IMPORTANT NEW UPDATE FOR CAREGIVERS

Caregiver Considerations for Duchenne Muscular Dystrophy

A Summary of the DMD Care Considerations Working Group for Caregivers

PURPOSE

Because of advancements in the diagnosis and treatment of Duchenne muscular dystrophy (DMD) and how treatment is approached, the 2010 guidelines for the recommendations of care for patients who are affected by Duchenne have been updated.

With the support of the U.S. 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 has reviewed the literature and asked Duchenne experts for their opinions about the necessity and appropriateness of specific assessments and interventions in Duchenne.

DISCLAIMER

The following is a summary of the guidelines for the recommendations of care for Duchenne patients and is being disseminated for your information only. Caregivers should carefully assess all relevant subject matter regarding treatment options before making decisions related to each individual patient. Full article published in *The Lancet Neurology*, 2018.

8 ORIGINAL TOPICS OF CARE CONSIDERATIONS[1,2]

A multidisciplinary approach to treating Duchenne can greatly improve your loved one's long-term health and quality of life, including the following.

- Diagnosis
- Neuromuscular management
- Rehabilitation
- Psychosocial management
- Gastrointestinal and nutritional management
- Respiratory management
- Cardiac management
- Orthopedic and surgical management

A TREATMENT TEAM APPROACH TO GLUCOCORTICOIDS AND DUCHENNE[1]

To date, treating Duchenne with glucocorticoids remains the standard of care and has been confirmed by updated guidelines. Currently, deflazacort is the only FDA-approved glucocorticoid for the treatment of Duchenne.

3 NEWLY ADDED TOPICS IN THE MANAGEMENT OF DUCHENNE[3]

Addressing 3 additional specialties and how they contribute to the care of your loved one.

- Primary care and emergency management
- Endocrine management
- Transitions of care across the life span

ORIGINAL TOPICS OF CARE CONSIDERATION

A multidisciplinary approach to treating Duchenne can greatly improve your loved one's long-term health and quality of life.[1,2]

In the past, a multidisciplinary approach to managing Duchenne has greatly slowed the natural progression of the disease.[1] The new Care Considerations will introduce potential members of a Duchenne care team and focus on the caregiver's and patient's needs by addressing 3 main points:

- Providing more guidance to caregivers about early diagnosis and interventions
- Considering new therapies and how they impact long-term health
- Prolonging survival

TOPIC	KEY POINTS	CARE CONSIDERATIONS
DIAGNOSIS[1]	An early and timely diagnosis of Duchenne is crucial. It's usually the caregivers who are the first to notice subtle delays in their child's development and often bring it to the attention of their primary care physician (PCP). Unless there is a family history of Duchenne, an early diagnosis may be difficult due to the slow progression of the disease and lack of visible symptoms.	• The PCP can order a blood test to see if serum creatine kinase (CK) levels are elevated. Children with Duchenne typically have CK levels 10 to 100 times the normal level. • If increased CK levels are found, further testing and referral should be recommended. • Recently, certain tests have been recommended including: ○ Deletion and duplication testing, genetic sequencing, muscle biopsy to help with dystrophin protein detection, and carrier testing for female relatives of a male confirmed to have Duchenne.
NEUROMUSCULAR MANAGEMENT[1]	Children with Duchenne lack the dystrophin protein, which eventually leads to muscle weakness due to irreversible damage. Because of this, regular checkups with a neuromuscular specialist (NMS) familiar with Duchenne are important. The NMS is well aware of the disease progression and can help prepare for the next steps in treatment and recommend preventative therapies as early as possible. Around the world, glucocorticoids are used to treat a variety of conditions, including Duchenne.	• An NMS can help assess and keep track of your loved one's progress by monitoring strength, range of motion, timed tests, motor function scales, activities of daily living, and drug treatment for skeletal muscle. • Recent studies show that starting children on glucocorticoids earlier is more beneficial than waiting until there is a noticeable physical decline. • Long-term use of glucocorticoids has been shown to prolong ambulation and preservation of upper-limb and respiratory function, and help delay the need for invasive scoliosis surgery. Use of glucocorticoids, both prednisone and deflazacort, has been shown to improve muscle strength even after loss of ambulation.

TOPIC	KEY POINTS	CARE CONSIDERATIONS
REHABILITATION[1]	For those living with Duchenne, a variety of rehabilitation options is necessary to help combat well-known patterns of progressive muscle degeneration and weakness, postural compensations, and functional losses resulting from dystrophin deficiency. A rehabilitation team can include physicians, physical therapists, occupational therapists, speech-language pathologists, orthopedists, orthotists, and durable medical equipment providers.	• For continuous preventative care that will help to minimize the effect of Duchenne on your loved one's life, the rehabilitation team should take into consideration all of your loved one's goals and lifestyle choices. • Evaluations like the North Star Ambulatory Assessment (NSAA) and timed function tests are the cornerstones for testing motor function during the ambulatory phase and should be done every 6 months. • Direct physical, occupational, and speech therapy should be performed in an outpatient or school setting and continue for the entire course of the patient's life. • The CCWG has noted that increased use of standardized testing in children is key because of new therapies that are useful for younger children.
PSYCHOSOCIAL MANAGEMENT[1]	Living with Duchenne can be difficult. An all-inclusive approach should be adopted to help manage psychosocial issues as well as emotional quality of life, cognitive development, and factors that affect family functioning across home, school, and work. These issues may arise at any time, so it is important to remember that support is available and very much a part of the treatment journey. Don't hesitate to inform the treatment team of any issues that may arise.	• Support for your loved one spanning the course of the disease through his transitions of care may help to promote age-appropriate independence and social maturity. This is vital because it can encourage your loved one to set expectations about actively taking part in his daily activities and care. • Speaking with a mental health specialist is encouraged to help the entire family learn new strategies to cope with the disease process.
GASTROINTESTINAL AND NUTRITIONAL MANAGEMENT[1]	When caring for your loved one with Duchenne, gastrointestinal and nutritional issues should remain at the forefront of the caregiver's mind. These children may gain or lose weight, become constipated, and may find it difficult to swallow (dysphagia). To help with these issues that are sometimes caused by glucocorticoid use, your loved one may need to see a registered dietician nutritionist, physical therapist, speech language pathologist, and a gastroenterologist.	• Constipation and gastroesophageal reflux (acid coming from the stomach up into the throat, which causes heartburn) are the two most common gastrointestinal conditions seen in people living with Duchenne. It is important to discuss your loved one's bowel habits at every gastroenterologist visit. • A registered dietician can help to evaluate how many calories the patient may need per day by calculating the resting energy expenditure (REE) using measurements of height, age, and activity level. • Weakness in the muscles of the face, jaw, and head may lead to difficulty swallowing. This can exacerbate nutritional issues by reducing caloric intake and increase the risk of pneumonia due to potential aspiration (food or fluid entering the lungs).

TOPIC	KEY POINTS	CARE CONSIDERATIONS
RESPIRATORY OR PULMONARY MANAGEMENT[2]	Typically, those affected by Duchenne will develop breathing problems after they lose their ability to walk. This is why having a pulmonologist on the treatment team is crucial. As your loved one gets older, the muscles involved in coughing may become weak, leading to an increased risk of chest infection. As the disease continues to progress, the muscles that support breathing may also become weak, leading to problems with breathing while asleep. The neuromuscular specialist and pulmonologist should be consulted if waking symptoms of shallow sleep breathing are noticed because a sleep study may be necessary. Some of these symptoms include morning fatigue and headaches.	• If a weakening cough is noticed, there are devices available that will improve the quality of your loved one's cough as well as therapeutic exercises. • If oxygen levels are found to drop while asleep, your loved one may need to use bilevel positive airway pressure (BI-PAP) to help improve breathing and keep oxygen levels at a normal range. • The guidelines provide an update to the criteria on initiation of pulmonary interventions, which can help with better planning for respiratory care and early intervention where indicated throughout the stages of Duchenne.
CARDIAC MANAGEMENT[2]	Duchenne affects all the muscles of the body, including the heart (cardiomyopathy). Cardiomyopathy due to a lack of dystrophin causes a decrease in heart function and eventually heart failure. Varying degrees of heart failure can be managed with medications and regular visits to the cardiologist. It is important to involve a cardiologist as a member of the care team soon after a diagnosis is made and maintain regular visits to monitor heart function and medications as needed.	• Medication should be started at the first sign of cardiomyopathy or decreased heart function or by age 10 even if there are no obvious symptoms. • Any new or worsening cardiac symptoms should be immediately reported to your loved one's cardiologist for evaluation. • Potential side effects such as hypertension (high blood pressure) and hypercholesterolemia (high cholesterol) can be managed by adjusting your loved one's medication dose.
ORTHOPEDIC AND SURGICAL MANAGEMENT[2,3]	It is very important to add an orthopedist to your loved one's care team. When managing Duchenne, one of the main goals is to preserve walking and motor function as long as possible. Both of these basic body functions are affected by Duchenne and can ultimately lead to decreased bone health and osteoporosis. As a result of decreased muscle strength and impaired bone health, common pathologies that may arise include scoliosis, joint contracture, long bone fracture, and fat embolism syndrome, which is a medical emergency. A number of situations may arise either related or unrelated to Duchenne that may require surgery and general anesthesia. Those who are affected by Duchenne are at higher risk of complications, so all precautions should be considered, like using a hospital that is familiar with Duchenne.	• An orthopedist on your loved one's care team can help manage and delay the progressive effects of Duchenne. • Knowledge of vertebral fractures and initiating baseline testing with regular spine imaging for monitoring is a key update in the guidelines when discussing bone health monitoring. • When considering a surgical option, it is important to choose a hospital that has a knowledgeable staff and experience with Duchenne. • Fracture prevention for your loved one is key and potentially lifesaving. Important considerations for prevention include safety of the home environment, avoiding falls from a wheelchair or other mobility device, safety on slippery or uneven surfaces, and any potential home modifications that may be necessary.

A Treatment Team Approach to Glucocorticoids and Duchenne

To date, treating Duchenne with glucocorticoids remains the standard of care and has been confirmed by updated guidelines.

KEY POINTS TO CONSIDER WHEN GLUCOCORTICOIDS ARE USED

When to begin treatment	What is the appropriate dose for my loved one?	What about side effects?	How long does he need to take glucocorticoids?
Key findings			
Studies show the benefits of starting a steroid regimen before there is substantial physical decline[1]	When deciding on an appropriate dose, talk to the prescribing doctor about potential side effects and how they may affect your loved one[1]	Though glucocorticoids are the standard of care for Duchenne, it is important to remember that side effects may arise[1,2]	Glucocorticoids should be taken at the time of diagnosis and continued even after loss of ambulation to help preserve upper-limb function[1]
Expert recommendations			
Steroids should be introduced soon after diagnosis and should be viewed as the cornerstone for a multidisciplinary approach[1]	The recommended starting dose of deflazacort is 0.9 mg/kg/day, and 0.75 mg/kg/day for prednisone[1]	If side effects become unmanageable, discuss with the prescribing physician. Do not abruptly stop taking corticosteroids	Even older steroid-naive patients may benefit from initiation of a steroid regimen[1]

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.

CAUTION

Patients with Duchenne should not stop taking their medication abruptly. This may lead to an increased risk of adrenal insufficiency (suppression of the hypothalamic-pituitary-adrenal axis or HPA, which is considered a medical emergency). Because of this, caregivers and family members should be educated on the signs and symptoms of acute adrenal crisis.[1]

3 NEW TOPICS OF CONSIDERATION FOR DUCHENNE TREATMENT

Newly adopted specialties to enhance the treatment journey

In the ever-evolving world of treating Duchenne, it's important to stay current on topics that may aid in making his care a little easier. Here are 3 new topics that may contribute to the care of your loved one.

TOPIC	KEY POINTS	CARE CONSIDERATIONS
PRIMARY CARE AND EMERGENCY MANAGEMENT[3]	Most emergency medicine and primary care physicians lack experience when treating Duchenne. Here are several points you should bring to their attention:	• Have your loved one's emergency information on hand. This should include all results from recent evaluations and the contact information of his care team staff. • When possible, contact the PCP or NMS before you get to the hospital so they can call ahead to inform the hospital staff of potential hazards and advise them on best practices. • Make clear on arrival that your loved one has Duchenne. Provide a list of all medications and current cardiac or pulmonary issues. • Healthcare providers who are not connected to the Duchenne care team should be made aware of treatment goals, potential management strategies, quality of life issues, and, in some cases, life expectancy.
ENDOCRINE MANAGEMENT: GROWTH, PUBERTY, AND ADRENAL INSUFFICIENCY[3]	Complications with the endocrine system (the body's chemical messenger system consisting of hormones) may arise as a result of continued glucocorticoid use, including delayed puberty, altered growth, and adrenal insufficiency. It's imperative that both caregivers and patients alike are educated on the potential impact glucocorticoids may have on the patient.	Long-term glucocorticoid use, though necessary to treat Duchenne, has an impact on endocrine function. As a result, the NMS should refer your loved one to an endocrinologist, who can help by prescribing, when indicated: • Growth hormone therapy to help with short stature. • Testosterone therapy to support healthy bone growth and psychosocial/emotional development. • The endocrinologist can also advise on the risks of and help monitor for adrenal insufficiency, which is considered a medical emergency.
TRANSITION OF CARE ACROSS LIFE SPAN[3]	As your loved one grows, so will his healthcare needs. He will face a unique set of challenges that require special attention, especially as he enters adulthood at around 13 to 14 years. Full coordination and communication among the care team, caregiver, and young man is vital for transition planning.	• Develop a plan that includes potential new services and providers, and how it will be paid for. • Develop transition plans based on the needs and wants of your loved one and family. • Research legal and financial planning to cover unexpected medical, legal, and end-of-life decision-making needs. • Consider a care coordinator or social worker to act as a central point of contact for the patient and caregiver and the rest of the treatment team, and ensure the facilitation of adolescent healthcare with referral to adult providers as well as the transition of medical records.

YOUR DUCHENNE TREATMENT TEAM

Contact Information

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Address: _____

Telephone: _____

Email: _____

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KEY TAKEAWAY SUMMARY

A CAREGIVER'S GUIDE TO NAVIGATING DUCHENNE MUSCULAR DYSTROPHY

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