

Dear Respected Emergency Care Providers:

This person has a condition called Duchenne muscular dystrophy (DMD), a degenerative neuromuscular disease which results in loss of ambulation, as well as cardiac and respiratory deterioration with age. This abbreviated guide is intended to help you provide emergency care for individuals with DMD.

Like with many other chronic illnesses, older individuals and parents often have exceptional insight into the condition, and we encourage you to engage with individuals and families as feasible.

General Treatment Considerations

Caution with:

- Oxygen therapy in hypoxic individuals—need to monitor for the presence of hypercapnia
- IV fluids due to frequency of heart failure in individuals with DMD age 10 years and older
- Succinylcholine and inhaled anesthesia are absolutely contraindicated; consider listing as drug allergies

Common abnormal test results:

- CK, AST/SGOT, ALT/SGPT may all be elevated due to normal course of underlying DMD
- Decreased LVEF
- Decreased FVC%

Common medical equipment:

- Assess for trach, G-tube, ICD, VAD/port, non-invasive ventilatory support
- Individuals should be encouraged to bring personal cough assist to ER as soon as possible

Overview of Key Issues Related to Duchenne Muscular Dystrophy Emergency Care

Initial Assessment:

- Advance directives, determine whether there are restrictions on resuscitation
- Scan the individual's MyID medical bracelet (if applicable) or ask for the individual's emergency card and baseline test results, including ECG results
- Obtain a brief history with a focus on baseline respiratory and cardiac status, including the use of relevant devices and medications (chronic steroid therapy, gene therapy or enrolled in drug trial)
- Contact the individual's neuromuscular specialist and/or clinical trial contacts

Breathing problems (most common):

- **Individuals may not appear distressed due to inability to manifest increased work of breathing; individual's perception of symptoms should be taken seriously.** The importance of initiating noninvasive ventilatory (NIV) support and assisted coughing early cannot be overstated

- Consider pneumonia or atelectasis, untreated chronic or acute hypoventilation, pulmonary/fat embolism (in presence of recent extremity fracture), worsening heart failure, aspiration, or airway obstruction
- Intubating individuals with DMD is usually difficult and likely to be avoided using NIV support. If intubation is deemed necessary, it should be considered a high-risk procedure and may be complicated by macroglossia and limited jaw motion
- Ask about respiratory symptoms and home equipment
- Obtain portable CXR where possible, noting that fat embolism syndrome (FES) may present as pneumonia
- Monitor SpO₂ levels; even mild hypoxemia (SpO₂ <95% in room air) is a concern; do a blood gas analysis if necessary
- Treat with noninvasive ventilation and frequent application of a cough assistance device (or manual assisted coughing if device is unavailable); use the individual's home equipment when available

Cardiac problems:

- Ask about cardiac history, function and symptoms; note that all individuals with DMD have associated cardiomyopathy. Individuals are followed with routine Echo (LVEF declines with age) and ECG.
- Monitor heart rate and rhythm. Obtain an ECG (this is typically abnormal and Q waves might be expected)
- Consider worsening cardiomyopathy, congestive heart failure, and arrhythmias
- Diuretic administration is acceptable
- Obtain early consultation with a cardiologist

Endocrine problems:

- Most individuals are treated with deflazacort or prednisone; determine whether stress dosing is necessary
- For critical adrenal insufficiency, administer intravenous/intramuscular hydrocortisone (50mg for children <2yr; 100mg for children >2yr and adults)
- In less critical situations, consult the PJ Nicholoff Steroid Protocol for Duchenne (available on PubMed)
- Obtain early consultation with an endocrinologist

Orthopedic problems:

- Assess for long-bone or vertebral fractures as indicated
- Consider FES if individual has dyspnea or altered mental status; can be associated with seizure activity. FES may appear as pneumonia on CXR

Disposition after discharge from emergency care:

- Be aware that most individuals will need hospital admission (e.g., to initiate or intensify respiratory or cardiac therapy, or to manage fractures)
- Initiate emergency transport to a center specializing in the care of individuals with DMD, in cooperation with the individual's neuromuscular specialist