Health Watch Table — Down Syndrome

Forster-Gibson and Berg 2011

CONSIDERATIONS	RECOMMENDATIONS
1. HEENT (HEAD, EYES, EARS, NOSE, THROAT)	
Children and Adults: Vision: ~15% have cataracts; ~ 20%-70% have significant refractive errors 5%-15% of adults have keratoconus Hearing: 50%-80% have a hearing deficit	☐ Neonatally: refer immediately to an ophthalmologist if the red reflex is absent or if strabismus, nystagmus or poor vision is identified ☐ Arrange ophthalmological assessment: first by 6 months for all; then every 1-2 years, with special attention to cataracts, keratoconus, and refractive errors ☐ During childhood: screen vision annually with history and exam; refer as needed ☐ Arrange auditory brainstem response (ABR) measurement by 3 months if newborn screening has not been done or if results were suspicious ☐ During childhood: screen hearing annually with history and exam; review risks for frequently occurring serious otitis media ☐ Undertake auditory testing: first at 9 – 12 months, then every 6 months up to 3 years, annually until adulthood, then every two years
2. DENTAL	
Children and Adults: tooth anomalies are common Increased risk of periodontal disease in adults	☐ Undertake initial dental exam at 2 years, then every 6 months thereafter. Encourage proper dental hygiene. Refer to an orthodontist if needed ☐ Undertake clinical exams every six months with referral, as appropriate
3. CARDIOVASCULAR	
Children: 30%-60% have congenital heart defects (CHD)	☐ Newborn screening: Obtain an echocardiogram and refer to a cardiologist, even in the absence of physical findings ☐ 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 SBE 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 follow-up depending on the type of cardiovascular problem present or refer to an Adult Congenital Heart specialist or Disease clinic ☐ Monitor regularly those that have had surgery in childhood ☐ An echocardiogram is indicated to assess new abnormal physical findings or if unable to assess adequately by physical exam. Consider echocardiogram to establish baseline cardiac anatomy and function if not previously done or records are unavailable [1]
4. RESPIRATORY	
Children and Adults: 50%-80% have obstructive sleep apnea (OSA)	☐ Newborn: Refer to an ENT surgeon if recurring otitis media infections ☐ Treat infections promptly and aggressively
Adults: 50%-80% have obstructive sleep apnea (OSA)	☐ Ascertain a detailed sleep history, with special attention to OSA symptoms. Refer to an ENT surgeon, including sleep study, if OSA is suspected ☐ If aspiration pneumonia is suspected, investigate for possible swallowing disorder and gastro-esophageal reflux disease

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5. GASTROINTESTINAL	
Children: ~ 50% 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 ☐ Infants and children: anticipate constipation; treat with fluid/fibre/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: ~ 95% are obese; ~ 7% have celiac disease	☐ Monitor for obesity ☐ Screen for celiac disease, which may present in adulthood; screening tests used are the same as in the general population [2] ☐ Test for Helicobacter Pylori and treat if positive, regardless of symptoms ☐ Manage constipation proactively
6. GENITOURINARY	
Children: Cryptorchidism is common	☐ 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 [3]
7. SEXUAL FUNCTION	
Adults: Fertility has been documented in women Fertility in males rarely reported	☐ Counsel regarding fertility possibility and the 50% [4] risk of Down syndrome in offspring
8. MUSCULOSKELETAL (MSK)	
Children: ~15% have atlanto-axial instability (AAI)	☐ Arrange lateral cervical spine X-rays (flexed, neutral, and extended positions) between 3-5 years of age ☐ 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	☐ Undertake an annual neurological exam and assess for evidence of spinal cord compression ☐ Arrange lateral cervical spine X-rays if not previously done, if presenting with signs and symptoms of AAI or if participating in Special Olympics ☐ Take detailed history and attend to joint complaints, scoliosis, and hip abnormalities ☐ If suspected, undertake bone mineral density (BMD) screening and refer to an appropriate specialist if indicated ☐ Encourage ambulation/mobility and weight reduction if obesity is present to decrease the risk of osteoarthritis

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9. NEUROLOGICAL	
Children: Epilepsy in up to 22%	☐ Take careful neurological history with particular attention to seizures (infantile spasms or tonic-clonic-type) ☐ Arrange an EEG and refer to a neurologist
Adults: Dementia is frequent and occurs earlier: 11%: 40 – 49 y, 77%: 60 – 69 y, Up to 75% with dementia have seizures with frequency increasing with age	☐ Obtain a neuropsychiatric history at every visit with particular attention to change in behaviour, loss of function/activities of daily living, and new onset seizures ☐ 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) ☐ For possible seizures, arrange an EEG and refer to a neurologist
10. DERMATOLOGICAL	
Children and Adults: Dry skin, atopic dermatitis, seborrheic dermatitis, chelitis, impetigo, and alopecia areata are more common than in general population	☐ Examine skin as part of routine care ☐ Treat as per general population, with referral to dermatologist as needed
11. BEHAVIOURAL/MENTAL HEALTH	
Children: Self-talk is very common; autism spectrum disorder occurs in 5%-10% of children with DS	☐ Review regularly with respect to behavioural concerns ☐ Review for positive or negative signs suggestive of psychosis
Adults: ~ 30% have a psychiatric disorder, including depression	☐ Review regularly with respect to behavioural concerns ☐ Ascertain neuropsychiatric history at every visit, with particular attention to changes in behaviour, loss of function/activities of daily living, and new onset seizures
12. ENDOCRINE	
Children: ~ 1% 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 ☐ If signs of hyperthyroidism in adolescence, check for autoimmune thyroiditis

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Adults: 15%-50% are hypothyroid Subclinical hypothyroidism, hyperthyroidism, and autoimmune thyroiditis are more common than in the general population	☐ For adults who are euthyroid, check TSH and free T4 levels at least once every 5 years [5] (some recommend annually) [6] ☐ If subclinical hypothyroidism (i.e., elevated TSH with normal free T4), follow free T4 every 6 months 4 to one year [7] (some recommend treatment if thyroid antibodies are positive) ☐ Consider checking thyroid function whenever there are changes in mental status, behaviour or functional abilities
13. HEMATOLOGICAL	
Children and Adults: Increased frequency of transient myeloproliferative disorder and leukemia No increased risk of leukemia in adults	☐ Neonates to 1 month olds: investigate for polycythemia and thromobocytopenia ☐ Assess history periodically for symptoms of leukemia, with close attention to those with a history of transient myeloproliferative disorder

Resources

22 published Down syndrome health care guidelines were reviewed and compared (For full list of references, see **www.surreyplace.on.ca/Clinical-Programs/Medical-Services/Pages/PrimaryCare.aspx**)

Down syndrome websites that may be helpful for families and caregivers
Canadian Down Syndrome Society **www.cdss.ca/**
Down Syndrome Education International [DownsEd] **www.downsed.org/**
Down Syndrome: Health Issues by Dr. Len Leshin **www.ds-health.com/**
Down Syndrome Medical Interest Group [DSMIG-UK] **www.dsmig.org.uk/**
National Down Syndrome Society [USA] **www.ndss.org/**

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