PREVALENCE OF CANCER AMONG LARGE COHORT OF INDIVIDUALS WITH DOWN SYNDROME: IMPLICATIONS FOR SCREENING GUIDELINES

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Background

- Cancer is the second leading cause of death globally.
- Current US cancer screening guidelines reflect the general population.
- Individuals with Down syndrome (DS) have significantly different odds of cancers compared to the US general population.
- To adhere to current US cancer screening guidelines, individuals with DS may be subject to unnecessary risk and trauma.

Objective

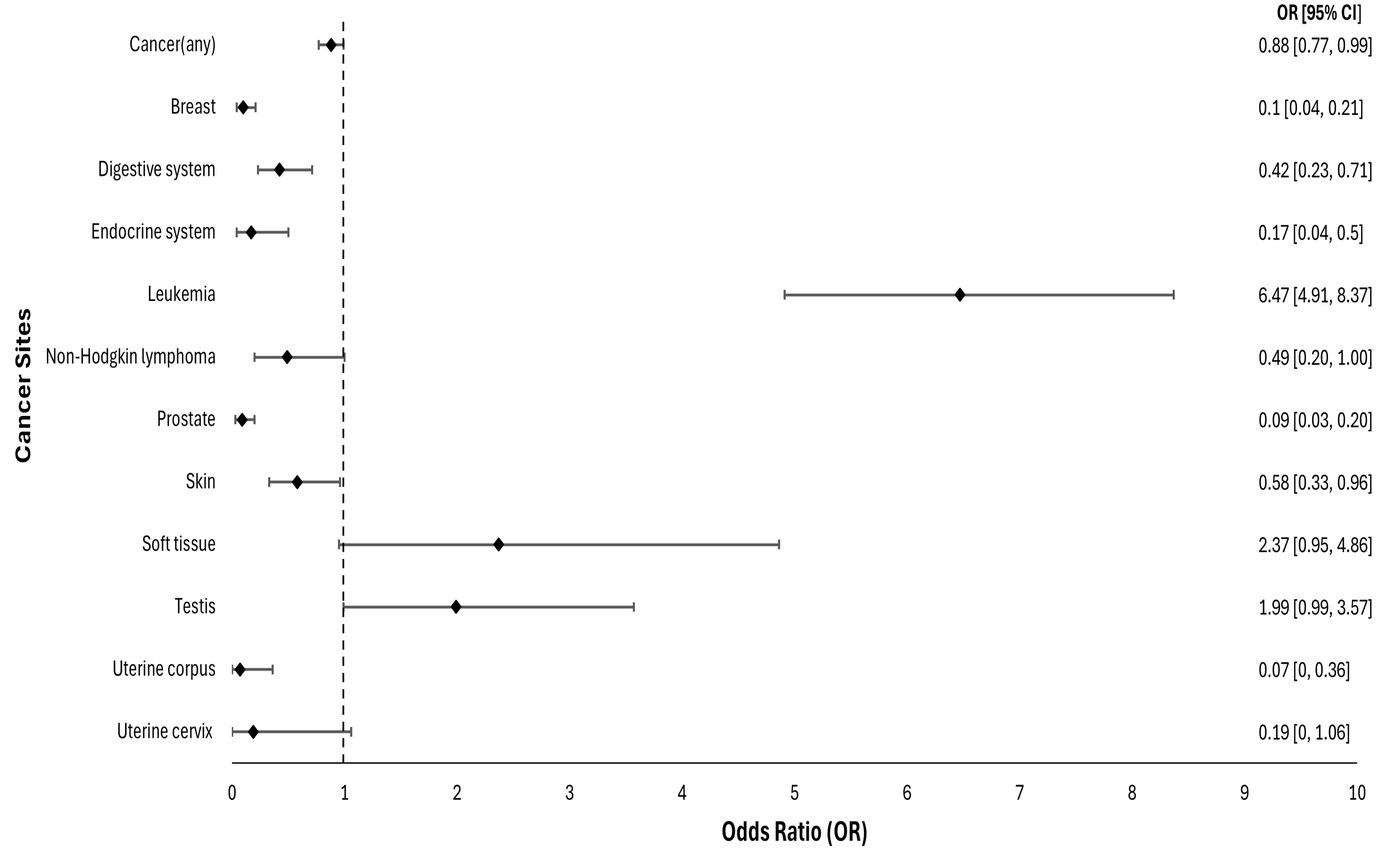
- To calculate prevalence across specific cancer types among a large U.S.-based sample of patients with DS.

Methods

- Retrospective cohort study of 24 years of electronic health record (EHR) data (2000-2024) in the Advocate Health (AH) system.
- Calculated point prevalence of cancer types as defined by ICD-10 codes.
- Compared prevalence in DS study sample to US population using 2022 SEER data.

Patient Characteristics

Patient Characteristics (N = 5895)	
Patient's Age	
Mean (SD)	30 (19)
Median (Q1, Q3)	28 (14, 44)
Total encounters	
Mean (SD)	39 (55)
Median (Q1, Q3)	19 (7, 47)
Sex	
Male	3,040 (51.57%)
Female	2,855 (48.43%)
Race/Ethnicity	
NH White	3,720 (69.55%)
NH Black	526 (9.83%)
Hispanic	847 (15.83%)
NH Asian	173 (3.23%)
NH Other Race	83 (1.55%)
Missing	546
Insurance status	
Commercial	1,736 (29.49%)
Medicare	2,208 (37.51%)
Medicaid	1,647 (27.98%)
Other/Uninsured	296 (5.03%)
Missing	8



Results

Reduced odds of cancer in DS

- 12% overall (p=0.0308)
- 83% endocrine system (p<0.0001)
- 51% Non-Hodgkin lymphoma (p=0.0472)
- 42% skin (p=0.0295)
- 58% digestive system (p=0.0003)
- 81% uterine cervix (females only) (p=0.0746)
- 90% breast (females only) (p<0.0001)
- 91% prostate (males only) (p<0.0001)

Increased odds of cancer in DS

- 137% soft tissue (p=0.0321)
- 547% leukemia (p<0.0001)
- 99% testes (males only) (p=0.0301)

KEY TAKEAWAYS

- **To prevent unnecessary risk and trauma, current national screening guidelines should be modified to reflect lower or higher cancer prevalence in the DS population.**
- **While screening the general population might be beneficial for early detection efforts, the burden to screen individuals with DS for low-prevalence cancers might not be necessary.**
- **More research is needed to ensure the most accurate screening guidelines for the DS population.**

Discussion

Screening recommendations for people with DS should be reviewed due to differences in screening and treatment risks, life expectancy, and differences in cancer prevalence.

Conclusions

- Given the low prevalence of top cancers among individuals with DS, it could be argued that invasive or physically difficult screenings, such as mammograms and colonoscopies, may not be necessary or could be de-emphasized in this population.
- There are screening guidelines for colon, breast, and prostate cancer; however, the prevalence of these cancers in people with DS is significantly lower than the general population.
- Screening is not recommended for testicular cancer for the US population. However, it is significantly more prevalent in DS and presents at more advanced stages due to limited self-report of symptoms.
- Protection from some cancer types potentially could be explained by behavioral differences in people with DS, such as lower rates of sexual activity leading to lower risk of cervical cancer, or lower rates of smoking leading to lower risk of lung cancer. The protection from other cancer types is likely driven by genetic effects of trisomy 21.

See authors for references.

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