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Language Phenotypes and Intervention Planning: Bridging Research and Practice

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Abstract

This paper focuses on the communication and language pheno-types associated with three genetic disorders: Down syndrome, Williams syndrome, and fragile X syndrome. It is argued that there is empirical evidence that these disorders predispose children to specific profiles of strength and weakness in some areas of speech, language, and communication, and that intervention planning for children with each syndrome may take an approach informed by these profiles. Issues related to within-group variability, shared outcomes among syndromes, and the need for empirical validation for syndrome-specific recommendations are discussed.

Keywords

behavioral phenotypes; genetic disorders; communication; language development; Williams syndrome; Down syndrome; fragile X syndrome; intervention

The study of development in children with genetic syndromes associated with intellectual disabilities has been evolving over the past 30 years. The monolithic view of general developmental delays was first challenged by Zigler [1967, 1969], who argued that at the very least, there are two distinct subgroups of children with intellectual disability to consider: individuals with organic etiologies for their delays and children whose delays originate in environmental or familial factors. Since then, the study of the organic subgroup of children wirh intellectual disabilities has become increasingly refined, leading to our current conceptualization of “behavioral pheno-types,” or the specific probabilistic behavioral outcomes that are associated with known genetic syndromes [Dykens, 1995].

At the present time, phenotype research in developmental disabilities has been primarily descriptive, with the goals of differentiating aspects of functioning that are specific to a certain condition and identifying prevalence of specific outcomes within the population of individuals presenting with that condition. But beyond its scientific importance, this type of pheno-type research may have implications for intervention for children with different genetic disorders, particularly in the area of communication and language development. Although a literature has begun to emerge concerning etiologically-specific intervention

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practices in various aspects of developmental functioning [Levine, 1993: Miller et al, 1999; Buckley and Bird, 2000; Kumin, 2003; Mirrett et al, 2003; Rondal and Buckley, 2003; Semel and Rosner, 2003], very few empirical investigations comparing approaches within or across syndromes have been conducted. Additionally, the current literature around etiologically-specific intervention practices does not compare the efficacy of difrerent procedures within a syndrome, consider the differential effects of dosage, or address goal attack (i.e., the manner and timing in which multiple treatment goals are addressed). Thus, with respect to certain aspects of vulnerability within the communication phenotype of children with a developmental disability, modifications and the use of strategies compatible with supporting growth may be alternately warranted.

In this chapter, we focus primarily on findings related to communication and language development in three different genetic disorders: Down syndrome, Williams syndrome, and fragile X syndrome. Beginning with a summary of what is known about the language and conmumication phenotypes of each disorder, we will then describe how phenotypic information can inform the assessment and intervention process, relying on what is known about Down syndrome as an example. We then discuss issues surrounding the idea of differential efficacy for specific treatments in different syndrome groups. We hope that this chapter will provide an illustration for how phenotype information can inform practice; however, we must emphasize that the empirical foundations for etiologically-specific interventions remain relatively undocumented and the issues surrounding the potential for developing phenotype-specific intervention practices are complex.

PHENOTYPIC DESCRIPTION OF COMMUNICATIVE AND LINGUISTIC FEATURES

Down Syndrome

The development of language and communication in Down syndrome has been well characterized in many areas (please see Table 1 for a summary of phenotypic characteristics in the area of communication and language by genetic syndrome). One theme that is consistently reported across studies relates to a profile of relative strengths in receptive language and relative deficits in expressive language. This profile emerges in children with Down syndrome throughout the first tew years of life and becomes more pronounced as children enter middle childhood [Miller, 1999].

In addition, Miller [1999] reports that advances in mental age seem to be instrumental in these developments, and he notes that as mental age increases, children with Down syndrome appear to make greater gains in receptive language than in expressive language skills. Miller [1999] proposes that the variable onset of this profile may point to two subgroups of children with Down syndrome: one evidencing expressive language delays from the early stages of vocabulary acquisition, and a second whose expressive language delays emerge with the development of syntax. In addition, many individuals with Down syndrome arc argued to evidence language deficits like a specific language impairment (SLI), a language profile characterized by difficulties using morphemes and marked syntactical weaknesses relative to other linguistic domains (i.e., semantics and pragmatics:

Chapman et al., 1998: Eadie et al.. 2002]. It is notable that this protile of stronger receptive and weaker expressive language persists into later childhood and adolescence with pronounced dcticits observed in fluent expressive communication, particularly with respect to syntactic skills (Chapman, 19991. However, not all areas of receptive language continue to develop on par with mental age. Although receptive vocabulary continues to develop in mental age appropriate ways, relative deficits in other areas, such as receptive syntax, are reported in adolescents with Down syndrome (Chapman et al ., 1991: Abbeduto et al. 2003].

The origins of expressive language difficulties may be observed as early as infancy in Down syndrome, when delays in the onset of some aspects of prdinguistic vocalizing have been reported, particularly in the area of canonical babbling, a specific type of multisyllabic babbling with varied consonant and vowel content that emerges in typically-developing infants between 6- and 12-months-old [Lynch et al., 1995, though sec Smith and Oller, 1981]. While not every area of prelinguistic development shows pronounced impairments [Smith and Stoel-Gammon, 1983], these delays may have important downstream effects, as prelinguistic vocal development has been linked with general speech and language skills later in childhood [Stoel Gammon, 1992]. In fact, there is evidence that delays and atypicalities in the development of canonical babbling in early development in Down syndrome are correlated with social and conununication behaviors that predict later expressive language ability [Lynch et al., 1995]. Rondal [2003] notes that additional research attention on the development of prelinguistic vocalizing in Down syndrome is warranted, in that it may “yield additional clues as to why the onset of conventional language is markedly delayed in children with Down syndrome” (p 20).

Expressive vocabulary development is delayed, with the majority of children with Down syndrome showing vocabulary skills that score at very low percentiles [Mervis and Robinson, 2000]. It is unclear whether children with Down syndrome show evidence of a vocabulary spurt as would be expected in typical language development, or whether their vocabulary acquisition follows more of a linear trend [Mervis and Becerra, 2003 for a discussion of this issue]. However, as observed in typically developing children, the use of social, referential gestures (i.e. joint attention) precedes vocabulary acquisition in Down syndrome [Franco and Wishart, 1995]. In fact, children with Down syndrome have been characterized as showing strengths in this particular area of gestural communication. with stronger gesture performances observed than would be expected for overall mental age [Singer-Harris et al., 19971], though these strengths do not appear to offset the subsequent delays in vocabulary acquisition.

Children with Down syndrome also begin to combine words when their vocabularies reach 50–100 words, similar to the pattern observed in typically developing children [Mervis and Becerra, 2003]. However, the emergence of two- and three-word utterances is offten quite delayed and does not appear tor many children with Down syndrome until they are 3–4 years of age, roughly 2 years behind that seen in typical development [Kumin et al., 1999; Rondal, 2003]. This is likely reflective of the specific deficits with morpho-syntax notable in children with Down syndrome [Chapman et al., 1991: Fowler et al., 1994], skills that become increasingly important as phrase length increases. In particular, even when nonverbal mental age and expressive vocabulary are accounted for, children and adults with

Down syndrome show delays in specific areas of morphology and syntax [complex sentences, tense and aspect, modals, and prepositions; Fowler et al., 1994; Kernan and Sabsay. 1996: Eadie et al., 2002] engendering comparisons to the language difficulties observed in children with SLI (Chapman et al., 1998].

Speech difficulties play a role in the larger difficulties experienced by children with Down syndrome in the area of expressive language, as do difficulties with the verbal shon-term memory [Jarrold et al., 2000], and in particular, the phonologiral loop [Jarrold et al., 1999]. Though some speech errors common in Down syndrome are characterized as developmentall errors, made by many young children during typical language acquisition, there are some features of speech development in Down syndrome that do not follow this normal course. One study found, for example, that children with Down syndrome make a greater number of phonological errors in their speech [Dodd, 1976]. Stod-Gammon [2003] points to several contributing factors, including anatomical differences in the central and peripheral nervous system; a typical development of vocal cords, oral cavity, palate, and muscle tone; and hearing impairments. Additionally, based upon over 1 ,600 parent surveys, a diagnosis of verbal apraxia (i.e., difficulties with motor planning) was reponed by 15% of parents of children with Down syndrotnc, though the symptoms reported suggested that perhaps even more children with Down syndrome may warranr such a diagnosis [Kumin, 2006].

Despite showing deficits in speech and some aspects of bnguage development, children with Down syndrome show competence in other areas of communicative development, including pragmatics. In particular, most studies report that children with Down syndrome show relative strengths in the development of joint attention behaviors, or the use of eye contact, gesture, and vocalization for the purposes of nonverbal social sharing [Mundy et al., 1988: Kasari et al., 1995; Mundy et al., 1995; Fidler et al., 2005]. By the time children with Down syndrome arc verbal, pragmatics have been suggested to be an area of relative strength [Johnston and Stansfield, 1997; Laws and Bishop, 2004]. In addition, the use of nonverbal communication to initiate or request social contact seems to develop in mental age appropriate ways as well[Fidler et al., 2005]. However, children with Down syndrome do show relative delays in the development of nonverbal requesting behaviors, or the use of eye contact, gesture, or vocalization for instrumental purposes [Mundy et al., 1988; Fidler et al., 2005]. These communication delays may be related to a more global delay in the development of instrumental thinking skills, which may be foundational to the ability to develop instrumental conummication skills [Fidler et al., 2005; Fidler, 2006].

Williams Syndrome

As a result of its remarkable presentation, language in Williams syndrome has received substantial amounts of research attention. A typical development in young children with Williams syndrome has been reported in the emergence of nonverbal precursor behaviors related to language acquisition [Laing et al., 2002]. For example, children with Williams syndrome speak their first words before they point to refer to objects [Mervis and Robinson, 2000] and demonstrate reduced overall frequencies of nonverbal joint attention and nonverbal requesting communicative behaviors compared to mental-age matched typically

developing children [Laing et al., 2002]. Additionally. Masataka [2001] reported the delayed onset of canonical babbling in a longitudinal study of eight infants with Williams syndrome between the ages of 6 and 30 months. Contrary to older sophistications in language usage, substantial evidence exists to suggest that most young children with Williams syndrome evidence delays in the emergence of spoken language [Singer-Harris et al., 1997: Masataka, 2001: Semel and Rosner, 2003]. In contrast with the pattern observed in Down syndrome, once language emerges, articulation has been a suggested relative strength for many children with Williams syndrome [Meyerson and Frank, 1987; Semel and Rosber, 2003].

Additionally, specitic to early lexical development, children with Williams syndrome evidence a number of anomalous patterns [as reviewed by Stevens and Karmiloff-Smith, 1997: Mervis et al., 1999]. For example, children with Williams syndrome have been suggested to rely more on the verbal short-term memory than on semantics when acquiring new words, a pattern that deviates from that observed in typically developing children [Vicari et al., 1996]. In contrast to children with Down syndrome, strength in the verbal short-term memory of children with Williams syndrome has been argued to form an alternate path from typical development to expressive vocabulary and syntactic language acquisition [Mervis and Klein-Tasman, 2000].

Furthermore, comprehension difficulties have been identified as emerging early in Williams syndrome, such that many toddlers with the syndrome evidence comprehension abilities at about half of their CA [Paterson, et al., 1999]. A case study of a 3-year-old girl with Williams syndrome provided results to show a CA-appropriate expressive language standard score (i.e., 99), with a receptive language Standard Score lagging behind by almost 40 points on the preschool language scale-4 [PLS-4; Hepburn et al., 2005]. Additionally, toddlers with Williams syndrome evidence similar receptive vocabularies compared to a matched comparison group of toddlers with Down syndrome despite a significant advantage in expressive vocabulary of 2-year-olds with Williams syndrome over 2-year-olds with Down syndrome [Paterson et al., 1999; Mervis and Robinson, 2000]. Mervis and Klein-Tasman [2000] reported relatively even development of the receptive and expressive language standard scores from the Mullens scales of early learning, that was comparable to the overall visual reception standard score, in 13 preschool children with Williams syndrmme. Many questions remain concerning the foundation, emergence, and course of early language skills, and how they lead to the unique language outcomes observed in this population, and how they are related to other aspects of development, including hyper-sociability.

Notably, the bulk of the research has focused on school-age children and adolescents, with a number of studies using children with Down syndrome or specitic language impairment as the comparison groups [Wang and Bellugi, 1994; Bellugi et al., 1999a; Klein and Mervis, 1999; Bellugi et al., 2000; Mervis and Klein-Tasman, 2000]. Bellugi et al. l2000] found that the overall language performance of children and adolescents with williams syndrome far outstripped age and intelligence quotient (IQ)-matched children with Down syndrome who demonstrate distinctly different cognitive profiles. This finding has significance because individuals with Williams syndrome perform similarly on language tasks to children who have impairments specific to language but do not evidence a global cognitive impairment [Bellugi et al., 2000]. However, the use of Down syndrome and SLI as comparison groups

weakened the initial claims of marked language strengths in Williams syndrome, as Down syndrome and SLI are conditions identified as involving significant language difficulties [Fowler, 1990: Chapman, 1995, 1999; Sigman and Ruskin, 1999].

Subsequent research has shown that individuals with Williams syndrome do show some distinct areas of relative strength in language development, but they also show areas of pronounced weakness as well. Relative strengths have been observed in lexical development [Bello et al., 2004]. Good recall of previously heard words and phrases and an unusual, sophisticated diction compared to other children [Bellugi et al., 1999b; Bellugi et al., 2000], as well as a socially engaging use of prosody, discourse, and narrative skills [Bellugi et al., 1999b) may all serve to augment an overall illusion of linguistic strength.

In terms of vocabulary performance in older children, Mervis et al, [2004] report strong peabody picture vocabulary test-III (PPVT-III) performances in school-age children with Williams syndrome, but also note that the PPVT-III primarily measures receptive concrete vocabulary knowledge, and not aspects of vocabulary related to reasoning ability or visuo-spatial processing. Despite this strength, there is also evidence of a word-tinding difficulty in this population, as older rhildren with Williams syndrome have been shown to use more iconic gestures than developmentally-matched children on a naming test [Bello et al., 2004]. On the test of relational concepts, children with Williams syndrome show poorer abstract relational vocabulary size than vocabulary-size-matched typically developing children [Mervis et al., 2004]. The authors note that it is unclear whether these differences result from developmental disability in general, or are specific to the phenotypic expression of Williams syndrome.

Findings in the area ot morphology and syntax in Williams syndrome are less clear. Some studies report that morpho-syntactic development is an area of strength in Williams syndrome [Volterra et al., 1996; Karmiloff-Smith et al., 1997; Karmiloff-Smith, 1998], while others report this is an area of delay, especially in languages with complex morphologies [Volterra et al., 1996; Levy and Hermon, 2003). However, compared to children matched for mental and chronological age with developmental delays of mixed etiology, children with Williams syndrome perform comparably on measures of expressive syntax and syntactic complexity is generally on par with mean-length utterance in this population [Mervis et al., 1999; Mervis and Klein-Tasman, 2000]. Studies considering languages with more complex morphologies than English have suggested that children with Williams syndrome show difficulty compared to mental-age-matched, younger, typically developing children [Karmiloff-Smith et al., 1997; Volterra et al., 1996]. In terms of receptive morpho-syntax, limited research suggests that in school-age and adolescent children some receptive morpho-syntactic skills tend to show a relative strength compared to children with Down syndrome and SLI and be on par with mental age [Ring and Clahsen, 2005].

The pragmatic language abilities of individuals with Williams syndrome may show impairment, relative to typically developing children and individuals with Down syndrome. Rather, more similarities may be seen with respect to the deficits associated with autism spectrutn disorders although the profile is slightly different [Laws and Bishop, 2004;

Philofsky. 2006]. While both school-aged children with autism and Williams syndrome show impairment on a parent report measure of pragmatic language functioning, the Children's Communication Checklist-2 (CCC-2; Bishop, 2003]. children with Williams syndrome may show relative strength compared to children on the autism spectrum in pragmatic areas relating to social-emotional aspects, nonverbal communication (including affective expression and understanding), and prosody, although their performances were still often within the impaired range and significantly worse than a younger, typically developing group of children [Philofsky, 2006]. Conversely, comparable deficis have been observed between children with an ASD and children with Williams syndrome in the following pragmatic areas: use of context (which includes difficulties with humor and abstract language), inappropriate initiation, and variety of interests [Philofsky, 2006]. Further, Stojanovik [2006] analyzed the conversational structure of five school-aged children with Williams syndrome by coding 100–150 utterances obtained from conversations generated by looking at pictures of familiar situations (i.e ., a birthday party, vacation, and children playing together), in comparison to typical development and children with specific language impairment (SLI). Results suggested that the conversations of children with Williams syndrome were more immature and inappropriate, in comparison with both other groups of children [Stojanovik, 2006].

Additional areas under the domain of difficulties in pragmatics for children with Williams syndrome relate to incessancy including: “cocktail party chatter” (i.e., superficial talking that lacks meaningful content), constant, inappropriate requests for ateention. excessive greeting behaviors, asking the same question over and over again, and use of indiscriminate flattery [Udwin and Yule, 1990; Schreiber, 2000; Semel and Rosner, 2003]. Finally, limitations in conversational skills relate to difficulties with “giving up the floor”; making irrelevant and tangential comments; topic maintenance; providing limited information concerning the needs of [he conversational partner: an over-reliance on the conversational partner's leads; and over-literal interpretation of conversational information suggesting difficulty in the broader construct of conversational reciprocity for these children [Meyerson and Frank, 1987; Udwin and Yule, 1990; Levine, 1993; Semel and Rosner, 2003: Stojanovik, 2006].

Fragile X Syndrome

Although fragile X syndrome remains among the least understood of disorders with known behavioral pheno-types with respect to language symptoms [Rice et al., 2005], a growing body of work has begun to create an emerging communication phenotype for children with the disorder [Paul et al., 1987; Abbeduto and Hagerman, 1997; Belser and Sudhalter, 2001; Roberts et al., 2001: Sudhalter and Belser, 2001; Rice et al., 2005]. Although the majority of language studies in tragile X syndrome have focused on older children and adolescents with the disnrder, more recent studies haw begun to consider younger children with fragile X syndrome [Roberts et al., 2001; Roberts et al., 2002; Mirrett et al., 2004; Philofsky et al., 2004; Brady et al., in press]. Language delays, particularly in the area of expressive language, are typical [Roberts et al., 2001; Rice et al., 2005]. Mirrett et al. [2004] found that by the time they were 12 months old, 88% (i.e., 14/16) of their infant sample with fragile X syndrome were evidencing ddays on an early receptive and expressive language screening

tool, the Early Language Milestone Scale-2 (ELM-2; Mirrett et al., 2004]. Further, both better cognitive abilities, as well as fewer autism symptoms, have been related to better language outcomes for young children with fragile X syndrome [Roberts et al., 2001; Philofsky et al., 2004].

Given that children with fragile X syndrome evidence language delays. Roberts et al. [2002] published a article to consider the development of social-commuuicative behaviors (i.e., language gestures, symbolic behaviors, reciprocity. etc.), considering the broader scope of behaviors involved in communication beyond just language, in young children with fragile X syndrome and the relationship of these behaviors to language development I year later [Roberts et al., 2002]. Following careful subject selection criteria to only include children (a) with a diagnosis of full mutation fragile X syndrome, (b) with developmental receptive language skills at or above 12 months and expressive language skills between 15 and 28 months (as a function of the developmental levels necessary for the study measures selected) and, (c) without a comorbid diagnosis of autism, 22 males between the chronological ages of 21 and 77 monchs (M = 49.2) were included. Participants were administered the Communication and Symbolic Behavior Scales (CSBS) and Reynell developmental language scales (Reynell) initially, as well as 1 year later. Their results confirmed delayed overall communication development in their sample, though there was considerable variability among the children. While variability was also considerable in the communication profiles of the children, relative strengths were noted in verbal and vocal communication, and relative weaknesses were noted in gesturing, reciprocity, and symbolic behaviors (i.e., response to simple directions in a play context and play skills). Further, additional relative strengths were noted in certain aspects of social communication (i.e ., behavior regulation, joint attention, and social referencing). Affect sharing occurred in the sample with scaled scores reflective of the children's mental ages, suggesting that while positive atfect was shared with typical frequency, negative affect was shared more frequently. Children in their sample who performed becter in overall use of communicative functions, vocalizations, verbalizations, and reciprocity, scored better in overall language comprehension 1 year later, while only higher scores in verbal communication predicted better expressive language development 1 year later [Roberts et al., 2002]. While this study represents an initial look at early gesture use in fragile X syndrome, the gencralizability of the findings is limited as a function of the strict inclusion criteria that precluded participation of many young children with fragile X syndrome into the study. Further, a lack of comparison to children with other types of developmental disability limits any attributable differences to cognitive delays rather than to fragile X syndrome. in particular.

Thus, not only is language development delayed in young children with fragile X syndrome, but other types of communicative behaviors appear likely to be affected in many children, as well. However, a profile of relative strength and weakness in early social communicative abilities may be characteristic of young children with tragile X syndrome. Of additional note, although some aspects of social communication have been reported to be a particular weakness in older individuals with fragile X syndrome (i.e ., communicative eye contact), this skill surprisingly proved to be a relative strength for young children with the disorder [Roberts et al., 2002].

A relative strength in receptive language compared to expressive language has been suggested to contribute to the fragile X syndrome communicative phenotype of young boys with fragile X syndrome rRobcrrs et al., 20011. This advantaged receptive language profile has been found in children with a mean age as young as 34 months, as well as in three other studies of older children with fragile X syndrome [Paul et al., 1984; Paul et al., 1987; Roberts et al., 2001; Philofsky et al., 2004]. Specitically, receptive language skills have been reported to grow at about half the rate of typically developing children, while expressive language skills were reported to increase at about one-third the rate of typical development owr time in young children with fragile X syndrome (Roberts et al., 200 I). Furthermore, by young adulthood, receptive language has been reported to be mental-age appropriate in boys with tragile X syndrome who do not meet criteria for an autism spectrum disorder (Abbeduto et al., 2003).

A number of atypicalities have been observed in the expressive language of older individuals with fragile X syndrome. Spcech-related expressive difficulties have been reported in terms of articulation and sound-sequencing difficulties, poor overall intelligibility, a harsh vocal quality, and a variable, impulsive, and rapid speech rate [Newell et al., 1983; Paul et al., 1984; Hanson et al., 1986; Wolf-Schein et al., 1987; Abbeduto and Hagerman, 1997]. Some of the speech difficulties reported in this population have: been a suggested function of a developmental dyspraxia of speech (i.e., difficulties related to the motor planning involved in speech sound production [Dykens et al., 1994; Abbeduto and Hagerman, 1997]. Although limited, syntactic usc studies of children with FXS haw suggested delays relative to MA-expectancies; however, receptive morpho-syntactic skills appear to be on par with mental age in school-age and adolescent boys [Paul et al., 1984; Abbeduto et al., 2003]. Fluency-related expressive difficulties have been noted in an increased number of dysfluencies, though it has been suggested that these dysfluencies are fewer than those observed in a stuttering disorder [Newell et al., 1983; Paul et al., 1984; Hanson et al., 1986; Wolf-Schein et al., 1987]. Pragmatics-relatcd expressive language difficulties have been distilled into the fragile X syndrome-specific use of deviant, repetitive, and tangential language that is distinguishable, in terms of an increased frequency, from children with autism and children with mental retarelation not caused by fragile X syndrome [Wolf-Schein et al., 1987; Sudhalter et al., 1990; Belser and Sudhalter, 2001; Sudhalter and Belser, 2001]. Additionally, while gaze avoidance and eye contact have been reported as problematic for adolescents with fragile X syndrome [Cohen et al., 1989: Cohen et al., 1991], these ditliculties are not necessarily problematic earlier in development [Roberts et al., 2002]. Finally. several descriptive explanations have been used for the expressive language of individuals with fragile X syndrome, including such terms as, “jocular,” “staccato,” “litany-like phraseology,” and “sing-songy” [Turner et al., 1980; Dykens et al., 1994].

IMPLICATIONS FOR INTERVENTION

At this time, there is no consensus regarding the role that this etiology-specific information should have in shaping intervention and service delivery for young children. For example, despite the fact that the vast majority of young children with Down syndrome show pronounced delays in expressive language development relative to their overall mental age,

it is still somewhat controversial ro suggest that the Down syndrome diagnosis is sufficient to warrant speech-language intervention services from the earliest stages of development [Miller, 1999]. Miller argues that, “Because language production impairments are likely to be a part of the learning impairments associated with Down syndrome, eligibility should be automatic rather than waiting to document the inevitable language delay ...”; [Miller, 1999, p 37]. While ideas such as these have been introduced in the literature tor some time [Fey, 1986], such an anticipatory stance has not been evaluated rigorously, and as such is not widely accepted by the larger research and practice communities. For example, many intervention programs do not include speech and language intervention components until the child reaches a specific age or communication milestone [Kumin, 2002].

Despite these debates within the protessional communities, there is some preliminary evidence of parental recognition that phenotypic outcomes may have important implications for how we structure intervention and therapeutic planning for children with diflerent genetic disorders. In the limited research that has been conducted to date, the preferences expressed by parents seem to show evidence of syndrome specificity [Fidler et al., 2003]. For example, compared to parents of children with other syndromes, parents of children with Down syndrome are more likely to express spontaneous concerns regarding speech-language intervention services for their children, with comments targeting a desire for more one-on-one time and increased quality of services [Fidler et al., 2003]. In conjunction with the issue of communication, parents in this study also reported other syndrome-specific desires regarding modifications to their child's current educational plan.

One challenge to taking an etiology-specific approach to intervention planning relates to the definition of behavioral phenotypes, and the degree to which this information can be used to anticipate potential areas of strength and weakness prospectively in development. Dykens [1995] presents a definition of behavioral phenotypes that takes a probablistic view, suggesting that there is a heightened probability that children with a given syndrome will show a phcnorypic outcome relative to other children with developmental disabilities, but who do not have the specific syndrome. Within this probabilistic framework lies the notion that not all children with a given syndrome will evidence the particular phenotypic outcomes that have been characterized in research studies.

Planning individualized interventions based upon what is known broadly about a phenotype must, therefore, he approached with a probabilistic interwnrion model. By this we mean, using general information about a genetic syndrome to (1) anticipate potential developmental vulnerabilities and resiliencies, and (2) incorporate an anticipatory guidance approach. After a preliminary exploration of this possible model, the separate question of differential efficacy of particular intervention approaches across groups will be discussed.

Anticipating Developmental Vulnerabilities and Resiliencies

Phenotypic research may make it possible to “know where to look” for potential vulnerabilities in early development. In essence, parents and practitioners could use information regarding phenotypic predispositions to take anticipatory stance, and to monitor poten tial areas of vulnerability that haw been linked to the child's syndrome more closely than they might otherwise. Such an approach would provide for the timely application of

incervention techniques for at-risk children. For example, since morphological development is an area of vulnerability tor children with Down syndrome compared to other children with developmental disabilities, the development of early word combinations and morpheme use could be monitored in a targeted way, with strategies employed for promoting an optimal developmental course. For children with Down syndrome who do not show emerging difficulties in this area, monitoring can continue, but intervention resources can be placed elsewhere. Table 1 provides an illustration of how one might utilize general phenotypic information to guide assessment efforts at an individual level when a diagnosis is known. This notion of using syndrome-specific predispositions tnay not only help educators make more informed choices about how to plan early intervention, bur it may also increase the likelihood that the earliest, most subtle aspects of a particular developmental problem can be detected. In knowing, for example, that a child with Down syndrome shows difficulty with expressive language development, it may be possible for interventionists to identify markers during the earliest stages of speech development—including the first year of lite—that could be evidence of this emerging difficulty. For example, it has been suggested that the atypical development of some aspects of babbling in infants with Down syndrome may be the earliest precursor of later expressive language deficits [Lynch et al., 1995]. In using this knowledge strategically, an interventionist might be able to document subtle delays in the emergence of babbling skills and perhaps target those skills at that point in early development, rather than waiting for a pronounced deficit in expressive language to become evident later in early childhood. In addition to anticipating areas of potential weakness in a child's development, it is also important to consider areas of relative strength, which, as with deficits, can be expected to be expressed probabilisrically within a given genetic syndrome. Table 2 depicts a summary of what has been written about empirically-based language and communication strengths in specitic syndromes.

When incorporating etiology-specific information in educational planning, there are two important issues to address. First, there is a risk that educators and practitioners could squander intervention resources on problems that a child does not actually demonstrate, despite a child's higher probability of demonstrating certain difficulties as a function of their diagnosis. An overly rigid application of the usc of syndrome-specific information could Jead to poor intervention planning and wasted resourses. Second, at this time, there is insutlicient empirical research that has explored the issue of the efficacy of specific intervention techniques in one group over another, dosage, or goal attack (discussed further below). There have been very few studies to examine whether specific techniques are more or less effective in treating the same targets in groups with ditferem phenotypic trajectories. Nonetheless it is possible to speculate, for example, that interventions that rely on one skill area—for example, verbal short-term memory—might be more effective tor children with Williams syndrome than for children with Down syndrome. Thus recommendations within the model presented here focus primarily on identit)ring targets and using this information to guide the selection of general techniques that have been empirically supported tor children with developmental disabilities in general, rather than specitic syndrome groups.

Adopting an Anticipatory Guidance Approach

Anticipatory bruidancc approaches refer to those interventions that actively target symptoms of a disorder that Jrc more likely to be impaired, with the general notion that if the interventions are natural and without harm, then potential “over-employment” is of low risk, while the gains may be of high benefit. This kind of approach has been employed by pediatricians in talking with parents about a child's temperament and how specific environmental demands may challenge that temperament. For example, a highly active child (temperament characteristic) is likely to behave poorly when required to sit still and wait (environmental demand). Similarly. we can take what we know about the behavioral and learning tendencies of an etiologically-defined subgroup of children with developmental disabilities and postulate amicipatory guidance strategies that may be most relevant. What follows is an example of how phenotypic information can be linked with intervention practice, within an anticipatory guidance framework .

An Illustration: Down Syndrome: Linking Phenotype to Intervention

The idea of taking an anticipatory stance throughout development may make it possible to address emerging issues earlier in development. Based on his work on the developing language profile of young children with Down syndrome, Miller [1999] recommends that the earliest stntegies for intervention in this population target two areas: (1) strengthening speech and oral motor development, and (2) promoting the use of functional communication modalities. Others have suggested that targeting critical component processes, such as verbal short-term memory and means-end reasoning, would have important implications for communicative intervention [Jarrold et al., 1999; Fidler, 2006]. Anticipatory guidance, therefore, for a young child with Down syndrome, suggests incorporating ongoing assessment of oral-motor, functional communication, verbal short-term memory, and means-end reasoning. Given the relatively high probability that these areas of development wi11 be impaired, the following are some empirically-based recommendations aimed toward ameliorating and accommodating these difficulties.

Strengthening speech development—Rondal [2003] recommends active stimulation of babbling by parents, which involves providing parents, with information both about the various stages ofbabbling development, as well as with intervention guides to promote development at each stage. Specific techniques, such as vocal stimulation [Warren and Yoder, 1998], place an emphasis on increasing the production of specchlike babble and other key prelinbruisric vocalization benchmarks. Rondal[2003] further advises that facilitating reciprocal, back-and-forth interactions with infants and toddlers with Down syndrome may stimulate greater child participation in communicative contexts.

Articulation exercises may involve heavy visual and auditory bombardment of targeted sounds including finding toys that play certain sounds and mirror imitation games [Kumin, 2003]. Knowledge that children with Down syndrome have phenocypic-specific difficulties with thdr verbal short-term memory underscores the addition of the visual modality along with heavy repetition of sounds for treating articulation difiiculcies. Others have suggested targeting articulation in Down syndrome during naturalistic interactions. One approach irwolvcs training parents to respond only to a specific pronunciation of a set of preselected

words [Dodd et al., 1994]. Other recommcndations specifically aimed at sound bombardment and practice include having “sound days” where each activiity begins with the designated sound, using children's books that offer examples of a specific sound, and highlighting a spcciiic phoneme during the use of familiar songs [Kumin, 2003].

While specific interventions for teaching phonological awareness in children with Down syndrome have been shown to improve phoneme isolation skills. as wdl as alliteration and rhyme detection, training in this area may not necessarily generalize to increased speech intelligibility [Kennedy and Flynn, 2003]. However, given the pronounced verbal processing difliculties observed in many individuals with Down syndrome, additional research is warranted to explore this connection further. In addition. while it is noted that many of these techniques were not specifically developed for children with Down syndrome, the selection of such techniques within an anticipatory guidance approach for children based on a known specific impairment, and the timeliness of implementation based on probabilistic trajectories, has the potential to advance the practice of intervention in this situation.

Targeting component processes—Targeting verbal short-term memory has been recommended for children with Down syndrome [Jarrold et al., 1999]. Jarrold et al. [1999] note that “ if one can improve the short-term memo1y skills of these individuals via some form of interveution program, then this may also lead to important benefits in terms of language and reading skills” (p. 68). Deficits in chis area haw been primarily linked to receptive grammar in empirical research [Laws, 1998], though they may also be implicated in vocabulary acquisition as well. However, empirical examination of different approaches to improve the verbal short-term memory functioning in individuals with Down syndrome has yielded limited success [Comblain, 1994]. Therefore, to address these issues, it may be important to make specitic accmmnodations (as opposed to treating the problem) to support aspects of language acquisition in children with Down syndrome, including capitalizing on visual modality strengths, increasing the density and duration of exposure to new vocabulary words, and contexrualizing language instruction.

Use of alternative modes of communications—While early speech and babbling skills are being targeted, encouraging the use of an alternative communication system, like sign language, promotes the competent development of expressive communication skills and may also reduce the level of frustration experienced by both the child and caregiver [Miller, 1999; Rondal 2003]. Use of alternative modes of communication is thought to be a bridge (rather than a hindrance) to the development of more sophisticated language forms and vocabulary, bypassing the delays imposed by speech on expressive language development [Rondal and Buckley, 2003].

Means-end thinking—Recent research suggests that children with Down syndrome are at increased risk of having difficulty with means-end reasoning, or basic instrumental problem-solving, relative to children with other developmental disabilities [Fidler et al., 2005; Fidler, 2006]. Means-end thinking may contribute to many of the motivational components involved in the use oflanguage, as well as the development of other critical functional skills in child development. To address the emerging split between the use of nonverbal communication tor instrumental versus social purpose, interventions that target early

instrumental thinking skills may be beneficial [Fidler, 2006]. For example, targeting early means-end thinking through work on contingencies and chaining behaviors together toward a goal, may, in turn, facilitate the development of instrumental communication. It may be that strengthening these skills not only has an eflect on specific early instrumental communication competence, but it may also have downstream effects on other developmental areas like motivational orientation and task persistence [Fidler, 2006].

Differential Efficacy of Interventions by Group

In conjunction with use of an anticipatory guidance approach, there may be additional possibilities tor incorporating etiology-specific information into intervention planning. There is an intriguing—though relatively unexplored—possibility that some interventions may be more effective for one group of children than another, and that these differential effects may be rooted in etiology-specitic difierences in motivational orientation, behavior, and development. This topic must be approached with tentativeness at this time in that the research community has not generated sufficient empirical evidence to support even the most basic claim of differential interwntion ctlicacy based on syndromic differences. However, it may be that some techniques are more or less effective at addressing a given dday as a result of other mediating factors associated with the larger developmental profile associated with a particular genetic disorder. Thus, it could be possible that in addition to identifying specitic targets for intervention, etiology specitic research could also be useful in helping interventionists select specific techniques, dosages, and goal attacks that may be more maximally effective.

One example of such differential etferts romes from findings relating to the nonverbal requesting deficit in young children with Down syndrome that was characterized earlier. Yoder and Warren [2002] found differential effectiveness for their responsive education/ prelinguistic mileiu teaching (RPMT) approach for children wirh and without Down syndrome. While gains in nonverbal requesting were observed in children with developmental disabilities without Down syndrome, children with Down syndrome actually showed a faster rate of growth in requesting skills when they had not received the intervention as opposed to those who had [Yoder and Warren, 2002]. The authors suggested that the persistence required of the child in RPMT may have, in fact, interacted with the motivational style of children with Down syndrome such that the increased requirement tor persistence actually inhibited children with Down syndrome from “sticking with it” to complete a request [Yoder and Warren, 2002], providing evidence that some techniques may be differentially effective for one group of children over another.

In a follow-up study, Fey et al. [2006] modified the RPMT approach by reducing the demands and requiring the child only to shift their gaze from the object of interest to the adult to obtain wanted objects. Unlike Yoder and Warren [2002], this study observed no negative impacts on growth of requests as a result of using RPMT and a trend toward positively affecting skill growth in children with Down syndrome [Fey et al., 2006]. However, due to different variables between the two studies, the authors could not affirmatively conclude that the modification of reducing the demands of the RPMT treatment was the reason children with Down syndrome difierentially demonstrated potential

beneficial effects in growth of requesting behaviors compared to the Yoder and Warren [2002] study [Fey et al., 2006]. Despite many remaining questions around how to best go about treating a requesting deficit in Down syndrome, there exists a special need for improvements in this area for children with Down syndrome (i.e., an etiology-specific goal).

Thus, it may be that some techniques are generally effective across groups, for any child with a given communication delay, and there also may be cases where specific interventions are more appropriately suited to children with a specific disorder, who may bring mediating ftctors to the intervention setting as a result of their larger phenotypic profile. Differential response to treatment within syndromes cannot yet be ruled out as a possibility for difterential treatment efficacy, as well. As intervention science continues to advance in this area, these issues may emerge as potentially vital in maximizing the efficacy of intervention implementation from the perspective of cost-efficiency and sound developmental practice.

DISCUSSION

While there has been tremendous progress in research regarding the links between genetic disorders and communication-language outcomes. the link between science and practice has only begun to be explored. As these connections are made, there are several issues that will need to be addressed. The first issue relates to specificity, and in particular, how specific a phenotypic trajectory must be to warrant attention from the practice community. As noted in Table 1, while each of the syndromes presentcd showed specific patterns of early communication and language development, there are certain areas of shared vulnerability across syndrome groups. For example, both children with Down syndrome and fragile X syndrome evidence challenges with articulation/speech issues, expressive language delays, and strengths in some aspects of receptive language. Some anticipatory guidance approaches will be applicable across phenotypes, particularly with the recommended use of compensatory communication strategies while building speech and articulation skills. Yet, while building up instrumental requesting abilities may comprise a portion of the intervention package addressing expressive language deficits in Down syndrome, such work may or may not be warranted for children with fragile X syndrome though the research in this particular area of development in fragile X syndrome remains quite limited.

In addition, there is some overlap betwecn the protiles observed in children with Down syndrome and Williams syndrome, in that both groups show delays in the onset of expressive language. It is notable, however, that the shared difficulty may ditler in the degree to which this area is impaired, with perhaps a more pronounced impairment in children with Down syndrome if one were to consider the later pcrt(mnance in expressive language in each population. Thus, while intervention strategies to improve prelinguistic development may benefit young children in either group, the specific strategies, goal attack, and dosages chosen by interventionists may differ relative to profile of speech and babbling evidenced within each group. Finally, there are areas of overlap between children with Williams syndrome and fragile X syndrome as well, specifically in the area of pragmatics. Thus, strategies that address the social uses of language and other related pragmatic skills may be warranted for both groups though approaches may vary depending upon the child.

Despite these areas of overlap, it is important to note that each syndrome presented predisposes children to a distinctive developmental linguistic profile, necessitating informed and targeted decision making to plan effective intervention in the area of speech, language, and communication. Numerous other syndromes may have their own distinct profiles in these areas as well, including Smith–Magenis syndrome. Velo-cardiofacial syndrome, Prader–Willi syndrome, and others, though relatively less is known about the language and communication trajectories associated with these disorders.

There is much promise in the use of phenotypic information to inform intervention planning even at this early stage. Perhaps adopting such an approach will prompt the research community to engage in more rigorous empirical testing of the efficacy of specific techniques tor dearly defined and different groups of children. With this scientific rigor, it may be possible not only to understand the impact of genetic disorders on developmental pathways, but to change the course of these pathways through targeted and empirically validated interventions, modifications, and treatments.

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Planning individualized interventions based upon what is known broadly about a phenotype must, therefore, be approached with a probabilistic intervention model. By this we mean, using general information about a genetic syndrome to (1) anticipate potential developmental vulnerabilities and resiliencies, and (2) incorporate an anticipatory guidance approach.

There is much promiise in the use of phenotypic information to inform intervention planning even at this early stage. Perhaps adopting such an approach tvill prompt the research community to engage in more rigorous empirical testing of the efficacy of specific techniques for clearly defined and different gro11ps of children. With this scientific rigor, it may be possible to not only understand the impact of genetic disorders on developmental pathways, h11t to change the course of these pathways through targeted and empirically validated interventions, modifications, and treatments.

Table 1

Considering the Behavioral Phenotypes: Implications for Clinical Intervention Across Genetic Syndromes

Communication/Language Feature That Is Impaired	Estimated Probability of Occurrence Within Specific Genetic Syndrome Based on Empirical Studies		
	DS	WMS	FXS[a]
Prelinguistic			
Delayed babbling	High	High	NR
Poor requesting gesture use	High	High	NR
Poor joint attention gesture use	Low	High	NR
Receptive/expressive profile			
Delayed expressive skills (early childhood)	High	High	High
Delayed expressive skills (middle childhood and beyond)	High	Low	High
Delayed receptive skills (early childhood)	High	High	High
Delayed receptive skills (middle childhood and later)	Low	Low	Low
Develop into a profile of stronger receptive than expressive language skills	High	Low	High
Speech			
Intelligibility issues	High	Low	High
Dyspraxia	Moderate	Low	High
Structural language			
Morpho-syntax use problems	High	Low	High
Receptive morpho-syntactic difficulties	High	Low	Low
Pragmatic Language			
Problems with inhibition	Low	High	High
Repetitive speech	Low	High	High
Tangential speech	Low	High	High

NR not sufficiently researched

[a]Children with comorbid autism appear qualitatively distinct to those without significant autism symptoms; these estimates refer to the general population of children with fragile X syndrome, without significant autism symptomology. Children with comorbid conditions tend to demonstrate more severe impairments across all areas of language and communication (Philofsky et al., 2004).

Table 2

Implications of Probable Strengths for Intervention by Genetic Syndrome

Communication/Language Feature That Is a Strength	Estimated Probability of Occurrence As a Strength Within Specific Genetic Syndrome		
	DS	WMS	FXS
Prelinguistic			
Good nonverbal requesting	Low	Low	NR
Good nonverbal joint attention	High	Low	NR
Speech			
Clearly intelligible	Low	High	Low
Structural language			
Expressive language strengths	Low	High	Low
Receptive language strengths	High	Low	High
Good expressive syntax	Low	High	Low
MA-appropriate receptive syntax	Low	High	High
Pragmatic language			
Shares positive affect	High	High	Moderate
Conversational abilities	High	Moderate	Moderate

NR. not sufficiently researched.