PCP Handout

Caring for Persons with Neurofibromatosis 1

Neurofibromatosis type 1 is a neurocutaneous disorder in 1 in 3,000 people. Presentation is variable, even within families, with median life expectancy 10-15 year below average. Germline mutations in NF1 tumor suppressor gene alter protein production of neurofibromin, which regulates the RAS/mitogen-activated protein kinase (MAPK) cell signaling pathway. Dysregulation leads to benign / malignant tumors and various non-tumor manifestations. It is inherited in an autosomal dominant pattern; 50% are new mutations.

System	Information	Follow up
Cognition	▪ Neurocognitive issues 50–80%, learning disabilities 50-75%, ADHD 40-50%, autism 30-40%, intell dis 6-7% ▪ Most common - visual spatial, social, attention issues ▪ Frequent motor, executive, memory, language issues	▪ Developmental screening ▪ Neuropsych testing and support services as needed
Skin	▪ = 6 cafe-au-lait macules > 5 mm prepubertal, > 15 mm post-pubertal, not palms/soles ▪ = 2 neurofibromas, later childhood, 1 plexiform neurofibroma 50%, overlying brownish purple or hairy ▪ Freckled axilla or inguinal, over age 5 ▪ Juvenile xanthogranuloma - transient, nevus anemicus 50%, pruritus	▪ Dermatology surveillance ▪ ~20% plexiform neurofib need intervention (likely congenital, grow in first 2 decades of life)
Eye	▪ Iris hamartomas (lisch nodules) school-age 50%, most teens ▪ Optic gliomas 15-20% - varied growth & visual loss ▪ Rarer neovascular glaucoma	▪ Annual ophtho exam starting age 6-8 mo then every other yr after age 8 if nl, MRI for sx ▪ Screen for progressive visual impairment <50%, proptosis, hypothalamic dysfnc
Cardio-vascular	▪ HTN 30%, renal artery dysplasia 1-2%, pheochromocytoma, primary essential ▪ Inc. NF1-assoc. vasculopathy, arterial stenosis / aneurysms, younger ages, moyamoya carotid stenosis ▪ Rarer - adult pulm HTN, congen heart defects 2% - pulmonary stenosis, ASD, VSD, Ao coarct	▪ Annual BP monitoring ▪ Renal vascular evaluation for HTN ▪ Imaging for sx
Oncology	▪ Cumulative inc. lifelong malignancy ▪ Increased breast cancer 2-4%, before age 50 ▪ Malignant peripheral nerve sheath tumors 8-15%, age 20-40, GI stromal tumors, esp. small intestines 5-25%, pheochromocytomas 0,1-5,7%, rhabdomyosarcomas esp. GU / head/neck ▪ Inc. juvenile myelomonocytic leukemia <1%	▪ Annual mammogram &/or MRI start age 30 ▪ MRI for clinically suspected intracranial / other tumors ▪ Endoscopy for GI sx, bleeding
Endocrine	▪ Short stature, precocious / delayed puberty, macrocephaly, typically benign ▪ Inc. neurofibromas & HTN w/ pregnancy, higher pregnancy complications, 50% NF1 in fetus	▪ Watch growth curves for signs of hypothalamic dysfnc
GI	▪ Constipation 30%, abdominal migraines	▪ Migraine rx can help abd pain
Musculo-skeletal	▪ Scoliosis 10-30%, cervical/thoracic, pectus excavatum/ carinatum 30%, tibia dysplasia 2%, pseudoarthrosis, sphenoid wing / vertebral dysplasia assoc.w/ nearby neurofibroma or dural ectasia ▪ Low bone density, inc fracture risk	▪ Scoliosis surveillance ▪ Consider Dexa scan ▪ Check 25 vit D
Neurology	▪ Hyperintense lesions on T2 brain MRI pathognomonic, ages 8-16, brain/spinal cord gliomas, esp brainstem, cerebellum, aqueduct stenosis 1.5% ▪ Headaches, often migraines, seizures 4-6%, onset across lifespan w/ mean 14 yo, abn. sleep limb movements, sleep apnea	▪ Surveillance for neuro sx, MRI for sx ▪ Screen for seizure-like sx ▪ Evaluate sleep ▪ Sleep study for sx
Other	▪ Noonan features 12 % – short, facies, heart defect ▪ Inc anxiety, depression	▪ Mental health screening

Reference: Miller DT et al, Health Supervision for Children with Neurofibromatosis Type 1. Pediatrics May 2019.

Summary by CYACC: Dec 2025

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