Imperatives for DUCHENNE MD

a guide for providers

Diagnosis • If developmental delay or elevated liver enzymes, do a creatine kinase (CK) ChildMuscleWeakness.org • If CK is high (CK>800), order full genetic testing for Duchenne muscular dystrophy • Discuss carrier testing/reproductive options for mother and testing for other family members

Use Support • Direct to trustworthy, reliable online resources; Offer contact with patient organizations (ParentProjectMD.org, TREAT-NMD.eu, UPPMD.org) • Organize follow up via a comprehensive neuromuscular center with expertise in caring for people living with Duchenne

Corticosteroids • **Start early!** *Discuss the benefits and possible side effects of corticosteroids by age 3yo, or as young as possible* • Evaluate efficacy and manage side effects of corticosteroids at each neuromuscular visit • Discuss the rationale for long term steroid treatment

Heart • Cardiology visit with imaging (echocardiogram or cardiac MRI) at diagnosis or by age 6, then every two years until age 10 (or as needed), then annually (or more often if needed) • Discuss cardiac medications if fibrosis is seen on cardiac MRI, for any decrease in cardiac function decreases from baseline or for heart failure (SF or shortening fraction <28%, EF or ejection fraction <55)

Every visit • Monitor weight • Assess/discuss diet (healthy eating, calcium, vitamin D) • Evaluate swallowing/need for intervention • Treat GERD and constipation as necessary

Never forget Physical and Occupational therapy, physical medicine and rehabilitation • Specialized evaluation every 4-6 months • Discuss contracture prevention (splints, stretches), appropriate exercise, assistive mobility devices (strollers, scooters, wheelchairs) and other assistive devices (beds, arm assistance, lifts, etc.)

Nor Bone density • If taking steroids, check 25-OH vitamin D prior to starting steroids, then annually • Supplement vitamin D as needed • Nutrition discussions of adequate calcium and vitamin D intake • Discuss measuremen of bone density and use of bisphosphonates • Assess spine for scoliosis while ambulatory and with any sign of scoliosis

Evaluate breathing • Pulmonary function test at least once while ambulatory and every year after loss of ambulation • Discuss cough assist when cough peak flow is <270 liters per minute or if cough becomes weaker (use during respiratory illnesses while ambulatory and daily and as needed after loss of ambulation) • Discuss nighttime Bi-PAP as needed or when forced vital capacity (FVC<30) • Keep immunizations (including pneumonia and annual flu) up to date • Treat respiratory infections promptly and aggressively

Mental health • Assess adjustment, coping, behavioral and emotional disorder and social isolation for the patient and family at each visit • Screen for learning disability, speech and language problems, attention deficit disorder (ADD), attention deficit and hyperactivity disorder (ADHD), autism and obsessive compulsive disorder (OCD) • Neurocognitive evaluation done at diagnosis and prior to formal schooling; screening/management as needed • Discuss the need for individualized/special educational plan

Do have patients/parents carry a copy of their last visit note/summary (including medications and neuromuscular contact information) and a Duchenne emergency card with them at all times • Use caution with all anesthesia • Avoid succinycholine

For more information:

Center for Disease Control and Prevention Care Considerations
ParentProjectMD.org/CareGuidelines

Family Friendly Version of the Care Considerations
ParentProjectMD.org/CareGuidelinesFamilyPF

Care for Duchenne
ParentProjectMD.org/Care

Parent Project Muscular Dystrophy
LEADING THE FIGHT TO END DUCHENNE

TREAT-NMD Neuromuscular Network