

Assessing and Managing Autism in Down Syndrome: Practice Patterns and Barriers to Care

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INTRODUCTION

Autism spectrum disorder (ASD) is common in individuals with Down syndrome (DS), with a prevalence of 16-18%. Receiving a dual diagnosis of Down syndrome and ASD (DS+ASD) is often delayed, effective interventions are poorly studied, and barriers to diagnosis and treatment for this unique patient population have yet to be described.

This study explores clinicians' practices in diagnosing and treating DS+ASD, and the barriers their patients face in connecting to recommended evaluations and services.

METHODS

The Down Syndrome Medical Interest Group (DSMIG) DS+ASD workgroup, consisting of physicians, psychologists, and researchers, created an anonymous, prospective, web-based survey. This 40-item, multiple choice survey, with one open ended response, was divided into 3 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 encountered in pursuing a diagnostic evaluation
- 3) Referral patterns for a patient with DS+ASD, including perceived barriers to families accessing additional services.

Survey Dissemination:

- Email listservs to professional societies
- Direct solicitation at DSMIG annual meeting, July 2024

Survey responses were collected between 7/24/2024 and 11/8/2024. Descriptive statistics were utilized to detail respondents’ demographics, clinical practice setting, and responses to other survey questions. Mann-Whitney U tests were conducted to test for group differences (when sizes were large enough to permit statistical comparisons).

RESPONDENTS

106 professionals completed the survey; 8 were excluded because they practice outside of the US. Of the 92 remaining:

- 78% (n=76) were physicians
- 78% (n=76) work in an academic medical center
- 44% (n=43) work in a DS specialty clinic
- 37% (n=37) work in an ASD specialty clinic
- 76% (n=74) see primarily pediatric patients

RESULTS

“Diagnosing co-occurring autism in children with Down syndrome has an important impact on management” – 96% agreement
“I sometimes hold back on an ASD diagnosis” – 35% agreement
“I make an ASD diagnosis myself, or refer to someone in my practice” – 57% agreement

Most **common** developmental/behavioral concerns leading to targeted referrals:

- Communication impairment (65%)
- Aggressive behaviors (39%)
- Self-injurious behaviors (34%)
- Adaptive skills (28%)

Most helpful resources for **communication concerns**:

- Speech therapy (63%)
- Augmentative and alternative communication (59%)
- Applied Behavior Analysis (56%)

Most helpful resources for **self-injurious behaviors**:

- Applied Behavior Analysis (79%)
- Psychotropic medications (54%)
- Parent training (41%)

Most **difficult** services to access:

- Respite care (77%)
- Psychiatrists (74%)
- Psychologists, for assessment (73%)

DISCUSSION/CONCLUSION

- Despite the above-average expertise in the respondent sample, there was significant heterogeneity in practice patterns once ASD was suspected in an individual with DS.
- Despite 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.
- Providers identified communication, behavioral dysregulation and difficulties with adaptive skills as the primary therapeutic targets for individuals with DS+ASD.

RESULTS

Figure 1: *Most common referrals for new DS+ASD diagnosis.* ABA=Applied Behavior Analysis. “Other” includes physical, local resources (e.g., DS associations), and education advocacy

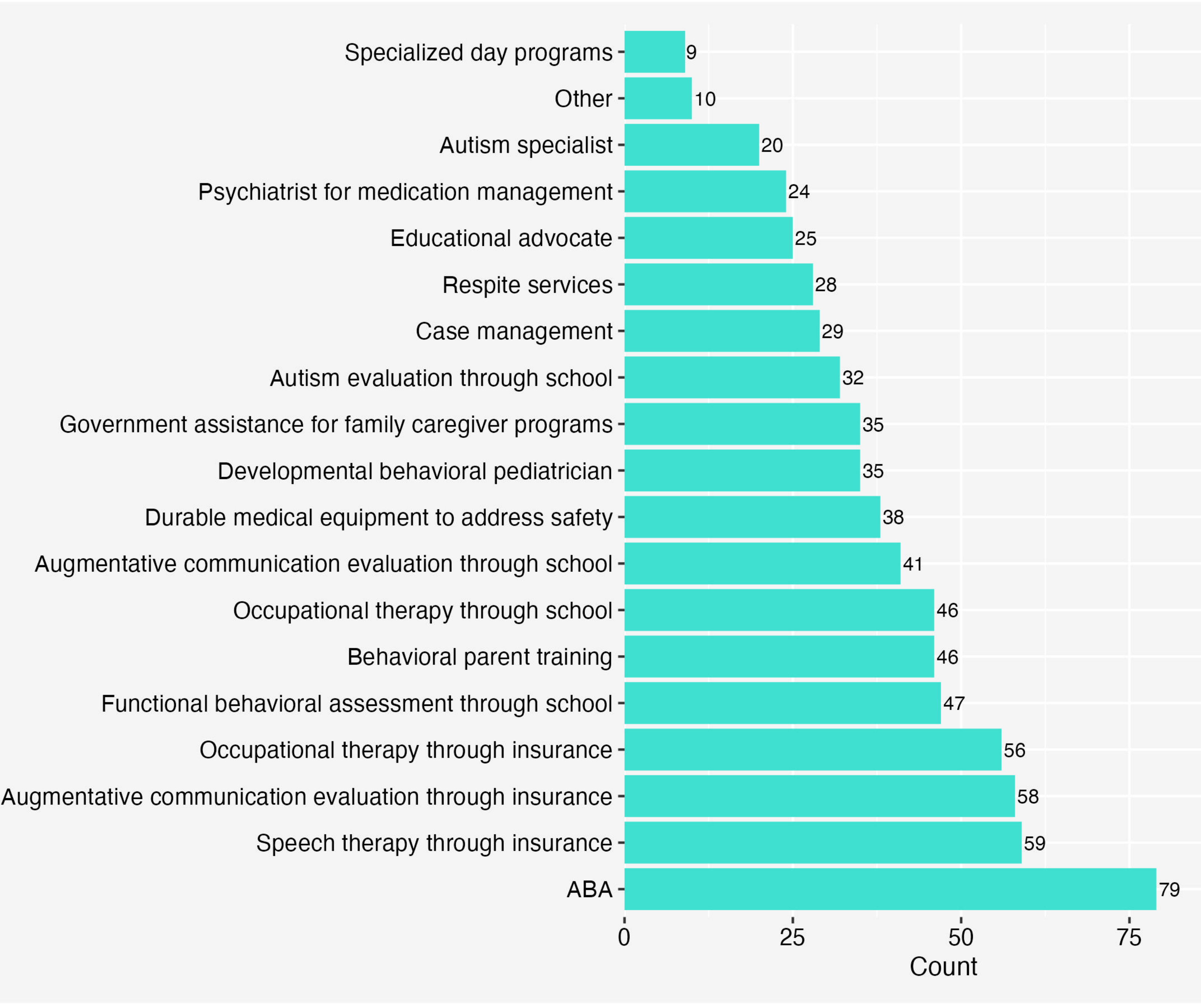


Figure 2: *Most commonly discussed caregiver resources.*

DS=Down syndrome; ASD=autism spectrum disorder. “Other” includes Home and Community-Based Services waivers, Supplemental Security Income waivers, Local Children’s Board

