Down Syndrome and Dementia: Guide to Identification, Screening, and Management

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ABSTRACT

Down syndrome (DS) is an intellectual disability due to the genetic disorder trisomy 21. Many individuals with DS are living into middle and older adulthood, experiencing chronic health problems, and are at risk for dementia. The purpose of this article is to describe the primary care management of adults with DS, the relationship between DS and Alzheimer's dementia, and screening protocols for primary care.

Keywords: aging, dementia, developmental disability, Down syndrome, screening

INTRODUCTION

Down syndrome (DS) is a common genetic disorder with over 300,000 persons affected in the United States.[1] Many individuals with DS are living into middle and older adulthood and experiencing chronic health problems related to DS as well as aging. Described as "the triple challenge," adults with DS have baseline cognitive and functional deficits, experience accelerated aging, and are at high risk for development of Alzheimer's disease (AD).[2] With a median life expectancy of approximately 60 years, up from just 12 years in the 1950s,[2] nurse practitioners (NPs) are now evaluating aging persons with DS who have chronic disease along with new cognitive deficits. This population is also expected to grow in the future. From 1979 to 2003, the prevalence of DS cases increased by 31%, from 9 to 12 per 10,000 live births in 10 US regions.[3] Thus, NPs can expect to care for adults with DS in primary care.

Adults with DS have a unique risk for developing AD. Risk estimates vary, but the National Down Syndrome Society (NDSS) reports that nearly 25% of individuals with DS over the age of 35 show signs of Alzheimer's-type dementia and this percentage increases with age.[4] By age 40, most will have some characteristic brain lesions (amyloid plaque and neurofibrillary tangles) that meet neuropathologic criteria for AD,[5,6] and the risk of developing AD increases incrementally with age.[2] The clinical signs of AD may not be as obvious in adults with DS, due to their baseline cognitive impairment, and often present as a change in function, behavior, or performance of activities of daily living. Our purpose in this article was to describe the common primary care health problems, tools, and diagnostics useful in evaluating new-onset cognitive, behavioral, or functional (CBF) changes as well as health management of middle-age and older adults with DS.

PATHOPHYSIOLOGY OF DS AND ITS RELATIONSHIP TO AGING

The relationship between DS, AD, and accelerated aging is explained by the genetic changes in protein expression. DS is a genetic disorder that occurs due to an extra copy (or partial copy) of chromosome 21. The abnormal cell division results in abnormalities in physical and developmental features due to extra DNA. The extra DNA on chromosome 21 leads to overexpression of amyloid precursor protein, which

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is broken down (cleaved sequentially by β- and γ-secretases to release Aβ) to form senile plaques in the brains of persons with AD and DS.[7] The additional DNA is also believed to explain the body's rapid aging process (accelerated aging) due to overexpression of various proteins related to oxidative damage in the brain.[8] The normal aging process in adults with DS is not well understood and more evidence is needed on expected cognitive changes.[4]

Adults with DS are at high risk for AD, yet it can be difficult to detect early signs of AD due to the baseline memory and cognitive impairment. When evaluating adults with DS who present with cognitive, behavioral, or functional (CBF) change, it is important that NPs rule-out alternative etiologies by taking the following steps:

- Medication review.
- Thorough health history for chronic diseases.
- Comprehensive physical examination.
- Ordering and evaluation of diagnostic study results.

MEDICATION REVIEW FOR CBF CHANGES

The risk of "cascading pharmacy" (medicines that are overlooked as a likely cause of new symptoms)[9] and performance of medication reconciliation at each encounter cannot be overstated.[10] NPs need to thoroughly review medications with a focus on the newly added agents. Medications with psychoactive, sedating, antiepileptic, or anticholinergic properties are common causes of cognitive changes (delirium).[11] Polypharmacy is common and thus drug-drug interactions should be reviewed assessed for nonspecific signs and symptoms, such as somnolence, gait instability, and urinary retention. Collaboration with a pharmacist may be useful in evaluating drug-drug interactions and for the pharmacy cascade. The NP should consider assessing for adherence to medication dose and frequency, as nonadherence can also be a source of CBF changes.[11,12]

CHRONIC HEALTH PROBLEMS

Many chronic health conditions (multimorbidity) are associated with DS, can be exacerbated during aging, and may be associated with CBF changes. Adults with DS may have multiple (4 or more) comorbid conditions due to underlying physical abnormalities as well as metabolic effects of long-term medication use, such as seizure or antipsychotic medications.[11] Health problems associated with aging and DS are numerous and may be etiologies of cognitive changes, and their presentations may differ from the usual aging patterns (see Box 1).[13] For example, thyroid disorders have been reported to have a 40% prevalence in the DS population as compared with a 10% prevalence in an age-matched population.[2] Thus, screening for hypothyroidism is recommended more often in this population and the presenting symptoms may include fatigue or difficulty concentrating.

Sensory changes are a common primary health care problem and may contribute to CBF changes. Age-associated visual deficits develop at a younger age in DS than that in the general population.[4] Adults with DS often develop vision loss due to cataracts and keratoconus.[10] Cataracts develop earlier in adults with DS, which impairs vision. Keratoconus causes the round cornea to become cone-shaped, which can lead to a distortion of vision.[11]

Persons with DS often have hearing impairment. The accelerated aging process places them at high risk for conductive hearing loss. Craniofacial anomalies associated with DS also place them at risk for hearing

Box 1. Differential Diagnoses

The differential diagnoses for cognitive, behavioral, or functional changes may include[10,16,34]:

- Delirium
- Hypothyroidism
- Depression or other psychiatric disorders (eg, anxiety or phobias)
- Obstructive sleep apnea
- Medication—adverse response, especially anticholinergics
- Sensory changes or deprivation (hearing and vision loss)
- Acute or chronic hepatitis
- Urinary tract infection
- Changes in environment
- Bereavement and grief
- Emotional or psychological abuse
- Neoplasms
- Normal pressure hydrocephalus
- Anemia due to folic acid deficiency
- B_{12} deficiency
- Sexually transmitted infections