing individuals, it was a LH lesion that led to aphasia with disruption to both expressive and receptive abilities. While commenting on this paradox, the authors state (p. 533) "an intact LH is not required for development of language, yet it appears to be critical for normal language functions in the adult. If the LH is not essential for normal language development, how and why does the characteristic form of brain organization for language observed in adults come about in the first place? Something at the beginning of life must favour LH mediation of language under normal circumstances. If that something is not language itself, what is it?". The implication is not that high level cognitive modules exist in the left hemisphere right from the start, but that the processes underlying language learning in different environments themselves may contribute to LH mediation.

Bates *et al* (1999) also stated that representational nativism (that the LH comes into this world with hard-wired linguistic knowledge) is incompatible with 15 years of evidence in developmental neurobiology regarding neural plasticity and the role of experience in determining cortical specialization. It is also incompatible with the fact that children with severe LH injuries go on to acquire language abilities in the normal range. It appears that genetic constraints on cognition are less direct especially in the case of complex disorders (see Elman *et al* 1996). I shall elaborate this point with reference to two well-researched disorders of language learning, developmental dyslexia and Williams syndrome.

5. Static vs dynamic explanations of complex disorders affecting language learning

5.1 *Developmental dyslexia*

Drawing on his earlier research (Pennington 1997), Pennington (1999) offered the following description with regard to the genetic basis for dyslexia:

Dyslexia is not an X-linked disorder. Little if any published evidence of parental gender effects on transmis-

sion is available. Simple polygenic or multi-factorial transmission can be rejected because a major locus effect is seen in several samples. This effect reportedly acts in an additive or dominant (but not recessive) fashion. A monogenic hypothesis is also rejected because dyslexia is genetically heterogenous. A necessary disease allele hypothesis can be rejected because evidence exists of a major locus effect on the transmission of normal variation in reading skills. Therefore, traditional linkage analysis is not the most appropriate method by which to identify the major loci [that is, the quantitative trait loci (QTLs)]. Instead, a type of sibling-pair linkage analysis is more appropriate. Such a study has provided significant evidence of a QTL located in a two centimorgan (2–cM) region of the interval between the D65105 and TNFB markers situated in chromosome 6p21·332 with deficits in phoneme awareness phenotype in certain dyslexic families. There is additional evidence linking the same sample for a different phenotype, deficits in word recognition and markers in the centromeric region of chromosome 15. Pennington (1999: 312) states that "once we have a better understanding of the genetic mechanisms, we can also conduct much more revealing studies of environmental factors, both risk factors and protective ones, which undoubtedly operate in the transmission of both abnormal and normal reading skill. Once the genes are identified clearly, we can begin to trace the dynamic developmental pathway that runs from gene to brain to behaviour."

To clinicians who have to assess the phenotypic outcomes that show a great deal of variability, the static descriptions hardly help. The realization that in developmental dyslexia, over the course of development the gene differences lead to differences in brain structure which in turn will influence cognitive ability such as phonological processing has led to models that began to look more seriously at the environmental factors such as orthographic principles characterizing the writing system of a given language. To elaborate, deficits in phonological processing are known to lead to problems in rapid naming, spelling and poor performance in phonological awareness tasks. However, the actual difficulty in reading experienced by the child depends upon the orthography and the kind of literacy instruction he or she receives. Thus, for instance, what precipitates dyslexia to a greater extent in an English speaking population compared to an Italian speaking population is not differences in genetic make-up (the same gene may be defective in both populations). Instead, it is the complexity of the mapping between orthography and phonology in English that results in English speaking dyslexic children experiencing greater difficulty in naming and spelling than Italian children (Italian is characterized by more transparent phonology-orthography relations). Such cross-lingustic observations (Lindgren *et al* 1985) have led to reconceptualizing earlier models of dyslexia (see Morton and Frith 2001).

Cognitive scientists also began to look beyond the behavioural phenotypes. The search is on for a neurophysiological phenotype for developmental dyslexia. McCandliss and Noble (2003) published a review of cognitive neuroscience investigations into the biological basis of developmental dyslexia. They too pointed out that the cognitive phenotype associated with this disorder is a deficit in phonological processing as revealed in poor performance in analysis and synthesis of sounds within syllables. Some of the studies they looked at revealed that there are two distinct cortical areas connected to this dysfunction: the left perisylvian area typically involving the superior temporal gyrus (STG) and a portion of the left occipito-temporal extra-striate visual system near the fusiform gyrus (FG). Studies have shown that significant structural and functional differences exist in relation to STG between dyslexic and non-impaired people and that FG is associated with visual word-form perception in skilled adult readers. They proposed that these two regions interact during the typical development of reading skills. The neuro-imaging studies have revealed that dyslexics exhibit a reduced tendency to recruit left perisylvian regions (STG in particular) when faced with a phonologically challenging task such as, for instance, deciding whether the names of two letters rhyme. Electrophysiological, anatomical and neuro-imaging studies have also shown that the later stages of skilled reading are linked to specialization of the left fusiform gyrus in the extrastriate visual system. Specialization emerges slowly over the first several years of reading experience. This region, referred to as visual word form area (VWFA), responds to visually presented alphabetic characters and not to pseudo letters or random letter strings. In contrast to non-impaired readers, dyslexics have been shown to underutilize this region during word reading tasks. This was found to be true across different writing systems.

These researchers argued that the advantage of pursuing a biological phenotype (seeking evidence at the brain level) rather than a cognitive phenotype (seeking evidence at the functional level) for dyslexia is that it accounts for the fact that dyslexia may take on different forms in different writing systems and yet different behavioural outcomes may share one underlying biologically defined core deficit. We need to understand the developmental implications of this perspective (McCandliss and Noble 2003). The functional and structural abnormalities in the STG that subserve phonological processing may have a cascading effect on the development of rapid word recognition processes during the years when VWFA is becoming increasingly specialized to respond to the regularities in the writing system. Studies cited in McCandliss and Noble (2003) demonstrate a

relationship between decoding ability (converting letters into sounds) and the development of VWFA specialization, thereby suggesting a relationship between the degree to which a child might successfully engage in decoding and the degree to which his or her left fusiform gyrus becomes tuned via experience to become responsive to orthographic principles of a given writing system. They go on to cite intervention studies that succeeded in improving not only the reading skills, but also had an impact on the degree to which the STG was recruited in phonologically challenging tasks. The intervention involved having children to delete a single grapheme (letter unit) within a word to make a new word. In other words, evidence is accumulating to lend support to the idea that functional properties of certain brain areas change with learning, development and intervention. This view, termed neuroconstructivism by Karmiloff-Smith and her colleagues, has also informed current debates about fractionated cognitive abilities in yet another neurodevelopmental disorder, the Williams syndrome.

5.2 *Williams syndrome*

Williams syndrome is a genetic disorder characterized by dissociations in cognitive functions both within and across domains – below average IQs, extreme deficits in spatial cognition, but excellent face processing skills, spared linguistic abilities and so on. The static description of the syndrome at the genetic level is as follows (taken from Karmiloff-Smith 1997):

Elastin gene deleted – Not expressed in the brain but claimed to explain abnormalities of connective tissue.

LimKinase 1 gene deleted – Expressed in the brain and claimed to explain spatial impairments.

F2D3 gene deleted – Expressed in the brain and other areas and claimed to explain some of the musculo-skeletal abnormalities.

These characteristics may not be present in every case diagnosed to have Williams syndrome.

Early research in the 1960s made a connection between supra valvar aortic stenosis (SVAS) and Williams syndrome and debated on the possible genetic relations between the two conditions. Linkage analysis in families with autosomal dominant SVAS demonstrated a linkage between SVAS phenotype and DNA markers on the long arm of chromosome 7. Elastin or ELN was one of the candidate genes whose mutation results in SVAS. Later studies discussed sub-microscopic deletions of chromosome 7q11·23 resulting in Williams syndrome (for more details about genetic as well as neurobiological profiles of Williams syndrome see Mervis *et al* 1999).

Considerable behavioural evidence exists today that questions the dissociation between visuo-spatial abilities (supposedly impaired) and language abilities (supposedly intact). Paterson *et al* (1999) have demonstrated through systematic studies of infants with Williams syndrome and Downs syndrome that Williams syndrome subjects do well in infancy on numerosity judgments but poorly in adult hood. (Numerosity refers to a test in which the subject is presented a series of displays in which pictures of everyday objects are shown, with their number and type being varied. The aim of the test is to monitor the subject's sensitivity to numerical changes.) For certain aspects of language (e.g. vocabulary), they perform poorly in infancy and well in adulthood, suggesting that researchers cannot depend on phenotypic outcomes to make generalizations about impaired or intact modules in the initial state. It must be noted that these results reflect relative comparison and not absolute judgments. It has been demonstrated that even within the language domain, there are multiple dissociations. Specifically, the longitudinal studies reported by Bellugi *et al* (1999b) showed that Williams syndrome children (in the age range 3 to 23 years), unlike Down syndrome children, showed developmental trajectories in the cognitive domains of lexical knowledge, spatial cognition and face processing that are dramatically different from one domain to the other. On lexical knowledge, the Williams syndrome subjects began very low, but showed a sharp increase with age; on face processing Williams syndrome children performed well right from the beginning. Their spatial cognition abilities were consistently lower than those of age matched Down syndrome subjects. Since a given behavioual outcome in Williams syndrome seem to depend upon the age of the child, the linguistic environment (monolingual or multilingual), the comparison groups used, the techniques and materials employed to test the various abilities, caution should be exercised in reaching conclusions about absolute linguistic abilities. Paterson *et al*'s (1999) studies of normally developing children, Williams syndrome children and Down's syndrome children has concluded that it is not advisable to assume that the phenotypic outcomes noted at middle childhood are reflections of infant starting states; neither are we justified in assuming that modules subserving the abilities under examination start out as either intact or impaired. Neurophysiological studies of Williams syndrome indicate that the well-developed language system in these individuals may not be lateralized to the left hemisphere as it is in normal populations. The studies, based on event related potentials (ERP) in 10 subjects with Williams syndrome in the age range of 10–32 years, have suggested that the neural systems subserving cognitive functions such as language and face processing in Williams syndrome may be different from normal (see Bellugi *et al* 1999b for details of these studies).

Linguistic abilities, especially in the area of lexical development of English and Italian speaking Williams syndrome children, revealed a number of atypicalities.

The first words of children with Williams syndrome precede pointing; children with Williams syndrome do not obey certain lexical constraints on learning new words; children with Williams syndrome show similar lexical priming as normals, but integration of semantics into online sentence processing is abnormal; children with Williams syndrome have phonological short-term memory abilities significantly lower than vocabulary knowledge and mental age. In other words, the manner in which the lexicon is learned and used often involves different processes compared to normal children. Drawing on studies that made use of a developmental perspective, Karmiloff-Smith (1997) concluded that cognitive neuroscientists must:

- Focus on how genes interact amongst themselves and how they are expressed through interaction with the environment.
- Focus on how environments of atypical children may differ from normal environments.
- Go beyond behavioural equivalence.
- Consider the possibility that abnormal development may show associations or dissociations due to factors that may differ from normal development where the same may not hold.
- Consider the process of gradual modularization as the product of development rather than its starting point.
- Consider the trade-off in human infants between minimal cortical pre-specification and maximum plasticity via a long period of postnatal brain development.

In Williams syndrome individuals the authors found a strong relationship between vocabulary knowledge and short term memory that is not seen beyond the age of 4 years in normal development, suggesting that people with Williams syndrome exhibit relative advantage with regard to language over other domains. It is erroneous to conclude that their language domain is intact in the face of damage to other congnitive domains. Further, even within the subdomain of language, namely lexical acquisition, there are relative strengths and weaknesses. In a later article Karmiloff-Smith *et al* (2003) argue that impairments to complex cognitive outcomes are unlikely to be explained by single gene/cognitive phenotype mappings and that it is time that myths of static intactness be dethroned in favour of studying the dynamics of developmental trajectories.

This brief review of studies based on Williams syndrome and dyslexia reveals that searching for behavioural phenotypes within a model that gives importance to the environment and developmental time scale is more productive for the purposes of assessment and remediation than determining the genetic bases of the disorder using expensive DNA based tests.

To conclude, since genetic constraints interact with both internal and external environmental influences in giving rise to the phenotype, and since the relationship between the genome and the phenotype is highly non-linear in complex disorders affecting language learning, we need to pay attention to the adaptive aspects of behaviour. To understand adaptation, we need to look at the environment. Basically, to paraphrase Elman *et al* (1996), we need to keep reminding ourselves that development itself is at the heart of knowledge acquisition and use.

Acknowledgments

Preparation of this paper was made possible by a grant made to the author from the University Grants Commission, New Delhi under their Research Awards scheme for the period 2002–2005. I have benefited from the comments of all the anonymous reviewers and the editors on earlier versions of this paper.

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*e*Publication: 3 March 2005