Myotonic

# Care Guidelines for Speech and Language Pathologists Treating Adults and Children with Myotonic Dystrophy

Myotonic

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# Care Guidelines for Speech and Language Pathologists Treating Adults and Children with Myotonic Dystrophy

In order to improve and standardize care, six Speech and Language Pathology clinicians specialized in myotonic dystrophy (DM) joined in a process started in January 2019 and concluded in June 2020 to create the *Care Guidelines for Speech and Language Pathologists Treating Adults and Children with Myotonic Dystrophy*. The project was organized and supported by Myotonic. A complete author list and reference list is available in the appendix.

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## Abbreviations that are commonly used in this guide:

**ASD:** Autism Spectrum Disorders

**ASHA:** American Speech-Language-Hearing-Association

**AAC:** Augmentative and Alternative Communication

**CDM:** Congenital-onset Myotonic Dystrophy

**DM:** Myotonic Dystrophy

**DM1:** Myotonic Dystrophy Type 1

**DM2:** Myotonic Dystrophy Type 2

**ID:** Intellectual Disability

**OT:** Occupational Therapy

**SLP:** Speech-Language Pathologist

**ST:** Speech Therapy

# Speech:
## Assessment of Adults with Myotonic Dystrophy

Speech is, despite the rise of social media, indispensable for daily communication. When a neurological disease like myotonic dystrophy (DM) affects speech, this is called dysarthria. The International Classification of Functioning (ICF) defines speech as a bodily function, which if affected, can lead to problems in communication and conversation (World Health Organization, 2001). Assessment of speech should cover all ICF levels.

### Level of Functioning

Myotonic Dystrophy (DM) is a neuromuscular disorder characterized by weakness and myotonia. In the speech of patients with DM, signs of weakness and myotonia can occur, although weakness seems to be the main characteristic (de Swart, van Engelen, & Maassen, 2007; Sjögreen, Mårtensson, & Ekström, 2018). Assessment of speech includes the five aspects of speech production; respiration, phonation, resonance, articulation and prosody, because weakness can be present in all aspects (Duffy, 2013). Myotonia can occur in the tongue, leading to irregularities in articulation (de Swart et al., 2007).

To assess speech at the level of functioning, some standardized dysarthria assessments have been developed, such as the Frenchay Dysarthria Assessment (FDA) (Enderby & Palmer, 2008) and the Robertson Dysarthria Profile (Robertson, 1982) in the UK, the Swedish 'Dysartritest' (Hartelius, Svensson, & Bubach, 1993), the Dutch Radboud Dysarthria Assessment (Knuijt, Kalf, Van Engelen, De Swart, & Geurts, 2017) and the German 'Bogenhausener Dysarthrieskalen' (Ziegler, Staiger, Scholderle, & Vogel, 2017). These dysarthria assessments include assessment of speech characteristics. The speech tasks used in the dysarthria assessments are different. Spontaneous or conversational speech is the most important task to evaluate speech because it is the most representative task of daily communication. Text reading or picture story can be helpful because there are no interruptions and prosodic features that will become more evident. One can choose to use maximum performance tests of speech production, like maximum repetition rate, maximum phonation time, fundamental frequency range, and maximum phonation volume, to interpret speech characteristics. For example, patients who tend to slow down their speaking rate during spontaneous speech can exhibit some difficulties during maximum repetition rate.

Examples of observation items and characteristic speech deficits in DM are detailed in **Table 1** on the following page.

Dysarthria assessments include oromotor tasks. Oromotor tasks can be used to interpret speech characteristics (e.g. hypotonia) or to elicit myotonia in the tongue or the masseter muscle.

The dysarthria assessments named above include severity classification. Scoring the severity of dysarthria is important for various reasons. First, a severity score is helpful to monitor dysarthria over time, to evaluate progression, recovery and treatment effects. Second, a severity score is important for treatment focus. Generally, in the case of minimal dysarthria, patients can be helped satisfactorily with subtle adjustments of their speech habits. In mild to moderate dysarthria, the therapy usually will be more focused on practicing speaking techniques, whereas in severe dysarthria speech functions are compensated with augmentative and alternative communication strategies.

To conclude, speech assessment at the level of functioning in DM can include a description of the deviant speech characteristics in all aspects of speech production, accompanied by a severity score.

**Table 1: Observation Items Regarding the Five Aspects of Speech Production and Characteristic Speech Deficits in Myotonic Dystrophy.**

| Aspects of Speech Production | Observation Items | Myotonic Dystrophy |
|---|---|---|
| Respiration | Breathing pattern, breath groups, place | Short breath groups, short breath support |
| Phonation | Vocal quality, vocal use, loudness, pitch | Hoarseness, decrease in volume |
| Resonance | Nasality | Hypernasality |
| Articulation | Articulation movements, vowels, consonants, clusters | Imprecise, flaccid consonants, interdental articulation, substitutions*, irregularities due to myotonia |
| Prosody | Melodic accent, dynamic accent, temporal accent | Monopitch, monoloudness |

**Bilabials are compensated for with interdental or labiodentals articulation, dentals are substituted with interdental articulation, and velars with dental or palatal articulation (Sjögreen et al., 2018).*

## The Use of Objective Measurements

Certain measurements may be helpful to distinguish pathological from non-pathological performances and to evaluate progression, recovery, or treatment effects. When distinguishing pathological from non-pathological performances, language specific reference values are indispensable (Icht & Ben-David, 2014). Sample measurements are speaking rate, maximum repetition rate, maximum phonation time, maximum phonation volume and fundamental frequency range. Speaking rate can be easily assessed by counting the number of words or syllables per minute. The maximum phonation time can be analyzed with a stopwatch and maximum phonation volume with a dB meter. Fundamental frequency range and maximum repetition rate are more elaborate to analyze. The software package PRAAT (Boersma & Weening, 1995) can be used for acoustic analysis.

## Level of Activity

From a clinical perspective, assessment at the levels of activity and participation is most important because of the impact of unintelligible speech on daily life. Deviant speech characteristics may negatively influence intelligibility. Standardized dysarthria assessments (discussed in the paragraph 'levels of functioning') include measurement of intelligibility. However, these assessments are restricted most of the time to a limited amount of words or sentences, leading to familiarity by the SLP after a couple of patients. There are more elaborate intelligibility assessments, such as the Sentence Intelligibility Test (K. Yorkston, Beukelman, & Tice, 1996), the Swedish Intelligibility Assessment (Lillvik, Allemark, Karlström, & Hartelius, 1999), the Computerized Assessment of Intelligibility and Dysarthric Speech (K. Yorkston, 1984), the Dutch Speech Intelligibility Assessment at Word (de Bodt, Guns, & van Nuffelen, 2006) and Sentence Levels (Martens, Van Nuffelen, & De Bodt, 2010). Intelligibility can also be scored on a severity scale, like the one used in Sjögreen et al. (Sjögreen et al., 2007).

## Level of Participation

Communicative participation is defined as "taking part in life situations where knowledge, information, ideas or feelings are exchanged" (Eadie et al., 2006). People with dysarthria are at risk for restricted communicative participation. Assessment of communicative participation must include patient-report, because the person living with dysarthria is the only one who can judge their participation problems. There are several questionnaires which can be used, like Living with Dysarthria (Hartelius, Elmberg, Holm, Lovberg, & Nikolaidis, 2008) and the Communication Participation Item Bank Questionnaire (K. M. Yorkston et al., 2007).

# Speech:
## Assessment of Children with Myotonic Dystrophy

For the assessment of speech of a child with congenital-onset myotonic dystrophy (CDM), it is recommended to start with a short conversation (spontaneous speech). Spontaneous or conversational speech is an important task to evaluate speech because it is the most representative task of daily communication. Spontaneous speech can be scored with a four-point scale[1] or with a scale developed for the severity of dysarthria, based on the Therapy Outcome Measure, ranging from 0 = normal, to, 5 = most severe.[2]

It is important to include a test for speech sound production (phonological and phonetic inventory), because for children with CDM, there is often a component of speech and language development. These tests are often performed with picture naming and evaluated with language based reference norms (for example, the Hodson Assessment of Phonological Patterns by B. Hodson).

The use of maximum performance tasks (maximum repetition rate, maximum phonation time, fundamental frequency range, maximum phonation volume) are recommended to facilitate the interpretation of specific characteristics that are observed in spontaneous speech, such as signs of weakness.[2]

These three different tasks will lead to a diagnosis related to speech. The diagnosis will often be a combined diagnosis of phonological problems and dysarthria, with weakness and myotonia of the face and tongue (problems with initiation of speech) as main problems.[3] Facial weakness is associated with problems with bilabial consonants, interdental articulation and hypernasal speech.[1] Phonological problems, defined as omissions and substitution of speech sounds, reflect cognitive-linguistic development.[4] Because cognitive delays are often noticed in children with CDM, attention to the development of speech sounds is important.[5]

# Speech:
## Symptoms and Diagnosis of Adults with Myotonic Dystrophy

Speech depends on phonation, respiration, articulation, and resonance. In individuals with adult-onset myotonic dystrophy type 1 (DM1), all of these areas may be affected, with features consistent with flaccid dysarthria with the following characteristics:[1–3]

- Hypernasality
  - Nasal emissions in more severely affected individuals
- Imprecision of speech
  - Especially for bilabial and labiodental phonemes secondary to reduced ability to approximate lips for adequate closure
- Slowed speaking rate
- Breathiness or fewer than average syllables produced per utterance
- Reduced vocal intensity
- Reduced use of prosody

Additionally, individuals with DM1 report lingual and jaw myotonia though exact prevalence is unknown.

Clinical speech evaluation of any individual with DM1 should include:

- Thorough history taking regarding duration, scope, and perception of symptoms
- Assessment of cranial nerves[4]
  - Function of lips
    - At rest
      - Frequently lips are in a "tented" posture and open
      - Observe symmetry, tone, size
    - In motion
      - Pucker
      - Smile
      - Alternating smile/pucker
      - Observe symmetry of movement, range of motion, strength, speed
      - Observe if patients are able to approximate lips
    - Presence of other movements
      - Myotonia, spasms, tremor, myoclonus, fasciculations, etc.

- Function of tongue
  - At rest
    - Observe symmetry, tone, size
  - In motion
    - Lateralization
    - Protrusion/retraction
    - Elevation/depression
    - Observe symmetry of movement, range of motion, strength, speed
  - Presence of other movements
    - Myotonia, spasms, tremor, myoclonus, fasciculations, etc.
- Function of jaw
  - At rest
    - Is jaw closed or open?
    - Observe symmetry, tone, size
  - In motion
    - Open/close
    - Lateralizing
    - Observe symmetry of movement, range of motion, strength, speed
  - Presence of other movements
    - Myotonia, spasms, tremor, myoclonus, fasciculations, etc.
- Soft palate
  - At rest
    - Observe symmetry, tone, size
  - In motion
    - Sustained elevation with /a/
    - Alternating between elevation/relaxation with repeated /a/s
    - Observe symmetry of movement, range of motion, strength, speed
  - Presence of other movements
    - Myotonia, spasms, tremor, myoclonus, fasciculations, etc.
- Oral agility/diadochokinetics
    - /pʌ/, /tʌ/, /kʌ/ and /pʌtʌkʌ/

Individuals with DM1 have been shown to have slower syllable repetition rates (diadochokinetics) prior to "warming up" the musculature used in speech.[3] Additionally, the variability of rate of syllable production decreases following warm up.

Thorough evaluation of orofacial musculature and function can help with the diagnosis and degree of dysarthria as well as document the presence or absence of myotonia. This information may be of benefit during therapy for identification of strategies to help minimize dysarthria.

# Speech:
## Symptoms and Diagnosis of Children with Myotonic Dystrophy

Speech disturbances are frequently reported in children with congenital-onset myotonic dystrophy (CDM) and childhood-onset myotonic dystrophy. Children with CDM generally have more impaired speech than children with childhood-onset DM.[1] A comprehensive speech language assessment is always necessary to judge the intelligibility and to diagnose speech problems. In addition, information from parents about the intelligibility of the child can be collected, for example with the Intelligibility in Context Scale (ICS) by McLeod (**https://www.csu.edu.au/research/multilingual-speech/ics**).

Speech problems in CDM and childhood-onset DM are multifactorial, therefore assessment should begin with a broad view, then progress to a more focused assessment as described in the section *"Symptoms and Diagnosis of Adults with Myotonic Dystrophy."*

# Speech:
## Therapy of Adults with Myotonic Dystrophy

Speech language pathologists help individuals learn to compensate for weakness of the muscles involved in speaking and to improve functional communication across communication partners and environments. Recommendations for speech and language therapy for myotonic dystrophy (DM) are usually based on clinical practice due to limited evidence-based research on treatment. The American Speech-Language-Hearing-Association (ASHA) has contrived two guidelines consisting of evidence-based or consensus-based recommendations to assist with management and treatment of DM. There is also one systematic review on muscular dystrophies.

Common challenges facing individuals with DM are muscle weakness of the face, tongue and jaw, especially the lips, tongue and velum musculature causing flaccid dysarthria (Ercolin, B., et al., 2013). Consequences of dystrophy and hypotonia of the speech musculature include hypernasality, resulting in decreased intelligibility. Consequences of decreased respiratory support/weakness of respiratory muscles are decrease in volume and shorter utterance lengths. Bushby, K., Finkel, R., et al. (2010) indicate that adult patients can benefit from compensatory strategies, voice exercises and speech amplifiers to improve intelligibility for those impacted by decreased respiratory support for speech and volume. It should be noted that for some individuals, voice exercises can be inefficient as the disease progresses and weak respiratory and/or speech muscles interfere with maintaining adequate breath support required for speech. Pacing methods or augmentative and alternative communication (AAC) might be more appropriate at that time. Individuals can also benefit from cognitive-linguistic therapy addressing deficits in cognition, executive dysfunction, and avoidant personality traits.

Frequency and duration of therapy services and goals will vary depending on the individual's needs. A speech-language pathologist may play a more consultative role by monitoring changes in speech and language skills, developing a home program with exercises, or providing a communication device to assist with increasing functional communication (Ho, G., Cardamone, M., & Farrar, M. 2015).

Articulation therapy can benefit clients with weakened speech. Due to weakened articulatory muscles, pressure consonants (/b, p, d, t, g, k, v, f/, "voiced and voiceless th, dj, sh, ch) can be affected. The clinician will determine if articulation exercises or compensatory strategies will increase speech intelligibility. The effect of compensatory strategies is directly clear but articulation exercises should be tried before one can hear the effect on intelligibility. Articulation exercises should be considered only in mild dysarthria, using principles of motor learning. Therapists can teach individuals how to slow their rate of speech and increase speech intelligibility, for example with on demand, pacing boards, prosody trainers. According to researchers, consistent muscle activity can result in a decrease in myotonia. A warm-up can help improve speech production. De Swart et. al (2006) found that a warm-up with two three-minute connected speech samples improved the number of syllables per second and repetition rates for the third three-minute connected speech sample. The study also found that patients who presented with at least a mild case of flaccid dysarthria showed no evidence of muscular fatigue. In addition, behavioral therapies such as Lee Silverman Voice Treatment and Clear Speech (Lam & Tjaden, 2012) could be useful in improving speech intelligibility in individuals who exhibit voice and articulation deficits.

Voice therapy can also benefit clients experiencing hypernasality as muscle weakness increases. Nose clips or palatal lift prosthesis may be considered in order to improve velopharyngeal closure due to velum weakness, as they have a positive effect on pressure consonants. For decreased vocal volume, voice amplifiers may be used to increase loudness without creating fatigue.

# Speech:
## Therapy of Children with Myotonic Dystrophy

The goals of speech interventions for children with myotonic dystrophy (DM) and speech impairment are to stimulate the development of speech, to improve or maintain the oral motor strength and range of motion that is a prerequisite for speech intelligibility, and to support communication. The child's individual intervention plan should be established closely with caregivers, clinical team members, and school personnel. Most children with DM will eventually use speech as their primary way of communication, however, delayed speech development is common (Sjögreen et al., 2007; Berggren et al., 2018). Early introduction of augmentative and alternative communication (AAC) to children with congenital-onset myotonic dystrophy (CDM) and their family members is fundamental to the development of speech, language and communication (O'Neill et al., 2018). Some children with childhood-onset DM may also benefit from AAC. Manual signs and picture support are especially important for children with DM who do not develop speech due to autism spectrum disorders (ASD) and/or intellectual disabilities (Ekström et al., 2009; Brignell et al., 2018).

Some children with DM will benefit from periods of more intensive speech therapy (ST) aimed at a specific goal. Motor learning principles including type of feedback, length and number of therapy sessions, and target complexity should be considered in treatment (Maas et al., 2008). Goals include improving lip articulation, producing specific speech sounds, and focusing on speech clarity. Hypernasality is a major cause for impaired intelligibility in DM caused by velopharyngeal incompetence, a symptom difficult to treat with speech therapy only (Sjögreen et al., 2018). Try a nose clip to explore if intelligibility improves, but if further intervention is needed, palatal lift prosthesis or pharyngeal flap surgery can facilitate the closing of the velo-pharyngeal port in selected cases. Periods of intensive ST are followed by periods where the SLP plays a more consultative role but still monitors the child´s development of speech, language and communication skills.

There are a variety of oral motor therapy tools available on the market; oral screens for improving lip strength, chewy tubes for improving jaw stability and jaw strength, palatal plates for facilitating speech sound production and z-vibes with different tips for activation and differentiation of lip, tongue and jaw motions fundamental for speech. The intervention program should be individualized to age, cognitive development, general condition, motivation and available support. It is important to have a baseline evaluation before implementation of oral motor exercises and to have regular follow-ups to monitor and modify the program when necessary. For children who cannot participate in structured exercises, lip, tongue and jaw muscles can be activated in daily activities such as singing, talking, eating, drinking, making facial expressions, chewing gum, blowing bubbles, playing on wind instruments, and making animal sounds.

# Speech:
## Preserving Skill Set of Adults with Myotonic Dystrophy

Advanced planning is essential for maintaining quality of life due to the gradual increase of symptoms over time in adult-onset myotonic dystrophy (DM). Management of the symptoms and their changes are critical to avoiding further complications and progression. The childhood-onset and congenital forms of DM appear earlier in life with more severe symptoms, consequently creating more challenges and different management techniques than the adult forms. It is important to note that all children with congenital-onset DM1 will develop the adult form of DM when they reach adolescence or adulthood. Lower intellectual ability can be a problem in congenital and juvenile-onset DM, thus requiring more dependence on caregivers and loved ones to help manage day-to-day operations.

Support through social networks is vital to ensure that speech therapy consultations are completed as deemed appropriate. Determining additional speech and language needs as the disease progresses is important to maintaining function. The individual's ability to demonstrate executive functioning skills such as planning and organizing their life becomes increasingly vital. Organizational and planning skills are important to completing tasks associated with independence. These skills include keeping a calendar to manage appointments, arrange schedules, and manage bill payments. Caregiver and communication partner training and education are needed to preserve speech and language skills. Encouraging patients to talk and verbally express themselves as much as possible can assist with preserving speech. This can include, but is not limited to, conversations with family and friends, reading a book out loud, having phone conversations, and reading the subtitles from a television show.

# Speech:
## Preserving Skill Set of Children with Myotonic Dystrophy

In order to preserve speech for children with myotonic dystrophy, consider communication interventions such as the early introduction of AAC (augmentative and alternative communication) to facilitate communication and stimulate speech development in children with delayed speech. For preserving oral sensory-motor development skills, introduce toys and mouth play to activate the oral muscles and give tactile stimulation. To continue language development, stimulate language through language awareness and practice. For hearing, make sure that the child has no hearing impairment that could interfere with speech and language development. Therapists can encourage speech production by teaching the child to use both lips when learning to say words including bilabial consonants (/m/, /b/ and /p/). Introduce strength training to improve or preserve oral muscle strength and speech clarity, including lip exercises (blowing, straw drinking with thicker liquids, training with an oral screen), jaw exercises (with food, chewing gum, chewy tube), and tongue exercises (pushing the tongue against the palate and cheeks or against a tongue depressor, a toothbrush or a z-vibe).

# Swallowing:
## Assessment of Adults with Myotonic Dystrophy

Pneumonia is one of the leading causes of death in myotonic dystrophy type 1 (DM1)[1,2] and aspiration when eating is a significant factor in the development of pneumonia. Swallowing difficulties (dysphagia) are common in DM1, however reports on prevalence vary from 25%– 80%.[3] Individuals with DM1 report swallowing as one of the symptoms of concern.[4,5] A screening of swallow function in DM1 should be completed at least annually[6] with a referral to a speech-language pathologist if symptoms of dysphagia are present or suspected. An assessment of swallowing function entails a thorough history taking regarding the symptoms present when eating and/or drinking. Assessments should include:[7]

- Case history
  - Interview patient and care partners, review available medical records
  - Patient insight into symptoms and frequency of dysphagic events
- Assessment of anatomy and physiology
  - Cranial nerve function
  - Oral cavity shape
  - Facial, oral, and pharyngeal strength
  - Presence of myotonia in tongue and/or jaw
  - Esophageal motility issues and myotonia
  - Reflux
  - Coughing when eating, particularly with drier foods and/or thin liquids
  - Head and neck control and overall posture
  - Respiratory function including cough strength
  - Incidences of respiratory infections and/or pneumonia
- Assessment of behavior
  - Oral hygiene
  - Are there any foods that are avoided?
  - How many meals/day and what are the length of mealtimes
  - Unintended weight loss or gain
  - Rate of ingestion
  - Bite and sip size
    - Use of straws
    - Do they drink large volumes at a time?
  - Thoroughness of mastication
  - Are there distractions while eating
  - Compliance with common strategies for eating safely
- Clinical swallow evaluation

Additionally, an instrumental evaluation can be performed to look more thoroughly at anatomy and physiology during feeding and swallowing. There are two commonly used instrumental analyses that may be performed; FEES (fiberoptic endoscopic evaluation of swallow), and VFSS (videofluoroscopic swallow study). For the clinical swallow evaluation and any instrumental analyses, food and liquids of various consistencies should be trialed. Additionally, bolus size and method of presentation of liquids should be varied. Strategies to help improve safety of swallow can be trialed at this time as well.

# Swallowing:
## Assessment of Children with Myotonic Dystrophy

Congenital-onset myotonic dystrophy (CDM) is characterized by severe hypotonia and weakness at birth, often with respiratory insufficiency, feeding difficulty, respiratory failure, intellectual disability and autistic features as the child gets older.[1] A multidisciplinary treatment approach is recommended with a speech language pathologist (SLP) to assess feeding and swallowing issues, starting at birth. Because of the wide range of severity of symptoms in CDM, a tailored health care approach is recommended.[2]

Newborns with CDM often have profound difficulties with sucking and breathing because of neonatal hypotonia.[3] These infants often experience problems in the development of eating and drinking functions in the first years of their life, such as eating from a spoon and chewing solid food. Therefore, it is important that a comprehensive assessment by an SLP is performed as soon as possible and is repeated over the years.[4] This assessment is focused on 4 major issues: (a) orofacial anatomy and muscle tone; (b) suck-swallow-breath capability in neonates and efficient swallowing in older infants; (c) state of the infant before, during and after feeding; (d) parents' concerns about feeding issues.

# Swallowing:
## Symptoms and Diagnosis of Adults with Myotonic Dystrophy

Adults with myotonic dystrophy type 1 (DM1) commonly demonstrate reduced strength in oral, facial, and pharyngeal musculature leading to difficulty swallowing (dysphagia). Some may also report myotonia of the jaw and/or tongue. Symptoms of dysphagia can range from very mild to severe and often increase in severity during the lifespan. Diagnosis via a thorough assessment can include eating/swallowing questionnaires, disease specific patient reported outcomes with dysphagia questions,[1,2] clinical swallow evaluations, and instrumental evaluations.

Common symptoms reported:

- Facial, oral, and pharyngeal weakness
  - Labial weakness and reduced ability to create good labial seal can lead to anterior loss of solids and liquids and inability to use straws.
  - Lingual weakness can lead to reduced oral manipulation of food and poor propulsion in the oral stage of swallow.
  - Nasal regurgitation is possible if soft palate movement is reduced.
- Presence of myotonia in tongue and/or jaw
- Esophageal motility issues and myotonia
- Reflux
- Fatigue when chewing
- Coughing when eating, particularly with drier foods and/or thin liquids
- Difficulty with morning and evening meals
  - Fatigue later in the day is commonly reported in DM1 and patients often report increased coughing, longer mealtimes, and increased difficulty with more complex food items.
  - Some individuals with DM1 benefit from warming up muscles for optimal use.[3]
- Reduced oral hygiene habits
  - More severely affected individuals can present with open-mouth posture and altered dentition.
- Eating too fast
  - Gulping liquids
  - Not chewing thoroughly
  - Taking very large bites and not clearing oral and pharyngeal cavities clearly between bites

- Becoming distracted while eating
- Reduced adherence to safe swallow strategies
- Reduced insight and/or interest in health and potential complications
    - This symptom is more commonly reported by care partners or observed over repeated clinical visits when adherence to safe swallow strategies is limited.

Additionally, if an instrumental analysis is performed, abnormal anatomy and physiology is often observed. Modified barium swallow and electrophysiological studies show:[4,5]

- Incomplete laryngeal closure
- Incomplete oral and pharyngeal clearance
- Reduced upper esophageal sphincter opening
- Incomplete closure of the nasopharynx with the soft palate
- Delayed initiation of oral and pharyngeal phases of swallow
- Prolonged oral and pharyngeal transit times of the bolus
- Delay in initiation of movement of structures in oral and pharyngeal cavities
- Reduced pharyngeal constriction resulting in residue in pharynx
- Early opening of upper esophageal sphincter and delayed closing

# Swallowing:
## Symptoms and Diagnosis of Children with Myotonic Dystrophy

The orofacial anatomy of infants with congenital-onset myotonic dystrophy (CDM) is characterized by hypotonia and open mouth posture (tent shaped mouth), which is not normal in healthy newborns.[1] It is described that the tent shaped mouth, in combination with polyhydramnios due to reduced fetal swallowing is already present intrauterine.[2] The orofacial function often shows impaired facial expression, drooling, reduced muscle strength and open mouth posture.[3] These features will lead to open bites and other occlusal contact problems. Together with reduced strength, these features will hamper the development of eating, drinking and speech.

The suck-swallow-breathe sequence consists of the ability to make a sufficient vacuum for sucking and the coordination between sucking, swallowing and breathing. In the neonatal period, the usual SLP assessment consists of an observation of drinking from the bottle or breast. This observation is preferably quantitatively using the Neonatal Oral Motor Assessment Scale.[4] Cervical auscultation during drinking to detect swallowing and the coordination with inspiration and expiration is useful.[5] The development of subsequent eating functions (spoon feeding and mastication) should be carefully monitored in order to support orofacial development, and safe and sufficient intake.

Because of severe hypotonia and respiratory problems, the condition of a newborn with CDM is often limited. Careful observation of the infant's state and signals of exhaustion during and after feeding is important, for example with the Early Feeding Scale (EFS).[6] Although this scale is primarily for preterm infants, it can also be used in term born infants. Because of hypotonia and reduced facial strength, mealtimes can be prolonged and influenced by fatigue, thus food adaptations (making food easier to eat) must be considered.[7] If there are concerns about safe swallowing (e.g., aspiration), a videofluoroscopic swallow study (VFSS) should be performed. Tube feeding (or additional tube feeding) might be necessary in the case of aspiration, respiratory problems or reduced condition.

Parents with a child with CDM with feeding or swallowing problems have greater challenges and stresses than those of typically developing children.[8] Attention to these problems and specific support is necessary. The Feeding/Swallowing Impact Survey (FS-IS) as an instrument designed to measure and improve understanding of caregiver issues and can be helpful for this parent group.

# Swallowing:
## Therapy and Preserving Skill Set of Adults with Myotonic Dystrophy

Historically, the idea of swallowing therapy in neuromuscular diseases has been discouraged out of concern for overtaxing the muscles with exercise and potentially leading to even greater disability. There is no literature to date indicating a specific dysphagia therapy that is most efficacious for adults with myotonic dystrophy type 1 (DM1).[1] More recent studies in skeletal muscles in general have shown that moderate intensity exercise in DM1 is generally not harmful though the benefit is unclear.[2,3]

Literature regarding the rehabilitation of swallow functioning following neurologic injury (cerebrovascular accident, traumatic brain injury, tumor resection, etc.) indicates that the McNeil Dysphagia Therapy Program, expiratory muscle strength training (EMST), and exercises such as Mendelsohn, Masako, and Shaker seem to have the most promise.[4] While there is ongoing research into frequency, intensity, and duration of exercise in the rehabilitation field, there has not been a consensus to date in the dysphagia literature on these factors for any patient population.

More recent literature has shown that EMST in amyotrophic lateral sclerosis (ALS) can help maximum inspiratory pressure and hyoid displacement during swallowing, though it is unclear if this will improve swallowing during daily life.[5] These are promising results for a neuromuscular disease, but more research is needed in general for dysphagia treatment in neuromuscular diseases and in DM1 specifically.

In general, for individuals with DM1 who are interested in therapeutic exercise targeting potential improvement in swallow function, these exercises should be done cautiously under the guidance of a speech-language pathologist, preferably one familiar with neuromuscular diseases.[6]

# Swallowing:
## Therapy and Preserving Skill Set of Children with Myotonic Dystrophy

Dysphagia may affect one or more phases of swallowing; the oral, the pharyngeal or the esophageal phase. Impaired swallowing may also be the cause of saliva leakage ("drooling"). The goals of the SLP intervention are to support feeding development, safe and effective swallowing, and enjoyable mealtimes. Ideally, the SLP is part of a multidisciplinary team and is well informed about the general condition of the patient and current interventions. In congenital-onset myotonic dystrophy (CDM), the SLP may be involved in feeding therapy when the child is born and then will continue to monitor and stimulate the feeding development throughout childhood. In childhood-onset DM, dysphagia may occur as the child grows older and has increased muscle weakness. Close cooperation with caregivers and other professionals involved in the management of the child is essential. School personal should support the child at mealtimes, though supervision from an SLP is often needed for specific exercises.

Neonatal hypotonia in newborns with severe CDM leads to difficulties with breathing and sucking. In most cases, these children will need tube feeding during the first weeks of life or for a longer period. Some infants may be able to suck from a feeding bottle with an enlarged hole in the nipple or a special care feeding bottle that can regulate the milk flow and facilitate sucking. The SLP can help the caregivers find the safest and most effective feeding equipment. The occupational therapist (OT) can try a feeding position that provides stability and is efficient for feeding. Some children will also need chin and lip support from the feeder (ASHA). A dietician can create a menu that meets the nutritional needs of the child. Children with swallowing difficulties often have gastrointestinal manifestations that needs treatment. Mealtimes should be safe but should not take too long to eat a full meal. The ability to chew and swallow, not the age of the child, determines which food consistency is accurate to serve the child. Specialized feeding equipment can sometimes facilitate sucking, eating and drinking. An OT or SLP can help the family find the most effective feeding bottle, spoon or cup for the child. Children with severe dysphagia may require thickened liquids to prevent aspiration to the airways. Tactile stimulation by finger stroking of lips, cheeks, gums, tongue and palate is often recommended to activate the oral muscles and to prevent oral hypersensitivity in children who are tube fed (nasogastric tube or gastrostomy) (ASHA; Tian X et al., 2015).

An oral sensory-motor intervention program should include exercises aiming at improving or maintaining muscle strength and range of motion. There are a variety of therapy tools available on the market, for example, oral screens for improving lip strength, chewy tubes for chewing exercises and Z-vibes with different tips for activation of lip, tongue and jaw muscles. The therapy program should be individualized to age, cognitive development, general condition, motivation and available support. It is important to have a baseline evaluation before implementation of oral sensory-motor exercises and to have regular follow-ups to monitor and modify the program when necessary. Daily activities include stimulation and activation of lips, tongue and jaw, including singing, talking, eating, drinking, brushing teeth, making facial expressions, chewing gum, playing with mouth toys, chewing on "chewelry", blowing bubbles and playing on wind instruments. It is important to note that there is a disagreement between some researchers regarding 'functional training' (using food in different consistencies to learn to chew and swallow) and oral sensory-motor intervention.

Oral health care is of extra importance for children with impaired swallowing and risk for aspiration of food, liquids or saliva. Good occlusion and jaw function as well as enough saliva production are important for effective chewing. A referral to specialized dental care is therefore recommended when indicated.

## Recommendations for infants and younger children

### Tactile stimulation of cheeks, lips, gums, tongue and palate

**Goal:** To activate the oral muscles by sensory-motor stimulation and to prevent oral hypersensitivity in children who are tube fed (nasogastric tube or gastrostomy).

**Intervention:** Give oral tactile stimulation by:

- Finger stroking.
- A vibrating therapy tool (z-vibe with different tips).
- An electric toothbrush.

### Mouth toys and mouth play

**Goal:** To activate the oral muscles and to stimulate the oral sensory-motor development.

**Intervention:**

- Offer the child sensory-motor stimulation through mouth play and mouth toys of various textures.
- Make sure the toys are non-toxic and safe to suck, bite and chew on.

### From bottle feeding to cup drinking

**Goal:** To facilitate the maturation of the swallowing pattern (without tongue protrusion).

**Intervention:**

- Introduce cup drinking as soon as the child can sit without support.
- Avoid sippy cups if they stimulate tongue protrusion.

### Spoon feeding

**Goal:** To activate lip closing and lip rounding.

**Intervention:**

- Await the child´s own attempt to empty the spoon with lips.
- You can stimulate lip rounding by feeding from the tip of the spoon.
- You can stimulate lip closing by feeding from the side of the spoon.

## Recommendations for children and adolescents

### Straw drinking

**Goal:** To activate lip closing, lip rounding and tongue retraction.

**Intervention:** To have a stimulating effect, only a short piece of the straw should go inside the mouth. If this is difficult, a lip block can be used.

### Strength/mastication training

**Goal:** To improve or preserve oral muscle strength.

**Intervention:** Exercise 5 minutes, 3-5 days a week .

- Lip exercises (blowing, straw drinking with thicker liquids, training with an oral screen).
- Jaw exercises (with food, chewing gum, chewy tube).
- Tongue exercises (pushing the tongue against the palate and cheeks or against a tongue depressor, a toothbrush or a z-vibe).

(Rosenfeld-Johnson S; Morris SE & Klein MD)

## Special dental care

Oral health, dental occlusion and jaw function have a major impact on the ability to chew and swallow safely. All children with DM should have regular dental check-ups and receive preventive oral health care (Mårtensson et al., 2016).

# Language:
## Assessment of Adults with Myotonic Dystrophy

Language assessment for adults with myotonic dystrophy (DM) should be completed by a licensed speech-language pathologist. A speech and language evaluation will assess memory, expressive language, receptive language, auditory comprehension, problem solving, planning sequencing, attention and writing. A detailed medical and developmental history will be collected. When appropriate, patients and caregivers can also determine areas that impact their activities of daily living. Appropriate goals for therapy will be dependent on the clinician and patient determining areas that impact activities of daily living. Adults can be assessed at an outpatient center or department of rehabilitation in their area. A summary of the evaluation as well as specific recommendations for treatment and/or additional services will be provided to the family.

# Language:
## Assessment of Children with Myotonic Dystrophy

Language assessment for children with myotonic dystrophy (DM) should be completed by a licensed speech-language pathologist. The assessment can combine standardized (when appropriate) and non-standardized methods, informal observation, a review of medical records, and family input to develop an accurate picture of a child's needs. A speech-language pathologist will assess receptive and expressive language, pragmatic language, play skills, gestures, vocabulary and articulation. If suspected, other areas such as oral motor weakness, feeding and swallowing, apraxia of speech, pragmatics, fluency and auditory processing will be assessed. The evaluation can be conducted through the child's local school district, outpatient services/clinic, or early intervention programs. A summary of the evaluation as well as specific recommendations for treatment and/or additional services will be provided to the family.

# Language:
## Symptoms and Diagnosis of Adults with Myotonic Dystrophy

Language symptoms in adult-onset myotonic dystrophy (DM) can include deficits in intellectual and/or cognitive function, social and/or emotional impairments, hearing impairment, and receptive and expressive language impairment. Some individuals will present with a mixed receptive and expressive language impairment while others will only have speech sound production difficulties due to poor muscle strength. Adults with DM may experience difficulties with reading and writing.

# Language:
## Symptoms and Diagnosis of Children with Myotonic Dystrophy

Language symptoms in children with congenital-onset myotonic dystrophy (CDM, ages 0-3) include developmental delays in speech or motor skills, which may impact the development of language skills due to limited practice with language. Lowered cognition and psychological symptoms, including attention deficit disorder and autism spectrum disorder (ASD), can impact speech and language production. Children with CDM have receptive language impairments, including difficulty processing and understanding language. They also have impairments in expressive language, with difficulty using vocabulary appropriately in social settings with peers of a similar age and gender.

Language in childhood-onset myotonic dystrophy (sometimes referred to as juvenile-onset myotonic dystrophy, ages 3-21) can be affected by intellectual or cognitive function, thus may require assistive technology, devices, executive functioning support, and speech therapy to improve language skills. Children with myotonic dystrophy (DM) often have Autism, developmental delays, emotional disturbance, intellectual disabilities, and other health impairments (attention deficit disorder, attention deficit hyperactivity disorder). They can be diagnosed with speech and/or language impairments, including cognitive-communication disorders, difficulty with communication skills involving perception, memory, organization, problem solving and other cognitive functions. They also can be diagnosed with social language disorders, meaning difficulty understanding the meaning of what others are conveying, communicating their desires and needs with others, and difficulty interacting with others. Children with DM have impairments with receptive language, including difficulty processing and understanding language and limited skills drawing meaning from language and communication. They can also have impairments in expressive language, meaning difficulty putting words together to convey ideas, wants and needs. Some children with DM have hearing impairments and difficulties with reading and writing.

# Language:
## Therapy of Adults with Myotonic Dystrophy

The primary focus for language therapy for adults with myotonic dystrophy (DM) is to repair communication breakdowns between patients and their communication partners. Speech therapy supports language maintenance and improvement in the following areas: listening/perception, vocabulary development, and understanding/comprehension of functional information. Effective therapy supports cognitive and emotional development, while reinforcing social skills. Therapy supports adults with their communication exchanges in diverse settings. Therapy can also improve quality of life by building self-confidence and self-esteem, increasing the ability to interact in social settings, increasing independence, increasing problem solving skills, and increasing the ability to express thoughts and ideas with novel and familiar communication partners.

# Language:
## Therapy of Children with Myotonic Dystrophy

Language therapy for children with myotonic dystrophy (DM) is designed based on the needs of the child. Therapy for each child will look different depending on the needs determined during the evaluation. The speech-language pathologist will use their knowledge of communication development and the needs of the child to structure the therapy session. At the end of each session, the caregiver should receive information on how treatment went and the targets addressed within the session. Strategies and homework are reviewed with the caregiver to support consistent practice in natural settings. Evidence based practices confirm that parent or caregiver involvement is essential for treatment success, so the SLP should involve these groups whenever possible. For children with DM, an early introduction of augmentative and alternative communication (AAC) is recommended in order to stimulate communication and language development.

# Language:
## Preserving Skill Set of Adults with Myotonic Dystrophy

Preserving language in adults with myotonic dystrophy (DM) is necessary due to the complexity and nature of the disease as well as meeting the person's individual needs. These needs will greatly depend on the patient's educational level, cognitive abilities, pragmatic functioning and skills, and challenges with executive functioning. Any deficits in these areas can impact the individual's ability to make friends, make appropriate choices to limit risks, and participate in novel activities.

Research notes that individuals with myotonic dystrophy may be uninterested and impartial in their health, possibly due to apathy and personality issues that are present for some patients. Furthermore, we can see an impact on the individual's ability to maintain relationships, continue higher-level education, complete activities of daily living (ADLs) and instrumental activities of daily living (IADLs), and maintain employment. Due to the possible cognitive and psychosocial changes in individuals with DM, a speech-language pathologist (SLP) can be an important support figure and resource. SLPs can support cognitive and psychosocial changes through education on current and possible future functioning. SLPs can also assist patients and caregivers with the development of compensatory strategies to increase independence and quality of life within the limitations of the individuals existing abilities.

During the assessment, clinicians should note cognitive difficulties, including:

1. Impaired recall of information
2. Problem solving
3. Temporal orientation and schedule management
4. Impaired social participation

Clinicians can assist with:

1. Developing compensatory calendars and schedules to assist the patient with activity participation.
2. Developing routines
3. Simplifying complex tasks to support safety
4. Enhancing problem solving skills
5. Setting reminders for medication management

It is imperative that the individual has a good support system to assist with maintaining skills and encouraging engagement with others. Good support can help improve an individual's quality of life. This support can be from family, friends, caregivers, psychologists, or other medical professionals.

# Language:
## Preserving Skill Set of Children with Myotonic Dystrophy

For children with congenital-onset myotonic dystrophy (CDM), it is important for the family to target rehabilitation of deficits in receptive and expressive language, social development, and cognitive growth using age-appropriate developmental care and infant stimulation programs. Support from medical professionals (e.g., speech-language pathologist, occupational therapist, special education, behavioral interventions, etc.) is imperative to continue to support learning and overcome the many language difficulties impacting children with CDM. Moderate to severe intellectual disability (ID), severe problems regarding adaptive skills, Autism spectrum disorders (ASD), attention deficit disorders without hyperactivity, and anxiety can be comorbid conditions with CDM.

It is recommended that children with congenital-onset myotonic dystrophy have assessments upon diagnosis, at preschool age, and 2-3 times before adulthood. Children with CDM in the United States generally have at least a 504 Plan, if not an individualized edustion plan (IEP). Speech and language assessment intervals before adulthood will depend on the child's level of functioning, though are recommended to be done yearly. Modifications to the child's treatment plan, long-term, and short-term goals will be determined at the time of these assessments. For children with CDM who have skeletal muscle weakness, speech therapy may require specific attention to language acquisition delays, as well as the potential for augmentative and alternative communication (AAC) needs.

Ongoing communication between health care professionals and the child's school district should be encouraged to develop educational modifications and strategies, as well as the child's IEP, which is updated yearly.

It is important for parents to be aware of the mental health differences that a child with CDM or childhood-onset DM1 may experience, which may impact overall functioning and preservation of language. Although not all children with DM will experience anxiety and depression, families should be aware of the many challenges that children may face as their awareness of others' perceptions of their differences increase. Adherence to social and behavioral norms become more expected around middle school, thus families should be extra attentive to their child's needs at this stage. Some children with DM may need support to access specific school curriculum and extracurricular activities that support learning, in order to navigate and express their feelings with others.

Children with CDM or childhood-onset DM1 may also demonstrate emotional responses to their situation, such as hostility and depression due to the feeling of learned helplessness. Mental health conditions, such as anxiety and depression can be noted in some children with DM. Emotional responses can include hostility and depression due to the feeling of learned helplessness. This feeling of helplessness or being stuck can lead to emotional reactions, such as apathy and depression, and behavioral problems like tantrums. During this time an individual can decrease communication with others, which impacts their preservation of language. Clinicians can assist by having the child complete activities that they succeed in to create a positive experience. For the child, an increase in control can lead to an increase in motivation to continue learning and/or engaging with others.

# Language:
## Functional Communication for Work and Home for Adults with Myotonic Dystrophy

Maintaining functional communication is essential for quality of life for an individual with myotonic dystrophy (DM). Research has noted that in some cases, cognitive impairment, executive dysfunction, and avoidant personality traits are noted to deteriorate with age for individuals with DM. These resulting communication restrictions can affect social relationships and education. Adults with DM and their caregivers should be educated about the impact and impairment that DM can have on cognition and behavior. Caregivers and significant others have the role of monitoring the appropriateness of the individual's augmentative and alternative communication device and assisting with updating the device as needed. This process can include removing and adding relevant vocabulary words.

It is important for caregivers to assist with motivating the adult with DM to complete daily living tasks and engage with family and community members. One role of caregivers is to encourage independence when ordering food, paying for expenses, and making appointments, using functional language. Clinicians can facilitate functional language learning by creating goals that address greetings, such as introducing yourself, asking for/giving advice, explaining rules, apologizing, answering personal questions, and agreeing/disagreeing. Clinicians can educate caregivers on their role of educating community members and extended family to understand the personality differences, challenges, and social irregularities that a person with DM may exhibit.

# Language:
## Functional Communication for School and Home for Children with Myotonic Dystrophy

It is important for the child to have a strong supportive network consisting of family, friends, psychotherapists, and healthcare professionals, as well as other members of the myotonic dystrophy (DM) community who live with the disease and have life experiences similar to their own. Speech therapists can help clients with communication skills that improve independence in settings such as school, supported or unsupported employment, continuing education, higher education, and community outings. Speech therapists can assist the child with expression of fundamental needs or preferences in the home, interpersonal relationships with family, peers, educators, counselors, and other professionals on their multidisciplinary team, and overall self-advocacy. It is important for the child to be able to carryover strategies and techniques learned in therapy to the home, community, and work environments, thus the speech therapist should consider this in their treatment plan.

Depending on the severity of DM, augmentative and alternative communication (AAC) devices can supplement the natural speech of children with DM, which can assist with functional communication. When determining best practices, it is important to consider the impact that myotonia has on a child with DM, specifically function that is hindered by muscle contraction and release. AAC systems should be programmed to support expressive communication and highlight/complement the individual's stronger receptive language abilities. Although research evaluating the use of AAC for individuals with DM is severely lacking, studies in other neuromuscular diseases revealed that quality of life improves with the use of AAC. AAC systems also provide assistance based on the individuals changing needs. AAC devices can vary from low-tech to high-tech devices. An example of a low-tech device is an alphabet board; this type of device can be used to assist the individual by providing an initial sound or letter to the approximated word.

Visual speech aids such as alphabet boards can also be used to eliminate the misinterpretation of initial sounds in words, thus reducing communication breakdowns. A high-tech device can be computerized, such as dedicated communication devices or Apple iPads that use communication applications.

The type of AAC device used greatly depends on the client. Low-tech AAC or visual speech aids have been successful for younger children who are developing language. These devices can include portable communication books, Boardmaker addendum libraries, picture cue cards, large communication books, and varied size communication boards.

# Appendix
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## Section 3

**Language (all parts)**


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Myotonic's mission is to enhance the quality of life of people living with myotonic dystrophy and accelerate research focused on treatments and a cure.

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