HEALTH GUIDELINES FOR ADULTS WITH DOWN SYNDROME

This article focuses on the specific needs for adults with Down syndrome and is based on Dr. Leigh Wilson’s “Preventive Care for Adults With Down Syndrome,” first published by the American College of Preventative Medicine. It is important to start looking for a knowledgeable primary care physician for your loved one with Down syndrome as early as his or her 14th birthday. The transition from the pediatrician to an adult healthcare provider can be intimidating, so ask your own PCP what they feel about caring for an adult with Down syndrome. If they are hesitant, ask them if they could make a referral to a colleague who has experience and knowledge in this field, or ask other parents. Please remember that you are the advocate for your loved one and it is ok for you to ask questions and to make suggestions to your chosen MD. Many providers, despite their many years of practice, lack a comfort in caring for the adult with special needs. For this reason, it is essential that you empower yourself with the knowledge of the specific needs of individuals with Ds. If you have any further questions, please email me at KathleenF@dsagc.com.

Autoimmune and endocrine diseases. Diseases, such as celiac, diabetes mellitus, and hypothyroidism, have higher prevalence in adults with Down syndrome than in the general population. Adults with Down syndrome face increased risks for celiac disease that can occur at any time. Please note any excessive diarrhea, nausea, abdominal distension, or unexplained lethargy. The initial blood test should be a complete blood count to assess for anemia. If suspicion for disease is high, the next test to have is an antiendomysial and/or antitissue transglutaminase antibody followed by referral to a gastroenterologist.

- Type 1 diabetes is another disorder with a higher prevalence in adults with Down syndrome. Most diagnoses of type 1 diabetes are made in childhood; however, adult healthcare clinicians should be aware of this association and with the increased risk for type 2 diabetes. Fasting blood glucose level should be performed yearly.

- Hypothyroidism is estimated to occur in 10%-40% of all patients with Down syndrome, and can be difficult to detect clinically; it may even masquerade as early-onset dementia. Thyroid-stimulating hormone levels should be checked annually in adults with Down syndrome.
Cancers. Mammography and colonoscopy are recommended at the same time intervals as for similar-age peers. If patients are not able to tolerate mammography, annual breast ultrasounds have been proposed as an alternative. Testicular cancer has been noted to have a higher prevalence in men with Down syndrome than in the general population; therefore, performing annual testicular exams may aid in detection of this cancer.

- Cervical cancer screening with pelvic exam and Pap smear should be performed yearly; however, the screening may be difficult to perform if patient education is not sufficient to gain patient cooperation. If the provider is unable to complete a pelvic exam, a modified exam has been suggested that forgoes a speculum, and instead the provider's gloved finger is inserted to obtain cells for the Pap cytology. Providing reproductive health services to persons with Down syndrome and other cognitive disabilities is extremely important.

Cardiac. From late adolescence on, there is an increased risk for pulmonary hypertension, mitral valve prolapse and aortic valve regurgitation in patients with Down syndrome. The ACPM recommends adults with Down syndrome be followed by a cardiologist if they have a had a congenital heart defect. It is also recommended that adults with DS with no history of heart defect undergo a thorough cardiac exam in early adulthood.

Neurologic. Symptoms of dementia, such as decline in function, memory loss, ataxia, seizures, and incontinence, should first be evaluated for a treatable medical etiology. Spinal stenosis, adult-onset epilepsy, and hypothyroidism have higher prevalence in adults with Down syndrome than the general population. If any signs or symptoms of atlantoaxial subluxation arise, such as gait disturbance, weakness, clonus, or spasticity, ask for a neck x-rays. An association between Alzheimer's disease and Down syndrome has long been noted. However, before making a diagnosis of Alzheimer's disease, make sure your doctor considers differential diagnoses such as hypothyroidism, urinary tract infection, or diabetes.

Mental health. Disorders, such as depression, obsessive-compulsive disorder, abuse, and conduct disorder, occur more frequently than other mental health disorders in individuals with Down syndrome. To differentiate between dementia and depression, the Dementia Screen for Down Syndrome (DSDS) is a validated scale for adults.

Osteoporosis. Osteoporosis is more common in adults with Down syndrome than in the general population. Calcium supplementation of 1000 mg for men and nonmenopausal women and 1500 mg for postmenopausal women (along with vitamin D) has been
suggested. Ask your physician to consider obtaining a dual-energy x-ray absorptiometry scan to screen these patients for osteoporosis.

**Pulmonary.** Sleep apnea is estimated to occur in 50% of people with Down syndrome as a result of physical characteristics associated with the disorder. Sleep apnea can occur in the absence of obesity and can manifest as a change in behavior, such as increased irritability, daytime somnolence, or psychological symptoms -- such as depression. For this reason, sleep studies are recommended throughout adulthood.

**Vision.** Cataracts and keratoconus may develop in adolescence or adulthood and are found to be approximately 4 times more common than in the general adult population. Hypermetropia can develop at any age. Screening by an optometrist or ophthalmologist should occur yearly.

**Hearing.** More than 50% of patients with Down syndrome may have mild-to-severe hearing loss. Conductive and/or sensorineural loss can begin at any age; therefore, audiometry should be performed at least every 2 year

**References**