

Emergency Information Card for Patients with Spinal Muscular Atrophy (SMA): An Evidence-Based Tool for Acute Care in Rare and Complex Diseases

Cattinari M.G.¹, De Lemus M.^{1,2,3}

1. Fundación Atrofia Muscular Espinal, FundAME, Madrid, Spain. email: cattinari@fundame.net. 2. SMA Europe, Freiburg, Germany. 3. Committee of Advanced Therapies at the European Medicines Agency, Amsterdam, The Netherlands.

Introduction

SMA is a rare genetic neuromuscular disorder marked by progressive muscle weakness, bulbar and respiratory involvement, and multisystemic impact.

Due to its low prevalence, SMA is often unfamiliar to many emergency healthcare professionals (HP), especially in non-specialized settings. In the time-pressured environment of acute medical care, it is unrealistic to expect that non-SMA specialist HP can swiftly access and apply the specialized knowledge required to manage complex rare diseases like SMA.

This limited exposure increases the risk of suboptimal clinical decisions. For this reason, raising awareness of SMA-specific emergency considerations is crucial for ensuring patient safety and preventing avoidable complications.

Systemic Complications and Treatment Considerations in SMA Acute Care

Beyond the classical complications of SMA, emergency HP must also consider less well-known manifestations related to systemic SMN protein deficiency, which may affect other organs and systems.

Additionally, while disease-modifying therapies have significantly improved the prognosis of SMA, they may also pose medical risks or introduce limitations in the use of other treatments during acute episodes. Therefore, they must be carefully factored into their emergency care.

SMA specific emergency information card

In response to recurrent concerns from families about emergency care in SMA, FundAME launched a disease-specific emergency medical information card in 2020.

Designed to support safe, appropriate, and individualized emergency care by providing concise, critical information to HP.



Clinical Basis and Endorsement

Content based on the 2018 internationally recognized SMA Standards of Care. Developed with input from multidisciplinary clinical experts. Endorsed by Spanish scientific societies for:

- Neurology
- Pediatrics
- Pediatric Neurology
- Emergency Medicine



These endorsements reinforce the clinical reliability and practical utility of the card in emergency settings.

Card Features

- Personalized, including confidential personal and clinical information.
- Property of the patient; distributed on request to individuals in the RegistrAME registry.
- Two formats available: a physical card and an online version.
- There are two versions, one for SMA type I and another for types II and III.
- QR code linking directly to the official standards of care guidelines. Supports informed clinical decisions, particularly in acute scenarios.

Monitoring During SMA Treatment

For patients receiving treatments, commercially and through clinical trials, monitor for:

- Coagulopathies
- Thrombocytopenia
- Hepatotoxicity
- Cardiotoxicity
- Nephrotoxicity

Intrathecal therapies require surveillance for procedure-related complications.

Drug Interactions with Risdiplam

Use caution with medications metabolized by CYP3A, including:

Ketoconazole, Erythromycin, Carbamazepine...

Monitor interactions with OCT2 substrates, such as: Ranitidine, Acyclovir, Cephalexin...

Respiratory Management in SMA

Preferred Ventilation Strategy

- Non-invasive bilevel mechanical ventilation (BPAP) is preferred.
- Avoid CPAP as it may lead to respiratory fatigue and clinical deterioration.

Oxygen Supplementation

- Do not administer oxygen empirically without BPAP or CO₂ monitoring.
- Optimize BPAP settings (backup respiratory rate and secretion management) before giving supplemental oxygen.

Secretion Management

- Respiratory support should begin with proper secretion management with manual chest physiotherapy combined with mechanical insufflation-exsufflation (Cough Assist).
- Use caution with Cough Assist in infants <6 months or with significant bulbar dysfunction.
- Recommend ≥2 sessions/day of respiratory physiotherapy.

Endotracheal intubation

- Involve the family in decision-making regarding endotracheal intubation.
- Extubation may be difficult and require tracheostomy.
- Consider limited cervical and mandibular mobility.
- Minimize O₂ supplementation before extubation.
- Do not replace positive pressure ventilation with oxygen alone.
- In cases of recent atelectasis, resolve prior to extubation.

Bronchodilator Use

- Use nebulized bronchodilators in cases of asthma or a positive bronchodilator response.

Infection-Related Decompensation

- Patients with SMA are highly vulnerable to respiratory decompensation during infections.
- Initiate early, aggressive respiratory protocols with proactive interventions.

Nutrition

- Low fasting tolerance – avoid fasting longer than 6 hours.
- Risk of metabolic acidosis, fatty acid metabolism abnormalities, and hyper/hypoglycemia.
- Prone to dehydration – monitor hydration status.
- High risk of bronchoaspiration – use non-oral feeding alternatives and do not resume oral intake until a safety assessment has been performed.

Fracture Risk

- Spontaneous and fragility fractures are common.
- Patients with hip fractures generally benefit from surgical stabilization.
- Casting is recommended, avoiding prolonged immobilization (<4 weeks).

Cardiac and Neurological Complications

- Severe SMA cases may include congenital cardiac malformations.
- Autonomic dysfunction may also occur in advanced disease.
- Communicating hydrocephalus has been reported in some cases.

Impact and Outcomes

It is not possible to isolate the impact of the card from other parallel advancements –such as improved professional awareness, broader access to treatments, or better implementation of standard care– since its launch. Nevertheless, there has been a significant reduction in emergency-related calls to the foundation. The card has thus been consolidated as a practical, reliable, affordable, and patient-centered tool that contributes to improving safety and quality of care in acute settings.