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Clinical Focus

Down Syndrome and Autism Spectrum Disorder Dual Diagnosis: Important Considerations for Speech-Language Pathologists

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Purpose: Individuals with Down syndrome (DS) often receive speech-language therapy services starting in infancy or toddlerhood. When providing speech-language therapy services for children with DS, speech-language pathologists (SLPs) need to consider the impact of other developmental and comorbid disorders that can affect language development, such as the presence of a dual diagnosis of DS and autism spectrum disorder (DS + ASD). The prevalence rate of ASD in DS is ~20%, which is higher than in the general population.
Method: This clinical focus article aims to provide SLPs with additional knowledge about DS + ASD to improve service delivery and support parents' ability to advocate for their child with confirmed or suspected DS + ASD. This is accomplished by summarizing the current evidence base on the presence of ASD in DS and discussing implications of a DS + ASD diagnosis for clinical practice with SLPs.
Conclusions: SLPs play a key role in supporting families of those with DS + ASD by advocating and educating. By understanding the unique profiles of strengths and weaknesses of individuals with DS + ASD, SLPs can provide appropriate service delivery (i.e., treatment and intervention approaches) and advocacy for their clients and their families.

Down syndrome (DS) is a neurogenetic syndrome caused by a triplication of all or part of chromosome 21 (trisomy 21), leading to mild-to-moderate intellectual disability (Parker et al., 2010). DS is characterized by a unique behavioral profile that includes relative weakness in expressive language and relative strengths in receptive language and nonverbal social communication (Luyster et al., 2011; Philofsky et al., 2007). Given these early known difficulties with language, most children with DS start receiving speech-language therapy in their first 2 years of life (Cuckle & Maymon, 2016). When providing speech-language therapy services for children with DS, speech-language pathologists (SLPs) need to consider the impact of intellectual disability, as well as other comorbid disorders that can affect language and communication development. Another important consideration is the presence of a dual diagnosis of autism spectrum disorder (ASD) and the potential overlap in symptoms between intellectual disability, DS, and ASD.

Many SLPs are familiar with DS and ASD as individual neurodevelopmental disorders, but they may not be as familiar with DS and ASD (DS + ASD) comorbidity, which until recently has received little research or clinical attention. This may be due in part to a historical belief that an individual with DS could not have ASD because of the stereotype that individuals with DS are always social (Reilly, 2009). However, there is now an evidence base indicating that individuals with DS can have comorbid ASD (Reilly, 2009). Although research on DS + ASD is ongoing, this evidence must be provided to SLPs. SLPs' expertise in social communication and the likelihood of providing services to children with DS starting in early development make SLPs a key figure in identifying early signs of ASD in DS. Also, children with DS + ASD may need additional or different approaches to treatment than those with DS or ASD only. This clinical focus article aims to provide SLPs with additional knowledge about DS + ASD to improve service delivery and support parents' ability to advocate for their child with confirmed or suspected DS + ASD. This is accomplished by summarizing the current evidence base on the presence of ASD in DS and discussing implications of a DS + ASD diagnosis for clinical practice.

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ASD

ASD is a neurodevelopmental disorder characterized by two core behavioral domains: (a) persistent deficits in social communication and social interaction across multiple contexts and (b) restricted, repetitive patterns of behaviors, interests, or activities (American Psychiatric Association [APA], 2013; World Health Organization [WHO], 2018). It is estimated that, in the United States, one in 59 children have ASD (Baio et al., 2018). Common features of ASD include difficulty with social–emotional reciprocity, understanding relationships, nonverbal communication, and fixation on interests and/or routines (APA, 2013; WHO, 2018). Characteristics of ASD must be present within the early developmental period to warrant an ASD diagnosis. ASD may co-occur with intellectual disability, language impairment, medical conditions, and other neurodevelopmental and neurogenetic syndromes, like DS.

DS and Comorbid ASD

Comorbidity Estimates

To date, there has not been a population-based prevalence study of ASD in DS. However, the available research suggests that ~20% of individuals with DS meet diagnostic criteria for ASD (range: 7%–42%; Capone et al., 2005; DiGuiseppi et al., 2010; Ghaziuddin et al., 1992; Lowenthal et al., 2007; Oxelgren et al., 2017; Warner et al., 2014). This range in the estimated prevalence of DS + ASD may be due to differences in the studies' methodologies (sampling methods, diagnostic tools, etc.) and the ASD diagnostic criteria used (i.e., *Diagnostic and Statistical Manual of Mental Disorders, Fourth Edition* [*DSM-IV*] *or Fifth Edition* [*DSM-5*]; *International Classification of Diseases* [*ICD-10* or *ICD-11*]; APA, 1994, 2013; WHO, 1992, 2018). Nonetheless, this prevalence rate is higher than the 2% prevalence rate in the general U.S. population (Baio et al., 2018).

Age of Diagnosis and Onset

The diagnosis of ASD in DS is often made later in life than it is in individuals with ASD only. The age of diagnosis for individuals with ASD only occurs, on average, by 4.5 years of age (Baio et al., 2018), but can be reliably diagnosed as early as 2 years (Lord et al., 2006). However, for individuals with DS + ASD, the available, but limited, research suggests that this dual diagnosis is not made until later in development (range: 4–33 years; Rasmussen et al., 2001; Reilly, 2009) due, in part, to challenges with early identification of ASD. The challenges associated with early diagnosis of ASD may be more difficult in children with DS because of overlap in the early behavioral features of these disorders (Capone, 1999; Reilly, 2009; Stone et al., 1999). Therefore, it is important to be familiar with the profiles associated with DS, ASD, and DS + ASD. Furthermore, some behavioral features of ASD may not be as clear in young children with DS due to delays in motor development (i.e., presence of restricted, repetitive patterns of behaviors, interests, or activities; Reilly, 2009). This later age of diagnosis likely negatively impacts development and functioning for individuals with DS + ASD because they are not receiving the best interventions and treatments for their needs (Reilly, 2009).

Despite the challenges associated with diagnosing ASD in DS, two developmental patterns for ASD onset in DS have been identified by Capone based on clinical experience and observation (personal communication, December 11, 2019). The available cross-sectional research studies are consistent with the onset patterns reported in those with ASD only (Kalb et al., 2010; Ozonoff et al., 2008; Ozonoff & Iosif, 2019): (a) early onset and (b) developmental regression or plateau. However, it is important to note that the developmental course of ASD symptoms in DS has yet to be examined in a longitudinal study. Therefore, more research is needed to confirm these patterns. For both of these onset patterns, SLPs are positioned to be among the first professionals to notice these symptoms or have them reported to them by parents.

In the first pattern of development, individuals with DS + ASD, like those with ASD only, present with atypical behaviors early in infancy and toddlerhood (Capone, 1999). To date, only one study describes early signs of ASD in infants and toddlers with DS (7–18 months; Hahn et al., 2020). This study suggests that seven of 18 infants and toddlers with DS were designated at risk for ASD. Similarly, a longitudinal study of ASD in 2- to -3-year-olds with DS (M_{age} = 34 months; Hepburn et al., 2008) indicates that 15% (n = 3/20) of the sample met criteria for ASD using a comprehensive diagnostic evaluation. Diagnostic stability was high with all children with DS diagnosed with ASD retaining the diagnosis and all those not diagnosed with ASD not meeting criteria at the 2-year follow-up. Longitudinal follow-up indicated that the severity of ASD features increased for two of the three children with DS diagnosed with ASD, whereas it did not change for the third child (Hepburn et al., 2008). Children in both studies demonstrated impairments in communication and social skills (Hahn et al., 2020; Hepburn et al., 2008). Collectively, these data provide initial evidence that ASD symptoms emerge early in development (Hahn et al., 2020; Hepburn et al., 2008).

The second developmental pattern that has been noted in individuals with DS + ASD, and those with ASD only (Ozonoff & Iosif, 2019), is developmental regression or plateauing (Capone, 1999; Castillo et al., 2008). This group of children experiences a dramatic loss or inability to advance in developmental milestones related to acquisition and use of language and social skills. Before regression, development follows patterns typical for children with DS. In comparison to children with regressive onset ASD, children with DS with regressive onset ASD appear to show regression later in development (Castillo et al., 2008). This later onset may be related in part to the overall level of developmental delay associated with DS. However, it is important to note that the pattern of regression in those with DS + ASD is similar to those with ASD only (Castillo et al., 2008).

Behavioral Profile

The behavioral profile of DS alone is relatively well-studied, but less is known about the DS + ASD behavioral profile. There is an ongoing effort by researchers across medical, behavioral, and genetic disciplines to understand how the presence of DS may influence the presentation of core features of ASD (Channell et al., 2015, 2019; DiGuiseppi et al., 2010; Godfrey et al., 2019; Ji et al., 2011; Molloy et al., 2009; Moss & Howlin, 2009). In doing so, this line of research seeks to clarify the shared phenomenology between the two disorders and identify any unique features of the DS + ASD profile (Glennon et al., 2017). Nonetheless, the available evidence base can provide SLPs with important and relevant information to support their clients with DS + ASD by drawing their attention to social communication challenges and informing their intervention approaches for this population.

Similar to those with ASD only, the presence of ASD symptomatology emerges early in DS (Hahn et al., 2020; Hepburn et al., 2008). Individuals with DS + ASD have marked difficulties in areas of social and emotional reciprocity early in development that may be similar in severity to individuals with DS alone with severe cognitive impairment (Hepburn et al., 2008; Howlin et al., 1995; Molloy et al., 2009). Additionally, individuals with DS + ASD may have poorer communication skills than individuals with DS alone and may present with little to no meaningful symbolic communication; however, this impairment may be reduced relative to ASD alone (Capone, 1999; DiGuiseppi et al., 2010; Godfrey et al., 2019; Moss et al., 2013). Interestingly, a recent study indicates the high correlation between lower verbal abilities and social communication impairments in children with DS + ASD is not as significant in children with DS only and those with ASD only (Godfrey et al., 2019).

It has been suggested that individuals with DS + ASD may have more severe intellectual disability than those with DS alone (Capone et al., 2005; Carter et al., 2007; Molloy et al., 2009; Starr et al., 2005; Wester Oxelgren et al., 2019). However, severe intellectual disability in an individual with DS does not alone yield an additional diagnosis of ASD. Cognition may be further affected by the increased risk of anxiety, irritability, difficulty with transitions, hyperactivity, withdrawal and attention problems, and sleep disturbances associated with the dual presentation of DS + ASD (Capone, 1999; Moss et al., 2013). Additionally, individuals with DS + ASD show higher rates of stereotyped behaviors than those with DS alone (Godfrey et al., 2019; Moss et al., 2013). These behaviors include self-injurious behaviors (e.g., biting, hitting), repetitive motor behaviors (e.g., teeth grinding, hand flapping), vocalizations (e.g., howling, growling), response to sensory input (e.g., attraction to lights and spinning objects), and feeding problems (e.g., refusal, strong preferences; Center for Disease Control and Prevention [CDC], 2019a; Moss et al., 2013). It is important to remember that ASD is a spectrum; individuals with DS + ASD demonstrate a continuum of ASD-like features (Starr et al., 2005).

Risk Factors for ASD in DS

By understanding the risk factors for ASD in DS, SLPs also can play a role in identifying individuals with DS who may be at a greater risk for ASD and should be referred for further screening and evaluation. There is emerging evidence about the potential risk factors associated with the development of DS + ASD. These risk factors include familial history of ASD-related disorders in first- or second-degree relatives (similar to those with ASD only), infantile spasms or seizures, early hypothyroidism, brain injury secondary to complicated surgeries (i.e., heart surgeries, etc.), and/or a combination of these factors (Rasmussen et al., 2001). Furthermore, higher rates of impaired social skills have been reported in family members of individuals with DS + ASD in comparison with individuals with DS alone (Lowenthal et al., 2007). It has also been suggested that seizures across the life span may increase the presentation of ASD-like behaviors in individuals with DS + ASD (Molloy et al., 2009). While there is evidence that ASD symptoms present early in DS, as in ASD only, to date, only one study has examined early markers of ASD in infants with DS (Hahn et al., 2020). This preliminary, cross-sectional study presents the first evidence that early risk markers of ASD are present and detectable in infants with DS (Hahn et al., 2020). Furthermore, like other populations at risk for ASD (Kalb et al., 2010), impairments in social communication may signal elevated risk for the emergence of ASD in DS (Hahn et al., 2020, Hepburn et al., 2008; Starr et al., 2005).

Diagnosing ASD in DS: Role of the SLP

Early Identification

SLPs working with young children with DS may be among those first to notice early signs of ASD. Early identification of ASD in DS is important for these children to receive the best interventions and treatments for their needs (Reilly, 2009). These early signs of ASD, in general, include repetitive behaviors such as lining up toys, echolalia, or hand flapping; sensory fixations such as aversions to or preferences for certain tastes, textures, lights, or sounds; and lack of language comprehension or expression, though this final variable may not be as clear due to the language impairments associated with ASD (CDC, 2019b). Given that the presence of intellectual disability is common in DS, it can be difficult to discern between repetitive behaviors that are associated with intellectual disability and those associated with ASD due to similar behaviors being present (Capone et al., 2005; Howlin et al., 1995; Vatter, 1998). Based on the available research, an early sign of ASD in DS are impairments in communication and social skills that are greater than would be expected for a child with DS (Hahn et al., 2020; Hepburn et al., 2008). Nonetheless, early signs of ASD in children with and without DS may be better associated with the intellectual disability and do not always lead to an ASD diagnosis. Therefore, knowledge of the behaviors associated with each neurodevelopmental disorder (i.e., intellectual disability, DS, and ASD) will help with the differentiation of symptoms.

A Team-Based Approach to the Diagnosis of ASD in DS

As stated by the American Speech-Language-Hearing Association (ASHA), SLPs make up an important part of the interdisciplinary team needed to diagnose ASD (ASHA, n.d.; Autism Practice Portal). An interdisciplinary team-based approach to diagnosing ASD often requires close collaboration with parents, clinical psychologists, pediatric physicians, and other specialists, such as SLPs (Gerdts et al., 2018). Unless an SLP has received specialized training in ASD diagnostics, diagnosing ASD does not fall within the SLP's scope of practice (ASHA, n.d.: Autism Practice Portal). However, SLPs working with individuals with DS, ASD, and DS + ASD should be familiar with the current diagnostic criteria and tools to support their client's needs and participate as part of the diagnostic team. SLPs are an important component of the diagnostic evaluation for ASD as their expertise in language development, social communication, and pragmatics is integral to the diagnosis of ASD. Also, the familiarity of the SLP with the language and communication profile associated with DS can help a team determine if the behaviors of an individual with DS are best explained by the behavioral profile associated with DS alone or the dual diagnosis of DS + ASD (Reilly, 2009).

Facilitating Parent Involvement in the Identification and Diagnosis of ASD in DS

Infants and toddlers with DS are often referred to SLPs due to the known difficulties with language development, especially spoken language. If parents have not been referred to an SLP, they may seek out these services very early in development due to recommendations from other parents or through their research on how to support their child with DS. As always, it is important SLPs are responsive to caregiver concerns about their child's behavior. Parents are often the first to identify personality or behavioral changes that lead to the initiation of the early ASD diagnostic process (Grønborg et al., 2013; Messinger et al., 2013), and likely, this is also true for parents of children with DS. Familiarity with the patterns of behavior associated with DS, ASD, and DS + ASD is essential for interpreting parental concerns. It is important to note that parents of children with DS may not realize their child can also have ASD, so their concerns may be described in comparison to what they know about DS or to other children with DS they know. Therefore, SLPs need to be vigilant responders to the concerns of these parents and work to decode parents' concerns or descriptions of their child's behavior to discern whether their concerns are associated with early signs of ASD.

If parents are concerned about ASD, SLPs can refer families to appropriate resources. This includes local, state, and federal resources. The Center for Disease Control has launched "Learn the Signs, Act Early," which was developed to support early identification of ASD and other developmental disabilities by providing information and videos about child development that is accessible for parents and professionals (CDC, 2019b; https://www.cdc.gov/actearly). The Autism Navigator also provides video examples to help parents learn more about the behaviors and symptoms associated with ASD (https://autismnavigator.com/family-resources/; Autism Navigator, 2018). Both of these resources can be beneficial for SLPs who are not as familiar with ASD.

Consideration of these identified behaviors in conjunction with comprehensive assessments may warrant a dual diagnosis of DS + ASD. Early identification is critical as it opens the door to services that promote positive outcomes related to social, educational, and vocational aspects of daily life, and children with DS + ASD may require additional supports to achieve these outcomes (Koegel et al., 2014; Reilly, 2009). By helping families learn the signs of ASD, SLPs can support the early identification of ASD in DS and in young children broadly.

Assessment and Measurement of ASD in DS

Clinical judgment and experience are key to early identification and diagnosis of ASD, as this opens the door to services that promote positive outcomes related to social, educational, and vocational aspects of daily life. The current diagnostic criteria for ASD are outlined in the *DSM-5* (APA, 2013) and the *ICD-11* (WHO, 2018). Both the *DSM-5* and *ICD-11* criteria have moved to the use of "autism spectrum disorder" instead of using the category of "pervasive development disorders." Furthermore, this diagnostic category in either the *DSM-5* or *ICD-11* is no longer subdivided into "autistic disorder," "Asperger's syndrome," and "pervasive developmental disorder, not otherwise specified" (APA, 1994; WHO, 1992). Thus, *autism spectrum disorder* acts as a single umbrella term. Individuals who received a diagnosis of either autistic disorder, Asperger's syndrome, or pervasive developmental disorder, not otherwise specified under the *DSM-IV* or *ICD-10*, still meet criteria for ASD under the *DSM-5* and the *ICD-11* (APA, 2013; WHO, 2018). In both the *DSM-5* and *ICD-11*, individuals can be diagnosed with co-occurring intellectual disability and/or language impairment. However, per the *ICD-11*, individuals with ASD cannot also be diagnosed with developmental language disorder (WHO, 2018), and in the *DSM-5* criteria, they cannot also be diagnosed with social (pragmatic) communication disorder (APA, 2013). The nuances associated with language and communication when diagnosing ASD highlight the importance of having SLPs on the diagnostic team.

Aligned to one or more of the aforementioned diagnostic standards, a variety of tools have been developed to assist in the detection of ASD and/or to describe the severity of ASD and can be used with DS populations. These tools fall broadly into three categories: screeners, diagnostic measures, and symptomatology measures. Below, we provide a summary of common tools used in ASD, including their sensitivity (i.e., percentage of kids who are identified who go on to have an ASD diagnosis) and specificity (i.e., percentage of kids who are not identified and do not go on to have an ASD diagnosis), and their use in populations with DS to familiarize SLPs with these tools.

Common Screeners

Screeners are used to quickly assess the likelihood that an individual has ASD. That is, they provide a risk cutoff score. If an individual fails a screener (i.e., has a score that indicates they are at risk), he or she must undergo further evaluation to determine the presence or absence of ASD. The information gathered in the administration of a screener is not extensive enough to make the diagnosis accurately and appropriately. While screeners attempt to have both strong sensitivity and specificity, both false positives and false negatives may occur, making the referral for a full diagnostic evaluation necessary before diagnosing ASD. Thus, when a screener has been administered, it is important to not make diagnostic statements to parents until a comprehensive diagnostic elevation has been completed. Table 1 provides a summary of common screeners for ASD, many of which have also been used in samples with DS.

Of these screeners, the Social Communication Questionnaire (SCQ; Rutter et al., 2003) has been used the most with populations with DS. Collectively, these studies indicate that the SCQ can be used as either a measure of ASD symptoms in DS or to identify those with who are at an increased risk for ASD (Channell et al., 2015, 2019; DiGuiseppi et al. 2010; Moss et al. 2013; Oxelgren et al., 2017; Warner et al., 2014). It has been suggested that some individuals with DS who do have ASD may score just below the cutoff on the SCQ (Oxelgren et al., 2017). This pattern has been seen in other neurodevelopmental disorders, highlighting the need to develop cutoff scores for different known conditions associated with ASD (Glennon et al., 2017).

Common Diagnostic Measures

There are two common diagnostic measures that, when combined with a comprehensive evaluation (a family history interview; assessment of cognition, adaptive behavior, and language; etc.) and clinical judgment, are used to formally diagnose ASD. Clinical judgment must be used in the interpretation of comprehensive diagnostic evaluation results to ensure that the diagnosis is made appropriately. Both of these measures require specialized training to administer and score them. Without this training, these measures cannot be administered.

Autism Diagnostic Observation Schedule. The Autism Diagnostic Observation Schedule (ADOS), currently in its second edition (ADOS-2), is a standardized, semistructured assessment of communication, social interaction, and play for individuals suspected of having autism (Lord et al., 2012). The ADOS consists of five modules appropriate for young children through adults of differing developmental and language levels, ranging from nonverbal to verbally fluent. It is important to note that eligibility criteria for the ADOS-2 toddler module require a minimum mental age of 12 months and that the child is walking or "cruising" (Lord et al., 2012). Also, the toddler module provides a range of concern for ASD instead of the likelihood of ASD provided by the other modules. Within each module, the individual's behaviors and responses are coded. The examiner determines the presence or absence of behaviors consistent with the diagnosis of across the life span. The ADOS yields a total score that is used to indicate the likelihood of ASD based on the cutoff score for each module. The second edition of the ADOS (i.e., ADOS-2) is consistent with diagnostic criteria for ASD according to the *DSM-5*, while the ADOS–General is consistent with the *DSM-IV* diagnostic criteria (Lord et al., 2012). The ADOS-2 has established specificity (.86–.94) and sensitivity (.83–.91).

Autism Diagnostic Interview–Revised. The Autism Diagnostic Interview–Revised (ADI-R) is a standardized, semistructured interview concerning current functioning, behaviors displayed at 4–5 years of age, and lifetime occurrence (Lord et al., 1994). This interview is conducted with a parent or caregiver and is a valid and reliable diagnostic tool. Items of the ADI-R correspond with the diagnostic criteria for ASD according to the *DSM-IV*/*ICD-10*. There are three areas of focus on the ADI-R: quality of communication and language; quality of reciprocal social interactions; and repetitive, restricted, and stereotyped interests and behaviors. For each area, cutoff scores are used to indicate the likelihood of ASD, with higher scores indicating more characteristics and/or severity of ASD. Also, the onset of ASD must be present before 36 months. The ADI-R has established specificity (.70–.81) and sensitivity (.80–.96; Kim & Lord, 2012).

Several studies have used the ADOS and/or the ADI-R to examine the presence of ASD in DS (Godfrey et al., 2019; Hepburn et al., 2008; Oxelgren et al., 2017; Starr et al., 2005; Wester Oxelgren et al., 2019). For example, in a study of 13 individuals with DS (7–31 years), six met the criteria for ASD on either the ADOS or ADI-R (Starr et al., 2005). However, other studies are using the ADOS and/or ADI as part of a comprehensive evaluation for ASD—administering the ADOS, ADI-R, and other developmental measures along with clinical judgment to make a diagnosis of ASD. For example, Hepburn et al. (2008) examined ASD in 20 toddlers with DS (2–3 years) using this comprehensive approach. Three toddlers met the cutoff for ASD for both social and communication on the ADOS, and these three toddlers also met cutoff criteria for ASD for communication on the ADI-R (Hepburn et al., 2008). Three other toddlers exceed the cutoff for only the communication domain of the ADOS, and one toddler exceeded criteria on the social domain of the ADOS, but these four toddlers were not identified as having ASD. Taken together, the results of this study highlight the importance of using a comprehensive evaluation of ASD in DS because there may be overlapping features associated with both DS and intellectual disability that lead to children with DS exceed criteria on some components of these measures.

Symptomatology Measures

Symptomatology measures are used to assess and describe the severity of symptoms and behaviors associated with ASD, including how they may be affecting activities of daily life. These measures differ from a screener because they do not provide a risk score. These measures are used to describe the type, frequency, and/or severity of symptoms and behaviors associated with ASD, which can provide

Table 1. Common screening measures for ASD.

Name of measure	Reference	Age range	Description	Scoring	Cutoff score[a]	Sensitivity	Specificity	Used with DS
Social Communication Questionnaire (SCQ)	Rutter et al., 2003	4+ years (mental age 2+ years)	Focuses on behaviors that are rare in nonaffected individuals and are consistent with the ASD diagnostic criteria of the *DSM-IV/ICD-10* in the domains of Reciprocal Social Interaction; Communication; and Restricted, Repetitive, and Stereotyped Patterns of Behavior	yes/no	15+	.85–.95	.67–.80	Channell et al., 2015, 2019; DiGuiseppi et al., 2010; Magyar et al., 2012; Moss et al., 2013; Oxelgren et al., 2017; Warner et al., 2017, 2014
Modified Checklist for Autism in Toddlers (M-CHAT)	Robins et al., 2001	6–30 months	Identifies early signs of ASD based on the child's achievement of developmental milestones	yes/no	0–2: no follow-up necessary 3–6: refer for evaluation 7–23: at risk for ASD	.87–.97	.95–.99	DiGuiseppi et al., 2010
Modified Checklist for Autism in Toddlers–Revised/Follow-up (M-CHAT-R/F)	Robins et al., 2014, 2009	16–30 months	Uses simpler wording than the M-CHAT to identify early signs of ASD based on the child's achievement of developmental milestones. If a child screens positive on the M-CHAT-R, then the follow-up questions for the items the child failed are asked of the parent.	pass/fail	0–2: Low risk 3–7: Medium risk 8–20: High risk	.67–.86	.992–.995	—
Social Responsiveness Scale (SRS; SRS-2)	Constantino et al., 2012	2.5+ years	Measure of the presence and severity of social impairments associated with ASD. The SRS-2 can be completed by a parent or teacher, and for individuals who are older than 19 years, it can also be completed by a relative, friend, or as a self-report.	1 = *not true* to 4 = *almost always true* Higher scores indicate greater severity of social impairments.	< 59T: within normal limits 60T–65T: mild range 66T–75T: moderate range 76T+: severe range	.92[b]	.92[b]	Channell, 2020; Channell et al., 2015
Autism Behavior Checklist (AutBC)	Krug et al., 1980	2–14;11 (years; months)	Indicates the presence or absence of ASD-associated behaviors in the following categories: sensory, relating, body and object use, language, and social and self-help	weighted score from 1 to 4 Higher scores indicate a greater frequency of behaviors	< 54: no follow-up necessary 54–67: refer for evaluation 68+: suggests "high probability" of autism	.75–.88	.81	Carter et al., 2007

Note. Score that indicates the need for a more complete ASD diagnostic evaluation. ASD = autism spectrum disorder; *DS = Down syndrome; DSM = Diagnostic and Statistical Manual of Mental Disorders, Fourth Edition*; *ICD = International Classification of Diseases*.

[a]Score that indicates the need for a more complete ASD diagnostic evaluation. [b]Sensitivity and specificity from Bruni (2014).

Table 2. Common symptomatology measures for ASD.

Name of measure	Reference	Age range	Description	Scoring	Used with DS
Aberrant Behavior Checklist (ABC; ABC-2)	Aman et al., 1985	5 years to adult	Indicates the severity of problem behaviors associated with ASD across five categories: irritability, lethargy/social withdrawal, stereotypy, hyperactivity, and inappropriate speech	0 = *not problematic* to 3 = *severely problematic*	Capone et al., 2011, 2005; Ji et al., 2011; Salehi et al., 2018
Childhood Autism Rating Scale (CARS; CARS-2)	Schopler et al., 2010	2+ years	Assess the severity of behaviors associated with ASD to differentially diagnose ASD from the other developmental disorders in the areas of relating to people, imitative behavior, emotional response, body use, object use, adaptation to change, visual response, listening response, perceptive response, fear or anxiety, verbal communication, nonverbal communication, activity level, level and consistency of intellective relations, general impressions	1 = *behavior appropriate for age level* to 4 = *severe deviance from expected behaviors of typical development*	Davis et al., 2018; Dressler et al., 2011; Kent et al., 1999

Note. Score that indicates the need for a more complete ASD diagnostic evaluation. ASD = autism spectrum disorder; DS = Down syndrome.

helpful information for treatment planning. Table 2 provides a summary of common symptomatology for ASD, some of which have also been used in samples with DS. Of the symptomatology measures, the Aberrant Behavior Checklist (ABC; Aman et al., 1985) may be the most commonly used to rate problematic behaviors associated with ASD in DS populations. Taken together, studies have shown a correlation between *DSM*-based criteria for ASD and scores on the ABC's maladaptive behavior subscales in children with DS (Capone et al., 2005; Ji et al., 2011; Salehi et al., 2018). Children with DS who have a higher severity of maladaptive behaviors on the ABC may be at a heightened risk for comorbid ASD.

After the Dual Diagnosis Is Made

The identification of ASD within individuals with DS impacts the educational, medical, and therapeutic interventions accessible to the individual. Without a dual diagnosis of DS + ASD, these individuals may not be offered certain treatments that have been developed to target the symptoms and behaviors associated with ASD. These supports may be critical in allowing the individual to effectively engage in activities of daily living. Those with DS + ASD are more likely to be referred to beneficial services associated with ASD (CDC, 2020), such as applied behavioral analysis, social interventions, sensory integration therapy, and augmentative and alternative communication (AAC) systems (e.g., Picture Exchange Communication System, Paterson, 1999; speech-generating devices, etc.). Overall, increased access to supports for individuals with DS + ASD is beneficial to daily functioning (Reilly, 2009).

A Challenge to Receiving DS + ASD Diagnosis: Parental Reactions

Some professionals who suspect ASD in a child with DS may be hesitant to suggest an additional developmental disorder, fearing the reaction of the parents (Capone, 1999; Reilly, 2009). They may feel that, by withholding their suspicions about the presence of comorbid ASD, they are protecting parents. However, this could be considered unethical and is to the detriment of the child who would not receive the most appropriate treatments. Fear is a natural response and to support the individual and the parents, it is important to explore what is driving these feelings or why they feel this way. Some parents of children with DS may find relief when their child receives a diagnosis of ASD, as it allows for a better understanding of their child (Moss & Howlin, 2009). By pairing the individual's presenting profile with a name—DS + ASD—parents can begin to understand their child's unique needs. They can better accommodate these needs, read cues, and be directed

to relevant resources and research when the diagnosis is made. Giving a diagnosis a name allows families to seek the support they need within their communities. When the dual diagnosis is made, SLPs can support parents by helping them to access resources and research that can best inform and support the family and the child (see Appendix for an informative resource for parents).

Implications for Speech-Language Therapy

Individuals with DS + ASD may exhibit greater developmental delays and developmental plateaus in social communication and social–emotional reciprocity than those with DS alone (Channell et al., 2019; Molloy et al., 2009). Additionally, individuals with DS + ASD may present with more severe intellectual disabilities, higher rates of stereotyped behaviors, and increased irritability, anxiety, and rigidity in communication or routine (Godfrey et al., 2019; Molloy et al., 2009; Moss et al., 2013; Philofsky, 2008). It is important to understand that clinical approaches often used for DS or ASD profiles may not be adequate for an individual with DS + ASD. An SLP providing services to a child with DS + ASD must recognize that some characteristics overlap between DS and ASD (such as language impairments and intellectual disability) and other characteristics may be more distinct between the two disorders. The intersection of these profiles, as described above, should be considered when developing assessment and treatment plans.

In addition to gathering information about medical history, behaviors, speech, expressive language, receptive language, and social communication for clients with DS + ASD, SLPs should also consider social relatedness, imitation, pragmatics, play, and nonverbal communication to best understand the individual's unique repertoire of communication strengths and weaknesses. While this may seem like a standard assumption when providing services, it is easy to assume certain patterns of behavior based on prior experience with DS or ASD, but the co-occurrence of ASD in DS is likely to lead to deviations from both of these individual profiles. For example, for children with DS alone, a clinician may expect the child to already have foundational skills for social interaction (coordinated eye gaze, gestures, etc.) but may lack the ability to apply these skills appropriately (Fidler et al., 2007; Luyster et al., 2011). However, for a child with DS + ASD, the SLP may have to first directly target foundational skills (i.e., prelinguistic communication skills), including directed eye-gaze, gestures, facial expressions, joint attention, and shared enjoyment as treatment targets. In doing so, an individual's relative strengths can promote growth in areas of difficulty targeted in speech-language therapy. These goals should be functional and target the fundamental skills necessary to promote the child's successful engagement with others in activities of daily life.

To address likely impairments in social communication and emotional reciprocity (Hepburn et al., 2008; Howlin et al., 1995; Molloy et al., 2009), the SLP can develop goals targeting the child's social communication and interaction skills. Starting first with their prelinguistic skills while children with DS only demonstrate relative strengths in prelinguistic communication, based on the available research in children with DS + ASD, there are likely early impairments in these skills (Hahn et al., 2020; Hepburn et al., 2008). Recognizing the repertoire of prelinguistic communication skills the child possesses will provide the SLP with a platform for adding linguistic input (Harris et al., 1988). The SLP can implement one of many intervention programs that support these foundation skills (e.g.,Hanen program; Girolametto & Weitzman, 2006; responsivity/prelinguistic milieu teaching, Fey et al., 2006; Olswang et al., 2006; Yoder & Warren, 2002) that teach caregivers to recognize the child's communicative intent and map language onto these communicative offers. When a parent or SLP puts the child's communicative act into words, the process can motivate the child to increase patterns of communication when the child's interest or request is recognized, validated, and met (Olswang et al., 2006; Yoder & Warren, 1999). The SLP can further promote communication acts through sabotage or the process of withholding the desired object until the child intentionally communicates.

Once foundational skills are established, the SLP may work with the child to continue advancing social communication skills. The SLP may target social communication by providing direct instruction related to the interpretation of social cues (e.g., such as body language, voice pitch or tone, and facial expressions), instruction on how to initiate and maintain appropriate communication topics (e.g., such as asking questions, making comments, and actively listening), as well as social skills training in group settings (how to navigate conflict with others, how to interpret figurative language, how to write appropriate e-mails, etc.). The SLP may also promote social acceptance by interpreting behaviors of the individual with DS + ASD for peers and/or teach the individual to advocate for him/herself in social contexts. The SLP should provide social and pragmatic communication treatment within contexts relevant to the individual's life, such as child playgroups, routines within the home, or community organizations and businesses.

Additionally, the SLP may consider implementing an AAC system as a temporary or permanent communication mode to augment or replace spoken language when it is limited. Alternative and augmentative communication is an effective tool to promote vocabulary expansion because (a) high-tech systems can generate spoken words and (b) both low-tech and high-tech systems can serve as a platform for language modeling by a verbal partner (Holyfield et al., 2019). When the child selects a symbol, the SLP should provide linguistic mapping and follow through with the request to increase the child's motivation to initiate communication (Holyfield et al., 2018).

The above methods can be incorporated into a variety of contexts, such as therapeutic play to ask questions, make comments, or elicit requests. For example, an SLP can engage the child in a play scenario involving toy cars. The SLP may roll a car out of the reach of the child (sabotage) and then wait for the child to gesture, vocalize, or indicate "car" via AAC selection. The SLP can then linguistically map onto

the child's request (i.e., "Do you want the car?" "Here is the car"). They can also model the request on the AAC device for the child (i.e., selecting symbols to say "I want the car"). Finally, the SLP validates the child's communication by following through with the request (i.e., returning the car to the child). The SLP can teach parents how to follow similar procedures to promote communication while playing or during other routines at home.

Conclusions

ASD is reported in 7%–42% of individuals with DS. SLPs are key figures in identifying early signs of ASD in DS. Based on the available research, early signs of ASD in DS that SLPs can be on the lookout for include impairments in social and emotional reciprocity and communication and higher rates of stereotyped behaviors that are greater than would be expected for a child with DS (Capone, 1999; DiGuiseppi et al., 2010; Godfrey et al., 2019; Hepburn et al., 2008; Howlin et al., 1995; Molloy et al., 2009; Moss et al., 2013). However, SLPs need to be familiar with common patterns of social communication and behavior in DS to be able to identify ASD risk in children with DS. While there is no one generally accepted diagnostic tool used to determine the presence of ASD in individuals with DS, having an interdisciplinary diagnostic team that is familiar with the profile of ASD and DS is necessary for accurate diagnosis in this population. SLPs are key members of these diagnostic teams. Receiving a dual diagnosis may allow certain educational, medical, and therapeutic services to become more accessible to families. Additionally, SLPs can support families by advocating, educating, and directing them to supportive resources. All individuals with DS + ASD have unique profiles of strengths and weaknesses, and the clinicians must understand and incorporate both when providing treatment to these individuals.

Author Contributions

Theresa M. Versaci: Conceptualization (Lead), Writing – original draft (Lead), **Laura J. Mattie:** Conceptualization (Supporting), Supervision (Lead), Writing – review & editing (Lead), **Laura J. Imming:** Conceptualization (Supporting), Writing – review & editing (Supporting).

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Appendix

Down Syndrome and Autism Spectrum Disorder Comorbidity: Addressing Questions of Parents

Versaci, T. M., Mattie, L. J., & Imming, L. J. (2020). Down syndrome and autism spectrum disorder dual diagnosis: Important considerations for speech-language pathologists. *American Journal of Speech-Language Pathology*. Advance online publication. https://doi.org/10.1044/2020_AJSLP-20-00050

This Q and A resource is intended to provide parents with some foundational knowledge regarding the diagnosis of autism spectrum disorder in individuals with Down syndrome (DS + ASD). This resource may serve as a launching point for parents to begin addressing concerns with relevant clinical and medical professionals.

What is DS + ASD?

DS + ASD is the dual presentation of two neurodevelopmental disorders: autism spectrum disorder and Down syndrome. In other words, DS + ASD occurs when an individual with Down syndrome also falls on the autism spectrum.

What's the difference between DS and DS + ASD?

Individuals with DS often use nonverbal social communication (gestures, eye gaze) with ease. For those with DS + ASD, these skills may be more difficult. Individuals with DS + ASD may have more severe intellectual impairments than those with DS without autism spectrum disorder. Additionally, individuals with DS + ASD are at an increased risk of anxiety, irritability, difficulty with transitions, hyperactivity, withdrawal and attention problems, sleep disturbances, sensory-related feeding problems, and restrictive-repetitive behaviors (i.e., teeth grinding, hand flapping, fixation on sensory input).

What is the likelihood my child with DS has DS + ASD?

It is currently estimated that between 7 and 42% of individuals with DS also have ASD. An individual with DS may be more likely to have ASD if there is a familial history of ASD in first- or second-degree relatives, infantile spasms or seizures, early hypothyroidism, and/or brain injury secondary to complicated surgeries.

What signs of DS + ASD should I be looking out for?

Early signs to look for include: limited social communication, repetitive motor behaviors, fixation on lights and spinning objects, oral texture aversion, lack of response to auditory language input, and/or repetitive or absent spoken language. If your child is older, you may look out for regression or plateauing language and social skills. You may also notice increased irritability, anxiety, and the onset of repetitive behaviors in these older individuals.

How do I know if my child does have DS + ASD?

It is the job of clinical professionals (i.e., clinical psychologists, behavioral pediatricians) to make the diagnosis of DS + ASD. If you are concerned your child may have DS + ASD based on the detection of early signs, request an evaluation. This evaluation will be comprehensive, including many diagnostic tools, interviews, and reviews of developmental and medical history. Gathering as much evidence as possible, the clinician may make the dual diagnosis of DS + ASD.

My child already has one diagnosis. Why would I want them to have another?

The additional diagnosis of autism spectrum disorder may allow your child to access more services. This includes educational, medical, and treatments. This includes applied behavioral analysis (ABA), social interventions, sensory integration therapy, and augmentative and alternative communication systems, medications, and medical screenings. Access to these services may help your child find more success in activities of daily life.

What if it is too late for my child to get this diagnosis?

Your child may be diagnosed at any time, so long as his profile is consistent with developmental patterns associated with DS + ASD.

How can I best advocate for my child?

You know your child best. You are tuned into how your child communicates, what your child likes, and what behaviors are expected based on his personality. If you notice unexpected behaviors and are concerned, seek out an evaluation. Look for professionals with a strong knowledge base in both autism spectrum disorders and Down syndrome. The professionals you work with, such as speech-language pathologists, should be open-minded and able to see your child as a unique individual, with unique strengths and weaknesses.

For more information, please see:
Your local DS, ASD, or DS + ASD network community
http://www.ds-asd-connection.org/
https://www.ndss.org/