Best Practice Recommendations:

therapy interventions for swallowing and speech in Children with Congenital Myopathy, Duchenne Muscular Dystrophy, Myotonic Dystrophy Type 1 and Spinal Muscular Atrophy Type 2

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Reading guide

These best practice recommendations consist of several sections. The general section describes the motivation, goal, target group, terminology and methodology. The methods describes how these best practice recommendations came about. The generic section and the sections about the four specific neuromuscular diseases describe the clinical presentation, a summary of the literature, considerations by the expert group and recommendations for SLT intervention. The recommendations are not equally specific for every disease. This is due to the varied experiences and consensus per disorder. For reasons of legibility, where 'he' is cited in the text, this refers to both 'he' and 'she', and where 'parents' is cited, this refers to 'parents/caregivers'. Where the term 'patient' is used, this can refer to 'patient', 'resident' and 'client (system)', depending on the context.

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1. General

1.1 Motivation

In children with a neuromuscular disease (NMD), also called muscle disease, problems with chewing, swallowing, speaking, saliva control and oral hygiene are common.[1] Depending on the muscle disease, there are various problems for which customised treatment is important. In addition to direct therapy (exercise therapy), indirect therapy (explanation, advice and adjustments) is also often provided. There is currently little evidence for these interventions[2], all the while there is a great need for more information about how to treat these problems. Parents of children with a muscle disease often indicate they need advice about what can be exercised, compensated for and adjusted.

In order to provide well-founded advice about chewing, swallowing, speaking, saliva control and oral hygiene, proper diagnostics and knowledge of underlying pathological mechanisms in these functions is required. To improve care, clinical reasoning is important so as to formulate a good treatment plan. An overview of the possible recommendations and exercises for SLT (SLT) in children with a neuromuscular disease might be able to increase the quality of care for these children. Such an overview was not available until now.

1.2 Goal

The goal of these best practice recommendations is to develop an overview of SLT interventions for children with congenital myopathy, Duchenne muscular dystrophy, myotonic dystrophy type 1 and spinal muscular atrophy type 2, based on consensus. These muscle diseases are seen most frequently within the range of muscle diseases in children. Experience in assessment and treatment for these diseases has been amassed by speech language therapists (SLT's) in rehabilitation centres and hospitals. SLT's can use this overview in their clinical reasoning in order to formulate an intervention plan. It also offers insight to other healthcare professionals and parents.

1.3 Target group

SLT's involved in the care for children with a neuromuscular disease are the primary target group of these best practice recommendations. Additionally, the recommendations may be informative to other involved healthcare professionals and parents. SLT intervention for these children requires specific skills. A specialised team at an (academic) hospital and/or rehabilitation centre is involved in the treatment of many children with a muscle disease. Given the rarity of most muscle diseases, having background knowledge and experience with treating a child with a muscle disease is not usual for SLT's working in a different setting. For parents it is important that the SLT is having sufficient expertise and knows what he is doing, or to acquire knowledge from a healthcare provider with this specific knowledge. It is important for the involved SLT to know his limitations and take the initiative towards peer consultation or referral, if necessary.

1.4 Terminology

1.4.1 Dysphagia

Eating problems may occur from the moment that a patient accepts food and liquids orally to when these enter the stomach. This is a broad term that can include food refusal, disruptive behaviour while eating, rigid food preferences, suboptimal growth and autonomy while eating.[3, 4] Swallowing problems are defined as problems with transporting food and liquids from the mouth to the stomach, in one or more phases of swallowing.[3, 5] For various reasons, eating and swallowing problems in children are different and more complex (and therefore more difficult to differentiate from each other) than in adults. First of all, children experience complex anatomical changes of the oral cavity and pharynx due to their growth. Additionally, their physiology changes, due to which the swallowing effort evolves from fully reflexive to partially voluntary. Dealing with gradually changing food substances and the acquisition of eating independently also plays a role. Children must continually adapt to these changing factors.[6-9] That is why these best practice recommendations use the umbrella term dysphagia when talking about problems with eating and swallowing.

1.4.2 Dysarthria

Dysarthria is a neuromuscular speech disorder caused by damage of the peripheral and/or central nervous system that affect the tone, strength and coordination of (a part of) the muscles that are used for speech. Problems occur in various partial aspects of speech, specifically breathing, phonation, articulation, nasal resonance and/or prosody.[10, 11]

1.4.3 Drooling

The problems with saliva control which we discuss in these best practice recommendations are about excessive drooling after the fourth year of life. Anterior drooling is when the saliva drops from the lips to the chin. When there is inadequate swallowing and the saliva spills over the tongue and runs towards the throat, this is called posterior drooling.[6, 12]

1.4.4 Oral hygiene

Daily care of the teeth and mouth with tools such as a toothbrush and toothpaste is called oral hygiene

1.5 Methodology

Due to the very limited evidence available, it was not possible to go through a regular guideline process, where all evidence and recommendations are weighed and graded with the help of the GRADE method (Grading of Recommendations Assessment, Development and Evaluation).[13] For this reason, a Delphi consensus study was conducted for the development of these best practice recommendations, whereby consensus was obtained about recommending interventions through expert opinion. The Delphi method is a phased method used worldwide for compiling group consensus from individual opinions about a specific topic.[14-16]

2. Methods

2.1 Stakeholder input

The draft of these best practice recommendations was submitted to a number of stakeholders in order to include the perspectives of patients, their parents and other healthcare disciplines. The comments received were incorporated into the final version of the best practice recommendations.

Stakeholders

- Dutch Association for SLT and Phoniatrics (Nederlandse Vereniging voor Logopedie en Foniatrie) (NVLF), subject content guideline panel
- Dutch Neuromuscular Disease Association (Spierziekten Nederland) (congenital and metabolic muscle diseases guideline panel)
- Duchenne Parent Project
- Ms A.M.B. van der Heul, MSc, speech language therapist and researcher SMA, UMC Utrecht
- Ms A. de Baaij-Daalmeyer, dietician, Radboudumc, Nijmegen
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- Ms S. van de Kamp, paediatric physical therapist, Klimmendaal Rehabilitation Center, Arnhem
- Dr M.J. Poelma, paediatric rehabilitation physician, Sint Maartenskliniek, Nijmegen

2.2 Methodology

Om tot consensus van experts over interventies te komen is een Delphi-procedure met Rand/UCLA-modificaties gebruikt.[17] In een aantal stappen, waaronder twee rondes met vragenlijsten en een consensusbijeenkomst, heeft een panel van experts een aantal interventie-opties voorgesteld bij een aantal casussen. Vervolgens hebben zij de mate van geschiktheid van deze interventies gescoord, gebaseerd op evidentie in de literatuur en hun ervaring als professional met de doelgroep. Specifiekere informatie over de werkwijze is te vinden in een wetenschappelijk artikel dat in voorbereiding is.

2.2.1 Practical implementation

Exploratory questionnaire (step 1)

A group of SLT's with experience in treating children with a muscle disease (experts) answered open-ended, broad and exploratory questions about methodology, advice and exercises in the area of dysphagia, dysarthria, drooling and problems with oral hygiene in 29 cases. These cases consisted of the most frequent symptoms and were categorised into four types of muscle diseases: Duchenne muscular dystrophy, congenital myopathy, myotonic dystrophy type 1 and spinal muscular atrophy type 2. These diseases were chosen because of their high frequency (within the range of muscle diseases in children) and because speech language therapists have gained the most experience with these children in rehabilitation centres and hospitals.

Literature search and second exploratory questionnaire (step 2)

After an analysis of the answers from the first questionnaire and an exploratory review of literature about interventions, the interventions identified were linked to the cases. In a second questionnaire round, the experts could see all of the suggested interventions from step 1 and the linked literature. They were then asked to rate the degree of appropriateness for each of the listed interventions on a 9-point Likert scale.

Consensus meeting (step 3)

During a consensus meeting, the panel of experts provided feedback about the results from step 2 for each case. The interventions for which there was consensus were verified by a show of hands. The interventions without consensus were discussed and revised in semi-structured group discussions. In order to assess the degree of appropriateness of the adjusted interventions, the experts were asked to rate this on a 9-point Likert scale. For appropriateness scores of 7 or higher for more than 75% of the panel, agreement was assumed and the intervention was included in the best practice recommendations.

Final best practice recommendations (step 4)

Under the editorship of Mieke Kooi-van Es, the research group processed the data into final best practice recommendations. During this step the data were first ordered by muscle disease and then by category (dysphagia, dysarthria, drooling and oral hygiene problems). Then a general description was added for each muscle disease (background), the encountered literature was described and the considerations from the consensus meeting were described (evidence and considerations). The encountered literature was also linked to the recommendations where possible. An introduction and methods were also written. Draft versions of the best practice recommendations were revised by means of review cycles and research group meetings. The participating experts were the last to review the draft version. Their comments were incorporated into the last draft of these best practice recommendations. The draft of these best practice recommendations was then submitted for review to the stakeholders, whose comments were incorporated into the final version of the best practice recommendations.

2.3 Financing

This research and product came about with the financial support of Johanna KinderFonds (Johanna Children's Foundation), Stichting Janivo (Janivo Foundation), the Damsté-Terpstra Fonds (Damsté-Terpstra Foundation) and the Christina Bader Stichting (Christina Bader Foundation). A number of sponsors also assisted with the layout: Ipsen NV, Hanssen Footcare, Proreva and Livit. The financial backers had no influence on the creation of the best practice recommendations.

3. Neuromuscular diseases

3.1 Background

There are around 600 different muscle diseases, with muscle weakness being the main symptom. Some of these occur in childhood. In the Netherlands, the diagnosis is usually done by a (paediatric) neurologist. After a clinical exam, a follow-up exam is conducted if a muscle disease is suspected. This exam can entail a heredity, blood (CK level) and/or clinical neurophysiology test (EMG/nerve ultrasound), imaging (muscle ultrasound/MRI scan) or a biopsy of a muscle or nerve.

The various types of diseases are sub-divided based on the location where the problem appears: diseases of the anterior horn cell, the nerve, the motor endplate (neuromuscular junction) or the muscle itself (see Figure 1). There are congenital and hereditary forms, slowly progressing and rapidly progressing forms.

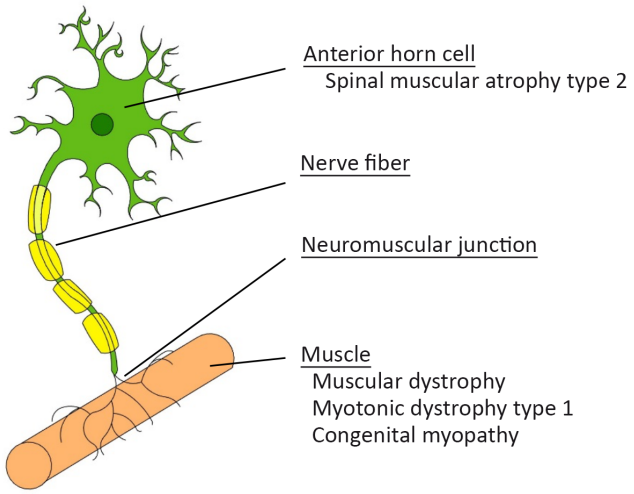

Figure 1. NMD classification based on anatomy (image: Tom Slegers)

3.2 Generic principles and collective considerations

Because treatment of children with a muscle disease requires a multidisciplinary approach, a (rehabilitation) physician, physician assistant and/or nursing specialist and occupational therapist, dietician and paediatric physical therapist or other care providers are usually also involved in addition to a SLT. One of these medical professionals coordinates the care.

Caregivers decide which intervention is needed with the help of clinical reasoning. Observation and testing take place based on the need for assistance and anamnestic data. Based on these data and the capacities of the child and parents, the most suitable intervention is validated. SLT intervention may entail explanation and advice, treatment (training), recommendation of aids and adjustments, referral and monitoring. Table 1 provides an explanation of these types of interventions.

Table 1. Explanation of intervention types

INTERVENTION	EXPLANATION
Explanation/advice	This concerns: - providing information and explanation to the parents, child and their environment about the normal development and the (possible) problems related to the child's muscle disease with regard to eating, swallowing and speaking - giving advice with regard to eating, swallowing and speaking (compensatory or training at home)
Training/treatment	This concerns: - integrated assessment and treatment (whereby assessment and treatment are integrated with a continuous interaction between the two) - training/treatment during direct SLT - multidisciplinary treatment (such as use of occupational therapy for determining the best sitting position during a meal) Because training and treatment frequencies depend on more factors than just the problem or symptom, no specific recommendations are given. We assume that explanation and advice is always given about the selected training or treatment and that transfer to home (or place of residence) always takes place.
Referral	This concerns a referral to or request for consultation of occupational therapy, paediatric physical therapy, dietetics or dentistry, for example, or for an instrumental swallowing assessment (such as a videofluoroscopic swallow study) within or outside of one's own treatment team, the institution or the practice.
Aids/adjustments	This concerns aids such as a slab, modified eating utensils and/or chair, food modifications, Augmentative and Alternative Communication (AAC). Food modifications concern the use of a thickening agent, modified consistencies and/or liquid nutrition. AAC include non-tech, low-tech and high-tech aids (https://www.asha.org/public/speech/disorders/aac/). Use of aids and adjustments is described under EXPLANATION/ADVICE. Therapy materials, such as a cork for stretching the mouth opening, are not considered aids. Aids can be provided by SLT's; this is frequently done in combination with occupational therapy.
Monitoring	The basic principle is that every child is screened by an NMD team or comes in for a check-up every 12 months. The check-ups described under this heading are check-ups in addition to the standard check-ups, aimed at current problems and interventions.

4. Congenital myopathy

4.1 Background information

Congenital myopathy (CM) is the umbrella term for a group of rare muscle diseases where a genetic mutation causes a defect in the structure of the muscle. The disease often occurs in young children, and sometimes the muscles are already weak at birth (floppy infant syndrome). These babies have severely delayed motor development. A CM diagnosis can also be made at a later age in childhood. These children start walking at a later age then normal and have difficulty running, climbing stairs and playing sports. The course of disease is not the same in every child; some children get worse while others remain stable. Due to the hypotonia, the face is often small, the palate is narrow and high and there may be a tented upper lip. In addition, a misaligned jaw may occur (underbite) during nocturnal mask ventilation.[18, 19] There may also be cardiac and/or respiratory problems or feeding problems. Scoliosis may also occur, depending on the type of CM. Examples of CM are nemaline myopathy, central core myopathy, congenital fibre type disproportion myopathy and myosin storage myopathy.[20, 21] Dysphagia often occurs in children with CM (60%).[1] Dysarthria occurs in 30% of the children.[1]

4.2 Dysphagia: evidence and considerations

Longer mealtimes can be stressful for the child and the family. They are caused by weakness of the chewing and swallowing muscles and may result in inadequate food intake and weight loss. Feeding through a feeding tube is recommended if the child cannot orally ingest the required daily quantities of food. Measurements of growth are recommended every 3 months and a dietician should be consulted annually.[21]

Due to muscle weakness in the oropharyngeal region, some children need multiple swallows in order to swallow a bite, especially with pureed and solid food. This is caused by weakness of the tongue and the tongue base. This in turn causes decreased retraction of the tongue base and reduced activity of the submental and pharyngeal muscle groups.[22, 23] Solid/thick liquid food is more likely to cause a residue (in the throat) after swallowing than thin liquid. This may create a risk for aspiration and pneumonia. Both the consensus group and scientific research indicate that thickened food is not always appropriate for children with CM, even though this is an oft-given recommendation for other target groups, such as children with cerebral paresis.[22] It is recommended that swallowing capabilities be monitored and the appropriate consistencies be properly examined.[24] If there are signs of possible risk full swallowing, a video fluoroscopic swallow study is recommended. The lack of coughing during observation of eating and drinking does not provide sufficient information. There may still be silent aspiration because the residue can easily leak into the airways.[22] In particular noisy or wet breathing and voice changes after swallowing are associated with hypopharyngeal residue.[9] Flexible Endoscopic Evaluation of Swallowing (FEES) may be indicated for older children but is not always possible and is not 'standard care'.[21] Tube feeding may be advised for severe, recurrent aspiration.[25, 26]

There is consensus that few specific skills can be trained in children with congenital myopathy. The intervention consists primarily of explanation, advice, aids and referral. A SLT may decide, however, to try out the advice in the therapy situation for a brief duration, such as testing which consistency or which bite size works best.

If there are lower respiratory infections not related to swallowing, then the medical causes will have to be examined before SLT intervention is initiated.

4.3 Dysphagia: recommendations

Table 2. Overview of possible SLT interventions aimed at dysphagia due to congenital myopathy

Anatomic characteristics, functions, activities and participation: possible problems	There may be difficulties elevating the hand to the mouth and there may be problems with maintaining posture. There may be weak oral muscles[27], due to which abnormal oral structures (malocclusion, such as open bite) may occur, due to which digestion of solid food may be difficult and swallowing problems may occur and residue may occur in the pharynx.[22, 28] This can result in long mealtime durations[21], weight loss[21, 27], recurring illness and recurring lower respiratory infections.[21, 27, 29]
Indications for intervention	- chewing problems - increased risk of choking - suspicion of silent aspiration - long mealtime duration - weight loss - (inexplicable) lower respiratory infections

	Oral (digesting and chewing)	**Pharyngeal** (swallowing)
Explanation/advice	EXPLANATION Provide information to the patient and his parents and give advice about: - normal eating and drinking processes - possible eating and drinking problems in children with CM - possible consequences and risks - the impact of posture - the possible interventions POSTURE ADVICE - provide interdisciplinary advice ADJUSTMENTS (for chewing more easily and transport) - softer, smoother, creamier consistency[24] - thick or thin liquid[24] - adjusted bite size - drinking water during and after eating - energy-rich food or supplemental liquid nutrition - meals not longer than 30 min - multiple smaller portions per day	EXPLANATION Provide information to the patient and his parents and give advice about: - normal eating and drinking processes - possible eating and drinking problems in children with CM - possible consequences and risks - the impact of posture - the possible interventions POSTURE ADVICE - provide interdisciplinary advice ADJUSTMENTS (easier swallowing) - softer, smoother, creamier consistency[24] - thick or thin liquid[24] - drinking water during and after eating - always eat and drink sitting down
Training/treatment	- integrated assessment and treatment: implement and evaluate adjustments	
Aids/adjustments	adjustments	
Referral	- dietician[21] (annually [standard]; when there are signs of weight loss or malnutrition)	- videofluoroscopic swallow study (when unsafe swallowing is suspected)[21, 26, 28, 30] - dietician[21] (annually [standard]; when there are signs of weight loss)
Monitoring	- evaluate advice and monitor problems - after 3 months	- after a videofluoroscopic swallow study (if appropriate), for evaluation of the findings/advice - evaluate advice and monitor problems[24] - after 1 month

4.4 Dysarthria: evidence and considerations

The only sources found where treatment of children with CM with speech problems is mentioned are the Consensus Statement on Standard of Care for Congenital Myopathies from 2012 (Wang et al) and an article by Bagnall et al. from 2006.[21, 26] According to both authors, problems with speech are caused by weakness of the oral motor muscles (bad lip closure), a weak voice, difficulty with breathing control and abnormal oral structures (malocclusion, open bite, facial deformity). Due to this, it may be difficult for the patient to produce bilabial sounds, and because of that compensations such as tongue-lip or tongue-tooth articulations are used. Sometimes tracheostoma is also a negative factor.[26]

Both Wang (2012) and the panel of experts state that SLT treatment is indicated if the speech causes communication problems. SLT by offering strategies for improving intelligibility, such as articulation training, respiratory training and supported communication, are ways that can be used according to the literature and the panel of experts. However, these are not always successful. The panel of experts mentioned that if articulation is trained, this must be trained in functional expressions. With nemaline myopathy it is known that articulation training or muscle strength training of the lips or palate do not improve muscle strength or muscle range, given the underlying neuromuscular weakness.[26] Both Wang et al., Bagnall, and the experts state that AAC may be needed, for example gestures, sound amplification or speech computers. In the presence of severe nasality, use of a speech prosthesis may be considered. Pharyngoplasty has been effective in some cases with respect to articulation, but there was no experience with this within the consensus group.[21, 26] Sound amplification is not indicated if the patient has severe dysarthria where the speech is very distorted.[26]

4.5 Dysarthria: recommendations

Table 3. Overview of possible SLT interventions aimed at dysarthria due to congenital myopathy

CONGENITALE MYOPATHIE- DYSARTRIE	
Anatomic characteristics, functions, activities and participation: possible problems	Weak oral muscles[27], possibly causing abnormal oral structures, malocclusion (e.g. open bite), difficulty closing the lips (tented upper lip), limited facial expression, limited respiratory control, a weak voice, hypernasal speech, difficulty articulating and nasal speech (dysarthria).[21, 26]
Indications for intervention	- inability to adequately express oneself - inability to make oneself adequately understood
Explanation/advice	Provide information to the patient and his parents and give advice about: - the relationship between the muscle disease and difficulty expressing oneself - communication advice to the child's/parents' environment - discuss AAC device options, introduce them and start the process[21]
Training/treatment	- respiratory/voice linking and articulation training[21, 30] in short, functional expressions/words - AAC training (gestures/pictograms/AAC devices)[21, 26, 30]
Aids/adjustments	- AAC devices[21]
Use of other disciplines	
Referral	
Monitoring	- evaluate advice - evaluate/adapt AAC - after 3 months

4.6 Drooling: evidence and considerations

Excessive drooling can be a characteristic problem for children with CM. This is often caused by bulbar and facial weakness, resulting in difficulty closing the lips and swallowing saliva.[21, 26] The problem of choking on saliva in particular is a good reason to initiate intervention according to the experts. Various treatment options, such as exercises for lip strength, are suggested in the literature, but there is no consensus on this either in the literature or by the expert group. Botox and surgeries on the salivary glands are not recommended but can be considered in individual cases. Medication can be effective but may have negative effects, such as causing more difficulty in airway clearance and constipation because this may result in thickened secretions. That is why great caution must be exercised with medication use.[21] The panel of experts is of the opinion that a team specialised in treating excessive drooling (a so-called saliva control team) can be of assistance when considering possible interventions.

4.7 Drooling: recommendations

Table 4. Overview of possible SLT interventions aimed at drooling due to congenital myopathy

CONGENITAL MYOPATHY - DROOLING	
Anatomic characteristics, functions, activities and participation: possible problems	Drooling due to bulbar and facial weakness, causing difficulty closing the lips and swallowing saliva. [21, 26] Drooling through atrophy of the tongue, whereby sometimes the lips can be closed and swallowing is possible, but with inadequate slurping due to insufficient strength in the tongue.
Indications for intervention	- the chance of choking on saliva (posterior drooling) - recurring lower respiratory infections
Explanation/advice	Provide information to the patient and his parents and give advice about: - anterior and posterior drooling - the relationship between drooling and open mouth - the risk of aspiration - the impact of food on saliva - the relationship with being ill - discussing possibilities of decreasing drooling - multidisciplinary advice about posture (sitting posture, head posture) - swallowing as regularly as possible - stimulating lip closure/lip activity
Training/treatment	- integrated assessment and treatment (influenceability) - practicing becoming aware of drooling - consciously practicing swallowing
Aids/adjustments	- a wristband or cloth for wiping away saliva - a bib or shawl - a reminder
Use of other disciplines	
Referral	- saliva control team with the question of whether there may be a treatment for the drooling
Monitoring	- evaluate advice and monitor problems - after 6 months

4.8 Problems with oral hygiene: evidence and considerations

In the consensus statement by Wang et al. (2012), referral to a paediatric dentist is recommended from one year of age. In addition, they advise brushing the teeth twice daily as soon as the children get their first teeth. Desensitising techniques can be taught to parents if the brushing is challenging. If necessary, adjustments can be made so that the children can brush their teeth independently in consultation with an occupational therapist or a dentist.[21] Starting from the age of 6-8 years, a child can be referred to an orthodontist in order to assess a possible malocclusion and a misaligned jaw (e.g. due to an excessively high palate). However, the severity and progression of the muscle disorder must be considered in the treatment by the dentist or orthodontist.[21] Wang (2012) writes the following about this: 'Extensive orthodontic treatment in children with very weak facial muscles is not recommended given the high recurrence risk'. Surgical procedures for severe malocclusion are not recommended due to the high risk of complications of the intubation and anaesthesia.[21]

During the consensus meeting it came to the fore that oral hygiene is important for the airways and the general health, which justifies intervention by a SLT. The SLT can provide explanation, advice and possibly integrated assessment and treatment. If the problems are significant or cannot be influenced, then referral to a specialised dentist is recommended.

4.9 Problems with oral hygiene: recommendations

Table 5. Overview of possible SLT interventions aimed at oral hygiene problems due to congenital myopathy

CONGENITAL MYOPATHY – PROBLEMS WITH ORAL HYGIENE	
Anatomic characteristics, functions, activities and participation: possible problems	Facial deformities, malocclusion (open bite), high palate, jaw contractures, bad oral hygiene, hypersensitivity in the mouth area, non-acceptance of teeth brushing.[21]
Indications for intervention	- problems with teeth brushing
Explanation/advice	Provide information to the patient and his parents and give advice about: - the relationship between the muscle disease and oral hygiene - the consequences of bad oral hygiene - making brushing teeth comprehensible and predictable (in steps, e.g. with the help of pictograms) - brushing teeth with an electric toothbrush
Training/treatment	- integrated assessment and treatment; problem analysis (what is the cause of the problem) and searching for a good approach/trying out of advice then transfer to/advice for home. - the child brushing his teeth in the presence of the parents, during which tips can be given about how to hold the brush, the brushing order (least sensitive places first) or the type of toothbrush or toothpaste
Aids/adjustments	- recommend a different type of toothbrush (e.g. smaller, electric) - use gauze or swabs - pictograms
Use of other disciplines	
Referral	- specialised dentistry (evaluation of teeth)
Monitoring	- evaluate advice and monitor problems - after 3 months

5. Duchenne muscular dystrophy

5.1 Background information

Duchenne muscular dystrophy (usually abbreviated as DMD) is a progressive diseases whereby the muscles become continually weaker. With DMD the muscle cells become increasingly damaged due to the lack of the protein dystrophin in the muscle cell wall. Dystrophin cannot be produced due to a genetic mutation of the X chromosome, something which affects primarily boys. Dystrophin ensures the strength and resilience of the muscle cell but also plays a role in the brain, which explains why boys can also have learning difficulties.[31] The damaged muscle cells are increasingly replaced by fat and connective tissue and eventually die off. The diagnosis is often made around age 5, although the initial symptoms already occur in the first year of life. Duchenne's course of disease is divided into various phases; see Bushby et al. (2010) for an overview.[32] Between age 8 and 12, boys lose the ability to walk and continue to get worse.[20, 33, 34] The heart muscle is also damaged, and ultimately almost all boys must be ventilated. The prevalence of dysphagia and dysarthria is 36% and 10%, respectively.[1] The oral muscles are affected and deteriorate as the patients become older.[35] Drooling does not occur or hardly occurs with DMD. There is no literature about the prevalence and treatment of drooling in DMD. It is conceivable for drooling to occur in combination with other problems, such as anatomical defects in the nose/mouth cavity or severe mental disabilities.

5.2 Dysphagia: evidence and considerations

Chewing problems may occur due to decreased bite force and decreased contact between the molars (including due to an enlarged tongue) and may cause decreased fragmentation (especially when biting hard and sticky food). It also becomes more difficult to move food through the mouth due to decreased mobility of the enlarged tongue. The larger food chunks can lead to complications, such as choking, due to residue after swallowing.[36, 37] Chewing problems in boys with DMD are typically observed starting around the age of 8.[28] Many boys modify their food in order to make it easier to chew: soft food, breaking hard or chewy food into small pieces, adding sauce and/or drinking water during meals.[38, 39] A study by Van Bruggen describes that chew training with chewing gum, (4 weeks, 3x per day) improves the chewing efficiency and helps keep the chewing stable (Note: Not for strength training). The recommendation is to have boys with DMD start low-intensity chew training in the ambulatory and late ambulatory phase. [36, 40]

If food remains stuck in the throat (pharyngeal residue of solid food), this is often caused by bad fragmentation in combination with weak pharyngeal cleaning. This increases the risk of choking. Explain about this and advice about food modifications in order to make swallowing easier, drinking water during and after the meal to clean the throat and the importance of sitting up straight after the meal can help.[38] For cumulative residue and penetration of food to above the vocal folds, the recommendation is to completely avoid solid, minced and mashed food and only use fluids and purees.[41]

Modified consistencies can result in lower intake of the required nutrients, such as iron and fibre.[42] That's why regular evaluations by a dietician are important.[36] The Dutch guideline on Duchenne recommends half-yearly monitoring of weight and annual check-ups with a dietician.

Tube feeding may be considered with moderate to severe dysphagia. This is done when interventions for improving oral caloric intake in the presence of malnutrition and maintenance of adequate hydration are insufficiently effective. In undernourished boys with DMD, tube feeding can lead to stabilisation or (in many cases) improvement of the nutritional status.[34]
In general, orthodontic treatment is offered for malocclusions, in combination with exercises for keeping the tongue in the mouth and promoting nose breathing (oral myofunctional therapy [OMFT]). However, there are no studies about the effect of these interventions in boys with Duchenne. These treatment options should be carefully considered, because the malocclusions are caused by the affected chewing muscles and hypertrophy of the tongue. The chance of relapse is high because the cause cannot be treated. A limited mouth opening sometimes occurs and may be an impediment to intake, oral hygiene and dental care. It's important to undertake measures for counteracting this change through training.

Gastro-oesophageal reflux occurs primarily in the non-ambulatory phases. In combination with swallowing problems, there is a chance of aspiration. Risk factors for reflux in boys with DMD are: oesophageal dysmotility, delayed gastric emptying, medication use (corticosteroids) and scoliosis. Acid inhibitors or proton pump inhibitors can be prescribed by a physician (gastroenterologist) as treatment, but these do cause side effects at times. Dietary changes can also help: more frequent small meals and eating less fat.[43]

Thickening liquids is often recommended in general for swallowing problems. Due to their weakness, it is easier for boys with DMD to swallow thin liquid food than thick liquid or solid food.37

5.3 Dysphagia: recommendations

Table 6. Overview of possible SLT interventions aimed at dysphagia due to Duchenne muscular dystrophy

DUCHENNE MUSCULAR DYSTROPHY - DYSPHAGIA	
Background	
Anatomic characteristics, functions, activities and participation: possible problems	The problems regarding eating, drinking and swallowing are progressive in nature. The initial symptoms may be: difficulty chewing and swallowing solid foods, a feeling that food remains stuck in the throat and long mealtime duration [44]. There may be difficulties raising the hand to the mouth, problems with maintaining posture, weak orofacial muscles[38], open mouth[38, 45], low-lying, wide tongue[36, 37, 45], tongue thrust[45], reduced jaw opening[35, 38-40], decreased bite force[35], weak chewing muscles[35, 39], less force when biting off/chewing hard foods[36, 37], difficulty with bolus formation and transport, decreased fragmentation of food[36, 37, 40], swallowing multiple times for 1 bolus (especially with solid food), a feeling that food remains stuck in the throat/residue[36-38], more residue with solid food[37, 38], coughing during the meal, weak cough force[41], choking, feeling of suffocation[36, 37], reflux problems. The above aspects can cause problems at the social level and in terms of quality of life[39], needing help when eating (especially in the evenings)[38], leaning over sideways when raising the arm, malocclusions of the teeth[35, 38, 39, 45,] insufficient oral intake[38], decreasing BMI (Body Mass Index)[38], decreased appetite, weight loss, malnutrition, dehydration, increased risk of penetration and aspiration[9, 38] pneumonia due to aspiration

DUCHENNE MUSCULAR DYSTROPHY - DYSPHAGIA		
Indications for intervention	- difficulty bringing food to the mouth - posture problems when eating/drinking - mealtime duration is longer than 30 min. - decreased appetite and pleasure from eating (due to the energy it costs) - decreased jaw opening - chewing problems - food getting stuck in the throat - coughing when eating or drinking - increased risk of choking - difficulty coughing up - pneumonia - reflux - decreasing BMI GOALS - safe eating and drinking - saving energy (maintaining sufficient energy for other activities) - shortening the meal duration[38] - sufficient intake	
	Oral (digesting and chewing)	**Pharyngeal** (swallowing)
Explanation/advice	EXPLANATION Provide information to the patient and his parents and give advice about: - normal eating and drinking processes - possible eating and drinking problems in boys with Duchenne - limitations in bite force, chewing, swallowing - limitations and dealing with independence - possible consequences and risks - the impact of posture - the importance of sufficient food and liquid intake with the least possible loss of energy and strength - the importance of continuing to chew - the importance of keeping the oral musculature active - discussing liquid nutrition and/or (partial) tube feeding[34, 44] - possible interventions	EXPLANATION Provide information to the patient and his parents and give advice about: - normal eating and drinking processes - possible eating and drinking problems in boys with Duchenne - possible consequences and risks - risks of choking and suffocation Give information what to do in the event of choking and suffocation[38] - the effect of the decreased functioning of the muscles when swallowing - discussing liquid nutrition or (partial) tube feeding[34, 44] - the impact of posture - possible interventions

DUCHENNE MUSCULAR DYSTROPHY - DYSPHAGIA		

Explanation/advice	POSTURE) ADVICE
	- provide interdisciplinary advice
	- to improve the jaw opening
	- in combination with arm support or seating facility (in connection with fatigue and independence)
	- have the patient chew gum often[40]
	
	ADJUSTMENTS (for more easily chewing and transport)
	- softer, smoother, creamier consistency (costs less energy, is easier to increase intake)[38]
	- thick or thin liquid
	- drinking water during and after eating to facilitate transport and clean the mouth and throat[37, 38]
	- maximum meal duration: 30 minutes[38], otherwise further modify consistency, give more breaks, give calorie-rich food (to prevent malnutrition and save energy)
	- modify the bread: make it creamier (e.g. with a creamy topping) and cut off the crust.
	- modify the food consistency such as making it easier to chew or (enriched) thick or thin liquid (entire or part of the meal)
	- offer smaller pieces for more control when chewing.[36]
	- calm eating situation
	- chewing advice: being aware of proper chewing for improved bolus formation
	- ensure enough breaks during meals so that there is energy for the next meal
	- decrease time pressure
	- utensil recommendation: small utensils due to small jaw opening and decreased strength in the hands/ arms
	- longer straw or elevated cup
		POSTURE ADVICE
		- provide interdisciplinary advice
		
		ADJUSTMENTS (easier swallowing)
		- softer, smoother, creamier consistency[38]
		  - decreased risk of choking and residue[38]
		  - less strength needed for chewing and swallowing
		  - ensures easier transport and swallowing[38]
		- thick or thin liquid
		- avoiding hard and chewy consistencies due to risk of choking[36, 37, 41]
		- drinking water during and after eating to facilitate transport and clean the mouth and throat[37, 38]
		- smaller bites or pieces for more control during swallowing
		
		- advice on how to cough properly (in combination with paediatric physical therapy)
		- discussion of parents sounding the alarm if there is more frequent choking

DUCHENNE MUSCULAR DYSTROPHY - DYSPHAGIA		
Training/treatment	integrated assessment and treatment: implement and evaluate adjustments - together with the occupational therapist, look at the sitting facility in connection with improving the initial posture/sitting position - training with chewing gum[40] - practicing with arm support while eating - stretching the jaw opening	- integrated assessment and treatment: implement and evaluate adjustments - practicing with chewing as best possible, collecting and swallowing smaller bites as forcefully as possible - training of conscious swallowing
Aids/adjustments	- food adjustments - chair adjustment (sitting with more support) so that maintaining the posture costs as little energy as possible and the amassed energy can be dedicated to eating - arm support (together with the occupational therapist) - cup stand/lengthened straw and/or plate raiser - modified utensils (e.g. small or light utensils)[38]	- food adjustments
Referral	- dietician: consultation about food intake, overweight and underweight, food schedule and advice[34, 36, 44] - feedback to rehabilitation physician about the situation (if the BMI decreases and eating remains difficult, despite the adjustments)	- primarily referral if the problems remain despite the given advice - dietician: evaluate the nutritional status[34, 36, 44] - examine whether the cough force can be stimulated (together with the paediatric physical therapist), especially train the patient's environment on how they can stimulate this - videofluoroscopic swallow study (when unsafe swallowing is suspected) - NMD team university hospital - gastroenterologist (reflux medication)
Monitoring	- eating observation after one month in order to see whether the advice is helpful - evaluate advice and monitor problems - after 3 months	- eating observation after one month in order to see whether the advice is helpful - evaluate advice and monitor problems - after 1 month

5.4 Dysarthria: evidence and considerations

Exercises for the muscles that are involved in speaking are required for both young boys with DMD who have trouble in this area as well as for older boys whose oral muscle strength is deteriorating and/or intelligibility is decreasing. For older boys, compensatory strategies, speech exercises and sound amplification are needed if it becomes more difficult to understand them.

The consensus study showed that there is especially agreement about the use of SLT for assessing whether (short-term) improvement of intelligibility is possible. Boys can learn to employ this during moments of the day that intelligibility is important. If this is not possible, non-verbal communication, compensatory techniques and possibly AAC aids are indicated.

5.5 Dysarthria: recommendations

Table 7. Overview of possible SLT interventions aimed at dysarthria due to Duchenne muscular dystrophy

DUCHENNE MUSCULAR DYSTROPHY - DYSARTHRIA	
Anatomic characteristics, functions, activities and participation: possible problems	Lip closure problems, mouth breathing, macroglossia[35, 45] Soft voice (low volume), nasal voice[43]
Indications for intervention	- limitations in intelligibility
Explanation/advice	Provide information to the patient and his parents and give advice about: - the cause, type and severity of intelligibility problems. - distributing energy throughout the day as best possible, resting if the patient is fatigued - instructing the patient's environment to place less pressure on communication when he is tired - accepting non-verbal communication - encouraging use of AAC aids
Training/treatment	- dysarthria treatment (voice and articulation) and learn to apply this at specific moments - training of efficient communication (short sentences, including timely breathing breaks, pronouncing all sounds, speaking slowly -not focusing on strength or volume-)
Aids/adjustments	- AAC
Use of other disciplines	
Referral	
Monitoring	- evaluate advice and monitor problems - after 3 months

5.6 Problems with oral hygiene: evidence and considerations

Some children with DMD have a limited mouth opening, making eating, oral hygiene and dental care difficult. In addition to monitoring and providing explanation about this to parents/caregivers, it is also important to take measures to counteract this development. To date, there is no training programme.

In the literature, a correlation is described between bad oral hygiene, nutritional condition and aspiration pneumonia in older patients.[46] Nothing is written about this for DMD, but the relationship is conceivable.

During the consensus meeting, primarily compensatory interventions were considered usable

5.7 Problems with oral hygiene: recommendations

Table 8. Overview of possible SLT interventions aimed at problems with oral hygiene due to Duchenne muscular dystrophy

DUCHENNE MUSCULAR DYSTROPHY – PROBLEMS WITH ORAL HYGIENE	
Anatomic characteristics, functions, activities and participation: possible problems	Limited mouth opening, difficulty inserting a toothbrush into the mouth, thereby not permitting proper brushing of the teeth. The large tongue makes brushing (especially lingual and sometimes occlusal) difficult. Natural cleansing by the tongue, cheeks and chewing is also limited. The molars of the mandible are often characterised by buccal tipping, which creates extra buccal plaque retention.
Indications for intervention	- problems with oral hygiene
Explanation/advice	Provide information to the patient and his parents and give advice about: - the importance of good dental care - the relationship between the jaw opening, oral hygiene and the disease - keeping the teeth clean - using a small (toddler's) or electric toothbrush - drinking after the meal to rinse the mouth - order of teeth cleaning (such as: difficult areas or in the back of the mouth first) - advice on jaw stretching
Training/treatment	- practicing teeth cleaning together with the occupational therapist
Aids/adjustments	- arm support
Use of other disciplines	
Referral	
Monitoring	- evaluate advice and monitor problems - after 3 months

6. Myotonic dystrophy type 1

6.1 Background information

Myotonic dystrophy type 1 (MD1) is a dominant hereditary muscle disease. There are four forms of this disease, which are classified based on the onset of symptoms (congenital, juvenile, classic and late onset adult form). In addition to muscle weakness, there may also be other characteristics in adults, such as cataracts at a young age, frontal balding and/or hormonal problems, arrhythmias or hypoventilation problems. The disease progression is usually slow and is characterised by increasing disease symptoms (anticipation) per younger generation in a family. Cognitive impairments may occur in both children and adults. Because of this there may be limited disease insight, causing other disease characteristics not to be recognised and leading to limited therapy compliance, among other things.[47] With the congenital and juvenile forms, the developmental problems are centre stage. The disease is characterised by the onset of myotonia starting in puberty and by weakness in the distal muscles of the arms and legs, the neck, tongue and face.[48] The prevalence of dysphagia and dysarthria is 64% and 64%, respectively in the juvenile form. Children with the congenital form are very hypotonic, and dysphagia and dysarthria occur in all cases.[1]

6.2 Dysphagia: evidence and considerations

The symptoms can be very diverse with MD1. Clinical reasoning should be employed to determine a course of action; there are many possible interventions about which there is consensus.

Due to limited disease insight, advice for self-management of chewing and swallowing may not be as effective as expected. Umemoto et al. (2012) state that it is important to provide very clear nutritional advice and posture advice for preventing aspiration and choking.[49, 50]

Practicing lip strength with an oral screen (16 minutes, 5 days per week) improved lip strength and endurance in children with MD1 in a study by Sjogreen et al. (2010). Though it cannot be expected that the improvement of lip strength alone has an positive effect on eating and drinking in DM1 population.[51] The panel of experts was in agreement on this. There are no targeted exercises or training that can be recommended. It is recommended that the patient not do any isolated oral-motor exercises but rather train as functionally as possible.

6.3 Dysphagia: recommendations

Table 9. Overview of possible SLT interventions aimed at dysphagia due to myotonic dystrophy type 1

MYOTONIC DYSTROPHY TYPE 1 - DYSPHAGIA	
Anatomic characteristics, functions, activities and participation: possible problems	The patient may have a tented upper lip due to facial weakness[52] and weak oral muscles[53]. Because of this, liquid and solid food may spill out of the mouth[52], pocketing may occur and the patient may hold food and drink in the mouth for a long time. There may also be weak chewing[53], difficulty with oral transport of thin liquid and solid food[53], long duration of swallowing[53], difficulty swallowing solid food, residue, a feeling that food remains stuck in the throat[53], coughing and choking when drinking thin liquid food[53] and wet vocal quality.
Indications for intervention	- wet vocal quality (as a sign of choking risk) - increased risk of choking (residue and coughing/choking) - aspiration pneumonia

MYOTONIC DYSTROPHY TYPE 1 - DYSPHAGIA		
	Oral (digesting and chewing)	**Pharyngeal** (swallowing)
Explanation/advice	EXPLANATION Provide information to the patient and his parents and give advice about: - normal eating and drinking processes - possible eating and drinking problems - the impact of posture - the impact of the tongue strength in relation to the difficulty digesting food and the duration (explanation can promote awareness and help the child understand)[49] - possible interventions POSTURE ADVICE - posture advice: stable, properly supported and upright ADJUSTMENTS (for chewing more easily and transport) - adjusting consistency: softer, smoother, creamier - offering food in small pieces; have the patient take small bites - giving advice for decreasing pocketing: first swallow a bite before taking the next bite/offer food in small bites/plate with separate compartments - placing pieces of solid food between the molars - swallowing more consciously and powerfully - chewing on both the left and right sides - drinking water during and after eating - carefully monitoring weight in order to respond promptly if the nutritional condition worsens - taking into account fatigue, decreased capacity	EXPLANATION Provide information to the patient and his parents and give advice about: - posture, swallowing, eating and drinking - the impact of the tongue strength in relation to the difficulty digesting food and the duration (explanation can promote awareness and help the child understand)[49] - possible interventions POSTURE ADVICE - posture advice: stable, properly supported and upright to assist with best possible swallowing of food, prevent flexion of the head (less chance of choking and less drooling) ADJUSTMENTS (easier swallowing) - adjusting consistency: softer, smoother, creamier - thickening of thin liquids to get more control of the sip - always taking time for eating and drinking - dry swallowing - drinking water during and after eating - swallowing with the chin on the chest to increase strength - taking smaller bites of solid food - taking into account fatigue, decreased capacity - being alert to lower respiratory infections - discussing tube feeding as an option

MYOTONIC DYSTROPHY TYPE 1 - DYSPHAGIA		
Training/treatment	- integrated assessment and treatment: implement and evaluate adjustments	- integrated assessment and treatment: implement and evaluate adjustments - training to become aware of swallowing - checking to see if the temperature of the food has an effect on swallowing
Aids/adjustments	- food adjustments - adjustments to the chair and table to improve the posture at the table - a different cup or a straw in order to better dose thin liquids	- food adjustments
Referral	- dietician - saliva control team: can the drooling be reduced?	- saliva control team: can the drooling be improved? - videofluoroscopic swallow study (when unsafe swallowing is suspected) - NMD team of a university hospital
Monitoring	- evaluate advice and monitor problems - after 3 months	- evaluate advice and monitor problems - after 1 month

6.4 Dysarthria: evidence and considerations

Oral muscle weakness is the main cause of dysarthria in children with MD1.[54] There are no known intervention studies on the effect of SLT.[55] In some children with unintelligible speech, supported communication therapy can be useful.

Improved lip strength (by practicing lip strength) cannot be expected to have an effect on articulation in this target group (see also section 6.2). According to Sjogreen, training with an oral screen can be used as a supplement to SLT[51]; no consensus was reached about this by the expert panel.

6.5 Dysarthria: recommendations

Table 10. Overview of possible SLT interventions aimed at dysarthria due to myotonic dystrophy type 1

MYOTONIC DYSTROPHY TYPE 1 - DYSARTHRIA	
Anatomic characteristics, functions, activities and participation: possible problems	Tented upper lip[52, 56], difficulty lifting the tongue, dysarthria (difficulty with bilabial sounds (no or transient lip closure), vowel distortion, compensatory movements of the tongue, monotonous speech, hypernasality, hoarse voice, difficulty moderating volume, decreased intelligibility), worsening of symptoms when tired.
Indications for intervention	- hindrance of the intelligibility problems

MYOTONIC DYSTROPHY TYPE 1 - DYSARTHRIA	
Explanation/advice	Provide information to the patient and his parents and give advice about: - the relationship between muscle weakness and intelligibility - prognosis - how best the child can speak in situations where he/she is more tired - place in classroom - teaching the child's environment to take decreased intelligibility into account - allocating more time in the interaction - good initial posture (properly supported) - keeping a communication notebook about activities the child undertakes, so that the context is known for the school and at home in order to facilitate faster understanding of the child - examining the possibilities of using a AAC device
Training/treatment	- integrated assessment and treatment (influenceability of articulation, intelligibility and/or adjustment of speaking speed) - practicing talking in single words, short sentences, with (breathing) breaks (aimed at speaking more clearly in the short term) - learning in which situations speaking clearly is/is not important - initiation of AAC if the child is almost unintelligible - if opting for practicing: only functional - indirectly by instructing the child's environment
Aids/adjustments	- AAC for severe intelligibility problems
Use of other disciplines	Depending on the setting: - AAC team and/or - occupational therapist and/or - remedial education specialist/psychologist
Referral	
Monitoring	- evaluate advice and monitor problems - after 3 months

6.6 Drooling: evidence and considerations

Drooling occurs regularly in children with myotonic dystrophy type 1 (MD1).[53] Children with MD1 often have difficulty following instructions and some children don't respond at all to instructions, making it difficult to implement advice.

Lip strength improvement does not help reduce drooling (see also section 6.2).[51] This corresponds to the experiences of the panel of experts. Explanation and advice with respect to posture and awareness in particular are considered to be helpful. A saliva control team may be able to recommend treatment or give targeted advice for improving the swallowing of saliva.

6.7 Drooling: recommendations

Table 11. Overview of possible SLT interventions aimed at drooling due to myotonic dystrophy type 1

MYOTONIC DYSTROPHY TYPE 1 - DROOLING	
Anatomic characteristics, functions, activities and participation: possible problems	Drooling, wet vocal quality
Indications for intervention	- bothersome drooling after age 4[12]
Explanation/advice	Provide information to the patient and his parents and give advice about: - the relationship between posture, swallowing and saliva control - posture advice: (flexion of the head worsens drooling), stable, properly supported and upright
Training/treatment	- implementing advice given to the patient during treatment in order to monitor its efficacy - training to become aware of swallowing - effect of dry swallowing
Aids/adjustments	
Use of other disciplines	
Referral	
Monitoring	- evaluate advice and monitor problems - after 3 months

6.8 Problems with oral hygiene: evidence and considerations

Engvall et al. (2009) indicate that the oral health of many children with myotonic dystrophy type 1 (MD1) is worse than that of healthy subjects. This worsens over the course of time. Keeping the teeth clean is especially difficult in the posterior part of the mouth, because children with MD1 have diminished hand strength. This decreases the agility when cleaning the teeth. Additionally, children and adolescents with MD1 have problems cooperating during dental treatments (due to fear and tiredness, among other things), and this does not appear to get better with age. It is important for prophylactic care (tooth plaque removal and fluoride treatment) to be offered from a young age[57].

A peripheral dental practice for prophylactic care in short intervals in cooperation with a specialised dentist for specialised care should be recommended according to Engvall et al.[57]

The panel of experts reached agreement about the importance of explanation and advice in particular. Direct therapy may be indicated for carrying out a multidisciplinary task analysis (what goes well under which circumstances) and for the joint giving of instructions.

6.9 Problems with oral hygiene: recommendations

Table 12. Overview of possible SLT interventions aimed at problems with oral hygiene due to myotonic dystrophy type 1

MYOTONIC DYSTROPHY TYPE I – PROBLEMS WITH ORAL HYGIENE	
Anatomic characteristics, functions, activities and participation: possible problems	Difficulty brushing teeth due to limited hand strength and agility during brushing, tooth plaque, cavities
Indications for intervention	
Explanation/advice	Provide information to the patient and his parents and give advice about: - why brushing teeth is difficult - explanation of oral hygiene, the importance of brushing properly in relation to general health - parental assistance with teeth brushing - trying out whether using an electric toothbrush works better - parents rebrushing the teeth after the child has brushed - visualising the steps of teeth brushing
Training/treatment	- together with the occupational therapist, observing the teeth brushing and jointly giving advice, if necessary
Aids/adjustments	- electric toothbrush or adapted toothbrush
Use of other disciplines	
Referral	- special dentistry (advice and treatment in connection with dental care and cavities) - oral hygienist
Monitoring	- evaluate advice and monitor problems - after 6 months

7 Spinal muscular atrophy type 2

7.1 Background

Spinal muscular atrophy (SMA) is a group of autosomal recessive muscle diseases caused by degeneration of the anterior horn cells in the spinal cord. The illness primarily manifests itself through axial and symmetric muscle weakness in the upper arms and thighs, with the legs often being more affected than the arms. The initial symptoms of SMA type 2 appear between six and 18 months of age. Children achieve the milestone independent sitting, but they cannot achieve independent standing or walking.[58]

The prevalence of dysphagia and dysarthria is 47% and 11%, respectively.1 These children generally have weak muscles and decreased efficiency of tongue movements and often problems with head posture.[58] Drooling rarely occurs in children with SMA type 2. There is no literature about the prevalence and treatment of drooling in SMA type 2. It is conceivable for drooling to occur in the presence of co-morbidity, such as defects of the ear, nose and throat or severe mental disabilities.

7.2 Dysphagia: evidence and considerations

Problems with limited jaw opening occur frequently, regardless of the severity and type of SMA.[59] A study by Van Bruggen (2011) showed that parents and children are often unaware of the limited jaw opening and the possible consequences thereof. Little scientific research has been done on the treatment of limited jaw opening. Only one study is known with three SMA patients. Stretching exercises aim to maintain maximum jaw opening and prevent further exacerbation of the condition. It is important to start treatment early. Limited jaw opening occurs at an early age (7 years) in some children. Early detection and starting training when the opening is still adequate could have more effect than late intervention.[60]

Jaw, chewing and swallowing problems often occur in combination with each other and are described as bulbar problems. Fatique during chewing is also frequently observed, which may result in limited oral intake or aspiration.[59,60] Softer or semi-solid food can help with swallowing problems, preventing choking and decreasing meal duration.[61, 62] Sitting adjustments and eating aids (such as the Neaty Eater, arm support, adapted straws) may contribute to independent eating. This may also promote the efficiency and safety of the swallowing.[63] There is no supporting evidence that oral motor treatment affects the safety or efficiency of oral feeding.[61] A more forward position of the head can facilitate a wider range of forward movement of the hyoid, thereby making swallowing easier.[64]
Children with SMA type 2 often need several swallows in order to process one bite. There may be residue in the valleculae and above the upper oesophageal sphincter (visualised by a videofluoroscopic swallow study).[22, 64] A study with videofluoroscopic swallow studies described that the problems eating solid food are greater than with liquids. Large quantities of residue increase the risk of aspiration.[9] Van den Engel-Hoek et al (2009) suggest "an integrated treatment with an adapted postured during meals, adjusting meals (avoiding or reducing solid foods) and the advice to drink water after meals, claring the oropharyngeal area to prevent aspiration pneumonia".[64] A videofluoroscopic swallow study is recommended in the presence of coughing/choking or other signs of pharyngeal dysphagia.

The presence of pneumonia and a weak cough may indicate aspiration, which are all serious (possibly life-threatening) problems.[34] Reflux can also occur. Fatty food delays gastric emptying and increases the risk of reflux.[61] Another consensus study did not recommended a standard videofluoroscopic swallow study, but did recommended referring the patient for a videofluoroscopic swallow study in the event of concerns about swallow safety.[61] Coughing support (cough assist machine, assistance from caregivers with coughing and sputum mobilisation) is important for these patients.[61]
Eating problems may affect the food intake and general nutritional condition. It is important for advice to be given regarding dietary adjustments. A dietician must be involved to determine the caloric intake, fluid intake and food composition and to perform regular check-ups.[62] If inadequate oral intake is determined, then proactive offering of additional food is indicated. If swallowing (after intervention) is not safe or in the presence of malnutrition, tube feeding is strongly recommended.[62] The aim is to keep each child on his own growth curve.

7.3 Dysphagia: recommendations

Table 13. Overview of possible SLT interventions aimed at dysphagia due to spinal muscular atrophy type 2

SMA 2 - DYSPHAGIA	
Anatomic characteristics, functions, activities and participation: possible problems	There may be difficulties bringing the food to the mouth[58], especially in the presence of fatigue, limited jaw opening[59, 60, 65,66], malocclusion[66], head retraction[60], limited bite force[66], difficulty biting off large pieces[60], difficulty eating hard foods[60, 66], problems with oral transport and chewing solid food[59, 65], chewing takes a long time and energy and is tiring[58, 59, 65, 66], swallowing problems[65], multiple swallows needed for solid food (due to reduced movements and weakness of the tongue)[64], the feeling that food remains stuck, residue in the valleculae and above the upper oesophageal sphincter (not with thin liquids)64, choking[58, 59], weak coughing[62], regular spitting, reflux[62, 65], pain when yawning and brushing teeth[66]. Due to the above-mentioned problems, the mealtime duration can be long[58, 65, 66], there is an increased risk of penetration and aspiration[62, 64, 65] and pneumonia[58, 62, 65] (even without signs of choking) and inadequate intake[59, 62] and underweight[58, 65] may occur.
Indications for intervention	- chewing problems - fatigue when chewing and swallowing - symptoms may increase without intervention[60] - (slight) limited jaw opening[60] - (risk of) inadequate intake[62] - risk of choking[62, 64] - decreased appetite - problems with independent eating and drinking - decreased cough force - risk of pneumonia[62] - possible reflux[62]

SMA 2 - DYSPHAGIA		
	Oral (digesting and chewing)	**Pharyngeal** (swallowing)
Explanation/advice	EXPLANATION Provide information to the patient and his parents and give advice about: - jaw opening: deterioration if nothing is done (use it or lose it), maintenance of the current situation (this advice also applies to a slightly limited jaw opening without symptoms)[60] - teeth brushing and oral hygiene - posture in relation to the swallowing process and food - the possible interventions POSTURE ADVICE - general posture advice[64] ADJUSTMENTS (for chewing more easily and transport) - adjustment of consistency[62, 64, 65] (softer food, thick liquids, creamier) for easier chewing and easier oral transport[61] - avoiding hard foods[64] - enriched liquid nutrition for (partial) meal replacement[62] - discussion of how complete and sufficient intake can be effectuated[62, 65] - adjusting the bite size[65] - continuing to encourage chewing - drinking water during and after the meal[64] - practicing jaw opening for 10 minutes 3x per day (e.g. before or after brushing teeth) to maintain jaw opening / - opening the mouth as wide as possible several times per day (if needed, first practice this during therapy and then switch to doing it at home)[60] - advice about arm support and adjusted eating and drinking utensils - if the above things are already being done but with inadequate effect: discuss tube feeding	EXPLANATION Provide information to the patient and his parents and give advice about: - why food gets stuck - what happens during swallowing - possible consequences and risks - the possible interventions POSTURE ADVICE - advice on optimal initial posture[64]: stable, properly supported, upright, proper head posture (as much as possible in the middle position, with no pulling in or jutting out of the chin) ADJUSTMENTS (easier swallowing) - adjusting the consistency[62, 64, 65] to softer and smoother food, thick and thin fluids - smaller bites[65] - drinking water during and after the meal[64] (to decrease the risk of penetration and aspiration and make swallowing easier) - raising the possibility of enriched liquid nutrition or tube feeding, stressing the importance of a good nutritional condition and safe swallowing[62, 65] - advice on coughing up, together with the paediatric physical therapist

SMA 2 - DYSPHAGIA		
Training/treatment	- integrated assessment and treatment: implement and evaluate adjustments - posture optimisation (in combination with other therapists, if needed) - trying out which aid works best - parents should be present - training for maintenance of jaw opening (e.g. with Therabite or cork)[60]	- integrated assessment and treatment: implement and evaluate adjustments - consciously learning how to swallow, as forcefully as possible - training swallowing with the correct tongue position, during eating and drinking (functional training), to facilitate transport
Aids/adjustments	- food adjustments - reviewing seating facility together with the occupational therapist - arm support[63] - adjusted eating and drinking utensils (Neaty Eater, adjusted cup, plate, cutlery, straw)[63]	- food adjustments
Referral	- dietician for nutritional advice[62, 65] - occupational therapist (arm and hand function) - physician for advice or starting tube feeding	- dietician for nutritional advice[62, 65] - videofluoroscopic swallow study (when unsafe swallowing is suspected)[62, 65] - physician for advice for possible reflux[65]
Monitoring	- evaluate advice and monitor problems - after 3 months (in the presence of risk of malnutrition and/or choking) - after 1 month (longer meal duration, underweight)	- evaluate advice and monitor problems - after 3 months

7.4 Dysarthria: evidence and considerations

Limited intelligibility affecting daily communication is observed in these children, but no literature on intervention is available. The recommendations regarding speech are entirely based on the experiences of the panel of experts.

7.5 Dysarthria: recommendations

Table 14. Overview of possible SLT interventions aimed at dysarthria due to spinal muscular atrophy type 2

SMA 2 - DYSARTHRIA	
Anatomic characteristics, functions, activities and participation: possible problems	Dysarthria (reduced speaking volume, decreased intelligibility)
Indications for intervention	- decreased intelligibility
Explanation/advice	Provide information to the patient and his parents and give advice about: - the relationship between the muscle disease and speech - advice about the location in the classroom where the patient should sit, tranquil environment - instructing the child's environment about communication strategies - use of AAC, such as gestures, agreeing on signals for busy surroundings, teacher approaching the child more often to ask if he wants to say something - use of AAC devices: such as a sound amplification or speech computer - posture advice for better, most optimal breathing support possible
Training/treatment	- integrated assessment and treatment: Is it possible to improve the voice volume? Is it possible to improve diaphragmatic breathing and the coordination between breathing and voice for a short time? - practicing timely breathing breaks during speaking or decreasing speaking speed - practicing with an AAC device (amplification or speech computer/app) - lung function training, e.g. with the help of the magic flute or electric saxophone or GroovTube (in collaboration with other disciplines, e.g. physical, music or occupational therapist) (https://mybreathmymusic.com/magic-flute# https://www.groovtube.nl/)
Aids/adjustments	- AAC device (such as a speech computer or voice amplification)
Use of other disciplines	
Referral	
Monitoring	- evaluate advice and monitor problems - after 3 months

7.6 Problems with oral hygiene: evidence and considerations

Problems with the jaw occur frequently, regardless of the severity and type of SMA.[59] Limited jaw opening occurs at an early age (7 years) in some children, which can cause problems with oral hygiene. The above-cited study by Van Bruggen (2011) shows that there is little awareness among parents and children about the limited mouth opening.[60] It is important to start treatment early. Early detection and starting training when the jaw opening is still adequate could have more effect than late intervention

7.7 Problems with oral hygiene: recommendations

Table 15. Overview of possible SLT interventions aimed at problems with oral hygiene due to spinal muscular atrophy type 2

SMA-2 – PROBLEMS WITH ORAL HYGIENE	
Anatomic characteristics, functions, activities and participation: possible problems	Limitation in the jaw opening (mild to severe), which may result in oral care difficulty.
Indications for intervention	- symptoms will increase without intervention - do not wait even if the limitation is mild
Explanation/advice	Provide information to the patient and his parents and give advice about: - problems with tooth brushing and oral hygiene due to a small jaw opening - small jaw opening: deterioration if nothing is done (use it or lose it) - maintaining what is still possible (this advice also applies to a slightly limited jaw opening without symptoms) - practicing jaw opening for 10 minutes 3x per day (e.g. before/after brushing teeth) to maintain jaw opening/opening the mouth as wide as possible several times per day (if needed, first practice this during therapy and then switch to doing it at home) - brushing teeth with a toothbrush with a small head - small electric toothbrush
Training/treatment	- integrated assessment and treatment: which head posture and which aid may improve brushing
Aids/adjustments	- a toothbrush with a small head
Use of other disciplines	
Referral	
Monitoring	- evaluate advice and monitor problems - after 6 months

Literature

1. Kooi-van Es, M., et al., *Dysphagia and Dysarthria in Children with Neuromuscular Diseases, a Prevalence Study.* Journal of Neuromuscular Diseases, 2020. **7**(3): p. 287-295.

2. Hill, M., T. Hughes, and C. Milford, *Treatment for swallowing difficulties (dysphagia) in chronic muscle disease.* Cochrane Database of Systematic Reviews, 2014(8): p. 4.

3. American Speech-Language-Hearing Association. *Guidelines for Speech-Language Pathologists Providing Swallowing and Feeding Services in Schools [Guidelines].* 2007; Available from: www.asha.org/policy.

4. European society for swallowing disorders, *3rd congress. Paediatric dysphagia—Position statements [Position Statement].* 2013.

5. Prasse, J.E. and G.E. Kikano, *An overview of pediatric dysphagia.* Clinical Pediatrics, 2009. **48**(3): p. 247-251.

6. Van den Engel-Hoek, L., et al., *Eet- en drinkproblemen bij jonge kinderen. Een leidraad voor logopedisten en andere hulpverleners in de gezondheidszorg.* 5 ed. 2018, Assen: Uitgeverij Koninklijke Van Gorcum.

7. Jansonius-Schultheiss, K., L. van Coppenolle, and E. Beyaert, *Afwijkende mondgewoonten. Inleiding, onderzoek en behandeling*. 1997, Leuven: Uitgeverij Acco.

8. Logeman, J.A., *Evaluation and treatment of swallowing disorders*. 1998, Austin: TX: Pro-Ed.

9. Weir, K., et al., *Clinical signs and symptoms of oropharyngeal aspiration and dysphagia in children.* European Respiratory Journal, 2009. **33**(3): p. 604-611.

10. Pennington, L., N. Miller, and S. Robson, *SLT for children with dysarthria acquired before three years of age.* Cochrane Database of Systematic Reviews, 2009(4): p. 21.

11. Pennington, L., et al., *Intensive speech and language therapy for older children with cerebral palsy: a systems approach.* Developmental Medicine and Child Neurology, 2010. **52**(4): p. 337-344.

12. van Hulst, K., et al., *Development of the Drooling Infants and Preschoolers Scale (DRIPS) and reference charts for monitoring saliva control in children aged 0-4 years.* Infant Behavior & Development, 2018. **50**: p. 247-256.

13. Guyatt, G.H., et al., *GRADE: an emerging consensus on rating quality of evidence and strength of recommendations.* British Medical Journal, 2008. **336**(7650): p. 924-926.

14. Okoli, C. and S.D. Pawlowski, *The Delphi method as a research tool: an example, design considerations and applications.* Information & Management, 2004. **42**: p. 15-29.

15. van der Lee, L., A.M. Hill, and S. Patman, *Expert consensus for respiratory physiotherapy management of mechanically ventilated adults with community-acquired pneumonia: A Delphi study.* Journal of Evaluation in Clinical Practice, 2019. **25**(2): p. 230-243.

16. Junger, S., et al., *Guidance on Conducting and REporting DElphi Studies (CREDES) in palliative care: Recommendations based on a methodological systematic review.* Palliative Medicine, 2017. **31**(8): p. 684-706.

17. Fitch, K., et al., *The RAND/UCLA Appropriateness Method User's Manual.* 2001: RAND.

18. Bariani, R.C.B., et al., *The impact of positive airway pressure on midface growth: a literature review.* Brazilian Journal of Otorhinolaryngology, 2020. **86**(5): p. 647-653.

19. Simonds, A.K., et al., *Outcome of paediatric domiciliary mask ventilation in neuromuscular and skeletal disease.* European Respiratory Journal, 2000. **16**(3): p. 476-481.

20. Dubowitz, V., *Muscle Disorders in childhood.* 2 ed. 2000, London: W.B. Saunders Company Ltd.

21. Wang, C.H., et al., *Consensus Statement on Standard of Care for Congenital Myopathies.* Journal of Child Neurology, 2012. **27**(3): p. 363-382.

22. van den Engel-Hoek, L., et al., *Children with central and peripheral neurologic disorders have distinguishable patterns of dysphagia on videofluoroscopic swallow study.* Journal of Child Neurology, 2014. **29**(5): p. 646-653.

23. Williams, R.B., et al., *Biomechanics, diagnosis, and treatment outcome in inflammatory myopathy presenting as oropharyngeal dysphagia.* Gut, 2003. **52**(4): p. 471-478.

24. van den Engel-Hoek, L., et al., *Neonatal Swallowing Assessment and Practical Recommendations for Oral Feeding in a Girl With a Severe Congenital Myopathy.* Journal of Child Neurology, 2011. **26**(8): p. 1041-1044.

25. Ryan, M.M., et al., *Nemaline myopathy: A clinical study of 143 cases.* Annals of Neurology, 2001. **50**(3): p. 312-320.

26. Bagnall, A.K., M.A. Al-Muhaizea, and A.Y. Manzur, *Feeding and speech difficulties in typical congenital Nemaline Myopathy.* Int J Speech Lang Pathol, 2006. **8**(1): p. 7-16.

27. Maggi, L., et al., *Congenital myopathies - Clinical features and frequency of individual subtypes diagnosed over a 5-year period in the United Kingdom.* Neuromuscular Disorders, 2013. **23**(3): p. 195-205.

28. van den Engel-Hoek, L., et al., *Feeding and swallowing disorders in pediatric neuromuscular diseases: an overview.* Journal of Neuromuscular Diseases, 2015. **2**(4): p. 357–369.

29. Tsabari, R., et al., *Congenital myopathy due to myosin heavy chain 2 mutation presenting as chronic aspiration pneumonia in infancy.* Neuromuscular Disorders, 2017. **27**(10): p. 947-950.

30. Bagnall, A., M.A. Al-Muhaizea, and A.Y. Manzur, *Feeding and speech difficulties in typical congenital nemaline myopathy.* Developmental Medicine and Child Neurology, 2006. **48**: p. 29-29.

31. Hendriksen, J.G.M., et al., *249th ENMC workshop: The role of brain dystrophin in muscular dystrophy: Implications for clinical care and translational research.* Neuromuscular Disorders, 2020.

32. Bushby, K., et al., *Diagnosis and management of Duchenne muscular dystrophy, part 1: diagnosis, and pharmacological and psychosocial management.* Lancet Neurology, 2010. **9**(1): p. 77-93.

33. Bushby, K., et al., *The diagnosis and management of Duchenne muscular dystrophy: Internationally generated care recommendations.* Neuromuscular Disorders, 2009. **19**(8-9): p. 640-641.

34. Birnkrant, D.J., K. Bushby, and C.M. Bann, *Diagnosis and management of Duchenne muscular dystrophy, part 1: diagnosis, and neuromuscular, rehabilitation, endocrine, and gastrointestinal and nutritional management (vol 17, pg 251, 2018).* Lancet Neurology, 2018. **17**(6): p. 495-495.

35. Botteron, S., et al., *Orofacial dysfunction in Duchenne muscular dystrophy.* Archives of Oral Biology, 2009. **54**(1): p. 26-31.

36. van den Engel-Hoek, L., et al., *Dystrophic changes in masticatory muscles related chewing problems and malocclusions in Duchenne muscular dystrophy.* Neuromuscular Disorders, 2016. **26**(6): p. 354-360.

37. van den Engel-Hoek, L., et al., *Oral muscles are progressively affected in Duchenne muscular dystrophy: implications for dysphagia treatment.* Journal of Neurology, 2013. **260**(5): p. 1295-1303.

38. Aloysius, A., et al., *Swallowing difficulties in Duchenne muscular dystrophy: Indications for feeding assessment and outcome of videofluroscopic swallow studies.* european Journal of Paediatric Neurology, 2008. **12**(3): p. 239-245.

39. Archer, S.K., et al., *Dysphagia in Duchenne muscular dystrophy assessed by validated questionnaire.* International journal of language & communication disorders/Royal College of Speech & Language Therapists, 2013. **48**(2): p. 240.

40. van Bruggen, H.W., et al., *Fighting Against Disuse of the Masticatory System in Duchenne Muscular Dystrophy: A Pilot Study Using Chewing Gum.* Journal of Child Neurology, 2015. **30**(12): p. 1625-1632.

41. Toussaint, M., et al., *Dysphagia in Duchenne muscular dystrophy: practical recommendations to guide management.* Disability and Rehabilitation, 2016. **38**(20-21): p. 2052-2062.

42. van der Bilt, A., *Assessment of mastication with implications for oral rehabilitation: a review.* Journal of Oral Rehabilitation, 2011. **38**(10): p. 754-780.

43. Jaffe, K.M., et al., *Symptoms of upper gastrointestinal dysfunction in Duchenne Muscular-Dystrophy - Case-controle study.* Archives of Physical Medicine and Rehabilitation, 1990. **71**(10): p. 742-744.

44. Brumbaugh, D., et al., *Nutritional and Gastrointestinal Management of the Patient With Duchenne Muscular Dystrophy.* Pediatrics, 2018. **142**: p. S53-S61.

45. Symons, A., G. Townsend, and T. Hughes, *Dental characteristics of patients with Duchenne muscular dystrophy.* Journal of Dentistry for Children, 2002. **69**(3): p. 277-283.

46. van der Maarel-Wierink, C.D., et al., *Oral health care and aspiration pneumonia in frail older people: a systematic literature review.* Gerodontology, 2013. **30**(1): p. 3-9.

47. Baldanzi, S., et al., *Disease awareness in myotonic dystrophy type 1: an observational cross-sectional study.* Orphanet Journal of Rare Diseases, 2016. **11**: p. 11.

48. Kiliaridis, S. and C. Katsaros, *The effects of myotonic dystrophy and Duchenne muscular dystrophy on the orofacial muscles and dentofacial morphology.* Acta Odontologica Scandinavica, 1998. **56**(6): p. 369-374.

49. Ercolin, B., et al., *Oral Motor Movements and Swallowing in Patients with Myotonic Dystrophy Type 1.* Dysphagia, 2013. **28**(3): p. 446-454.

50. Umemoto, G., et al., *Dysphagia in Duchenne Muscular Dystrophy versus Myotonic Dystrophy Type I.* Muscle & Nerve, 2012. **46**(4): p. 490-495.

51. Sjogreen, L., et al., *The effect of lip strengthening exercises in children and adolescents with myotonic dystrophy type 1.* International Journal of Pediatric Otorhinolaryngology, 2010. **74**(10): p. 1126-1134.

52. Echenne, B., et al., *Myotonic dystrophy type I in childhood Long-term evolution in patients surviving the neonatal period.* European Journal of Paediatric Neurology, 2008. **12**(3): p. 210-223.

53. Sjogreen, L., et al., *Orofacial dysfunction in children and adolescents with myotonic dystrophy.* Developmental Medicine and Child Neurology, 2007. **49**(1): p. 18-22.

54. Sjogreen, L., A. Martensson, and A.B. Ekstrom, *Speech characteristics in the congenital and childhood-onset forms of myotonic dystrophy type 1.* International Journal of Language & Communication Disorders, 2018. **53**(3): p. 576-583.

55. Sjogreen, L., A. Martensson, and A.B. Ekstrom, *Speech characteristics in the congenital and childhood-onset forms of myotonic dystrophy type 1.* International journal of language & communication disorders/Royal College of Speech & Language Therapists, 2018. **00**(0): p. 1-8.

56. Hageman, A.T.M., et al., *Congenital myotonic dystrophy; a report on thirteen cases and a review of the literature.* Journal of the Neurological Sciences, 1993. **115**: p. 95-101.

57. Engvall, M., et al., *Oral health status in a group of children and adolescents with myotonic dystrophy type 1 over a 4-year period.* International Journal of Paediatric Dentistry, 2009. **19**(6): p. 412-422.

58. Chen, Y.S., et al., *Prevalence and risk factors for feeding and swallowing difficulties in spinal muscular atrophy types II and III.* Journal of pediatrics, 2012. **160**(3): p. 447-451.

59. Van der Heul, A.M.B., et al., *Bulbar problems self-reported by children and adults with spinal muscular atrophy.* Journal of Neuromuscular Diseases, 2019. **6**: p. 361–368.

60. van Bruggen, H.W., et al., *Impaired mandibular function in Spinal Muscular Atrophy Type II: need for early recognition.* Journal of Child Neurology, 2011. **26**(11): p. 1392-1396.

61. Wang, C.H., et al., *Consensus statement for Standard of Care in Spinal Muscular Atrophy.* Journal of Child Neurology, 2007. **22**(8): p. 1027-1049.

62. Mercuri, E., et al., *Diagnosis and management of spinal muscular atrophy: Part 1: Recommendations for diagnosis, rehabilitation, orthopedic and nutritional care.* Neuromuscular Disorders, 2018. **28**(2): p. 103-115.

63. Willig, T.N., et al., *Swallowing problems in neuromuscular disorders.* Archives of Physical Medicine and Rehabilitation, 1994. **75**(11): p. 1175-1181.

64. van den Engel-Hoek, L., et al., *Dysphagia in spinal muscular atrophy type II: More than a bulbar problem?* neurology 2009. **73**(21): p. 1787-1791.

65. Messina, S., et al., *Feeding problems and malnutrition in spinal muscular atrophy type II.* Neuromuscular Disorders, 2008. **18**: p. 389–393.

66. Granger, M.W., et al., *Masticatory muscle function in patients with spinal muscular atrophy.* American Journal of Orthodontics and Dentofacial Orthopedics, 1999. **115**(6): p. 697-702.

Appendix 1. Experts

(SLT's, workplace at the time of the study)

- Ms Suzanne Franssen, Klimmendaal Rehabilitation Centre, Arnhem
- Ms Marcia van den Hove, Klimmendaal Rehabilitation Centre, Arnhem
- Ms Martina Quint, Libra Libra Rehabilitation and Audiology, Eindhoven
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- Ms Wilma Faber, Rehabilitation Friesland, Beetsterzwaag
- Ms Yvonne Stel, Rehabilitation Friesland, Beetsterzwaag
- Ms Fleur Roosenboom, Tolburg Rehabilitation Center, Den Bosch
- Ms Priscilla Bevers, Tolburg Rehabilitation Center, Den Bosch
- Ms Ellen Hoogland, Treant Healthcare Group, Emmen
- Ms Marie Pruyn, Sint Maartenskliniek, Nijmegen
- Ms Els Overdiep, Breda University of Applied Sciences, Breda
- Ms Marije Kraak, Heliomare, Wijk aan zee
- Ms Anke Bulsink, Adelante, Venlo
- Ms Marina Bil, Basalt, Delft
- Ms Richelle te Kempel, Rehabilitation Center Roessingh, Enschede
- Ms Marijke Hijlkema, Centre for Rehabilitation UMCG, Beatrixoord
- Ms A.J.P. van Daal, Reade, Amsterdam
- Ms Annette de Ruijter-Oosting, Vogellanden, Zwolle

Appendix 2. Acronyms

AAC	Augmentative and Alternative Communication
CM	Congenital myopathy
DMD	Duchenne muscular dystrophy
MD	Myotonic dystrophy
NMD	Neuromuscular disease
SLT	Speech Language Therapy/Therapist
SMA	Spinal muscular atrophy