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Down Syndrome Health Watch Table

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CONSIDERATION	RECOMMENDATION
1. HEENT (HEAD, EYES, EARS, NOSE, THROAT)	
<i>Children:</i> ±15% have cataracts; >50% have significant refractive errors; 50 – 80% have a hearing deficit	<ul style="list-style-type: none"> <input type="checkbox"/> Neonatally: refer immediately to specialist if red reflex is absent or if strabismus, nystagmus, or poor vision is found <input type="checkbox"/> Arrange ophthalmological assessment: first by 6 months for all; then annually (or more frequently as needed) <input type="checkbox"/> During childhood: screen with history and exam; then 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 with history and exam; review risks for frequently occurring serous otitis media <input type="checkbox"/> Undertake auditory testing: first at 9 – 12 months, then every six months up to 3 years and annually thereafter until adulthood
<i>Adults:</i> ± 15% have cataracts; 5 – 15% have keratoconus; 20 – 70% have significant refractive errors; 50-90% have a hearing deficit	<ul style="list-style-type: none"> <input type="checkbox"/> Arrange ophthalmological assessment every 1 – 2 years, with special attention to cataracts, keratoconus and refractive errors <input type="checkbox"/> Undertake auditory testing every 2 years
2. DENTAL	
<i>Children:</i> tooth anomalies are common	<ul style="list-style-type: none"> <input type="checkbox"/> Undertake initial dental exam at 2 years and bi-annually thereafter. Encourage proper dental hygiene. Make orthodontic referral if needed
<i>Adults:</i> have increased risk of periodontal disease	<ul style="list-style-type: none"> <input type="checkbox"/> Undertake bi-annual clinical exam with referral as appropriate
3. CARDIOVASCULAR	
<i>Children:</i> 30 % - 60% have congenital heart defects (CHD)	<ul style="list-style-type: none"> <input type="checkbox"/> Newborn screening: Arrange pediatric consult with echocardiogram <u>even in the absence of physical findings</u> <input type="checkbox"/> In children and adolescents: review cardiovascular history and check for physical signs with specialist referral if indicated <ul style="list-style-type: none"> <input type="checkbox"/> Refer for echocardiogram if not previously done <input type="checkbox"/> Undertake SBE prophylaxis as indicated by findings

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<p><i>Adults:</i> >50% have cardiovascular concerns, commonly including acquired mitral valve prolapse (MVP) and valvular regurgitation</p>	<ul style="list-style-type: none"> <input type="checkbox"/> Ascertain comprehensive cardiovascular history <input type="checkbox"/> Undertake annual cardiac exam, with echocardiogram to confirm new abnormal findings and follow-up depending on the type of cardiovascular problem present and any repair undertaken – consider referral to Adult Congenital Heart Disease clinic <input type="checkbox"/> Echocardiogram is indicated if new abnormal physical findings or inability to assess adequately in physical exam. Echocardiogram should be considered to establish baseline if not previously done or records unavailable (1)
4. RESPIRATORY	
<p><i>Children:</i> 50%-80% have obstructive sleep apnea (OSA)</p>	<ul style="list-style-type: none"> <input type="checkbox"/> Newborn: Arrange ENT consult if recurrent otitis media infections occur <input type="checkbox"/> Infants and children: Arrange ENT consult and sleep study if history suggests obstructive sleep apnea <input type="checkbox"/> Treat infections promptly and aggressively
<p><i>Adults:</i> 50%-80% have obstructive sleep apnea (OSA)</p>	<ul style="list-style-type: none"> <input type="checkbox"/> Ascertain detailed sleep history, with special attention to obstructive sleep apnea (OSA) symptoms. Refer to ENT, including sleep study if OSA is suspected <input type="checkbox"/> Investigate possible GE reflux/swallowing disorder if aspiration pneumonia is suspected
5. GASTROINTESTINAL	
<p><i>Children:</i> ≤ 50% have gastrointestinal (GI) tract abnormalities including imperforate anus, duodenal atresia, celiac disease and Hirschsprung disease</p>	<ul style="list-style-type: none"> <input type="checkbox"/> Newborn: with vomiting or absent stools check for GI tract blockage and refer to 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 obesity
<p><i>Adults:</i> ≤ 95% have obesity; ±7% have celiac disease</p>	<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 H. Pylori and treat if positive regardless of symptoms <input type="checkbox"/> Manage constipation
6. GENITOURINARY	
<p><i>Children:</i> Cryptorchidism is common</p>	<ul style="list-style-type: none"> <input type="checkbox"/> Examine clinically to detect hypogonadism, undescended testes and possible testicular germ cell tumors, with surgical referral as appropriate
<p><i>Adults:</i> Have increased risk of testicular cancer</p>	<ul style="list-style-type: none"> <input type="checkbox"/> Annual clinical exam with surgical referral as appropriate (3)
7. SEXUAL FUNCTION	
<p><i>Adults:</i> Fertility has been documented in women Fertility in males rarely reported</p>	<ul style="list-style-type: none"> <input type="checkbox"/> Counsel regarding fertility possibility and risk of Down syndrome in offspring

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8. MUSCULOSKELETAL	
<i>Children:</i> ≤15% have atlanto-axial instability (AAI)	<ul style="list-style-type: none"> <input type="checkbox"/> Arrange lateral cervical spine radiographs (flexed, neutral and extended positions) between 3-5 years of age <input type="checkbox"/> Then screen as needed for high risk activities (e.g., tumbling) and Special Olympics participation <input type="checkbox"/> Undertake neurological exam for signs of spinal cord compression and consider MRI and neurosurgical consult if present <input type="checkbox"/> Obtain detailed history with particular attention to possible joint subluxations/dislocations, scoliosis and hip abnormalities
<i>Adults:</i> Continued risk for spinal cord compression secondary to AAI	<ul style="list-style-type: none"> <input type="checkbox"/> Undertake annual neurological exam to look for signs of spinal cord compression <input type="checkbox"/> Arrange lateral cervical spine films 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
9. NEUROLOGICAL	
<i>Children:</i> Seizures in ≤22% ;Dementia has <i>not</i> been documented in childhood	<ul style="list-style-type: none"> <input type="checkbox"/> Take careful neurological history with particular attention to seizures (infantile spasms or tonic-clonic) <input type="checkbox"/> Arrange EEG and possible neurology consult for seizures
<i>Adults:</i> Dementia: 40 – 49 y, 11% 60 – 69 y, 77% ≤75% with dementia have seizures with frequency increasing with age	<ul style="list-style-type: none"> <input type="checkbox"/> Ascertain neuropsychiatric history at every visit with particular attention to change in behaviour, loss of function/ADL 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., chronic pain, medication side effects, depression, obstructive sleep apnea, menopause, hearing/vision deficits, hypothyroidism, low folic acid/vitamin B12) <input type="checkbox"/> Undertake EEG and possible neurology consult for seizures
10. DERMATOLOGICAL	
<i>Children & Adults:</i> Dry skin, atopic dermatitis, seborrheic dermatitis, cheilitis, impetigo and alopecia areata are more common than in general population	<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
11. MENTAL HEALTH/BEHAVIOURAL	
<i>Children:</i> Self-talk very common; Autism spectrum disorder in 5-10%	<ul style="list-style-type: none"> <input type="checkbox"/> Review for other positive or negative signs suggesting psychosis <input type="checkbox"/> Review regularly with respect to behavioural concerns
<i>Adults:</i> ≤ 30% have psychiatric disorder – including depression	<ul style="list-style-type: none"> <input type="checkbox"/> Review regularly with respect to behavioural concerns <input type="checkbox"/> Ascertain neuropsychiatric history at every visit, with particular attention to change in behaviour, loss of function/ADL and new onset seizures

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12. ENDOCRINE	
<i>Children:</i> 1% have congenital hypothyroidism; ≤ 20% develop hypothyroidism after birth	<ul style="list-style-type: none"> <input type="checkbox"/> Review neonatal screening. <input type="checkbox"/> Ascertain TSH and free T4 levels at 6 and 12 months, then annually <input type="checkbox"/> In adolescence, if signs of hyperthyroidism, check for autoimmune thyroiditis
<i>Adults:</i> 15 – 50% are hypothyroid Hyperthyroidism, autoimmune thyroiditis, and subclinical hypothyroidism are more common than in the general population	<ul style="list-style-type: none"> <input type="checkbox"/> If subclinical hypothyroidism (i.e., elevated TSH with normal free T4) then follow free T4 every 6 months (4) to a year (5) <input type="checkbox"/> For adults who are euthyroid, then check TSH and free T4 levels every 5 years (6) <input type="checkbox"/> Consider checking thyroid function with changes in mental status
13. HEMATOLOGICAL	
<i>Children:</i> Increased frequency of transient myeloproliferative disorder and leukemia	<ul style="list-style-type: none"> <input type="checkbox"/> Neonate to one month of age: Investigate for polycythemia or thrombocytopenia <input type="checkbox"/> Assess history periodically for symptoms of leukemia, with close attention to those with a history of transient myeloproliferative disorder
<i>Adults:</i> do not have an increased risk of leukemia	

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Down syndrome websites that may be useful 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/>