:: Steinert myotonic dystrophy

- *This document is a translation of the French recommendations drafted by Professor Bruno Eymard, Dr. David Orlikowski, Dr. Karim Wahbi and the team belonging to Professor Denis Duboc, reviewed and published by Orphanet in 2010.*
- *Some of the procedures mentioned, particularly drug treatments, may not be validated in the country where you practice.*

Synonyms:

Type I myotonic dystrophy, MD1, Steinert's disease

Definition:

Steinert myotonic dystrophy (MD) is the commonest form of muscular dystrophy in adults (affecting between 1/8 000 and 1/20 000 people in Europe) and exhibiting autosomal dominant transmission. It is characterised by a wasting muscle disorder affecting primarily the distal, axial, facial, pharyngeal and respiratory muscles, causing myotonia in the hands and affecting multiple systems, with cataracts, cardiac conduction blockade and arrhythmia, diabetes and drowsiness.

There are **four important characteristics** that need to be borne in mind: 1) signs do not necessarily manifest themselves in a set pattern (heart problems may develop in apparent isolation, with no muscle deficiency); 2) the contrast between the severity of the heart or respiratory disorder and the low level, if any, of symptoms; 3) the number of different mechanisms involved: cardiac disorders - conduction and/or rhythm disturbances and, less commonly, ventricular dysfunction; respiratory disorders - weakness in the respiratory muscles, aspiration, central hypoventilation, pulmonary embolism; 4) a tendency in patients towards a degree of apathy, with underestimation of the problems.

Often, blood gases are abnormal (hypoxaemia, hypercapnia) although there will not, necessarily, be a correlation between these 2 parameters.

The severity of the disorder arises as a result of complications: 1) **cardiac**: risk of sudden death, accounting for 20 to 30% of deaths; conduction and/or rhythm disturbances; risk of embolic accident due to persistent AF arrhythmia; 2) **respiratory**: implicated in the majority of deaths (lung disease, alveolar hypoventilation) made worse by aspiration and deficient respiratory muscle function. On average, the lifespan will be reduced by about ten years. There are **paediatric forms**, initially either **neonatal** (congenital Steinert) - which is very severe - with joint contractures, severe hypotonia, respiratory failure, frequent death, or **infantile** (infantile Steinert), manifesting itself through motor delay, learning difficulties leading to schooling problems and a varying reduced IQ.

Further information:

See the Orphanet abstract

Pre-hospital emergency care recommendations
Call for a patient suffering from Steinert myotonic dystrophy

Synonyms

- type 1 myotonic dystrophy, MD1, Steinert disease

Mechanisms

- dominant genetic myopathy, causing a wasting muscle disorder affecting multiple systems

Specific risks in emergency situations

- respiratory distress due to lung disease, aspiration, pulmonary embolism or central hypoventilation
- respiratory decompensation after a common or post-operative infection
- conduction or rhythm problems, alternating between bradycardia and tachycardia
- arterial embolism complicating atrial fibrillation
- sudden death, particularly during physical exertion
- ENT haemorrhage complicating tracheotomy

Commonly used long-term treatments

- respiratory physiotherapy

Complications

- be wary of underestimation by patients of the severity of their symptom(s) or of their illness
- be wary of any manifestation of sensation of faintness, syncope, palpitations
- a cardiac form may develop in isolation (with no muscle disorder)
- paediatric forms
- sudden death with a pacemaker *in situ*

Specific pre-hospitalisation medical care

- ask for any patient-held information on current treatment or precautions required
- routine ECG with continuous monitoring
- be wary of the respiratory risk associated with morphine-type medicines and sedatives
- contraindications: depolarising curare-type medicines; class I anti-arrhythmic agents, amiodarone, beta-blockers, anticholinergic medicines, bronchial fluidifying agents (in the event of obstruction)
- give preference to non-invasive ventilation and avoid intubation (at-risk)
- be wary of high-flow oxygen in patients, whether ventilated or not
- patients who have undergone tracheotomy: cannula change or fitting of a balloon cannula
- orthopnoeic patients: do not lay down flat (risk of respiratory arrest)
- admit to Cardiology Resuscitation or Intensive Care for fitting of a pacemaker

Further information

- MDSG - Myotonic Dystrophy Support Group: www.myotonicdystrophysupportgroup.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

Ask for any patient-held information on current treatment or precautions required.

- **Onset of acute respiratory decompensation:**
  - common infectious events (during colds or episodes of rhinopharyngitis)
  - certain medical-surgical situations (post-operative period or during anaesthesia)
- **Onset of cardiac conduction or rhythm disturbances**
- Onset of gastrointestinal problems:
  - pain $^{+}$/- vomiting due to biliary lithiasis
  - pseudo-surgical abdomen: gastrointestinal pseudo-occlusion treated medically

Emergency recommendations

A. Onset of respiratory decompensation

The major problem here is the presence of **alveolar hypoventilation**, sometimes an acute flare-up of a chronic condition, or revealed by a factor such as bronchial obstruction due to weakness in the cough muscles. This may require **invasive ventilation**, with a risk of **difficult, if not impossible, weaning** and, hence, of tracheotomy. Subsequently, this may create difficult social management problems (solitude and frequent inability to socialise).

- **Emergency diagnostics:**
  - Assess severity: assessment criteria are based on **respiratory tolerance**
  - Severity criteria:
    - dyspnoea
    - orthopnoea
    - recession
    - paradoxical respiration
    - bronchial obstruction
    - desaturation in ambient air or need for oxygen therapy
    - patient already ventilated: increase in ventilation time
    - patients who have undergone tracheotomy: intra-tracheal aspiration is impossible if there is heavy bleeding
  - Emergency investigations:
    - **arterial blood gases** (sometimes capillary in some patients from whom it is difficult to obtain a specimen) to identify:
      - **alveolar hypoventilation** (with or without respiratory acidosis): **$PaCO_2$>45 mmHg**
      - **hypoxaemia**
    - **chest X-ray**
      - screening for causes: parenchymatous (lung disease, atelectasis, pulmonary oedema, etc.) or pleural (pneumothorax, pleural effusion, etc.)

- **Immediate treatment:**

  Treatment is primarily symptomatic.
  - **Oxygen therapy** in cases of desaturation (hypoxaemia). Pay attention to high-flow oxygen in patients, whether or not they are being ventilated
  - If there are clinical signs of **respiratory failure** present, or if blood gas results so suggest, **mechanical ventilation, preferably NIV**

- **Clearance of the bronchial obstruction**, either manual (physio) or using a mechanical technique (assisted cough)
- Strict indications for endotracheal intubation and invasive ventilation:
  - problems with level of consciousness
  - shock status
  - respiratory arrest
  - cardiac arrest
- Patients who have undergone tracheotomy: change the cannula in cases of obstruction or use a balloon cannula if there is alveolar hypoventilation
- **In the event of tracheal bleeding**, rapid ENT or Respiratory Disorders assessment

B. Onset of cardiac conduction or rhythm disturbances

This happens far more frequently than ventricular dysfunction (in the context of dilated cardiopathy): there is no correlation between the heart problem and limb weakness which means that the heart problem may reveal the disease.

- **Conduction disturbances** (atrioventricular $^{+}$/- intraventricular) are **generally proportional to the length of the period of illness**
- **Rhythm disturbances** (atrial fibrillation, ventricular rhythm disturbances) **may develop during adolescence,** exacerbated by physical exertion
- The **risk of sudden death** is higher where there is a combination of clear AV (PR>200 ms) and ventricular (QRS>100ms) conduction disturbances
- **There have been reports of sudden death in patients** who have had a **pacemaker** implanted to deal with conduction disturbances; one explanation may be onset of ventricular rhythm disturbances or pulmonary embolism, but sometimes the cause remains unexplained

Emergency diagnostics:

- Assess severity: assessment criteria are based upon **haemodynamics**
- Severity criteria:
  - **syncope,faintness, palpitations**
  - **bradycardia <40, tachycardia >120, arrhythmia**
  - factors that indicate **poor haemodynamic tolerance**
    - **hypotension** (it is not always easy to recognise this since **previous BP readings** are often low)
    - low cardiac output: mental confusion, cardiac liver
- Emergency investigations:
  - The **ECG** will often exhibit:
    - high-grade paroxysmal **conduction disturbances** (pauses, atrioventricular block (AVB): AVB II or AVB III - full AVB)
    - or **paroxysmal rhythm disturbances**, mainly in patients with a history of syncope or conduction disturbances, *e.g.* AVB I or bundle-branch block

Immediate treatment:

- Symptomatic measures
- There is a high risk of **alternating** episodes of bradycardia and tachycardia:
  - **extreme care** must be taken when using **anti-arrhythmic agents** (amiodarone, beta-blockers) **in cases of tachycardia** since this may be associated with the onset of severe bradycardia
  - **class I anti-arrhythmic agents are formally contraindicated**, particularly via the intravenous route

Orientation

- Where?:
  - Resuscitation or Respiratory Intensive Care with specialist knowledge of the pathology in cases of respiratory failure.
  - Cardiac ICU in cases of pure heart-failure (non-ventilated patients) with the option of rapid fitting of a pacemaker in cases of high-grade conduction disturbances
- When?: in an emergency
- 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)**

Drug interactions

- **Bronchial fluidifying agents** strictly **contraindicated** in cases of obstruction
- **Avoid sedatives** 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
- **Class I anti-arrhythmic agents are formally contraindicated**
- **Medicines that induce bradycardia** (amiodarone, beta-blockers) must be used with **extreme care**
- **Risk of urine retention with anticholinergic medicinal products**

Precautions for anaesthesia

- Pre-operative **cardiac** assessment to exclude the presence of associated conduction disturbances that may cause decompensation during administration of anti-arrhythmic agents. Onset of paroxysmal rhythm disturbances during or after surgery is not uncommon.
- 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) **contraindicated: risk of hyperkalaemia**
  - **The technique of choice appears to be intubation under propofol sedation** (endoscope in cases of facial dysmorphia)
- Maintenance of anaesthesia:
  - **Halogenated agents contraindicated (malignant hyperthermia)**
  - **Intravenous hypnotic agents of the propofol type** appear to be the products **of choice**, combined with **short-acting morphine-type substances** such as remifentanil 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**
  - **Temperature monitoring** with prevention of hypothermia and detection of malignant hyperthermia

Preventive measures

- Be wary of respiratory failure (frequent hypoxaemia): consider the value of NIV combined, where necessary, with oxygen (risk of exacerbation of hypercapnia and hypercapnia that is, paradoxically, well tolerated)
- Early respiratory and motor function physiotherapy
- Avoid sedative medicines and hypnotic agents (frequent sleep problems)
- Prevention of thrombo-embolic complications (frequent thrombosis)
- "Passive" and relatively undemanding patients who are not, therefore, very symptomatic
- Social aspects to be anticipated

Additional therapeutic measures and hospitalisation

- Where the **patient's next of kin** are present, they will be a key **component in coordinating any care** that is given, including emergency care: they are generally the people who are most familiar with the pathology and with the risks as it develops.
- Check that care instructions are well understood (possible cognitive and/or depressive problems)
- Bear in mind difficulty with **mobilisation** and **positioning** (patients who need to be mobilised frequently)
- Take account of pain (suitable mattress)
- Frequent **swallowing problems**. Pass a **nasogastric tube in cases of aspiration**. Tailor the texture of foods, allow time to eat calmly
- Do not forget **mobilisation physiotherapy** (motor exacerbation; less commonly: **recession**)
- Tailor the individual's environment and setup to any motor problems that he/she may have: risk of falls, difficulty in grasping, difficulty in moving limbs (the bell needs to be accessible, the patient may need to be given things to drink and eat)
- Tailor the individual's environment and setup to any eyesight problems that he/she may have (early-onset cataract, particularly in forms that started during childhood): lighting, avoid back-lighting

Organ donation

- The disease features myocardial, sometimes central neurological, pathology; in theory, this pathology is not associated with any type of kidney disorder.
- **No heart, liver or cornea donation**.

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

- Harper PS, van Engelen B, Eymard B, Wilcox DE: **Myotonic Dystrophy: present managment, future therapy.** *Oxford: Oxford University Press* 2004.
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- Groh WJ, Groh MR, Saha MSC, Kincaid JC, Simmons Z, Ciafaloni E, Pourmand R, Otten RF, Bhakta D, Nair GV, Marashdeh MM, Zipes DP, Pascuzzi RM: **Electrocardiographic abnormalities and sudden death in myotonic dystrophy Type 1.** *N Engl J Med* 2008, 358:2688-97.

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