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Assessing co-occurring mental health conditions in a multidisciplinary Down syndrome clinic and the role of family history

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Abstract

Compared to the general population, individuals with Down syndrome (DS) are at a significantly increased risk to develop mental health conditions. This study sought to examine individuals with DS and co-existing mental health comorbidities at one DS specialty clinic. Retrospective chart review of medical records including demographics, genetic testing history, personal and familial mental health history, referrals for mental health indications, and recommendations was performed. Summary statistics, logistic regression, and log of odds were converted to odd ratios to assess associations and significance. The charts of 327 patients, average 19.4 years of age (1–70), were reviewed. Nearly half the participants (42.2%) had at least one diagnosis of a mental health condition. Those with a family history were significantly more likely to have a personal diagnosis of a mental health condition than those without a family history ($p < 0.01$). Moreover, those who completed referrals often received medical management recommendations (86%). This study highlights the prevalence of mental health comorbidities among individuals with DS, and the referral process for mental health conditions, at one DS specialty clinic. Further research is needed to investigate our family history findings, and to determine if these results are generalizable across other DS clinics.

KEYWORDS
Down syndrome, trisomy 21, mental health

1 | INTRODUCTION

Individuals with Down syndrome (DS) are at a significantly increased risk to develop mental health conditions including behavioral, emotional, and psychiatric complications in comparison to those without DS (Capone et al., 2006; Dykens, 2007). In particular, autism spectrum disorder (ASD) is one co-occurring condition which is more common in individuals with DS compared those without DS (DiGuiseppi et al., 2010; Ersoy et al., 2018; Pandolfi et al., 2018); 6%–18% of individuals with DS meet diagnostic criteria for ASD (Ersoy et al., 2018; Rachubinski et al., 2017). However, individuals with DS tend to receive an ASD diagnosis later than those with an isolated ASD diagnosis (Pandolfi et al., 2018) at an average age of 14 years of age versus 3–6 years of age, respectively (Kent et al., 1999; Mandell et al., 2005; Rasmussen et al., 2001). Importantly, if ASD is diagnosed earlier, patients receive earlier access to services and changes to their medical management (Marshall et al., 2015; Pandolfi et al., 2018).

While there is a considerable amount of literature linking individuals with DS to dementia or ASD, there are limited data on additional mental health conditions that are prevalent in individuals with DS (Mantry et al., 2008; Pandolfi et al., 2018; Roizen & Patterson, 2003). The studies to-date highlight other mental health conditions including

obsessive compulsive disorder (OCD), anxiety, major depressive disorder (MDD), and attention deficit hyperactivity disorder (ADHD), which are also prevalent in individuals with DS (Capone et al., 2006; Dykens, 2007; Mantry et al., 2008; Palumbo & McDougle, 2018; Roizen & Patterson, 2003). While reported values vary, studies show that about 18%–38% of individuals with DS experience significant mental health conditions (Capone et al., 2006; Tasse et al., 2016; Vicari et al., 2013); this is greater than the 21% of United States adults in the general population with any mental illness (Mental Illness, n.d.; Merikangas et al., 2010; Substance Abuse and Mental Health Services Administration, 2021), and the 5.2% with a severe mental illness (Substance Abuse and Mental Health Services Administration, 2020).

Studies describing the family history of mental health conditions in individuals with DS are limited; however, research has shown that history of mental illness in biological relatives is the greatest risk factor for most psychiatric conditions in the general population (Nicol & Erlenmeyer-Kimling, 1986; Rasic et al., 2014; Weissman et al., 2000). In those without DS, the children of parents with severe mental illness are at increased risk to develop a range of psychiatric disorders themselves (Rasic et al., 2014).

In addition to co-occurring mental health conditions, various medical comorbidities exist in individuals with DS. Some of these medical conditions could contribute to or mimic mental health conditions, such as thyroid disease, B12 deficiency, and sleep apnea. Prior to 2020, there were no clinical guidelines for the care of adults with DS experiencing these associated conditions (Tsou et al., 2020). Due to the complexity of comorbidity combinations, multidisciplinary clinics have been created to ensure that patients with DS stay up-to-date on screenings, and are receiving appropriate and complete care. One study examined testing and referrals ordered as a result of clinic visits; referrals to clinical psychology were made for 29.5% of patients to conduct further diagnostic work-up of difficult-to-manage behaviors (Skotko et al., 2013). Furthermore, diagnosis of co-occurring mental health conditions for individuals with DS is more complex than isolated diagnoses for a variety of reasons (Ersoy et al., 2018; Vicari et al., 2013). Substantial gaps exist in regard to the knowledge and treatment of patients with DS suspected of having co-occurring mental health conditions (Dykens, 2007).

We began this study to understand our clinic procedures and to investigate the clinical care of patients with DS and co-occurring mental health conditions. The mental health symptoms and diagnoses we studied were anxiety, MDD, ASD, ADHD, OCD, post-traumatic stress disorder (PTSD), bipolar, suicide attempts, and schizophrenia. We aimed to understand the care of patients in the Massachusetts General Hospital Down syndrome Program (MGH DSP) suspected of having a co-occurring mental health condition, including how often are we referring to mental health professionals, which mental health professionals we are referring to, how well referrals were followed through from order to visit, and how referral to a mental health clinician impacted the patient's medical management. By studying our clinical workflow and baseline processes, we hoped to identify gaps in our process, barriers to care, and drivers of referrals to inform future quality improvement research in the MGH DSP. We also anticipated that our results could further support the importance of including psychological services in DS clinic models as previously shown (Santoro, Campbell et al., 2021).

2 | METHODS

2.1 | Context

The MGH DSP is a multidisciplinary clinic for people with DS of all ages. Team includes a physician, nutritionist, social worker, and resource specialist. In the MGH DSP, social workers not only provide resources and social support to families, but also take on a unique role of triaging behavioral and mental health concerns. When indicated, the social worker and physician will confer to decide next steps such as a referral to other mental health services including psychiatry, psychology, neuropsychology, and behavioral health. Additional services including mental health services, access to a feeding specialist, and access to an educational advocate are available on an individualized, as-needed basis. Children under age 5 years are seen by speech/language pathologists, physical therapists, and occupational therapists.

2.2 | Participants

This retrospective chart review study included individuals enrolled in the MGH DSP research database. Patients are eligible for inclusion in this research database if they have a diagnosis of DS documented in their chart, attend at least one visit at the MGH DSP, and provided consent to be enrolled, as previously described (Lavigne et al., 2015). The medical records were reviewed for patients with visits to the MGH DSP, visits from 2015 to 2019, and consent to be enrolled in the registry database.

2.3 | Data collection

Records that were reviewed came from the following sources: (1) Prior to a scheduled clinic visit to the MGH DSP, parents and caregivers of patients fill out an electronic intake form, previously described (Krell et al., 2021). The electronic intake form is reviewed by the MGH DSP team prior to a visit and provides background on interval history, past medical history, medication reconciliation, family history, and physicians/members of a patient's clinical care team. (2) Physician progress notes from visits to the MGH DSP include medical and family history details written by the physicians as well as notation of referrals made. Data were collected through reviewing clinical charts from the MGH DSP database, electronic intake form and EPIC, and the electronic medical record. The information recorded included demographics, personal and familial mental health history, and referrals to psychological services.

1. Demographic characteristics included age, race, ethnicity, gender, and type of DS (trisomy 21, translocation, or mosaic). Patient age was calculated using patient birthdate and age at the time of chart review.
2. Mental health diagnoses included ASD, OCD, anxiety, MDD, ADHD/ADD, bipolar disorder, schizophrenia, PTSD, suicide attempts, and others if any. Personal mental health history included patient information regarding evaluation for mental health concerns (Y/N), and the presence of symptoms of the mental health diagnoses above and the diagnosis of the mental health diagnoses above were recorded.
3. Family mental health history was defined as a history of a mental health diagnosis in the patient's family members, and the relationship to the patient (father, mother, etc.). The genetic counselor reviewing charts then coded this relationship to patient's degree of relative (1st, 2nd, 3rd degree, or other).
4. Referral information included the specialty department to which the mental health referral was placed (psychology, psychiatry, neuropsychology, behavioral health), if the referral was placed within MGH or outside of MGH, other non-mental health referrals, the total number of referrals made, the length of time from visit when the referral was placed to visit with the mental health specialist, the indication for the referral, and if the referral led to modifications in medical management. "Medical management" referred to any recommendations from the visit that could change medical care (e.g., referral to another physician or specialty, recommendations for specific types of therapy, etc).

All electronic progress notes from the genetics, social work, psychology, psychiatry, and neuropsychology departments were reviewed for each patient. Search terms were then used to collect additional information from any documentation in the electronic health record (see Table S1). In addition, content included under the EPIC media tab was reviewed for relevant data. Data from record reviews were collected and managed using REDCap electronic data capture tools hosted at Mass General Brigham (MGB), formally Partners Healthcare. REDCap (Research Electronic Data Capture) is a secure, web-based software platform designed to support data capture for research studies (Harris et al., 2009, 2019).

2.4 | Statistical analysis

Summary statistics and analysis were calculated using JMP® Pro 15 (2021 SAS Institute). Logistic regression was applied, and log of odds were converted to odd ratios to assess associations and significance. Given the importance of family history as a risk factor for mental illness (Nicol & Erlenmeyer-Kimling, 1986; Rasic et al., 2014; Weissman et al., 2000), we compared referral frequencies and outcomes between groups with and without family history. We present results for two cohorts in our total group: those with any family history of a mental health condition present, and those without any family history of a mental health condition.

This effort to understand our clinical workflow, processes, and collect baseline quality improvement data was one of many ongoing quality improvement initiatives in the MGH DSP (Santoro, Brenner-Miller, et al., 2021; Santoro, Donelan, et al., 2021). A waiver of documentation of consent was obtained.

This study was reviewed and approved by the MGB (formerly Partners) Human Research Institutional Review Board.

3 | RESULTS

3.1 | Cohort demographics

The medical records of 327 patients were included in this study, with mean patient age of 19 years ranging from 21 months to 70 years old (Table 1). Our total cohort had slightly more females (52.6%) than males, and was mostly White and non-Hispanic (76%). While all participants had a diagnosis of DS, not all had cytogenetic records scanned into, or clearly noted in, their charts. Of those with a cytogenetic diagnosis listed in the chart, the majority of patients were diagnosed with Complete Trisomy 21 (n = 219, 94.4%). In our total cohort, 20% had a history of previous neuropsychological or psychological testing, either at school (10%) or elsewhere. Demographic characteristics of the two cohorts with and without family history of a mental health condition were similar to the demographics of the total cohort (Table 1).

3.2 | Patient mental health history

Charts were reviewed for (1) mental health symptoms and (2) mental health diagnoses.

Out of the 327 charts reviewed, 50% of patients with DS had at least one mental health symptom present and documented in their chart (Table 2), and of the symptoms present, the most frequent type of symptom in the total cohort with DS was symptoms of anxiety (29%).

Among the 327 patients with DS whose charts were reviewed, 42.2% of patients (n = 138) had at least one diagnosis of a mental health condition (Table 2).

3.3 | Mental health family history

In our cohort of 327 patients with DS, 52% (n = 170) of patients had a family history of a mental health condition (Table 3). Of those with a family history of a mental health diagnosis, the most common diagnoses were anxiety (n = 107, 62.9%), MDD (n = 105, 61.8%), ADHD (n = 53, 31%), and ASD (n = 28, 16%) (Table 3). The family member with a mental health diagnosis varied, but most often was a first degree relative (71%; Table 3). Of our 170 patients with DS with family members with a mental health diagnosis, patients had on average 2.2 family members with a mental health diagnosis. Of those, 37%

TABLE 1 Demographic traits of 327 patients with Down syndrome included in retrospective chart review

Characteristic	Total cohort ($N = 327$)	Family history of mental health condition present[a] ($N = 170$)	Family history of mental health condition absent[a] ($N = 146$)
N (%)			
Sex			
Female	172 (53)	87 (51[b])	81 (55[b])
Male	155 (47)	83 (49)	65 (45)
Race/ethnicity[c]			
White or Caucasian, and non-Hispanic	247 (76)	141 (83)	106 (73)
Hispanic	35 (11)	14 (8)	21 (14)
Black or African-American	17 (5)	9 (5)	8 (5)
Asian or Pacific Islander[d]	12 (4)	5 (3)	7 (5)
American Indian	2 (1)	2 (1)	0 (0)
Other	5 (2)	2 (1)	3 (2)
Missing	28 (9)	13 (8)	15 (10)
Type of Down syndrome documented	232 (71)	122 (72)	104 (71)
If documented, type of Down syndrome			
Trisomy 21	219	115	100
Translocation	10	5	4
Mosaicism	3	2	0
History of previous neuropsychological or psychological testing, outside of the MGH community	66 (20)	36 (21)	28 (19)
School	33 (10)	20 (12)	12 (8)
Other	29 (9)	17 (10)	11 (8)
Mean ± *SD*			
Age (years)	19.4 ± 14.7	20.7 ± 13.9	16.9 ± 14.9

[a]11 patients with Family History unknown are not listed in either cohort, but are included in the total cohort.
[b]% shown are the frequency within each group—e.g., 87 of the total 170 = 51%, 81% of the total 146 = 55%.
[c]Patients may have selected more than one race/ethnicity identifier; sum of percentages >100%.
[d]Includes Asian, Asian Indian, Chinese, and Japanese.

had one, 27% had two, 16% had three, 5% had four, and 9% had five or more relatives with a mental health diagnosis (Table 3).

3.4 | Referrals to psychological services

In our cohort of 327 patients with DS, 203 (50%) patients received at least one referral for mental health (Table 4). Among those patients that received at least one referral, the most common referrals were to MGH Psychology ($n = 165$; 81% of referrals and 50% of patients), MGH Psychiatry ($n = 96$; 47% of referrals and 29% of patients), and MGH Neuropsychology ($n = 118$; 58% of referrals and 36% of patients; Table 4). Referrals outside of the MGH system were less common (4%). The most common indications for referral were cognitive concerns (34% of patients) and behavioral concerns (20%). On average, patients had 1.7 mental health referrals placed.

Of the 203 referrals placed for our total cohort, most (85%) patients followed up on the referral and were evaluated for the mental health concern (87%). In tracking the outcome of the referral, we found that many (69%) of those referrals had changes made to medical care as a result of the referral. The most commonly seen recommendations to care were referrals to another clinician (74% of the referrals), changes in medications (65%), additional psychological testing (21%), and educational recommendations and guardianship recommendations (15%; Table 4).

3.5 | Family history analysis

Patients with DS, with a family history of a mental health condition present were more likely to have at least one mental health symptom (64%; Table 2), than those without a family history (33%, $p < 0.01$). Mental health diagnoses in those with a family history included

TABLE 2 Mental health diagnoses in patients with Down syndrome in the MGH Down syndrome program ($n = 327$)

Mental health diagnosis	Total cohort ($N = 327$)	Family history of mental health condition present[a] ($N = 170$)	Family history of mental health condition absent[a] ($N = 146$)
No mental health symptoms	164 (50)	62 (36)	98 (67)
At least one mental health symptom present	163 (50)	108 (64)	48 (33)
If mental health symptoms present, type			
Anxiety	95 (29)	63 (37)	26 (18)
Major depressive disorder (MDD)	52 (16)	38 (22)	14 (10)
Autism spectrum disorder (ASD)	38 (12)	25 (15)	11 (8)
Attention deficit hyperactivity disorder (ADHD)	36 (11)	26 (15)	9 (6)
Obsessive compulsive disorder (OCD)	33 (10)	23 (29)	7 (5)
Post traumatic stress disorder (PTSD)	4 (1)	3 (2)	1 (1)
Bipolar disorder	3 (1)	3 (2)	0 (0)
Suicide attempts	1 (<1)	1 (1)	0 (0)
Schizophrenia	0 (0)	0 (0)	0 (0)
Other[+]	27 (8)	15 (9)	11 (8)
Any mental health diagnosis	138 (42)	100 (59)	31 (21)
If mental health diagnosis, type			
Anxiety	67 (20)	47 (28)	14 (10)
MDD	37 (11)	30 (18)	7 (5)
ASD	33 (10)	23 (14)	8 (5)
ADHD	30 (9)	22 (13)	7 (5)
OCD	17 (5)	12 (7)	3 (2)
Bipolar disorder	3 (1)	3 (2)	0 (0)
PTSD	1 (<1)	0 (0)	1 (1)
Schizophrenia	0 (0)	0 (0)	0 (0)
Other	17 (5)	13 (8)	3 (2)

[a]11 patients with Family History unknown are not listed in either cohort, but are included in the total cohort.

anxiety (37%), MDD (22%), ASD (15%), and OCD (29%) and those without a family history included anxiety (18%), MDD (10%), ASD (8%), and OCD (5%; Table 1). In the subgroup with a family history of mental health condition present, 59% of patients with DS were diagnosed with a mental health condition, with anxiety, MDD, and ASD the most frequent diagnoses (28%, 18%, and 14%, respectively; Table 2). In the subgroup without a family history of mental health condition, 21% of patients with DS were diagnosed with a mental health condition.

Patients with or without a family history of mental health condition present were referred to the same departments; those with a family history were referred more (59% vs. 38% to MGH Psychology; 39% vs. 18% to MGH Psychiatry, and 42% vs. 29% to MGH Neuropsychology; Table 4). In comparing referrals based on the presence/absence of family history of mental health conditions, we found that our patients with DS with a mental health diagnosis present in the family history were 3.7 times more likely to receive an initial psychological service referral than those without a family history of mental health diagnoses (CI: 2.29, 5.95; $p \leq 0.01$). When comparing the outcome from those referrals between those patients with DS with family history of mental health conditions present and those without a family history of mental health condition present, there was no significant difference in referral follow-up based on family history ($p > 0.05$). In the subgroup of patients with DS with family history of a mental health condition present, 68% had additional referrals placed, 67% had medication changes, and 22% had additional psychological testing; in those without a family history, rates were 86%, 57%, and 21%, respectively; Table 4.

In our logistic regression analysis, the odds of a patient being diagnosed with a mental health condition were significantly higher in those who had a family history of mental health condition, compared to those who do not (OR: 5.77, z: 6.75, Confidence Interval [CI]: 3.47, 9.61; $p < 0.01$, Figure 1). The breakdown of the 304 patients with both patient information and family history available are summarized (Table 5). There were no statistically significant differences between those with a family history of mental health diagnosis and those

TABLE 3 Family history details for patients in the MGH Down syndrome program with a family history of mental health diagnosis ($N = 170$)

	N (%)
Any family history of mental health diagnosis	170 (100)
Type of mental health diagnosis in family member	
Anxiety	107 (63)
Major depressive disorder (MDD)	105 (62)
Attention deficit hyperactivity disorder (ADHD)	53 (31)
Autism spectrum disorder (ASD)	28 (16)
Bipolar disorder	20 (12)
Schizophrenia	5 (3)
Suicide attempts	5 (3)
Obsessive compulsive disorder (OCD)	3 (2)
Posttraumatic stress disorder (PTSD)	1 (1)
Other+	15 (9)
Degree relative with mental health diagnosis	
First	121 (71)
Second	89 (52)
Third	31 (18)
Other	2 (1)
Number of family members with any mental health diagnosis	
1	63 (37)
2	46 (27)
3	27 (16)
4	8 (5)
5+	15 (9)
	Mean ± *SD*
Average number of family members with any mental health diagnosis	2.2 ± 1.3

without on our demographics collected (age, race, ethnicity, gender, and type of DS).

4 | DISCUSSION

We began this study to understand the mental health conditions that co-occur with DS and to understand the current referral process for mental health conditions at one specialty clinic. We found that co-occurring mental health diagnoses were common in our cohort of individuals with DS. In studying our clinical workflow, we were pleased to find that the referrals to psychological services were often completed, patients with DS often met with a mental health professional, and these referrals led to increased recommendations to medical management. We did not identify barriers to care, but this could be investigated further in the future with knowledge of our baseline processes. Family history may be important a driver of referrals; patients with DS and a family history of mental health conditions were more likely to be diagnosed with a mental health condition than those without such family history.;

Co-occurring mental health diagnoses are a frequent consideration for individuals with DS. Literature to date has shown that individuals with DS are at higher risk of developing mental health conditions including behavioral, emotional, and psychiatric complications (Capone et al., 2006; Dykens, 2007). Indeed, in our cohort of individuals with DS, we found that many (42%) of our patients had a co-occurring mental health diagnosis. Specifically, the two most common mental health comorbidities among participants in our study, anxiety and depression, align with previous studies (Mantry et al., 2008; Tasse et al., 2016).

Families often followed through with referrals to mental health departments with impacts on outcomes. For those who received a referral to a mental health clinician, the majority had additional medical management recommendations made by that clinician, and it is known that improved medical management could increase the quality of life of patients with DS (Skotko et al., 2013). With so many individuals in this study having a mental health diagnosis, in combination with the frequency and value of referrals on medical management recommendations and potential to improve quality of life, our results demonstrate the value of referrals to mental health care for patients with DS. Increasing access to psychological services for patients with DS and considering incorporating mental health roles, including psychiatrists, psychologists, and neuropsychologists into multidisciplinary DS clinics may be of value. Indeed, a published survey of members of DS specialty clinics showed that many of the top roles valued as "must haves" for a DS clinic were psychological service roles, including: social worker, psychologist/neuropsychologist, and psychiatrist, but these roles were not always present in DS specialty clinics (Santoro, Campbell, et al., 2021). Further, parents have reported behavior problems as a top concern to be addressed at the DS clinic (Skotko et al., 2013).

Awareness of family history of mental health diagnoses is of known value in the general population (Trotter & Martin, 2007; Valdez et al., 2007), and our study highlights the importance of collecting a comprehensive psychiatric family history when evaluating patients with DS. Psychiatric genetic counselors could be helpful to provide guidance on risks for common mental health conditions based on family history and could be a resource for providers discussing these risks with families with or without DS (Austin & Honer, 2008; Saxton et al., 2022). We identified that patients with DS and family history of mental health diagnoses were (1) at greater risk for a mental health diagnosis, and (2) more likely to be referred to psychological services. In theory, patients with DS with a family history *could* be more likely to have a personal diagnosis due to genetic and shared environmental factors. Parents' knowledge, experience, and awareness of the signs of a mental health condition, if they have this or a similar type of condition in their family (Herlihy et al., 2013), might have contributed to the increased likelihood of referrals. While patients without a family history may attribute behavioral changes to the DS diagnosis rather than a mental health condition (Appleton

TABLE 4 Referrals for mental health and the subsequent medical management recommendations from those referrals for patients with DS in the MGH Down syndrome program

Referral information	Total cohort ($N = 327$)	Family history of mental health condition present[a] ($N = 170$)	Family history of mental health condition absent[a] ($N = 146$)
	N (% of *N* patients in cohort)		
Patients with mental health referrals through MGH Down Syndrome Program	203 (62)	128 (75)	66 (45)
Type			
MGH Psychology	165 (50)	101 (59)	55 (38)
MGH Psychiatry	96 (29)	66 (39)	26 (18)
MGH Neuropsychology	118 (36)	71 (42)	42 (29)
MGH Behavioral Health	1 (<1)	0 (0)	1 (1)
External Psychology	2 (1)	2 (1)	0 (0)
External Psychiatry	2 (1)	2 (1)	0 (0)
External Neuropsychology	1 (<1)	1 (1)	0 (0)
External Behavioral Health	0 (0)	0 (0)	0 (0)
Other	9 (3)	6 (4)	3 (2)
Indication for referral			
Cognitive concerns	112 (34)	66 (39)	41 (28)
Behavioral concerns	65 (20)	42 (25)	20 (14)
Academic concerns	23 (7)	12 (7)	9 (6)
Emotional concerns	21 (6)	16 (9)	4 (3)
Social concerns	5 (2)	3 (2)	2 (1)
Other[b]	169 (52)	107 (63)	55 (38)
	Mean ± *SD*		
Total # of referrals	1.7 ± 0.7	1.7 ± 0.7	1.8 ± 0.7

Outcome of referral	$N = 203$ referrals	$N = 128$ referrals	$N = 66$ referrals
	N		
Patients followed up on the referral(s)	173	113	53
If the referral was followed up on, patients with changes made to the patient's medical care as a result of the referral	141	93	42
If changes to medical care, type of management recommendation	*N*		
Additional referrals	104	63	36
Medications	91	62	24
Additional psychological testing	30	20	9
Educational recommendations	21	12	8
Guardianship recommendations	18	13	4
Behavioral interventions	10	8	2
Discussed electroconvulsive therapy (ECT)	2	2	0
Drug trials	1	0	1
Recommendation of the use of a continuous positive airway pressure (CPAP) mask	1	0	1
Additional imaging	2	2	0
Over-the-counter medications	1	0	1

[a]Other included (*N*): Baseline neuropsychological evaluation (78), initial developmental and psychiatric consultation (24), general consult (10), diagnostic clarification (9), guardianship (6), medication related (4), ADHD (3), initial mental health consultation for Weight Center (2), adaptive skills evaluation (1), adjustment to illness/condition (1).
[b]11 patients with Family History unknown are not listed in either cohort, but are included in the total cohort.

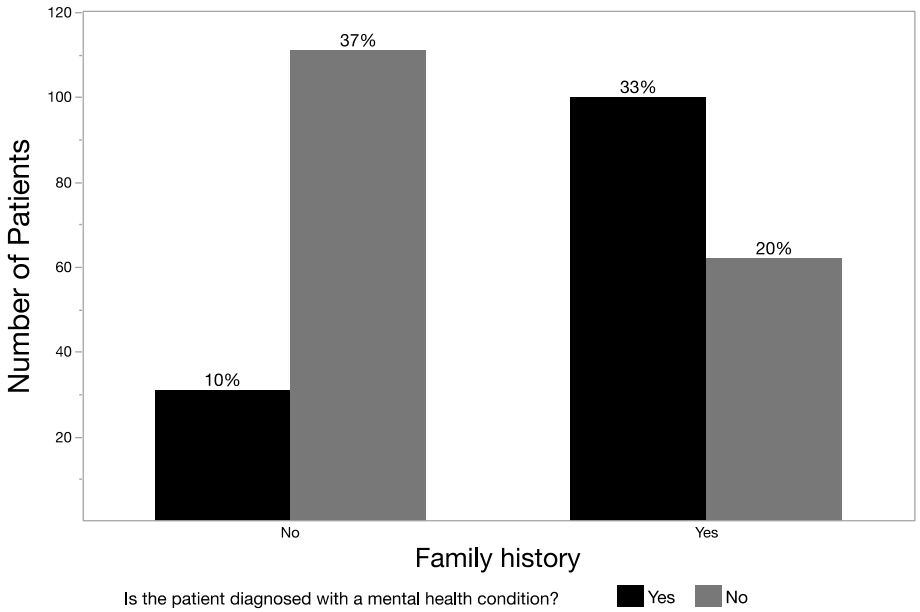

FIGURE 1 Relationship between a mental health diagnosis and family history. This graph represents individuals with a reported family history of a mental health condition compared to those who did not. Those with unclear and pending diagnoses were excluded

TABLE 5 Comparison of 304 patients with Down syndrome by diagnosis of mental health condition in patient and presence of family history of mental health condition

	No family history of mental health condition, *N*	Family history of mental health condition, *N*	Total
Patient diagnosed with mental health condition	31	100	131
Patient not diagnosed with mental health condition	111	62	173
Total	142	162	304

et al., 2019; Deb et al., 2001; Tasse et al., 2016). Documented family history might influence referral patterns, or family history might impact the prevalence of mental health conditions in those with DS and lead to an increased need for referrals; additional studies with larger samples sizes could further investigate the drivers of differences in referrals by family history of mental health condition. In the literature for those without DS, there are multiple studies demonstrating the link between family history and risk for patient's mental health diagnosis (Rasic et al., 2014), but we did not identify studies to-date which associate family history of mental health diagnosis to patient referral patterns to mental health clinicians.

Our cohort consisted of patients in one DS clinic, the MGH DSP, which uses a multidisciplinary model of care with physicians, social workers, and nutritionists, among others. Though there is no mental health professional embedded in the MGH DS clinic, a social work professional attended the clinics, worked to triage mental health concerns, and coordinated referrals to psychiatry and neuropsychology; in our clinic they were on the "front lines" of mental health care. The role and availability of SW to all patients may be unique to our program, and may not apply to other DS clinics without a mental health professional embedded in the clinic. In fact, although many DS clinics consider social workers a "must have" role, only 64% of adult DS clinics surveyed had a social worker at DS clinic sessions (Santoro, Campbell, et al., 2021). Our clinic model may not generalize to other DS clinics; other clinics may need to adapt our findings to their model of care, considering how social workers and other psychological services interface with mental health care in their program and system. However, there is a role for mental health professionals in DS clinics given the high percent of individuals who need evaluation for these symptoms, and a need to support the mental health of patients with DS.

Additional limitations may exist. As records reviewed for this study were based on a population of patients seen at one specialty DS clinic, the cohort may not be representative of the larger DS population. In addition, the majority of participants were White and non-Hispanic which may impact how our findings translate to communities with greater diversity. Specifically, when considering psychiatric referrals, it is important to note that the MGH DSP is a large well-ranked tertiary medical center, and patients with greater medical complexity may seek out the MGH DSP for "second opinions"; our patient population may not generalize to other DS clinics. Record review relied on the information available in the medical record, and our low rate of neuropsychological testing at schools (10%) may be the result of limited documentation. Further, other sites may have more limitations in mental health resources (such as long wait lists for providers, insurance coverage limitations) or in other locations, the processes may

differ (e.g., in some states all individuals will have neuropsych testing done as part of their diagnosis of ID and to qualify for services). In the future, further studies are needed to evaluate these factors on mental health care, and to evaluate if the rate of mental health diagnoses made for individuals with DS differs in other clinics or based on other factors (such as access, availability, and visits to a mental health professional).

Our study compared two groups of patients with DS by family history; we did not compare to a control group without DS, but in the future it would be interesting to compare rates of family history of mental health diagnoses to the general population rates. Future study would be useful to evaluate the types of mental health diagnosis; for example, if family history of a specific mental health condition predicts the same mental health diagnosis in the individual with DS or predicts any mental health diagnosis. Future study could track further details on asymptomatic patients with positive family history of mental health diagnosis to determine if earlier referral based on family history alone improves care.

5 | CONCLUSION

Family history of mental health diagnosis was associated with greater rate of mental health diagnosis for the patient with DS, and higher rates of referrals to psychological services; highlighting the need for a comprehensive family history to be collected in DS clinics. Psychological service referrals had impacts on medical management recommendations. DS clinics should be prepared to address the co-occurring mental health diagnoses that are common in DS, and should consider hiring mental health professionals in their clinics, genetic counselors to assess family history, and/or utilizing clinic social workers to help facilitate mental health referrals.

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CONFLICT OF INTEREST

Stephanie L. Santoro has received research funding from LuMind Research Down Syndrome Foundation to conduct clinical trials for people with Down syndrome within the past 2 years. She serves in a non-paid capacity on the Medical and Scientific Advisory Council of the Massachusetts Down Syndrome Congress, the Board of Directors of the Down Syndrome Medical Interest Group (DSMIG-USA), and the Executive Committee of the American Academy of Pediatrics Council on Genetics. The other authors have no conflicts of interest to disclose.

DATA AVAILABILITY STATEMENT

The data that support the findings of this study are available on request from the corresponding author. The data are not publicly available due to privacy or ethical restrictions.

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