



Dual Diagnosis Down Syndrome and Autism: A Survey of Caregiver Experiences

Alyssa Bianca Velasco, MD¹, Noemi Spinazzi, MD¹, Drew Wodecki, MPH², and Lina Patel, PsyD³

¹UCSF Benioff Children's Hospital Oakland, ²University of California Berkeley, ³Children's Hospital Colorado

BACKGROUND

The incidence of autism spectrum disorder (ASD) is higher in children with Down syndrome (DS) than in the general population. Estimates range greatly from 5-39%, depending on the assessment and diagnostic criteria used. This suggests that children with DS are at minimum 3 times more likely to have ASD than children in the general population, where the estimated prevalence is 1.7%.

ASD is often diagnosed at a later age in patients with DS, if they are diagnosed at all. Until the late 1980s, it was believed that ASD is rare in individuals with DS. Their relative strength in social skills was thought to be protective against ASD. Further, shared symptomatology between DS and ASD can lead to diagnostic overshadowing and delays in diagnosis.

OBJECTIVE

This study aims to understand the lived experience of families caring for individuals with dual diagnosis of DS-ASD in order to shed light on how to improve care for this patient population by identifying barriers to obtaining diagnosis and appropriate services.

STUDY DESIGN

A survey of 17 questions in free text format was distributed to members of the DS-ASD Connection, a closed online support group for families who have a child with confirmed or suspected dual diagnosis DS-ASD. The survey asked families to reflect on their experience in the early years prior to diagnosis, interactions with the medical and education systems, and the overall impact that the dual diagnosis has had on their family unit.

Responses from completed surveys were coded into categories, which were created through consensus discussions, to quantitatively describe the results. Multiple clinicians independently coded the data for survey questions, and interrater concordance kappa statistic was 0.92. Initial aggregation, cleaning, and sorting of the data was done on Microsoft Excel. All analytical computing was completed on RStudio and RMarkdown.

RESULTS

Table 1: Age of Participant in Years (n = 45)

Category	Mean	Median	Range	SD
Age at time of survey	14.0	16.8	5.0-47.0	8.4
Age at first concern for ASD	3.9	3.0	0.3-12.0	2.6
Age at diagnosis of ASD	8.5	7.0	1.5-21.0	4.8
# of years between first concern for ASD and diagnosis	4.7	3.5	0.0-17.5	3.7

Table 2: Who Caregivers Reported First Concerns to and Whether they had the Knowledge to Guide Further Evaluation (n = 47)

Recipient of first concern	Frequency	Knowledge to guide further evaluation	
		Yes	No
Family	6 (13%)	0 (0%)	6 (100%)
Educator/therapeutic provider	27 (57%)	6 (22%)	21 (78%)
Pediatrician/PCP	22 (47%)	4 (18%)	18 (82%)
Medical specialist*	12 (26%)	10 (83%)	2 (17%)

*Including developmental-behavioral specialists, Down syndrome specialists, geneticists, neurologists

Table 3: Greatest Impact of DS-ASD on Family (n=42)

Social isolation	16 (38%)
Stronger and more empathetic family	16 (38%)
Time demand to care for child	10 (24%)
Stress (including financial and marital)	6 (14%)
Difficulty managing child's behavior	6 (14%)
Career change for caregiver	6 (14%)
Less time to care for other children	4 (10%)
Uncertainty over the future	3 (7%)

The survey was distributed to 486 caregivers. The survey response rate was 35% (n=172), and of those, the survey completion rate was 39% (n=67). There were 67 survey entries, and 47 surveys were included- 9 surveys were excluded as they only submitted demographic information, and 11 were excluded because the child was never formally diagnosed with ASD.

Caregivers developed concerns about their child's development at an average age of 3.9 years. There was a mean 4.7-year gap between the age that families first noticed signs of autism and the age of diagnosis, which occurred on average at 8.5 years.

Most participants expressed their initial concerns to the child's primary care provider (PCP) (47%), or educators and therapeutic service providers (57%). Of respondents who expressed initial concerns to a PCP, 82% felt that the provider did not have the knowledge or experience required to address their concerns.

The most commonly reported initial presenting concerns were stereotypic behaviors (53%), lack of interest in social relationships or poor shared attention (49%), expressive language deficits (40%), sensory sensitivity or defensiveness (29%), poor eye contact (27%), slower progress in developmental milestones (27%), mood or behavior abnormalities (24%), abnormal patterns of play (22%), and developmental regression (18%).

When asked about the most helpful interventions for their child, 37% of respondents mentioned Applied Behavioral Analysis (ABA therapy), and 35% listed other therapeutic services (including speech, physical, and occupational therapy). Respondents were divided when asked whether school was a helpful resource—only 51.3% felt that the school met their child's needs.

Overall, caregivers noted both positive and negative effects of the dual diagnosis on their families. 38% of participants experienced feelings of isolation, but another 38% felt that their family unit became stronger and more empathetic through the process.

CONCLUSIONS

- Most caregivers noticed that their child was different from others with DS in early childhood.
- Though caregivers are presenting with concerns that are consistent with ASD, the diagnosis of ASD in children with DS is typically delayed by years after families first notice warning signs.
- The PCP is often the first recipient of parental concern, underscoring the need for education on DS-ASD among pediatricians.
- This study is limited by the lack of control group of caregivers of children with DS alone or ASD alone, who may experience similar challenges with diagnosis and access to services.
- There is a need for ongoing research on how children with DS-ASD differ from those with ASD alone to lead the way for the development of DS-specific autism screening tools, diagnostic algorithms, and therapeutic interventions.

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