Health Maintenance Checklist for Adults with DS
A Guided Assessment Tool - For annual visits and follow-ups

History:
- Review family history and most recent lab requisition results with caregiver
- Review current and past medications taken for all conditions
- Past and current health and comorbid conditions
- Obtain cardiac history - prior surgeries, cardiac defects, and symptoms of concern: shortness of breath, history of fatigue, or exertional dyspnea (murmur or gallop)
- Sleep history and concerns regarding sleep
  - Determine current sleep issues/concerns
  - How long issues have persisted
  - Daytime behavioural and activity issues
  - Childhood sleep issues
  - Any sleep assessments or consultation conducted or medications given
History:

☐ Discuss with family any noticeable changes in typical behaviours, social states, or overall deterioration of regression (both in early and late adulthood)
  - Evaluate for medical problems that may underlie behaviour changes (thyroid disorder, sleep apnea/issues, celiac disease, gastroesophageal reflux, constipation)
  - Any history of psychiatric issues (i.e. depression, OCD, other mood disorders)

☐ Discuss with family any myopathic or neurological issues that have been observed
  - Onset of seizures; greater risk with age and comorbidity with dementia
  - History of strange movements, gait and balance problems

☐ Discuss sexual function and fertility for both men and women with DS
  - Counsel fertility and risk of recurrence of DS (50%) for women with DS; few reported cases in men with normal phenotype in all cases
  - Discuss contraceptive use and options as well as STD prevention for both genders
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Behavioural Observations and Clinical Exams:

- Perform physical examination and measure BMI
- Cardiac and pulmonary health - listen to heart and determine breathing; look for signs of infection and fluid buildup in lungs
- Celiac disease - chronic symptoms present of dyspepsia, diffuse abdominal pain, flatulence, mouth sores, and skin rashes
- Dementia - conduct yearly cognitive screen and assessment of activities of daily living (ADLs and IADLs) - observe for changes in behaviour or new onset seizures
- Changes in behaviour - in women, determine symptoms for early menopause; look for medical causes for changes- otherwise conduct screening for regression or dementia
- Neurological exam - conduct annually to test for musculoskeletal or myopathic issues; risk assessment for gait/balance issues done through Get-up and Go test
- Domestic violence and sexual abuse screening for traumas and abuse at every clinical encounter
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Lab Tests:

☐ TSH concentration - once every 5 yrs; yearly with positive results

☐ Diabetes mellitus - screen every 3 yrs until 45; yearly for ≥50 yrs
  - FPG (mmol/L) no caloric intake for 8 hours = 6.1-6.9 IFG; ≥7.0 diabetes
  - 2hPG in a 75g OGTT 9 (mmol/L) = 7.8-11.0 IGT; ≥11.0 diabetes
  - A1C (%) standardized, validated assay in absences of factors that affect accuracy of A1C and not for suspected type 1 diabetes = 6.0-6.4 prediabetes; ≥6.5 diabetes
  - Random PG (mmol/L) = ≥11.1 diabetes

☐ Fasting lipid panel - every 5 yrs; increased freq if patient on atypical antipsychotics or diagnosed with diabetes

☐ IgA tissue transglutaminase and total IgA testing - when symptoms of celiac diseases suspected

☐ Hemoglobin concentration, ferritin, and CRP - done annually to monitor anemia and iron deficiency
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Diagnostic/other tests:

☐ Measure BMI and BP - yearly to monitor for obesity risk or follow-up on diagnoses, hypertension
☐ Auditory testing - every 2 yrs
☐ Ophthalmic exam - every 1-2 yrs; vision screen- yearly
☐ Oral exam - done yearly
☐ Echocardiogram - for suspected acquired mitral valve prolapse or valvular regurgitation; monitor/follow-up those with heart surgery
☐ Neurologic exam - for suspected myopathy and spinal cord compression
☐ Bone mineral density screening - regular testing for suspected or confirmed osteoporosis
☐ Lateral cervical spine X-rays - to confirm neutral position prior to any surgeries or anesthetic procedures
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Diagnostic/other tests:

☐ Polysomnography/sleep study - for suspected obstructive sleep apnea and other sleep issues
☐ DS specific baseline battery of dementia testing - once before age 35, every 1-5 years after
☐ Testicular exam - done yearly
☐ Mammography - inconclusive value and low risk for breast cancer in DS population; done for suspected cases
☐ Screen for colorectal cancer - starting age 50
☐ Pap smear assessments - to screen for cervical cancer and gynecologic concerns after age 21; performed every 2-3 years with aging
☐ STD screening - between 13-64 screen once; test annually for at risk patients (multiple partners, unprotected sex, pregnant women, history of STD’s)
Consultations:

- ENT specialist - review audiological evaluation; assess for otitis media
- Ophthalmologist - review vision screen; assess for glaucoma, cataracts, refractive errors and keratoconus
- Dentist - regular care and maintenance every 6 months; assess for gum disease and tooth decay
- Cardiologist - review echocardiogram; assess for acquired mitral and aortic valvular disease
- Radiologist - conduct imaging for atlanto-axial subluxation, bone density (DEXA) scans for assessing signs of osteoporosis
- Oncologist - for specific cancers that increase in prevalence with age; particular attention for testicular cancer screening
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Consultations:

☐ Gynecologist - review specific reproductive health care needs of women with DS
☐ Dermatologist - review routine care evaluation; address concerns for atopic dermatitis, chelities, impetigo and alopecia areata
☐ Neuropsychiatrist - referral for specialized evaluation for chronic behavioural problems, acute deterioration in function or suspected dementia
☐ Sleep specialist - assess for underlying causes of sleep issues/disorders; referral to sleep study and/or CPAP evaluation
☐ Geriatrician - issues with aging and critical/palliative care for the elderly with DS