Advance Care Planning Discussions Among Adult Patients with Down Syndrome: A Single-Institution Retrospective Study

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BACKGROUND

- **Public health significance:** Adults with Down syndrome (DS) are living longer while facing accelerated aging, increasing the need for advance care planning.[1-4] Serious illness conversations (SIC) can lead to well-informed decision-making and improved quality of life.[5]
- **What is known:** Adults with DS are disproportionately affected by early-onset dementia, which is their #1 cause of death. These patients also frequently require assistance in making complex medical decisions.[6,7]
- **Gaps:** Advance care planning (ACP) is underutilized in the general population, with even lower rates among older adults with DS.[8,9]

OBJECTIVE

To evaluate the occurrence and content of documented ACP conversations between healthcare providers, adults with DS, and their caregivers at the Massachusetts General Hospital Down Syndrome Program.

METHODS

- **Study design:** Retrospective chart review of adults with Down syndrome aged ≥21 years enrolled in the Massachusetts General Hospital (MGH) Down Syndrome Program between February 1, 2023 to February 12, 2025.
- **Patient identification:** Patients were identified by registration with the MGH Down Syndrome Program. Patients with at least one healthcare encounter in the electronic health record (EHR) were included.
- **Data source:** All data were abstracted from the EMR and securely stored in Dropbox in accordance with MGH institutional guidelines.
- **Natural language processing:** Clinical Regex software identified ACP-related keywords within four domains: goals of care (GOC), code status limitations (CSL), palliative care (PC), and hospice (HOS).
- **Data abstraction:** Three independent reviewers evaluated the context surrounding keyword-identified notes using the Serious Illness Conversation Guide. Meaningful documentation was defined as sufficient detail for other providers to understand the goals or outcomes discussed and was coded as present (1) or absent (0) for each ACP domain.
- **Analysis:** This was a descriptive study with results reported as counts (n) and percentages (%).

RESULTS

Study cohort

- Included 327 Down syndrome patients with a cumulative 9,953 clinical notes.

Overall ACP documentation:

- 31 (9.5%) patients had any documented ACP discussion across the four evaluated domains.
- 296 (90.5%) patients had no documented ACP discussions in any domain across their medical records.

ACP documentation by domain:

- GOC discussions were the most frequent, identified in 27 patients (8.3%)
- CSL were documented in only 7 patients (2.1%)
- Discussions involving HOS and PC were rare, each present in just 3 patients (0.9%).

Figure 1. Patients with any meaningful documented ACP discussion

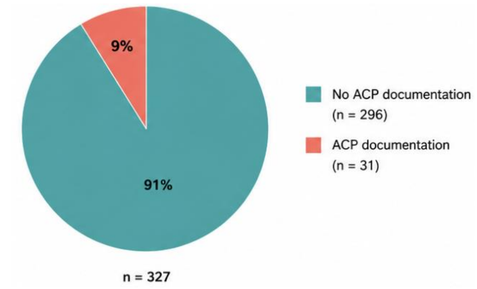

Figure 2. Distribution of patient-specific ACP documentation by domain.

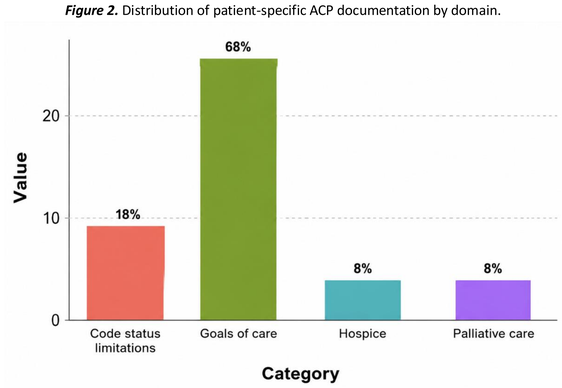

Table 1. Patient demographics (n = 335)

Age		**Sex**	
Mean	36.8	Male	178 (53.1%)
Median	34.6	Female	157 (46.9%)
Range	22.1 - 69.6		
Race		**Ethnicity**	
White	292 (87.2%)	Not Hispanic	310 (92.5%)
Black/African American	13 (3.9%)	Hispanic	15 (4.5%)
Asian	7 (2.1%)	Unavailable/Declined	10 (3.0%)
Race not listed	15 (4.5%)	**Clinical Status**	
Other/Declined	8 (2.4%)	Alive	304 (90.7%)
		Deceased	15 (4.5%)
		Missing	16 (4.8%)

- **Age distribution:** Predominantly young-to-middle-aged adult population (mean = 36.8 years).
- **Race distribution:** Predominantly White (87.2%), with much smaller representations of Black/African American (3.9%), Asian (2.1%), and other or unreported racial groups.

Table 2. Inter-rater reliability with Cohen's Kappa across ACP domains

Domain	Kappa	Interpretation
CSL	0.7872	Substantial agreement
GOC	0.7248	Substantial agreement
HOS	0	No agreement
PC	0	No agreement

- **Cohen's Kappa:** Cohen's kappa indicated substantial interrater agreement for CSL and GOC, with no agreement observed for HOS and PC.
- **Double-coding inter-rater test:** Interrater agreement calculated was 98.125%.

CONCLUSIONS

- Our findings highlight a significant opportunity to prioritize conversations about advance care planning for adults with Down syndrome and their caregivers.
- As the population continues to age, supporting advance care planning will become increasingly important to help individuals and families navigate end-of-life decision-making.
- Future quality improvement initiatives could focus on implementing structured ACP documentation using standardized communication frameworks, such as the Serious Illness Conversation Guide.
- Future research could evaluate the impact of co-occurring conditions, including Alzheimer's disease, on ACP documentation and compare rates between individuals with and without an AD diagnosis.
- Future efforts should also focus on connecting patients and caregivers with resources, including the Massachusetts Down Syndrome Congress (MDSC) Aging Center, Mary-Frances Garber, and the Alzheimer's Association.

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