PCP Handout

Caring for Persons with Turner Syndrome

Turner syndrome is the most common chromosomal abnormality in female live births, affecting ~ 1:2,500. In the individual's cells, the X chromosome can be absent (40-50%), structurally abnormal (15% isodicentric), or mosaic (15-25% XX, 10-12% XY). X deletion distal to Xq24 are not included. Symptoms vary in individuals.

System	Information	Follow-up
Cognition	▪ 90% avg intelligence. Small ring X with risk of severe intellectual disability. ▪ Generally higher verbal reasoning. Increased nonverbal impairments i.e. visual spatial, math learning disability, autism, ADHD, executive / social emotional issues.	▪ Neuro-psych and behavioral screening ▪ Review school function, decision making, other functional supports as needed
Nutrition	▪ Obesity common -? Estrogen deficiency, ▪ Anorexia nervosa -? More common	▪ Maintain health BMI ▪ Monitor for eating disorder
Cardio-vascular	▪ Congen. heart disease (40-60%) - Bicuspid AV 14-40%, aortic coarctation 4-15%, hypoplastic left 4-5%, common brachiocephalic/L carotid 6-29%, aberrant R subclavian 6-8%, persistent L SVC 2-13%, anomalous PVR 4-16% ▪ Inc. HTN (60%), pre-eclampsia, ischemic heart dis ▪ Thoracic aortic aneurysms 25%, aortic dissection. Further inc. risk in pregnancy.	▪ BP in both upper/lower extremities (screen for coarct) ▪ EKG as infant, age 9-11 years, q5-10 years as adult. ▪ Echo as infant, age 9-11 years, q 5-10 years as adult. ▪ Cardiac MR as teen, w/in 2 yrs of planned pregnancy. ▪ Surveillance for aortic dissection, urgent for any symptoms. If at risk, frequent cardiac re-eval in pregnancy. ▪ Rx aorta dilation w/ or w/o HTN, beta blocker, ACE. ▪ If mild-mod aortic dilate, low/ moderate static / dynamic sports, avoid intense weight-training. If mod-sev., limit sports.
ENT/ Respiratory	▪ Otitis media (75%), cholesteatomas (7%), conductive / sensorineural-hearing loss (36-84%, 61% >35 years), inc. sleep apnea	▪ Audio q 5 yrs as adult. Assess balance if hearing loss. ▪ Screen for sleep apnea symptoms.
GI	▪ 40-80% increased liver enzymes, 4.5% celiac. ▪ Inc. IBD 18%, intestinal telangiectasias, inc. colon cancer (despite no IBD),	▪ Screen LFT q 1-2 yrs start at 10. If increased x 2, Fibroscan. ▪ Anti TTG and IgA q 2-5 yrs, start age 2. ▪ Screen for iron deficiency w/ CBC q 1-2 yr in teen/ adult
GU	▪ Spontaneous puberty (5-20%), higher w/ mosaic; premature ovarian failure ▪ Inc. gonadoblastoma (27% by age 30) ▪ Inc. risk Breast & endometrial cancer with HRT ▪ Spontaneous pregnancy 10% (most in mosaics), 29-48% miscarriage, inc. C/S for small pelvis, ? inc. risk chromosomal abn. ▪ 20% horseshoe kidney/ duplicate collecting system. 5% malrotation/multi-cystic.	▪ LH, FSH, anti-Müllerian (AMH) start age 8-9 yearly to 11-12 years. Lo ERT age 11-12 if FSH abn. x 2. Inc. over 2-4 yr, goal E2 100-150 pg/ml. Prefer transdermal ERT, add prog 1.5-2 yr later, dec. ERT to low dose age 50-55 ▪ Screen for Y chromosomal material if virilized, consider prophylactic gonadectomy. ▪ Preventive cancer screening. ▪ Genetic counsel, consider early fertility preservation ▪ Annual U/A if abn. kidney. Check BUN/Cr, UTI by sx.
Endocrine	▪ Short stature (98%) absent short stature homeobox-contain (SHOX) gene, ▪ Hypothyroidism (25-30%) ▪ Glucose-stimulated insulin secretion defect, DM2 more common, hyperlipid unrelated to BMI, hypertriglyc. w/ obesity & hyperinsulinemia.	▪ Growth hormone (GH) as early as age 2 for abn. growth, continue to bone age ≥14 yrs, ideal > 4 yr prior to puberty. Measure IGF-I at least annually. ▪ On GH, risk of intracranial HTN, SCFE, scoliosis. ▪ TSH q 1-2 yrs, start age 2. If inc., antibodies. ▪ Lipids q 3 yrs. A1c q 1-2 yr after 10-2yo. If inc, check antibodies.
MSK	▪ Scoliosis (10 %), kyphosis, cubitus valgus, genu valgum, Madelung (bayonet) deformity of wrist, short 4th/5th metacarpals/tarsals ▪ Osteoporosis 23.8%; decrease peak bone mass (density normal if spontaneous puberty)	▪ GH improves bone density if used for 1 year in teens ▪ 25OH vit D every 2-3 yrs, start age 9-11. ▪ Dexa prior to age 21, every 5-10 yrs ▪ As adult - HRT, weight bearing exercise, calcium; bisphosphonate role unclear
Skin	▪ Lymphedema 12-27% -common early (may recur with HRT), webbed neck 18-25% (inc. w/ ao coarct) ▪ Nevi in non-sun exposed areas, ? inc. malignant transform; Inc. psoriasis, alopecia, vitiligo, nail pitting/convexity, ? inc. keloid risk.	▪ Annual skin exam. Counsel re: sunscreen, keloid prior to surgery ▪ Compression garments for lymphedema.
Ophthal	▪ Eye abn. 63%; refractive error 40%, strabismus 25%, ptosis, amblyopia, color blindness 8%.	▪ Monitor regularly
Psych	▪ Self-esteem, body image issues, social anxiety, higher depression / mood disorders	▪ Screening and counseling as needed

Reference: Gravholt CH et al Intl Turner Syndrome Consensus Group. Clinical practice guidelines for the care of girls and women with Turner syndrome: Euro J Endocrin Jun 2024, 190 (6), G53–G151.

Summary by CYACC: Dec 2025

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