PCP Handout

Caring for Persons with Muscular Dystrophy

Muscular dystrophies are characterized by progressive muscle weakness and atrophy. Duchenne and Becker types are related conditions, primarily affect skeletal and cardiac muscle. Occur almost exclusively in males. Together affect 1:3500 - 5000 newborn males as X-linked recessive. Different mutations in the DMD gene alter instructions for the protein dystrophin located primarily in skeletal and cardiac muscle. Dystrophin helps stabilize and protect muscle fibers and plays a role in chemical signaling within cells.

System	Information	Follow-up
Cognition	▪ IQ average ~ 1 SD below norm, limits esp. in working memory, increased dyslexia, dyscalculia, dysgraphia w/ 20% moderate + 20% serious reading problem. ▪ Co-morbidities 23% autism, 35% ADHD, and OCD.	▪ Evaluate neuropsych if concerns re: performance. ▪ Consider nonstimulants for ADHD due to arrhythmogenic risk of stimulants
Neurology	▪ Progressing muscle weakness, affects hip/legs early. ▪ New treatments include gene therapy, exon skipping treatments, novel corticosteroids and histone deacetylase enzyme inhibitor.	▪ Rapidly changing treatments required authorization and oversight by teams that are expert in care.
Pulmonary	▪ Respiratory failure with decline in function ~ 4-6.9%/ yr from age 9, some improve/delay w/ corticosteroids. ▪ Night arousals, day sleepiness from nocturnal desaturation and hypercapnia. ▪ Progressive hypoventilation/ atelectasis/ reduced cough assoc w/ lung infections. PCF > 270 l/min = effective cough. ▪ If cushingoid, think about OSA. ▪ Poor swallow causes aspiration. ▪ Need for ventilatory support increases as FVC <50%, particularly FVC < 30%.	▪ Routine pulmonary care. ▪ Annual FVC/PFC measures. ▪ Resting O2 sat > 95% unlikely to be hypercapnic. ▪ Shared decision-making regarding supports. ▪ Cough assist, therapy vest, non-invasive vent options.
ENT	▪ Sialorrhea	▪ Rx scopolamine patch, glycopyrollate, botox.
Cardio-vascular	▪ Cardiomyopathy in most by age 18, often w/ atypical symptoms - anorexia, abdominal pain, fatigue. Some delay ~ 2 yr w/ steroids. Risk from arrhythmias.	▪ Annual cardiology visit w/ echo or MRI. ▪ BNP not predictive. Early use of ACE inhibitor. More benefit w/ salcubitril/valsartan. ▪ ICD post VT or arrest.
Renal/urology	▪ Progressive GFR decreases, often w/ underhydration and hypertension. ▪ 10% renal stones. ▪ Small capacity, hyperreflexic bladder, detrusor sphincter dyssynergia.	▪ Push hydration. Watch GFR especially w/ CHF. For small muscle mass, use cystatin C for GFR. ▪ If incontinence, evaluate bladder emptying. Reduce lordosis to help bladder emptying.
Heme	▪ Assess risk for DVT/PE	▪ Consider prophylaxis for risk of DVT/PE with severe LVH/a fib.
GI	▪ Steroids induce overweight. ▪ Adult - higher rates of cachexia w/ dysphagia. ▪ High rates constipation, GI dysmotility, GER.	▪ Monitor BMI at least semi-annually. ▪ Treat constipation, bloating/GERD. ▪ Evaluate swallowing issues.
Endocrine	▪ Risks of steroid dependence -hypertension, cataracts, glucose intolerance, obesity, infections, short stature, friable skin, GI perforation, pubertal delay d/t hypogonadism, osteoporosis, adrenal insufficiency, fat embolism d/t osteoporotic fractures. ▪ Watch for acanthosis nigricans.	▪ Adrenal crisis alert. ▪ Watch for metabolic syndrome children on steroids. If weaning steroids, go slow. ▪ Lateral spine film or DXA for bones. Use vitamin D. Consider bisphosphonates. ▪ Testosterone to initiate puberty. Check LH, FSH, AM testosterone if concern for adult central hypogonadism
MSK	▪ 68-90% scoliosis w/in 2 years of loss of ambulation w/ risk of rapid progression, contractures.	▪ Monitor curve
Mental health	▪ 17–29% depression and/or anxiety, increases as adult, social isolation, grief.	▪ Manage pain, gabapentin helps muscle pain. ▪ Use mental health, palliative care and advance care planning.

Reference: Quinlivana R et al. Adult North Star Network (ANSN): Consensus Guideline of Standard Care for Adults with DMD. *J Neuromusc Dis*, 2021.

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