PCP Handout

Caring for Persons with Sickle Cell Disease

Sickle cell disease (SCD) is an inherited, chronic disorder of sickling erythrocyte, microvascular occlusion, hemolytic anemia affecting multiple organ systems. Universal newborn testing began in 1987. Types include homozygous (HbSS), HbSC, sickle beta plus thal (HbSβ+ thal.), sickle beta null thal (HbSβ0 thal.) plus trait.

System	Information	Follow up
Cognition/ psych	- change in intellectual abilities / academic achievement assoc. with silent CVA	- GAD7, PHQ9, functional abilities - Neuropsych test all at age 12-18 + as needed
ID	- encapsulated organisms—UTI, pneumonia, osteomyelitis - early childhood penicillin px 0-5yo, continue if higher risk - vaccinate for asplenic state	- Pneumovax every 5 years, flu yearly - if risk or transfused past yr: HIV, Hep C & B
Cardio-vascular	- dyspnea alone common in SCD (40-50%) w/ and w/o PH - pulmonary hypertension 6-11% adults (mPAP >=25) , alert to sx of progressive exertional dyspnea, esp. stair climbing, +/- exertional chest pain, light-headedness, syncope - Assoc. mortality with prolonged QTc in SCD	- evaluate exertional hypoxemia, elevated JVP, loud/fixed split S2, tricuspid regurg, S3 or S4, parasternal lift, pulsatile/large liver, periph edema. - BNP, echo doppler, if abn refer to cardiology/ right cath, caution against over-diuresis - screen EKG esp if QT prolonging med
Pulmonary	- sleep disorders, nocturnal hypoxemia, infiltrative lung disease, exertional dyspnea - acute chest crisis pain, fever, cough, low O2, triggers = hypoventilation, pulm infections, vascular occlusions	- PFT, ventilation/perfusion, sleep study - consider HIV disease, sarcoidosis, connective tissue disease - acute chest 4% adult mortality, Rx aggressively
GI	- surveillance for transfusion related iron overload - acute splenic sequestration in younger patients - LUQ pain, pale, inc HR/RR - transfuse - acute abd pain, cholelithiasis 11% esp teen to young adult	- check ferritin/transferrin, chelation as needed - consider increase hearing loss risk (13%) esp w/ chelation, consider hearing screening
GU	- Hyposthenuria, enuresis, dehydration - Proteinuria—early indicator of nephropathy - Risk papillary necrosis - check hematuria - Delayed pubertal development - Ischemic priapism emergency, risk corporal fibrosis, erectile dysfunction; 33% adult males - Stuttering priapism = recurring unwanted painful erections - Exacerbation of hemolysis or anemia during pregnancy	- screen for enuresis, encourage high hydration - ACE/ARB protect kidney for proteinuria or HTN - screen growth/puberty progress, hx priapism - emergent aspiration, prn sympathomimetic - considered progesterone-only contraception for menstrual assoc vaso-occlusive sx - OH-urea impact sperm/ova, avoid pregnancy
Skin	- leg ulcers	- evaluate for venous stasis skin changes - consider echo & change in disease modifiers
MSK	- vaso-occlusive pain crises - assess person's usual pattern, often symmetric bilateral, 2 or more sites - avascular necrosis up to 50% esp shoulder/hip, chronic arthritis, vertebral deformity (fish mouthing), osteopenia	- VOC correct inciting causes, control pain, maintain euvolemia, transfuse as needed - work up diff dx, esp if different pattern - annual joint mobility, symptoms
Heme	- Hgb 6-12, retic 5-20, mild disease phenotype = no organ damage or very infrequent pain meds, Rx disease modifiers (hydroxyurea, crizanlizumab, voxelotor, L-glutamine) - aplastic crisis esp w/ parvo, rare hemophagocytic lymphohistiocytosis, hemolytic crisis esp G6PDase def. - Trait—risk micro-vasoocclusive events, rhabdomyolysis assoc. across mild to moderate exertion	- annual & w/ acute illness—CBC, diff, retic, comp chemistry, total /direct bili, LDH, ferritin, transferrin, U/A, Ur Alb/Cr, 25-OH Vit D, RBC Ab screen. Once geno/phenotype (C/D/E/Kell) - HLH = fever, hepato or splenomegaly, bi or trilineage cytopenia, elevated ferritin - in trait, metabolic gap acidosis = high risk factor
Neurology	- silent cerebral infarction 39% by age 18 - esp male, lower baseline Hgb, higher baseline systolic BP, previous seizures - also berry aneurysms, moyamoya disease	- assess cranial nerve & gait at least yearly - Hx transcranial Doppler U/S (TCD/TCDi) yearly ages 3-16, adult brain MRI if Hgb SS/S beta 0 - Refer to neurology for silent CVA
Ophthal	- Retinopathy, central retinal artery inclusion	- annual dilated eye exam start age 10
Other	- Genetic counseling regarding risk to offspring. - Untreated dental disease is source of infection	- every 6 month dental care

Reference: https://sicklecellcenters.org/consensus_recommendations/general

Summary by CYACC: April 2025

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