

Prevalence of Common Conditions in a Large Population of Individuals with Down Syndrome

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BACKGROUND

The majority of what we know about the Down syndrome population is surrounding genetics and youth-onset effects, like congenital heart defects and learning/cognitive issues. More recently, comorbidities among individuals with DS have been explored in other countries, particularly the UK, where conditions across healthcare systems are tracked in unified registries and incidence can be easily described. This study is a novel, foundational look at prevalence of comorbidities among a large sample of individuals with DS across one healthcare system in the U.S. The comorbidities investigated in this study were selected by an expert and pioneer in the field of DS based on clinical experience treating individuals with DS in the U.S. for almost 30 years.

DESIGN/METHODS

Study Design

- Retrospective, descriptive cohort study
- Approximately 28 years of data
- Data collected on the Advocate Health Care patient population, which includes the Adult Down Syndrome Center

Analysis

- Prevalence reported as counts (%) with odds ratio (OR)
- Corresponding Pearson Chi-square tests, or Fisher's Exact tests where prevalence ≤ 5 , as indicated by \wedge in table
- P-values were calculated to represent statistically significant differences, with $p < 0.05$ indicated by * in table
- Conditions coded per AHRQ's CCS for ICD-9-CM ([here](#))
- Controls were matched on year of birth (+/- one year) and sex at up to a 1:5 ratio

Table 1. Demographics and Baseline Characteristics of Cases and Controls at Most Recent Visit

	DS Cases (N=6078)	Controls (N=30326)
	27.88 (20.37); 25.00 (9.46)	29.04 (20.96); 25.00 (11.48)
Age (Mean, Median)		
Total Encounters (Mean, Median)	11.23 (15.19); 6.00 (2-14)	194.73 (1656.66); 18.00 (8-42)
Sex	N=6075	N=30324
Male	3171 (52.19%)	15816 (52.15%)
Female	2904 (47.79%)	14508 (47.84%)
Race	N=4495	N=21296
White	3477 (77.35%)	13198 (61.97%)
Black	650 (14.46%)	6667 (31.31%)
Asian	150 (3.34%)	1183 (5.56%)
American Indian	57 (1.27%)	174 (0.82%)
Other	161 (3.58%)	74 (0.35%)
Ethnicity	N=3212	N=21066
Hispanic	851 (26.49%)	3851 (18.28%)
Non-Hispanic	2361 (73.51%)	17215 (81.72%)
Insurance	N=5991	N=29666
Private	2848 (47.54%)	22532 (75.95%)
Medicare	2050 (34.39%)	1464 (4.93%)
Medicaid	644 (10.75%)	4097 (13.82%)
Other	409 (6.83%)	1573 (5.30%)

RESULTS

Table 2. Prevalence of Conditions of Interest among Cases as Compared with Controls

Conditions	DS Cases (N = 6078)	Controls (N=30,326)	Odds Ratio (CI)	P-VALUE
Cancer				
Breast	9	232	0.19 (0.10, 0.37)	<0.0001*
Cervix	23	371	0.31 (0.20, 0.47)	<0.0001*
Testis	10	19	2.63 (1.22, 5.66)	0.0102*
Colorectal	3	110	0.14 (0.04, 0.43)	<0.0001**
Leukemia	43	50	4.31 (2.87, 6.49)	<0.0001*
Lymphoma (Hodgkins and Non-Hodgkins)	7	43	0.81 (0.37, 1.81)	0.609
Melanomas of Skin	6	59	0.51 (0.22, 1.17)	0.1063
Heart Disease				
Hypertension	343	5345	0.28 (0.25, 0.31)	<0.0001*
Stroke	22	95	1.16 (0.73, 1.84)	0.5404
Coronary Atherosclerosis and Other Heart Disease	119	703	0.84 (0.69, 1.02)	0.0844
Acute MI	58	343	0.84 (0.64, 1.11)	0.2281
Moyamoya	14	2	35.04 (7.95, 154.06)	<0.0001*
Mental Health				
Anxiety	519	1283	2.11 (1.90, 2.35)	<0.0001*
QCD*	0	0	-	-
Attention-Deficit, Disruptive Behavior Disorders	210	1070	0.98 (0.84, 1.14)	0.7772
Mood Disorders	614	2856	1.08 (0.99, 1.18)	0.0973
Schizophrenia and other Psychoses	79	100	3.92 (2.96, 5.35)	<0.0001*
Impulse Control Disorders NEC	27	34	3.98 (2.40, 6.59)	<0.0001*
Catatonia	27	51	2.65 (1.66, 4.23)	<0.0001*
Infections				
STDs (not including HIV or Hepatitis)	3	79	0.19 (0.06, 0.60)	0.0005**
HIV	6	60	0.50 (0.22, 1.15)	0.0973
Hepatitis	10	39	1.28 (0.64, 2.56)	0.4857
Flu	125	1004	0.61 (0.51, 0.74)	<0.0001*
Bronchitis	210	479	2.23 (1.89, 2.63)	<0.0001*
Sinusitis	227	227	5.14 (4.27, 6.19)	<0.0001*
Pneumonia	679	961	3.84 (3.47, 4.26)	<0.0001*
UTI	29	166	0.87 (0.59, 1.29)	0.4934
Cellulitis	186	931	1.00 (0.85, 1.17)	0.9679
Gastrointestinal Conditions				
Intestinal Obstruction without Hernia	82	167	2.47 (1.89, 3.22)	<0.0001*
Esophageal Disorders	1052	3677	1.52 (1.41, 1.63)	<0.0001*
Gastroduodenal Ulcer except Hemorrhage	27	80	1.69 (1.10, 2.62)	0.0177*
Celiac	0	0	-	-
Endocrine Conditions				
Thyroid	2291	2008	8.53 (7.96, 9.14)	<0.0001*
Diabetes*	471	3105	0.74 (0.67, 0.81)	<0.0001*
Neurological Conditions				
Epilepsy; Convulsions	803	869	5.16 (4.67, 5.70)	<0.0001*
Alzheimer's Disease*	0	0	-	-
Myelopathy, cervical	12	31	1.93 (0.99, 3.77)	0.0486*
Orthopedic Conditions				
Atlanto-axial Subluxation	129	8	82.18 (40.21, 167.93)	<0.0001*
Osteoporosis	12	26	2.31 (1.16, 4.57)	0.0138*
Osteoarthritis	151	1309	0.56 (0.48, 0.67)	<0.0001*
Miscellaneous Conditions				
Dysphagia	194	236	4.20 (3.47, 5.10)	<0.0001*
Diseases of the Eyes and Adnexa	1710	6177	1.53 (1.44, 1.63)	<0.0001*
Diseases of the Ears and Mastoid Process	2548	8267	1.93 (1.82, 2.04)	<0.0001*
Sleep Apnea	1117	2179	2.91 (2.69, 3.15)	<0.0001*

* Due to this study's coding framework neither QCD nor Alzheimer's Disease were present in either cases or controls. We believe this is an EMR classification issue and not due to absence of condition.

CONCLUSIONS

Relative to their non-DS counterparts, individuals with DS have **decreased odds of experiencing:**

- Breast, cervical, and colorectal cancers, lymphomas, and melanomas of the skin
- Hypertension, atherosclerosis, acute MI
- STDs, HIV, Flu, UTI
- Diabetes
- Osteoarthritis

Relative to their non-DS counterparts, individuals with DS have **increased odds of experiencing:**

- Testicular cancer and leukemia
- Moyamoya
- Anxiety, mood disorders, schizophrenia and other psychoses, impulse control disorders, and catatonia
- Hepatitis, bronchitis, sinusitis, pneumonia
- Intestinal obstruction, esophageal disorders, gastroduodenal ulcers
- Thyroid disorders
- Epilepsy, cervical myelopathy
- Atlanto-axial subluxation, osteoporosis
- Dysphagia
- Diseases of the eyes and diseases of the ears
- Sleep apnea

Currently, medical care for individuals with DS follow general population guidelines when it comes to treating comorbidities and anticipating co-occurring conditions based on common risk factors. *Our findings indicate that individuals with DS have drastically different odds of experiencing medical conditions than age- and sex-matched individuals without DS. Considering this, prevention and treatment guidelines should more accurately reflect the needs of this unique population.*

REFERENCES

This study was influenced by the foundational work of: Alexander M, Petri H, Ding Y, Wandel C, Khwaja O, Foskett N. Morbidity and medication in a large population of individuals with Down syndrome compared to the general population. Dev Med Child Neurol. 2015;58(3):246-254. doi: <https://doi.org/10.1111/dmcn.12868>

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