

Individuals with Duchenne Muscular Dystrophy (DMD) are often cared for in multidisciplinary clinics, with pediatric neuromuscular specialists serving as their primary DMD care provider. General pediatricians as well as first responders, urgent care providers, and emergency department personnel may need to provide acute care to individuals with DMD, though they may be unfamiliar with the disease.

This resource was designed to provide individuals with DMD and their families with information to help them advocate for the highest quality of emergency care. When you or a loved one are faced with a medical emergency, stress and worry can get in the way of clear thinking. We advise you to read this guide *before* you need it, and that you keep a copy handy in the event of an emergency by bookmarking it on your smartphone or printing a version to keep in your vehicle.

The CureDuchenne Cares team ([cares@cureduchenne.org](mailto:cares@cureduchenne.org)) is available to answer any questions or concerns you may have about the information we have provided below.

### **Medical Alert Bracelets**

CureDuchenne knows the stress that individuals and families face during medical emergencies, particularly given the specialty care required. That's why we've partnered with MyID to provide free, [next-generation medical alert bracelets](#) to individuals with DMD. When scanned by a smart phone or other device, MyID bracelets provide emergency contact information as well as Duchenne-specific care considerations for first responders.

### **Emergency Prevention**

One key way for individuals and families to proactively reduce the risk of emergency care is to work with their primary care providers or neuromuscular specialists to keep up-to-date on routine vaccinations.

- Not all vaccinations are 100% effective, but those that aren't reduce the risk of severe illness.
- Families should be aware that vaccinations which contain live viruses are not recommended for individuals receiving certain doses of steroids.
- The annual flu vaccine is recommended (unless contraindicated due to allergy); consult with a primary care provider or neuromuscular specialist prior to receiving a nasal spray version.
- For more information on the US recommendations for childhood vaccinations, visit the Center for Disease Control (CDC) website at <https://www.cdc.gov/vaccines/schedules/easy-to-read/child-easyread.html>

In addition to keeping up to date on vaccinations, individuals and families can help prevent emergencies through the following techniques:

- Always wear a seatbelt or custom restraint when in a wheelchair and vehicle
- Eliminate distracted driving
- Install home smoke and carbon monoxide detectors and change batteries twice annually (tip: change them when you change your clocks!)

## Emergency Preparedness

In addition to taking the measures above to prevent emergencies, families find reassurance in being prepared.

- Ensure you have emergency evacuation plans for home and school, and consider plans for power outages if you rely on home medical equipment.
- Have a plan for weather-related emergencies, including an evacuation route.
- Keep the 24-hour on-call number for your neuromuscular specialist or primary care provider programmed in your phone and notify them as soon as you can—always call 911 first in the event of a true medical emergency.
- Know what tests (e.g., electrocardiogram, also referred to as ECG or EKG) or lab results (e.g., liver function tests, also referred to as LFTs or transaminases; creatine kinase, CK) are typically abnormal so you can inform emergency room staff.

Additionally, gather the following information so it's ready to bring with you to the Emergency Room (ER):

- A copy of the patient's medication list, noting the day and time of last steroid dose (if applicable)
- A list of the patient's allergies
- Contact information for the patient's primary care and/or neuromuscular provider
- Most recent multidisciplinary note (with information such as test results)
- Cough assist device, if applicable (label this with name and contact information in the event it is lost)
- Noninvasive mechanical ventilation device, if applicable (label this with name and contact information in the event it is lost)
- Clinical trial information, if applicable
- A copy of this guide for your reference
- A copy of the CureDuchenne guide for Emergency Care Providers, to provide to ER staff

Some clinics and doctor's offices use electronic medical records that allow for patient/parent access. If you have the ability to access electronic copies of records, create an account to access the records before you need it—in some cases, access can be delayed by a few business days.

Additionally, if appropriate, we encourage families to discuss advanced directives. Though these conversations are difficult, knowing an individual's wishes in the event that he is unable to advocate for himself will empower family members to advocate on his behalf.

## **Acute Respiratory Distress**

Parents and providers should know that individuals in acute respiratory distress may not appear to be having a difficult time breathing. This is because DMD-associated muscle weakness can result in the inability to display classic signs of respiratory distress, such as gasping for air. If an individual with DMD complains of difficulty breathing, you should seek emergency care. You may also wish to purchase a pulse oximeter—a relatively inexpensive device which measures blood oxygen levels through a light sensor applied to a fingertip. If you decide to purchase a “pulse ox” monitor, talk to your neuromuscular team about how to interpret the readings and what readings require emergency evaluation.

Understanding acute respiratory emergencies requires an understanding of DMD physiology. As DMD progresses, decreased strength of the muscles in the chest wall make it harder for individuals to take deep breaths in (inhalation) and out (exhalation); this may be referred to as “poor ventilation.” Providing supplemental oxygen does not help lungs inflate and deflate effectively, and therefore doesn’t help with poor ventilation. Additionally, the decreased strength also impacts the ability to cough forcefully which can result in difficulty moving respiratory secretions or clearing food or drink lodged in their airway. Poor ventilation, difficulty swallowing and weak cough may increase the risk of acute issues such as infection, aspiration, and airway blockage.

In general, the typical provider response to a patient’s complaint of respiratory distress is to provide supplemental oxygen. In individuals with DMD, this often does not help treat the cause of the respiratory distress and, in fact, may cause greater harm by decreasing the drive to breathe. Individuals with DMD in acute respiratory distress often need ventilation support (frequently with noninvasive ventilation) along with supplemental oxygen. Cough assistance devices and techniques should also be employed, when appropriate. If being seen in an ER, ask for a respiratory therapy consultation, though this may not always be possible. Always bring your cough assistance device to the ER with you.

Individuals with DMD who have respiratory and chest wall muscle weakness are at an increased risk for pneumonia; this risk is increased for those on daily steroids, which can weaken the immune system. A chest Xray can help diagnose the pneumonia, though you should discuss the use of “empiric antibiotics,” those given before a bacterial infection is confirmed, with the consulting doctor. If being seen in an ER, “portable” Xrays can routinely be done in hospital rooms rather than radiology departments and allow for patients to remain on ventilation and monitoring equipment. Discuss this option with the ER provider.

Though it rarely develops rapidly, shortness of breath can be the result of worsening heart failure; this is particularly true of shortness of breath that worsens with physical activity or lying flat. Having access to the most recent results from an echocardiogram may be helpful for the treating team’s assessment. Diuretic medications (often referred to as “water pills”), given by mouth or IV, are safe for most individuals with DMD and can pull extra fluid from the body and may help relieve the shortness of breath.

Very rarely, acute respiratory distress occurs as a result of a fat embolism, a condition where a fat globule blocks a blood vessel. Sudden onset of rapid breathing or difficulty breathing with or without confusion, particularly if the individual has had a recent fracture or trauma of an arm or leg, should prompt an urgent evaluation in an ER. These symptoms mimic pneumonia and may be misdiagnosed as a result; relay information about recent fractures and the increased risk of fat embolism syndrome (FES) in individuals with DMD to the ER staff.

Should a patient require invasive ventilation, intubation (the process of inserting a breathing tube) can be complicated by limited jaw muscle motion or by an enlarged tongue. Ask that an anesthesiologist be present to assist, if possible. If a breathing tube (called an endotracheal or ET tube) is inserted, preparation for it to be removed should be taken. Assisted cough devices and non-invasive ventilation (such as CPAP and BiPAP) may be helpful in weaning a patient from mechanical ventilation. Additionally, make sure that supplemental oxygen is given concurrently with this non-invasive ventilation.

### **Altered Consciousness**

Those who arrive in an ER with altered consciousness are less able to advocate for themselves, increasing the importance of medical alert bracelets, wallet cards and advanced directives.

Altered consciousness can be due to many things, including a fall or other accident with head injury, decreased blood oxygen from difficulty breathing, fat embolism associated with arm or leg fracture, or adrenal insufficiency due to sudden stoppage of steroids.

No special instructions exist for individuals with Duchenne who experience a concussion. For other causes, see information elsewhere in this guide.

### **Anesthesia**

Local and intravenous (IV) anesthesia are generally well tolerated. **Make sure the emergency and/or anesthesia team know the patient has Duchenne muscular dystrophy. Drugs that cause neuromuscular blockage, such as succinylcholine (sometimes referred to as “succs”), are often given during intubation but are strongly contraindicated in DMD patients.** Consider documenting this as an allergy in the patient’s medical record to reduce the likelihood of administration.

Caution should also be used with inhaled anesthetics.

Because cardiomyopathy affects all individuals with DMD, we encourage you to talk to providers about cardiac anesthesia precautions prior to sedation.

## **Chest Pain**

All individuals with DMD have associated cardiomyopathy, a condition that makes it harder for the heart to pump blood throughout the body. As a result, electrocardiograms (also referred to as EKGs or ECGs)—tests that record the electric signals—may not be “normal,” even when an individual with DMD is not experiencing a medical emergency. It’s important to relay this information to ER providers and, if feasible, to connect them with the individual’s primary care provider, cardiologist, or neuromuscular specialist to provide specific information.

The ER team may also obtain an echocardiogram, or ultrasound of the heart. Due to cardiomyopathy, they may be concerned about the individual’s ejection fraction (EF), a measure of how well the heart is pumping. This is often reduced in individuals with DMD, particularly those diagnosed with heart failure; if you know the most recent EF (a percentage generally less than 65), relay this to the ER staff.

## **Steroid Dosing**

**Steroids such as prednisone or deflazacort should never be stopped suddenly.** Instead, steroids should be weaned gradually (called “steroid tapering”) over weeks to months under the supervision of a neuromuscular specialist or endocrinologist.

Additionally, individuals experiencing a medical crisis such as severe illness, trauma, or surgery may require “stress dosing” of steroids (even if this event occurs during tapering)—a higher than normal dose required because the adrenal glands may not be able to produce adequate levels of cortisol, a hormone normally produced under physical stress which, among other things, helps regulate blood pressure and boost energy levels. Stress doses of steroids will be calculated by the individual’s care team based on his height and weight. Point the individual’s emergency care team to the [PJ Nicholoff Steroid Protocol for Duchenne and Becker Muscular Dystrophy and Adrenal Suppression](#), a comprehensive peer-reviewed article that provides further details.

## **Vomiting**

In the event that an individual experiences vomiting and is unable to keep down fluids and/or steroid doses, notify your primary care provider or neuromuscular specialist. The individual may need to be hospitalized in order to receive IV fluids or IV steroids.

Difficulty swallowing, as well as ineffective cough, put individuals with DMD at increased risk of accidentally inhaling vomit into their windpipe or lungs; this is known as *pulmonary aspiration*. Ensure that an individual who is actively vomiting and unable to sit independently is rolled onto their side, cleaning up the vomit quickly to reduce the risk of aspiration.