

Database to Assess Medical and Neurodevelopmental Co-Occurring Conditions in Down syndrome

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BACKGROUND

Despite a common genotype, individuals with Down syndrome (DS) experience a wide range of medical and neurodevelopmental conditions, and behavioral outcomes.¹⁻²

Reports of patterns and prevalence for co-occurring neurodevelopmental disorders (NDD) [specifically Autism Spectrum Disorder (ASD) and Attention Deficit Hyperactivity Disorder (ADHD)] have been varied and the impact of medical conditions on neurodevelopment in DS remains unknown.

The Boston Children's Hospital Down Syndrome Program (BCH DSP) created an ongoing prospective clinical database of patients to characterize:

- Patterns of development
- Prevalence and interplay of co-occurring medical, neurodevelopmental, and mental health conditions
- Impact of therapeutic interventions on these conditions

OBJECTIVES/AIMS

This research aims to :

1. Describe medical conditions, NDD, and mental health conditions in children with DS in a clinical DSP at a tertiary medical center
2. Examine prevalence of ASD and ADHD in DS, and
3. Examine prevalence of associated co-occurring medical and mental health conditions in children/adolescents with DS who have ADHD, ASD, Combined ADHD and ASD (ADHD+ASD), and no neurodevelopmental disorders (Non-NDD).

DESIGN/METHODS

Database: 761 patients seen at the BCH DSP from 2018 –2021

- 617 patients had complete medical, mental health and neurodevelopmental data and were included for analysis

Data: Medical (caregiver reported), neurodevelopmental (clinician reported), and mental health (clinician reported) conditions, and sociodemographics, were collected and entered into the database

Caregiver form: All caregiver forms are completed in English; non-English speaking families completed forms with the help of an interpreter, when available

Cohorts:

ASD only group: individuals with a clinical diagnosis of ASD

ADHD only group: individuals with a clinical diagnosis of ADHD

ASD+ADHD group: individuals with a clinical diagnosis of both ASD and ADHD

Non-NDD group: all individuals without ASD or ADHD

Analysis: Descriptive frequencies were utilized to describe prevalence

Table 1A. Population demographics and characteristics	N (%) N=761
Sex	
Male	421 (55%)
Female	340 (45%)
Age:	
0-3 years	217 (29%)
4-6 years	140 (18%)
7-13 years	271 (36%)
14-17 years	99 (13%)
18+ years	34 (4%)
Race:	
White	491 (65%)
Black/African American	74 (10%)
Asian	20 (3%)
Native Hawaiian/Pacific Islander	1 (0.1%)
Other	111 (15%)
Missing/Not reported	64 (8%)
Median Income based on Zip Code:	
≤\$34,282	49 (6%)
≥\$34,282 but < \$4,850	311 (41%)
≥\$4,850 but < \$82,276	319 (42%)
≥\$82,276	82 (11%)
Primary Language:	
English	657 (86%)
Spanish	54 (7%)
Other	50 (7%)
Table 1B. Co-Occurring medical, mental health conditions, and neurodevelopmental disorders	N(%) N = 617
Medical Conditions:	
Vision Problem	358 (58%)
Congenital Heart Disease (CHD)	291 (47%)
Cardiac Surgery	141 (23%)
ENT Surgery	289 (47%)
Respiratory Problem	238 (39%)
Ear Problem	226 (37%)
Constipation	225 (36%)
Obstructive Sleep Apnea	160 (26%)
Thyroid Disease	159 (26%)
Gastroesophageal Reflux Disease	133 (22%)
Celiac	31 (5%)
Duodenal Atresia	30 (5%)
Scoliosis	26 (4%)
Atlantoaxial Instability	21 (3%)
Hirschprung's	16 (3%)
Epilepsy	16 (3%)
Leukemia	13 (2%)
Transient Myeloproliferative Disorder	9 (1%)
Mental Health Conditions:	
Behavior disorder	41 (7%)
Anxiety	31 (5%)
Regression/Catatonia	10 (2%)
Depression	6 (1%)
Obsessive Compulsive Disorder (OCD)	2 (0.3%)
Psychosis	1 (0.2%)
Neurodevelopmental Disorders	
Non-NDD	526 (85%)
ASD	56 (9%)
ADHD	29 (5%)
ASD +ADHD	6 (1%)

RESULTS

Table 2. Frequency of co-occurring medical and mental health conditions in ASD only, ADHD only, ASD+ADHD, and Non-NDD groups	Non-NDD N= 526	ASD Only N = 56	ADHD Only N = 29	ASD+ADHD N =6
Medical Conditions:				
Vision Problem	295 (56%)	32 (57%)	26 (90%)	5 (83%)
Congenital Heart Disease (CHD)	252 (48%)	22 (29%)	15 (52%)	2 (33%)
Cardiac Surgery	121 (23%)	12 (21%)	7 (24%)	1 (17%)
ENT Surgery	238 (45%)	27 (48%)	20 (69%)	4 (67%)
Respiratory Problem	201 (38%)	22 (39%)	12 (41%)	3 (50%)
Ear Problem	186 (35%)	23 (41%)	14 (48%)	3 (50%)
Constipation	170 (32%)	32 (57%)	18 (62%)	5 (85%)
Obstructive Sleep Apnea	128 (24%)	13 (23%)	16 (55%)	3 (50%)
Thyroid Disease	135 (26%)	12 (21%)	11 (38%)	1 (17%)
Gastroesophageal Reflux Disease	106 (20%)	17 (30%)	8 (28%)	2 (33%)
Celiac	24 (5%)	4 (7%)	2 (7%)	1 (17%)
Duodenal Atresia	25 (5%)	3 (5%)	2 (7%)	-
Scoliosis	17 (3%)	6 (11%)	2 (7%)	1 (17%)
Atlantoaxial Instability	16 (3%)	4 (7%)	-	1 (17%)
Hirschprung's	15 (3%)	1 (2%)	-	-
Epilepsy	9 (2%)	4 (7%)	1 (3%)	2 (33%)
Leukemia	12 (2%)	1 (2%)	-	-
Transient Myeloproliferative Disorder	8 (2%)	1 (2%)	-	-
Mental Health Conditions:				
Behavior disorder	25 (5%)	7 (13%)	9 (31%)	-
Anxiety	21 (4%)	1 (2%)	6 (21%)	3 (50%)
Regression/Catatonia	8 (2%)	2 (4%)	-	-
Depression	3 (1%)	3 (5%)	-	-
Obsessive Compulsive Disorder	1 (1%)	-	1 (3%)	-
Psychosis	1 (1%)	-	-	-

SUMMARY

Preliminary analysis revealed:

- Most individuals experienced 1-6 co-occurring medical conditions
- Vision problems, ENT surgery, and respiratory problems were frequent co-occurring medical conditions in all groups
- OSA, ENT surgery, vision problems and behavior disorder were especially common in ADHD
- Constipation was especially common in ASD and ADHD groups

Larger samples and further analyses are needed to compare medical and mental health conditions between groups.

CONCLUSIONS

Findings highlight that co-occurring medical, neurodevelopmental, and mental health conditions are prevalent in a clinical DS populations.

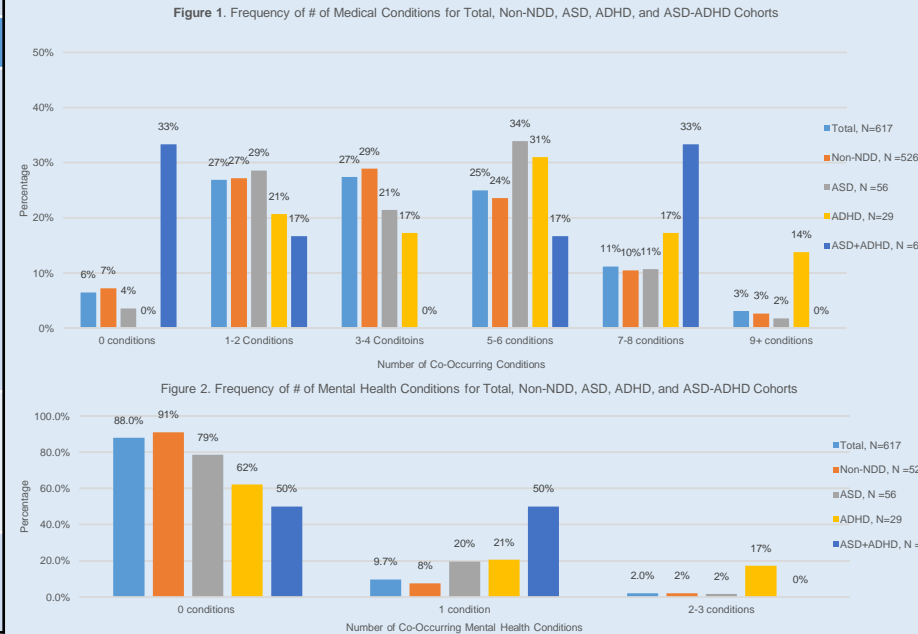
A clinical databases can elucidate patterns of development and risk factors at various ages for individuals with DS.

Limitations:

- Caregiver forms were only available in English
- 81% completion rate for caregiver-completed forms
- Specialty clinic at tertiary care center may skew toward a more complex patient population

Future Directions:

- Examine relationships between co-occurring medical, neurodevelopmental, sociodemographic, educational, therapeutic, and behavioral outcomes
- Create a process for verification of caregiver reported medical history and comparison with clinician provided information



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