The Emerging Down Syndrome Behavioral Phenotype in Early Childhood Implications for Practice

Deborah J. Fidler, PhD

Previous studies have reported a specific behavioral phenotype, or a distinct profile of behavioral outcomes, associated with Down syndrome. Until recently, however, there has been little attention given to how this behavioral profile emerges and develops over time. It is argued here that some aspects of the Down syndrome behavioral phenotype are already emerging in infants and toddlers, including emerging relative strengths in some aspects of visual processing, receptive language and nonverbal social functioning, and relative weaknesses in gross motor skills and expressive language skills. Research on the early developmental trajectory associated with Down syndrome (and other genetic disorders) is important because it can help researchers and practitioners formulate interventions that are time-sensitive, and that prevent or offset potential future negative outcomes. This article reviews evidence for the emerging Down syndrome behavioral phenotype in infants, toddlers, and preschoolers. This is followed by a discussion of intervention approaches that specifically target this developing profile, with a focus on language, preliteracy skills, and personality motivation. Key words: behavioral phenotypes, Down syndrome, early intervention

Down syndrome is the most common genetic (chromosomal) mental retardation syndrome, occurring in from 1 in 700 to 1 in 1000 live births (Hassold & Jacobs, 1984; Stoll, Alembik, Dott, & Roth, 1990). In 95% of cases, Down syndrome is caused by an extra chromosome 21 (trisomy 21). Common physical features associated with Down syndrome are a distinctive craniofacial structure and health-related issues like congenital heart disease, middle ear disease, and immune and endocrine system abnormalities (Pueschel & Pueschel, 1992).

Over the past few decades, research has begun to converge on a specific behavioral phenotype, or a distinct profile of behavioral outcomes, associated with Down syndrome as well. According to recent studies, the Down syndrome behavioral phenotype includes relative strengths in some aspects of visuospatial processing (Jarrold & Baddeley, 1997; Jarrold, Baddeley, & Hewes, 1999; Klein & Mervis, 1999; Wang & Bellugi, 1994), and social functioning (Gibbs & Thorpe, 1983; Rodgers, 1987; Wishart & Johnston, 1990), as well as relative deficits in verbal processing (Byrne, Buckley, MacDonald, & Bird, 1995; Hesketh & Chapman, 1998; Jarrold et al., 1999; Laws, 1998) and some aspects of motor functioning (Chen & Woolley, 1978; Dunst, 1988; Fidler, Hepburn, Mankin, & Rogers, in press; Jobling, 1998; Mon-Williams et al., 2001). Language has been described as a “major area of deficit” in Down syndrome...
Early Development in Down Syndrome

(Sigman & Ruskin, 1999), with particular difficulties manifested in expressive language (Miller & Leddy, 1999). In addition, individuals with Down syndrome have been described as showing a distinct personality motivation profile (Pitcairn & Wishart, 1994).

Researchers often acknowledge two important issues when studying behavioral phenotypes (Dykens & Hodapp, 2001). First, they acknowledge that behavioral phenotypes are probabilistic. As such, groups with a certain syndrome are more likely to show one or more "characteristic" behaviors than other individuals with mental retardation, but not every child with a specific syndrome necessarily shows all etiology-specific behaviors (Dykens, 1995; Hodapp, 1997). Additionally, researchers note that some syndromes share certain behavioral outcomes with other genetic disorders, so outcomes are often not specific to a particular syndrome. In several genetic disorders of mental retardation (eg, fragile X syndrome, Williams syndrome), for example, many children show hyperactivity or attention problems (Hodapp, 1997).

DEVELOPMENTAL CONSIDERATIONS

Amidst the recent advances in behavioral phenotype research (Dykens & Hodapp, 2001), researchers have also begun to argue for the importance of understanding how phenotypes develop and change over time (Karmiloff-Smith, 1997). Rather than considering outcomes as preserved or damaged modules that are wholly intact or impaired uniformly throughout development, Karmiloff-Smith (1998) argues that "tiny variations in the initial state" can become magnified throughout development into domains of relative strength and weakness. Early development may be a crucial window of opportunity for intervention, as these "tiny variations" have not yet snowballed into impairments in whole domains of processing. Studies to date that have taken a developmental approach to behavioral phenotypes have shown that areas of purported relative strength at one stage of development (middle childhood or adolescence) may not have been relatively stronger at other stages of development (early childhood; Paterson, Brown, Gsoedl, Johnson, & Karmiloff-Smith, 1999). There may be crucial windows of opportunity in early development to target areas that pose potential problems to children with Down syndrome before they become pronounced areas of weakness. Thus, understanding how the Down syndrome behavioral phenotype emerges over the first few years of early development may help shape effective, time-sensitive intervention for young children with Down syndrome and their families.

EARLY EMERGENCE OF THE DOWN SYNDROME BEHAVIORAL PHENOTYPE

Compared to other genetic disorders, early development in Down syndrome has received a good deal of research attention. Development in infancy and toddlerhood has rarely been studied in other genetic disorders such as Prader-Willi syndrome, Williams syndrome, or Smith-Magenis syndrome. Even in those few existing studies on early development in other syndromes, the focus is primarily on issues such as early feeding in infancy (Morris, Demsey, Leonad, Dilts, & Blackburn, 1988), rather than on the development of various aspects of cognitive-linguistic or social-emotional functioning.

The wealth of research on early development in Down syndrome may be attributed to the higher incidence of Down syndrome than that of other genetic syndromes, as well as technological advances enabling early identification of Down syndrome. Since the late 1960s, it has been possible to screen pregnant women for Down syndrome via amniocentesis and karyotyping of fetal cells. In addition, in most neonatal units, diagnostic testing is standard procedure for any newborns showing the cardiovascular, craniofacial, or other physical features associated with Down
syndrome. This stands in contrast with the delay of diagnosis often seen in other genetic disorders, for example, Williams syndrome (Huang, Sadler, O’Riordan, & Robin, 2002; Morris et al., 1988).

Thus, early identification has facilitated the description of early social-emotional functioning, cognitive-linguistic development, personality motivation, and motoric functioning in young children with Down syndrome over the first few years of life. This research can be seen as a description of the early initial states of an emerging behavioral phenotype and can help shed light on how the specific outcomes in genetic disorders change and become more pronounced throughout development.

The following section discusses research on different domains of development, including cognitive, linguistic, motor, social-emotional, and motivational functioning. For each domain of development, functioning in older children, adolescents, and young adults with Down syndrome is discussed. Then, research on related findings in early development—or what could be considered the “developmental precursors” to these later outcomes—is discussed.

Cognitive functioning

Children, adolescents, and young adults

Research on the cognitive phenotype in adolescents and adults with Down syndrome has most recently focused on deficits in verbal working memory, and on how they relate to poor expressive language and learning outcomes (Byrne et al., 1995; Hesketh & Chapman, 1998; Jarrold, Baddeley, & Phillips, 2002; Laws, 1998). In addition, studies have found relative strengths in visuospatial processing in this population, and many individuals with Down syndrome have a profile of stronger visuospatial than verbal processing skills (Jarrold et al., 1999; Klein & Mervis, 1999; Wang & Bellugi, 1994). Amidst these relative strengths in visuospatial processing, there is preliminary evidence that some aspects of visuospatial processing are stronger than others in older children and young adults with Down syndrome (Fidler, 2005). In particular, visual memory visual-motor integration, and especially visual imitation seem to be areas of relative strength within visuospatial processing, whereas spatial memory and visuoconstructive tasks seem to be areas of relative weakness (Fidler, 2005).

Early developmental precursors

Can evidence of this cognitive profile already be found in early development? Later deficits in auditory processing could be linked to atypical auditory brain-stem responses in infants with Down syndrome in the first year of life (Folsom, Widen, & Wilson, 1983). In addition, the high incidence of congenital anomalies of the ear in this population—otitis media, for example—has been linked to deficits in auditory processing in early development as well (Downs & Balkany, 1988). But a distinction must be made between auditory perception and short-term/working memory for auditorily presented information, as Jarrold et al. (2002) have identified a short-term memory deficit for auditorily presented verbal information in older individuals that cannot be attributed to sensory deficits. It may be that poor vocal imitation in infants with Down syndrome is linked to later deficits in verbal working memory in this population, but this connection should be explored further (Mahoney, Glover, & Finger, 1981; Rondal, 1980). It may also be important to consider that precursors to deficits in verbal working memory and related cognitive skills may not be present in early childhood and may emerge later in development.

Evidence of strengths in visual processing in early development in Down syndrome can be found in studies of infant visual recognition memory, where infants with Down syndrome show similar event-related brain potential morphology, visual attention, and visual fixation to typically developing infants (Karrer, Karrer, Bloom, Chaney, & Davis, 1998; Karrer, Wojtascek, & Davis, 1995). Infants with Down syndrome have even shown evidence of faster
information processing than have typical in-
fo
fers on some components of visual mem-
ory (Karrer et al., 1995). In line with findings
of strong visual imitation skills in older chil-
dren, there are also reports of early visual (not
vocal) imitative competence in infants with
Down syndrome, similar to the performance
of typically developing infants (Heiman &
Ulstadius, 1999).

However, not all areas of visuospatial func-
tioning are relatively strong in young children
with Down syndrome, a finding that could
be associated with ocular abnormalities com-
monly found in this population (Niva, 1988;
Woodhouse et al., 1996). Gunn, Berry, and
Andrews (1982) report that 6-month-old in-
fants with Down syndrome show delays in vi-
sual exploration in play situations with their
mothers (Gunn et al., 1982). Other reports de-
scribe impaired visual attention performance
on a habituation task in infants with Down
syndrome (Miranda & Fantz, 1973), and de-
lays in various aspects of eye contact in infants
with Down syndrome, including the func-
tional use of eye contact to explore the en-
vironment in a parent-child interactive set-
ting (Berger & Cunningham, 1983). These
early development findings also suggest that
in both infancy and later development, mix-
tures of strengths and weaknesses can be
found within this area of functioning.

Language, speech, and communication
Children, adolescents, and young adults

Many children with Down syndrome have
severe language delays (Sigman & Ruskin,
1999). Part of the Down syndrome lan-
guage phenotype includes pronounced im-
pairments in expressive language relative to
receptive language, including large deficits
in vocabulary size relative to mental age
(Chapman, 1999; Fabretti, Pizzuto, Vicari, &
Voterra, 1997). In terms of receptive lan-
guage, whereas receptive vocabulary is MA
appropriate in later childhood and adoles-
cence, comprehension of syntax lags behind
(Abbeduto et al., 2003; Chapman, Schwartz,
& Kay-Raining Bird, 1991). Individuals with
Down syndrome also show particular deficits
in the development of grammar, and many
adults with Down syndrome do not progress
beyond the early stages of morphological
and syntactic development (Fowler, 1990). In
terms of speech, Miller and Leddy (1999) re-
port that articulation and speech intelligibil-
ity is a major challenge for many individuals
with Down syndrome as well. However, de-
spite deficits in language and speech, older in-
dividuals with Down syndrome show relative
strengths in nonverbal communication (Miller
& Leddy, 1999).

Early developmental precursors

A similar profile of deficits in language
and speech development, but strengths in
communicative competence, can already be
found in early development of children with
Down syndrome. In terms of speech and ex-
pressive language, atypical vocalizing is al-
ready evident in infants with Down syndrome
from 2 to 12 months, who produce atypical
prelinguistic phrases compared to those pro-
duced by typically developing infants (Lynch,
Oller, Steffens, & Buder, 1995). In the first 6
months of life, infants with Down syndrome
also produce more non–speech-like sounds
than speech-like sounds, which may nega-
tively impact the later development of nor-
mal vocal behavior (Legerstee, Bowman, &
Fels, 1992). Additionally, delays in age of on-
set of canonical babbling have been found in
infants with Down syndrome (Lynch, Oller,
Steffens, Levine, et al., 1995). In contrast
with the relatively strong visual imitative com-
petence in young children with Down syn-
drome, as mentioned earlier, vocal imitation
seems to be greatly impaired (Mahoney et al.,
1981; Rondal, 1980). Decreased vocal imi-
tation in Down syndrome has been shown
to be associated with lower expressive and
receptive language skills (Mahoney et al.,

Nevertheless, other aspects of prelinguis-
tic vocal development seem to be on par
with typically developing infants, including the amount of vocalization produced, developmental timetable of vocalizations, and characteristics of consonants and vowels produced during babbling (Oller & Seibert, 1988; Smith & Oller, 1981; Smith & Stoel-Gammon, 1996; Steffens, Oller, Lynch, & Urbano, 1992).

One of the most important studies of early speech and language functioning in Down syndrome has demonstrated that the majority (64%) of children with Down syndrome aged 0 to 5 years fit a profile of receptive language that is mental age appropriate while expressive language lags behind (Miller, 1999). In addition, this study found that over time, the number of children who fit this profile increased to 72%, suggesting that some children may be “growing into” this profile as they develop. Miller (1999) reported that there seemed to be 2 distinct groups of young children with Down syndrome—one group that showed impairment from the onset of first words and a second group that acquired vocabulary but showed expressive language lags when language learning advanced to more difficult skills, such as the combining of words into phrases.

In terms of early communicative competence, some areas seem to be intact whereas others are impaired. Young children with Down syndrome show MA-appropriate levels of nonverbal joint attention (Fidler, Philofsky, Hepburn, & Rogers, in press; Mundy, Kasari, Sigman, & Ruskin, 1995; Mundy, Sigman, Kasari, & Yirmiya, 1988; Wetherby, Yonclas, & Bryan, 1989). In addition, despite deficits in expressive language development, the early use of gestures in children with Down syndrome seems to be intact. One study found a “gesture advantage” in young children with Down syndrome compared with controls matched for word comprehension (Caselli et al., 1998). Another study found that despite a smaller repertoire of gestures, no differences could be found for overall usage of gestures between young children with Down syndrome and a comparison group of language-age-matched children (Iverson, Longobardi, & Caselli, 2003). Yet, even in the context of these communicative strengths, other aspects of early communicative competence seem to be impaired. In particular, young children with Down syndrome show deficits in nonverbal requesting behaviors (Mundy et al., 1988, 1995; Fidler et al., in press; Wetherby et al., 1989).

Social-emotional functioning

Children, adolescents, and young adults

Although deficits in speech, language, and communication are common, many older individuals with Down syndrome nonetheless show relative strengths in social functioning. For example, individuals with Down syndrome may show relative competence in forming relationships with others. Freeman and Kasari (2002) found that the majority of children with Down syndrome in their sample showed relationships with peers that met criteria for true friendships—reciprocal nomination in the friendship dyad, convergence between parental and child nomination, and at least 6-month stability of friendship in that dyad. Children with Down syndrome have also been shown to be more empathic than other children with developmental disabilities, showing more prosocial responses in a simulated distress situation (Kasari, Freeman, & Bass, 2005). Children with Down syndrome may also “overuse” their social skills to compensate for other weaker domains of functioning (Freeman & Kasari, 2002). In an impossible task study, for example, young children with Down syndrome showed looks to the experimenter and more “party pieces,” or charming off-task behaviors that engaged the experimenter socially (Pitcairn & Wishart, 1994).

Children with Down syndrome may also send more positive emotional signals than may other children with mental retardation. In one study, 5- to 12-year-olds with Down syndrome smiled more frequently than children with other mental retardation syndromes, although this finding of increased smile frequency changed as individuals with Down
syndrome approached adulthood (Fidler & Barrett, in press).

**Early developmental precursors**

Some aspects of this socioemotional profile are already present in infancy. Visual imitative competence in infancy has been described as evidence of “an innate social competence” (Heiman & Ulstadius, 1999). In terms of early looking behavior, Crown, Feldstein, Jasnow, and Beebe (1992) found that infants with Down syndrome look longer at their mothers than typically developing infants even at 4 months of age, a behavior that may promote connections with others. These findings are echoed in a study by Gunn et al. (1982), who found that 6- and 9-month-olds with Down syndrome spent nearly half of their interaction time looking toward their mother, and by Kasari, Freeman, Mundy, and Sigman (1995), who found increased looking behavior at parents during an ambiguous situation. However, in the context of increased looking behavior, Kasari et al. (1995) and Walden, Kneips, and Baxter (1991) found decreased social referencing.

Other evidence of social competence in infancy can be found in increased melodic sounds, vocalic sounds, and emotional sounds in 4-month-old infants with Down syndrome when interacting with people rather than with objects (Legerstee, Bowman, & Fels, 1992). Evidence of continued social competence seems to continue throughout toddlerhood and pre-school-aged children with Down syndrome. At 17.5 months, infants with Down syndrome show responses to maternal requests that are similar to those responses made by typically developing infants (Bressanutti, Sachs, & Mahoney, 1992). In a modified strange situation, 24-month-olds with Down syndrome show distress when their mothers are absent, with increased crying and noncrying distress and increased looks at the door—behavior described as similar to that observed in typically developing children (Berry, Gunn, & Andrews, 1980; see also Vaughn et al., 1994). Toddlers and preschoolers with Down syndrome also display relative strengths in certain types of nonverbal social interaction including more play acts, turn taking, invitations, and object shows compared to typically developing children (Mundy et al., 1988; Sigman & Ruskin, 1999).

One aspect of social-emotional functioning in Down syndrome that may be of particular interest is the ability to communicate positive affect through frequent emotion displays such as smiles. Initial studies of emotion communication in infants with Down syndrome reported muted emotion displays and less emotional lability than typically developing infants (Berger & Cunningham, 1986; Buckhalt, Rutherford, & Goldberg, 1978; Cicchetti & Sroufe, 1978; Emde & Brown, 1978; Rothbart & Hanson, 1980). Later studies, however, that were conducted with more objective coding systems (ie, MAX and FACS), suggested that although there may be more frequent low-intensity smiling in young children with Down syndrome, this may be in addition to frequent high-intensity smiling, such that there may actually be more smiling and increased emotional lability in young children with Down syndrome than in typically developing children (Kasari, Mundy, Yirmiya, & Sigman, 1990; Kneips, Walder, & Baxter, 1994). These findings are in line with the finding of increased smiling behavior in older children and preadolescents with Down syndrome (Fidler & Barrett, in press).

**Motor functioning**

**Overview**

Another aspect of the Down syndrome behavioral phenotype described in older individuals involves difficulties with motor skills and motor planning (Jobling, 1998; Mon-Williams et al., 2001). Jobling (1998) reported that 10- to 16-year-old children with Down syndrome have specific motor impairments, including difficulty with precise movements of limbs (eg, stepping over a stick while on a balance beam) and fingers (eg, pivoting thumb and index finger) as well as gross motor tasks such as sit-ups and push-ups. Similar relative
weaker weaknesses have been demonstrated in motor planning or praxis (Mon-Williams et al., 2001). However, in other domains such as running speed and agility and visual-motor control, Jobling (1998) reports that child performance in Down syndrome can be at CA levels.

**Developmental precursors**

Most infants and toddlers with Down syndrome show extreme motor delays relative to CA-matched typically developing infants, moving through stages of early motor development more slowly and exhibiting more within-group variability than typically developing infants (Chen & Woolley, 1978; Dunst, 1988). Abnormal movement patterns, hypotonia, and hyperflexibility are common in this population (Harris & Shea, 1991). In addition, delays in the emergence and termination of reflexes are prevalent in early motor development in this population (Block, 1991; Harris & Shea, 1991). These atypical outcomes seem to become more evident toward the end of the first year of life (Dunst, 1988; Henderson, 1985).

Dmitriev (2001) describes 4 different types of infants with Down syndrome on the basis of muscle tone and motor functioning. Type 1 (15%-25%) babies have good muscle tone and show milestones like head control, bearing weight on feet with support and lifting the torso on extended arms by 4 months. Types 2 and 3 (50%-60%) babies show a discrepancy between upper and lower body motor functioning. Type 2 infants have strong upper back, neck, shoulders, and arms, but are unable to bear weight on their legs as other infants are able to do, whereas Type 3 infants have strong legs and lower torso, but weaker upper torso, neck, head, shoulders, and arms. Finally Type 4 babies (15%-25%) are weak all over, with flaccid arms and legs, and often have accompanying cardiovascular challenges. These groupings suggest that although there is variability within Down syndrome motor functioning in infancy, the majority of infants do face serious motor challenges that warrant intervention.

In terms of motor planning, infants with Down syndrome show more deviation from straight lines and changes in plane of motion during reaching behavior than typically developing infants, evidence of a deficit in the organization of reaching movement (Cadoret & Beuter, 1994). Fidler et al. (in press) explored whether motor delays in Down syndrome include deficits in motor planning, and whether motor planning is related to adaptive functioning in this population. Toddlers with Down syndrome in this study performed significantly worse on a battery of motor planning tasks, including reaching into a jar to grasp a nerf ball, and stringing beads, a finding specific to Down syndrome and not attributable to disability status in general. Furthermore, partial correlations demonstrated a strong association between overall adaptive motor functioning and motor planning performance in both disability groups even when age was partialed out. Similar associations were found between motor planning and daily living skills, suggesting that motor planning deficits in Down syndrome may also be associated with day-to-day adaptation, and not only motor-related adaptive skills.

**Personality motivation**

**Overview**

Individuals with Down syndrome have frequently been described as having charming personalities, often in accordance with a positive Down syndrome personality stereotype (Gibbs & Thorpe, 1983; Rodgers, 1987; Wishart & Johnston, 1990). Older children and young adults with Down syndrome are described as of primarily positive mood and predictable in their behavior, but less active and persistent and more distractible than other children as well (Gunn & Cuskelly, 1991). In one study, over 50% of 11-year-old children with Down syndrome were described as “affectionate,” “lovable,” “nice,” and “getting on well with other people,” and many children were also described as “cheerful,” “generous,” and “fun” (Carr, 1995).
Alongside these positive perceptions, many individuals with Down syndrome are also described as showing inconsistency in motivational orientation. Many children with Down syndrome also show lower levels of task persistence and higher levels of off-task behavior tasks, interfering with task completion (Landry & Chapieski, 1990; Pitcairn & Wishart, 1994; Ruskin, Kasari, Mundy, & Sigman, 1994; Vlachou & Ferrell, 2000). These individuals are sometimes described as stubborn or strong willed, traits that may contribute to inconsistent performance on tasks due to task refusal (Carr, 1995; Gibson, 1978).

**Developmental precursors**

Several studies report no significant temperament differences between infants with Down syndrome and typical infants in early infancy, at 2 months (Ohr & Fagen, 1994) and later at 12 to 36 months (Vaughn, Contreras, & Seifer, 1994). Other studies, however, report that young children with Down syndrome ($M = 30$ months) are rated as of more positive mood, more rhythmic, and less intense than CA-matched children (Gunn & Berry, 1985). These findings echo the findings of increased predictability, increased positive mood, and decreased persistence in older children with Down syndrome. However, nearly one third of children with Down syndrome in Gunn and Berry’s (1985) study showed signs of difficult temperament as well, a possible precursor to stubbornness and other behavior problems.

The developmental precursors of task persistence findings may also be identifiable in early development. Young children with Down syndrome often show inconsistent performance on assessment measures from time-point to timepoint (Morss, 1983; Wishart & Duffy, 1990). Wishart and Duffy (1990) found that children with Down syndrome aged 6 months to 4 years show highly inconsistent performances on the same testing battery across sessions 2 weeks apart. The authors suggested that this inconsistency is the result of motivational issues, often the result of refusal to engage fully in tasks at either time-point (Wishart & Duffy, 1990).

Morss (1983) has similarly reported that infants with Down syndrome repeat their successes on tasks less often than mental, age-matched, typically developing children. Hasan and Messer (1997) found that children with Down syndrome in their sample showed more stability in performance on executive function/object permanence and other cognitive tasks although 20% of their sample did show some regressions. Researchers suggest that these regressions often result from a child’s unwillingness to engage in a task, suggesting that motivation may be an important factor for assessing development in Down syndrome (Pitcairn & Wishart, 1994; Wishart & Duffy, 1990).

According to Wishart (1993), “[F]rom a very early age, it would appear that the Down syndrome children are avoiding opportunities for learning new skills, making poor use of skills that are acquired, and failing to consolidate skills into their repertoires.” Along these lines, increased level of help elicitation has also been found in Down syndrome and may relate to persistence issues as well. In the motor planning study described above (Fidler, Hepburn, Mankin, & Rogers, in press), it was also found that toddlers with Down syndrome elicited significantly more help on the object retrieval task than did children in both comparison groups, a finding also reported in other studies (Freeman & Kasari, 2002).

**Overall profile of early development in Down syndrome**

In addition to exploring the early development of various aspects of functioning in Down syndrome, it may also be important to explore “cross-domain relations,” or how different domains of functioning develop together (Hodapp, 1996). Are pronounced dissociations between areas of strength and weakness already observable in early childhood? In a recent study, Fidler et al. (in press) described the performances of young children with Down syndrome on measures of visual processing, expressive language, receptive language, fine and gross motor functioning, and social functioning on the Mullen...
Toddlers with Down syndrome in this study did show relative strengths in the areas of visual processing and receptive language, and relative weaknesses in gross motor skills and expressive language, although it is important to note that these dissociations were small in magnitude. In terms of parent-reported skills in adaptive behavior in real-life situations, the children with Down syndrome in this study showed relative strengths in socialization and relative weaknesses in communication and motor skills. This is evidence that phenotypic pattern of strengths and weaknesses associated with Down syndrome is emerging by the age of 2, with between-group differences in sociability, and within-group patterns of relative strengths and weaknesses that foreshadow the phenotype described in studies of older persons.

It is notable that the dissociations observed within the individuals with Down syndrome were significant, but also relatively small at these early developmental ages. Even in the significant difference between expressive and receptive language, differences averaged only 2.5 months in age-equivalent scores. In other studies with older children with Down syndrome, dissociations between domains of functioning can be much larger. This does not minimize the rapid changes that take place over several months in early development. But the relatively small dissociation is also notable for intervention purposes—because areas of strength and weakness are less pronounced early on, it may be possible to reduce these dissociations and set areas of potential weakness on more optimal pathways.

With this understanding of the early emergence of the Down syndrome behavioral phenotype in infants and toddlers, it may be possible to shape intervention that is sensitive not only to the current functioning level of the child but also to the developmental trajectory associated with their genetic disorder. Rather than waiting for a dissociated pattern to take its full form, interventions can focus on preventing these dissociations from taking place.

USING BEHAVIORAL PHENOTYPE RESEARCH TO INFORM EARLY INTERVENTION IN DOWN SYNDROME

One of the most interesting questions that arise from behavioral phenotype research concerns the influences of child transactional history on the developing behavioral phenotype. Is it possible to help children with Down syndrome follow more optimal development pathways? This section will explore approaches that show promise—intervention approaches that are informed by behavioral phenotype research.

Amidst the many proposed theoretical approaches to intervention in Down syndrome, researchers have introduced yet another approach—focusing on behavioral phenotype research (Hodapp & DesJardin, 2003; Hodapp & Fidler, 1999). This approach argues that education and intervention may be more effective when it specifically targets the developmental trajectory associated with a particular syndrome. The behavioral phenotype approach is housed within the larger movement of developmental interventions, where programming decisions are informed by developmental theory (see Spiker, 1990, for a review).

The importance of time sensitivity and early implementation in intervention has also been demonstrated in this population. In one study, a 2-month delay of treatment for young children with Down syndrome was associated with lower gross motor, fine motor, language, and social outcomes at 18 months (Sanz & Menendez, 1995). In another study, infants who received language intervention beginning as newborns showed more optimal outcomes than did infants who started the intervention at 90 and 180 days of age (Sanz & Balana, 2002).

Yet, the efficacy of intervention in Down syndrome and other groups remains in
question (for reviews, see Gibson & Fields, 1984; Gunn & Berry, 1989; Guralnick, 1996; Nilholm, 1996). The implementation of just any intervention is not sufficient for improving developmental outcomes (Crombie & Gunn, 1998; Gibson & Fields, 1984). In addition, some interventions that have become popular at different points have been ineffective. For example, popular high-dosage multivitamin and mineral supplements that have been administered to infants and children with Down syndrome aged 7.5–63 months have been shown to be associated with decreased, rather than increased, developmental progress, according to one study (Bidder, Gray, Newcombe, & Evans, 1989). These high-dosage multivitamins and supplements may also be associated with unpleasant side effects as well. Nevertheless, parents report improvements in child appearance and skin tone with these products, and some parents report that they would recommend the vitamin therapy to other parents of children with Down syndrome (Bidder et al., 1989).

With the increased prevalence of alternative and unconventional therapies aimed at parents of children with disabilities and their children, there is a strong need for interventions that are rooted in good science. At this point in time, interventions that are informed by behavioral phenotype research have not been tested in the literature. Testing the tenability of such an approach will need to involve scientific rigor and the high standards found in other types of treatment trials (Kasari, 2002). Yet, there is promise in this approach that it is rooted in good science, and it is in line with recommendations that educational programs target “the specific learning abilities and disabilities of Down syndrome individuals” (Nadel, 1996). A preliminary sampling of intervention ideas that are informed by behavioral phenotype research is described in the following section.

Cognitive-linguistic functioning and intervention

If individuals with Down syndrome do show an advantage for processing visuospatial, rather than verbal information, might this information be used to improve developmental outcomes in this population? Several suggestions have been made to this effect (Byrne et al., 1995; Chapman, 1995; Gibson, 1991). Pueschel, Gallagher, Zartler, and Pezzullo (1987) noted that “[t]eaching strategies should capitalize on Down syndrome children’s strengths and should focus on visual-vocal and visual-motor processing modalities in remediation” (p. 35). They also note that “increasing emphasis on auditory teaching strategies may lead to frustration in the child and may impede academic process” (p. 35). Indeed, a recent study demonstrates that children with Down syndrome respond better to scaffolding that involves both speech and gestures (visual) than to scaffolds that involve only speech (Wang, Bernas, & Eberhard, 2001).

Yet amidst the many recommendations for an increased attention on visual processing in Down syndrome, there have been relatively few efforts to utilize this processing mode to improve outcomes. According to Nadel (1996), “there has been scant application of knowledge about the specific learning abilities and disabilities of Down syndrome individuals to the development of these programs” (p. 22).

One exception is the movement to emphasize early reading in young children with Down syndrome (Buckley, Bird, & Byrne, 1996; Oelwein, 1995; Oelwein, Fewell, & Pruess, 1985). Buckley and colleagues suggest that it is possible to improve language and memory functioning by establishing early sight vocabularies in children with Down syndrome (Buckley et al., 1996). They argue that [T]he benefits of learning to read go beyond simply acquiring a functionally useful level of reading and writing skill . . . reading can develop speech and language skills, auditory perceptual skills and working memory function; all areas where children with Down syndrome usually display difficulties. (p. 269)

Early sight vocabularies for children with Down syndrome capitalize on their strengths.
in visual memory to recognize and identify words, making logographic reading possible at young ages. Buckley et al. (1996) cite case studies of children with Down syndrome who are 2 and 3 years old and who have greatly benefited from the establishment of sight vocabularies, findings that have been echoed by parents as well (eg, Carter, 1985; Duffen, 1976).

Another group advocating the use of early sight vocabularies is team at the University of Washington Model Preschool Program for Children with Down Syndrome and Other Developmental Delays (Oelwein, 1988). Acknowledging that most preschoolers do not receive formal reading instruction, Oelwein described the decision to teach reading to preschoolers with Down syndrome as one that provided a solution for children who “had very well-developed visual discrimination skills, but virtually no spoken language.” This approach advocates scheduling 5 to 7 minutes of reading instruction during short, individualized sessions 2 to 4 days per week. Data collected on children in this program suggest that children with Down syndrome can develop sight vocabularies at all levels of IQ, and that reading level is highly related to receptive language scores—not IQ. These suggestions are well justified and target areas of distinct strength in the developing Down syndrome behavioral phenotype.

However, more evidence is needed to substantiate the claim that reading can impact other areas of development (Kemp, 1996; Lorenz, Sloper, & Cunningham, 1985).

**Language outcomes**

More direct routes to improving language outcomes can also be informed by behavioral phenotype research in Down syndrome as well. Miller (1999) argues that it is unnecessary for—and may be detrimental to—children with Down syndrome to wait for almost inevitable deficits in expressive language to become apparent and then documented. He argues that linguistic phenotype research in Down syndrome demonstrates the inevitability of expressive language deficits, and as such, a diagnosis of Down syndrome should automatically make a child eligible for speech and language intervention services. Miller (1999) also argues that language intervention should focus on targeting and preventing expressive language impairments in Down syndrome before they become pronounced. Continuous reinforcement for vocalizations in infants between 2 and 8 months has been shown to increase vocalization rates (Poulson, 1988), a promising technique to encourage precursors to expressive language.

In addition, some have suggested that language intervention should promote oral motor functioning in Down syndrome, while facilitating communication regardless of modality (verbal or nonverbal; Miller, 1999). As such, it may be beneficial to target oral dyspraxia early in speech therapy. Parents can be taught to use techniques such as back-chaining, prompt-fading, and social praise as a reward for effort. Other recommendations have been made to target the difficult transition in Down syndrome from babbling to meaningful speech. For example, Stoel-Gammon (2001) recommends that adults offer phonetically contingent responses to prelinguistic vocalizations during infancy. These responses can help an infant understand and start to produce meaningful utterances. Stoel-Gammon (2001) also suggests the use of sound games to facilitate the awareness of sound-meaning relationships in infants and toddlers with Down syndrome. It may also be helpful to provide nonverbal means of communicating in the first years of life (eg, gestures, picture exchange, sign language), in addition to building language skills, as a way of minimizing frustration. These and other recommendations specifically target the developing linguistic phenotype in Down syndrome and may be more effective because they keep an eye toward Down syndrome language outcomes in general.

**Motivation orientation, social functioning, and intervention**

In planning early interventions, caregivers, educators, and therapists must be aware of
the propensity of children with Down syndrome to avoid challenging tasks via social initiation. The development of cognitive, language, and motor skills relies upon frequent practice with supports. Avoidance of these tasks will lead to a broader gap in skills over time and thus significantly inhibit the emergence of adaptive skills.

Help elicitation observed in some studies of children with Down syndrome (Fidler et al., in press) can be interpreted in several ways. On one hand, children may be using their ability to relate to others in ways that help them complete tasks more successfully. This may bode well for individuals who might otherwise not be able to perform certain daily living skills tasks. On the other hand, most new tasks are difficult for children at various points of development, and part of the growth process involves challenging oneself to develop skills in order to overcome obstacles. If individuals with Down syndrome are eliciting help this early in development, they may be missing out on important challenging early experiences that may promote their growth.

To address the motivational issues in Down syndrome early development, errorless learning techniques may be important ways to prevent task abandonment in Down syndrome (Fidler, in press; Oelwein, 1995). To maximize task persistence during interventions, practitioners and parents may opt for alternate activities by skill domain (ie, social, expressive language, receptive language, motor), beginning and ending with domains of strength (eg, social, receptive language). Intervention approaches can also more readily target areas of deficit by imbedding them in tasks that involve areas of strength. For example, to increase practice of motor foundation skills, it may be useful to imbed motor tasks in play and other social contexts.

Dmitriev (2001) recommends an operant conditioning-grounded approach in Down syndrome that involves rewarding desired behaviors. He suggests that actions that result in success or the attainment of a desired goal—the fun of playing with a new rattler, the feeling of accomplishment and a mother’s praise when a toddler successfully pulls on a pair of socks…quickly teach the child which behaviors guarantee success. (p. 68)

An operant conditioning approach may be particularly helpful for children with Down syndrome, who can be prone to inconsistent performance due to motivational issues (Wishart & Duffy, 1990). A steady flow of positive motivational feedback may serve to continue to motivate children as they proceed through a challenging task, especially given the social orientation of many children with Down syndrome.

In addition, children with Down syndrome should be encouraged to use their social skills in adaptive and appropriate ways. Lloveras and Fornells (1998), for example, recommend symbolic play group approaches that facilitate “the construction of relational competencies which are needed for…social integration and…global satisfactory development” (p. 89).

Motor skills and intervention

Recent findings suggest that various approaches to early motor intervention—approaches that focus on developmental functioning and approaches that focus on functional skills—may have little effect on improving developmental outcomes in Down syndrome and other disability groups (Mahoney, Robinson, & Fewell, 2001). However, these findings may not be generalizable to all intervention studies, particularly because parents were not included in the intervention approaches studied (see discussion below).

Building a stronger motor foundation involves participation in purposeful, relevant activities that incorporate specific components of motor foundation. For example, to improve and maintain appropriate posture and position, a child may work on these skills while sitting at a table engaging in a task. The skill development component of the Denver Model intervention involves targeting initiation actions, imitation of others, hand
development, coordination, and dexterity. This may be especially helpful for young children with Down syndrome who may have difficulty with initiation due to hypotonia and a less persistent temperament, but who show strengths in visual imitation due to social and other factors. In this model, skills are taught using shaping and other prompting techniques, with each skill broken down and chaining procedures are used in multistep sequences (Osaki, Roger, & Hall, 2000). This recommendation is echoed by others for children with Down syndrome (Dmitriev, 2001).

Finally, the compensatory strategies component of the model includes adaptations to the task that allow for independence rather than a dependence on prompting and help elicitation throughout the task.

**General recommendations**

Other more general recommendations may also be effective in Down syndrome early intervention. For example, interventions that are informed by an understanding of the emerging phenotype in Down syndrome may enable practitioners to focus on areas of strength as a “way in” for interventions that target potential areas of weakness. While intervention is typically drawn to the relative weaknesses in an individual’s developmental phenotype (Hodapp & Zigler, 1990), it may also be interesting to consider the emerging phenotype as a reflection of strengths, a compensatory pattern constructed from areas of greatest competency that promotes adaptation and access to preferred people and activities. Working from this framework, intervention may choose to target strengths as strongly as weaknesses, in helping people build a life that highlights their talents and interests. For example, children with Down syndrome may be encouraged early on to pursue tasks that involve potential future strengths like visual processing and visual-motor coordination, as well as their relative strengths in social functioning and forming social relationships. Furthermore, promotion of strengths in targeted ways may facilitate the bootstrapping of weaker skills.

In addition to focusing on the specific child profile, a focus on family ecology and the parent-child may be crucial for successful intervention in Down syndrome and other disability groups (Spiker, 1990). Bronfenbrenner (1974) was among the first to argue that early intervention is most effective if the family is an active agent in implementation. Early motor intervention that includes parental involvement has been shown to have a positive effect on early development in Down syndrome (Torres & Buceta, 1998), while intervention studies that do not involve parents have been shown to be less effective (Mahoney et al., 2001; see Spiker, 1990, for a review). Children may also show better outcomes when parents are trained directly by practitioners, as studies show that parents who are trained directly by clinicians fare better than those given written instructions (Sanz, 1988, 1996).

Thus, the larger movement toward targeting intervention to both the child and the context in which the child develops may play an important role in improving developmental outcomes in Down syndrome as well.

**SUMMARY**

As a part of a larger movement toward studying the behavioral phenotypes associated with different genetic disorders, this article explored the early developmental precursors to the Down syndrome behavioral phenotype. There is evidence that cognitive, linguistic, social, emotional, motivational, and motoric aspects of the Down syndrome behavioral phenotype are already emerging in the earliest years of life. In addition, cross-domain relations observed in older individuals with Down syndrome also seem to be emerging already in toddlers, although findings are less pronounced at this early stage than they are in older children and adults.

In light of this new understanding of development in genetic syndromes, it may now be possible to target domains of development, such as expressive language, before they become areas of pronounced weakness. These
areas can be targeted via time-sensitive interventions that are informed by phenotype research on older individuals with the syndrome. It may also be possible to use areas of relative strength as a “way in” to those areas of weakness, to prevent or offset future delays, as suggested by early reading approaches that seek to impact language and memory outcomes. The behavioral phenotype approach remains unsupported by evidence at this point in time and warrants rigorous scientific testing to verify its utility. But this approach is grounded in good science and may prove to be the next shift in how services are delivered to young children with Down syndrome and other genetic disorders.

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