Continuing Education

Care of Adults With Down Syndrome: The Nurse Practitioner Perspective

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Abstract

Down syndrome is a chromosomal disorder that is commonly diagnosed in childhood. The most referenced care guidelines need updating and focus on children. As a result of advancements in health care, life expectancies for people with Down syndrome have reached middle to late adulthood, leaving providers unprepared for the management of common comorbidities in adults with Down syndrome. To promote quality of life in this population, this overview of emerging recommendations and evidence-based literature through a systems-based approach may help nurse practitioners foster high-quality and cost-effective services in providing care for adults with Down syndrome.

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This activity is designed to augment the knowledge, skills, and attitudes of nurse practitioners and assist in their ability to identify common health risks and co-occurring conditions that adults with Down Syndrome experience, offer new recommendations from evidence-based research, and help develop a transition plan in early adulthood to provide a high level of care for these individuals.

At the conclusion of this activity, the participant will be able to:

a. Explore the genotype determined and phenotypical features that contribute to common co-occurring conditions and health risk in adults with DS
b. Identify the differences in the care guidelines between adults in the general population and those with DS
c. Explain the benefits of transition planning for individuals with DS and their families

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This activity has been awarded 1 Contact Hour of which 0 credit is in the area of Pharmacology. The activity is valid for CE credit until October 01, 2024.

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Background and Significance

Down syndrome (DS) is the most common chromosomal disorder globally and within the United States, where 1 in every 700 births is an infant with DS.1,2 This syndrome is highly associated with advanced maternal age. The prevalence of babies with DS is 20 per 10,000 in women less than 35 years of age, but greater than 120 per 10,000 in those who are 35 years or older.3 Moreover, a mathematical function used in some studies, including a 1978 New York State seminal study, demonstrated an exponential relationship between maternal age and DS where 1 in 1,925 births in women at 20 years old have the diagnosis compared with 1 in 12 in those 49 years of age.4

Currently, cases of DS can be detected prenatally using a cell-free DNA test that utilizes a sample of the mother’s blood as early as 7 weeks.5 An ultrasound to determine the presence of nuchal translucency (fluid behind the neck) should be done in those with a positive result.6 Nuchal translucency of greater than 3.5 mm is associated with chromosomal abnormalities in approximately 25% of pregnancies.6 Definitive confirmation is done with an invasive prenatal test such as chorionic villus sampling, amniocentesis, or percutaneous umbilical blood sampling.1 After birth abnormal phenotypic features require a blood test for karyotyping for accurate diagnosis.1,6,7

In the primary type of DS, an additional chromosome 21 is present in all cells. This results from an error during cell division and replication resulting in an egg with 2 copies of the chromosome instead of 1.1,7,8 Less common genetic derangements may also occur, involving translocations of genetic material from chromosome 21 onto chromosomes 14 and 22.1,7,8 Mosaic trisomy 21 is a mixture of normal cells (46 chromosomes) as well as cells with 47 chromosomes, typically an extra chromosome 21.7,8 Individuals with Mosaic trisomy 21 typically have fewer medical issues and less intellectual disability.7,9

Duplication of multiple critical regions (eg, 21q21.22 and 21q22.13) cause the phenotypical features of DS, including...
hypotonia, short fingers, larger spaces between the first and second toe, and craniofacial dysmorphisms such as upward-slanting palpebral fissures, flat nasal bridge, flat occiput, epicanthic folds, round face, and open mouth (Table 1).2,7,10,11 These genetic aberrations are also directly linked to impaired physical development, intellectual disability, and organ system anomalies such as congenital heart defects and enlargement of adenoids and tonsils.2,7,11

The evidence-based literature and guidelines largely focus on the health challenges encountered by children with DS and provides significant guidance for pediatric care.2,13-14 The health risks and care needs for the adult with DS are less well understood and will be the focus of this discussion. Specific recent recommendations for care will be provided for the practitioner to optimize health.

**Systems-Based Approach to Care**

Although special screenings for adults with Down syndrome are highlighted in this article, it is necessary to perform all preventive care measures within the recommended age as with the general population.15 These screenings include (but are not limited to) mammograms, Pap smears, human immunodeficiency virus, and hepatitis C.15 Although mental illness in this population is of low prevalence and studies are limited, the well-being and mental health of adults with DS should be monitored annually (Table 2).2,13,15,17-19 Common medical conditions experienced by adults with Down syndrome are best reviewed using a systems-based approach.

**Immunology**

Some pathophysiologic findings can contribute to multiple disease states; among these is immune dysregulation. Individuals with Down syndrome have abnormal innate, humoral, and adaptive immunity; functionality of the immune system can be increased, depressed, or aberrant in response.18 For example, monocyte dysregulation is responsible for reduced phagocytosis and increased inflammatory response, T-helper cells are suppressed, as is immunoglobulin (Ig)G, IgM, and IgA.18 These abnormalities put the individual at increased risk for respiratory infections and dictate the need for additional boosters for vaccines to ensure effectiveness.18 Immune dysregulation also contributes to the higher incidence of autoimmune disorders, specifically immune-mediated thyroid dysfunction and celiac disease.19,20 Undertreated and chronic inflammation may be implicated in the earlier onset of dementia related to an increased neuroinflammatory response.19,21 However, overproduction of certain T cells may explain the low incidence of solid malignancies in people with DS20,22 except for a small number of cancers such as testicular in young adult males.22,23

**Cardiology**

Approximately half of the people with DS have congenital heart defects often diagnosed in early childhood.2,12,21 Surgical interventions reduce mortality but do not guarantee lifelong results. For example, atrioventricular septal defect repairs may require surgical revision to avoid obstruction of blood flow in the left ventricle or the development of Eisenmenger syndrome.24 Among those born without cardiac disease, there is a higher risk of developing valvular disease in adulthood.2 Due to the higher prevalence of congenital heart disease and acquired cardiac valve disease, a minimum of 1 echocardiogram is recommended in the individual’s lifetime; additionally, an echocardiogram should be included in the workup for those presenting with a new murmur or signs of heart failure.2,21,25,26 Although the physiologic explanation is unclear, adults with Down syndrome have a lower risk of coronary artery disease and myocardial infarction.27 Moreover, with conflicting evidence regarding the prevalence of stroke in adults with DS, the most recent recommendations suggest management of risk factors such as congenital heart disease and elevated cholesterol levels, which are similar to guidelines for the general population.2,12

**Respiratory**

Respiratory conditions remain a leading cause of hospitalization, morbidity, and mortality in individuals with DS28,29 due to a compromised immune system and anatomical airway differences.2,13,25 Pneumonia and influenza in individuals with Down syndrome account for 11.8% of US deaths compared with 2.07% in the general population.18 Because annual rates of influenza immunizations were 11%–16% in individuals with DS compared with 41% in adults in the general population,30 a clear recommendation is to administer influenza, pneumonia, and pertussis vaccines as recommended by the Centers for Disease Control and Prevention.2,13,26 Asthma medications are given to control respiratory difficulties, but this airway disease is uncommon in this population.2

Adults with Down syndrome have an increased vulnerability to COVID-19. Individuals with DS who are ill enough to require hospitalization are younger (aged 32–59), compared with those in the general population (aged 55–78), have more severe symptoms, and experience additional complications.28,29 Overall, the risks and treatments are similar, but due to increased congenital heart disease in this population, there is higher risk of severe complications and death.28 Early detection of common comorbidities and symptoms (eg, altered level of consciousness and confusion), which are common in COVID-19 cases in this population, can help prevent mortality or a more severe course of the disease.2,28,29

Obstructive sleep apnea (OSA) has a prevalence of up to 90% in adults with DS,2,13 OSA likely develops because of enlarged adenoids and tonsils, macroglossia, and general hypotonia, in addition to co-occurring conditions such as hypothyroidism and obesity, which may progress to pulmonary hypertension in more advanced stages of the disease.2,7,13,21 Because communication is a challenge for individuals with DS, it is good practice for the nurse practitioner (NP) to monitor for typical symptoms (eg, fatigue and daytime sleepiness) as well as atypical symptoms such as maladaptive behavior or decline in adaptive skills when suspecting OSA.2,13 Polysomnography and a comprehensive sleep study provide the best diagnostic results.2,26 A continuous positive airway pressure machine, interventions for weight loss, or surgical removal of enlarged structures of the airway, if applicable, are beneficial and reduce associated risks.2,21,26
Celiac disease is commonly found in individuals with DS, a diet free of gluten eliminates symptoms effectively. If left undiagnosed and untreated, adults will likely experience severe gastrointestinal problems. Observed symptoms include abdominal pain, diarrhea, weight loss, and maladaptive behaviors. The NP should order celiac screening labs and consider a referral to a gastroenterologist to confirm the diagnosis, which often includes an intestinal biopsy.

A yearly targeted history and physical assessment investigating for signs and symptoms of developing celiac disease should suffice instead of regular testing thus reducing the need for an invasive procedure. Involving family members or caregivers during the assessment process may be necessary because adults with DS may have difficulty expressing themselves.

Dysphagia is reported in more than half of the adults over 50 years of age with DS. Esophageal motility disorders (such as achalasia), hypertonia, obesity, and dental abnormalities are the main contributors to dysphagia and gastrointestinal reflux. Because of an increased risk of aspiration, education for caregivers should address meal supervision, dietary restrictions and modifications, as appropriate (eg, thickened liquids), and advising adults to eat slowly and chew their food thoroughly.

### Endocrinology

Hypothyroidism is present in 50% of adults with DS older than 30 years of age. In addition to advancing age, those at risk are adults with a family history of thyroid disease or congenital hypothyroidism. Misdiagnoses of weight gain, constipation, and depression are common, but symptoms such as cold intolerance, dry skin, and reduced activity should prompt the NP to perform a thyroid function assessment. Tsou et al recommend that adults with DS starting at 21 years of age should undergo a serum thyrotropin test every 1–2 years instead of thyroid stimulating hormone and T4 testing in symptomatic adults in the general population. This approach will avoid an incorrect diagnosis and promote early treatment with hormone replacement.

Adults with DS have difficulty maintaining a healthy weight, with 25%–55% clinically overweight and 33% classified as obese. Weight issues are associated with reduced exercise, decreased resting metabolic rate, satiety problems due to leptin resistance, and associated conditions (eg, hypothyroidism and OSA). In the general population, a body mass index (BMI) of greater than 30 and an increased waist circumference confirms a diagnosis of obesity requiring a weight management referral. Some sources suggest that BMI measurements have low sensitivity for obesity in individuals with DS because of their short stature and limb length and a generally higher percentage of adipose tissue, but others still recommend annual BMI screenings and body adiposity index and dual-energy X-ray absorptiometry (DEXA) as alternative methods. For this population, multiple interventions such as swimming, dancing, and portion control are beneficial for weight control.

There is a slight increase in the prevalence of diabetes mellitus associated with obesity and leptin resistance. Although type 1 diabetes is more common than type 2 in individuals with DS, evidence-based guidelines only exist for the latter, even when the prevalence rate is in a low range of 4%–8%. Because diabetes is prevalent in individuals with DS in their younger years, screening guidelines of glycated hemoglobin or fasting plasma glucose levels should be measured at age 30 years instead of 45 in asymptomatic adults. Those with known risk factors for diabetes should undergo screening more frequently—every 2–3 years, starting at 21 years of age.

### Musculoskeletal

Osteoporosis is found in 43% of adults 50 years of age or greater; this is significantly earlier and in a greater percentage than in the general population. Instead of excessive bone resorption, osteoporosis in DS results from decreased bone formation and low turnover that may cause critical fractures and other issues. Osteoporosis and serious complications can be prevented with regular exercise, adequate sunlight exposure, weightbearing exercise, and daily calcium and vitamin D supplements. Because of the risks of developing osteoporosis, DEXA screening should start at 40 years of age. The NP should evaluate for alternative causes
such as vitamin D deficiency, celiac disease, and thyroid problems.\textsuperscript{2,12}

**Sensory**

Most adults with DS experience visual\textsuperscript{2,7,13,21,26} and hearing impairments.\textsuperscript{2,7,13,21,22,26} Childhood problems such as refractive errors, strabismus, and cataracts may cause issues in adulthood.\textsuperscript{7} Therefore, an ophthalmologic exam every 1–3 years is recommended.\textsuperscript{2,7} Hearing impairments are associated with bone malformations in the ear, a history of childhood ear infections, and cholesteatoma.\textsuperscript{22} These justify an audiology screening every 1–2 years.\textsuperscript{2,7} Treatment with hearing aids and cochlear implants is associated with moderate improvement in hearing in the adult with DS.\textsuperscript{2,21,26}

**Oncology**

The risk of developing most cancers is less in individuals with DS; however, the current screening recommendations (eg, mammography) are no different from those for the general population.\textsuperscript{2,7,12,21,26} Three cancers are more likely to occur in those with DS: testicular cancer in young adults, liver cancer, and stomach cancer.\textsuperscript{22,23} The patient should be trained to do testicular self-exams, if possible, and screening for testicular cancer should be included in annual physical exams. Given a comprehensive history and an increased risk of gastrointestinal cancers,\textsuperscript{22,23} a referral to a gastroenterologist should be considered in addition to routine exams, such as fecal occult blood tests and colonoscopies.\textsuperscript{2,7,12,26}

Lastly, the NP should also raise suspicion of leukemia for adult patients who are fatigued and bruise easily even if this condition is more often found in childhood.\textsuperscript{2,26}

**Neurology and Mental Health**

The presence of the amyloid precursor protein gene on chromosome 21 increases the risk of Alzheimer’s disease in people with DS.\textsuperscript{31} Given that adults with DS have 3 copies of chromosome 21, studies show that half of the adults with DS will develop dementia; the prevalence is 40% in those over 45 years and 49% in those who are 65 years and older.\textsuperscript{31} Although the development of Alzheimer’s disease is linked to the accumulation of amyloid protein in the brain,\textsuperscript{2,7} recommendations for the NP involve conducting a careful assessment of medical, cognitive, and psychiatric factors to avoid misdiagnosis of dementia.\textsuperscript{2,12} Similar signs and symptoms found in dementia such as memory loss, a decline in adaptive skills, and mood changes are also present in hypothyroidism, OSA, sensory changes, mental health issues such as anxiety and depression, and early onset of menopause in women with DS.\textsuperscript{2,12,26} A clinical workup and routine vision and hearing screens will assist in making the diagnosis. To make a clinical diagnosis of anxiety and depression, an interview with adults with DS and their caregivers should occur to identify any recent life changes like separation or losing a loved one, then an application of guidelines under the *Diagnostic and Statistical Manual of Mental Disorders* will provide accuracy of the evaluation.\textsuperscript{2,7,12,21,26}

Of note, the prevalence of dementia in DS increases after age 40 but remains relatively low in younger persons;\textsuperscript{21} therefore, NPs should avoid diagnosing dementia in adults aged 39 and below without a comprehensive workup.\textsuperscript{12}

Mental illness is uncommon in adults with DS.\textsuperscript{32} Behavioral problems and depression are less prevalent in this population compared with other groups affected with intellectual disability.\textsuperscript{32} Communication difficulties are often misinterpreted for misbehavior in adults with DS.\textsuperscript{2,7,12,21,22,26} Although commonly diagnosed in childhood, a co-occurring diagnosis of autism with DS results in higher severity of social and communication impairments in adulthood.\textsuperscript{7,25,33} Management and treatment for adults with DS with autism require combined efforts from specialists in medicine and behavioral health, as well as caregivers who will maintain a stable and stress-free home environment.\textsuperscript{2} Additionally, a high level of coordinated care and support is necessary for all adults with DS with significant co-occurring conditions such as severe mental illness and epilepsy due to increased mortality rates.\textsuperscript{34}

**Implications for NPs**

**Social considerations**

The majority of individuals with DS are born to parents of advanced age who typically become primary caregivers for their children. However, as parents start to have more medical issues due to aging, adults with DS also experience increasing concerns, such as loss of parents and disruption in care. In addition, some parents believe that there is a lack of opportunities for their children to make social connections and learn to live independently.\textsuperscript{2} In this case, a primary care NP can initiate advanced or transition planning in early adulthood. Advanced planning for adults with DS upholds their right to consent for preferred health care services and increases their quality of life.

Furthermore, transition planning covers important decisions regarding medical care with an adult provider, living situations, and employment.\textsuperscript{36–38} Studies show that those who succeed in delaying cognitive decline, maintaining adaptive skills, and decreasing depression in adulthood more often live with their parents or siblings.\textsuperscript{2,7,39} Some adults live independently and keep regular jobs,\textsuperscript{36,37} but the less ideal situations are group homes or institutional residences.\textsuperscript{2,16,17,25} Although living with family members is most beneficial, the NP should provide information about local resources, social groups, and respite care for elderly parents or siblings because caregiver burnout is highly likely.\textsuperscript{36,37}

Families in this population have experienced past discrimination and health inequities. Prenatal screenings have decreased the birth of infants with DS globally.\textsuperscript{41} Furthermore, individuals with DS along with their families have met insensitive and unsympathetic care from providers. When receiving the diagnosis of DS, mothers often hear comments from health care providers about future hardships and the negative aspects instead of up-to-date information regarding care and positive sentiments.\textsuperscript{42} Therefore, NPs should avoid these mistakes and become more sensitive in their approach to care to build trust with individuals with DS and their families.

As mentioned throughout this article, communication challenges are often mistaken for behavior problems and depression in adults. In addition, consent to care often becomes an ethical issue because parents or conservators make decisions for the adult with DS.\textsuperscript{33} Because communication abilities are at their peak in young adulthood,\textsuperscript{30,34} individuals with DS can provide a baseline for their health preferences at the time of transition planning. Moving forward, whether inquiring about health issues or making important decisions toward treatment and management, the adult with DS needs to be the primary informant.

**Safety**

As with typical adults, individuals with DS experience sexual desires.\textsuperscript{43,44} However, due to communication difficulties, social challenges, and trusting nature, sexual assault is a concern in persons with DS.\textsuperscript{43,45} Although the data on sexual assault in DS are limited, studies exist in those with intellectual disability with an
estimated prevalence rate of 32%-39% in females and 40% in males. 

Sexual health education should include family members and patients for awareness and prevention of sexual assault, pregnancies, and sexually transmitted infections. Some parents believe that sterilization or neglect of their children’s sexual desires will fix the problem, but research demonstrates that sex education promotes awareness and helps adults with DS make appropriate decisions.

**Recommendations to Improve Care for This Population**

Due to the complexity of providing health care for the adult with DS, the optimal care delivery setting may be in centers specializing in care for this population. However, these centers may be geographically restricted or operate for limited hours. As such, it is essential that comprehensive, evidence-based guidelines be developed and become available for the practitioner providing primary care, where most of these patients will be seen. To achieve these aims, ongoing research is needed to identify modalities, specific treatments, and screening parameters as well as effective strategies to educate all primary care providers about the care of adults with DS.

**Conclusion**

A holistic approach is needed to provide a high quality of care to individuals with DS. To meet the patient and family needs, the NP should be aware of medical conditions with high prevalence, needed screenings, specialty education, and be skilled in communicating with intellectually disabled individuals and their families.

**References**


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