

Health Watch Table - Down Syndrome

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CONSIDERATIONS	RECOMMENDATIONS
1. HEENT (HEAD, EYES, EARS, NOSE, THROAT)	
<p><i>Children & Adults:</i> <i>Vision:</i> ~15% have cataracts; ~ 20-70% have significant refractive errors; 5-15% of adults have keratoconus <i>Hearing:</i> 50 – 80% have a hearing deficit</p>	<ul style="list-style-type: none"> <input type="checkbox"/> Neonatally: refer immediately to an ophthalmologist if the red reflex is absent or if strabismus, nystagmus or poor vision is identified <input type="checkbox"/> Arrange ophthalmological assessment: first by 6 months for all; then every 1-2 years, with special attention to cataracts, keratoconus, and refractive errors <input type="checkbox"/> During childhood: screen vision annually with history and exam; refer as needed <input type="checkbox"/> Arrange auditory brainstem response (ABR) measurement by 3 months if newborn screening has not been done <i>or</i> if results were suspicious <input type="checkbox"/> During childhood: screen hearing annually with history and exam; review risks for frequently occurring serious otitis media <input type="checkbox"/> Undertake auditory testing: first at 9 – 12 months, then every six months up to 3 years, annually until adulthood, then every 2 years
2. DENTAL	
<p><i>Children & Adults:</i> tooth anomalies are common Increased risk of periodontal disease in adults</p>	<ul style="list-style-type: none"> <input type="checkbox"/> Undertake initial dental exam at 2 years, then every six months thereafter. Encourage proper dental hygiene. Refer to an orthodontist if needed <input type="checkbox"/> Undertake clinical exams every six months with referral, as appropriate
3. CARDIOVASCULAR	
<p><i>Children:</i> 30% - 60% have congenital heart defects (CHD)</p>	<ul style="list-style-type: none"> <input type="checkbox"/> Newborn screening: Obtain an echocardiogram and refer to a cardiologist, <u>even in the absence of physical findings</u> <input type="checkbox"/> In children and adolescents: review cardiovascular history and assess for physical signs with specialist referral if indicated <ul style="list-style-type: none"> <input type="checkbox"/> Refer for an echocardiogram if not previously done <input type="checkbox"/> Undertake SBE prophylaxis as indicated by findings
<p><i>Adults:</i> ~ 50% have cardiovascular concerns, commonly acquired mitral valve prolapse (MVP) and valvular regurgitation</p>	<ul style="list-style-type: none"> <input type="checkbox"/> Ascertain a comprehensive cardiovascular history <input type="checkbox"/> 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 <input type="checkbox"/> Monitor regularly those that have had surgery in childhood <input type="checkbox"/> 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

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4. RESPIRATORY	
<i>Children & Adults:</i> 50%-80% have obstructive sleep apnea (OSA)	<ul style="list-style-type: none"> <input type="checkbox"/> Newborn: Refer to an ENT surgeon those with recurring otitis media infections <input type="checkbox"/> Treat infections promptly and aggressively
<i>Adults:</i> 50%-80% have obstructive sleep apnea (OSA)	<ul style="list-style-type: none"> <input type="checkbox"/> Ascertain a detailed sleep history, with special attention to OSA symptoms. Refer to an ENT surgeon, including sleep study, if OSA is suspected <input type="checkbox"/> If aspiration pneumonia is suspected, investigate for possible swallowing disorder and gastro-esophageal reflux disease
5. GASTROINTESTINAL	
<i>Children:</i> ~ 50% have gastrointestinal (GI) tract anomalies including duodenal atresia, celiac disease, Hirschsprung disease, and imperforate anus	<ul style="list-style-type: none"> <input type="checkbox"/> Newborn: with vomiting or absent stools, check for GI tract blockage and refer to a gastroenterologist <input type="checkbox"/> Infants and children: anticipate constipation; treat with fluid/fiber/laxative/stool softener/exercise/dietary change <input type="checkbox"/> From 2-3 years of age, screen for celiac disease <input type="checkbox"/> Establish good dietary and exercise habits to prevent or manage obesity
<i>Adults:</i> ~ 95% are obese; ~ 7% have celiac disease	<ul style="list-style-type: none"> <input type="checkbox"/> Monitor for obesity <input type="checkbox"/> Screen for celiac disease, which may present in adulthood; screening tests used are the same as in the general population ² <input type="checkbox"/> Test for <i>Helicobacter Pylori</i> and treat if positive, regardless of symptoms <input type="checkbox"/> Manage constipation proactively
6. GENITOURINARY	
<i>Children:</i> Cryptorchidism is common	<ul style="list-style-type: none"> <input type="checkbox"/> Assess for hypogonadism, undescended testes, and possible testicular germ-cell tumors, or refer to a urologist, as appropriate
<i>Adults:</i> Have increased risk of testicular cancer	<ul style="list-style-type: none"> <input type="checkbox"/> Assess annually by clinical exam, and refer to a urologist as appropriate ³
7. SEXUAL FUNCTION	
<i>Adults:</i> Fertility has been documented in women Fertility in males rarely reported	<ul style="list-style-type: none"> <input type="checkbox"/> Counsel regarding fertility possibility and the 50%⁴ risk of Down syndrome in offspring

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8. MUSCULOSKELETAL (MSK)	
<p><i>Children:</i> ~15% have atlanto-axial instability (AAI)</p>	<ul style="list-style-type: none"> <input type="checkbox"/> Arrange lateral cervical spine x-rays (flexed, neutral, and extended positions) between 3-5 years of age <input type="checkbox"/> Screen, as needed, prior to high risk activities (e.g., tumbling) and if participating in Special Olympics <input type="checkbox"/> Undertake an annual neurological exam for signs or symptoms of spinal cord compression. If present, refer urgently to a MSK neurosurgeon and arrange an urgent MRI <input type="checkbox"/> Obtain a detailed MSK history with particular attention to possible joint subluxations/dislocations, scoliosis, and hip abnormalities
<p><i>Adults:</i> Continued risk for spinal cord compression secondary to AAI</p> <p>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</p>	<ul style="list-style-type: none"> <input type="checkbox"/> Undertake an annual neurological exam and assess for evidence of spinal cord compression <input type="checkbox"/> Arrange lateral cervical spine x-rays if not previously done, if presenting with signs and symptoms of AAI or if participating in Special Olympics <input type="checkbox"/> Take detailed history and attend to joint complaints, scoliosis, and hip abnormalities <input type="checkbox"/> If suspected, undertake bone mineral density (BMD) screening and refer to an appropriate specialist if indicated <input type="checkbox"/> Encourage ambulation/mobility and weight reduction if obesity is present to decrease the risk of osteoarthritis
9. NEUROLOGICAL	
<p><i>Children:</i> Epilepsy in up to 22%</p>	<ul style="list-style-type: none"> <input type="checkbox"/> Take careful neurological history with particular attention to seizures (infantile spasms or tonic-clonic-type) <input type="checkbox"/> Arrange an EEG and refer to a neurologist
<p><i>Adults:</i> Dementia is frequent and occurs earlier:</p> <p>11%: 40 – 49 y, 77%: 60 – 69 y,</p> <p>Up to 75% with dementia have seizures with frequency increasing with age</p>	<ul style="list-style-type: none"> <input type="checkbox"/> 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 <input type="checkbox"/> 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) <input type="checkbox"/> For possible seizures, arrange an EEG and refer to a neurologist
10. DERMATOLOGICAL	
<p><i>Children & Adults:</i></p> <p>Dry skin, atopic dermatitis, seborrheic dermatitis, chelitis, impetigo, and alopecia</p>	<ul style="list-style-type: none"> <input type="checkbox"/> Examine skin as part of routine care <input type="checkbox"/> Treat as per general population, with referral to dermatologist as needed

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11. BEHAVIOURAL/MENTAL HEALTH	
<i>Children:</i> Self-talk is very common; Autism spectrum disorder occurs in 5-10% of children with DS	<input type="checkbox"/> Review regularly with respect to behavioural concerns <input type="checkbox"/> Review for positive or negative signs suggestive of psychosis
<i>Adults:</i> ~30% have a psychiatric disorder, including depression	<input type="checkbox"/> Review regularly with respect to behavioural concerns <input type="checkbox"/> 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	
<i>Children:</i> ~1% have congenital hypothyroidism; ~20% develop hypothyroidism after birth	<input type="checkbox"/> Review neonatal screening. <input type="checkbox"/> Ascertain TSH and free T4 tests to confirm euthyroid status at six and twelve months, then annually <input type="checkbox"/> If signs of hyperthyroidism in adolescence, check for autoimmune thyroiditis
<i>Adults:</i> 15 – 50% are hypothyroid Subclinical hypothyroidism, hyperthyroidism, and autoimmune thyroiditis are more common than in the general population	<input type="checkbox"/> For adults who are euthyroid, check TSH and free T4 levels at least once every five years ⁵ (some recommend annually) ⁶ <input type="checkbox"/> If subclinical hypothyroidism (i.e., elevated TSH with normal free T4), follow free T4 every six months ⁴ to one year ⁷ (some recommend treatment if thyroid antibodies are positive) <input type="checkbox"/> Consider checking thyroid function whenever there are changes in mental status, behaviour or functional abilities
13. HEMATOLOGICAL	
<i>Children & Adults:</i> Increased frequency of transient myeloproliferative disorder and leukemia No increased risk of leukemia in adults	<input type="checkbox"/> Neonates to one month olds: investigate for polycythemia and thrombocytopenia <input type="checkbox"/> Assess history periodically for symptoms of leukemia, with close attention to those with a history of transient myeloproliferative disorder

References

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<http://www.nice.org.uk/nicemedia/pdf/CG86QuickRefGuide.pdf>.

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7. Wallace RA, Dalton AJ. Clinicians' guide to physical health problems of older adults with Down syndrome. *Journal on Developmental Disabilities*. 2006;12(1 (Supplement 1)):1-92. Available from: http://www.oadd.org/publications/journal/issues/vol12no1supp/DS_supplement_1.pdf.

22 Published Down syndrome health care guidelines reviewed and compared (For full list of references, see <http://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 <http://www.cdss.ca/>

Down Syndrome: Health Issues by Dr. Len Leshin <http://www.ds-health.com/>

National Down Syndrome Society [USA] <http://www.ndss.org/>

Down Syndrome Medical Interest Group [DSMIG-UK] <http://www.dsmig.org.uk/>

Down Syndrome Education International [DownsEd] <http://www.downsed.org/>

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