PCP Handout

Caring for Persons with Down Syndrome

Down syndrome (trisomy 21) is the most common genetic cause of intellectual disability (ID) and occurs in 16 of 10,000 U.S. live births. Symptoms vary in each individual with a range of mild to moderate ID and average life expectancy of ~60 years. It is associated with distinct facial and body features. About 95% have classic trisomy 21, while 5% have mosaicism or translocation.

Comorbidity	Summary	Management
Depression	▪ Presents commonly w/ anhedonia, depressed mood, disturbed sleep OR with "deficit syndrome" (apathy, abulia, anhedonia, mutism) ▪ Disintegrative disorder as early adult – catatonic	▪ Use DSM-5 + DSM-ID-2. Screeners - adapted PHQ-9, Glasgow Depression. Screen medical issues eg. thyroid, sleep, hearing, vision. Rx SSRI. Refer to mental health w/ skills in IDD.
Autism	▪ 16-18% co-occurring autism, inc. GI sx, infantile spasm, scoliosis	▪ Evaluate autistic concerns as may change approach to treatment, eg. ABA.
Hypothyroidism	▪ Prevalence 40% < age 30, up to 50% in >30 ▪ Symptoms obscured eg. weight gain, constipation, xerosis common w/ & w/o thyroid dis, also difficulty communicating fatigue, cold intolerance	▪ Screen with TSH every 1 to 2 years from age 21 ▪ ~30% TPO Ab+, increase screening if positive or subclinical hypothyroid ▪ Rx improves cognitive function and weight
Dementia	▪ Over 50% of older adults develop dementia ▪ Caution dx if < age 40 due to low prevalence ▪ Consider hypothyroid, obstructive sleep apnea (50-75%), hearing loss (up to 75%), vision loss (DS higher risk keratoconus, cataracts, strabismus), ▪ Observe for co-occurring late onset seizures.	▪ Interview caregiver for functional decline yearly from age 40 (NTG-EDSD) in cognition/ memory/ executive/adaptive function, behavior / personality, communication, ambulation/motor, established skills ▪ Screen thyroid, sleep, hearing, vision, mental health. Address advance care planning.
Diabetes	▪ Higher prevalence vs. age matched controls ▪ Premature aging increases risks of cataracts, kidney, peripheral nervous system damage	▪ Screen A1c or fasting glucose every 2-3 years from age 30. If obese, start at age 21.
Celiac Disease	▪ ~11%, GI sx of constipation, loose stools, cramping, non-GI sx of rash, joint pain, bone loss symptoms can be difficult to recognize ▪ Other GI issues – constipation, GERD, Hirschsprung's, duodenal atresia	▪ Annual assessment for GI and non-GI symptoms ▪ IgA deficiency can obscure anti-TTG or anti-gliadin results.
ASCVD	▪ CAD prevalence low (< 1.5%), increased risk moya moya, cardioembolic stroke w/ CHD	▪ Assess 10-year ASCVD risk re: statin use every 5 years
CHD	▪ ~50%, esp. atrioventricular septal abn ▪ Risk arrhythmias, pulmonary hypertension (higher w/, still increased w/o CHD)	▪ Recommend one adult echo if no prior CHD. ▪ Periodic cardiac evaluation by adult congenital cardiology
Obesity & eating	▪ Poor appetite-satiety control, reduced physical activity, hypothyroid, OSA, adverse med effects ▪ Also risks of choking and dehydration	▪ Screening BMI annually ▪ USPSTF interventions (healthy diet, regular exercise, calorie control)
Atlantoaxial instability (AAI)	▪ 10% adults age <30, symptomatic ~1% ▪ Don't restrict physical activity ▪ Flex/extend cervical xray measures instability, no routine screen if asymptomatic	▪ Annual screen for myelopathy w/altered gait, new incontinence, brisk reflexes, clonus ▪ Shared decision re: high risk diving, horseback riding, gymnastics, skiing
MSK - Osteoporosis, ligament laxity, arthritis	▪ DEXA no data, bisphosphonates don't correct reduced bone formation, FRAX not applicable ▪ Pes planus, patellar/hip dislocate ▪ Increased gout, early osteoarthritis	▪ For fragility fracture, screen celiac, vit D def, hyperthyroid/parathyroid, adverse med use ▪ Gait changes - consider AAI, OA, gout, orthotics for pes planus
Midface hypoplasia	▪ Increased otitis media, sinusitis, cerumen impaction, sleep apnea. bruxism, dental caries.	▪ Regular dental and ENT exams. Lower threshold for sleep study. Re-evaluate post T & A.
Skin	▪ Xerosis 70%, atopy 50%, folliculitis, alopecia 6-9%, vitiligo 4%, hidradenitis suppurative 2%	▪ Non-perfumed mild soaps/lotions

Reference: Tsou et al. Global Down Syndrome Foundation Medical Care Guidelines for Adults with Down Syndrome Workgroup. Medical Care of Adults With Down Syndrome: A Clinical Guideline. JAMA. 2020 Oct 20;324(15):1543-1556.

Summary by CYACC: April 2025

Center for Youth and Adults with Conditions of Childhood (CYACC)