Congenital Muscular Dystrophy

Multidisciplinary care, clinical/imaging and genetic diagnosis, respiratory and nutritional complications, cardiac and periprocedural risk, and musculoskeletal/educational support.

2015 AAN/AANEM Evidence-Based Guideline
Evaluation, Diagnosis, and Management of Congenital Muscular Dystrophy
AAN/AANEM Guideline Subcommittee · Neurology 2015;84:1369–1378

LEVEL B · Should C · May

1 Multidisciplinary Care & Counseling

B	Consult a **pediatric neuromuscular specialist** for diagnosis and management; coordinate multidisciplinary care when accessible.
B	Help families access **genetic counselors** to interpret results and inform family planning, when available.
B	**At diagnosis:** advise families on areas of uncertainty, multisystem implications, and the value of interventions for longevity and quality of life.

2 Clinical & Imaging Diagnosis

B	Use **ethnicity/geography**, weakness/contracture pattern, CNS involvement, organ-involvement timing, and CK levels to guide diagnosis of collagenopathies and dystroglycanopathies.
B	Order **brain MRI** for suspected merosinopathies/dystroglycanopathies if sedation risk is acceptable and expert interpretation is available.
C	Consider lower-extremity **ultrasound/MRI** for suspected collagenopathies, or MRI for SEPN1-related myopathy.

3 Muscle Biopsy & Genetic Testing

C	Consider **biopsy** with immunohistochemical staining if subtype unclear after initial studies; perform/interpret at experienced centers.
C	Order **targeted genetic testing** for well-characterized CMD subtypes when available and feasible.
C	Consider **whole-exome/whole-genome sequencing** if no mutation found or genetics poorly characterized, as technology becomes accessible.

4 Respiratory Complications

B	Counsel that respiratory insufficiency and associated problems **may be inconspicuous** at the outset.
B	**Monitor** pulmonary function (spirometry, oxygen saturation) awake and asleep, individualized to clinical status.
B	**Refer** to pulmonary/aerodigestive care teams experienced in the oropharyngeal–reflux–nutrition–respiratory interface, when available.

5 Nutrition & Dysphagia

B	Coordinate **nutrition and growth** trajectory tracking with primary care.
B	Order **multidisciplinary evaluation** (swallow therapy, GI, radiology) if failure to thrive or respiratory symptoms.
B	Recommend **gastrostomy ± fundoplication** via multidisciplinary team, weighing medical and family considerations.

6 Cardiac & Periprocedural Risk

B	**Baseline cardiac evaluation** for all CMD regardless of subtype; interval further testing per baseline results and subtype.
B	**Before procedures:** discuss increased periprocedural complication risk with families before surgery/anesthesia to inform consent decisions.
B	**Postoperative:** monitor longer than usual after sedation/anesthesia for respiratory, nutritional, mobility, and GI-motility complications.

7 Musculoskeletal & Educational Support

B	**Referrals:** PT/OT/speech therapy, seating/mobility specialists, rehabilitation, and orthopedic surgery to maximize function.
B	Consider **range-of-motion exercises, orthotic devices, or heel-cord lengthening** in appropriate circumstances.
C	**Avoid neuromuscular blocking agents** (e.g., botulinum toxin) unless contractures cause substantially greater impairment than potential worsening of weakness.
B	**Education:** refer to special education advocates, developmental specialists, and education specialists as appropriate.

Kang PB, Morrison L, Iannaccone ST, et al. Evidence-Based Guideline Summary: Evaluation, Diagnosis, and Management of Congenital Muscular Dystrophy: Report of the Guideline Development Subcommittee of the AAN and the Practice Issues Review Panel of AANEM. *Neurology.* 2015;84(13):1369–1378.