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Practice Patterns and Barriers in the Assessment and Treatment of Autism Spectrum Disorder in Children With Down Syndrome

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ABSTRACT

Background: Autism spectrum disorder (ASD) is common in individuals with Down syndrome (DS), with an estimated prevalence of 16%–18%. However, receiving a dual diagnosis of Down syndrome and ASD (DS + ASD) is often delayed. Little evidence exists on the path to ASD diagnosis nor interventions to support individuals with DS + ASD. Barriers to diagnosis and treatment for this unique patient population have yet to be described. This study explores clinicians' practices and perceptions regarding the diagnosis and treatment DS + ASD, and the barriers their patients face in connecting to recommended evaluations and services.

Methods: The study used an anonymous web-based survey developed by a group of physicians, psychologists and researchers who work with individuals with DS, ASD and DS + ASD. The survey queried clinicians from various specialties about their practice patterns regarding assessment of suspected ASD in individuals with DS. The survey inquired about treatment recommendations for DS + ASD and perceived barriers to connecting families with evaluations and services. Data analysis involved descriptive statistics and Mann–Whitney *U* tests.

Results: Most respondents believe diagnosing ASD in individuals with DS significantly impacts management. Challenges were reported in accessing diagnostic evaluations, with heavy reliance on highly specialised DS and ASD clinics. Communication impairment ($n = 64$, 65%), aggressive behaviours ($n = 38$, 39%), self-injurious behaviours ($n = 33$, 34%) and adaptive skills ($n = 27$, 28%) are priority targets for intervention, and applied behavioural analysis (ABA) ($n = 80$, 82%), speech therapy through insurance ($n = 60$, 61%), augmentative and alternative communication evaluation through insurance ($n = 59$, 60%), and occupational therapy through insurance ($n = 57$, 58%) are the most frequent referrals following a diagnosis of DS + ASD. All respondents identified multiple barriers to care for individuals with DS + ASD, including waitlists, insurance networks and requirements, lack of experienced providers and high turnover.

Conclusion: This study highlighted the complexity of caring for individuals with DS + ASD, revealed the heterogeneity of practice patterns among providers, and reported multiple barriers to care for this underserved patient population. Results should

prompt work aimed at redressing barriers to care and additional research in the field of effective interventions for individuals with DS + ASD.

1 | Background

Down syndrome (DS) is the most common chromosomal cause of intellectual disability (Lee et al. 2023); the estimated prevalence of DS in the United States is 1 in every 640 births (Presson et al. 2013; Stallings et al. 2024). Autism spectrum disorder (ASD) is a neurodevelopmental condition characterised by persistent deficits in social communication and the presence of repetitive behaviours and/or intense restricted interests (American Psychiatric Association 2013). ASD co-occurs in 12%–41% of individuals with DS (Diniz et al. 2022), with meta-analysis data estimating a prevalence of 16%–18% (Richards et al. 2015). These estimates are approximately four to thirteen times the estimated prevalence of ASD in the general population, which is currently 1 in 31 children aged 4–8 years (Shaw et al. 2025). In recent years, there has been a growing effort to determine appropriate screening and diagnostic tools to identify individuals with the dual diagnosis of Down syndrome and autism (DS + ASD) (Baumer et al. 2024; Channell 2020; DiGuiseppi et al. 2010; Diniz et al. 2022; Hahn et al. 2020; Magyar et al. 2012; Pisula and Niedźwiecka 2021), describe their developmental and behavioural phenotype (Patel et al. 2025), identify co-occurring conditions (Harvey et al. 2022; Molloy et al. 2009; Rubenstein et al. 2024; N. A. Spinazzi, Santoro, et al. 2023) and describe the experience of their caregivers (Soccorso et al. 2023; N. Spinazzi, Velasco, et al. 2023).

1.1 | Diagnostic Delays

Receiving the initial dual diagnosis of ASD is often delayed in individuals with DS (N. Spinazzi, Velasco, et al. 2023). Clinicians with expertise in caring for this patient population speculate that this delay is attributable to a combination of diagnostic overshadowing (i.e., attributing all differences to DS and intellectual disability), lack of validated diagnostic tools, providers' hesitancy to suggest an additional diagnosis and multiple logistical barriers to accessing specialised evaluations.

Literature on idiopathic ASD suggests that proximity to a medical school, proximity to a neurology or psychiatric specialist, and a primary care provider's ability to refer to a nearby specialist affect time to diagnosis (Daniels and Mandell 2014). Some children with DS live in proximity to DS clinics, while many others do not (Joslyn et al. 2020). Little is known about the journey from clinical suspicion for DS + ASD to diagnosis, including information about steps that clinicians who interact with individuals with DS take if they suspect co-occurring ASD.

1.2 | Interventions for DS + ASD

Based on the described developmental profile and characteristics of those with DS + ASD (Baumer et al. 2024; Magyar et al. 2012; Patel et al. 2025; Warner et al. 2014), there are many potential targets for intervention including communication skills, decreased social reciprocity, elevated levels of hyperactivity, anxiety, stereotyped behaviours and maladaptive behaviours including self-injury, sensory integration dysfunction and difficulties in adaptive functioning. However, with little known about the effectiveness or efficacy of developmental, behavioural and psychopharmacologic interventions for individuals with DS + ASD, there are no evidence-based guidelines to help providers with appropriate referrals, leaving clinicians with no choice but to recommend interventions that have been studied in individuals with DS only or ASD only. For example, although there is recent controversy surrounding its use (Schuck et al. 2022), applied behaviour analysis (ABA) and/or ABA principles are widely used evidence-based interventions targeting challenging behaviours in children with DS or ASD (Alves et al. 2020; Gitimoghaddam et al. 2022; Granpeesheh et al. 2009; Ivy and Schreck 2016; Neil et al. 2021; Newcomb and Hagopian 2018; Peters-Scheffer 2011). Less research has been conducted with children with DS + ASD (Oxelgren, Westerlund, et al. 2019). Similarly, although there is published evidence on the effectiveness of various communication interventions for individuals with DS only (Neil and Jones 2018; O'Toole et al. 2018; Smith et al. 2020) and ASD only (Bejarano-Martín et al. 2020; Osman et al. 2023), only one case report has been published documenting improvement in communication skills in a child with DS + ASD following a naturalistic speech and language intervention (Kroeger and Nelson 2006). Psychopharmacologic interventions are also poorly studied in this underserved population, as outlined in a recent review (Baumer and Capone 2023). The lack of evidence on effective interventions for individuals with DS + ASD is a set up for heterogeneity in referrals among providers caring for individuals with DS + ASD, though little is known about clinicians' practice patterns or clinicians' perception of helpfulness of available therapeutic interventions.

1.3 | Barriers to Care

Caregivers face many potential barriers to both accessing diagnostic evaluations and interventions for individuals with DS + ASD which, in turn, affect access to the support services needed for daily living and optimal quality of life in this population. For individuals with ASD without DS, socioeconomic (Dickerson et al. 2017), linguistic (Bivarchi et al. 2021; Lim et al. 2021; St Amant et al. 2018), sociocultural (Ahmed and Farooq 2020; Fisher et al. 2023) and geographic (Daniels and Mandell 2014; Drahota et al. 2020; Liu et al. 2023; Mello et al. 2016) factors have been reported as barriers influencing disparate evaluation, diagnosis and treatment. However, it remains unknown if these barriers are also encountered by individuals with DS + ASD. It is essential to investigate barriers faced by this population in order for clinicians to provide adequate support to caregivers, and for policymakers to address them at a systems level.

1.4 | Study Aims

Little is known about practice patterns of clinicians in various specialties caring for individuals with DS in whom co-occurring ASD is suspected or confirmed. Similarly, there is little published evidence on the barriers that clinicians and caregivers encounter as they navigate services for this patient population. To move forward, it is important to understand the experiences of providers who work with individuals with DS in a variety of settings. This study aimed to explore clinicians' practice patterns in the diagnosis and treatment of DS + ASD and, when possible, evaluate group differences in practices between different types of providers. The study also sought to evaluate providers' perceptions of the helpfulness of various interventions related to core impairments in DS + ASD. The study intended to identify barriers to evaluation, diagnosis and referral to services for treatment in this population.

2 | Methods

2.1 | Ethics Statement, Funding and Data Availability

This study involved human subjects and adhered to ethical principles. The research was undertaken with consent of each participant. The study was reviewed and approved by the Children's Hospital Los Angeles/University of Southern California institutional review board (ID: CHLA-24-00190). This research received no specific grant from any funding agency in the public, commercial or not-for-profit sectors. Anonymised data are available to qualified researchers upon request.

2.2 | Study Design

An anonymous, prospective, web-based survey was designed to assess clinician attitudes towards the dual diagnosis of ASD in children with DS. The development of this survey was organised by the Down Syndrome Medical Interest Group (DSMIG) DS + ASD workgroup in June 2024. The workgroup was composed of seven physicians, psychologists and researchers who work with individuals with DS, ASD and DS + ASD. Questions were developed based on the decades of combined patient-care experience of the professionals in the workgroup. The survey (Supplementary Information A) included 40 questions, some with multiple-choice answers and some with 5-point Likert scale ratings that included a 'not applicable' option; the survey ended with one open-ended question. The measure was divided into three sections: (1) demographic and practice information of the respondent; (2) practice patterns when a patient with DS is suspected to have co-occurring ASD, including barriers to diagnostic evaluations; and (3) referral patterns for a patient with DS + ASD, including perceived barriers that families face trying to access services. All questions required majority approval by the workgroup (> 50%) for inclusion in the survey. Inclusion criteria for the study included being able to read and answer questions in English, be over 18 years of age and have cared for individuals with DS.

2.3 | Solicitation and Dissemination

Study data were collected and managed using REDCap electronic data capture tools hosted at Children's Hospital of Los Angeles. REDCap (Research Electronic Data Capture) is a secure, web-based software platform designed to support data capture for research studies (Harris et al. 2019; Harris et al. 2009). The survey was advertised to physicians, clinicians, psychologists and researchers through email listservs and through direct solicitation at the DSMIG annual meeting that took place in Phoenix, Arizona, in July 2024. Professional groups with practice areas including paediatrics, paediatric and adult psychology, development and neurodevelopment were approached about disseminating the surveys to their listservs or through other contact methods that they utilise with their members. A full list of professional societies who allowed dissemination is presented in Supplementary Information B. Recruitment efforts attempted to reach a broad range of clinicians, including those who care for both children and adults, because ASD is not uncommonly diagnosed in individuals with DS who have already graduated out of paediatric care, and because behavioural and developmental services are often indicated and prescribed throughout the lifespan. Outreach was inclusive of therapy providers because though they do not directly diagnose ASD, they may be the first to suspect a dual diagnosis in a patient with DS or be the first clinician to whom caregivers disclose their concerns (N. Spinazzi, Velasco, et al. 2023). Workgroup members' affiliation to professional organisations, many of which were DS-focused, facilitated recruitment outreach. Emails provided a brief summary of the study, QR code, and a direct link to access the study. The survey was disseminated from 7/24/2024–11/08/2024. After reviewing information about the study, participants consented to participation by proceeding with the survey. Participants received no compensation for participating in the study.

2.4 | Data Validation

Only completed surveys were included for analysis. To avoid duplicate surveys from the same participant (who may have received two or more solicitations for response), IP addresses were reviewed for potential duplication and removed if the IP address was identical for more than one response; this reduced, but did not eliminate, the possibility of duplicate responses.

2.5 | Statistical Analysis

Descriptive statistics were utilised to detail respondents' demographics, clinical practice setting and responses to other survey questions. When possible, group differences were also evaluated. To facilitate interpretation of ordinal data, Likert scale responses were dichotomised into *difficult* ('Very difficult' and 'Somewhat difficult' responses) or *not difficult* ('Neutral', 'Somewhat easy', 'Very easy'). Mann–Whitney *U* tests were conducted to test for group differences when cell sizes were large enough to permit statistical comparisons. A post hoc sensitivity power analysis (using G*Power 3.1) was conducted to characterise the minimum detectable effect size. Assuming an alpha of 0.05 and power of 0.80 for between-group comparisons (e.g.,

TABLE 1 | Demographic and clinical practice data.

	n (%)
Primary professional role	
Physician	76 (78%)
Psychologist	9 (9%)
Nurse practitioner	4 (4%)
Occupational therapist	2 (2%)
Physical therapist	2 (2%)
Speech therapist	1 (1%)
Social worker	1 (1%)
Researcher	1 (1%)
Other (BCBA, nurse coordinator)	2 (2%)
Geographic location of clinical practice	
Urban	71(72%)
Suburban	24 (24%)
Rural	3 (3%)
Region of practice	
South (TX, OK, AK, LA, MS, AL, GA, FL, NC, SC, TN, KY, VA, WV)	27 (27%)
West (CA, AK, HI, OR, WA, NV, ID, UT, AZ, NM, CO, WY, MT)	24 (24%)
Northeast (MD, DE, PA, NY, NJ, CT, MA, RI, NH, VT, ME)	17 (17%)
Midwest (ND, SD, NE, KS, MO, IA, MN, WI, MI, IL, IN, OH)	2 (2%)
Age group of primary population of practice	
Paediatric (0–18 years)	74 (76%)
Transition-aged youth	0 (0%)
Adult (18 + years)	6 (6%)
Older adults (50 + years)	0 (0%)
All age groups	18 (18%)
Work within a DS clinic	
Yes	43 (44%)
No	55 (56%)
Work within an ASD clinic	
Yes	37 (38%)
No	60 (62%)
Did not respond	1 (1%)
Type of clinical practice[a]	
Academic medical centre	76
Sub-specialty clinic	20
Outpatient primary care—hospital-affiliated	20
Community hospital	6
Outpatient primary care—private practice	5
Outpatient primary care—federally qualified health centre	4
Outpatient therapy (e.g., PT, OT, ST)	3
K-12 schools	2
Early childhood education	1
Community mental health practice	1
Governmental hospital (VA/Military)	1
Other	3

Note: Number and percent of individuals surveyed according to various categories.
Abbreviations: BCBA, board-certified behavioural analyst; OT, occupational therapy; PT, physical therapy; ST, speech therapy; VA, Veterans Affairs.
[a]Thirty-five respondents (36%) reported working in more than one setting.

ANOVA or χ^2 tests across clinician types or diagnostic focus), the sample size was determined sufficient to detect medium-to-large effect sizes (Cohen's $f \geq 0.30$ or Cramer's $V \geq 0.25$).

3 | Results

In total, 167 individuals accessed the survey's eligibility screening questions. Of those individuals, 50 (30%) did not complete the eligibility questions and 11 (7%) self-reported that they were ineligible (were not clinicians who treat individuals with DS), leaving 106 individuals who completed the survey (63%). There were no duplicate responses from the same IP address. Of those who completed the survey, a small number of respondents reported that they practice outside the United States ($n = 8$, 5%). Due to this small number and the differences in systems and structures of care in other countries, only individuals who provide care in the United States were included in the final sample ($n = 98$, 92%). In the final sample of 98 respondents, the majority were physicians ($n = 76$, 78%), and the most common work environment was an academic medical centre ($n = 76$, 78%); 43 (44%) respondents reported working in a DS clinic, and 37 (38%) reported working in an ASD clinic, with some working in both settings. Demographic and clinical practice data are summarised in Table 1. All results are reported out of the full sample of 98 respondents unless otherwise specified.

3.1 | Practice Patterns From Diagnosis to Intervention

Nearly all respondents ($n = 94$, 96%) agreed or strongly agreed with the statement: 'Diagnosing co-occurring autism in children with Down syndrome has an important impact on management'. However, as reported below, survey responses revealed

challenges with accessing a diagnosis, disparate patterns of referrals to evaluations and therapeutic supports, and multiple barriers to accessing both.

3.1.1 | Accessing a Diagnosis

When asked about practice patterns once they suspect a diagnosis of ASD in a person with DS, almost half of respondents ($n = 48$, 49%) reported that they were able to make a diagnosis within their clinic, either by making a diagnosis themselves ($n = 26$, 27%) or referring to a psychologist on their team ($n = 22$, 22%). Of those who reported the ability to make an in-clinic diagnosis, most ($n = 31$ of 48, 65%) worked in a specialty clinic: DS and/or ASD clinic.

Of the 50 other respondents who did not report making an in-clinic diagnosis, over half (62%) indicated they refer patients to highly specialised services (e.g., DS clinic, ASD clinic/centre or DS + ASD clinic; $n = 24$ of 50) or a psychologist in the community with known expertise in DS + ASD ($n = 7$ of 50). The remaining 38% reported they rely on referring for additional evaluations through insurance ($n = 8$) or the educational system ($n = 1$), feel unsure about next steps ($n = 3$) or use other referral strategies (e.g., primarily work with adults so make referrals to psychiatrists if complex presentation; referral depends on age or family constraints, $n = 7$). As expected, all therapy provider respondents endorsed referring to other clinicians for additional evaluation. Notably, only one respondent reported never having suspected ASD in a patient with DS.

Across the full sample, some participants agreed or strongly agreed that they sometimes hold back on discussing concerns for co-occurring ASD because of their own lack of expertise ($n = 18$, 18%) or because of concern for how the family might react ($n = 17$, 17%). For the 35 who reported holding back on an ASD diagnosis, there was no difference in whether or not the respondent worked in a DS clinic ($W = 1156$, $p = 0.3115$).

3.1.2 | Referrals for Therapeutic Intervention and Other Supports

We also explored clinicians' referral patterns for addressing developmental and behavioural concerns once their patient received a diagnosis of DS + ASD. When asked to select the top 3 behavioural and developmental concerns that they target/recommend via referrals, the most commonly endorsed were communication impairment ($n = 64$, 65%), aggressive behaviours ($n = 38$, 39%), self-injurious behaviours ($n = 33$, 34%) and adaptive skills ($n = 27$, 28%).

When asked where they refer their patient with a new diagnosis of DS + ASD, respondents selected a variety of services. Out of the 19 options, respondents endorsed referring patients to up to 16 resources ($M = 7.33$ resources endorsed per clinician). See Figure 1. The most endorsed services were ABA ($n = 80$, 82%), speech therapy through insurance ($n = 60$, 61%), augmentative and alternative communication evaluation through insurance ($n = 59$, 60%), and occupational therapy through insurance ($n = 57$, 58%). In a free response 'other' option, respondents noted referring individuals to physical therapy to promote mobility and toilet learning, local resources (e.g., DS associations, a DS-ASD Parent Connection), and education advocacy.

We asked participants what resources for caregiver support they discuss with families of individuals with DS + ASD: out of the 10 options listed, respondents endorsed referring caregivers to up to 9 resources ($M = 3.63$ resources endorsed per clinician). See Figure 2. In a free response 'other' option, respondents specified Home and Community-Based Services waivers, Supplemental Security Income waivers, and a Local Children's Board.

Respondents were also asked about what resources they perceived to be most helpful in targeting communication and self-injurious behaviours (common support needs in this population). Speech therapy ($n = 62$, 63%), augmentative and alternative communication ($n = 58$, 59%), and ABA ($n = 55$, 56%) were the most commonly endorsed resources for targeting communication. Psychotropic medications ($n = 53$, 54%), ABA ($n = 77$, 79%) and behavioural parent training ($n = 40$, 41%) were the most commonly endorsed resources for targeting self-injurious behaviour.

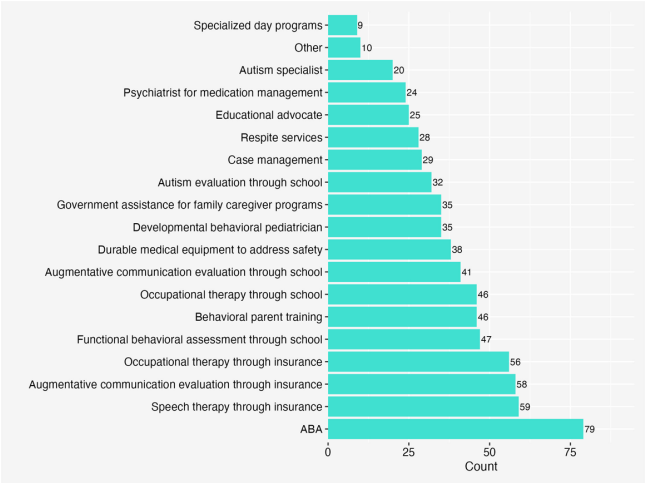

FIGURE 1 | Referrals for a new dual diagnosis of DS + ASD.

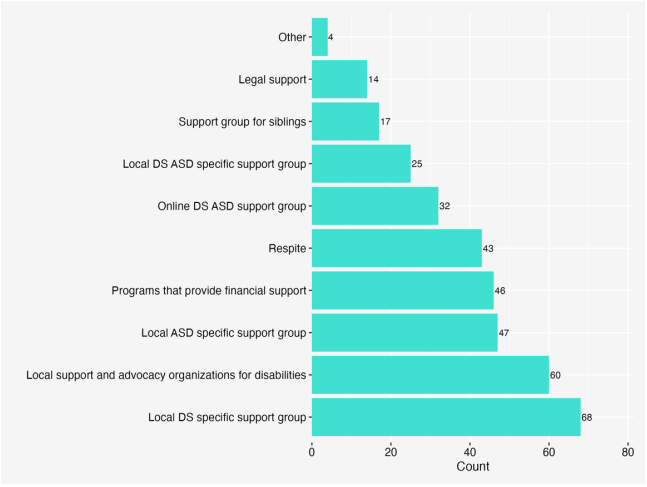

FIGURE 2 | Caregiver resources discussed with families.

3.2 | Barriers

3.2.1 | Level of Difficulty in Connecting Patients to Different Services

Clinicians were asked to rate the level of difficulty in connecting patients to different services. The services to which respondents most often reported as *difficult* to connect families to were respite care ($n = 75$, 77%) and psychiatrists ($n = 73$, 74%) or ($n = 72$, 73%) psychologists who can conduct assessments. In contrast, the services to which respondents most often reported as *not difficult* to connect with were occupational therapists for treatment ($n = 56$, 57%) or assessments ($n = 51$, 52%) and speech therapists for treatment ($n = 50$, 51%) or assessments ($n = 48$, 49%).

3.2.2 | Barriers to Patient Access of Specialty Services

Clinicians were asked to report common barriers their patients experience when attempting to access specialty evaluations and specialty therapies, specifically ABA or other (e.g., speech therapy, occupational therapy, or augmentative and alternative communication support). Table 2 outlines participants' responses. For specialty evaluations, the most commonly reported barriers were waitlists ($n = 88$, 90%), obstacles related to insurance coverage ($n = 47$, 48%) and a lack of access to specialists to refer to ($n = 32$, 33%). For ABA, the most commonly reported barriers were waitlists ($n = 94$, 96%), inexperienced providers ($n = 63$, 64%) and high provider turnover ($n = 58$, 59%). For other specialty therapies, the most commonly reported barriers were waitlists ($n = 83$, 85%), a lack of experienced providers ($n = 65$, 66%) and obstacles related to insurance ($n = 63$, 64%).

When asked, 'What else do you want to share regarding your experience evaluating and treating individuals with suspected or confirmed DS+ASD?' as a free-text response, respondents articulated even further challenges. Providers reported extreme difficulty accessing ASD evaluations for adults with DS; inadequate ability by general autism evaluators to recognise signs of ASD in a child with co-occurring DS, who frequently attribute presenting concerns to 'global developmental delay'; and financial barriers to sustaining DS clinics, where clinicians may feel most comfortable with providing a DS + ASD diagnosis. Furthermore, some respondents stated that the more complex presentation of DS + ASD may include a need for psychiatric intervention to address aggression or self-injurious behaviours, which is also difficult to obtain.

4 | Discussion

This study aimed to explore self-reported practice patterns and perceptions among clinicians in the diagnosis and management of co-occurring ASD in individuals with DS. Our findings revealed a heavy reliance on specialised care such as through a DS or ASD clinic for making a diagnosis of ASD within those with DS. Survey respondents also identified critical areas of need in the population of individuals with DS + ASD including interventions targeting communication, aggressive and self-injurious behaviours, and adaptive skills, as well as resources specific to those with DS + ASD, including clinicians with direct expertise treating this population. Respondents endorsed multiple perceived barriers to evaluation and therapeutic services including long waitlists and lack of available providers, insurance barriers, language and distance to services. These are similar barriers to those reported in other studies of individuals with DS (Joslyn et al. 2020; McCabe et al. 2011) and ASD (Daniels and Mandell 2014).

In the current study, survey respondents predominantly worked in the context of academic medical centres, with many working in the context of a DS clinic, ASD clinic or both. However, only

TABLE 2 | Clinician endorsements of family barriers to different services.

	Barriers to specialty evaluations, *n (%)*	Barriers to ABA, *n (%)*	Barriers to other specialty therapies, *n (%)*
Distance	27 (28%)	38 (39%)	40 (41%)
Insurance barriers	47 (48%)	48 (49%)	63 (64%)
Language	29 (30%)	41 (42%)	38 (39%)
Lack of available referrals	32 (33%)	30 (31%)	23 (23%)
Lack of follow through on referral by caregiver	28 (29%)	39 (40%)	32 (33%)
Inability to advocate for services through the IEP	N/A	N/A	45 (46%)
Lack of available providers/ long waitlists	88 (90%)	94 (96%)	83 (85%)
High provider turnover	N/A	58 (59%)	N/A
Lack of experienced providers	N/A	63 (64%)	65 (66%)
Other barriers	6 (6%)	3 (3%)	1 (1%)

Note: Number and percentage (out of all 98 respondents) who reported each barrier. N/A indicates that this barrier was not one of the listed options.
Abbreviations: ABA, applied behaviour analysis; IEP, individualised education program.

half reported the ability to make the diagnosis of ASD themselves or via a referral within their specialty clinic. Despite the above-average expertise represented in the respondent sample, there was significant heterogeneity in practice patterns once ASD was suspected in an individual with DS. These differences can be confusing to caregivers and providers alike and can serve as an additional barrier to diagnosis while contributing to the wide time gap between when concerns first arise and when a diagnosis is obtained (N. Spinazzi, Velasco, et al. 2023). Many respondents utilised the expertise of DS clinics when a diagnosis of ASD was suspected. This practice is unlikely to be generalisable, as recent data indicate that 35%–44% of individuals with DS do not live within 2 h of a DS clinic, and these clinics often have long waitlists (Jensen et al. 2024). Additionally, despite our survey respondents almost universally agreeing that it is important to diagnose and provide therapeutic supports for co-occurring ASD in those with DS, many reported occasional hesitancy in discussing a dual diagnosis with caregivers due to concerns that they lacked expertise or fear of how a family may react to the conversation. Taken together, these findings highlight both the need to increase access to specialised DS and ASD centres and expand education about DS + ASD among community providers in order to decrease reliance on these highly specialised services. Education of frontline clinicians is crucial, as caregivers will often express their initial concerns to their primary care provider, only to find that most do not have the knowledge or experience about DS + ASD to further guide them (N. Spinazzi, Velasco, et al. 2023).

In this survey, clinicians identified communication impairment, aggressive and self-injurious behaviours, and adaptive skills as the top three targets for intervention. This is consistent with challenges most frequently identified by caregivers of individuals with DS + ASD, namely communication deficits and mood and behavioural issues (N. Spinazzi, Velasco, et al. 2023), and thus reinforces the need for referral and access to targeted supports. Respondents identified speech therapy, augmentative and alternative communication, and ABA as the primary therapeutic intervention to target communication, and psychotropic medications, ABA and behavioural parent training as the most commonly endorsed resources for targeting self-injurious behaviour. Again, this is consistent with reports from caregivers, who have identified ABA therapy and therapeutic services (including speech, physical and occupational therapy) as the interventions that were most helpful for their loved one with DS + ASD (N. Spinazzi, Velasco, et al. 2023).

Despite congruency of impressions between caregivers and providers regarding the usefulness of ABA to address the most challenging symptoms in individuals with DS + ASD (Oxelgren, Aberg, et al. 2019), this study described many barriers to accessing ABA. In our survey, nearly all respondents cited long waitlists, and a majority endorsed inexperienced providers and high provider turnover as barriers to ABA. Half of respondents identified insurance as a barrier to accessing ABA services; indeed, many private insurance companies require use of specific diagnostic tools in order to provide access to evidence-based ASD services, thus deterring some experienced providers from making a clinical diagnosis themselves and further impacting access to timely intervention. These barriers are unlikely to be specific to the DS + ASD population, as similar therapies are also commonly prescribed for children with DS only and ASD only. Barriers identified in this study are similar to those reported for individuals with ASD alone such as lack of clarity of referral pathways, barriers related to insurance and access to providers with expertise for diagnosis (Malik-Soni et al. 2022). Distance to services and lack of available providers have also been identified as barriers to care for individuals with DS (Joslyn et al. 2020). More research is needed to explore similarities and differences between these three populations.

This study has several limitations that impact the interpretation and generalizability of its findings. Although attempts were made to recruit participants from a range of established listservs that include providers representing a variety of occupations/specialties, locations and other practice characteristics, the majority of respondents were from suburban/urban practice settings in the United States, with a large proportion identifying as working in a DS or ASD clinic; the possibility that multiple respondents worked within the same clinic could not be ruled out. The higher response rate by professionals who specialise in these diagnoses may have skewed the results towards reporting the perspectives of individuals who were a priori more likely to understand the complexities of the evaluation and referral process. Even so, multiple barriers to diagnosis and access to care were still identified. Generalists and primary care providers, who were under-represented in this sample, may experience greater barriers in access to diagnosis and intervention than those working in specialty clinics. The authors hope to expand the current study to include a combination of community, academic and state/federal-based generalists and primary care specialists; this will allow for enhanced understanding of whether barriers to care are similar or discrepant across unique practice areas. Another important limitation of the study is that the survey did not ask respondents to quantify the number of individuals with DS or ASD they treated nor their years of clinical experience caring for these patient populations, both factors that may influence results. Future work will ensure collection of data to quantify practitioners' degree of expertise in caring for individuals with intellectual and developmental disabilities.

Due to the under-representation of respondents practicing in rural settings, analyses of practice patterns and barriers by geographic location could not be conducted. Respondents who primarily care for adults with DS were also under-represented: Given the numerous differences in paediatric versus adult systems of care, including the medical and educational systems, this was a significant limitation that impacts the generalizability of this study's findings. Future research should target more diverse enrollment in terms of professional characteristics, to allow for more in-depth analysis. Due to the sample size and distribution of responses, the current study was underpowered to conduct nuanced regression analyses, focusing instead on dichotomous responses and descriptive statistics; it was powered to detect medium-to-large effect sizes and could not detect small statistical differences that could be clinically meaningful. Additionally, the decision to dichotomise ordinal data may have obscured distinctions, particularly for individuals who endorsed a neutral response. Future research that enrols a larger and more diverse sample of practitioners will fill this gap. In addition to the potential skew of participants, the questions asked in this prospective survey were designed by specialists in neurology,

psychiatry, and psychology, which may have resulted in more narrow querying of "perceived" problems that may not be generalisable to primary care providers. Importantly, this survey did not directly assess the effectiveness of interventions and relied on clinician perception of their helpfulness. The survey did not ask respondents to differentiate the referrals they make after diagnosing ASD in a person with DS from those they make for individuals with DS alone; many of the interventions endorsed by respondents are often prescribed to patients with DS regardless of ASD diagnosis, which complicates interpretation of the study's findings. Future work should more deliberately investigate how referral patterns differ for individuals with DS + ASD compared to those with DS or ASD alone. Finally, our study focused specifically on individuals with DS + ASD; the choice was motivated by current challenges in the field such as underidentification in community settings and lack of evidence-based provider guidelines for this population. Our data provide initial insight into providers' experiences and perceptions of access barriers. Future research should expand to directly compare these experiences to those of providers working with individuals with DS only or ASD only, in order to contextualise the unique and common barriers faced by each group. For example, future recruitment outreach should intentionally extend to ASD-specific listservs, in addition to DS-specific networks, to ensure a more balanced and representative dataset for robust group comparisons. Regardless of all its limitations, this study elucidated numerous barriers faced by individuals with DS + ASD and their families that warrant attention.

5 | Conclusion

In summary, this study surveyed clinicians who work with individuals with DS, most of whom worked in academic centres and other specialised settings and found disparate practice patterns in obtaining diagnostic evaluations for a dual diagnosis of DS + ASD. Providers identified communication, behavioural dysregulation and difficulties with adaptive skills as the primary therapeutic targets and endorsed ABA and developmental therapies as the most helpful supports for individuals with DS + ASD. Numerous barriers to care were described in this study, highlighting the urgent need to improve access to these crucial supports for this underserved population.

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The authors have nothing to report.

Conflicts of Interest

The authors declare no conflicts of interest.

Data Availability Statement

The data that support the findings of this study are available from the corresponding author upon reasonable request.

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Supporting Information

Additional supporting information can be found online in the Supporting Information section. **Supplementary Material A** Research survey. **Supplementary Material B** Participant recruitment outreach log.