Down Syndrome

Health Watch Table

1. Head, eyes, ears, nose, throat

Considerations	Recommendations
Children and Adults: Vision: ▸ 1%-3% of children have cataracts; 16% of adults have cataracts ▸ 36%-80% have significant refractive errors ▸ Adults have an increased incidence of keratoconus Hearing: ▸ 50%-80% have a hearing deficit ▸ 50%-75% experience otitis media with effusion	▸ Neonatally: Evaluate for a red reflex and refer immediately to an ophthalmologist if the red reflex is absent. ▸ Arrange ophthalmological assessment: first by 6 months for all; then every 1-2 years, with special attention to strabismus, cataracts, nasolacrimal duct obstruction, refractive errors, glaucoma, and nystagmus. ▸ During childhood: screen vision annually with history and exam, including photo screening if available; refer as needed. ▸ Arrange auditory brainstem response (ABR) measurement by 3 months if newborn screening has not been done or if results were suspicious; refer to otolaryngologist as needed. ▸ During childhood: screen hearing annually with history and exam; review risks for frequently occurring serious otitis media. ▸ Undertake brainstem auditory evoked response testing (BAER, behavioral, ear-specific): at 6 months and every 6 months until established baseline. Subsequently, screen annually until adulthood, then every two years. ▸ Treat otitis media promptly and aggressively. ▸ Refer to an ENT surgeon if recurring otitis media.

2. Dental

Considerations	Recommendations
▸ *Children and Adults*: tooth anomalies are common ▸ Increased risk of periodontal disease in adults	▸ Reassure parents that delayed dental eruption and irregular patterns are normal for children with DS. ▸ Undertake initial dental exam at 2 years, then every 6 months thereafter. ▸ Encourage proper dental hygiene. Refer to an orthodontist if needed.

3. Cardiovascular

Considerations	Recommendations
▸ *Children*: 40%-50% have congenital heart defects (CHD)	▸ Newborn: Obtain an echocardiogram and refer to a cardiologist, *even in the absence of physical findings* ▸ If child requires surgery, reassure parents with the success rate for heart defect repairs. ▸ In children and adolescents: review cardiovascular history and assess for physical signs with specialist referral if indicated. ▸ Refer for an echocardiogram if not previously done. ▸ Undertake subacute bacterial endocarditis prophylaxis as indicated by findings.
▸ *Adults*: 50% have cardiovascular concerns, commonly acquired mitral valve prolapse (MVP) and valvular regurgitation	▸ Ascertain a comprehensive cardiovascular history. ▸ Undertake an annual cardiac exam, with echocardiogram to confirm new abnormal findings and to assess for cardioembolic stroke risk and develop a monitoring plan depending on the type of cardiovascular problem present or refer to an adult congenital heart specialist or disease clinic. ▸ Monitor regularly those who have had surgery in childhood. ▸ Assess appropriateness of statin therapy every 5 years starting at age 40. ▸ Risk factors for stroke should be managed as specified by the American Heart Association/American Stroke Association's Guidelines for the Primary Prevention of Stroke.

4. Sleep

Considerations	Recommendations
▸ *Children and Adults*: 50%-80% have obstructive sleep apnea (OSA)	▸ An overnight polysomnography is recommended between the ages of 3 and 5, regardless of symptoms. ▸ Ascertain a detailed sleep history, with attention to OSA symptoms, such as snoring, noisy breathing, sweating in sleep, restless sleep, and morning dry mouth, nasal congestion or headaches. Refer to a sleep specialist for evaluation, including a sleep study, if OSA is suspected. ▸ Other sleep disorders, such as insomnia, parasomnias (sleepwalking, sleep terrors or confusional arousals) or daytime sleepiness can be ascertained by sleep history. ▸ Adenotonsillectomy is less effective in this population due to craniofacial abnormalities, hypotonia and other factors, so continuous positive airway pressure and ENT procedures (craniofacial surgery) should be considered. ▸ If showing signs of Alzheimer-type dementia, especially under age 40, evaluate for OSA.

5. Respiratory

Considerations	Recommendations
Children and Adults: Respiratory infections are more common and often more severe; also, choking with feeding is more common	For recurrent pneumonia or if aspiration pneumonia is suspected, investigate for possible swallowing disorder and gastro-esophageal reflux disease; for adults, also inquire about smoking. Stridor and croup are more common due to hypotonia and small trachea size; emphasize importance of influenza vaccine and respiratory syncytial virus vaccine for prevention.

6. Gastrointestinal

Considerations	Recommendations
Children: 31%-80% have feeding difficulties including gastroesophageal reflux and dysphagia *Children*: Up to 15% have gastrointestinal (GI) tract anomalies including duodenal atresia, celiac disease, Hirschsprung disease, and imperforate anus	Newborn: with vomiting or absent stools, check for GI tract blockage and refer to a gastroenterologist. Newborn: If difficulty feeding, conduct feeding assessment, video feeding study, or non-radiologic videofluoroscopic swallow studies; refer for occupational therapy. Reassure parents that babies with Down syndrome can successfully breastfeed, even through heart surgery. Infants and children: anticipate constipation; treat with fluid/fiber/laxative/stool softener/exercise/ dietary change. From 2-3 years of age, screen for celiac disease. Establish good dietary and exercise habits to prevent or manage obesity.
Adults: Obesity is common Up to 10% have celiac disease	Monitor for obesity and provide adults and families/ support persons with resources to encourage good nutrition and exercise. Assess adults with DS annually for gastrointestinal and non-gastrointestinal signs and symptoms of celiac disease using targeted history, physical examination, and clinical judgment of good practice. Test for *Helicobacter Pylori* and treat if positive, regardless of symptoms. Manage constipation proactively.

7. Genitourinary

Considerations	Recommendations
Children: Increased risk for cryptorchidism (undescended testicles)	Assess for hypogonadism, undescended testes, and possible testicular germ-cell tumors, or refer to a urologist, as appropriate.
Adults: Have increased risk of testicular cancer	Assess annually by clinical exam and refer to a urologist as appropriate.

8. Sexual function

Considerations	Recommendations
▸ *Adults*: Fertility has been documented in women ▸ Fertility in males rarely reported	▸ Counsel regarding fertility possibility and the 50% risk of Down syndrome in offspring. ▸ Suggest birth control when girls begin having periods to avoid unwanted pregnancy ▸ Starting in adolescence, regularly discuss healthy, typical sexual development and behaviors; sexuality; privacy and boundaries; contraception; and prevention of sexually transmitted diseases; and sex abuse prevention.

9. Musculoskeletal (MSK)

Considerations	Recommendations
▸ *Children*: 1%-2% have atlanto-axial instability (AAI)	▸ Current evidence does not support performing routine screening radiographs for potential atlantoaxial instability in asymptomatic children. ▸ A child with significant neck pain, radicular pain, weakness, spasticity or change in tone, gait difficulties, hyperreflexia, change in bowel or bladder function, or other symptoms of myelopathy should undergo cervical spine radiography in the neutral position. ▸ Screen, as needed, prior to high-risk activities (e.g., tumbling) and if participating in Special Olympics. ▸ Undertake an annual neurological exam for signs or symptoms of spinal cord compression. If present, refer urgently to a neurosurgeon and arrange an urgent MRI. ▸ Obtain a detailed MSK history with particular attention to possible joint subluxations/dislocations, scoliosis, and hip abnormalities.
▸ *Adults*: Continued risk for spinal cord compression secondary to AAI ▸ Though data are limited, osteoporosis (associated with increased fractures risk) may be more common in older adults with Down syndrome than in similar aged individuals in the general population or with other developmental disabilities	▸ Routine cervical spine radiographs are not necessary in asymptomatic individuals. Instead, annual screening should include a review of signs and symptoms of cervical myelopathy, such as altered gait, new incontinence, brisk reflexes, or clonus, using history and physical examination. ▸ For primary prevention of osteoporotic fractures, there is insufficient evidence to recommend for or against applying established osteoporosis screening guidelines; thus, good clinical practice would support a shared decision-making approach, to the extent possible. ▸ Adults who sustain a fragility fracture should be evaluated for secondary causes of osteoporosis, including screening for hyperthyroidism, celiac disease, vitamin D deficiency, hyperparathyroidism, and medications associated with adverse effects on bone health.

9. Musculoskeletal (MSK) continued

Considerations	Recommendations
	▸ Encourage ambulation/mobility and weight reduction if obesity is present to decrease the risk of osteoarthritis.

10. Neurological

Considerations	Recommendations
▸ *Infants*: Infantile spasms 1%-13% ▸ *Children*: Neurologic dysfunction: 1%-13% ▸ *Children and adults*: Seizures estimated to be between 8.1%–26%	▸ Take careful neurological history with particular attention to seizures (infantile spasms or tonic-clonic-type). ▸ Monitor for iron deficiency and anemia. ▸ Refer to a neurologist if history indicates possible seizures.
▸ *Adults*: Dementia is frequent and occurs earlier ▸ 9%: 40–49 years ▸ 32%: 50-59 years ▸ 26%: 60–69 years ▸ Up to 50% with dementia have seizures with frequency increasing with age	▸ Obtain a neuropsychiatric history at every visit with particular attention to change in behavior, loss of function/activities of daily living, and new onset seizures. ▸ Caution is urged when diagnosing age-related, Alzheimer-type dementia in those younger than 40 years because of its low prevalence before this age. ▸ Assess the patient and interview primary support persons about changes from baseline function annually, beginning at age 40. Decline in six domains specified by the National Task Group–Early Detection Screen for Dementia (NTG-EDSD) should be used to identify early-stage age-related Alzheimer-type dementia, or a potentially reversible medical condition, or both. ▸ If functional decline and/or signs/symptoms of dementia, use history, exam, and blood work to check for other conditions and treatable causes (e.g., hearing/vision deficits, obstructive sleep apnea, hypothyroidism, chronic pain, medication side effects, depression, menopause, low folic acid/vitamin B12). ▸ Refer to a neurologist if history indicates possible seizures.

11. Dermatological

Considerations	Recommendations
▸ *Children and Adults*: Dry skin, atopic dermatitis, seborrheic dermatitis, chelitis, impetigo, cutis marmorata, vitiligo, folliculitis, keratosis pilaris, and alopecia areata are more common than in general population ▸ *Older age:* inflammatory disorders such as hidradenitis suppurativa	▸ Examine skin as part of routine care. ▸ Treat as per general population, with referral to dermatologist as needed.

12. Behavioral/mental health

Considerations	Recommendations
▸ *Children*: Self-talk is very common; autism spectrum disorder occurs in 7%-19% of children with DS	▸ Refer all children to the state's Early Intervention system for services such as speech therapy. ▸ Encourage families to teach self-help skills and counsel to prevent wandering. ▸ Assess behavior and talk about behavioral management, sibling adjustments, socialization, and recreational skills. ▸ Monitor for behavior problems that interfere with function in the home, com- munity, or school. Attention problems, hyperactivity, obsessive-compulsive behaviors, and wandering off/elopement are some common behavior concerns reported ▸ Make referral for autism screening as soon as autism diagnosis is suspected. ▸ Review for signs suggestive of psychosis.
▸ *Adults*: 30% have a psychiatric disorder, including depression	▸ Review regularly with respect to behavioral concerns. ▸ Ascertain neuropsychiatric history at every visit, with particular attention to changes in behavior, loss of function/activities of daily living, and new onset seizures. ▸ When concern for a mental health disorder is present, refer, if possible, to a clinician knowledgeable about the medical, mental health disorders, and common behavioral characteristics of adults with Down syndrome. ▸ When a mental health disorder is suspected, clinicians should follow guidelines for diagnosis in the *Diagnostic and Statistical Manual of Mental Disorders (Fifth Edition) (DSM-5)*. The *Diagnostic Manual–Intellectual Disability 2: A Textbook of Diagnosis of Mental Disorders in Persons with Intellectual Disability (DM-ID-2)* also may be used to adapt diagnostic criteria from the DSM-5.

13. Endocrine

Considerations	Recommendations
▸ *Children*: 2%-7% have congenital hypothyroidism ▸ 20% develop hypothyroidism after birth	▸ Review neonatal screening ▸ Ascertain TSH and free T4 tests to confirm euthyroid status at 6 and 12 months, then annually. Refer to pediatric endocrinologist as needed.

▸ *Adults*: 50% have hypothyroidism ▸ Subclinical hypothyroidism, hyperthyroidism, and autoimmune thyroiditis are more common than in the general population	▸ Screen for hypothyroidism every 1 to 2 years using a serum thyrotropin test, beginning at age 21. ▸ For asymptomatic adults, screen for type 2 diabetes using hemoglobin A1C or fasting plasma glucose levels every 3 years, starting at age 30. ▸ For any adult with comorbid obesity, screen for type 2 diabetes every 2 to 3 years, beginning at age 21 years. ▸ Consider checking thyroid function whenever there are changes in mental status, behavior or functional abilities.

14. Hematological

Considerations	Recommendations
▸ *Children and Adults*: Increased frequency of transient abnormal myelopoiesis (TAM) ▸ *Children*: Increased frequency of leukemia 1% (increased risk of acute myeloid leukemia in the first 4 years of life) ▸ No increased risk of leukemia in adults ▸ Anemia/Iron Deficiency found in about 9% of children with DS	▸ *Newborn*: Obtain a complete blood cell count with differential by 3 days of age to evaluate for transient abnormal myelopoiesis (TAM) (formerly called transient myeloproliferative disorder), polycythemia, and other hematologic abnormalities. ▸ Assess history periodically for symptoms of leukemia, with close attention to those with a history of transient abnormal myelopoiesis (TAM). ▸ Families/caregivers of infants with TAM should be aware of the risk of leukemia and told about the signs, including easy bruising or bleeding, recurring fevers, bone pain, petechiae, onset of lethargy, or change in feeding patterns. ▸ Obtain a complete blood cell count (CBC) with differential and either (1) a combination of ferritin and C- reactive protein (CRP), or (2) a combination of serum iron and total iron-binding capacity (TIBC), at 1 year and annually thereafter.

Developed by Forster-Gibson C, Berg J, & Developmental Disabilities Primary Care Initiative Co-editors.

Modified with permission of Surrey Place. This tool was reviewed and adapted for U.S. use by the IDD Toolkit's Advisory Committee; for list, view here.

Resources

Down syndrome websites that may be helpful for families and caregivers

Canadian Down Syndrome Society	www.cdss.ca	Accessed February 2024
Down Syndrome Education International [DownsEd]	www.downsed.org	Accessed February 2024
Down Syndrome: Health Issues by Dr. Len Leshin	www.ds-health.com	Accessed February 2024
Down Syndrome Medical Interest Group [DSMIG-UK]	www.dsmig.org.uk	Accessed February 2024
National Down Syndrome Society [USA]	www.ndss.org	Accessed February 2024
National Down Syndrome Congress [USA]	ndsccenter.org	Accessed February 2024
Global Down Syndrome Foundation [USA]	www.globaldownsyndrome.org	Accessed February 2024

Resources for Families and Clinicians

- Lettercase National Center for Prenatal and Postantal Resources: Understanding Down Syndrome (10 languages)
- Down Syndrome Pregnancy
- Breastfeeding and Down Syndrome
- Health Supervision for Children and Adolescents With Down Syndrome
- Children with Down Syndrome: Health Care Information for Families (English and Spanish)
- GLOBAL Medical Care Guidelines for Adults with Down Syndrome
- Primary Care Provider's Guide to Women's Health and Down Syndrome

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