

# Health Care Management of Adults with Down Syndrome

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The family physician's holistic approach to patients forms the basis of good health care for adults with Down syndrome. Patients with Down syndrome are likely to have a variety of illnesses, including thyroid disease, diabetes, depression, obsessive-compulsive disorder, hearing loss, atlantoaxial subluxation and Alzheimer's disease. In addition to routine health screening, patients with Down syndrome should be screened for sleep apnea, hypothyroidism, signs and symptoms of spinal cord compression and dementia. Patients with Down syndrome may have an unusual presentation of an ordinary illness or condition, and behavior changes or a loss of function may be the only indication of medical illnesses. Plans for long-term living arrangements, estate planning and custody arrangements should be discussed with the parents or guardians. Because of improvements in health care and better education, and because more people with this condition are being raised at home, most adults with Down syndrome can expect to function well enough to live in a group home and hold a meaningful job. (*Am Fam Physician* 2001;64:1031-8,1039-40.)

▶ A patient information handout on health issues in adults with Down syndrome, written by the author of this article, is on page 1039.

Members of various family practice departments develop articles for "Practical Therapeutics." This article is one in a series coordinated by the Department of Family and Community Medicine at the Medical College of Wisconsin, Milwaukee. Guest editors of the series are Linda N. Meurer, M.D., M.P.H., and Douglas Bower, M.D.

**D**own syndrome (trisomy 21) occurs in about one in 1,000 live births.<sup>1</sup> Although the risk increases with increasing maternal age, most infants with Down syndrome are born to mothers of typical childbearing age. About 250,000 families in the United States are affected.<sup>2</sup> Average life expectancy of persons with Down syndrome has increased into the middle 50s,<sup>3</sup> and a person with Down syndrome who lived to age 83 has been reported.<sup>4</sup> Most persons with Down syndrome are functioning in our com-

munities, sometimes with minimal support, for many years of adult life.

Certain clinical conditions occur more commonly in persons with Down syndrome (*Table 1*). It is important to be aware of these common problems because clinical features of disease can be difficult to recognize. The physician may attribute symptoms to Down syndrome instead of to a new disease process because of the difficulty of obtaining a good history in light of limited expressive speech, a decreased tendency to complain of pain and a tendency to manifest medical problems as behavior problems.

TABLE 1  
Selected Medical Conditions with a Higher Prevalence in Adults with Down Syndrome\*

Endocrine	Otolaryngology	Cataracts, refractive errors and keratoconus
Thyroid disease—hypothyroidism and hyperthyroidism	Obstructive sleep apnea	Seizures
Diabetes mellitus	Hearing loss	Testicular cancer
Mental health	Musculoskeletal	Xerodermatitis
Depression	Spinal cord compression	Acquired valvular heart disease, including mitral valve prolapse
Obsessive-compulsive disorder	Atlantoaxial subluxation	
Abuse (physical or sexual)	Periodontal disease	
Conduct disorder	Alzheimer's disease	

\*—Listed in approximate order of clinical importance.

*Patients with Down syndrome often present with behavior problems when they actually have underlying medical problems.*

### Health Care Guidelines

Health-related conditions beyond those of the general population that should be screened for in patients with Down syndrome are presented in detail in the “Health Care Guidelines for Individuals with Down Syndrome,”<sup>5</sup> the most widely published health care guidelines that include adults. Health care screening in persons with Down syndrome has not been well studied. So far, most relevant studies have focused on descriptions of the higher prevalence of a condition in persons with Down syndrome as a cause of significant morbidity or on case studies of conditions affecting these patients. The Health Care Guidelines are derived from the consensus of a panel of the Down Syndrome Medical Interest Group and based on available evidence.

### Usual Health Maintenance Needs

Adults with Down syndrome have the same basic health care needs as typically developed people, including health screening and prevention. Immunization schedules are the same. Screening for hypertension and heart disease, and disease surveillance are no different. Although solid tumors are less common

in persons with Down syndrome than in the general population,<sup>6</sup> until more data are available, cancer screening should be the same in this group as it is in the general population.

Diabetes has a higher prevalence in adults with Down syndrome than in the general population.<sup>7</sup> Although there are no specific recommendations regarding diabetes in persons with Down syndrome, it may be reasonable to screen these patients for this disease. A fasting plasma glucose level of 126 mg per dL (7.0 mmol per L) or higher, a plasma glucose level of 200 mg per dL (11.1 mmol per L) or higher two hours after a 75-g glucose load, or a random glucose level of 200 mg per dL or higher on two occasions are diagnostic of diabetes mellitus.

An annual testicular examination in men may be prudent because of the higher prevalence of testicular cancer in this group.<sup>6</sup> Because the prevalence of abuse in patients with disabilities is higher than in the general population,<sup>8</sup> screening for and counseling about abuse has special importance. In addition, these patients should be counseled about diet, exercise, obesity, smoking, alcohol use, accident prevention and contraception.

### GYNECOLOGIC EXAMINATION

The gynecologic examination may be difficult to perform because of poor cooperation. If attempts to educate a sexually active patient about the Papanicolaou (Pap) smear and pelvic examination are not successful, a modified Pap smear may have some diagnostic value.<sup>9</sup> The physician can insert a finger into the vagina and slide the cytology brush or swab along the finger into the cervical os.<sup>9</sup> If the bimanual examination cannot be done, pelvic ultrasound examination is an option.

A risk-benefit decision must be made about doing the gynecologic examination with sedation,<sup>10</sup> if other measures fail. Oral ketamine and midazolam, administered under the supervision of an anesthesiologist, have been recommended.<sup>10</sup> The author has used oral midazolam (Versed) or intravenous

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conscious sedation when only mild sedation is needed.

If the examination is performed with sedation, it may be helpful to combine it with other procedures such as blood tests, echocardiography, flexible sigmoidoscopy or mammography. Invasive procedures, however, require informed consent from an adult patient with Down syndrome, unless there is a court-appointed guardian.

### Specific Health Maintenance Needs

Figure 1 outlines the specific elements of health maintenance in adult patients with Down syndrome.

#### CARDIAC DISEASE

Mitral valve prolapse and valvular regurgitation occur in as many as 57 and 17 percent of adults with Down syndrome, respectively.<sup>11-13</sup> Adults without known cardiac disease can develop valve dysfunction.<sup>5</sup> Careful auscultation is probably sufficient to screen for valvular regurgitation, which, if found, requires bacterial endocarditis prophylaxis. The diagnosis can be confirmed with echocardiography. Bacterial endocarditis prophylaxis started after repair of congenital heart disease should be continued if recommended by American Heart Association guidelines and the consulting cardiologist.

#### OTOLARYNGOLOGIC DISEASE

Conductive and sensorineural hearing losses occur in up to 70 percent of persons with Down syndrome and may not develop until early adulthood.<sup>14-17</sup> Because of their poorer communication skills, these persons may not be able to communicate that they are having difficulty hearing. Poor hearing will further complicate speech problems. These persons also may appear to be stubborn when they do not respond to requests they have not heard. The sensory deprivation associated with hearing loss may contribute to delirium. Auditory testing every two years is recommended in persons with Down syndrome.

*Adults with Down syndrome should have all of the same age-appropriate preventive care as unaffected persons, in addition to special screening.*

Obstructive sleep apnea occurs in up to 50 percent of persons with Down syndrome.<sup>18,19</sup> In this group, it is not always associated with obesity and may be related to the hypotonia and structural abnormalities associated with Down syndrome. Apnea, snoring, unusual sleeping positions, daytime somnolence, obesity and a patulous uvula with erythema are all associated with obstructive sleep apnea.

Apnea in a person with Down syndrome may be expressed as psychologic symptoms such as irritability, depression, paranoia and other behavior changes. A sleep study will usually confirm the diagnosis. If obstructive sleep apnea is present, an otolaryngologic evaluation is required. Untreated obstructive sleep apnea can lead to cor pulmonale. Although continuous positive airway pressure devices can be difficult for many persons to tolerate, some patients with Down syndrome readily accept them.

#### OPHTHALMOLOGIC DISEASE

Cataracts occur in up to 13 percent of persons with Down syndrome. Keratoconus occurs in up to 15 percent, and 25 to 43 percent of these persons have refractive error.<sup>20-23</sup> An ophthalmologic examination is recommended every two years.

#### HYPOTHYROIDISM

Hypothyroidism occurs in 10 to 40 percent of persons with Down syndrome.<sup>24,25</sup> In addition, this group has a slightly higher incidence of hyperthyroidism. Because many of the signs of hypothyroidism may be confused with features of Down syndrome, the clinical diagnosis of hypothyroidism is difficult.<sup>26</sup> Furthermore, undiagnosed hypothyroidism can contribute to dementia<sup>27</sup> or be misdiag-

## Health Care for Adults with Down Syndrome

Age										
Usual preventive medicine care*										
Cardiology† auscultation										
Audiologic evaluation										
Ophthalmology referral										
Thyroid (TSH)										
Neurologic examination‡										
Assess biannual dental examination										
Reproductive health§ and testicular examination										
Development, depression, stress										
Nutrition and obesity										
Family and parent group information										
Estate planning										
Living arrangements and employment										

\*—Common medical problems, as well as special medical problems, may affect the person with Down syndrome. It cannot be assumed that a problem is due solely to the condition of Down syndrome.

†—Examine by auscultation for mitral valve prolapse and aortic regurgitation. If suspected, obtain an echocardiogram. Start endocarditis prophylaxis as indicated by American Heart Association guidelines.

‡—To assess for spinal cord compression from atlantoaxial instability, test gait, tone, Babinski responses, deep tendon reflexes and clonus. Assess for neck pain, torticollis, gait disturbances, spasticity and weakness. Advise the family or caregiver of symptoms to report to the physician.

§—Assess need for contraception. Obtain Papanicolaou smears and mammograms as indicated by standard guidelines. Consider pelvic ultrasonography or examination under anesthesia if the patient refuses a bimanual examination. Perform testicular examination.

||—A periodic assessment should be performed using a team approach involving occupational, physical and speech therapists, as indicated. If there are areas of loss of function, assess for thyroid function, depression, stress, Alzheimer's disease, sensory deficits, sleep patterns suggestive of sleep apnea, and common medical conditions with an unusual presentation. Do not assume Alzheimer's disease. Include information about supportive employment and self-advocacy for adults. Refer parents or caregivers to a parent advocacy group.

**FIGURE 1.** Down Syndrome Health Care Guidelines for Adults record form. The physician should perform the evaluation if the space is not shaded and record the dates of the evaluations in the blank spaces. (TSH = thyroid-stimulating hormone)

*Adapted with permission from Cohen WI, Patterson B, eds. Health care guidelines for individuals with Down syndrome: 1999 revision (Down syndrome preventive medical checklist). Down Syndrome Medical Interest Group. Down Syndrome Quarterly 1999;4:1-15.*

nosed as dementia.<sup>24,28</sup> Thyroid-stimulating hormone levels should be assessed annually in patients with Down syndrome.

#### ATLANTOAXIAL INSTABILITY

Up to 14 percent of persons with Down syndrome have evidence of atlantoaxial instability on plain radiographs of the neck.<sup>29</sup> From 1 to 2 percent of all persons with Down syndrome have symptoms of atlantoaxial subluxation and, in some cases, symptoms have appeared after surgery for other conditions.<sup>30</sup> Cervical radiographic screening for atlantoaxial instability is no longer recommended routinely in adults, but cervical radiography in neutral, flexed and extended positions should be considered if any of the signs or symptoms are present (Table 2). There should be no more than 5 mm of space between the posterior segment of the anterior arch of C1 and the anterior segment of the odontoid process of C2.<sup>5</sup> Family members or supervising staff should be advised to monitor for symptoms (Table 2), and an annual neurologic examination should be performed to look for upper motor neuron signs of spinal cord compression. Even if atlantoaxial instability is not pre-

*In adults with Down syndrome, sleep apnea can be expressed as irritability, depression, paranoia and other behavior changes.*

sent, cervical arthritis can be severe and may manifest largely as a refusal to move.

#### MENTAL ILLNESS

Mental illness occurs in about 30 percent of persons with Down syndrome. Using Diagnostic Research Criteria (DCR-10), depression, obsessive-compulsive disorder and conduct disorder are the most common mental illnesses.<sup>31</sup> Depression is a common cause of reduced function in persons with Down syndrome.<sup>24</sup> Loss of a friend, family member or other significant person, or a change of work, school or living arrangement can trigger depression. In addition, medical conditions can be associated with depression. Schizophrenia and psychosis are considered to be less common in persons with Down syndrome. Self-talk, or soliloquy, is fairly common, particularly in stressful situations, and is usually mental-age appropriate. It is sometimes incorrectly considered hallucinatory behavior in adults with Down syndrome.<sup>32</sup>

Selective serotonin reuptake inhibitors (SSRIs) are preferred to tricyclic antidepressants in the treatment of depression in this group of patients because anecdotal evidence indicates a high incidence of anticholinergic side effects from the latter agents. Counseling can be difficult to obtain for the adult with mental illness and Down syndrome. Local parent-support groups may be able to provide names of counselors who have worked well with cognitively delayed adults.

#### ALZHEIMER'S DISEASE

Alzheimer's disease in persons with Down syndrome has prevalence rates of zero to 10 percent in ages 30 to 39, 10 to 25 percent in ages 40 to 49, 28 to 55 percent in ages 50 to 59,

TABLE 2

#### Signs and Symptoms of Atlantoaxial Instability in Down Syndrome

##### Signs

Gait disturbances  
Spasticity  
Abnormal neurologic reflexes  
Babinski responses  
Increased deep tendon reflexes  
Weakness  
Clonus

##### Symptoms

Neck pain  
Torticollis  
Change in gait  
Loss of upper or lower body strength  
Change in bowel or bladder function

and 30 to 75 percent in ages 60 to 69.<sup>33</sup> Typically, the person with Down syndrome will present for the evaluation of dementia because of a change in behavior or loss of function. Symptoms of dementia in persons with Down syndrome include a decline in function, psychologic changes such as depression, obsessive-compulsive disorder and behavior changes, and sleep disorders, memory loss, ataxia, seizures, and urinary and fecal incontinence. Unfortunately, other causes for the same symptoms may be overlooked, and Alzheimer's disease may be overdiagnosed.<sup>34</sup>

As in the general population, Alzheimer's disease in persons with Down syndrome is a diagnosis of exclusion. Assessment for possible Alzheimer's disease in this group includes evaluation for depression or delirium, drug effects, infection, alcoholism and systemic illness.<sup>35</sup> Neuropsychologic testing and radiologic imaging do not accurately diagnose dementia or reliably differentiate depression from dementia.

Care and treatment of patients with Down syndrome and Alzheimer's disease are similar to that of patients in the general population. Because it is sometimes difficult to differentiate pseudodementia from dementia and because depression can coexist with dementia, a trial of treatment with an SSRI may be helpful.<sup>36</sup>

#### **DENTAL DISEASE**

Gingivitis and periodontal disease are more common in persons with Down syndrome<sup>37</sup> and generally cause tooth loss. Dental caries are less common. Orthodontic problems and bruxism are more common than in the general population. The family physician should encourage good dental hygiene that includes brushing of the teeth and gums, and regular flossing. Dental visits should be twice annually and as needed.

#### **HEALTH COUNSELING**

Family physicians can help patients with Down syndrome develop good communica-

tion and social skills that will enhance their ability to live independently, have a job and interact with others. Although the foundation for these skills starts before adulthood, there are still ways to help an adult with Down syndrome function more effectively. Speech and language therapy may improve intelligibility of language. Vocational training and job coaches also are helpful.

A local parent-support group is a valuable resource for information on relationship and sexuality training, abuse prevention, estate planning and independent and group living. Local groups can be found in the telephone book or obtained from national organizations, such as the National Down Syndrome Society.

An adult with Down syndrome is considered competent to make medical decisions unless declared otherwise. The issue of whether or not guardianship is appropriate should be addressed early. When guardianship is not appropriate, the question of advanced directives, especially power of attorney for health care and finance, should be addressed.

Frequently, independent living with supervision is appropriate but not available. Group homes are not appropriate for everyone, and some behavior problems that occur in group homes may be due to the stress of people living together who do not like one another. Some parents have tried to alleviate this problem by buying houses or duplex apartments to better control the living arrangements. Independent but supervised housing is important long-term planning. Parents may die or become incapacitated, and other family members cannot always step in.

Local parent groups can also provide information about estate planning for the family. Frequently, supplemental trusts are used to handle money inherited by the adult with Down syndrome.

Physicians and family members should anticipate stresses that may overwhelm the adult with Down syndrome. Planning can

ensure a successful transition, such as those from home to apartment or from one job to another. Anticipatory planning can also lessen the impact of the loss of access to a friend or family member. The physician should monitor these patients for loss of independence, loss of living skills or function, depression and behavior changes at least annually.

### Special Considerations for the Office Visit

It is best to address the person with Down syndrome directly, to assess expressive language, hear the patient's story and gain the confidence of the patient and family or caregiver for a successful therapeutic alliance. It is unwise to assume that a patient with Down syndrome may refuse a treatment, such as eyeglasses or continuous positive airway pressure. Gentle persistence, a variety of approaches, and psychologic preparation of the patient by the physician and a family member or caregiver can make the difference between acceptance and rejection of a treatment. If more help is needed for the care of a patient with Down syndrome, the family physician can contact or refer to a specialty clinic for persons with Down syndrome.

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