

FAST FACTS AND CONCEPTS #243
PALLIATIVE CARE FOR PATIENTS WITH DOWN SYNDROME

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Background Down syndrome (DS), or Trisomy 21, is the most common chromosome abnormality among liveborn infants, characterized by dysmorphic features, impaired intellectual ability, various cardiac septal defects, short stature, and a reduced life expectancy. This *Fast Fact* discusses the natural history of DS and issues specific to palliative and end-of-life care for patients with DS. *Fast Facts* #192 & 193 discussed end-of-life care for patients with developmental disabilities in general.

Causes of Morbidity and Death in DS Patients with DS frequently live to 60 years of age, men somewhat longer than women.

- Childhood mortality is most often associated with congenital heart defects or leukemia. The risk of developing childhood acute lymphoblastic leukemia is ten to twenty times higher in DS than the general population. Leukemia is treatable, although recurrences typically occur with an aggressive and terminal course (3-5).
- Midlife mortality is associated most often with pulmonary disease and problems related to congenital cardiac defects. The incidence of coronary artery disease and solid-tumor malignancies is actually lower in people with DS than in the general population.
- People with DS have a much higher incidence of dementia of the Alzheimer's type (DAT) than the general population, and tend to develop DAT in their 40s and 50s. By age 60, 75% of individuals with DS have developed DAT. Brain autopsies reveal amyloid plaques and neurofibrillary tangles; this may be due to chromosome 21 housing the amyloid precursor protein gene.
- In addition to cognitive issues, other common medical issues may include hearing impairment, cataracts, sleep apnea, dental issues, congenital cardiac septal defects, thyroid dysfunction, seizures, arthroses, osteoporosis, chronic constipation, GERD, incontinence, congenital hip dislocation, behavioral issues, and recurrent respiratory infections (6).
- Partial or tonic-clonic seizures are most common in the first year of life and in or after the third decade (7,8). Once seizures occur, the pace of functional decline often increases.
- Routine symptom management principles apply to patients with DS, acknowledging communication limitations that can limit comprehensive assessment. Polypharmacy can contribute to or exacerbate issues so rigorous care must be paid when medications are started or stopped.

Psychosocial issues Psychosocial domains include communication, self-care, grief, and family circumstances. In addition, there are unique issues that may complicate end-of-life decision making.

- Patients with DS and their loved ones and caregivers have experienced a lifelong disease trajectory, which includes mental retardation, medical, and psychosocial issues. The lifelong toll on families is high. Part of a robust plan of care includes acknowledgment of this toll by healthcare providers.
- Many people with DS reside in institutional settings where the primary caregivers are not family members. Many have lost parents to death and sometimes have no contact with other family. While it is not known how people with DS process these losses, maximal supports regarding grief and loss should be put in place.
- Issues such as guardianship and advance care planning should be addressed as early as possible with caregivers of people with DS.
- Whenever possible, decision makers for people with DS should be encouraged to use substituted judgment to make key palliative care decisions. All efforts should be made to determine the preferences of the patient, however because of lifelong cognitive impairment, the views of the person with DS may not be known. There may also be disability rights concerns that make proxies/guardians wary of not instituting all life-sustaining measures. Intensive education needs to be provided regarding the benefits and burdens of any medical interventions, with particular emphasis on how these will impact quality of life. Healthcare providers need to make sure that their own views about quality of life do not interfere with respecting the wishes of designated decision-makers – see *Fast Fact* #193.

Key teaching points

- The signs of aging and dementia that occur in the general population occur in patients with DS at a much younger age and with a different pattern.
- Prognostication in people with DS is different than for the general population. Although no guidelines exist, the end of life may be recognized late in Down Syndrome and accompanied by a precipitous decline. The presence of DAT and/or seizures should be considered when making assessments such as eligibility for hospice.
- Care may be complicated by behavioral and assessment difficulties as well as caregiver issues.
- Because patients with DS often have a longstanding lack of competence, proxies/guardians will typically not be able to utilize substituted judgment in making key decisions. Intensive education regarding how medical interventions will affect quality of life is a key part of any plan of care.

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