The increased deployment of multidisciplinary care and advances in diagnostic and therapeutic approaches has driven the need to update the 2010 guidelines for the recommendations of care for patients who are affected by Duchenne muscular dystrophy (DMD).

With the support of the US Centers for Disease Control and Prevention (CDC), the TREAT-NMD network for neuromuscular diseases, Parent Project Muscular Dystrophy (PPMD), and the Muscular Dystrophy Association (MDA), the DMD Care Considerations Working Group (CCWG) was assembled to develop these updated guidelines and recommendations.

The DMD CCWG reviewed the literature and surveyed a number of experts for their determination of the necessity and appropriateness of specific assessments and interventions.

The following is a summary of the guidelines for the recommendations of care for DMD patients and is being disseminated for your information only. Physicians should carefully assess all relevant safety and efficacy data of treatment options before making decisions related to each individual patient.

**AREAS OF FOCUS**

### 8 ORIGINAL TOPICS OF CARE CONSIDERATION: A MULTIDISCIPLINARY APPROACH CAN IMPROVE SURVIVAL OF PATIENTS AFFECTED BY DUCHENNE<sup>1-3</sup>

- Diagnosis
- Neuromuscular
- Rehabilitation
- Psychosocial
- Gastrointestinal and nutrition
- Respiratory
- Cardiac
- Orthopedic and surgical

### GLUCOCORTICOIDS IN A MULTIDISCIPLINARY APPROACH<sup>1</sup>

### 3 NEWLY ADDED TOPICS IN DMD MANAGEMENT: ADDRESSING VARIOUS SPECIALTIES AND THE CONTINUUM OF CARE<sup>1-3</sup>

- Primary care and emergency management
- Endocrine management
- Transitions of care across the lifespan

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A Summary of the Report of the DMD Care Considerations Working Group

Intended for US healthcare professionals only.
**8 ORIGINAL TOPICS OF CARE CONSIDERATION:**
A MULTIDISCIPLINARY APPROACH HAS IMPACTED THE CARE OF PATIENTS AFFECTED BY DUCHENNE\(^1-3\)

Multidisciplinary management of Duchenne patients has contributed to slowing the progression of the natural history of the disease. The new care considerations address patient needs with these focal points:

- Providing guidance to clinicians on advances in assessments and interventions
- Considering the implications of emerging therapies (deflazacort is the first FDA-approved corticosteroid specifically for DMD)
- Prolonging survival

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<th>TOPIC</th>
<th>KEY POINTS</th>
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| DIAGNOSIS                    | Timely diagnosis is a crucial aspect of care of Duchenne. More widespread realization of prolonged survival has shifted to more anticipatory diagnostic and therapeutic strategies, which can help facilitate earlier detection. | - A range of testing is recommended that includes:  
  - Deletion or duplication testing  
  - Genetic sequencing  
  - Muscle biopsy to help detect dystrophin mutation  
  - Carrier testing for female relatives of a male confirmed to have Duchenne |
| NEUROMUSCULAR MANAGEMENT     | CCWG notes that recent studies confirm benefits of starting glucocorticoids in younger children, before physical decline. Long-term glucocorticoid benefits include delayed loss of ambulation, preserved upper limb and lung function, and avoidance of scoliosis surgery. | - Large-scale natural history and cohort studies confirm individuals treated with less than 1 year of corticosteroids prolonged ambulation from a mean of 10.0 years to 11.2 years in individuals treated with daily prednisone, and 13.9 years in individuals taking daily deflazacort  
  - In the studies reviewed by CCWG, deflazacort and prednisone demonstrated improved muscle strength and deflazacort was associated with less weight gain than prednisone  
  - Physiotherapy and treatment with glucocorticoids remain the mainstay of Duchenne treatment and should be continued after loss of ambulation |
| REHABILITATION MANAGEMENT    | DMD is characterized by well-known patterns of progressive muscle degeneration and weakness, postural compensations, and functional losses resulting from dystrophin deficiency. | Suggested assessments and interventions include:  
  - North Star Ambulatory Assessment (NSAA) and timed function tests are foundational approaches for testing motor function during the ambulatory phase and should be done every 6 months  
  - Direct physical, occupational, and speech therapy should be pursued in outpatient and school settings and continued throughout adulthood  
  - CCWG notes the increased use of standardized testing in children as key due to the number of new therapies that are useful for younger children |
| PSYCHOSOCIAL MANAGEMENT      | Patients affected by Duchenne require psychosocial coping strategies and skills, and ongoing emotional support to maximize their daily functioning across environments such as home, school, and the workplace. | - Comprehensive care should address social and cognitive development, quality of life, and factors that affect the patient and family functioning across home, school, and work  
  - Support should be implemented across the patient’s lifespan that promotes thinking about the future and sets the expectations that the individual will actively participate in his or her care and daily activities  
  - A mental health specialist is recommended to help families cope with their emotions |
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| GASTROINTESTINAL AND NUTRITIONAL MANAGEMENT | Decreased energy expenditure, immobility, and glucocorticoid treatment can lead to gastrointestinal or nutritional complications. These complications can adversely affect the respiratory, musculoskeletal, and cardiac systems. | • Individuals affected by Duchenne who are taking glucocorticoids are at an increased risk of weight gain and obesity due to increased appetite and caloric intake as well as sodium and fluid retention, so a registered dietician nutritionist should assess nutritional status, track weight and height, and establish a specific nutrition plan.  
• Bone health monitoring is needed to assess mineral deficiencies, dietary calcium intake, and vitamin D levels of individuals affected by Duchenne.  
• A gastroenterologist should be consulted to help manage constipation, gastroesophageal reflux, and gastrointestinal motility concerns, and when a gastrostomy tube placement is needed.  
• Dysphagia is common and progressive in Duchenne, so it is recommended that screening be done regularly in addition to consulting with a speech-language pathologist. |
| RESPIRATORY MANAGEMENT                   | Implementation of respiratory care considerations requires a multidisciplinary team, including home caregivers. CCWG endorses higher pulmonary function thresholds for initiation of assisted coughing and assisted ventilation than were recommended in the 2010 care considerations. | • Respiratory management includes monitoring respiratory muscle function and the timely use of pulmonary volume recruitment, assisted coughing, assisted ventilation during the night, and daytime ventilation (patients should be using these therapies by the age of 18-21 years).  
• Recommendations include higher pulmonary function thresholds (milder levels of respiratory impairment) to begin with assisted coughing and assisted ventilation, with the intent to begin interventions in slightly younger patients.  
• Patients should receive yearly immunizations with the inactive influenza vaccine (injectable vaccine) and pneumococcal vaccines (PCV13 and PPSV23). |
| CARDIAC MANAGEMENT                       | Cardiovascular complications are a leading cause of disease-related morbidity and mortality among patients affected by Duchenne. A proactive strategy and early diagnosis of cardiomyopathy is essential to maximize quality of life for patients. | • At early diagnosis of cardiovascular complications, a cardiologist from a multidisciplinary care team is recommended, ideally with expertise in cardiomyopathy and heart failure related to neuromuscular disease.  
• Annual electrocardiogram and noninvasive imaging should be done before 10 years, and after that age, asymptomatic patients should have a cardiac assessment annually due to increased risk of left ventricular dysfunction.  
• In the guidelines update, CCWG identifies that female carriers of Duchenne are at risk for skeletal muscle disease and weakened heart muscle, so a cardiovascular magnetic resonance (CMR) or noninvasive imaging is recommended to assess heart function during early adulthood. |
| ORTHOPEDIC AND SURGICAL MANAGEMENT       | The overall aim of musculoskeletal care is to maintain motor function as long as possible, minimize joint contractures, maintain a straight spine, and promote bone health. An interdisciplinary team should assess and treat musculoskeletal complications. In cases of surgical intervention, it is crucial to involve a respiratory physician and cardiologist. | • CCWG states that bone health monitoring should prioritize spinal radiographs over bone mineral density tests; the latter should now be used to identify early signs of bone fragility.  
• Children in the ambulatory stage might benefit from surgery, but it is less frequently recommended than in the past.  
• Surgical interventions to manage contractures of upper and lower extremities are not recommended during late nonambulatory stage, unless pain, positioning, or skin integrity is a concern. |
Treatment with glucocorticoids continues to be the foundation of DMD treatment. The updated guidelines confirm the benefits of glucocorticoids as part of a DMD treatment plan.

Although the benefits of glucocorticoid therapy are well established, careful consideration must be given on when to start therapy, how to administer treatment, and how to manage side effects.

**USING GLUCOCORTICOID TO TREAT DUCHENNE**

<table>
<thead>
<tr>
<th>When to start steroids</th>
<th>Proper dosing</th>
<th>Managing side effects</th>
<th>Continued use in nonambulatory stage</th>
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<tbody>
<tr>
<td>Recent studies have confirmed the benefits of starting steroid therapy before substantial physical decline</td>
<td>When deciding on the proper dose, consider the growth and response of the child, and consult with the family about side effects and nutrition</td>
<td>Glucocorticoids are the mainstay of recommended treatment for DMD. As with any drug, glucocorticoids should be closely managed for side effects</td>
<td>Steroid use should continue after the loss of ambulation</td>
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**Expert recommendations**

- Introduce steroids in younger children as the foundation of the multidisciplinary approach to DMD care
- The recommended starting dose of deflazacort is 0.9 mg/kg/day, and 0.75 mg/kg/day for prednisone
- If side effects unmanageable or intolerable:
  - Reduce steroids by 25%-30% and reassess in 1 month
  - Increase steroids to target dose per weight on the basis of starting dose
  - Reassess in 2-3 months
- Continue steroid use, but dose as necessary to manage side effects
- Older steroid-naïve patients might benefit from initiation of a steroid regimen

Adapted from Fig. 3. in Birnkrant DJ, Bushby K, Bann CM, et al.; DMD Care Considerations Working Group. Diagnosis and management of Duchenne muscular dystrophy, part 1: diagnosis, and neuromuscular, rehabilitation, endocrine, and gastrointestinal and nutritional management. Lancet Neurol. 2018;17(3):251-267.

**CAUTIONS**

**GLUCOCORTICOID DISCONTINUATION AND ADRENAL INSUFFICIENCY**

Patients should not stop taking glucocorticoids abruptly as there may be an increased risk of adrenal insufficiency.

Patient and family must be educated on signs, symptoms, and management of adrenal crisis.

For more information about adrenal insufficiency and discontinuing glucocorticoids, please see “Diagnosis and management of Duchenne muscular dystrophy, part 1” in *The Lancet Neurology.*
The latest recommendations for primary and emergency care, endocrine management, and assessments and interventions are below. They are designed to improve transitions of care across the lifespan, including:

- Functionality
- Quality of life
- Mental health and independence

CCWG strongly recommends the establishment of a medical home, characterized by accessibility and continuity of care, as the platform for all treatment.

### TOPIC

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<thead>
<tr>
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<td>Many primary care and emergency medicine clinicians are inexperienced at managing the complications of DMD. There are several factors that should be brought to the attention of first-line providers.</td>
<td>• Goals for the primary care provider include provision of first-line care for acute and chronic medical issues, coordination of care with specialists, provision of trusted advice, continuity of care across the lifespan, and optimization of well-being and quality of life for patients and their family members.</td>
<td>• The updated guidelines include a listing of key issues that are important to consider when delivering emergency care for patients affected by Duchenne. It may be helpful for families to carry this listing.</td>
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<td>The endocrine complications of DMD and its treatment include impaired growth, delayed puberty, and adrenal insufficiency. Patients and caregivers should be educated on the impact and management of glucocorticoids on endocrine function.</td>
<td>• Screenings and assessments are recommended by CCWG to assess endocrine hormone abnormalities associated with growth failure.</td>
<td>• CCWG made recommendations regarding therapeutic interventions:</td>
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<td>With prolonged survival, patients affected by Duchenne face a unique set of challenges related to transitions of care, especially as they mature into adulthood. Full coordination between patient and care coordinator should be expected for transition planning.</td>
<td>• A care coordinator or social worker should be responsible for transition planning as care can become fragmented if the patient's needs go unmet.</td>
<td>• Transition planning should include a plan for continuity of healthcare with pediatric providers until adult care is established.</td>
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<td>• Patients affected by Duchenne know that they are living with a progressive disease and should be involved in healthcare discussions, even at a young age.</td>
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**KEY TAKEAWAYS FOR USE IN YOUR PRACTICE:**

**CARE CONSIDERATIONS FOR DUCHENNE MUSCULAR DYSTROPHY**

1. A MULTIDISCIPLINARY APPROACH CAN IMPROVE SURVIVAL OF PATIENTS AFFECTED BY DUCHENNE

2. GLUCOCORTICOIDS IN A MULTIDISCIPLINARY APPROACH

   Glucocorticoids continue to be a mainstay for the neuromuscular management of Duchenne and should be started before substantial physical decline and be continued after loss of ambulation.

3. 3 NEWLY ADDED CARE CONSIDERATION TOPICS ADDRESS VARIOUS SPECIALTIES AND THE CONTINUUM OF CARE

   The newly added topics include:
   - Primary care and emergency management
   - Endocrine management
   - Transitions of care across the lifespan

References: