Orofacial dysfunction in children and adolescents with myotonic dystrophy

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Myotonic dystrophy (DM) is a neuromuscular disorder caused by an expansion of a CTG repeat sequence on chromosome 19q13. The aim of the present study was to describe the characteristics and prevalence of oral motor dysfunction in a cohort of children and adolescents with DM and to correlate different aspects of oral motor function with the type of DM and sex. Fifty-six individuals with DM (30 males, 26 females; median age 13y 2mo; range 2y 6mo–21y 5mo) were compared with healthy controls. They were divided into four subgroups: severe congenital DM (*n*=18); mild congenital DM (*n*=18); childhood DM (*n*=18); and classical DM (*n*=2). A speech-language pathologist assessed different variables of oral motor function, intelligibility, and lip force. The families used a questionnaire to report on eating difficulties and drooling. All individuals with DM had impaired facial expression. Intelligibility was moderately or severely reduced in 30 patients (60%), excluding six patients without speech. Most had a moderate or severe impairment of lip motility (76.0%), tongue motility (52.2%), and lip force (69.2%), causing deviant production of bilabial and dental consonants. The families reported problems with eating (51.9%) and drooling (37.0%). Oral motor dysfunction was most prominent in congenital DM, and males were more affected than females.

Myotonic dystrophy (DM) is a slowly progressive neuromuscular disorder with autosomal dominant inheritance. It is caused by an expansion of a CTG repeat sequence (trinucleotide expansion) on chromosome 19q13. The number of CTG repeats correlates broadly with the overall severity of the disease, but the correlation between the size of the CTG repeat sequence and individual clinical manifestations still needs to be elucidated.[1] DM can be congenital and can appear in childhood or later in life (adult or classical type). The cardinal symptoms are weak muscles, especially in the face, neck, hands, and feet, but smooth muscles are also affected.[2] Myotonia is a common feature in adults with DM but this can also be seen in children.[2–5] Most individuals with the congenital or childhood type have learning disability* and there are an increased number of children and adolescents with DM who have a neuropsychiatric disorder in comparison with the prevalence in the general population.[4–7]

Newborn infants with the congenital form of DM generally have profound difficulties with sucking and breathing because of neonatal hypotonia. Polyhydramnios during pregnancy, caused by poor fetal swallowing, is often noted.[2,4,5] Adults with DM commonly develop flaccid dysarthria with indistinct articulation and hypernasal speech caused by velopharyngeal impairment.[8–11] The speech characteristics of children and adolescents with DM have not been described previously in any detail. Although orofacial weakness is a characteristic symptom in congenital and childhood DM,[2,4,5] research into the consequences for feeding in infancy, chewing, swallowing, and speech is very limited. Different aspects of oral motor impairment in children and adolescents with DM still need to be elucidated. Are there any quantitative or qualitative differences in oral motor functions between the different subgroups of DM or between males and females? The aim of this study was, therefore, to describe the characteristics and prevalence of oral motor dysfunction in a cohort of children and adolescents with DM and to correlate different aspects of oral motor function with the type of DM and with sex.

Method

STUDY POPULATION

All children and adolescents (*n*=63) with a confirmed diagnosis of DM living in western and southern Sweden (3 million inhabitants) were invited by their paediatric neurologist to participate in a multidisciplinary study. A paediatric neurologist and a physiotherapist met all the patients, made a clinical medical examination, took a medical history, and reviewed the records. The patients were then divided into four subgroups according to age at onset and the clinical picture.[2,3] Congenital DM was divided into a severe form and a mild form depending on whether or not the patient had had a life-threatening condition at birth.[3] The diagnostic criteria for childhood DM were symptoms presenting between 1 and 10 years of age and an uneventful prenatal and postnatal history. In classical DM the first symptoms occurred at 10 years of age or later.[12]

For the present study, cross-sectional data on orofacial dysfunction in 56 diagnosed children and adolescents with DM aged between 2 years 6 months and 21 years 5 months (median 13y 2mo) was collected. Subgroups, sex distribution, and age at examination are presented in Table I. At the time of assessment 10 children were of preschool age (2y

*North American usage: mental retardation.

6mo–5y 8mo). They all had the congenital type of DM and delayed general development. Thirty-seven of the older children and adolescents went to special schools for pupils with learning disability and the remaining nine attended ordinary schools. A control group recruited from the Public Dental Service Clinic at the Department of Odontology, Göteborg University, consisting of 56 healthy children, was matched by sex and age before the collection of data.

Informed consent was obtained from each family and the study was accepted by the Ethics Committee of the Medical Faculty at Göteborg and Lund universities.

ORAL MOTOR AND SPEECH ASSESSMENT

The orofacial function and dysfunction described in this study were assessed by a speech-language pathologist in accordance with a standardized protocol for the examination of oral motor function[13] (Table II). Evaluation of spontaneous facial expression and intelligibility was included in the protocol. The assessment was made from video recordings obtained at a dental clinic close to where the patients lived. The digital video camera (Sony Handycam, 3 megapixels; Sony Corporation, Tokyo, Japan) was placed on a tripod about 1.5m in front of the patient. Each variable was scored on a 4-point scale (Table II). The rest position of the lips and tongue was observed during 1 minute while the patient was watching a picture. Lip motility was tested by eight variables, and tongue motility by four (Table II). If the patients were assessed to have moderate or severe impairment on any of the variables tested they were considered to have dysfunction in that specific area. The ability to produce bilabial, dental, and velar consonants was assessed in a repetition test with simple words.

LIP FORCE

Lip force was measured with the LF100 instrument (Detektor AB, Göteborg, Sweden). A prefabricated oral screen (Ulmer

Table I: Myotonic dystrophy (DM) group divided into four subgroups, sex, and age distribution at the time of examination

Subgroup (males/females)	*n*	*Age (y:mo)*[a]
Congenital DM, severe (14/4)	18	9:10 (2:6–21:5)
Congenital DM, mild (8/10)	18	13:2 (3:3–18:7)
Childhood DM (8/10)	18	13:10 (8:3–20:10)
Classical DM (0/2)	2	17:0 (16:6–17:6)
Males	30	10:10 (2:6–21:5)
Females	26	14:6 (3:3–20:10)
Total (30/26)	56	13:2 (2:6–21:5)

[a]Results are medians (range).

Table II: Variables scales and definitions for oral motor and speech assessment

Variables	*Scales and definitions*
Facial expression	0=Normal function 1=Mild deviation 2=Moderate deviation 3=Severe deviation
Rest position of lips while watching a picture for 1 minute	0=Closed mouth or changing between closed and half-open 1=Half-open mouth 2=Half-open to wide-open mouth 3=Wide-open mouth
Rest position of tongue while watching a picture for 1 minute	0=Tongue is inside teeth 1=Tongue is sometimes outside teeth (less than half the time) 2=Tongue is outside the teeth more than half of time 3=Tongue is constantly outside teeth
Intelligibility; spontaneous speech	0=Speech is fully understood 1=Speech is largely understood; repetitions and verifications are occasionally needed 2=There is an ongoing need for repetitions and verifications; listener effort is required 3=Only a few words or phrases recognizable; alternative and complementary methods of communication are required
Lip function; active lip closure for 20 seconds; smacking with lips 5 times; blowing out a candle; showing teeth; smiling and pouting, 5 times; lip closure to spoon; lip closure while chewing; lip closure to a sucking straw	0=Normal range of movement and coordination for age 1=Slightly reduced range of movement and/or slightly reduced coordination compared to peers 2=Clearly impaired range of movement and or coordination, position or target is reached with effort 3=Severely affected range of movement and coordination, position or target is not reached
Tongue motility; pushing out tongue; licking upper lip; licking lower lip; moving tongue between corners of mouth, 5 times	See lip function above
Sound production; repetition of simple words	Production of bilabial, dental, and velar consonants

The tests are those described in reference 13.

model; Dentarum, Pforzheim, Germany) was attached to a handle by a string and the handle was connected to the measuring instrument. The oral screen is made in two sizes and the smaller one was used for children younger than 7 years old. The patient was seated during the test. The oral screen was placed inside the lips and the patient was told to try their best to keep the screen inside the lips while the examiner pulled the handle. The instrument saved the highest value measured during a 10-second period. The best of three values obtained was compared with the result from a matched control.

QUESTIONNAIRE

The parents and/or the patient answered with either 'yes' or 'no' according to whether they had any of the problems with eating or drinking specified in the questionnaire.[14] If there was any problem with saliva control, they were asked to specify whether the drooling caused saliva on the lips only (slight drooling), on the chin (moderate drooling), on the clothes (severe drooling), or on hands and objects (profuse drooling).

RELIABILITY

To study the inter-observer and intra-observer reliability of the oral motor and speech assessment, video registrations of 40 participants (20 cases and 20 controls) were randomly chosen for evaluation by a speech-language pathologist not involved in the study, and for re-evaluation by the first observer. Only the first observer was informed about the category of DM. Inter-observer and intra-observer reliability was good or excellent for most variables. The mean of the exact (point by point) percentage agreement was 84.3% (range 71.0–100%). If the results were divided into two categories, either 'no to mild deviations' or 'moderate to severe deviations', the percentage agreement was 94.91% (86.6–100%). The percentage agreements for intra-observer reliability were 90.7% (80.6–100%) and 94.7% (89.0–100%) respectively.

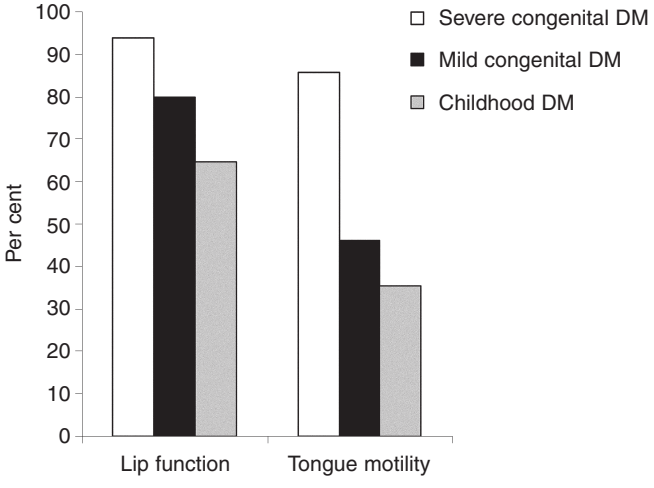

Figure 1: *Relative frequency of moderate and severe oral motor impairments in a cohort of children and adolescents with myotonic dystrophy (DM; n=56), distributed in three subgroups. Classical form of DM was represented only by two individuals in this study and is therefore not included in figure. No moderate or severe impairments were found in control group.*

STATISTICAL ANALYSIS

The statistical analysis was undertaken using the software SPSS for Windows. Kendall's tau_b was used for cross-tabulations between categorical data. Because of the small samples, non-parametric tests were used for comparison between groups: Wilcoxon's signed rank test (two related samples), the Mann–Whitney *U*-test (two unrelated samples), and the Kruskal–Wallis test (more than two independent samples).

Results

ORAL MOTOR AND SPEECH BEHAVIOUR

Spontaneous facial expression and the rest position of lips and tongue could be observed and evaluated in all patients. Facial expression was impaired in all the individuals with DM, in contrast with none in the control group. Nine were mildly affected, 22 were moderately affected, and 25 were severely affected. Forty-nine patients had open mouth at rest, 37 of them wide open. In addition to open mouth, nine patients had the tongue in a low and forward rest position (between the front teeth) most of the time. Nine controls had a half-open mouth and two occasionally had the tongue between the teeth at rest.

Five children had no speech or fewer than 10 spoken words, and one female had selective mutism. The intelligibility of spontaneous speech was evaluated in the other 50 patients with DM and in the controls. Most of these patients had affected speech, of whom 14 had mildly reduced, 21 moderately reduced, and 9 severely reduced intelligibility. Reduced intelligibility was more common and more severe in the congenital forms of DM than in the childhood and the classical form, and the younger children were significantly more affected (Kendall's tau_b=0.244, $p<0.05$). No one in the control group had reduced intelligibility.

The examinations of lip functions and tongue motility required collaboration and were completed or partly conducted in 48 patients. Five females and three males (median 5y 10mo; range 2y 6mo–13y 2mo) were unable to follow the examiner's instructions; two patients had severe congenital DM and five mild congenital DM, and one had childhood DM. In this group five had no speech and one had selective mutism. All had learning disability and behavioural problems. Another six patients failed to participate in one or two tasks. Moderate to severe dysfunction of the lips was found in 38 of 50 individuals. Tongue motility was assessed in 46 patients, and 24 of these had a moderate or severe impairment. Moderate to severe impairment of lip function (χ^2 10.209, $p=0.017$) and tongue motility (χ^2 10.393, $p=0.016$) was most frequently seen in severe congenital DM and was more common in congenital DM than in childhood DM (Fig. 1). Males were significantly more affected than females (lip function, $z=-2.250$, $p=0.024$; tongue motility, $z=-3.370$, $p=0.001$; Fig. 2). There was a clear association between reduced intelligibility and lip function (Kendall's tau_b=0.568, $p<0.001$) and tongue motility (Kendall's tau_b=0.406, $p<0.001$). All the examined children with DM who were younger than 6 years old had moderate to severe oral motor difficulties. The facial expression was moderately affected in one patient with the classical form of DM; otherwise both this group and the controls had no or only mild oral motor impairments.

The word-repetition task revealed that bilabial consonants

were produced with the tongue between the lips or with the lower lip against the upper teeth in 17 of 48 patients, and 16 of 46 patients produced dental consonants with the tongue-tip between the teeth. Velar consonants were dentalized in 4 of 44 patients, all with the severe congenital form of DM.

LIP FORCE

It was difficult for some of the patients to cooperate in the procedure of measuring lip force, especially the younger children. The results were therefore based on 39 patients and their matched controls: 10 had severe congenital DM; 12 had mild congenital DM; 15 had childhood DM; and 2 had classical DM. The ages of the patients in this group were 5 years 8 months to 21 years 5 months (median 14y 8mo).

The measurement showed that most patients with DM (69.2%) had weak or very weak lip muscles (lip force<8N, which was the cutoff score for the controls). The difference in lip force between cases and controls was highly significant (z=–5.410, p<0.001). Mean value for the study group was 7N (SD 3.5), in contrast with 21N (SD 7.8) for the control group.

INFORMATION FROM THE QUESTIONNAIRE

Information about eating, drinking, and ability to swallow the saliva was collected for 54 patients with DM and for 54 controls. The specific difficulties with eating and drinking reported by the parents and patients are presented in order of frequency in Table III. In all, 28 patients with DM (51.9%) were reported to have problems with eating and drinking, of whom 20 had one or two specified problems and the rest had three or more. Three controls reported one problem each. Most patients could chew ordinary food, but seven patients (median 13y 2mo; range 2y 10mo–21y 5mo) needed mashed foods, one 5-year-old child was still primarily bottle-fed, and two children had gastrostomy.

Twenty patients (37%) were reported to have problems with drooling: in nine this was slight, in 10 moderate, and in one severe. The problem with saliva control was evenly spread between sex and age groups. Three patients with slight drooling and one with moderate drooling had childhood DM; the other 16 had congenital DM. All patients with moderate and severe drooling had moderate to severe lip dysfunction and weak lips.

Discussion

There was a high prevalence of orofacial dysfunctions such as impaired facial expression, unintelligible speech, deviant production of consonants, dysphagia, and drooling in this cohort of children and adolescents with DM, in contrast with healthy controls. Different aspects of oral motor behaviour were shown to be affected such as muscle strength, muscle tone at rest, and range of movement in lips and tongue. All the examined preschool children with DM had moderately or severely impaired lip function and tongue motility. This could probably be explained as part of a general developmental delay, which is common in children with DM.[2,4–7] As expected, it was difficult for many of the children with DM to take part in the test procedure and some of them were unable to follow the examiner's instructions at all. Specific for this group was that they were relatively young, had severe communication disorders, and, all except one, had the congenital type of DM. The exclusions might have affected the results concerning lip function and tongue motility.

The high prevalence of impaired facial expression found in this study is in concordance with other studies of the younger population with DM.[4–7] Facial, masticatory, and velopharyngeal weakness have also been observed as prominent features in congenital and childhood DM.[4–6,15] By measuring the lip force we could confirm that the facial muscles were weak in most patients, in contrast with controls. Neither the bite force nor the velopharyngeal function was examined here, but the patients with wide-open mouth probably had weak jaw muscles, and velopharyngeal impairment was suspected in some patients because of clinically observed hypernasal speech.

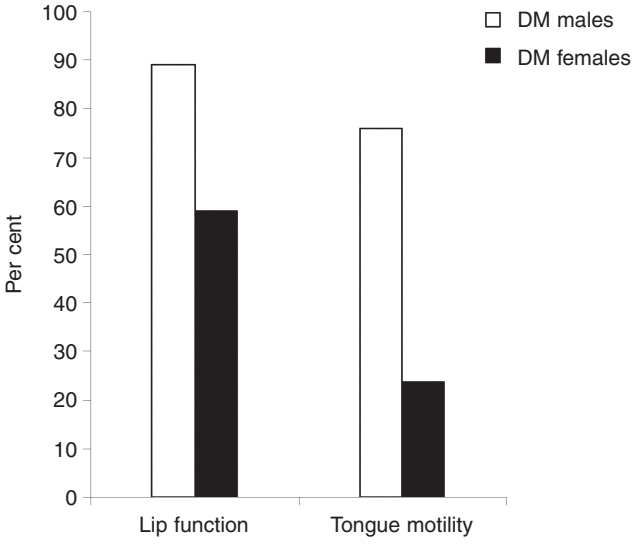

Figure 2: *Relative frequency of moderate and severe oral motor impairments in a cohort of children and adolescents with myotonic dystrophy (DM; n=56), distributed by sex.*

Table III: Frequency of specific eating and drinking difficulties in 54 individuals with myotonic dystrophy and in 54 controls

Eating and drinking difficulties	*Myotonic dystrophy*			*Controls*	
	n	*%*	*Age (y:mo)*[a]	*n*	*%*
Has difficulty in getting food off spoon with lips	11	20.4	13:2 (2:10–17:11)	0	0
Takes long time to swallow bites of food	11	20.4	13:2 (4:0–21:5)	0	0
Food and liquids leak out of corners of mouth	9	16.7	13:2 (4:0–17:0)	0	0
Food gets stuck in gums	8	14.8	10:9 (4:0–21:5)	0	0
Swallows large pieces of food without chewing	5	9.3	12:10 (3:2–21:2)	2	3.7
Chokes on food	4	7.4	10:4 (4:0–16:6)	0	0
Coughs when receiving liquids	4	7.4	15:7 (4:0–17:11)	0	0
Presses tongue forward when swallowing	4	7.4	6:9 (2:10–12:7)	0	0
Food and/or liquid goes up the nose	3	5.6	8:7 (2:6–9:7)	1	1.9

Information was collected from yes/no questions in a questionnaire to the families. [a]Results are medians (range).

The most frequently encountered difficulties in eating and drinking were associated with the oral preparatory phase of swallowing. According to the families' reports, there were some signs indicating swallowing difficulties in a few of the patients, such as coughing and choking during meal times, and nasal reflux. There was no other signs of aspiration, such as frequent pneumonias or gurgled voice after swallowing. Dysphagia with subclinical symptoms has been reported in adults with DM[16,17] and could, therefore, be suspected as a risk factor for younger patients as well. Swallowing cannot be evaluated by clinical observation only; it was therefore not included in the study. Neither did we investigate whether the reported eating problems were associated with gastrointestinal disturbances, respiratory problems, or malocclusion. We found no description of drooling as a symptom of DM in the literature. In the present study, one-third of the children and adolescents drooled. Drooling was reported in all age groups, which indicates that this problem will not be outgrown. About 50% of the questionnaires from the DM group were filled out by a parent who had the classical form of DM. Cognitive deficits are common in this diagnosis,[18] and in some cases this could have influenced the answers.

The ability to compensate successfully for orofacial dysfunction is striking in this patient group. The tongue was generally less affected than the facial muscles and could, therefore, compensate for impaired lip function to some extent. The children used the tongue, the teeth, the chin muscle, or the hand to compensate for impaired lip closure during speech, sucking, swallowing, and chewing.

It is well known that many of the symptoms associated with DM are more frequent and more severe in congenital DM than in the childhood-onset type.[2,4,6,7] This was also true for the different aspects of orofacial dysfunctions explored in this study. In contrast, our finding that males had a significantly higher frequency of oral motor impairments than females is not consistent with previous reports. The sex difference could be explained partly by the higher proportion of males in the severe congenital group and the fact that the median age of the males was lower than for females. A sex difference in the prevalence and symptoms of DM and between subgroups of DM has not to our knowledge been previously noted in the literature.

Flaccid dysarthria was clinically considered the main reason for reduced intelligibility in this study group, but communication was also influenced by cognitive and neuropsychiatric difficulties in some patients. There is a need for further investigation into what causes the communication problems in congenital and childhood DM. Other important areas for further research are oral motor development in this diagnosis and the effect of oral motor therapy.

Conclusion

We conclude that orofacial dysfunctions presenting as impaired facial expression, reduced intelligibility, eating and drinking difficulties, and drooling are common features in congenital and childhood DM. Children with DM should, therefore, be referred to a speech-language pathologist early, to obtain support for optimal development of feeding and communication. Speech-language pathologists should also be aware that facial weakness and speech problems are helpful clinical signs for recognizing DM.[5]

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