EMERGENCY+CARD

S.O.S

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METABOLIC (GLYCOGEN STORAGE) MYOPATHIES

ACUTE RESPIRATORY INSUFFICIENCY

- Respiratory complications are **frequent in Glycogen storage disease type II** (Pompe Disease) **both in infantile and late onset form.** Respiratory muscles weakness can compromise pump function of the respiratory system, upper airway muscles tone and efficiency of secretion clearance. The respiratory consequences are **secretion retention, upper airway obstruction, nocturnal and finally daytime hypoventilation.**
- **Respiratory infections** (i.e., tracheobronchitis or pneumonia) are the most frequent cause of acute respiratory failure and require early management. Low threshold for empiric antibiotic therapy is recommended for chest infections.
- If no infectious cause of acute respiratory failure is evident, consider non-infectious causes (e.g., **pneumothorax or atelectasis**). **Cardiogenic pulmonary** oedema should be also ruled out.
- Collect respiratory symptoms and **monitor SpO_2 levels via pulse oximetry**; even mild hypoxaemia (e.g., SpO_2 <95% in room air) is a concern and requires a chest x-ray and a blood gas analysis test. Chest x-ray may be difficult to interpret, especially in the presence of scoliosis. In this case chest CT scan may be useful in order to rule out pneumothorax, pneumonia or atelectasis. If even chest CT scan does not show any cause for acute RF, it is useful to deepen the examination by administering contrast medium to exclude a pulmonary thromboembolism.
- **NIV** is often required. In addition, assisted coughing (i.e., breath-stacking techniques with an Ambu bag combined with compression of the chest wall or abdomen) or cough assist device **(MI-E)** help to clear airways secretions. Use the patient's home equipment when available.
- **O_2** must never be used without associating it with NIV. If supplemental oxygen is required titrate oxygen therapy to achieve SpO_2 94-98% and monitor CO_2.
- In case of an acute, reversible event **intubation** and invasive ventilation is indicated when NIV failure occurs (unless there is a known advance directive stating otherwise). When indicated tracheal intubation must not be delayed. Consider that n these patients tracheal intubation may be difficult due to jaw ankylosis, atrophy of the masseter muscle and/or other masticatory muscles, macroglossia or limited mobility of the cervical spine.
- After recovery from the acute illness, these patients should be **promptly extubated to NIV combined with MI-E.** Tracheotomy can be evaluated in particular in patients with severe bulbar dysfunction. However, in the acute phase it should be considered only in the case of multiple failures of weaning protocol including preventive application of NIV combined with MI-E after extubation.

CHOCKING DUE TO SWALLOWING DIFFICULTIES

- Swallowing difficulties are frequent in Infantile onset Pompe Disease, rare in late onset Pompe Disease. Signs and symptoms of swallowing difficulties such as a meal time longer than 30 minutes, **recurrent chest infections**, unintentional weight loss, and **choking** when eating or drinking should be considered.
- Severe bulbar dysfunction increases the patient risk for **aspiration** and hampers the elimination of airway secretions. In addition, it may impede successful use of NIV.
- In case of choking use **MI-E** or manual assisted coughing; if it is ineffective consider **emergent tracheal intubation.**

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ACUTE CARDIAC COMPLICATIONS

- **Dilated cardiomyopathy** is very frequent in some subtypes (type II, III, IV, VII and IX). In the infantile form of Pompe disease hypertrophic cardiomyopathy may be present. **Conduction defects and arrhythmia** are frequent. However, clinical manifestations of heart failure are often unrecognized until very late, owing to musculoskeletal limitations.
- Consider worsening cardiomyopathy and rule out congestive heart failure, atrio-ventricular blocks and arrhythmias.
- Ask for the patient's baseline test results, including echocardiogram and electrocardiogram.
- Obtain a brief history with a focus on baseline cardiac status, including use of medications.
- Ask about cardiac symptoms and **monitor heart rate rhythm, blood pressure and SpO_2.**
- Measure blood levels of **B-type natriuretic peptide** and obtain an **electrocardiogram**; a chest radiograph and/or chest ultrasound may be useful if pulmonary oedema is suspected.
- Obtain an **echocardiogram** and early consultation with a cardiologist.
- In these patients blood level of cardiac Troponin T (cTnT) may be chronically high, while blood level of cardiac Troponin I (cTnI) are more rarely high. Consequently, in the case of suspected myocarditis or myocardial ischemia, it is recommended to measure cTnI.

ANAESTHETIC PRECAUTIONS AND PERIOPERATIVE MANAGEMENT

- Ideally, surgery should occur in a specialist centre with staff experienced in managing these individuals. Otherwise, the urgent surgical interventions may be performed in non-specialized centres following recommendations regarding anaesthesia and perioperative management.
- Obtain a preoperative evaluation including **lung function tests and cough assessment**; if respiratory muscle weakness is present (i.e., forced vital capacity less than 50% of predicted value or peak cough less than 270 l/min), familiarization with ventilatory support (i.e., MI-E and NIV) should be warranted prior to procedure whenever possible.
- Patients should also undergo careful **assessment of heart function** as well as optimization of cardiac therapies in the preoperative period. An electrocardiogram and echocardiogram should be performed before anaesthesia.
- **Use of succinylcholine and inhaled anaesthetics must be avoided** to prevent rhabdomyolysis.
- They may experience **increased sensitivity to sedatives, anaesthetic agents and muscle relaxants**; thus, the depth of anaesthesia and the neuromuscular function should be monitored in order to titrate the appropriate dose of those drugs. In addition, the effect of muscle relaxants should be completely reversed at the end of surgery (i.e., rocuronium should be used and must be reversed by sugammadex).
- In the infantile form of Pompe disease with significant hypertrophic cardiomyopathy, decreased cardiac output and myocardial ischemia have been observed during anaesthesia. In fact, stiffness of the hypertrophied ventricular walls can induce abnormal diastolic relaxation and lead to dynamic left ventricular outflow tract obstruction, elevated left ventricular end-diastolic pressure and reduced diastolic filling. Such a condition may precipitate as a consequence of a decrease in systemic vascular resistance, preload, or both eventually induced by anaesthetic agents, with an increased risk of intraoperative cardiac arrest.
- **Tracheal intubation may be difficult** in patients with NMDs and frequent use of fibreoptic-assisted endotracheal intubation is reported.
- The use of **regional or local anaesthesia** offers a significant advantage in term of avoidance of general anaesthesia and reduction of postoperative respiratory complications.

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ANAESTHETIC PRECAUTIONS AND PERIOPERATIVE MANAGEMENT

- **Morphine** infusions should be avoided, mainly in patients with reduced respiratory function or obstructive sleep apnoea.
- Admission to an **Intensive Care Unit** should be considered in any patient who is at risk for respiratory or cardiac complications. Patients with decreased respiratory muscle strength require close monitoring and aggressive postoperative respiratory management including **early extubation to NIV with aggressive use of MI-E.** O_2 must never be used without associating it with NIV.

FALLS AND FRACTURES

- Owing to weakness, contractures and poor balance, patients with NMDs are at high risk of frequent falls. On the other hand, osteoporosis increases the risk of fractures.
- In ambulatory adult patients, **internal fixation** of femoral fracture is preferable to **conservative treatment** because it allows early walking recovery, preserving muscle function.
- In non-ambulatory adult patients, **conservative treatment** can be considered in case of non-displaced sub capital femoral neck fracture. On the contrary, in diaphyseal or trochanteric femoral fracture **internal fixation is required.**
- In paediatric patients the treatment of femoral fractures is strictly related with the age of the child, the site of the fracture and the disability related to muscle weakness. **Conservative treatment** can be considered in patients under 5-6 years of age, with non-displaced fractures and when a short period of immobilization is expected. In the other cases surgical fixation using **minimally invasive techniques is preferred** (e.g., percutaneous fixation by Kirshner wires and plaster casts, Flexible Intramedullary Nailing or light external fixators).

ACUTE CONSTIPATION DUE TO BOWEL DYSFUNCTION

- Some patients can experience constipation due to abnormal gastrointestinal motility.
- **Gastric and/or abdominal distention may cause acute respiratory failure** in patients at high risk of respiratory complications. In these cases gastrointestinal decompression by using of a nasogastric tube and/or rectal tubes is often an effective therapy.

OTHER ISSUES

- In these patients blood levels of **transaminases and creatine kinase** may be increased. If other hepatic function tests (e.g., bilirubin and gamma GT) are normal, this pattern doesn't necessarily reflect hepatopathy and may be due to muscle involvement.
- Metabolic myopathy presenting with **exercise intolerance** (e.g., McArdle's disease) **may present with acute rhabdomyolysis** with severe hyperCKemia, muscle pain, and myoglobinuria. During such events, there is a risk of acute renal failure.

BIBLIOGRAPHIC REFERENCE
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