

## Provision of Care

Care will be provided according to the standards as identified by the Duchenne muscular dystrophy, Care Consideration Working Group, and specified by the Certified Duchenne Care Center Program. Measures for care include:

### Coordination and Communication

- A written summary with recommendations is sent to the patient's PCP at the time of diagnosis
- A summary with recommendations is sent to the patient's PCP with each appointment
- Disease specific information/resources available/sent to primary care providers
- Patients/families receive a copy of the note/summary sent to their primary care provider and/or will have access to an electronic copy of their note/summary

### AT EACH VISIT:

- Patients are measured for height/length by standing (when appropriate) and ulnar length
- Patients are weighed
- BMI is assessed
- %O<sub>2</sub> Sat measured at least annually in non-ambulatory patients
- CO<sub>2</sub> is measured in non-ambulatory patients
- Each patient is encouraged to have genetic testing.
- Genetic counseling is available at each clinic, and includes discussion of genetic risk, carrier testing and pre-implantation genetics (as appropriate)
- Patients are referred to endocrine with a downward height % or <4cm growth per year
- Sexual maturity is evaluated after age 10years
- Discussions of testosterone initiation if pre-pubertal by age 14 years or testosterone deficient
- Patients >14 yo spend at least a portion of their visit alone with a consistent provider at each visit
- Assess transition readiness at each visit after age 12yo
- Transition plans are in place and, at each visit, include:
  - Consistent person to monitor the transition plan
  - Discuss future goals
  - Set age appropriate goals
  - Discuss social and community participation and friendships/relationships
  - Plans for adult healthcare, education/employment, adult living
- Healthcare POA appointed by 18 yo
- Advanced care planning/advanced directives/emergency care plan in place by 18yo

### Pulmonary

- Patients see pulmonologist at least annually from diagnosis
- Ambulatory patients, for whom it is appropriate, receive standard spirometry (including FVC) at least annually
- Non-ambulatory patients receive some pulmonary function assessment at least every 6 mos, and more often as needed
- Lung volume recruitment (using self-inflating manual ventilation or mechanical insufflation-exsufflation device) is discussed/recommended to be used 1-2 times/day when FVC <60% predicted
- Cough peak flow is measured at least annually and more often as needed
- Assisted cough is offered when cough peak flow is below 270 LPM, FVC <50% or MEP <60 cm H<sub>2</sub>O

- Patients questioned regarding symptoms of sleep related hypoventilation (nighttime awakenings, morning headaches, behavioral changes) at each visit
- Sleep studies offered for signs/symptoms of obstructive sleep apnea or sleep disordered breathing
- Sleep studies discussed/offered when FVC is <50% predicted
- Nocturnal NIV (with back up rate of breathing) is offered to patients with FVC <50%, MIP <60 cm H<sub>2</sub>O or awake baseline SpO<sub>2</sub> <95% and or with evidence of sleep related hypoventilation on polysomnogram.
- Daytime NIV is discussed with patients whose daytime exhaled CO<sub>2</sub> >45 mmHg or greater, SpO<sub>2</sub> <95% or with symptoms of awake dyspnea are present

### **Cardiology**

- All patients see cardiology at least annually from diagnosis
- Patients have an ECG done at least annually
- Patients receive a printed copy of their ECG to use in emergencies
- Every patient receives cardiac imaging (cMRI if available and appropriate, echo if cMRI is unavailable, not appropriate or if q6 mos evaluations are needed) at least annually
- Cardiac medications are started with evidence of cardiac dysfunction or fibrosis or by age 10yo with normal findings
- Patients taking ACEi or ARB have their BUN, creatinine and electrolytes checked annually

### **Neuromuscular Specialist (NMS) (Neurology/PM&R)**

- Patients see the NMS every 6 mos
- The use of corticosteroids, and their side effects are discussed with every patient
- Corticosteroids are recommended to start during the toddler stage before significant decline
- Corticosteroids are continued, barring side effects necessitating discontinuation, life long
- Steroid complications (Obesity, Cushingoid features, acne, loss of bone density, behavioral changes, immune suppression, hypertension, short stature, delayed puberty, GERD/peptic ulcer disease, hirsutism, glucose intolerance, myoglobinuria) are discussed at each visit
- Approved medications are discussed and offered to each patient for whom they are appropriate
- Information on steroid conversion, stress dosing and prevention of adrenal crisis (PJ Nicholoff Steroid Protocol) is provided to each patient
- Patients are immunized/encouraged to receive influenza vaccines annually
- Childhood immunizations (including pneumovax) are encouraged to be UTD and given as recommended (in US, by the CDC)
- The back is visually inspected for scoliosis at each visit
- Radiographic evaluations of ambulatory patients are done when there are clinical signs of scoliosis
- Scoliosis evaluation is performed annually when the curve is <15-20 degrees and every 6 months when the curve(s) >20 degrees during skeletal growth, and after growth if clinically indicated
- There is an experienced orthopedic spine surgeon managing scoliosis and posterior spine fusions associated with this center
- Patients taking steroids get a lateral lumbar spine x-ray at the initiation of steroids and every 1-2 years thereafter (every 2-3 years if not taking steroids) with a Genant score (used to stage vertebral compression fractures)
- Patients are referred to endocrine/bone health for Genant score of Grade 1 or higher, evidence of VCF or first long bone fracture
- Bisphosphonates (IV preferred) are discussed for the management of bone fragility with evidence of VCF, Genant score 2+ and higher or low trauma fracture
- If appropriate, patients are referred to orthopedic surgery for evaluation of heel cord surgery

### **Palliative Care**

- Palliative care is discussed/offered/encouraged for patients
- Palliative care is available as part of the team or by referral

## **Rehabilitation Management**

- Patients see PT/PM&R every 6 mos for an evaluation
- Standardized strength measures are obtained longitudinally to track disease progression and therapeutic response at least annually
- Standardized functional measures are obtained longitudinally to track disease progression and therapeutic response at least annually
- ROM is assessed every 6 mos
- Stretching is recommended
- Ankle foot orthoses (AFOs) are encouraged (nighttime while ambulatory and day or night with LOA) from diagnosis and/or at a very young age
- Hand splints are prescribed if appropriate
- Safety/fracture prevention/complications are assessed and discussed at each visit
- The rehabilitation team provides activity and exercise recommendations at each visit
- The need for assistive devices and adaptive equipment is assessed at each visit

## **Nutrition**

- A registered or licensed dietitian (RD) or nutritionist is part of your team
- The RD is available to see patients at the start of steroids and at each visit, as appropriate, thereafter
- The RD is able to help each family develop a nutrition plan, helping to prevent obesity or treat underweight
- 25OH vitamin D is monitored just prior to starting steroids and repeated annually
- Intake of calcium is assessed at each visit and supplements encouraged as needed
- Swallowing, constipation, GERD and gastroparesis is assessed at each visit

## **Psychosocial/Neurodevelopmental**

- Evaluation of emotional adjustment/coping, speech and language, signs of autism spectrum disorder, emotional disorders and social isolation is done at each visit
- The mental health of the family is assessed at each visit
- Ongoing support is provided either at your center or referrals made to outside providers