

Prevalence of Cancer in a Large Population of Individuals with Down Syndrome

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BACKGROUND/AIMS

Research suggests that the prevalence of cancer in individuals with DS is different than in individuals without DS. It is well-established that individuals with DS show higher prevalence of leukemias and lymphomas in childhood and some research indicates a lower prevalence of solid tumors in adulthood, with a few notable exceptions. The aim of this study was to describe prevalence of cancer among individuals with DS (cases) relative to individuals without DS (controls).

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 is reported as counts (%) with odds ratio (OR)
- Corresponding Pearson Chi-square tests, or Fisher's Exact where prevalence ≤ 5 as indicated by * in table
- P-values were calculated to represent statistically significant differences, with $p < 0.05$ indicated by * in table
- Cancer sites/types were organized using *Surveillance, Epidemiology, and End Results (SEER) Program Malignant Neoplasm Site Groupings* reporting standards and associated ICD codes
- 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)
Age (Mean, Median)	27.88 (20.37); 25.00 (9-46)	29.04 (20.96); 25.00 (11-48)
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	2090 (34.89%)	1464 (4.93%)
Medicaid	644 (10.75%)	4097 (13.82%)
Other	409 (6.83%)	1573 (5.30%)

RESULTS

Cancer Site	DS Cases (N=6,078)	Controls (N=30,326)	Odds Ratio (CI)	P-value
<i>Buccal cavity and pharynx</i>	0	19	-	0.0594 [*]
<i>Digestive system</i>				
Esophagus	0	9	-	0.3721 [*]
Stomach	2	4	2.49 (0.46, 13.63)	0.2639 [*]
Small intestine	0	2	-	1.0000 [*]
Colorectal (including rectosigmoid junction)	2	72	0.14 (0.03, 0.56)	0.0003 ^{**}
Anus, anal canal, and anorectum	0	6	-	0.5981 [*]
Liver and intrahepatic bile duct	3	8	1.87 (0.50, 7.06)	0.4082 [*]
Gallbladder	0	0	-	-
Pancreas	0	19	-	0.0594 [*]
<i>Respiratory system</i>				
Nasal cavity, middle ear, and accessory sinuses	0	0	-	-
Larynx	1	8	0.62 (0.078, 4.99)	1.0000 [*]
Lung and bronchus	5	68	0.37 (0.15, 0.91)	0.0262 ^{**}
Trachea, mediastinum, and other respiratory organs	0	1	-	1.0000 [*]
<i>Bones and joints</i>	0	10	-	0.3865 [*]
<i>Soft tissue (including heart)</i>	2	12	0.83 (0.19, 3.72)	1.0000 [*]
<i>Skin</i>				
Malignant melanomas	4	43	0.46 (0.17, 1.29)	0.1695 [*]
Other malignant skin neoplasms	5	116	0.21 (0.09, 0.53)	<0.0001 ^{**}
<i>Breast (male and female)</i>	3	179	0.08 (0.03, 0.26)	<0.0001 ^{**}
<i>Female genital system</i>				
Cervix	0	16	-	0.0918 [*]
Corpus (Uterus, not otherwise specified)	2	18	0.55 (0.13, 2.39)	0.5597 [*]
Ovary	1	21	0.04 (0.01, 0.30)	<0.0001 ^{**}
Vagina	0	0	-	-
Vulva	0	2	-	1.0000 [*]
Other female genital organs	1	1	4.99 (0.31, 79.79)	0.3060 [*]
<i>Male genital system</i>				
Prostate	1	141	0.04 (0.01, 0.25)	<0.0001 ^{**}
Testis	8	12	3.33 (1.36, 8.15)	0.0052 [*]
Penis	0	3	-	1.0000 [*]
Other male genital organs	1	1	4.99 (0.31, 79.79)	0.3060 [*]
<i>Urinary system</i>				
Urinary bladder	3	25	0.60 (0.18, 1.98)	0.6105 [*]
Kidney and renal pelvis	1	0	-	0.1670 [*]
Ureter	0	0	-	-
Other urinary organs	0	2	-	1.0000 [*]
<i>Eye and orbit</i>	0	3	-	1.0000 [*]
<i>Brain and other nervous system</i>	5	23	1.09 (0.41, 2.86)	0.0814 [*]
<i>Endocrine system</i>				
Thyroid	1	48	0.10 (0.01, 0.75)	0.0031 ^{**}
Other endocrine (including thymus)	0	5	-	0.5980 [*]
<i>Lymphomas</i>				
Hodgkin's disease	3	9	1.66 (0.45, 6.15)	0.4353 [*]
Non-Hodgkin's lymphomas	5	44	0.57 (0.22, 1.43)	0.3351
<i>Multiple myeloma</i>	0	0	-	-
<i>Leukemias</i>	20	47	2.12 (1.26, 3.59)	0.0039 [*]
<i>Miscellaneous malignant neoplasms</i>	8	130	0.31 (0.15, 0.63)	0.0006 [*]

CONCLUSIONS

Our findings indicate that, relative to individuals without DS, individuals with DS experience less of the following cancers:

- colorectal
- lung and bronchus
- breast
- ovarian
- prostate
- thyroid
- malignant skin neoplasms
- miscellaneous malignant neoplasms

Relative to individuals without DS, individuals with DS experience more:

- testicular cancer
- leukemias

These findings support prior research, showing low incidence of solid tumors and increased incidence of blood cancers like leukemia among individuals with DS and testicular cancer among men with DS.

Considering this, cancer prevention and screening guidelines should be reconsidered in the context of this unique population. Identification of potential cancer often relies on self-report of symptoms, but this can be more difficult for individuals with DS. Research shows symptoms among individuals with DS are often detected later and by accident, typically by medical professionals during care for another issue or by a perceptive caregiver.¹

Additional research should be conducted to validate these findings using other data sources and to explore factors related to cancer development. Furthermore, research investigating protective genetic and lifestyle factors for cancer, particularly solid tumors, should be explored among individuals with DS.

REFERENCES

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