



:: Duchenne muscular dystrophy



- *This document is a translation of the French recommendations drafted by Dr. David Orlikowski and Dr Gilles Bagou reviewed and published by Orphanet in 2009.*
- *Some of the procedures mentioned, particularly drug treatments, may not be validated in the country where you practice.*

Synonyms:

Duchenne de Boulogne muscular dystrophy, DMD

Definition:

Definition: **Slowly-advancing** recessive X-linked **myopathy** starting during childhood and **ending in loss of motor function** (loss of mobility at around 10 years of age) and associated with **problems - cardiac** (heart failure) and **respiratory** (need for non-invasive ventilation, which may well become invasive in the long-term). Survival beyond 30 years of age with appropriate cardiorespiratory management.

Further information:

[See the Orphanet abstract](#)

Pre-hospital emergency care recommendations

Call for a patient suffering from Duchenne muscular dystrophy

Synonyms

- ▶ Duchenne disease, Duchenne de Boulogne muscular dystrophy , DMD

Mechanisms

- ▶ Recessive myopathy linked to the X chromosome, affecting smooth or striated muscle fibres, advancing slowly and ending in loss of motor function (loss of mobility at around 10 years of age), heart failure and respiratory failure

Specific risks in emergency situations

- ▶ heart failure (dilated cardiomyopathy) but with few complaints due to the peripheral impact upon motor function
- ▶ acute respiratory failure, exacerbated by difficulty with coughing
- ▶ metabolic: hyponatraemic dehydration, particularly on exertion or when it is very hot
- ▶ difficult intubation: orthopaedic deformity (scoliosis, etc.), macroglossia, small mouth aperture

Commonly used long-term treatments

- ▶ treatment for heart failure
- ▶ physiotherapy
- ▶ non-invasive ventilation
- ▶ tracheotomy

Complications

- be wary of respiratory depressants
- blockage of the tracheotomy cannula
- risk of difficult intubation (need to use endoscopy)
- frequent ECG abnormalities: rhythm or conduction disturbances (PAFA, severe AVB), PR shortening, lateral Q-wave, ST elevation

Specific pre-hospitalisation medical care

- ▶ do not lay an orthopnoeic patient down flat
- ▶ ventilation, preferably non-invasive
- ▶ succinylcholine is contraindicated (hyperkalaemia)
- ▶ intubation under propofol sedation
- ▶ frequent chronic pain
- ▶ difficulty with mobilisation and positioning (orthopaedic deformity)
- ▶ admission to Intensive Care or Resuscitation

Further information

- ▶ Action Duchenne: www.actionduchenne.org
- ▶ DFSG - Duchenne Family Support Group: dfsg.org.uk
- ▶ MDC - Muscular Dystrophy Campaign: www.muscular-dystrophy.org
- ▶ Please visit www.orpha.net and type the name of the disease → in the summary page click on “Expert centres” on the right tab → select “United Kingdom” in the “Country” field in the Expert centres page.

Recommendations for hospital emergency departments

Emergency issues

- ▶ Onset of **episodes of respiratory or cardiac decompensation**: patients exposed to the risk of acute respiratory decompensation even with a straightforward cold or rhinopharyngitis.
- ▶ Respiratory failure exacerbated by **difficulty with coughing**, particularly where this is associated with swallowing problems.
- ▶ Exposure to the **risk of invasive ventilation**, of **difficult intubation** and, thus, of complications in patients with major orthopaedic deformities, restricted mouth aperture or macroglossia.
- ▶ **Complications, sometimes serious** may be observed in **patients who have undergone tracheotomy**, in the form of cannula obstruction or heavy tracheal bleeding.
- ▶ **There are no specific features associated with DMD cardiopathy**, which develops in the form of **dilated cardiomyopathy (DCM)**: few complaints made in terms of cardiac function on account of low levels of motor activity, even in advanced cases of DCM. Peripheral oedema syndrome will be the primary indicator of heart failure. As with other DCM-type disorders, rhythm or conduction disturbances may develop.
- ▶ In themselves, **swallowing problems** are **rarely problematic in emergency practice** but they may result in aspiration, the management of which will be the same as for bronchial obstruction. Sometimes, where swallowing is difficult or even impossible, the patient may need to be admitted for enteral or even parenteral nutrition.

Emergency recommendations

1. Emergency diagnostics:

- ▶ Assess severity: assessment criteria will be based upon respiratory and haemodynamic tolerance
 - **Respiratory warning signs** include:
 - Dyspnoea
 - Orthopnoea
 - Recession
 - Paradoxical respiration
 - Bronchial obstruction
 - Desaturation in ambient air or the need for oxygen therapy
 - Patient already ventilated: increase in ventilation time
 - Patient with tracheotomy: intra-tracheal aspiration becomes impossible or heavy **bleeding**
 - **Warning signs for poor haemodynamic tolerance** are:
 - Hypotension (it is not always easy to recognise this since previous BP readings are often low)
 - Poor cardiac output: mental confusion, cardiac liver
- ▶ Emergency investigations:
 - **Arterial blood gases** (sometimes capillary in some patients from whom it is difficult to obtain a specimen): these will allow the patient to be **screened first and foremost for alveolar hypoventilation** (with or without respiratory acidosis) - **in practice PaCO₂ > 45 mmHg**. They will also allow screening for hypoxaemia.
 - **Chest X-ray**: screening for **causes - parenchymatous** (lung disease, atelectasis, pulmonary oedema, etc.) or **pleural** (pneumothorax, pleural effusion, etc.)
 - **ECG**: this will often exhibit the following specific features:
 - Q-waves in lateral territory (I, aVL and V₅, V₆) and, sometimes, inferior.
 - PR may be short.
 - Rhythm and conduction disturbances are fairly frequent, with episodes of PAFA, sinus dysfunction or a severe atrioventricular block.
 - There may be repolarisation problems, with ST elevation.
 - QRS widening is often seen.

2. Immediate treatment:

- ▶ Symptomatic:
 - **Oxygen therapy** in cases of desaturation or hypoxaemia.
 - **Diuretics** in the event of pulmonary oedema.
 - **Mechanical ventilation, preferably non-invasive** (non-ventilated or already mask-ventilated patient) if clinical signs or blood gas results point to respiratory failure.
 - **Bronchial clearance** either manual (physio) or using a mechanical technique (assisted cough).
 - **Endotracheal intubation and invasive ventilation**: problems with level of consciousness, shock status, respiratory or cardiac arrest are strict indications.
 - **Patients who have undergone tracheotomy**: change the cannula in cases of obstruction or use a balloon cannula for alveolar hypoventilation.
 - **Rapid ENT or Respiratory Disorders assessment of tracheal bleed.**
 - In cases of bronchial obstruction **other than established lung disease, the benefit of systemic antibiotic treatment has not been demonstrated.**

Orientation

- ▶ Where?: Resuscitation or Respiratory Intensive Care Unit with good knowledge of the pathology in cases of respiratory failure. Cardiac ICU in cases of pure cardiac failure (non-ventilated patients).
- ▶ When?: In emergencies.
- ▶ How?: **Fully-equipped ambulance** since the patient is problematic (**venous access, risky intubation**). Care with patient positioning: **do not lay an orthopnoeic patient down flat (risk of respiratory arrest)**. Care when mobilising (**risk of fracture**).

Drug Interactions

- ▶ **Bronchial fluidifying agents are strictly contraindicated** in cases of obstruction.
- ▶ **Sedatives must be avoided** due to the respiratory risk.
- ▶ **Morphine-type agents are to be handled with care** due to the respiratory risk and the risk of intestinal occlusion.
- ▶ **Risk of urine retention with anticholinergic medicinal products.**

Anaesthesia

- ▶ Induction: satisfactory control of the airways (frequent anatomical difficulties), avoid the risk of aspiration (delayed gastric emptying) and cardiovascular complications
 - **Curare-type depolarising agents** (succinylcholine) **are contraindicated**: risk of hyperkalaemia.
 - **The technique of choice appears to be intubation, using a flexible endoscope, under propofol sedation.**
- ▶ Maintenance of anaesthesia
 - **Halogenated agents are contraindicated (malignant hyperthermia).**
 - **Intravenous hypnotic agents of the propofol type appear to be the products of choice** in conjunction with short-acting morphine-type substances such as remifentanyl and, where necessary, combined with topical-regional anaesthesia techniques to **minimise, as far as possible, the risk of post-operative respiratory depression.**
 - **Anaesthetic respiratory and cardiovascular monitoring** is routine and needs to be tailored to the surgical procedure and to the extent of the patient's heart problems, along with **temperature monitoring** with **prevention of hypothermia** and **detection of malignant hyperthermia.**

Preventive measures

- ▶ Early respiratory physiotherapy.
- ▶ Vaccinations: anti-flu and anti-pneumococcal.

Additional therapeutic measures and hospitalisation

- ▶ **Allow the patient's next-of-kin to accompany him**, even in an emergency. The people around the patient are generally those who are most familiar with the pathology and with the risks as it develops.
- ▶ Bear in mind the difficulty with **mobilisation** and **positioning** (frequent posture changes), pain (suitable mattress).
- ▶ Often, feeding can only be carried out in a seated position; if the patient has to be laid flat for any length of time, pass a nasogastric tube (risk of fasting ketosis).
- ▶ Do not forget **mobilisation physiotherapy** (retractions)

Organ donation

In theory, liver and kidney donation are not contraindicated, but heart donation, on the other hand, is contraindicated (tropism-type disease affecting the myocardium).

Emergency telephone numbers

Please visit www.orpha.net and type the name of the disease → in the summary page click on “Expert centres” on the right tab → select “United Kingdom” in the “Country” field in the Expert centres page.

Documentary resources

- ▶ Prise en charge de l'insuffisance respiratoire aiguë d'origine neuromusculaire [Management of acute respiratory failure of neuromuscular origin]. D. Orlikowski , H. Prigent , T. Sharshar, J.-C. Raphael. Journal Européen des Urgences 21 (2008) 94–98.
- ▶ Intubation endotrachéale des patients atteints d'une maladie neuromusculaire [Endotracheal intubation of patients suffering from a neuromuscular disorder]. A. Polito, D. Orlikowski. Journal Européen des Urgences 21 (2008) 102–104.
- ▶ Quelle prise en charge anesthésique pour les patients atteints de pathologies musculaires? [How should anaesthesia be managed in patients suffering from muscular disorders?] Najla Akrouf, Andrea Polito, Djillali Annane and David Orlikowski. La revue du praticien. December 2008 (58) 20. 2265-2270.

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