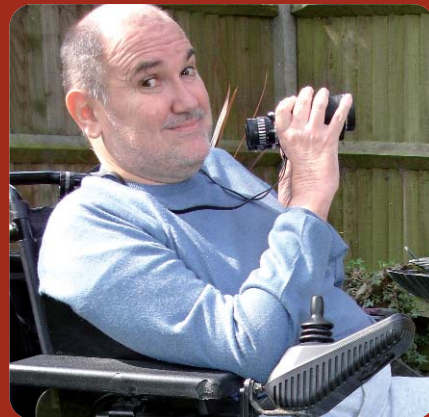


Standards of Care for Spinal Muscular Atrophy



Contents

This booklet is based on the *Consensus Statement for Standard of Care in Spinal Muscular Atrophy*, a document on guidelines for care of SMA patients which was published in the *Journal of Child Neurology* 2007:22 (p.1027-1049).

The *Consensus Statement* document was drawn up by an international group of experts, the International Standard of Care Committee for SMA (part of the ICC for SMA). TREAT-NMD, led by Thomas Sejersen of the Swedish Karolinska Institute, has worked with this group to create a user-friendly précis of these recommendations. This précis forms chapters 1-5 of this booklet and can also be downloaded from the TREAT-NMD website www.treat-nmd.eu/smacare/ where further information on TREAT-NMD's work on SMA care can be found, or from the Jennifer Trust website www.jtsma.org.uk



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Chapter 1

DIAGNOSTIC TESTING AND CARE OF NEW SMA PATIENTS



I. Clinical diagnosis and classification of SMA

Physicians encountering children with hypotonia and weakness should maintain a high index of suspicion for the diagnosis of SMA. Certain physical characteristics are readily identifiable. The weakness is usually symmetrical and more proximal than distal. Sensation is preserved. Tendon reflexes are absent or diminished. Weakness in the legs is greater than in the arms. The severity of weakness generally correlates with the age of onset. Classification and typical clinical features of SMA are listed in Table 1. Apart from these, Type IV SMA is also referred to. This is a mild form that presents in adult

age. Some patients will manifest features that are at the margins between groups.

Care for patients with SMA should be tailored according to their current functional status rather than the original classification of SMA types. Therefore, a classification of current functional level in the form of non-sitters, sitters, and walkers will be used here. The non-sitters include the group of children who currently are not able to sit independently. The sitters include those who can sit independently but cannot walk independently. The walkers can walk independently.

Table 1. Clinical classification of SMA

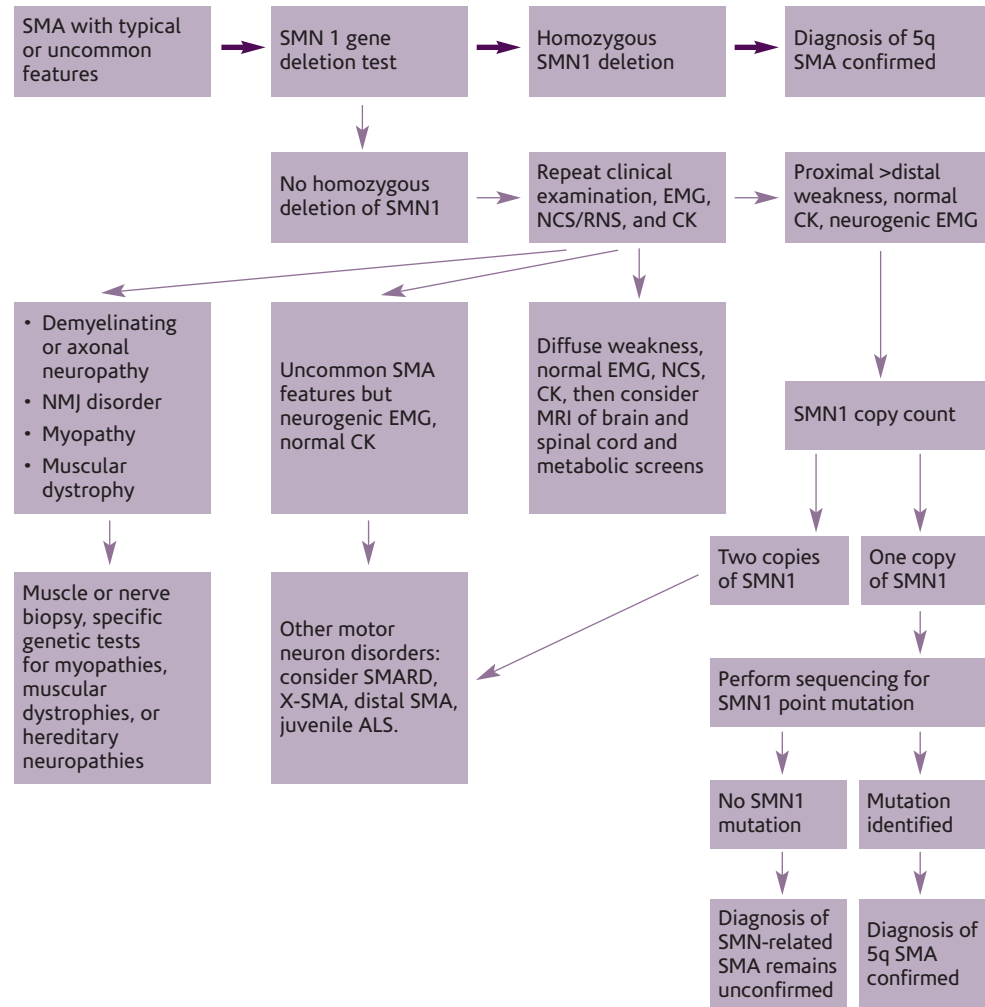
SMA TYPE	AGE OF ONSET	HIGHEST FUNCTION	NATIONAL AGE OF DEATH	TYPICAL FEATURES
Type I (severe) Werdnig-Hoffman disease	0-6 months	Never sits	<2 years	Profound weakness and hypotonia, impaired head control, weak cry and cough, difficulty with swallowing and handling of oral secretion, early morbidity due to respiratory insufficiency and aspiration pneumonia
Type II (intermediate)	7-18 months	Never stands	>2 years	Delayed motor milestones, poor weight gain, weak cough, fine hand tremors, joint contractures and scoliosis
Type III (mild) Kugelberg-Welander disease	>18 months	Stands and walks	Adult	Variable muscle weakness and cramp, joint overuse, loss of walking ability at some point in life

II. Diagnostic procedures

The stepwise algorithm of the diagnostic procedure is summarized in Figure 1. Briefly, the first diagnostic test for a patient suspected to have SMA should be the SMN gene deletion test. A homozygous deletion of the SMN1 gene exon 7 (with or without

deletion of exon 8) confirms the diagnosis of SMN-associated SMA (5q-SMA). The other diagnostic tests should be ordered only after a negative SMN gene test has been obtained.

Fig. 1. Diagnostic evaluation for Spinal Muscular Atrophy



III. Clinical management of newly diagnosed SMA patients

Many care issues arise when a patient is newly diagnosed with SMA. Clinicians need to address the various aspects of care issues as soon as possible.

Family education and counseling:

Because of the complexity of medical problems associated with a diagnosis of SMA, medical providers should designate a person to meet with the family.

During the first meeting with parents it is important to explain:

- The disease process
- Pathogenesis
- Phenotype classification
- Prognosis of the patient's SMA
- Online information on SMA and SMA patient advocacy groups
- Referral to clinical trial studies

The physician should also formulate a plan of multi-disciplinary intervention with the family. This usually includes referral to the following services:

- Pediatric neuromuscular clinic
- Genetics
- Pulmonary
- GI/nutrition
- Orthopedic/rehabilitation

Genetic topics:

Several genetic topics should be addressed with the diagnosis of SMA.

- Genetics of SMA such as autosomal recessive inheritance and genomic structure of the SMN genes - SMN1 and SMN2 copies.
- While higher SMN2 gene copy number is correlated with milder phenotype, predicting clinical severity using SMN2 copy number is not currently recommended because there can be substantial variation of clinical phenotype for any given SMN2 gene copy number.
- Recurrence risk.
- Carrier testing.
- Information for reproductive planning (prenatal or pre-implantational diagnosis).

Chapter 2

PULMONARY CARE



I. Overview of pulmonary problems in SMA

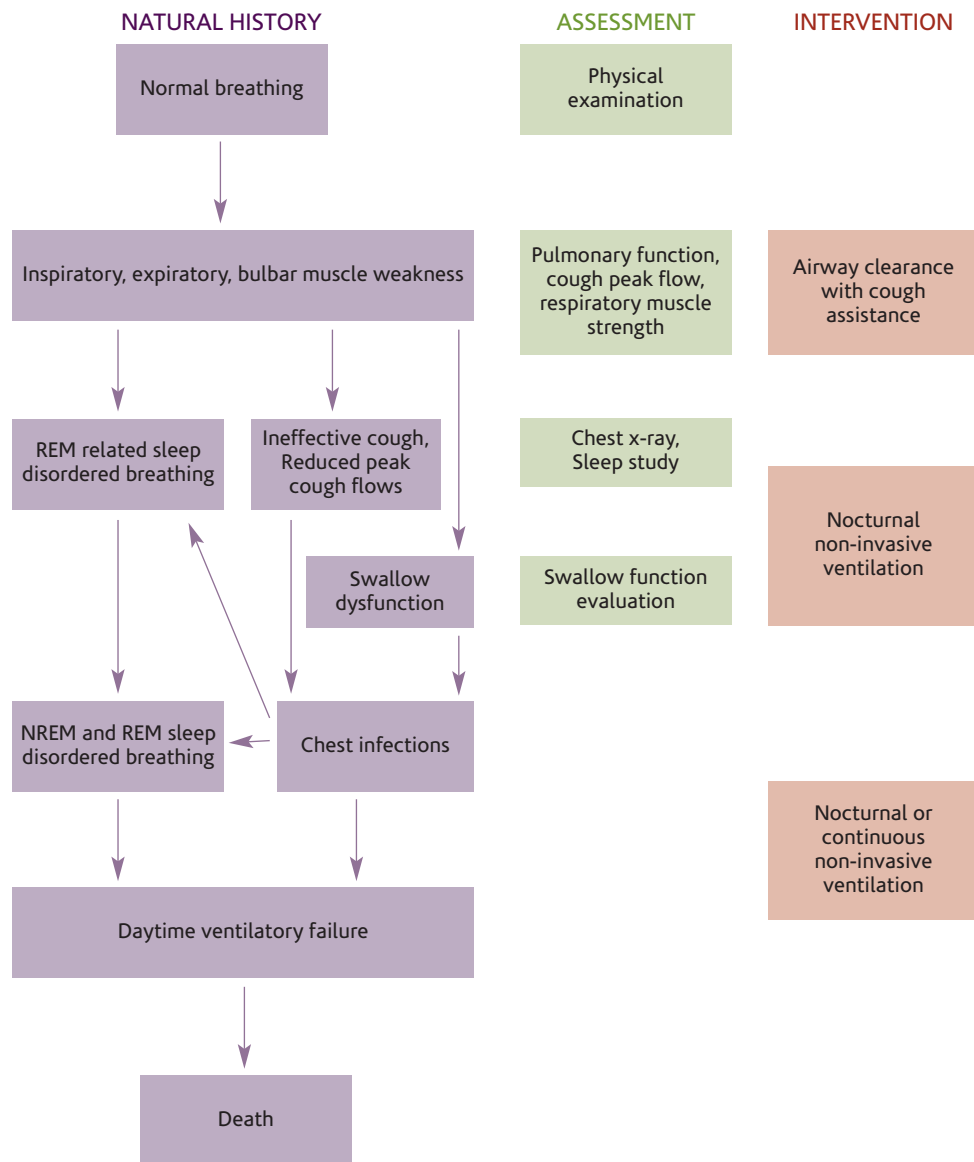
The key respiratory problems in SMA are:

- 1 **Impaired cough** resulting in poor clearance of lower airway secretions
- 2 **Hypoventilation** during sleep
- 3 **Chest wall and lung underdevelopment**
- 4 **Recurrent infections** that exacerbate muscle weakness

Pulmonary disease is the major cause of morbidity and mortality in SMA types I and II, and may occur in a small proportion of patients with SMA type III. Swallowing dysfunction and reflux are important contributors to pulmonary morbidity. Individuals tend to progress to daytime respiratory failure via a sequence of recurrent chest infections, nocturnal oxygen desaturation, followed by nocturnal hypoventilation and then daytime hypercarbia. Ventilatory support should be added at night if sleep disordered breathing is present, and cough assistance provided if cough efficiency is reduced. Airway clearance is very important in both acute and chronic management of all patients with SMA.



Fig. 2. Pulmonary natural history, assessment, and intervention



II. Assessment and monitoring

Suggested frequency of evaluation is every 3-6 months, less frequently in stable walkers, more frequently in clinically unstable non-sitters.

A. Non-sitters:

- **Physical examination:** monitor cough effectiveness, chest wall deformity, work of breathing, respiratory rate, paradoxical breathing, and skin color.
- **Polysomnography:** to document signs of hypoventilation.
- **Pulse oximetry:** to monitor oxygen saturation through transcutaneous sensor.
- **Pneumonias:** monitor frequency of infection and antibiotic treatments over past 6 months.
- **Chest x-ray:** baseline and during respiratory deterioration.
- **Swallow studies:** in unexplained acute respiratory deterioration and recurrent pneumonia.

B. Sitters:

- **Physical examination:** monitor cough effectiveness, chest wall deformity, work of breathing, respiratory rate, paradoxical breathing, and skin color.
- **Polysomnography:** to document signs of hypoventilation.
- **Pulse oximetry:** to monitor oxygen saturation through transcutaneous sensor.
- **Pneumonias:** frequency of infections and antibiotic treatments over the past 6 months.
- **Scoliosis:** inspection of spine and radiographic evaluations of scoliosis.

C. Walkers:

In general, SMA walkers have relatively preserved pulmonary function until late into their disease course.

- **Physical examination:** monitor cough effectiveness, chest wall deformity, work of breathing, respiratory rate, and skin color.
- **Pulmonary function testing:** spirometry, lung volumes, and respiratory muscle function.
- **Pneumonias:** frequency of infections and antibiotic treatments over the past 12 months.

III. Anticipatory respiratory care

Critical to respiratory management of SMA is to provide families with **information about options for chronic care, acute illness management, and perioperative care.**

- Non-sitters are the most fragile group and early discussions should include the option of **noninvasive ventilation (NIV) and secretion management** due to the rapid progression of the disease.
- Ongoing discussion of the family's desires for support should occur, and the result should be a negotiated care plan with **maximums and minimums outlined.**

Day to day management should include:

- Understanding the child's **baseline** and deviations from his/her baseline.
- Understanding **hypoventilation** and intervention.
- **Acute illness management** including rapid access to specialty medical care providers.
- **Airway clearance** and secretion management techniques.
- **Respiratory support** including NIV.
- **Nutrition and hydration.**
- A low threshold to start **antibiotics.**
- Routine **immunizations** including influenza vaccine, pneumococcus vaccine, and RSV prophylaxis (palivizumab).

IV. Chronic management

Discussion of the family's goals is essential. This may include balancing caring for the child at home for as long as possible, long term survival, quality of life and comfort, and the availability of resources. Goals of chronic management are to: **normalize gas exchange, improve sleep quality, facilitate home care, reduce hospitalizations and ICU care, and reduce the burden of illness.** Early aggressive and proactive intervention may prolong life without compromising quality of life.

Airway clearance:

- **Assisted cough**, manually or assisted with mechanical insufflation-exsufflation, is recommended daily in more severely affected patients. Caregivers of patients with SMA should learn to assist coughing.
- **Secretion mobilization techniques** are helpful and include chest physiotherapy and postural drainage.
- **Oximetry** should be used to guide therapy. Oral suctioning can assist in secretion management after assisted coughing.

Respiratory support:

This is clearly indicated in daytime hypercapnia. Nocturnal NIV reduces symptoms of disordered breathing during sleep and increases life quality.

- **NIV** should be combined with airway clearance techniques.
- In non-sitters, **care without ventilation support** is an option if the burden of treatment outweighs benefit.
- **CPAP** may be an option, with the goal of transition to bi-level positive airway pressure (**BiPAP**).
- Use of **NIV** with high span BiPAP, even for **short daytime periods**, may improve chest wall and lung development, and **reduce ribcage and sternal deformity** in non-sitters and sitters.
- **Tracheotomy:** In non-sitters, this is controversial and an **ethical dilemma.** There is a large spectrum of options that can be provided, ranging from no respiratory support, to NIV, to tracheotomy, and mechanical ventilation.
- **Palliative care** is an option for non-sitters. NIV can be used as a routine therapy or as a palliative tool. A key goal is to prevent PICU stays and avoid tracheotomy if possible. If supportive ventilation is chosen by the family, NIV is recommended.

V. Perioperative care

Patients with SMA are at high risk for post-anesthesia complications, which may lead to prolonged intubation, nosocomial infections, tracheotomy, and death. It is critical that the patient's respiratory status be optimized before surgery.

Pre-operative evaluation:

Physical examination

- Measurements of **respiratory function** and cough effectiveness.
- **Chest x-ray.**
- Evaluation for **sleep disordered breathing.**
- Consider **complicating factors** including jaw ankylosis, oropharyngeal aspiration, gastroesophageal reflux, nutritional status, and asthma.

If measurements of respiratory function and/or sleep study are abnormal, **nocturnal NIV** and **assisted coughing techniques** may be indicated before surgery. The patient should become familiar with these techniques prior to surgery. If **jaw ankylosis** is present, intubation should be performed by fiberoptic bronchoscopy.

Post-operative management:

- If **cough clearance is normal** and **muscle function is relatively preserved**, there is not an increased risk for post-operative complications.
- If **decreased respiratory muscle strength is present pre-operatively**, close monitoring and aggressive respiratory management is required.
- If **respiratory support** is required pre-operatively during sleep, similar respiratory support in the immediate postoperative course is required.

- **Extubation** in the recovery room to NIV should be planned as a bridge to weaning to the patient's baseline respiratory support. This requires careful planning and coordination. If the patient requires continuous ventilator support pre-operatively (via non-invasive interface or via tracheotomy tube) or use of muscular blocking agent during surgery then it is best to transferred patient directly from OR to ICU.
- Patients are encouraged to bring their **personal devices such as NIV and MI-E machines** to use in the post-operative period because hospitals may be limited in the availability of these devices.
- **Oxygen** must be applied with caution in the patient with SMA. Hypoxemia secondary to hypoventilation may be mistaken with hypoxemia due to other causes such as mucus plugging and atelectasis. **ETCO₂ or TcCO₂ monitoring** or arterial **blood gas analysis** will facilitate appropriate oxygen use.
- **Adequate pain control** will aid in preventing hypoventilation secondary to splinting. Pain management should be titrated to promote airway clearance and minimize respiratory suppression. Transient increased respiratory support may be needed while controlling post-operative pain.

VI. Acute care management

The goal of management during acute illness is to **normalize gas exchange** by reducing atelectasis and enhancing airway clearance where possible by non-invasive respiratory support. Blood gas monitoring may be of benefit.

Airway clearance:

- **Airway clearance** with manual cough assist or MI-E, oral or airway suctioning. Assisted cough techniques are preferred over deep suctioning and bronchoscopy.
- **Oximetry** feedback to guide airway clearance.
- **Chest physiotherapy.**
- **Postural drainage.**

Respiratory support:

(i) For non-sitters and sitters:

- **Acute use of NIV** reverses ventilatory decompensation caused by vicious cycle of added ventilatory load, increased respiratory muscle weakness, and ineffective secretion clearance.
- If already using nocturnal NIV, **daytime NIV** may be required, and airway clearance techniques carried out during NIV.
- **Oxygen therapy** entrained into the NIV circuit should be used to correct oxygen desaturation, after inspiratory and expiratory positive pressure settings are optimized and airway clearance techniques are optimally utilized.

- If a non-invasive approach fails, **intubation and mechanical ventilation** is a short-term measure. After recovery from the acute illness and arterial oxygen saturation on room air has normalized, they should be extubated back to NIV.
 - Decision-making about escalation to intubation should be carried out in advance as part of **anticipatory care planning.**
 - **Tracheotomy and ventilation** can be considered in case of frequent acute pulmonary infections in non-sitters, but may not improve quality of life or reduce hospitalizations. A tracheotomy is not an acute intervention. Tracheotomy is not appropriate in sitters.
 - With deteriorating function it may be appropriate to redirect care to a **palliative approach**, particularly for non-sitters.
- #### (ii) For walkers:
- **NIV** may be needed during an acute illness, in combination with airway clearance techniques.
 - **Oxygen therapy** and need for transient intubation should be carried out as outlined above for non-sitters/sitters.
 - **NIV for home use** should be considered if NIV was needed during an acute illness.

Additional management:

For non sitters, sitters, and walkers, recommended additional therapies are antibiotics, adequate nutritional support, hydration, and gastroesophageal reflux management.

Chapter 3

GASTROINTESTINAL (GI) AND NUTRITIONAL CARE



Overview of gastrointestinal and nutritional care

The key clinical problems associated with GI and nutritional complications in SMA are:

- 1 **Feeding and swallowing problems.**
Bulbar dysfunction is universal in SMA patients with severe weakness and can result in aspiration pneumonia, which is a common cause of death.
- 2 **Gastrointestinal dysfunction.**
GI dysmotility problems include constipation, delayed gastric emptying and potentially life-threatening gastro-esophageal reflux (GER).
- 3 **Growth and under/over nutrition problems.**
Without optimal management, growth failure is universal in non-sitters while excessive weight gain is more common in sitters and walkers.
- 4 **Respiratory problems.**
The presence of respiratory complications (weak cough, increased work of breathing, dyspnea, pneumonias and cyanosis or desaturation with feeds) raises concerns for feeding difficulty and increased risk of aspiration which can be life threatening. Increased work of breathing may also result in increased energy expenditure.

I. Feeding and swallowing problems

Feeding and swallowing difficulties are common in non-sitters and sitters but are rarely a concern in walkers.

1. Key symptoms of feeding and swallowing problems:

- **Prolonged mealtime.**
- **Fatigue** with oral feeding.
- **Choking or coughing** during or after swallowing.
- **Recurrent pneumonias:** potential indicator of aspiration, which may be silent, i.e. without evident choking or coughing.
- **Vocal cord paralysis** may be diagnostic sign of **silent laryngeal aspiration.**

2. Causes of feeding difficulty:

Pre-oral phase

- **Limited mouth opening** due to reduced range of mandibular motion.
- Difficulties in **getting food to the mouth** for self-feeding.

Oral phase

- **Weak bite force.**
- Increased **fatigue** of masticatory muscles.

Swallowing phase

- **Poor head control.**
- Inefficient **pharyngeal phase** of swallowing.
- **Poor co-ordination** of the swallow with airway closure.

3. Evaluation of feeding and swallowing problems:

- Feeding assessment by **feeding specialist**.
- A **feeding case history** with mealtime observation is desirable.
- **Examination of oral structures** which impact on feeding efficiency and consideration of the effect of **positioning and head control** on feeding and swallowing is essential.
- **Videofluoroscopic swallow studies (VFSS)** if concerns on concerns about swallow function and safety, and opportunity to evaluate therapeutic strategies.

4. Management of feeding and swallowing difficulties:

Treatment should aim at reducing the risk of aspiration and optimizing efficiency of feeding and promote enjoyable mealtimes.

- **Changing food consistency** and optimizing oral. A semi-solid diet can compensate for poor chewing and reduce length of mealtimes. Thickened liquids may protect against aspiration of thin fluids. Preferably, this intervention would be evaluated objectively on VFSS.
- **Positioning and seating alterations and orthotic devices** (e.g. Neater Eater®, elbow support, valved straw) to enhance self-feeding ability may improve swallow safety and efficiency. Plan in liaison with an occupational therapist and/or physiotherapist as required.

- **Proactive nutritional supplementation** as soon as inadequate oral intake is recognized. Whether or not a g-tube is placed in a particular child often requires extensive discussion with multiple caregivers. Nutritional supplementation via **nasogastric (NG) or nasojejunal (NJ) feeding** is desirable in the interim before g-tube placement. NJ feeding may be preferable in circumstances when GER with aspiration is a concern, especially when the patient is on ventilatory support. However, technical difficulty may prevent its feasibility.
- **G-tube feeding** is the optimal method of feeding for insufficient caloric intake or unsafe oral feeding. It prevents the potential morbidity and poor ventilatory mask fit associated with prolonged use of either NG or NJ tubes. A laparoscopic surgical technique for g-tube placement provides the best possible setting for immediate or early post-operative extubation. Care should be taken to minimize the amount of fasting preoperatively, and to quickly resume full nutritional support following the procedure.

II. Gastrointestinal dysfunction

Children with SMA suffer from the following GI problems: gastroesophageal reflux (GER), constipation, and abdominal distension and bloating. GER is an important determinant of mortality and morbidity in SMA patients. High fat foods delay gastric emptying and increase the risk of GER.

1. Key symptoms of GER:

- Frequent “**spitting up**” or **vomiting** post meals.
- **Emesis**.
- Complaints of **chest or abdominal discomfort**.
- **Bad breath**.
- Obvious **regurgitation** of feeds.
- **Refusal of feeds** when developing discomfort with swallowing.

2. Evaluation of gastrointestinal dysfunction:

- **Search early for symptoms** of GER (emesis, regurgitation, gurgling after feeds).
- A routine **upper gastrointestinal (UGI) series** for presurgical evaluation for gastrostomy tube (g-tube) placement to primarily rule out anatomical anomalies and secondarily to document reflux.
- **Motility studies** including scintigraphy can be helpful in documenting delayed gastric emptying which may contribute to GER and early satiety.

3. Management of gastroesophageal reflux (GER):

- Short-term use of **acid neutralizers** (e.g. magnesium or calcium carbonate) and/or **inhibitors of acid secretion** (e.g. histamine blockers and proton pump inhibitors (e.g. famotidine, ranitidine, omeprazole) for symptomatic management. However, prolonged use may be associated with a greater risk for gastroenteritis and pneumonia.
- When delayed gastric emptying or diminished motility is present, **prokinetic agents** (e.g. metaclopramide, erythromycin) may be useful.
- Use of **probiotics** such as acidophilus or lactobacillus to help maintain a healthy gastrointestinal flora, particularly following antibiotic treatment or in the setting of prolonged use of acid inhibitors, is an area deserving further study.
- Laparoscopic **anti-reflux Nissen fundoplication** during g-tube placement may be of value in the SMA patient with medically refractory GER, and in whom the benefit is deemed to outweigh the associated surgical and anesthetic risks.

III. Growth and under or over nutrition problems

Children with SMA are at risk for growth failure or excessive weight gain. Growth failure is commonly seen in non-sitters and some sitters, while obesity is a problem in the stronger sitters and walkers. Decreased activity and lean body mass will lead to reduced resting energy expenditure and increased risk of obesity.

Management of growth and under or over nutrition problems:

- The goal is to maintain each child on his/her own growth velocity.
- **Follow growth velocity curves** (weight, height/length, weight/height) followed over a period of time. Recumbent length, segmental measurements or arm span may be useful if contractures complicate length measurement.
- Assessment of **nutritional intake** by a **dietician** or other health care provider proficient in nutrition is recommended at each visit. A **3 day dietary record** is a simple and accurate tool to assess nutritional intake. A **24 hour food recall** is a practical method to highlight major nutritional concerns and inquire regarding use of any special supplements.

IV. Management of nutrition in acutely sick SMA patients

- With a reduction in lean body mass, **calculated body mass index (BMI)** will significantly underestimate body fat. This may result in inappropriate dietary recommendations which could lead to relative obesity.
- SMA patients at **risk of obesity** should have growth parameters in the lower percentiles for weight/height and BMI.
- It is important to document appropriate intake of **calcium and vitamin D**.
- Checking **pre-albumin levels** may help assess adequate **protein status**.
- SMA patients, particularly non-sitters and sitters, are particularly vulnerable to catabolic and fasting states, and are more likely to develop **hypoglycemia** in the setting of fasting. It is therefore necessary to avoid prolonged fasting, particularly during acute illness, in all SMA patients.
- Nutritional intake should be optimized to meet **full caloric needs within 4-6 hours** following admission for acute illness, via enteral feeding, parenteral feeding or a combined approach as necessary.
- Prompt **post-operative caloric supplementation** is recommended to avoid muscle catabolism, particularly in a child with reduced fat store. If enteral intake is not imminent then IV caloric feeding should be considered.

Chapter 4

ORTHOPEDICS CARE AND REHABILITATION



Overview of orthopedic care and rehabilitation strategies in SMA

A Key problems:

Muscle weakness resulting in **contractures, spinal deformity**, and increased risk of **pain, osteopenia and fractures**.

B Key evaluation procedures:

- Range of motion (ROM)
- Strength, function
- Seating and mobility
- Orthotics
- Radiographs (spine and other joints)
- DEXA scan
- Orthopedic surgery

I. Recommendations on evaluation and treatment by functional levels

A. Non-sitters:

Assessments:

- **Physical and occupational therapy evaluation** of function (CHOP-INTEND).
- **Speech therapy** evaluation if swallowing is impaired or speech affected by jaw contracture or inadequate voice.

Key interventions:

- **Nutritional support.**
- **Posture management:** Patient's primary posture should direct choice of equipment that supports function. Ensure comfortable seating.
- **Contracture management:** Splinting to preserve ROM and prevent pain may be indicated.
- **Pain management.**
- **Therapy for ADL and assistive equipment:** Play and occupational support should include lightweight toys and assistive technology with variable controls and a myriad of activation systems.
- **Wheelchair:** Ensure optimal independence and seating comfort.
- **Limb orthotics:** Upper extremity (UE) orthotics to aid in function includes the use of mobile arm supports or elastic slings that augment active range of motion and functional abilities.
- **Environmental controls and home modifications** to allow for safe accessibility and optimal independence.

B. Sitters:**Assessments:**

- **Functional assessment** (Hammersmith Functional Motor Scale for SMA, the Modified-Hammersmith functional motor scale for SMA, Gross Motor Function Measure (GMFM), and the Motor Function Measurement (MFM) scale for neuromuscular disease).
- **Contracture measurement** by goniometry.
- **Strength measurement** by manual muscle testing or myometry.
- **Spine and hip radiographs.**
- **Equipment evaluation** of seating, mobility, positioning, and self care equipment. Evaluations for manual and power mobility may be conducted as early as 18 to 24 months of age.

Key interventions (PT, OT, and orthopedics):

- **Wheelchair mobility.** Ensure optimal independence and seating comfort.
- **Environmental controls and home modifications** to allow for safe accessibility and optimal independence.
- **Contracture management** constitutes major focus of treatment with regular stretching and bracing program to preserve flexibility. Serial casting for contractures may improve standing and improve tolerance of bracing. AFO orthotics may delay development of Achilles tendon contractures. Upper extremity orthotics with mobile arm supports or slings augment active range of motion and functional abilities.
- **Regular exercise should be encouraged** to maintain fitness and endurance and might include swimming and adaptive sports.
- **Standing is encouraged.** Light weight ischial weight bearing KAFOs or reciprocal gait orthoses (RGO's) for standing or assisted ambulation for those with sufficient strength. Where this is not possible a standing frame or mobile stander with AFOs should be considered.
- **Spine orthotics and surgery** (see below).

C. Walkers:**Assessments:**

- **Balance and ambulation evaluations** include a specific survey of environmental adaptability and access.
- Evaluation of **joint ROM** and **spinal alignment.**
- **PT and OT assessments** to determine appropriate mobility aides, adaptive equipment, assistive technology and environmental access.
- **ADL assessment** for equipment and adaptation.
- **Non-spine x-rays and DEXA** are considered in the event of acute musculoskeletal injuries, as a result of overuse, an accident or a fall.

Key interventions:

- **Wheelchair** for longer distance transportation adds mobility and independence.
- **Contracture management** and education to maximize joint protection.
- **PT and OT** to maximize safety, endurance and independence or to prolong ambulation.
- **Walking should be encouraged** with appropriate assistive devices and orthotics.
- **Regular exercise** to maintain fitness and stamina. May include swimming, aquatic therapy, horseback riding and adaptive sports.
- **Driver's education** alternatives and consideration of customized driving controls.
- **Environmental controls and home modifications** to allow for safe accessibility and optimal independence.
- **Spine and limb orthotics** if scoliosis and contractures start to develop.
- **Spine surgery** (see below).

II. Orthotics

- It is important that the **orthotist, therapist and family work together** to ensure that the appropriate orthosis is fabricated and allows wearers to meet their functional goal.
- The **orthotist should have a good background and experience** in working with patients with neuromuscular disorders to choose proper materials and to make adaptations that allow for “best” fit and function.
- **Spinal orthoses** may be used for postural support but **there is insufficient evidence to support delayed curve progression**. When used, spinal orthoses should be fabricated with an **abdominal cut out** to allow appropriate diaphragmatic excursion and access to gastrostomy tubes where present.

III. Orthopedic surgery

1. Hip subluxation and contractures:

- **Hip subluxation** in SMA is rarely painful. Surgical reduction and osteotomy is frequently followed by redislocation. In most circumstances, this surgery is avoidable.
- Ankle and foot deformities make conventional shoes difficult to wear, and may be an indication for **soft tissue releases**. In walkers, if soft tissue releases are performed, rapid and aggressive physical therapy may improve outcome.

2. Scoliosis surgery:

- Scoliosis surgery provides benefits in sitting balance, endurance, and cosmesis. Earlier surgery results in better outcome.
- Scoliosis surgery appears to be **beneficial in patients that survive beyond two years of age** when curves are severe and progressive and should be performed while pulmonary function is adequate.
- Beneficial effects of scoliosis surgery on **pulmonary function** remain controversial, but the rate of pulmonary decline may be slowed.

IV. Perioperative management in SMA

1. Pre-operative management:

- **A plan for orthotic intervention** including timing and **modification of orthoses**.
- New **wheelchair** or wheelchair modification (seat, back, arm, leg or head rests) likely to be required.
- **Instruction in transfer** including arrangements for a mechanical lift, if necessary.
- Arrangements for **bathing, toileting and dressing** equipment and potential modifications to clothes.
- Pre-operative **spirometry, noninvasive (NIV) pulmonary supports** such as BiPAP and, if necessary, cough-assist devices.

2. Post-operative management:

- Confirm timing of appropriate **casting and fitting of orthoses**, allowed **ROM**, and **activity**, and that appropriate adaptive equipment is available.
- Appropriate use of incentive **spirometry and NIV pulmonary support**.
- Instruction of nursing staff and family on **bed mobility, transfers, dressing, bathing and toileting**.
- **Mobilization** as soon as possible as allowed by the procedure and surgeon.

Chapter 5

PALLIATIVE CARE



Palliative care

- Optimal clinical care for SMA patients should be **mindful of potential conflict of therapeutic goals**. This conflict is made more difficult by the natural involvement of surrogate decision makers for a dependent infant (parents, siblings, other relatives, caregivers, payers and the wider community).
- There is a deep responsibility to **present care options** in an open, fair and balanced manner, to be started soon after diagnosis.
- A choice for or against interventional **supportive care is not a single binary choice**, nor must it be unchanging with circumstance. Sufficient time, honest appraisal of the choices, openness to revisiting decisions made, and personal rapport are essential.
- Placement of **gastrostomy tube is better done relatively early** when associated risks are lower in order to provide more stable and comfortable nutritional support later when feeding is more tenuous.
- **Discuss and determine early the appropriate response to potential life-threatening respiratory insufficiency**, as emergency resuscitation and endotracheal intubation during times of crisis without prior respiratory support is associated with many more problems than when decisions are made in advance. If appropriate, other forms of non-invasive respiratory aid should be introduced in time and according to increasing need.
- **End of life care decisions need to be defined**, neither delayed nor aggressively foisted upon unsuspecting, grieving, stunned parents.
- Care is often best accomplished with **a multi-specialty team approach**, including appropriate medical, social, and spiritual assistance as appropriate. In addition, hospice referral or other provision for the specific issues regarding terminal care, grief and bereavement support are important.
- In the circumstance of a choice against mechanical ventilatory support, **appropriate provision for management of terminal dyspnea can be of comfort to patient and family alike**. Use of nebulized narcotics can avoid much of the concern that overdosing contributes to death and provide comfort to the patient.

SMA patient registry



What is the SMA patient registry?

A patient registry collects information about patients who are affected by a particular condition.

The EU funded network for neuromuscular disorders TREAT-NMD has created a registry for patients with Spinal Muscular Atrophy (SMA). This SMA patient registry collects genetic and clinical data about SMA patients with a confirmed mutation in the SMN1 gene.

How do I register?

Registration is voluntary and done by the patients/parents themselves. Patients with SMA, or their parents, can register either over the Internet (which allows them to view and update their data at any time) or by completing a paper form.

To register online, visit

www.treat-nmd.org.uk/registry

If you cannot register online, contact us at the address (right) and we will send you a paper registration form to fill in.

More information about the TREAT-NMD network is available at

www.treat-nmd.eu

Why should I register?

There are several good reasons for patients to register:

- Registered patients may be able to participate in clinical trials more easily
- Registered patients are kept informed about research results, such as new treatments for SMA
- Registries help specialists gain more knowledge about the prevalence, the epidemiology and the natural history of SMA
- Registries may help raise urgently needed funding for research

Contact details:

TREAT-NMD SMA Patient Registry
Institute of Human Genetics
Newcastle University
International Centre for Life
Newcastle upon Tyne
NE1 3BZ, United Kingdom

E: registry@treat-nmd.org.uk

The TREAT-NMD SMA patient registry in the UK is supported and co-sponsored by The Jennifer Trust for Spinal Muscular Atrophy and patients can also register through the Trust's website

www.jtsma.org.uk



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