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| Summary of Recommendations for the Adult Patient with Down Syndrome | | |
| **Condition** | **Prevalence** | **Recommendations** |
| Thyroid dysfunction |  | * Euthyroid-TSH, free T3/T4 every 5 years * Subclinical hypothyroidism- thyroid microsomal and thyroid autoantibodies once, if negative, then TSH, free T3/T4 every 5 years. If positive, consider more frequent screening. * Clinical hypo- or hyperthyroidism—treat with appropriate regimen same as in general population |
| Congenital heart disease | 40%-50% of children | * Awareness of higher all-cause mortality if CHD present. * Consider evaluation for surgical repair if clinically indicated |
| Cardiovascular disease |  | * Fasting lipid panel every 3-5 years after age 2 but no later than 10 if any of following present: overweight, obese, hypertension, tobacco use, diabetes mellitus, family history of dyslipidemia or CVD * Fasting lipid panel every 5 years beginning at age 18 if no other risk factors present |
| Hypertension |  | * Blood pressure screening every 2 years after age 18 * Baseline EKG at diagnosis of hypertension |
| Early menopause |  | * No screening recommended. * Treat symptoms (eg, hot flushes, mood swings) with hormone therapy as in general population |
| Ophthalmic disorders |  | * Vision screen yearly and ophthalmic exam every 1-2 years |
| Hearing loss |  | * Hearing assessment every 2 years in all patients with Down syndrome |
| Diabetes | Inconclusive evidence | * Follow USPSTF recommendations for type 2 DM screening * Evaluate for type 1 DM if indicated by symptomatology. |
| Leukemia |  | * No screening available. Evaluate refractory anemia and lymphadenopathy with high index of suspicion. |
| Other neoplasms |  | * Perform diligent yearly ophthalmic, testicular, and lymphatic exams (due to increased risk of retinoblastoma, germ cell tumors [ie, testicular carcinoma], and lymphoma.) * Mammography inconclusive evidence of value in Down syndrome |
| Seizures |  | * No screening indicated. * Increased incidence in 5th and 6th decade frequently associated with Alzheimer dementia |
| Alzheimer dementia | Universally present by age 40 | * Yearly cognitive screen and assessment of ADLs and IADLs * Down syndrome-specific baseline battery of dementia testing at least once before age 35, and every 1-5 years thereafter |
| Infectious diseases | 5th highest cause of death | * Respiratory illnesses are the most common and lethal * Yearly flu shot and serial boosters of Pneumovax |
| Gastrointestinal disorders | Celiac disease 6-fold increased risk, gut dysmotility extremely common | * IgA tissue transglutaminase and total IgA testing if chronic symptoms present (dyspepsia, diffuse abdominal pain, flatulence, mouth sores, and skin rashes) * Treat constipation with physical therapy and dietitian management, fiber supplements, and gentle laxatives |
| Contraception |  | * Low-dose OCPs, transdermal patch, and depot medroxyprogesterone generally preferred. * IUD can be considered when patient is a candidate for insertion. |
| Abuse | 68%-83% (females sexually assaulted) | * Screen for physical, psychological, and sexual abuse at every clinical encounter * Abuse screening required by many legal jurisdictions |

ADL, activities of daily living; CHD, congenital heart disease; CVD, cardiovascular disease; DM, diabetes mellitus; EKG, electrocardiogram; IADL, instrumental activities of daily living; IgA, immunoglobulin A; IUD, intrauterine device; OCP, oral contraceptives (or oral contraceptive pills); TSH, thyroid-stimulating hormone; USPSTF, US Preventive Services Task Force.