Language development in Down syndrome and fragile X syndrome

Current research and implications for theory and practice*

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In this chapter, we review research on the development of language in Down syndrome and fragile X syndrome, which are the two most common (known) genetic causes of mental retardation (Dykens, Hodapp, & Finucane 2000). In doing so, we address three goals. First, we hope to provide a concise but comprehensive characterization of the profile of language development associated with each syndrome. More extended reviews of the literatures on these syndromes can be found in Chapman (2003) and Murphy and Abbeduto (2003). Second, we argue that the data on language development in these two syndromes are relevant to long-standing controversies in the study of language development more generally. These controversies are embodied in the contrasting claims of the modularity and interactionist accounts of development (Abbeduto, Evans, & Dolan 2001a; Chapman 2000). The modularity account presumes strong innate constraints on development, a rather minimal and circumscribed role for experience, and a relative independence of language development from other facets of development. In contrast, the social-interactionist account supposes intimate bidirectional influences between language development and nonlinguistic developments in other domains (e.g., social cognition) and a critical role for experiences, particularly experiences in social interaction with caregivers and other supportive, competent language users. Third, we briefly sketch some of the implications for clinical practice of the empirical research we consider.

The chapter is organized as follows. First, we consider research on Down syndrome and its implications for developmental theory and clinical practice.

Next, we consider research on fragile X syndrome and its implications. We conclude by briefly considering the value of integrating research on the two syndromes.

1.   Language development and the behavioral phenotype of Down syndrome

Down syndrome results from a third copy of all or part of chromosome 21, with its attendant consequences of gene dosage effects on fetal development, physiology, and brain functioning. Extra copies of the 225 genes (Hattori, Fujiyama, Taylor, Watanabe, Yada, Park, Toyoda, Ishii, Totoki, Choi, Groner, Soeda, Ohki, Takagi, Sakaki, Taudien, Blechschmidt, Polley, Menzel, Delabar, Kumpf, Lehmann, Patterson, Reichwald, Rump, Schillhabel, Schudy, Zimmermann, Rosenthal, Kudoh, Schibuya, Kawasaki, Asakawa, Shintani, Sasaki, Nagamine, Mitsuyama, Antonarakis, Minoshima, Shimizu, Nordsiek, Hornischer, Brant, Scharfe, Schon, Desario, Reichelt, Kauer, Blocker, Ramser, Beck, Klages, Hennig, Riesselmann, Dagand, Haaf, Wehrmeyer, Borzym, Gardiner, Nizetic, Francis, Lehrach, Reinhardt, Yaspo; Chromosome 21 mapping and sequencing consortium 2000)[1] on chromosome 21 have more than 80 known physical and behavioral consequences (Epstein, Korenberg, Anneren, Antonarakis, Ayme, Courchesne, Epstein, Fowler, Groner, Huret, Kempter, Lott, Lubin, Magenis, Opitz, Patterson, Priest, Pueschel, Rapoport, Sinet, Tanzi, & de la Cruz 1991; Korenberg, Chen, Schipper, Sun, Gonsky, Gerwehr, Carpenter, Daumer, Dignan, Disteche Graham Jr., Hugdins, McGillivray, Miyazaki, Ogasawara, Park, Pagon, Pueschel, Sack, Say, Schuffenhauer, Soukup, & Yamanaka 1991; Reeves, Baxter, & Richtsmeier 2001). Thus, Down syndrome differs from a single-gene alteration, as in fragile X syndrome, or a small set of affected genes, as in Williams syndrome, in the number of potential genetic correlates for behavioral consequences. Phenotypic characteristics in the population thus reflect the over-expression of multiple genes on chromosome 21, the interaction of differing alleles with one another, and the genetic makeup of other chromosome pairs.

Intensive research on language and cognitive development in children and adolescents with Down syndrome has given us a detailed picture of the behavioral phenotype associated with the syndrome, its developmental emergence, and the wide individual variation in development that can occur (Abbeduto, Pavetto, Kesin, Weissman, Karadottir, O'Brien, & Cawthon 2001b; Chapman & Hesketh 2000; Miller 1999; Roizen 2001). This research has also revealed a

number of factors that affect language learning in specific domains (Chapman 2003; Chapman, Hesketh, & Kistler 2002).

2.   The emerging behavioral phenotype in Down syndrome

Infancy (*0–4 years*). Nonverbal cognitive delays on both standardized and Piagetian tasks emerge at ages 0 to 2 years and accelerate at ages 3 and 4; social skills appear commensurate with mental age, as does comprehension of vocabulary (Dykens, Hodapp, & Evans 1994). Studies of young children with Down syndrome make clear that problems emerge in prelinguistic communication, with less frequent nonverbal requesting behavior than children of comparable mental age (Mundy, Kasari, Sigman, & Ruskin 1995), and continue with the slower accumulation of productive vocabulary relative to mental age, even when signing is taken into account (Miller 1995). The proportion of preschool children with Down syndrome showing significant delays in productive vocabulary development increases with age (Miller 1999). Expressive language also lags behind social skills (Dykens, Hodapp, & Evans 1994). Comparison to typically developing children matched on overall language age shows no difference in use of gestures but fewer two-word combinations on the part of children with Down syndrome (Iverson, Longobardi, & Caselli 2003). Speech, too, is affected in Down syndrome, with a slower transition from babbling to speech and poorer intelligibility (Stoel-Gammon 1997).

Childhood (*4–12 years*). Expressive language delays in vocabulary, utterance length, utterance complexity, and grammatical morphology continue in childhood relative to receptive vocabulary, syntax comprehension, and nonverbal cognition (Chapman & Hesketh 2000; Cunningham, Glenn, Wilkinson, & Sloper 1985). Speech development shows a longer period of phonological errors and more variability in production (Stoel-Gammon 1997). Intelligibility of the speech produced by children with Down syndrome is a frequent concern of parents (Kumin 1994). Nonverbal cognitive development reveals specific deficits in verbal working memory (Marcell & Weeks 1988). Socially, children with Down syndrome have more behavior problems than siblings without Down syndrome, but fewer compared to other children with other types of cognitive disability (Stores, Stores, Fellows, & Buckley 1998). Problems that do occur tend to be anxiety, depression, and withdrawal, and these increase with age (Dykens & Kasari 1997).

Examination of social skills shows some specific deficits despite the reported high levels of sociability displayed by children with Down syndrome on average: skill in emotion recognition is delayed (Kasari, Freeman, & Hughes 2001), especially for fear and surprise (Wishart & Pitcairn 2000); and preference for social interaction rather than object manipulation is more prolonged developmentally than one would expect on the basis of other cognitive tasks (Kasari, Sigman, Mundy, & Yirmiya 1990; Kasari, Freeman, Mundy, & Sigman 1995). There is also evidence of excessive delays in some facets of understanding the mental states of other people (Abbeduto et al. 2001b; Yirmiya, Erel, Shaked, & Solomonica-Levi 1998; Zelazo, Burack, Benedetto, & Frye 1996). Children with Down syndrome respond more often to distress in others by looking to them more, and offering more comfort, than typically developing children matched for mental age; but are less likely to feel the same emotion as the protagonist in hypothetical situations (Kasari, Freeman, & Bass 2003).

Adolescence (*12–18 years*). Comprehension of words appears more advanced than comprehension of syntax and nonverbal mental age, when measures of vocabulary such as the Peabody Picture Vocabulary Test are used (Chapman, Schwartz, & Kay-Raining Bird 1991). More recent research (Chapman 2003), however, shows that the advantage relative to mental age disappears when a vocabulary test selected for conceptual difficulty, rather than frequency of occurrence (e.g. the vocabulary subtest of the Test of Auditory Comprehension of Language-3), is used, although grammatical morpheme and elaborated sentence comprehension are poorer yet (Abbeduto, Murphy, Cawthon, Richmond, Weissman, Karadottir, & O'Brien 2003; Chapman et al. 1991). Comprehension of syntax lags nonverbal cognition in adolescence (Rosin, Swift, Bless, & Vetter 1988). Longitudinal study shows actual loss of skills in receptive syntax in some individuals through late adolescence and the beginning of young adulthood (Chapman et al. 2002).

Expressive language deficits relative to nonverbal mental age persist, with grammatical morpheme production more deficient than predicted on the basis of MLU or the lexicon (Vicari, Caselli, & Tonucci 2000). Grammatical and lexical verb use per utterance is less frequent than would expect based on MLU, but lexical diversity of narrative samples is greater (Hesketh & Chapman 1998). The proportion of verbs that are metalinguistic or metacognitive, however, is significantly less than MLU controls (Hesketh & Chapman 1998), a finding which may be related to earlier emerging problems in emotion recognition or to differences in parent input (Tingley, Gleason, & Hooshyar 1994).

Importantly, however, adolescents continue to make progress, albeit it slow, in utterance length and sentence complexity (Chapman, Seung, Schwartz, & Kay-Raining Bird 1998; Chapman et al. 2002). Longer utterances as measured by MLU are associated with more complex sentence structures, and complex sentence use is as advanced as MLU-matched controls (Thordard-ottir, Chapman, & Wagner 2002; Grela 2003). Speech intelligibility continues to be a concern, with more variability in fundamental frequency, rate control, and placement of sentential stress than expected. Problems of auditory verbal short-term memory persist (Seung & Chapman 2000), and visual short-term memory begins to lag visual cognition on tests of these skills (Chapman et al. 1991). Social development continues, with fewer behavioral problems than peers with other cognitive disabilities (Pueschel 1996).

Young adulthood. Skill patterns in young adulthood (19–27 years) are similar to those of late adolescence. Longitudinal study shows loss of syntax comprehension skill (Chapman et al. 2002). Progress in expressive language learning continues and includes the acquisition of complex syntax (Thordardottir et al. 2002). Speech problems, still frequent, include a higher incidence of hypernasality and stuttering (Kumin 1994), but intelligibility improves with chronological age and hearing status (Chapman et al. 1998). Behavioral symptoms of dementia are not evident in young adulthood; indeed, they only begin to emerge at age 50 for approximately half the individuals studied, linked to the increase in beta-amyloid protein associated with three copies of the APP gene on chromosome 21 (Silverman & Wisniewski 1999) and the moderating influence of APOE alleles.

3. Predictors of individual difference

Although Down syndrome is associated with a typical behavioral phenotype, it is important to acknowledge that the syndrome is accompanied by wide individual differences in developmental rate. What predicts the individual variations? Evidence from language learning in children with Down syndrome can partially illuminate a long-standing controversy in theories of language acquisition: the question of whether nonverbal cognition determines language learning rate. The argument that nonverbal cognition should predict a significant proportion of the variance in language learning stems from the interactionist theory's belief that general cognitive mechanisms drive language learning, rather than language-specific ones (Abbeduto et al. 2001a). However, amount

of language input, enriched learning environments, social skills, motor skills, working memory skills, and hearing status, among other variables, will also contribute to language learning, in the interactionist account (Chapman 2000). A modular view, in contrast, predicts a reduced correlation of nonverbal cognition and language measures, and synchrony among the language measures.

Predictors of individual difference in comprehension skills (both syntactic and lexical) include chronological age and nonverbal cognition; hearing contributes significant explained variance to the measure of grammatical morphology comprehension (Chapman et al. 1991). Longitudinal evaluation, with the addition of auditory working memory to the predictor set, and separation of nonverbal cognition into pattern analysis and visual short-term memory skills, reveals that the visual and auditory working memory, together with chronological age, are the best predictors of overall syntax comprehension; and the rate of change in visual working memory skill predicts rate of change in syntax comprehension (Chapman et al. 2002). Auditory short-term memory skills do not change over this period, an observation also reported by Laws and Gunn (2004). Thus, cognitive variables predict language learning, as the interactionist view would expect; but they are the variables of working memory, rather than visual pattern analysis skill.

The best model for predicting individual difference in longitudinal measures of expressive language skill, as indexed by MLU, contains syntax comprehension at study start, to predict production at study start; and slope of comprehension change, to predict rate of expressive language growth. How can it be that losses in syntax comprehension and gains in syntax expression simultaneously occur? One possibility is that language input, to the extent that it is responsive to what individuals say rather than what they understand, will be targeted at the lower production level and hence prove less useful to continued development of syntax comprehension skills. A second possibility is that the shift to vocational training in mid-adolescence, and the end of language intervention in educational programs, reduces the overall effectiveness of the language learning environment, but that expressive syntax growth can continue, for a time, to take advantage of the greater syntactic knowledge in comprehension. In either case, we need to examine the language learning environment for older adolescents and young adults with Down syndrome.

4.   Implications for modular vs. interactive theories of language acquisition

How does language learning come apart? The study of specific behavioral phenotypes also assists in assessing the relative merits of modular vs. interactionist theories of language acquisition, which imply different patterns of strength and deficit (see, e.g., Chapman 2000). In particular, modular theories would imply that a particular linguistic domain could reflect a relative strength or weakness, and syntax has been proposed as the locus of the deficit in Down syndrome (Epstein et al. 1991). This view would imply deficits in both comprehension and production in the affected domain across the developmental span. Interactionist theories, in contrast, link linguistic strengths and deficits to language learning in social, emotional, and cognitive domains, working memory systems, comprehension and production requirements, and the communicative contexts encountered (e.g. Chapman et al. 1992; Elman, Bates, Johnson, Karmiloff-Smith, Parisi, & Plunkett 1996; MacWhinney 1999). The interactionist view would thus predict that deficits in nonverbal domains, short-term memory, or long-term store would have developmentally changing effects on phenotypic profiles.

The evidence just reviewed on language development in individuals with Down syndrome supports an interactionist rather than a modular perspective (Chapman & Hesketh 2000). Syntax is indeed identified as an area of deficit in children, but particularly in production, rather than comprehension, until late adolescence, when losses in syntax comprehension are encountered. Mean length of utterance is shorter in individuals with Down syndrome than one would expect on the basis of their nonverbal cognitive skills, and the grammatical constructions observed at each utterance length are typical of those found in MLU, rather than mental age, comparison groups (Thordardottir et al. 2002). The content of stories narrated after watching short wordless videos (Boudreau & Chapman 2000) or wordless picture books (Miles & Chapman 2002), however, is greater than that in the MLU-matched group, and similar to that of the group matched for syntax comprehension skill.

Grammatical morphology in production shows the most severe deficit, including more errors and omissions than the MLU-matched group (Chapman et al. 1998; Eadie, Fey, Douglas, & Parsons 2002). The deficit extends beyond tense inflections to include non-tense grammatical morphemes (Eadie et al. 2002). Comprehension of grammatical morphology, however, is consistent with nonverbal cognitive level unless hearing impairment limits comprehension (Chapman et al. 2002). Finally, the developmental trajectory for intelli-

gibility improves with age and hearing status (Chapman, Seung, Schwartz, & Kay-Raining Bird 2000).

Thus, the trajectories for growth of syntax comprehension and production separate, and within language production, the trajectory of thematic and plot content separates from sentence form, with grammatical morphology lagging even further behind. In other words, language learning in Down syndrome fractionates along the lines of comprehension vs. production, content vs. form, and grammatical elements vs. grammatical structure.

5.   The critical period hypothesis

The Critical Period Hypothesis of language learning is another major theoretical claim that can be evaluated in individual with cognitive disabilities. The question is whether there is a limit to the developmental period in which language can be easily learned; the onset of adolescence has been thought to be the end of the period. In her work with younger adolescents with Down syndrome, Fowler (Fowler, Gelman, & Gleitman 1994) reported plateauing of expressive language, a finding consistent with a belief in a critical period. Longitudinal research by Chapman and colleagues, however, has documented continued progress in expressive language learning throughout adolescence and young adulthood (Chapman et al. 2002). The difference in the two findings appears to be due to the method of language sampling: Fowler used a conversational, rather than narrative, sample; the latter offers more opportunity for later-learned syntactic structures to be used. Additionally, the Chapman et al. work included a larger, and older, sample of adolescents as well as young adults.

The finding of continued progress in expressive syntax in adolescence has implications for our understanding of the nature of "critical periods" observed in typical second language learners. If maturation is not a factor, as the data from the group with Down syndrome suggest, then perhaps it is the amount of first language learning itself (or learning in other domains that overtakes the usual neural locus of language) that ultimately makes learning a second language more difficult, rather than the age of the learner.

6.   Implications for clinical practice

We have reviewed evidence for a specific behavioral phenotype in language and cognition for individuals with Down syndrome that includes deficits in

expressive language syntax, especially grammatical morphology, and deficits in phonological working memory; as well as strengths in lexical comprehension. We find that both auditory and visual short-term memory measures, and chronological age, predict individual differences in syntax comprehension; syntax comprehension, in turn, predicts the course of syntax production.

There is no evidence of a critical period in adolescence; rather, losses in syntax comprehension, and gains in expressive syntax. Hearing status predicts intelligibility and grammatical morpheme comprehension. Visual support for story construction differentially increases expressive syntax for individuals with Down syndrome.

From this evidence, several implications for clinical practice can be drawn. First, goals in comprehension and production should have in mind the individual's differing developmental levels in the two domains. If all intervention work is addressed to production levels, future progress may be compromised. Second, hearing status, even within the range of mild loss, plays a critical role in intelligibility and grammatical morpheme comprehension; hearing should be monitored and aided, if needed. Third, language intervention work should continue in adolescence and young adulthood, focused on both production and comprehension, for these individuals are still developing language skill. The use of visual support for storytelling may be particularly helpful in supporting more complex syntax production. Additionally, a life-long learning approach to language and literacy skills is warranted.

7. Language development and the behavioral phenotype of fragile X syndrome

Fragile X syndrome is the leading inherited cause of mental retardation and is second only to Down syndrome as a genetic cause of mental retardation (Hagerman 1999). Fragile X syndrome is caused by a mutation in a single gene (FMR1) located on the X chromosome (Brown 2002). In the full mutation, a repetitive sequence of trinucleotides (i.e., the CGG repeats), which is typically characterized by 54 or fewer repeats, expands to more than 200 (Oostra 1996). This expansion results in a silencing of the gene, which blocks production of its associated protein (Oostra & Willemsen 2003). This protein (FMRP) has been found to play a critical role in experience-dependent maturation and functioning of neural synapses (Greenough, Klintsova, Irwin, Galvez, Bates, & Weiler 2001). In contrast to Down syndrome, then, the problem in fragile X syndrome is one of gene under-expression rather than over-expression. Also

in contrast to Down syndrome, in which the genetic anomaly has the same consequences for affected males and females, fragile X syndrome differentially affects the sexes. Thus, the prevalence of affected individuals is 1 in 4,000 births in males and 1 in 8,000 in females (Crawford, Acuna, & Sherman 2001). Moreover, whereas males with the full mutation typically meet diagnostic criteria for mental retardation, only half of females with the full mutation do so, with the remainder having normal-range IQs, but learning disabilities or social affective involvement (Mazzocco 2000).

The behavioral phenotype of fragile X syndrome has been intensely investigated for the past three or more decades, although research on language has been sparse compared to that on Down syndrome (Murphy & Abbeduto 2003). Nevertheless, there are features of the fragile X syndrome phenotype that distinguish it from Down syndrome in ways that are likely to have consequences for language development (Murphy & Abbeduto 2003). Most notable in this regard are the substantially higher rates of psychopathology observed in fragile X syndrome compared to Down syndrome (Mazzocco 2000). The behaviors associated with these psychopathologies, which are described further below, can lead the individual with fragile X syndrome to avoid or have difficulties with participation in social interaction and thereby interfere with the acquisition and use of language (Cornish, Sudhalter, & Turk 2004; Murphy & Abbeduto 2005). In contrast, individuals with Down syndrome are highly sociable and keenly interested in social interaction (Kasari et al. 1990, 1995), although they may lack some important social skills that their mental age-matched typical peers possess (Abbeduto et al. 2001; Kasari et al. 2003; Yirmiya, Erel, Shaked, & Solomonica-Levi 1998; Zelazo, Burack, Benedetto, & Frye 1996). This difference in psychopathology, particularly in the social-affective realm, suggests that comparisons between fragile X syndrome and Down syndrome may be informative about theoretical controversies concerning the mechanisms of language development, especially those regarding the role of social experience (Murphy & Abbeduto 2005). In this section, we briefly describe what is known about the behavioral phenotype and development of language in fragile X syndrome. In doing so, we have distinguished between research on males and females only to the extent that that there are different findings for the two (i.e., rather than simply differences in the degree of affectedness).

8.   The emerging behavioral phenotype in fragile X syndrome

In contrast to the case for Down syndrome, developmental changes in the behavioral phenotype associated with fragile X syndrome have not been well characterized (Murphy & Abbeduto 2005). In part, this reflects the fact that the diagnosis of fragile X syndrome is not confirmed until near the age of three or four years on average (Bailey, Skinner, Hatton, & Roberts 2000a) despite the fact that many of these children exhibit delays during the first year of life (Mirrett, Bailey, Roberts, & Hatton 2004). As a result of this delay in diagnosis, studies of behavioral development in infancy are rare and thus, a critical portion of the life span is unexplored for this population. It is also the case, however, that many studies of the behavioral phenotype of fragile X syndrome, especially those focused on language, have involved samples of affected individuals that are heterogeneous with respect to age, with comparisons made to various control samples but with little attention to age-related differences within the samples (Murphy & Abbeduto 2003, in press). This lack of a developmental perspective is particularly problematic in light of the fact that the physical stigmata associated with syndrome, including the elongated face, prominent ears, and (among boys) enlarged testicles, are actually exacerbated with age (Hagerman 1999). Moreover, there is now convincing evidence that the rate of cognitive development, at least as reflected in IQ, slows during late childhood and adolescence in both males and females with fragile X syndrome (Dykens, Hodapp, Ort, Finucane, Shapiro, & Leckman 1989a; Hagerman, Schreiner, Kemper, Wittenberger, Zahn, & Habicht 1989). More studies charting the longitudinal trajectory of language in fragile X syndrome are required before we can fully understand the mechanisms of development in this population.

Development in childhood.   Studies employing gross measures of language that summarize performance across the many domains of language (e.g., vocabulary, syntax) have generally found that delays in language during childhood are no more severe than observed in other domains, such as nonverbal cognition, at least for individuals with fragile X syndrome who do not also meet diagnostic criteria for autism (Bailey, Hatton, Mesibov, & Ament 2000b). Such summary measures, however, may obscure the existence of varying degrees of delay and differing trajectories across the different components of language (Abbeduto & Murphy 2004). Indeed, the need for attempting a more nuanced characterization of language is supported by the findings of a longitudinal investigation conducted by Roberts, Mirrett and Burchinal (2001). These investigators found

that between the ages of two and seven years, males with fragile X syndrome displayed greater delays relative to their typical age-matched peers in expressive language than in receptive language. In particular, the rate of development was one-third the typical rate in language expression and one-half the rate in the domain of receptive language for the boys with fragile X syndrome. This advantage of reception over expression, however, appears to diminish, at least for some individuals with fragile X syndrome, in adolescence and adulthood (Abbeduto et al. 2000; Madison, George, & Moeschler 1986).

The profile of delays is even less clear for other linguistic distinctions (Abbeduto & Hagerman 1997). In a study of three, 10- to 14-year-old males with fragile X syndrome, Paul, Cohen, Breg, Watson and Herman (1984) documented delays in the syntactic maturity of the boys' conversational language that exceeded their delays in nonverbal cognition. In a study of a single family affected by fragile X syndrome, Madison et al. (1986) found that the only young girl in their sample similarly achieved an MLU in conversation that was substantially below expectations based on her cognitive or receptive language ability. In contrast, Ferrier, Bashir, Meryash, Johnston and Wolff (1991) found no differences between the conversational syntax of males with fragile X syndrome and typically developing males matched to them on age and cognitive level; however, the groups included both children and adults, with no analyses conducted to examine possible age differences.

In summary, results to date suggest that children with fragile X syndrome have especially severe deficits in expressive language. At least some children with fragile X syndrome have especially severe delays in syntax, although it is not clear whether the majority of children with fragile X syndrome display such asynchrony between syntax and cognition. Finally, there have been no studies focused on other important domains of language (e.g., lexical ability, pragmatics) in children with fragile X syndrome (Abbeduto & Hagerman 1997).

Development in adolescence and adulthood. There is evidence from several longitudinal investigations that language in fragile X syndrome, like cognition, is characterized by a declining rate of development, or increasing delay, in later childhood and early adolescence for both males (Bailey et al. 1998; Dykens, Hodapp, Ort, & Leckman 1993; Fisch, Holden, Carpenter, Howard-Peebles, Maddalena, Pandya, & Nance 1999; Freund, Peebles, Aylward, & Reiss 1995; Prouty, Rogers, Stevenson, Dean, Palmer, Simensen, Coston, & Schwartz 1988; Roberts et al. 2001) and females (Dyer-Friedman, Glaser, Hessel, Johnston, Taylor, Wisbeck, & Reiss 2002). In these studies, however, the measures of language have been quite broad (e.g., verbal IQ) thereby making it impossible

to determine whether the trajectory of development is variable across different domains, with some domains showing more pronounced declines in rate of growth than other domains (Murphy & Abbeduto 2003).

Cross-sectional comparisons with typically developing individuals and other clinical groups matched on various dimensions of behavioral development (e.g., nonverbal mental age) suggest that, in contrast to the findings for childhood, developments in many domains of language keep pace with non-linguistic cognitive achievements during adolescence and adulthood. Lexical development has been found to be synchronous with cognitive ability in males and females with fragile X syndrome (Abbeduto et al. 2003; Paul, Dykens, Leckman, Watson, Breg, & Cohen 1987), although the emphasis in studies to date has been largely on the learning of concrete vocabulary and on current knowledge rather than on the nature of the strategies used to learn the meanings of new words (Abbeduto & Hagerman 1997).

Syntactic development, whether measured receptively (Abbeduto et al. 2003) or expressively (Abbeduto et al. 2000; Paul et al. 1987), is synchronous with nonverbal cognitive development in fragile X syndrome on average. Nevertheless, there do appear to be important individual differences in this regard. This is suggested by case studies of a few affected individuals who were found to make more rapid progress in syntax than in nonverbal cognition (Madison et al. 1986).

Together, the findings on lexical and syntactic development suggest that adolescents and young adults with fragile X syndrome acquire the linguistic tools needed to be successful communicators at a rate consistent with their rate of (nonlinguistic) cognitive growth. There also is evidence that the perceptual and oral-motor capabilities needed to hear and produce speech, although impaired relative to chronological age expectations, have developed to a mental age-appropriate level by adolescence (Abbeduto 2004).

In contrast to the results of studies focused on linguistic "tools," studies of various dimensions of language use in social interaction paint a picture of especially severe delay during adolescence and adulthood. Thus, perseveration (i.e., self-repetition of words, phrases, and topics) and the production of tangential language (i.e., utterances that are only loosely related in content to the conversational topic) have been found to be especially frequent in the language of adolescents and adults with fragile X syndrome (Abbeduto & Hagerman 1997; Mirrett, Roberts, & Price 2003; Murphy & Abbeduto 2003). Indeed, the rate of perseverative and tangential language distinguishes males with fragile X syndrome not only from typically developing age peers, but also from age- and developmental level-matched peers with other developmental dis-

abilities, including autism (Belser & Sudhalter 1995, 2001; Sudhalter, Cohen, Silverman, & Wolf-Schein 1990; Sudhalter, Scarborough, & Cohen 1991; Wolf-Schein, Cohen, Fisch, Brown, & Jenkins 1987). Although several methodological limitations of the studies in this area complicate interpretation (Murphy & Abbeduto 2003, 2005), the findings are consistent in documenting an especially severe problem in these domains.

Adolescence and adults with fragile X syndrome also have special difficulty producing utterances in a way that makes their intended referents clear to their listeners. In particular, Abbeduto and his colleagues (Abbeduto & Murphy 2004) found that when describing novel referents to another person, adolescents and young adults with fragile X syndrome often used the same description for different referents, and they did so significantly more often than typically developing 3- to 8-year-olds matched to them on nonverbal mental age. The youth with fragile X syndrome also were more likely than the typically developing comparison children to change their description of the referents, even when those descriptions were successful, as they recurred during the interaction. The failure to create one-to-one mappings of descriptions and referents and to retain successful descriptions will result in discourse that is difficult for others to understand (Abbeduto & Murphy 2004).

Not just the speaker role, but also the listener role, poses especially serious challenges for adolescents and adults with fragile X syndrome. In particular, they are less likely to recognize and take corrective action when they fail to understand a message addressed to them than are mental age-matched typically developing peers (Abbeduto & Murphy 2004). This is true even when the comprehension problem results from the inclusion in the message of an unfamiliar word, an ambiguous noun phrase, or a noun phrase that has no identifiable referent. Such failures are likely to "snowball" during an interaction, making comprehension and participation increasingly difficult as the interaction proceeds (Abbeduto & Murphy 2004).

In summary, although adolescents and adults with fragile X syndrome have many of the linguistic tools they need to participate at reasonably high (i.e., mental age-appropriate levels) in linguistic interactions, they often fail to do so; instead, they perseverate, produce tangential utterances, produce messages whose referents are difficult to determine, and they fail to resolve comprehension problems when in the role of listener. In the next sections, we consider some of the factors that might account for this profile of language development.

9.   Predictors of individual difference

The behavioral phenotype of fragile X syndrome includes cognitive limitations and various behaviors reflective of psychopathology. Although mental retardation is characteristic of virtually all males and many females with the full FMR1 mutation, some cognitive skills are more impaired than are others (Mirrett et al. 2003). Areas of special challenge include the processing of sequential information (Burack, Shulman, Katzir, Schaap, Brennan, Iarocci, Wilansky, & Amir 1999; Dykens, Hodapp, & Leckman 1989), arithmetic (Freund & Reiss 1991), and short-term memory (Freund & Reiss 1991). Areas of relative cognitive strength include the processing of simultaneous information (Dykens et al. 1987) and long-term memory, especially for holistic spatial information (Freund & Reiss 1991).

As noted previously, fragile X syndrome is also characterized by high rates of psychopathology. The psychopathology includes hyperarousal (Wisbeck, Huffman, Freund, Gunnar, Davis, & Reiss 2000), hyperactivity and attentional problems (Baumgardner, Reiss, Freund, & Abrams 1995; Bregman, Leckman, & Ort 1988; Dykens et al. 1989; Freund, Reiss, & Abrahms 1993; Mazzocco, Pennington, & Hagerman 1993), social anxiety (Bregman et al. 1988), and gaze avoidance (Cohen, Vietze, Sudhalter, Jenkins, & Brown 1989). Autistic-like behaviors are also frequent in fragile X syndrome (Feinstein & Reiss 2001); indeed, between 10% and 40% of affected individuals have a co-morbid diagnosis of autism (Demark, Feldman, & Holden 2003). The behaviors and limitations associated with these forms of psychopathology are likely to interfere with language learning and use within the contexts of social interaction (Belser & Sudhalter 1995; Cohen 1995; Cornish et al. 2004; Murphy & Abbeduto 2003, 2005).

Few longitudinal studies have been conducted to examine the predictive relationships between various aspects of the behavioral phenotype of fragile X syndrome and the subsequent development of language. In one of the few studies to do so (Roberts et al. 2001), it was found that cognitive ability (as reflected in IQ) predicted rate of growth in both expressive and receptive language for young boys with fragile X syndrome. This is consistent with research on mental retardation more generally: general cognitive ability appears to constrain many aspects of language development (Rosenberg & Abbeduto 1993).

In contrast to the scarcity of longitudinal studies, there have been several cross-sectional studies that have uncovered concurrent relationships between various aspects of the behavioral phenotype of fragile X syndrome and language development. Thus, perseverative and tangential language is correlated

with level of physiological arousal, at least in males (Belser & Sudhalter 1995); effectiveness in talking about referents is negatively correlated with the severity of attentional problems (Abbeduto & Murphy 2004); and the ability to resolve comprehension problems is correlated with achievements in social cognition (Abbeduto & Murphy 2004). Although such correlations are consistent with the notion that language learning and use are shaped by psychopathology at least in part through the latter's impact on social interaction, longitudinal tests of these relationships are needed to unambiguously determine the direction of causation (Murphy & Abbeduto 2005).

There is also emerging evidence that the course of language development is very different in individuals with fragile X syndrome who do and do not have a co-morbid diagnosis of autism. In particular, Philofsky, Hepburn, Hayes, Hagerman and Rogers (2004) have found that children with both diagnoses have more substantial deficits in receptive language than do children who have only a diagnosis of fragile X syndrome. Similar findings have been obtained by Murphy, Abbeduto, Giles, Bruno, Richmond and Schroeder (2004). In addition, Bailey et al. (2000b) found that the profile of impairments in children with both a fragile X syndrome and autism diagnosis is similar to that observed in children with only autism (e.g., communication is more impaired than are many other domains of behavioral functioning). At the same time, Bailey et al. (2000b) found that children with fragile X syndrome who did not meet diagnostic criteria for autism displayed more synchrony in their development across the behavioral domains examined.

In summary, various aspects of language learning and use are predicted by cognitive ability, social-cognitive ability, and various forms of psychopathology and maladaptive behavior. Many of these relationships, however, are concurrent, leaving questions about the direction of causation unanswered.

10. Implications for modular vs. interactive theories of language acquisition

Three sets of findings in the literature on fragile X syndrome argue against a modular account of language and in favor of accounts that ascribe an important role to more general learning mechanisms and experience, especially experience within the context of social interaction, such as emergentism (Abbeduto et al. 2001). First, are the findings reviewed in the previous section describing both longitudinal and concurrent relationships between language and various measures of nonlinguistic dimensions of the behavioral phenotype, such

as cognitive ability, social-cognitive ability, and psychopathology. Such relationships suggest that either there is a common causal mechanism for the linguistic and nonlinguistic domains examined or achievements in one domain are necessary for, or facilitative of, achievements in the others. Thus, these relationships are at odds with the notion that language is independent of other aspects of the mind as proposed by Chomsky and other modularity advocates. Moreover, to the extent that these relationships between the linguistic and non-linguistic domains are found to be mediated by the social interactions in which the individual participates, then an experience-dependent, interactionist position will be supported. In fact, Murphy and Abbeduto (2005) have developed a socially mediated model of language development in fragile X syndrome that attempts to account for the relationships described.

Second, Abbeduto et al. (2003) examined the concurrent relationships among various domains of language and cognitive ability for adolescents and young adults with fragile X or Down syndrome and typically developing children matched to them on nonverbal mental age. In particular, they examined correlations among measures of receptive vocabulary, receptive syntax, and cognitive ability. They found that for all groups the receptive language measures were highly correlated with cognitive ability, which is consistent with the longitudinal findings of Roberts et al. (2001). More importantly, Abbeduto et al. also found that, when cognitive ability was partialled out of the relationships among the receptive language measures, the latter were still significantly correlated for the typically developing children; however, there were fewer significant correlations among receptive language measures for the youth with fragile X syndrome and fewer still for the youth with Down syndrome. Such findings raise the possibility that increased maturity brings with it more integration of the different components of language; or, put differently, development works to reduce modularity. Such a conclusion favors an interactionist rather than modularity position.

Third, several investigators have sought to examine the relative contributions of genetic and environmental variation to the behavioral outcomes of children and adolescents with fragile X syndrome, with several such studies including gross measures of language. Dyer-Friedman, Glaser, Hessl, Johnston, Huffman, Taylor, Wisbeck and Reiss (2002) found that verbal IQs for both boys and girls with fragile X syndrome were predicted by a measure of responsiveness of the home environment, even after controlling for the effects of parental IQ and child FMRP levels. Similarly, Glaser, Hessl, Dyer-Friedman, Johnstone, Wisbeck, Taylor and Reiss (2003) found that variation in adaptive behavior, including in the communication domain, was explained in part by variations in

environmental responsiveness for males with fragile X syndrome. Such findings provide support for interactionist accounts of language development.

In summary, the results for fragile X syndrome, like the results for Down syndrome, challenge various tenets of the modularity position. Instead, the findings support the interactionist position in which language learning is seen to be influenced by, and influence, many other domains of development and is highly dependent on experience in a socially responsive and supportive environment.

11.   Implications for clinical practice

There remain many gaps in our knowledge about the extent, nature, and causes of the phenotype, including its linguistic dimensions, in fragile X syndrome. Nevertheless, it is possible to derive several implications for current clinical practice. First, it is clear that despite having the linguistic tools to perform at mental age-appropriate levels in communicative interactions, individuals with fragile X syndrome often fail to do so. This suggests that intervention must target not only the acquisition of new vocabulary and syntax, but also strategies for using new and existing forms in socially effective ways. Second, it is clear that there are intimate connections between the development and use of language and other nonlinguistic skills and behaviors. This implies that efforts to improve language and its use must also attempt to impart new cognitive and social-cognitive skills that may be prerequisites for language as well as remove barriers to langue learning and use (e.g., by reducing anxiety and hyperarousal). And finally, it is important to recognize that although there is a typical behavioral phenotype associated with fragile X syndrome, there is considerable individual variability that must be attended to in language assessment and intervention. In other words, clinicians should use their knowledge of the phenotype as a starting point to guide their assessment, while probing for the idiosyncratic strengths, weaknesses, and needs of the individual (Mirrett et al. 2003).

12.   Integrating research on Down syndrome and fragile X syndrome

Although there are commonalities in the behavioral phenotypes of Down syndrome and fragile X syndrome, there are, as we have seen, differences as well. These differences include speech intelligibility problems (more severe in Down

syndrome), auditory acuity (favoring those with fragile X syndrome), the relative delays of expressive and receptive language (more pronounced in Down syndrome, at least by adolescence), a syntactic deficit relative to nonverbal cognition (more pronounced in Down syndrome). Differences also extend beyond the domain of language to include differences in sociability (favoring Down syndrome), conceptual knowledge of the social world (favoring fragile X syndrome), and the presence of psychopathology and maladaptive behavior (more prevalent in fragile X syndrome). Direct comparisons of the two syndromes as regards language leaning and use may thus provide further insights not only into the mechanisms underlying the emergence of each syndrome's phenotype, but also of language development more generally.

Abbeduto and his colleagues have conducted such direct comparisons and with interesting results. Such comparisons have demonstrated, for example, that although youth with Down or fragile X syndrome are both poor at resolving their comprehension problems relative to mental age-matched typically developing children, the former are especially poor (Abbeduto & Murphy 2004). Additionally, youth with Down syndrome are less inclined to provide scaffolding for their listener's comprehension of referential descriptions when that scaffolding requires producing longer, more complicated utterances (Abbeduto & Murphy 2004). And finally, Abbeduto and colleagues (2003) have demonstrated that youth with Down syndrome acquire receptive vocabulary but not receptive syntax at a similar rate to their age peers with fragile X syndrome. To the extent that these differences in the domain of language are found to be related to differences on nonlinguistic dimensions of the behavioral phenotypes, then we will have further evidence against a modularity position and in favor of an interactionist position. In fact, some such relationships have been established (Abbeduto & Murphy 2004). It is hoped that further comparisons of Down and fragile X syndromes (and other syndromes) will be made in future research. Such comparisons hold promise for both clinical and theoretical work.

Notes

1. *Series editors' comment*: it is our editorial policy to list all authors at first mention, rather than just the first author and 'et al.'. Although this may lead to textually rather awkward situations, we feel that all authors, especially in groundbreaking work such as the DNA-research cited here, have a right to be mentioned in the text at least once.

The role of language and communication impairments within autism

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Delays in language development and impairments in communication ability constitute a defining feature of autism. However, these language and communication impairments can be quite varied, even in classic Autistic Disorder. By current diagnostic definition (ICD-10, World Health Organization 1993; DSM-IV, American Psychiatric Association 1994), these impairments can range from a delay in the development of expressive language to a total lack of expressive language, from problems with initiating or sustaining a conversation to use of stereotyped, repetitive, and idiosyncratic language. In this chapter we first describe the historical interpretation of the basis for the language and communication impairments in autism, beginning with Kanner's (1943) description of his 11 seminal patients and continuing through the 1990s. We then identify an emerging view of the role of language and communication impairments within autism, namely that they overlap, perhaps considerably, with the language and communication impairments observed outside of autism. We then review numerous empirical studies that have demonstrated this overlap. We conclude by offering recommendations for further, necessary empirical investigations and the theoretical implications of those investigations.

1. History of language/communication impairments in autism: Kanner's 11 patients

Communication impairments have been among the defining features of autism since Kanner (1943) first described his eleven seminal patients. The first child, Donald T., arrived at the Harriet Lane Home when he was 5 years, 1 month

of age. Before the age of 2, Donald could recite "short poems and even learned the Twenty-third Psalm and twenty-five questions and answers of the Presbyterian Catechism" (Kanner 1943/ reprinted 1985:11). However, his parents were concerned because "he was not learning to ask questions or to answer questions" (p. 11). During his two-week evaluation, Donald frequently engaged in "verbal rituals" (p. 13) using delayed echolalia by repeating phrases and questions his mother had asked him previously. As an example, when he wanted to get up after his nap, he would ask his mother to say "Don, do you want to get down?" and his mother would repeat the question to him verbatim. Donald would then tell his mother to say "All right" at which point Donald would be able to get up from his nap (p. 13). If his mother did not play her role in these verbal rituals, Donald would throw a temper tantrum. Donald believed in literal, inflexible meanings to words and "he seemed unable to generalize, to transfer an expression to another similar object or situation" (p. 14). By the age of 6;6,[1] his mother reported that "he talks very much more and asks a good many questions. Not often does he voluntarily tell me of happenings at school, but if I ask leading questions, he answers them correctly" (p. 16).

The mother of the second child, Frederick W., reported that "he had said at least two words ('daddy' and 'Dora') before he was 2 years old. From then on, between 2 and 3 years, he would say words that seemed to come as a surprise to himself. He'd say them once and then never repeat them" (p. 18). When Frederick was 4 years old, his mother tried to make him use words to ask for something he wanted or she would not give him the desired object, but he refused to comply. His mother also reported that he had great difficulty with the correct use of personal pronouns. Frederick was seen at the Harriet Lane Home when he was 6 years old. At that time, "when he responded to questions or commands at all, he did so by repeating them echolalia fashion" (p. 19).

Richard M. was brought to Johns Hopkins Hospital at 3;3 because his parents suspected that he was deaf as he did not talk or respond to questions. The intern who admitted Richard observed that "it is difficult to tell definitely whether he hears, but it seems that he does" as he obeyed commands "even when he does not see the speaker and he does not pay attention to conversation going on around him" (p. 20). During his evaluation, he "uttered short staccato forceful sounds – 'Ee! Ee! Ee!' He complied with a spoken and gestural command of his mother to take off his slippers" (p. 21). However, when she asked him a different command without gesture accompanying her speech, he again took off his slippers. At two subsequent visits to Johns Hopkins before his fifth birthday, he failed to display any expressive language gains.