The Down Syndrome Behavioral Phenotype: Implications for Practice and Research in Occupational Therapy

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ABSTRACT. Down syndrome (DS) is the most common chromosomal cause of intellectual disability. The genetic causes of DS are associated with characteristic outcomes, such as relative strengths in visual-spatial skills and relative challenges in motor planning. This profile of outcomes, called the DS behavioral phenotype, may be a critical tool for intervention planning and research in this population. In this article, aspects of the DS behavioral phenotype potentially relevant to occupational therapy practice are reviewed. Implications and challenges for etiology-informed research and practice are discussed.

KEYWORDS. Behavioral phenotype, Down syndrome, genetics, intellectual disability

Intellectual disability (formerly “mental retardation”) is defined by both low intelligence quotient scores and deficits in adaptive behavior (American Association on Mental Retardation, 2002). Throughout much of the twentieth century, both practitioners and researchers across disciplines considered individuals with intellectual disability as a homogeneous group. However, advances in genetics and development have improved our understanding of intellectual disabilities and atypical development across the lifespan (Dykens, Hodapp, & Finucane, 2000; Hodapp & Fidler, 1999). Presently, over 1,200 genetic disorders associated with intellectual disability have been identified (Moser, 2004). We now know that intellectual disability associated with genetic disorders is not merely the result of slowed cognitive development (Vicari, 2006). In fact, research indicates that in individuals with various genetic syndromes, atypical constraints are placed on neurodevelopment, leading to some areas that may appear to be severely affected and other areas that are less so (Hodapp, 2004; Karmiloff-Smith, 1998). Therefore, individuals with genetically

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based intellectual disabilities are predisposed to distinct developmental outcomes (Dykens, 1995; Hodapp & Dykens, 1994; see Dykens et al., 2000 for a review).

The interdisciplinary field of intellectual and developmental disabilities (DDs) keenly understands the need for research evidence. In Down syndrome (DS), research describing syndrome-related outcomes has outpaced the intervention evidence to date (Fidler & Nadel, 2007). However, researchers are calling for more etiology-specific intervention evidence, especially given that some researchers have found that intervention designed for individuals with DDs has had a differential effect for those with DS (Yoder & Warren, 2002). While there will always be individual variability in how the DS behavioral phenotype affects the occupations of an individual over the course of a lifespan, we believe that knowing the relative strengths and challenges associated with this genetic syndrome and the current state of inquiry on intervention in this population will be necessary for occupational therapists to maintain the professional currency to develop the most effective intervention plans for their clients.

In the field of occupational therapy, there has been a paucity of discussion regarding behavioral phenotypes or the patterns of developmental strengths and weakness associated with genetic disorders, such as DS. This has been highlighted by Fidler, Hodapp, and Dykens’ (2002) findings that parents of children with various genetic disorders, including DS, reported that the occupational therapists on their child’s team often do not provide etiology-specific information to the team for use in intervention planning.

In an effort to begin to bridge the gap between science and practice, this article reports evidence regarding phenotypic outcomes associated with DS, as well as emerging intervention evidence relevant to occupational therapy practice. Along with an overview of epidemiology and defining the concept of a behavioral phenotype, we review aspects of cognition and information processing, motor planning, social functioning, aging in DS, and participation in occupations. Suggestions are offered for application to etiology-specific research and intervention planning in occupational therapy.

**EPIDEMIOLOGY AND OVERVIEW**

Discovered over 150 years ago (Down, 1866), DS is a multisystem neurogenetic disorder that affects growth, development, and participation in daily activities across the lifespan (Fidler & Nadel, 2007; Krinsky-McHale, Devenny, Kittler, & Silverman, 2008). It is the most common genetic cause of intellectual disability (Sherman, Allen, Bean, & Freeman, 2007). In 95% of cases, DS is caused by an extra chromosome 21 (trisomy 21). However, approximately 1% of individuals with DS have mosaic DS (Connor & Ferguson-Smith, 1997). In mosaic DS, there are two different cell lines in a developing zygote/embryo: one cell line having the trisomy 21 error and one cell line not having that error (Connor & Ferguson-Smith, 1997). Also, a small percentage of cases of DS are caused by translocations between genetic material on chromosome 21 and another chromosome (Connor & Ferguson-Smith, 1997).

A recent study sampling areas across the United States indicates that in each region, the prevalence of DS is increased (Shin et al., 2009). The birth
rate of children with DS increased by 31.1% from 1979 to 2002, and DS prevalence rate during this period was 11.8 in every 10,000 live births (Shin et al., 2009). Because of advances in prenatal testing, an increasing number of parents are aware of their child’s diagnosis prior to birth (Malone et al., 2005). Given the increased prevalence of DS in the past 20 years (Shin et al., 2009) in conjunction with the improved life expectancy of this population (Krinsky-McHale et al., 2008), it is expected that the health and quality of life needs for individuals with DS will continue to be a growing concern for healthcare practitioners.

THE DOWN SYNDROME BEHAVIORAL PHENOTYPE

As defined by Nyhan (1972), a behavioral phenotype describes the patterns of behavioral outcomes associated with the genotype of a genetic disorder. These outcomes can involve a range of developmental areas, including motor functioning, cognition, language, social, and emotional functioning. While many aspects of the DS behavioral phenotype are now relatively well characterized, it is important to note that behavioral phenotypes are probabilistic. Thus, individuals with a DS are more likely to show one or more of these phenotypic behaviors than other individuals with intellectual disability, but not every individual with a DS will necessarily show all aspects of the syndrome’s phenotypic profile (Dykens, 1995; Hodapp, 1997). In this section, findings regarding phenotypic outcomes in DS are reviewed using components of the Occupational Therapy Practice Framework (American Occupational Therapy Association, 2008).

Client Factors: Body Structures

Most individuals with DS are born with characteristic physical features including brachycephaly (abnormally wide head), short neck, palpebral fissures, epicanthal folds, Brushfield spots, flat nasal bridge, dysplastic ear, a high-arched palate, muscular hypotonia, and musculoskeletal hyperflexibility. Short stature, small head circumference, and overweight are also widely observed in this population (Fidler & Daunhauer, in press). Additionally, DS is associated with a distinctive neuroanatomical profile. At birth, the brain development of individuals with DS presents as typical, yet delays in myelination and decreased brain structure volumes are soon notable (see Nadel, 2003, for a review). By adulthood volume reductions are apparent in the hippocampus, prefrontal cortex, and cerebellum (Nadel, 2003). In analyzing these findings, Nadel (2003) raises two issues: (1) he cautions that there is a great deal of individual variability and most individuals with DS fall in the lower end of the distribution for typically developing (TD) individuals and (2) he speculates that these findings based on postmortem assessment of primarily older individuals who may not have benefitted from the early intervention and enrichment promoted nowadays that may affect neural development.

Client Factors: Body Functions

DS is associated with specific medical conditions, including congenital heart disease, middle ear disease, celiac disease, obesity, and metabolic and endocrine dysfunction (for a review see Fidler & Daunhauer, in press; Pueschel & Pueschel, 1992). Additionally, an increasing incidence of individuals diagnosed with DS and comorbid
autism has been reported in the literature (Dykens, 2007; Hepburn, Philofsky, Fidler, & Rogers, 2008; Kent, Evans, Paul, & Sharp, 1999). While once thought to occur at low rates in the DS population (1%–2%, see Dykens, 2007, for a review), currently, 5%–10% of children with DS also meet the criteria for autistic spectrum disorder (DiGuiseppi et al., 2010; Hepburn et al., 2008; Kent et al., 1999; Lowenthal, Paula, Schwartzman, Brunoni, & Mercadante, 2007). Below we address strengths and challenges in performance skills pertinent to occupational therapy practice.

**Performance Skills**

**Cognitive Skills**

Most individuals with DS have a mild to moderate range of intellectual disability, with IQs ranging from 40 to 70 (Hodapp, Evans, & Gray, 1999). Over the past few decades, researchers have also characterized a profile in individuals with DS that involves a challenging learning style (Wishart, 1996; Kasari & Freeman, 2001), as well as difficulty with some aspects of goal-directed behavior (see Fidler, 2006, for a review).

**Learning style and motivation orientation.** Young children with DS have been observed to exhibit less pleasure in toy exploration including touching, banging, manipulating, as well as poorer mastery motivation than their peers matched for both mental and chronological age (Ruskin, Mundy, & Sigman, 1994). When compared with peers matched for mental age (MA), researchers have observed higher levels of off-task behavior in individuals with DS across childhood to young adulthood during play and visual-perceptual tasks (Kasari & Freeman, 2001; Daunhauer, Flabiano, & Fidler, 2009; Pitcairn & Wishart, 1994). Related to these challenges, individuals with DS may develop both positive and negative behaviors in response to challenging tasks. Toddlers with DS have been observed to use both positive behaviors (e.g., “party tricks” meant to charm such as clapping hands or blowing raspberries) and negative behaviors such as quitting a task or crying (Wishart, 1996). Older children have been observed to use relative strengths in social functioning to distract examiners from a difficult task (Pitcairn & Wishart, 1994). Many individuals with DS present with a distinct tendency to be “cognitively avoidant” (Wishart, 1996). According to Wishart (1996), cognitively avoidant behaviors observed in this population include resisting activities that involve new skills, difficulty achieving a high enough degree of mastery to add skills to their repertoire and poor generalization of acquired skills. The downstream effects of this profile cannot be underestimated, as Wishart (1996) notes; this style ultimately leads to an accumulation of missed opportunities for learning and adaptation.

**Goal-directed behavior.** Executive function is an umbrella term that describes the cognitive processes integral to adaptive, goal-directed actions, including working memory, inhibition, and planning (Blair, Zelazo, & Greenberg, 2005; Carlson, 2005; Fletcher, 1996). Researchers are beginning to characterize a behavioral profile in DS that involves specific difficulties with some aspects of executive function (Kogan et al., 2009; Rowe, Lavender, & Turk, 2006).

Challenges in working memory have been well documented in school-aged children and young adults with DS (see Baddeley & Jarrold, 2007, for a review; Jarrold, Baddeley, & Phillips, 2002; Lanfranchi, Cornoldi, & Vianello, 2004). This
impairment seems to be significantly more pronounced with a high amount of verbal information for individuals with DS, as impairments are minimal in the visual-spatial domain (see Nadel, 2003, for a review; Lanfranchi et al., 2004; Visu-Petra, Benga, Tincas, & Miclea, 2007).

Relevant to the practice of occupational therapy is the emerging evidence on planning and inhibition/self-regulation. Planning includes anticipating the steps needed to complete a task or selecting appropriate chains of behavior to successfully complete an activity. For example, when presented with an object retrieval task that involved minimal motor demands, Fidler, Hepburn, Mankin, & Rogers (2005) found that 2- to 3-year-old children with DS chose notably less optimal strategies than MA-matched children with other DDs for obtaining a desired object through the opening of a clear box. Children with DS were significantly more likely to attempt to reach through the top of the box, where there was no opening, than children in both the DD group and the typical comparison group. These planning difficulties were not observed in the DD comparison group, suggesting a challenge specific to the DS profile.

This planning difficulty is also evident in older children with DS. For example, the parents of children with DS, aged 3–10 years, reported that their children had significant difficulties in everyday activities requiring planning when compared with a mental-aged matched standardization population (Lee et al., 2010). Similarly, Kasari and Freeman (2001) found that 5- to 12-year-old children with DS took four times longer to initiate puzzles than the DD comparison group, and took nearly 70% longer to complete them. During aging, declining performance of executive functioning tasks may be an indicator of early-onset Alzheimer’s disease (AD) in this population (Ball, Holland, Treppner, Watson, & Huppert, 2008; Krinsky-McHale et al., 2008).

Studies of emotion regulation, delay of gratification, and response inhibition indicate that inhibition/self-regulation may be a specific area of challenge for individuals with DS (Kogan et al., 2009; Kopp, Krakow, & Johnson, 1983). In one study, 3- to 4-year-old children with DS showed significantly shorter latency to touch a prohibited toy than MA- or language-age-matched children (Kopp et al., 1983). Furthermore, the group with DS in this study generated fewer and less effective strategies for delaying gratification than comparison group children. Kogan and colleagues (2009) found in a sample of young adults to adults (mean age 16 years) that individuals with DS had significantly more difficulty in performing tasks requiring inhibition of an automatic response than those with fragile X syndrome. These studies suggest that individuals with DS may be less successful at controlling automatic responses, though additional empirical investigation in this area is needed.

Visual-Perceptual and Visual-Motor Skills

Visual-perceptual abilities appear to be an area of relative strength for individuals with DS (Jarrold & Baddeley, 1997; Jarrold, Baddeley, & Hewes, 2000; Klein & Mervis, 1999; Wang & Bellugi, 1994). For example, children and adolescents with DS have demonstrated strong short-term memory of visual images (Jarrold & Baddeley, 1997) and perform motor-free visual-perceptual tasks at a level expected for their MA (Wang, Doherty, Rourke, & Bellugi, 1995). However, individuals with DS have been described as having a global visual processing style (Annaz,
Karmiloff-Smith, Johnson, & Thomas, 2009). For example, adults with DS copy models or block designs with good attention to the overall organization, or gestalt, of a model, but poor attention to the internal details in comparison with adults with Williams syndrome (Bellugi, Lichtenberger, Mills, Galaburda, & Korenberg, 1999). Perhaps related to this global processing style, individuals with DS have been found to have difficulty with facial recognition that may involve integrating specific facial features (Annaz et al., 2009; Wang et al., 1995). In fact, Annaz and colleagues (2009) found in a modest-sized sample that included preschoolers through adolescents that the group with DS had as much difficulty recognizing faces as a comparison group with low-functioning autism.

**Motor and Praxis Skills**

Individuals with DS may demonstrate challenges in motor skills across the lifespan. Infants with DS grasp, sit, crawl, stand, walk, climb stairs, and jump at later ages than their TD peers (de Campos, Rocha, & Savelsbergh, 2010; Palisano et al., 2001). In fact, given the delayed onset of motor milestone acquisition in this population, Palisano and colleagues (2001) have researched expected motor milestone curves specifically for individuals with DS so that interventionists can determine if an individual is acquiring motor skills at a rate expected for someone with this genetic syndrome. Dmitriev (2001) has observed four patterns of muscle tone and motor functioning in infants with DS. Type 1 (15%–25%) babies have typical muscle tone and achieve 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, 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.

Despite challenges in motor functioning, as emphasized by Latash (2000), many individuals with DS develop strategies and compensations (such as early walkers using a wide gait) to facilitate mobility. In fact, Palisano and his colleagues (2001) suggested that their motor growth curve findings on children with DS could help therapists decide whether a child’s intervention goals should be focused on motor development or perhaps other areas. School-aged children and teenagers with DS continue to develop motor skills, though at a slower rate than a standardization sample and, of course, with a great deal of individual variability (Jobling, 1999). Jobling (1999) found that from the age of 10 to 16 years, students with DS clustered into one of two groups. The first group demonstrated greater proficiency at running speed and agility, while the second had better visual-motor scores—with poor balance being a consistent challenge across both groups (Jobling, 1999). In fact, Jobling (1999) reported that balance was notable for being significantly less than expected for MA and for being the one area that participants did not improve from ages 10 to 16 years. Later in the lifespan, aging adults with DS have demonstrated deteriorating praxis skills at a mean age of 56.9 years (Dalton, Mehta, Fedor, & Patti, 1999).

Individuals with DS have been described as “clumsy” with challenges in motor planning and coordination (Latash, 2007). They have been reported to have slower
velocity of movement execution and needing more time to initiate a movement than a MA-matched comparison group (Mon-Williams et al., 2001). Individuals with DS also demonstrate suboptimal strategies for joint co-contraction during fast movements and postural demands (for a review see Latash, 2007). Virji-Babul, Kerns, Zhou, Kapur, and Shiffrr (2006) conducted a short series of preliminary experiments to examine the integration of visual-perceptual and motor skills in this population since the two are typically combined in everyday activities. They found that while the group with DS could perceptually discriminate moving people and objects when they were just given the salient points of movement to view (point-to-light displays), they demonstrated difficulty when perception had a complex movement such as differentiating a typical gait from an atypical one. At a neurophysiological level, Virji-Babul, Moiseev, Cheung, Weeks, and Cheyne (2009) may have found preliminary evidence that adults with DS process perceptual-motor information in both the frontal and temporal lobes of their brains with delays and less activation. However, until a comparison with a group of individuals with intellectual disability is made, it will be difficult to determine whether their findings are applicable to DS in specific or intellectual disability in general.

Communication and Social Skills

Individuals with DS often have relatively stronger receptive language and comprehension than expressive capabilities throughout the lifespan (Chapman, 1999; Fabbretti, Pizzuto, Vicari, & Volterra, 1997; Sigman & Ruskin, 1999). The expressive language deficit is thought to be a result of deficits in the motoric components of speech as well as the acquisition of complex expressive language forms. Atypical development of the vocal cords, oral cavity, palate, and muscle tone all contribute to articulation and intelligibility issues in many individuals with DS (Miller & Leddy, 1999; Stoel-Gammon, 1992), and a low estimate of at least 15% of individuals show evidence of verbal apraxia (Kumin, 2006). Although expressive language development challenges many individuals with DS, nonverbal communication skills are usually an area of relative strength and may facilitate social interactions in this population (Fidler, Philofsky, Hepburn, & Rogers, 2005; Mundy, Kasari, Sigman, & Ruskin, 1995; Mundy, Sigman, Kasari, & Yirmiya, 1988; Wetherby, Yonclas, & Bryan, 1989).

Social skills have been found to be an area of relative strength for both younger and older children with DS who have been observed to have high levels of social engagement, social orientation, and competency in making friends in comparison to other children with developmental delays (Dykens, 2006; Fidler, Barrett, & Most, 2005; Kasari, Freeman, & Bass, 2003). However, individuals with DS have been observed to use their social strengths to avoid challenging tasks (Kasari & Freeman, 2002; Pitcairn & Wishart, 1994). In older childhood and adolescence, researchers have reported poorer interpersonal interactions with peers in students with DS who attend general education classrooms (Buckley, Bird, Sacks, & Archer, 2006).

Special Issues in Aging

DS has been called a syndrome of “precocious aging” (Lott, 1982). While aging in DS occurs with individual variability, adults with DS are at-risk for developing early-onset AD (Lott & Head, 2001). Research indicates that the symptoms of AD
in adults with DS may differ from those observed in the general population (Lott & Head, 2001). For example, Ball and colleagues (2008) found that (a) personality and behavior changes and (b) diminishing performance of executive functioning skills are early indicators of AD in people with DS. This finding contrasts with the changes in memory that often are associated with AD in the general population. However, decline is not an unavoidable outcome in this population. For example, Esbensen, Seltzer, and Krauss (2008) found that compared with other adults with intellectual disabilities, adults under 40 years of age with DS declined more quickly in health factors but maintained functional abilities better than the comparison group and had less behavioral problems. Importantly, poor family relationships and parental death were associated with more problems in health, behavior, and personal care skills in individuals with DS (Esbensen et al., 2008).

Areas of Occupation

Activities of Daily Living and Instrumental Activities of Daily Living

In much of the literature on individuals with DS, the term “adaptive behavior” is used to describe and measure functional skills such as dressing, meal preparation, safety, and the like (Dykens, 1993). Developmental researchers originally described “adaptive behavior” as a relative strength in children with DS that plateaus in middle childhood (Dykens, 1993; Dykens, Hodapp, & Evans, 2006). However, researchers from a rehabilitation science background (Coster, Deeney, Haltiwanger, & Haley, 1998) have argued that measures of adaptive behavior do not provide adequate information regarding levels of functional challenges and participation across different contexts. In contrast with the adaptive behavior findings in DS, a descriptive investigation of function related to participation in home and community life using parent report have found that children with DS demonstrate many challenges to participation, especially those related to self-care and safety (Dolva, Coster, & Lilja, 2004). Fidler, Hepburn, & Rogers (2005) have produced perhaps the only study addressing the connection between goal-directed behavior, a specific skill area found to be challenged in many individuals with DS and participation in daily life. They found that adaptive behavior skills were significantly correlated with planning skills on battery of planning tasks in toddlers with DS.

Education

Researchers have found evidence that preschoolers through teenagers with DS exhibit better progress, particularly in language, reading, and writing when included in general education classrooms (Buckley et al., 2006; Laws, Byrne, & Buckley, 2000). Indeed, the past decade has brought innovations that include a refined understanding of effective inclusive practice in DS (Wolpert, 2001) and new instructional approaches involving computer technology (Lloyd, Moni, & Jobling, 2006; Ortega-Tudela & Gomez-Arizal, 2006). These advances have led to notable milestones in the advancement of education in DS, including cases in the literature of young adults with DS attending university courses (Casale-Giannola & Wilson Kamens, 2006; Hamill, 2003). However, in a recent study conducted on a US sample, researchers found that students with DS spent on average over 4 hours of their school day in the special education resource room instead of attending general
education classes with academic support (Fidler, Most, & Daunhauer, in press). In this study, there also was a moderate, negative correlation between age and time spent in the general education. Therefore, the older the students, the less time they spent in general education classrooms. This may raise questions whether educating students in “the least restrictive environment” as proscribed by the Individuals with Disabilities Education Act (IDEA, 34 CFR §300.320[4]) is being met. Currently there is a paucity of evidence and little legal precedent for how to interpret these findings for students with DS (Farley, 2002; Roberts, 2008).

**Work and Leisure**

Hodapp (2004) hypothesized that the patterns of relative strengths and challenges associated with behavioral phenotypes would have both direct and indirect effects over the course of a lifetime for an individual with a genetic syndrome. He described one of those direct effects as being the environments/activities an individual with a genetic syndrome chooses. In the small body of research examining this phenomenon, researchers are finding that individuals with genetically-based intellectual disabilities are demonstrating activity choices that appear to be related to their genetic syndrome. For example, researchers have found that school-aged children with DS are reported to participate in visual-motor activities such as arts and crafts as well as engaging in activities involving dancing, singing, and listening to music more than individuals with other genetic syndromes such as Prader–Willi syndrome (Rosner, Hodapp, Fidler, Sagun, & Dykens, 2004). In this study, the DS group also was reported to participate in significantly more job-related activities and to exhibit lower skills and participation in sports. In a study of toddlers through adults, researchers found that the DS group participated more frequently in drawing and coloring as well as passive musical activities (listening as opposed to playing an instrument) than individuals with other genetic syndromes (Sellinger, Hodapp, & Dykens, 2006).

In a qualitative study, researchers examined individuals identified through their community for the “Stevie Award for Outstanding Persons with Down syndrome” (Li, Liu, Lok, & Lee, 2006). The researchers identified three recurring themes in the lives of these outstanding individuals with DS: involvement in extra-curricular activities and volunteer work, enthusiasm or persistence for learning, and supportive relationships from parents and/or teachers. Finally, supportive social relationships may promote a healthy lifestyle in individuals with DS, as well. One study found a relationship between friendships and engagement in physical recreation for individuals with DS and positive body mass indexes (Fujiura, Fitzsimons, Marks, & Chicoine, 1997).

Taken as a whole, the phenotypic evidence reviewed above supports Hodapp’s (2004) hypothesis that there may be both direct and indirect effects associated with genetic phenotypes such as DS. The evidence to date indicates that many individuals with DS may exhibit strengths in visual-perceptual skills, receptive language, and core social relatedness. However, many people with DS experience significant challenges including a cognitively avoidant learning style and a tendency for some individuals with DS to use their social strengths to avoid challenging tasks. This may result in a cascading chain of missed opportunities and exacerbate weaker skills, for example with in planning goal-directed actions, inhibiting automatic responses, and
performing skillful motor actions. However, much more information is needed to understand the interaction between phenotypic profiles and participation in daily life.

**IMPLICATIONS FOR RESEARCH AND PRACTICE IN OCCUPATIONAL THERAPY**

**Research Issues**

Although there has been substantial progress in the study of phenotypic outcomes, very little of this work has been conducted within an occupational therapy perspective. The occupational therapy profession can make important contributions to understanding of outcomes in DS by embarking on a research agenda that involves two goals. First, research from occupational therapy and occupational science perspective can assist in expanding our knowledge of the direct and indirect effects of the DS behavioral phenotype, particularly in relationship to how the relative strengths and challenges in performance skills may affect choices and success in areas of occupation in various contexts such as education, work, leisure, and play. Second, researchers from an occupational therapy perspective can examine how best to target the developing profile in DS with time-sensitive and effective intervention techniques and to test the effectiveness of etiology-specific interventions in this population.

In order to initiate this agenda, it would be useful for occupational therapy researchers to begin with a discussion of specific methodological issues that will be faced in this type of work. First, a discussion regarding comparison groups to be used in research on individuals with genetic syndromes such as DS will be critical. Determining the most appropriate comparison group for individuals with specific disorders such as DS has been a topic of much thoughtful discussion and debate in developmental research (Dykens & Hodapp, 2007; Hodapp & Dykens, 2001; Seltzer, Abbeduto, Krauss, Greenbert, & Swe, 2004). As Hodapp and Dykens (2001) note, in general, when interested in identifying whether a specific profile is unique to a particular etiology group such as DS, a comparison group of children with other intellectual disabilities can be useful. They reason that a TD comparison group only highlights general challenges that students with DS may exhibit in skills and engagement in activities. However, research using a TD group does *not* provide data regarding what may be distinct challenges and distinct strengths for individuals with a genetic syndrome such as DS in contrast to students with other types of intellectual delays (Hodapp & Dykens, 2001). Instead, a group with idiopathic or mixed etiologies of intellectual disabilities allows investigators to assess whether skills and behaviors are related to intellectual disability in general or to a specific genetic etiology (Hodapp & Dykens, 2001; Seltzer et al., 2004). Still other researchers propose to compare groups with genetic syndromes such as DS with groups of individuals with other genetic disorders, such as Williams syndrome (Burack, Iarocci, Bowler, & Mottron, 2002). It will likely be necessary for researchers in both occupational therapy and occupational science to engage with these issues within the context of occupational therapy-related research methodology and similar discussions may be useful in order to establish a consensus on how best to design studies that examine syndrome specificity.
**Practice Issues**

The connection between genetic condition and lifespan outcomes has become an important topic of discussion in the field of intervention and educational practice for individuals with intellectual disability (Fidler et al., 2002; Fidler & Nadel, 2007; Fidler, Philofsky, & Hepburn, 2007; Hodapp & Fidler, 1999; Roberts, Chapman, & Warren, 2008; Rondal & Buckley, 2003). However, there has been little discussion of this topic in peer-reviewed journals in the field of occupational therapy. There are potential practice issues. In a study, Fidler et al. (2002) examined parents’ knowledge of etiology-specific behavioral phenotypes in children with different genetic intellectual disability syndromes and found that parents of children with DS were aware of their children’s more prominent phenotypic traits such as relative strengths with visual processing and relative weaknesses with expressive language. However, they noted that parents were less aware of “…the more subtle and complex cognitive features associated with their children’s syndromes” (p. 86). Over half of the parents of children with DS reported that they wanted more occupational therapy services for their child. Despite this desire for more occupational therapy services, the parents also reported that the occupational therapist was less likely to bring etiology-specific information than any other team member. Furthermore, no parents in the other two groups, parents of children with Prader–Willi and Williams syndromes, reported that the occupational therapists had provided etiology-specific information to the team. In order to support families and individuals with etiology-specific intervention and accommodations specific to DS and other genetic syndromes related to intellectual disability, it will be critical for occupational therapists to both conduct research and actively stay informed of evidence-based outcomes being published in related disciplines.

These recommendations are particularly relevant to intervention with the DS population given the emerging evidence that some interventions may have differential effects for individuals with DS compared with individuals with other DDs (e.g., Landry, Miller-Loncar, & Swank, 2002; Yoder & Warren, 2002). For example, Landry and colleagues (1998) investigated the effects of a maternal play/support intervention on goal-directed behaviors and found that children with DS demonstrated no improvements, while a control group demonstrated treatment effects. Similarly, three studies (Fey et al., 2006; Warren et al., 2008; Yoder & Warren, 2002) found differential effects for interventions targeted for individuals with DS. Yoder and Warren (2002) found in a language intervention (Responsive Education/Prelinguistic Milieu Teaching—RE/PMT) that a comparison group of MA-matched children with DDs responded with greater gains in nonverbal requesting than the DS group. Paradoxically, the DS group not receiving RPMT actually demonstrated faster growth in requesting skills than the DS group in intervention. The researchers suggested that the intervention style that requires increasing amounts of persistence may have been moderated by the motivational style challenges associated with the DS behavioral phenotype.

Syndrome specificity in response to shaping behavior was found in a study examining parent approaches that facilitated on-task performance in a visual-perceptual activity for young children through young adults with DS. Researchers found that while parent approaches did not significantly differ between the DS group and
idiopathic intellectual disability group, the DS group responded significantly more frequently with off-task behavior, particularly when their parents used verbal instructions that were task oriented instead of emotionally supportive (Daunhauer, Flabiano, & Fidler, 2009). Fidler, Philofsky, and Hepburn (2007) noted that these increasing observed differential responses to intervention for children with DS may stem from the phenotypic profile associated with this disorder including challenges in motivation orientation, language development, and goal-directed behavior. These data emphasize that more specific research on DS may help interventionists select evidence-based techniques, dosage, and objectives to maximize intervention outcomes.

While acknowledging the paucity of empirical studies validating etiology-specific intervention approaches in occupational therapy practice, we propose that phenotypic information on DS can help guide occupational therapists in developing interventions for clients with DS and their families. For example, in early development Fidler and colleagues (2007) suggest interventionists use anticipatory guidance that is being on the “look out” for areas of potentially heightened vulnerability. With anticipatory guidance, it may be possible to identify early manifestations of outcomes before an area of difficulty becomes exacerbated. Therefore, an occupational therapist may observe a toddler with DS who exhibits motor planning challenges. Using anticipatory guidance, the occupational therapist may develop activities to encourage exploration and cause-and-effect play that minimizes cascading difficulties with goal-directed actions. In the educational context, an occupational therapist may analyze a student’s participation across the school day. From these observations, the occupational therapist may assist a team in determining the most beneficial cues to support participation in educational activities. For example, a student may benefit from shorter verbal cues or the addition of written cues to compensate for challenges in working memory. Likewise, using the available phenotypic evidence, an occupational therapist may create new social opportunities for older students, encourage social opportunities to promote exercise, or contribute to employment transition plans for graduating students. In assistive living, occupational therapists may help team members identify specific early signs of dementia in this population (e.g., declines in praxis and executive function). They also might help identify supports such as social and family support that will allow an individual to function as independently as possible in old age.

Modifiability
In discussing outcomes associated with genetic disorders, questions arise regarding the modifiability of outcomes through intervention. Evidence is emerging both from studying animals and examining child developmental outcomes to support behavioral phenotypes as modifiable. The extent that neural plasticity exists in individuals with DS still remains a question (Dierssen, Ortiz-Abalia, Arque, Martínez de Lágran, & Fillart, 2006). Currently, two studies using mouse models of DS provide conflicting evidence. One found that enriched early environments positively affected neural plasticity in very young female mice (Martínez-Cué et al., 2002), and one conducted on older mice found no effects (Dierssen et al., 2006). Clearly further research is needed. In examining modifiable outcomes, Buckley and colleagues (2006) found that British adolescents with DS who attended a school with an
“inclusive” educational model demonstrated no adaptive communication deficits relative to social and daily living skills as opposed to adolescents with DS who attended special education without inclusion. These findings are a potentially promising beginning to work that aims to modify and improve outcomes associated with the DS behavioral phenotype.

CONCLUSIONS
The characterization of the DS behavioral phenotype has great relevance for families, educators, and interventionists (Hodapp & Fidler, 1999). Though to date, there is a paucity of rigorous intervention research to support these assertions; arguments have been in the literature regarding the potential utility of this type of syndrome-specific behavioral information (Fidler, 2005; Fidler, Philofsky, & Hepburn, 2007; Hodapp & DesJardin, 2002; Hodapp & Fidler, 1999). This article reviewed the characterization of DS behavioral phenotype in the areas of client factors, performance skills, and occupation. Additionally, it examined issues related to both research and practice. While it is acknowledged that individual variability always exists in how the DS behavioral phenotype affects outcomes over the course of a lifespan, the intention of this article is to spark further discussion related to the needs of this population in the occupational therapy community.

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