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Nutrition in SMA

Nutrition in SMA Workshop

Sponsored by Roche, in collaboration with SMA Europe

Monday 21st November 2022, Basel, Switzerland

This meeting report for the Nutrition in SMA Workshop comprises topics and discussions on feeding support, weight management and measurement of swallowing and feeding. Insight sharing from patients, caregivers and expert panellists helped to define current unmet needs and next steps to address these gaps

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Contents

Nutrition in the spotlight of SMA care:
A joint mission for experts, patients and families

Moderator: Prof. Giovanni Baranello
Panel: Mrs Sara Almroth, Prof. Simona Bertoli, Dr Ramona De Amicis, Dr Yasemin Erbas

Overview

This session covered current practices and unmet needs in the management of nutritional care, feeding support and weight control in SMA. The speakers presented an overview of nutritional management for different types of SMA and current unmet needs in this field. Perspectives from people living with SMA and their caregivers were shared regarding the delivery of quality nutritional/feeding care.

Key points and unmet needs

Key points

- The main nutritional and feeding challenges for people living with SMA and their caregivers relate to meeting nutritional needs, gastrointestinal (GI) issues, psychological impact and managing swallowing status and choking risk
- Nutritional care is critical in SMA as people are at risk of being under- or overweight, which can be associated with alterations in glucose and lipid metabolism, growth deficiency and reduced bone mineral density
- It is important to regularly evaluate nutritional status and needs using assessments and measurements that are specific to SMA, such as anthropometry, growth charts and predictive equations for fat mass and for basal metabolic rate to estimate energy expenditure
- There is a lack of data and guidelines on recommended nutritional intake for people with SMA
- People with SMA may opt to experiment with their own nutritional and physical exercise plans to determine what works best for them
- The composition of diet should be individualised according to the person's phenotype, symptoms and growth patterns

Unmet needs

- To define a healthy diet for people living with SMA, as well as a healthy weight and how this can be maintained
- To address the psychological impact of nutritional and feeding complications
- To help people living with SMA understand what the most beneficial diet and training regimens are, based on scientific evidence
- To conduct further clinical studies on the following topics:
  - Longitudinal evaluation of nutritional approaches in SMA
  - Evaluation of high-fat versus low-fat diets and the tolerability of high-carbohydrate diets in SMA
  - Protein metabolism in SMA (*in vivo*)
  - Measurement of estimated energy requirements for total daily physical activity
  - The effects of the amino acid diet, including on GI issues and quality of life (QoL)

Speaker presentation: Dr Yasemin Erbas

Dr Erbas presented her experience of nutrition and swallowing challenges as a caregiver of a child with SMA.

Dr Erbas discussed the experiences of caregivers. She explained that it is important to understand how dietary needs may change with age, ability and disease state for people living with SMA. Dietary choices may have an impact on SMA symptoms and could influence strength and fatigue, so it is important to ensure caregivers have access to recommendations for diets and dietary supplements as well as what can be considered a healthy weight to maintain.

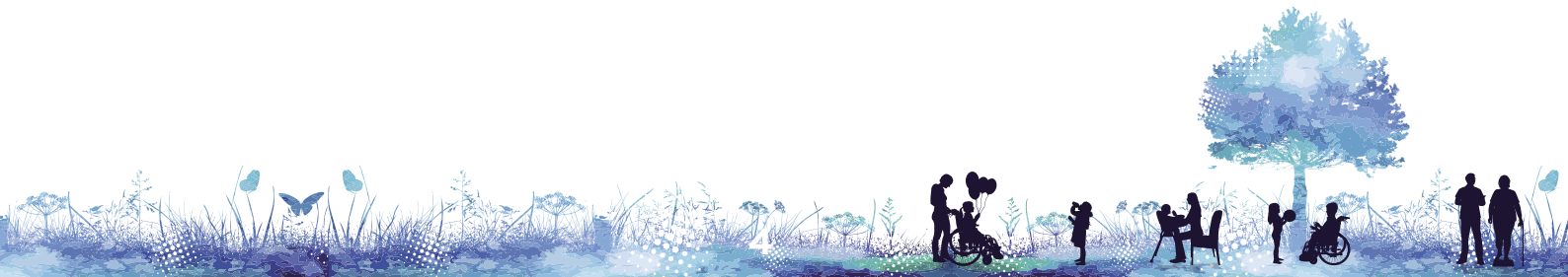
People living with SMA often have a small stomach and are only able to tolerate a small amount of food at one time. Common symptoms related to feeding include vomiting, bloating, acid reflux and constipation. People with SMA may experience an elevated heart rate if they have difficulties with digestion. Caregivers have to consider positioning when eating (sitting up vs lying down), timing of feeds and the nutritional value of each meal, which can be time-consuming.

Difficulty swallowing is a significant challenge for people living with SMA and their caregivers. In addition to reducing independence, weak swallowing increases the chance of choking, which can lead to anxiety. Children with SMA eat differently to other children their age, which can be psychologically challenging, particularly once they reach adolescence.

Food plays an important social role in many cultures, as many social activities centre around food. Family habits often need to change to support the need for several smaller meals rather than a few larger meals every day. This can add to feelings of stress, worry and guilt for family members supporting people living with SMA.

Dr Erbas explained that there is a need to understand the needs of people with SMA by conducting surveys of SMA communities, and for scientific researchers and dieticians to further investigate the needs of people living with SMA. The findings of such studies need to be disseminated and shared with physicians, people with SMA and their caregivers. Stronger communication between clinicians and caregivers is important when making decisions in the best interests of the person living with SMA.

The dietary needs of people living with SMA need to be further investigated and consensus shared with the entire community



Speaker presentation: Prof. Simona Bertoli

Prof. Bertoli presented an overview of the importance of active nutritional management in SMA.

Prof. Simona Bertoli discussed the importance of active nutritional support and metabolism surveillance in the management of people with SMA, as nutritional management may have a significant impact on clinical course and prognosis.

People with SMA are at risk of being either under- or overweight due to respiratory problems, increased energy expenditure during meals, GI disorders, or a decrease in physical activity due to severe neuromuscular deficit. It is important to optimise nutrition and growth and conduct early assessments of nutritional impairment.

Expert nutritionists should be included in the multidisciplinary team to monitor growth patterns, define appropriate energy intake and identify dietary requirements. Some dietary requirements such as vitamin D can be measured in the blood to define the level of deficiency and establish the amount of supplementation needed. Calcium intake is relatively easy to establish from the diet, so the amount that should be supplemented for each individual can be easily calculated.

It is important that nutritionists are included in the multidisciplinary team to ensure accurate measurement and surveillance of energy intake and dietary requirements

Click to read about nutritional studies in SMA

Speaker presentation: Mrs Sara Almroth

Mrs Almroth presented an overview of her experience of nutritional challenges as a person living with SMA.

Mrs Sara Almroth shared her experience of living with SMA. Mrs Almroth discussed the challenges she faces with nutrition in everyday life, and the lifestyle changes she has made to manage these. She outlined unanswered questions in relation to dietary and exercise requirements for people living with SMA and explained the guidance that is needed to help with their nutritional management.

Nutritional guidance and tools to support physical exercise are needed for people living with SMA

Understanding nutritional requirements for management of weight poses a significant challenge to people with SMA. It can be challenging to find scientific evidence on optimal nutrition and ideal weight for people living with SMA.

Mrs Almroth explained that she wanted to have more energy, lose weight and reduce GI issues. She experimented by changing her diet, including lowering her sugar intake and using supplements to manage her GI issues, and found that nutritional shakes provided her with increased energy as well as being easier to eat than solid food. Mrs Almroth experimented with an organised exercise regimen, including the use of a vibration plate to aid circulation, and found that singing is good training for breathing and neck/mouth muscles.

There is a need for guidance on nutrition to advise people living with SMA on the number of calories they should aim to eat every day, as well as the proportions of protein and carbohydrates they should aim for in their diet. There is a need for tools that support physical exercise beyond physical therapy, including electro muscle stimulation training.

Speaker presentation: Dr Ramona De Amicis

Dr De Amicis discussed current understanding and unmet needs surrounding dietary recommendations for people living with SMA.

Dr Ramona De Amicis discussed current dietary recommendations and the need for clinical studies on estimated energy requirement in the day-to-day life of people living with SMA. There is a need to understand the impact that dietary intervention has on new SMA phenotypes and further dietary clinical trials should be conducted to improve our current understanding.

Dietary guidelines cover recommended and optimal dietary intake that supports differences in sex, age, weight and disease, as well as the tolerable upper intake limit. There are very limited data available for dietary intake specific to SMA. Measurement of basal metabolic rate in Type 1 SMA is well understood, but there is a knowledge gap in estimating this in Types 2 and 3 SMA. There is a need to define the daily energy expenditure of people living with SMA, which must be defined by clinical studies that measure estimated energy requirements for total daily activities.

People living with SMA are more susceptible to dyslipidaemia (high cholesterol). One study has shown improvements in lifespan of people with SMA who have a high-fat diet, but another demonstrated improvements in those who have a low-fat diet. Expert experience suggests that people with SMA cannot tolerate more than 20–25% lipids in their total energy intake. There is a need for clinical trials to evaluate different diets, including those that incorporate either high- or low-fat intake.

There is currently a very limited understanding of carbohydrate intake in SMA. An impaired glucose profile is seen in 10% of people with SMA and is likely due to pancreatic defect.

The insulin profile of people with Type 1 SMA is generally poorer than those with Type 2 SMA. There is a need for clinical trials to evaluate tolerability of diets with a high carbohydrate volume in people with SMA.

A study in Italy of protein consumption in people with SMA found that protein intake was higher than recommended values by about 50%. No correlation was found between protein intake and fat-free mass, but it was instead correlated with fat mass. There is a need for *in vivo* studies of protein metabolism, considering faecal loss and urinary extraction, to better understand requirements for protein intake in people living with SMA.

Studies have demonstrated the usefulness of vitamin D, calcium and magnesium to improve body composition and bone density in people with SMA. There is currently no clinical evidence for the use of creatine or carnitine in people with SMA.

Recommendations include special consideration of fibre and fluid needs to address GI motility, types of feeding and food consistencies, timing of meals, drug–nutrient interactions and psychological issues.

There is a need for clinical studies to evaluate day-to-day energy requirements specific to people living with SMA to inform the development of clear nutritional guidelines

Nutrition in check:
Measuring and assessing swallowing and feeding in SMA

Moderator: Dr Tina Duong
Panel: Prof. Katlyn E McGrattan, Prof. Eugenio Mercuri

Overview

This session provided an overview of the tools available to measure swallowing and feeding functions in SMA, and how these are applied in clinical practice. The speakers explored some of the methods used to measure swallowing and feeding in SMA, including videofluoroscopy as the gold-standard approach for assessing swallowing function in neuromuscular disorders, and the Oral and Swallowing Abilities Tool (OrSAT), its rationale and the importance of patient-reported outcomes (PROs) in measuring swallowing function in Type 1 SMA. The speakers also presented case studies to demonstrate the advantages and limitations of certain swallowing assessments in clinical practice.

Key points and unmet needs

Key points

The importance of swallowing assessments

- Safe swallowing is one of the most important aspects to consider in SMA, as bulbar dysfunction can result in aspiration, infections and sometimes death
- Aspiration can sometimes be missed during examinations that are not performed properly
- Even mild deficits in swallowing are clinically relevant, because if untreated, they can progress to profound deficits

How to measure swallowing

- Swallowing can be evaluated using clinical assessments, instrumental assessments and functional outcome assessments (including PROs)
  - Each have strengths and weaknesses, and videofluoroscopy is considered to be the gold standard due to its standardisation and reliability
  - Functional outcome assessments should be used in conjunction with clinical assessments
  - PROs should be considered in combination with clinical assessment of swallowing to prevent bias if individuals have become accustomed to reduced function

When to measure swallowing

- Ongoing assessment is important, particularly to build the relationship between clinicians and people with SMA so that clinicians have a better idea of when to intervene. Despite observed improvements, deficits may remain, and the absence of clinical symptoms may not eliminate the need to continue monitoring

Factors that may impact swallowing ability

- Fatigue is more common in infants who have obvious signs of swallowing difficulties than those who do not
- Children who are treated early are less likely to develop serious bulbar dysfunction than untreated individuals. It is important to note the possibility that these children may still develop minor problems

Nutrition in check:
Measuring and assessing swallowing and feeding in SMA

Key points and unmet needs

Unmet needs

- To collect and disseminate real-world data as an evidence base for new guidelines on swallowing and assessments of bulbar dysfunction
- To develop more tools and increased access to existing tools that have been specifically developed for assessing swallowing in SMA
    - There is a need for a rollout of standard protocols for all key assessments, so that clinicians follow the same recommended procedures
    - There is a need for tools to be developed that measure fatigue
- To implement regular assessment by speech therapists
    - In many centres, after implementation of a disease-modifying therapy (DMT), it is often only those who show obvious signs and symptoms of swallowing dysfunction who are regularly assessed

Speaker presentation: Prof. Katlyn E McGrattan

Prof. McGrattan discussed the strengths and limitations of swallowing assessments for SMA.

Prof. Katlyn E McGrattan outlined the three primary categories of swallowing assessments: clinical, instrumental and functional. Each have strengths and limitations for interpreting different results.

Clinical assessment

During the clinical assessment of swallowing, visual observations are conducted for sucking (frequency, strength and length of time), swallowing (any visual signs that swallowing is challenging, e.g. coughing or choking), breathing (stopping feeding to take a pause) and intake (how much was eaten/drunk and length of time). There are new technologies becoming available that can measure sucking pressures through the course of a feed, which can pick up useful information.

Instrumental assessment

Videofluoroscopic swallowing studies are considered to be the gold standard method, due to the standardised approach in their execution and interpretation. This examination provides the clinician with a snapshot of around 3 minutes of eating time in total, so it can be challenging to assess the impact of fatigue.

Fibre optic endoscopic evaluation of swallowing can be an uncomfortable procedure. This examination can be challenging to interpret due to 'white out' of the camera image when the individual swallows. There are no standardised metrics for assessment of fibre optic endoscopy, which can introduce reliability concerns.

A key aspect of swallowing studies is the identification of impaired muscle groups so that appropriate targeted treatment can be used. If an individual eats minimally by mouth, it is important to know which muscles are impaired, and therefore which to rehabilitate, so that different therapeutics can be explored.

Functional outcome assessments

Prof. McGrattan shared examples of an oral intake scale that categorises individuals according to eating/drinking and modifications, and PROs, which are critical in capturing what is relevant to the person with SMA. There is a need for PROs to be combined with clinical assessment to prevent bias if individuals have become accustomed to reduced function.

Prof. McGrattan explained that the key to managing dysphagia (swallowing problems) is a combination of both X-rays and symptom assessments. Dysphagia is a clinical complaint regardless of whether it is observed in a swallowing study or not. Deficits may still remain even if improvements are observed, and the absence of clinical symptoms may not eliminate the need to continue monitoring. Ongoing assessment is important, particularly to build the relationship between clinicians and people with SMA so that clinicians have a better idea of when to intervene.

It is important that assessment of swallowing ability in people with SMA is ongoing so that intervention can be considered when changes are recognised

Click to read about aspiration problems

Click to read about considerations for clinicians conducting videofluoroscopy

Speaker presentation: Prof. Eugenio Mercuri

Prof. Mercuri discussed available tools for the assessment of swallowing and feeding in SMA clinical practice.

Prof. Mercuri discussed practical measures that can be used in clinical practice for the measurement of swallowing and feeding in people with SMA. It is important to consider differences in presentation of swallowing function in different types of SMA, including in those who have received DMTs. There is a need to focus more on fatigue when measuring swallowing and feeding, which has historically been underestimated in SMA, and it is important to understand the impact of new DMTs on swallowing function.

Survival prognosis of children with SMA has improved in recent years and assessments of swallowing function are becoming more common; the trajectory of swallowing dysfunction has changed dramatically. Standards of care suggest that regular assessment by speech therapists must be performed, but this is not always possible. In many centres, after implementation of a DMT, it is often only those who show obvious signs and symptoms of swallowing dysfunction who are regularly assessed.

It is important to collect and disseminate real-world data as an evidence base for new guidelines on swallowing and assessments of bulbar dysfunction. There are few tools available that have been specifically developed for assessing swallowing in SMA, especially with regard to the emergence of new phenotypes and recently introduced DMTs.

The tools available to assess swallowing function are not always appropriate in the real-world setting, and many that have been developed are not suitable for use in newborns and infants. Clinicians make an effort to conduct assessments as best they can, but these are not done in a structured or systematic way. Fatigue, which is a major challenge for people living with SMA, is often overlooked by the tools designed to assess swallowing and bulbar dysfunction.

The OrSAT is a tool specifically designed to record structured information on different aspects related to oral, swallowing and feeding abilities in infants with Type 1 SMA. The OrSAT can be used by a clinician or therapist experienced in neuromuscular disorders and does not replace the need for speech assessments, but offers a more structured format and scoring system for clinical history assessments.

There is a need to improve the accessibility of specific tools that can systematically assess swallowing in SMA. The use of a structured assessment, such as the OrSAT, helps to detect and record partial changes in the swallowing abilities of infants with Type 1 SMA over time. Particular attention should be paid to the assessment of fatigue during nursing; fatigue represents the most frequent finding in infants who have obvious signs of swallowing difficulties versus those who do not. A multidisciplinary team, involving speech and language therapists, is required in clinical services to adequately support children with SMA.

The role that fatigue plays in swallowing ability should not be underestimated and should be incorporated into the tools designed to assess swallowing and bulbar dysfunction in people living with SMA. These tools must be made accessible to the whole community

Click to read more about the OrSAT

Advances in nutritional care:
Best practice sharing

Moderator: Prof. Giovanni Baranello

Panel: Dr Tina Duong, Dr Yasemin Erbas, Prof. Tim Hagenacker, Prof. Katlyn E McGrattan, Prof. Valeria Ada Maria Sansone, Prof. Ludo van der Pol

Overview

This session provided an overview of ongoing country-specific initiatives to improve nutritional care for people living with SMA and discussed geographical differences in standards of care (SoC). The speakers provided an overview of current guidelines or SoC for nutritional care in SMA in their country and summarised the aims of their nutrition-focussed initiatives and the approaches taken. The initiatives presented included a study on amino acid diets in people living with SMA; a study on the timing and nature of swallowing deficits in people living with untreated Type 1 SMA; and a third study on the comparative effects of exercise and nutrition on muscle mass, muscle strength and physical performance in adults with sarcopenia. Further initiatives included integration of a multidomain protocol, clinical consequences of reduced maximal mouth opening (MMO) and results from the European Patient Expectation Survey.

Key points and unmet needs

Key points

Local initiatives

- In the absence of formal guidelines, Great Ormond Street Hospital (GOSH) in the UK has developed a strategy for nutritional management of children with SMA, encompassing dietician referrals, engagement of community services, standardised and regular nutritional assessments and calculations of nutritional requirements on an individual basis
- A consortia of speech pathologists in the USA have implemented initiatives to provide training and resources to clinicians on bulbar assessments and disseminate the latest research
- In Germany, interdisciplinary guidelines have been developed on the treatment and management of SMA, which cover nutritional management according to motor situation

Recent research

- A small pilot study has shown the potential benefits of the amino acid diet on GI symptoms. Larger studies are needed to assess this further
- A pilot study has indicated that swallowing studies should be performed upon diagnosis for baseline function and then every 2–3 months thereafter
- A study has shown that not only the pharyngeal aspect, but also tongue strength, mouth and jaw opening play a role in mastication (chewing) and swallowing and can be impacted in SMA
- Results from the European Patient Expectation Survey in SMA (EUPESMA) 2021 survey on nutrition showed that eating and swallowing abilities and nutrition vary greatly across the SMA population

Advances in nutritional care:
Best practice sharing

Key points and unmet needs

Key points (*Continued*)

SoC recommendations and requirements

- A multidisciplinary approach is optimal for SMA care; this should ideally include speech pathology and bulbar assessments, due to the frequency of bulbar deficits in people with SMA
- Trunk stabilisation is an important aspect in SMA as this allows effective respiration, eating and swallowing. Physical activity is SoC in SMA as it improves strength, balance, range of motion, endurance, physical well-being and oxidative capacity
- There are certain scales and assessment tools that are applicable for use as screening for swallowing/bulbar deficits in adults with SMA, including PROs; however, there is no SoC regarding these

Unmet needs

Guidelines

- Clear guidelines that can be easily implemented at the local level and guidelines about screening scales and assessments for adults with SMA
- A consensus is needed on what additional measures should be used to assess mastication and swallowing, as this is still a major issue in SMA and current measures can differ in objective or subjective perceptions. Although less prevalent in Type 2 and 3 than in Type 1 SMA, mastication and swallowing problems are still an issue and may be underestimated
- A consensus is needed on the best way to prevent reduced mouth opening in SMA

Diet and exercise

- Further studies on the amino acid diet, as the SMA community feel very strongly that this approach is beneficial
- Studies evaluating the combined effects of nutrition and exercise in SMA
- Effective measurement of physical activity in SMA to estimate metabolic costs and thereby inform nutritional requirements

Services and reimbursement

- Inclusion of speech pathologists in multidisciplinary teams
- Even though there are established SMA expert centres in Germany, there is a need to focus more on certain domains, such as bulbar function
- Only a small proportion of people with SMA are fully reimbursed for services and materials for nutrition, and the vast majority do not have access to the care they need to optimise their feeding situation

Speaker presentation: Prof. Giovanni Baranello

Prof. Baranello discussed Great Ormond Street Hospital's approach to nutritional management.

Prof. Baranello shared his experience of nutritional management at GOSH for people with SMA. There are no clear guidelines for nutritional management, and the existing guidelines can be difficult to implement at local level. A strategy for nutritional management for people with SMA has been developed at GOSH, based on the experiences accumulated there as well as global experiences reported.

GOSH have a dedicated referral to dietitian immediately after SMA diagnosis, especially in Type 1 SMA as these infants are monitored more regularly than those with other SMA types (every 6 months). People with Types 2 and 3 SMA have more individualised monitoring schedules. GOSH rely on local community services as it is important to engage them in the management of children with SMA.

Nutritional assessments include detailed anthropometry assessment and recording of dietary intake (3-day or 7-day diaries), as well as discussions with people about their eating habits. Limited physical activity in SMA means that most energy is used in basal metabolic rate. GOSH use predictive equations to calculate energy requirements and, for some cases, a calorimetry machine for individualised calculation. GOSH have implemented a standardised nutritional assessment that incorporates body composition, GI symptoms, use of indirect calorimeter (to get a precise and individualised requirement for calories, especially if people are overweight) and the collection of information on family eating habits, which can influence nutritional management.

There is a need to define how best to manage nutrition in SMA. Some families attempt to address this by themselves due to limited support from healthcare professionals. There has been strong support by people with SMA for the amino acid diet (branch chain amino acids, high carbohydrates and low fat), so GOSH started focus groups with families to better understand the perceived benefits. After receiving the amino acid diet, people with SMA reported improvement in airway secretions and respiratory management, constipation, reflux, strength, fatigue and sleep. This may have a considerable impact on quality of life for people living with SMA.

Standardised nutritional assessments that can be implemented at the local level, like the one developed by Great Ormond Street Hospital, need to be established within guidelines and distributed to all centres that care for people living with SMA

Click to read about the amino acid diet pilot study at GOSH

Speaker presentation: Prof. Katlyn E McGrattan

Prof. McGrattan discussed management of bulbar and swallowing dysfunction in the United States.

Prof. Katlyn E McGrattan shared her experience and perspective on management of bulbar and swallowing dysfunction in the US. Management of SMA in the US comprises multidisciplinary teams covering respiratory, neurological, motor control and nutrition factors for optimal care of people living with SMA. Where deficits are identified, consult services (e.g. speech therapists) are looped in for evaluation to communicate their findings to the team.

Swallowing/bulbar problems are pervasive across people living with SMA. Historically, all untreated people with Type 1 SMA experience dysphagia (swallowing problems). Centres in the US are currently working to integrate speech pathologists into the multidisciplinary team to bring bulbar assessment into the SMA clinic. Very few speech pathologists have the right expertise, and thus a key initiative is underway to help train clinicians (through resources, webinars and up-to-date research) and focus on collating more evidence through clinically relevant studies.

Data from a pilot study have shown degradation in swallowing over time in untreated people with SMA. These data were used to develop guidelines that recommend conducting a videofluoroscopic examination as soon possible (after newborn screening or diagnosis) to assess functional and physiological impairments at baseline.

The guidelines recommend:

- A follow-up videofluoroscopy is conducted every 2–3 months, regardless of baseline results, as people with SMA may experience complete loss of swallowing by 3 months of age
- Monitoring is conducted for the first 9–12 months of life, as the study showed that degradation can occur up to this age

A full study is in progress; guidelines are expected to be updated based on the evidence collected.

Speech pathologists should be trained in bulbar assessment of people with SMA so that they have the right expertise to offer as part of a multidisciplinary team

Speaker presentation: Dr Tina Duong

Dr Duong discussed the impact of nutrition on exercise in SMA.

Dr Tina Duong discussed the role of exercise in SMA and the impact of nutrition on a person's ability to exercise. There is a need for further studies to evaluate nutrition and exercise in parallel, as the metabolic costs of physical activity need to be better understood in order to advise nutritional requirements for people living with SMA.

Trunk stabilisation, posture and breathing can affect a person's ability to eat and digest food, as well as contributing to fatigue. Postural control relies on different systems working together (musculoskeletal, neuromuscular and cardiopulmonary). The accepted theory is that the trunk is a force of stabilisation – the spine, ribs, diaphragm and intercostal muscles create pressure that allows effective respiration, eating and swallowing.

Nutritional requirements may need to be changed in order to meet exercise needs. It is recommended that moderate-intensity exercise is used to improve strength, balance, range of motion, endurance and physical well-being.

A Cochrane review found that there are very few randomised controlled trials exploring the effect of physical exercise training in SMA, and most evidence has come from single observational studies. Existing literature shows that resistance training is safe, feasible and tolerable in people with SMA and helps to improve oxidative capacity.

No studies have been conducted that have looked at nutrition and exercise together in SMA. A meta-analysis of people with sarcopenia (muscle loss) studied the effects of exercise alone, nutrition alone and exercise plus nutrition together. Exercise improved gait speed, muscle strength and physical performance. Sensitivity analyses showed that combination treatment (exercise plus nutrition) was most sensitive to change in muscle mass.

Poor nutrition can contribute to obesity, which can significantly impact physical function. Current nutritional recommendations lack advice on the nutritional requirements for maintenance of muscle mass and changes required to support physical activity.

Metabolic cost of physical activity must be defined, including for people with limited mobility.

- Indirect calorimetry is understood to be the best way to measure metabolic rate; physical activity questionnaires are easier to perform, but lack evidence-based support
- Physical activity monitors can quantify the level of activity but may not offer the best solution for people who are non-ambulatory or weak walkers

There is a current need to measure physical activity in those with SMA so that metabolic costs can be calculated to inform nutritional recommendations.

Nutrition and exercise need to be studied in parallel to better understand the metabolic costs of exercise and help improve physical function for people living with SMA

Speaker presentation: Prof. Valeria Ada Maria Sansone

Prof. Sansone discussed real-world data collection and exploratory measures introduced by the International SMA Consortium to provide more data on mouth opening, swallowing ability, the strength of lip and tongue muscles and neurophysiology of muscles in the face in people with SMA.

Prof. Sansone discussed the need for exploratory measures in nutritional care. Swallowing challenges are a prevalent issue and dysphagia (swallowing problems) is one of the main causes of death in people with neuromuscular diseases. Swallowing involves input from muscle and posture as well as motor neurons. DMTs are having a positive impact, but more data are needed to provide physicians, regulators and people living with SMA with the confidence they need to understand and manage swallowing complications.

The International SMA Consortium (iSMAc) manages several worldwide data collection sites. More than half of children with Type 1 SMA were reported to use a percutaneous endoscopic gastrostomy (PEG) tube in 2017 (N=60); this proportion was the same in 2022 (N=90), likely because the same children continued to use a PEG tube. In total, 44% of children reported no problems with oral feeding. People living with Types 2 and 3 SMA reported fewer problems than those with Type 1 SMA, but challenges with oral feeding were still prevalent in this population.

Some Italian sites that are part of the iSMAc have introduced exploratory measures to provide additional data on the following:

1. Mouth opening
2. Time taken to swallow a cracker (to assess ability to bite, number of bites and how food is pushed into the pharynx)
3. Strength of lip muscles and tongue (how these are impacted by SMA and how they are influenced by treatment)
4. Neurophysiology of muscles in the face (some muscles can begin recovering with treatment. Surface electromyography is used to assess any changes in neurophysiology and collect data)

Further data on the impact of DMTs on muscle input and posture are needed to provide the community with the confidence they need to understand and manage swallowing complications

Click to read about results from a study conducted using the exploratory measures introduced by iSMAc

Speaker presentation: Prof. Tim Hagenacker

Prof. Hagenacker discussed German interdisciplinary disease management guidelines for SMA.

Prof. Hagenacker discussed current approaches in Germany, outlining the interdisciplinary guidelines for phenotype-specific disease management of people living with SMA. In Germany, there are established interdisciplinary disease management guidelines, which recommend phenotype-specific disease management, particularly for nutritional needs and associated risks. The guidelines are categorised by non-sitters, sitters, and walkers rather than by type of SMA.

Non-sitters require a swallowing examination by flexible endoscopy. Sitters require measurement of tongue pressure and bite force using a dental device to assess swallowing and mastication. Treatment modification may impact these functions, and data to support this are currently being collected. There is a need to further evaluate chewing ability in people with SMA, as well as bulbar function, which is a current focus of expert centres in Germany.

Guidelines in Germany recommend phenotype-specific disease management for people living with SMA, to inform nutritional needs and phenotype-associated risks

Speaker presentation: Prof. Ludo van der Pol

Prof. van der Pol discussed available tests for the assessment of bulbar function.

Prof. van der Pol discussed tests for assessing bulbar function and mastication (chewing) challenges experienced by people living with SMA. There are several tests for bulbar function, including the ruler test (to assess maximal mouth opening [MMO]), cracker eating, the 6-minute mastication test and videofluoroscopy. Neurologists are not trained to measure or assess bulbar function, instead speech and language therapists are needed to do this. The ruler test is simple to carry out and can provide information about mastication and swallowing. Reduced MMO is a common complication of SMA.

People with SMA experience inefficient chewing, reduced endurance and smaller mastication movement if they have reduced MMO, as well as reduced quality of muscle tissue. Feeding abnormalities are common in people with SMA (particularly those with Type 1, 2 or 3), which are partly due to reduced MMO, so there is a need to help prevent this where possible.

Click to read about a study which assessed MMO, mastication and swallowing in people with SMA

Reduced MMO and quality of muscle tissue have an impact on chewing efficiency and endurance for people living with SMA, and should be prevented where possible

Speaker presentation: Dr Yasemin Erbas

Dr Erbas discussed results from EUPESMA 2021, a survey on eating and drinking ability completed by people living with SMA and caregivers.

Dr Yasemin Erbas presented results from the EUPESMA 2021 survey on nutrition. The survey was completed by 1808 participants from 57 countries, with a broad distribution of people living with different types of SMA and their caregivers.
The survey evaluated the eating and drinking abilities of people with SMA, which are greatly impacted by motor function and the amount of support needed to feed. The survey highlighted that there are a lot of differences regarding eating and swallowing abilities and nutrition across the population of people living with SMA.

Swallowing and choking risk

A total of 84% of participants could swallow their own saliva, whereas only 63% of participants could swallow solid food. Overall, 25% of participants reported considerable secretions that were difficult to manage; 30% of participants reported high risk of choking, of whom almost 60% reported having experienced at least one life-threatening event relating to choking. Choking risk has an impact on independence and daily life: over half of participants said the possibility of choking negatively impacts their daily life, and a large number do not go out by themselves because of the risk of choking.

Use of feeding tubes

Use of a feeding tube was reported by 12% of participants. The reasons given for this were broad, including swallowing problems, digestive problems, fatigue and weight issues.

GI issues

65% of participants report having experienced GI issues. Constipation was reported by 42% of participants, but other frequently-reported issues included difficulty chewing, gas, low appetite and acid reflux.

Weight

Doctor assessment of weight showed that half of participants were either under- or overweight. A blended diet was the most common, but other diets were used by some.

Dietician/nutritionist support

A total of 62% of participants had never had help from a dietician or nutritionist. Those that did have help commonly reported that dieticians and nutritionists did not have extensive knowledge on the needs of people living with SMA.

Impact of eating on well-being

Most participants reported having a positive attitude. For many, their eating situation did not affect well-being or daily life, but some participants felt that their family life was impacted. Eating situation had a negative impact on self-esteem for some participants and made them feel uncomfortable eating in public.

Reimbursement

Many participants reported that they did not have reimbursement for services and materials for nutrition: 60% of participants did not have any reimbursement and 13% had some. This means that only 16% of participants were fully reimbursed, and the majority did not have access to the care they need to optimise their nutrition.

The EUPESMA 2021 survey highlighted a large number of differences regarding nutrition, as well as eating and swallowing abilities across the population of people living with SMA

Aiming to standardise nutritional care and support in SMA

Moderator: Prof. Eugenio Mercuri

Panel: Mrs Sara Almroth, Prof. Giovanni Baranello, Prof. Simona Bertoli, Dr Ramona De Amicis, Dr Tina Duong, Dr Yasemin Erbas, Dr Nicole Gusset, Prof. Tim Hagenacker, Prof. Katlyn E McGrattan, Prof. Valeria Ada Maria Sansone, Prof. Ludo van der Pol

Overview

In this session, the panel discussed the unmet needs and gaps in nutritional care and assessments in SMA, and how the SMA community can start to address some of these needs. The panel also discussed how standards of care (SoCs) regarding nutrition in SMA have evolved since disease-modifying therapies (DMTs) have become available.

Key points and unmet needs

Key points

Developments since DMTs have become available

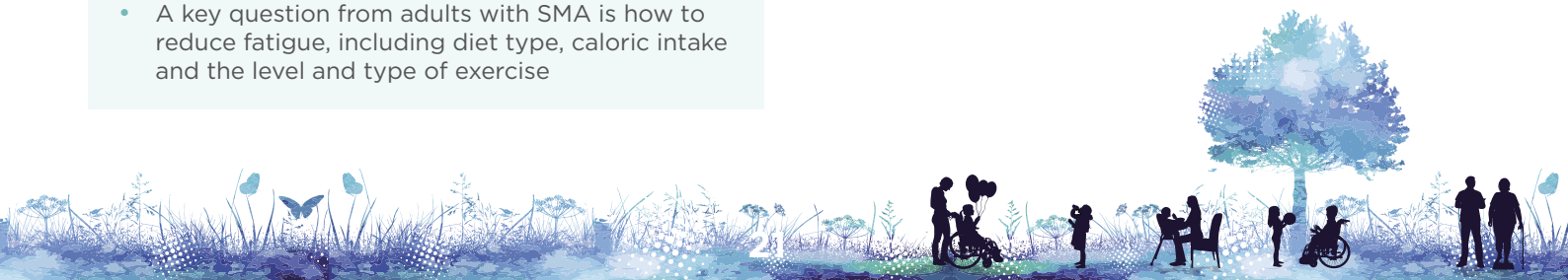
- The historic concept that all people with Type 1 SMA would have deterioration of bulbar function has changed with the introduction of DMTs. If treated early, people with SMA may not develop bulbar dysfunction
- In addition, the advent of DMTs has altered the previous principle that swallowing symptoms are progressive, and gastrostomy is irreversible. Now, the criteria for feeding via a tube are different to those used before DMTs were available, and gastrostomy can potentially be reversed with rehabilitation
- The approach to treating swallowing issues has changed completely. Swallowing problems in SMA are now treated in a similar way to other patients with dysphagia

Adults with SMA

- Adults with SMA are a heterogeneous population. Little is known about the disease course and how best to assess bulbar dysfunction
- A key question from adults with SMA is how to reduce fatigue, including diet type, caloric intake and the level and type of exercise

Standards of care

- When using feeding tubes, the risk of aspiration should be weighed against the potential benefits to maintain nutritional needs at night. However, tube feeding should not necessarily be initiated prophylactically
- One approach to improve SoC in the US setting is to create speech pathology centres of excellence that specialise in videofluoroscopic swallowing studies, which can be accessed via telehealth
- SoC should be followed, but treatments should also be personalised based on individual needs and expectations, especially in specialist centres
- More data are needed and should be collected before changing SoC, such as reimbursement or availability information and current unmet needs



Aiming to standardise nutritional care and support in SMA

Key points and unmet needs

Unmet needs

- To capture real-world data on the use of DMTs across a large spectrum of age and disease severity
- Nutrition recommendations for people with SMA in specific situations (e.g. in the peri-operative period, during infections)
- Speech pathologists with expertise in the use of videofluoroscopy to assess bulbar function in people with SMA. More speech pathologists need to be trained in this field
- Increased understanding of how adults with SMA can reduce fatigue, including diet type, caloric intake and the level and type of exercise
- Increased understanding of the variables involved in someone who is overweight becoming underweight
- Increased understanding of how quality of life (QoL) can be improved in people with profound swallowing deficits who are gastronomy tube dependent, being managed with suctioning, and with minimal oral intake, and how much room there is for improvement with rehabilitation

Speaker presentation: Prof. Eugenio Mercuri

Prof. Mercuri discussed the impact of DMTs on bulbar function for people with SMA.

Prof. Mercuri discussed the use of DMTs for people with SMA who are diagnosed early and for those who are not. Prof. Mercuri explained that before DMTs became available, all people living with Type 1 SMA experienced deterioration of bulbar function. Because of this, it is important to intervene as soon as possible. DMT studies suggest that if people are treated, there is a high possibility of maintaining bulbar function; this result is supported by results from long-term studies.

DMTs are now used across a larger spectrum of age and disease severity in the real world than those assessed in clinical trials, including in people who have already developed bulbar dysfunction. Improvements have been seen in some people who undergo gastronomy rehabilitation. For some people, gastrostomy tubes are no longer needed, and some are able to be partially fed by mouth. Different criteria to assess the need for tube feeding may be considered now than those used previously. The degree of functional impairment at the time of treatment initiation can help to predict the progression of swallowing ability. There is a need to change the management of bulbar dysfunction, be more systematic in the approach to assessment and rehabilitation, and to capture real-world data.

Panel discussion

Prof. Giovanni Baranello explained that preserving bulbar function is attracting more attention among HCPs working in SMA. In the past, for more severe forms of SMA, there was no specific approach to maintaining function. Now, speech and language therapists are focussing more on maintaining bulbar function, which in turn will provide people with SMA with the joy of eating; this is likely to have a significant psychological benefit. There is a need to build evidence and experience around progression of bulbar dysfunction and define other approaches to treatment that can be adopted.

In the US, the approach to a single episode of aspiration or coughing was very aggressive as it would get worse with time and could be rapid. After new therapies became available, have approaches changed?

Prof. McGrattan explained that the approach has changed completely. Swallowing problems in SMA are now treated in a similar way to others with dysphagia. Instrumental assessment has never been conducted in healthy infants, so we do not know what can be considered as abnormal. Swallowing studies in people who are newly diagnosed with SMA have shown that all participants have laryngeal penetration. This could be a normal finding as this population does not have any feeding functional issues. These people are not treated for the laryngeal penetration but are followed up to observe whether they develop any issues that require intervention. Some cases remain the same, whereas others experience respiratory infections, so treatment (e.g. thickened liquids) is provided, and the deficits resolve.

Dr Erbas explained that she sees a lot of variability in care centres for managing aspiration. As a parent, Dr Erbas would not advise prophylactic use of a feeding tube, but it is important to educate parents on the symptoms of aspiration. Parents could be reluctant to accept that a feeding tube is needed, so it is key not to be too conservative in the approach as it is important to keep children safe.

Prof. Sansone explained that evaluating possible differences in care needs to be monitored and conducted in a safe context. There is still a need for more data regarding when changes can be implemented and how they can be measured. Children may not have structured swallowing difficulty but can get tired easily, and fatigue may cause aspiration. Fatigue plays an important role in mastication and swallowing.

Dr Erbas explained that fasting can be very dangerous for some children. Without a feeding tube, it is difficult to feed every 4 hours. Feeding tubes can help at night.

Prof. Hagenacker suggested that in the future, special attention should be paid to nutrition during specific situations (e.g. peri-operatively and during infections). Hypoglycaemia is common in those with low muscle volume.

Dr Duong explained that there has been a change from use of a prophylactic tube being the standard method towards monitoring fatigue, growth charts and motor milestones, and discussing any deficits and concerns as a multidisciplinary team. There is a need to be more pervasive in screening.

Panel discussion

There are still unmet needs, but these needs have now changed following the introduction of DMTs. What are the unmet needs of adults with SMA?

Prof. Hagenacker explained that adults are not a homogeneous group, and it is important to be aware that there are many different phenotypes. People with SMA who have less severe disease should still be assessed for bulbar dysfunction, swallowing difficulties and nutritional needs. There is limited knowledge about disease course in adulthood (e.g. motor function of extremities), and there has been a lack of focus on bulbar dysfunction. There is a need to understand which phenotypes are at risk of developing bulbar or swallowing dysfunction, particularly in late adulthood, and how it can be assessed. There is not one single questionnaire or examination that will provide the necessary information across all adults with SMA.

Has the advent of therapies changed what you are doing in clinical practice?

Prof. Hagenacker explained that it is not known how many people with SMA who are untreated might not be aware of treatment options or do not want to be treated. Many people with SMA ask for nutritional information and for advice on their diet—there is a need to define an optimal, structured approach to sharing this information with people with SMA in clinical practice. Prof. Hagenacker shared that he expects this to change in the next couple of years.

Do we still have unmet needs with the advent of DMTs?

Mrs Almroth explained that a common question for people living with SMA is what they can do to increase their energy levels. Treatment for adults is not available in many countries, so it is even more important to address fatigue in other ways, such as changes to diet type, caloric intake and type of exercise.

Dr Duong explained that there is a need to understand how to individualise nutritional plans that consider change in activity levels, for example when people gain motor skills, increase endurance or during transitional periods (e.g. from adolescence to adulthood). There are challenges in measuring exercise and physical activity, therefore working closely with a dietician is important to better inform people with SMA of their nutritional needs. There is also a need to understand what variables are involved in someone who was overweight now becoming underweight.

How can we make sure that the research on diet and exercise is spread outside of tertiary care centres?

Dr Erbas emphasised the importance of the role of patient organisations to disseminate information, not only to reach clinicians and therapists in clinical trials but also the healthcare professionals who are directly involved in the everyday lives of people with SMA. It is important to involve patient organisations in research to disseminate findings.

Panel discussion

Is there a need for training and guidelines on videofluoroscopy?

Prof. McGrattan explained that telehealth can be used to access those with speciality training. There is a lack of speech pathologists with expertise in the use of videofluoroscopy to assess bulbar function in people with SMA. More speech pathologists need to be trained in this field. One approach to address this in the US setting would be to create speech pathology centres of excellence that specialise in videofluoroscopic swallowing studies, which can be accessed via telehealth. Prof. McGrattan explained that, for her, the question is how can we improve QoL in individuals with profound swallowing deficits who are gastronomy tube dependent, on suctioning and with minimal oral intake, and how much room for improvement is there with rehabilitation? This is currently unknown. In these people, X-rays may show some cranial nerve intact, some movement and some potential, so they could establish new neural networks, but there have been no studies that have looked systemically at rehabilitation.

Do we know enough to modify SoC, or are we still collecting evidence?

Prof. McGrattan explained that routine assessments are still really important but should be modified, so that people with all types of SMA receive a baseline assessment.

Dr Erbas explained that, with the use of DMTs, there are now even more differences between individuals, so decisions should be individualised, rather than based solely on SMA type. In addition to observations in the clinic, people's needs and expectations should drive how often assessments should be conducted and what to assess.

Prof. Mercuri agreed that more specialist centres are aware of SoC, but other centres with less expertise need guidelines and suggestions.

Prof. Sansone explained that there is a need to collect more information before future directions and recommendations can be considered.

Prof. van der Pol explained that it would be helpful to emphasise in future SoC that there is a need for reimbursement of studies in order to continue with further research.

Prof. Mercuri explained that it is important that unmet needs are addressed first. There are not enough data yet to change SoC.

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- **Mrs Sara Almroth** has no financial disclosures
- **Prof. Giovanni Baranello** is an investigator in SMA trials sponsored by F. Hoffmann-La Roche and Novartis Gene Therapies, and has received consultancy honoraria from Biogen, PTC Therapeutics, Sarepta Therapeutics, F. Hoffmann-La Roche and Novartis Gene Therapies
- **Prof. Simona Bertoli** has received honoraria for teaching and consultancy activities from F. Hoffmann-La Roche and Novo Nordisk
- **Dr Ramona De Amicis** has received honoraria for teaching and consultancy activities from F. Hoffmann-La Roche
- **Dr Tina Duong** is a board member of Cure SMA, and has received advisory and consultancy honoraria from Audentes Therapeutics, Biogen, Cytokinetics, Dyne Therapeutics, Edgewise Therapeutics, F. Hoffmann-La Roche, Pfizer Inc, Scholar Rock, Sarepta Therapeutics, Bristol-Myers Squibb and Novartis
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