Development of Orofacial Functions in Young Individuals with Myotonic Dystrophy: A Retrospective Study

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In a previous cross-sectional study of 56 children and adolescents with myotonic dystrophy type 1 (DM1) and 56 controls, orofacial dysfunction was found to be common in this disease. Using the same cohort, the present retrospective follow-up study describes changes in facial expression, intelligibility, eating and drinking ability, and saliva control that occur during the progression of the disease. Thirty-five patients (mean age 10 years) had been assessed with the same protocol 3–4 years earlier. A speech-language pathologist assessed facial expression and intelligibility, and the families answered a questionnaire. The progressive weakening of the orofacial muscles often began before puberty and manifested as deteriorated facial expression, reduced intelligibility, and increased drooling. Orofacial functions improved during childhood in some patients.

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Myotonic dystrophy type 1 (DM1) is a disease with autosomal dominant inheritance caused by a trinucleotide repeat expansion on chromosome 19q13. There are four subtypes of DM1 defined by age at onset and predominant symptoms: congenital, childhood, classical (or adult), and mild (Koch, Grimm, Harley, & Harper, 1991). DM1 is a multisystemic disorder affecting skeletal muscle, heart, lungs, smooth muscle, brain, peripheral nerve, endocrine regulation, and skin. Distal muscles, including the orofacial area, are generally more affected than proximal muscles (Koch et al., 1991). Newborns with DM1 are severely hypotonic, but muscle tone and muscle strength improve as the children grow older (de Die-Smulders, 2004; Roig, Balliu, Navarro, Brugera, & Losada, 1994). Muscle weakness and wasting is slowly progressive in DM1. It is still unclear at what age improvement in muscle strength turns to deterioration in children with congenital DM1, but around puberty has been suggested (Hageman, Gabreels, Liem, Renkawek, & Boom, 1993). Myotonia in the hands, tongue, and jaw muscles is a common symptom in adults with DM1. In children, clinical myotonia usually becomes successively more common with age (Kroksmark, Ekström, Björck, & Tulinius, 2005). Most children with DM1 have a developmental delay and cognitive deficits and in some children, DM1 is associated with a neuropsychiatric diagnosis (Martinello, Piazza, Pastorello, Angelini, & Trevisan, 1999; Steyaert, de Die-Smulders, Fryns, Goossens, & Willekens, 2000; Steyaert et al., 1997). Another sign of central nervous system involvement is excessive tiredness (Hilton-Jones, Damian, & Meola, 2004). Whether brain deficits associated with DM1 are progressive is still uncertain (Marchini et al., 2000; Staeyaert et al., 1997; Tuikka, Laaksonen, & Somer, 1993). Changes in orofacial functions over time, such as the pattern of improvement or deterioration of facial expression, speech, sucking, chewing, and swallowing in young individuals with DM1 has not yet been described in the literature.

A speech-language pathologist examined the orofacial functions of 56 children and adolescents with DM1 (ages 2–21 years) and age- and gender-matched healthy controls as part of a multidisciplinary survey of clinical manifestations of DM1 in children and adolescents living in the western and southern regions of Sweden (Sjögreen et al., 2007). This cross-sectional study found a high prevalence of orofacial dysfunction with major (moderate or severe) impairment of facial expression, intelligibility, lip function, tongue motility, and lip force. Wide-open mouth at rest was common, and deviant production of bilabial and dental consonants was a frequent finding. Self-reported prevalence of dysphagia was 52% and drooling 37%. Impaired lip function and tongue motility were significantly more common in males than in females, and orofacial dysfunction was most common and most severe in congenital DM1. Facial expression was moderately affected in one patient with classical DM1; otherwise, this group and the controls had no or only mild impairments.

The same speech-language pathologist (LS) had assessed a majority of the participants in the cross-sectional study about 4 years earlier. The same methods were used, except for lip force measurement, and all examinations were documented by videotape recordings. This enables a comparison of results from the cross-sectional study with earlier findings and, thereby, a retrospective description of changes in orofacial functions over time in young individuals with DM1. Kroksmark et al. (2005) reported muscle strength and motor function findings in this group of patients and Engvall et al. (2007) oral health findings.

In the present retrospective follow-up study we endeavored to explore changes in facial expression, intelligibility, eating and drinking ability, and saliva control in young individuals with DM1 from a retrospective perspective. Furthermore, we investigated whether improvement or deterioration could be related to gender, DM1 subgroup, or age group and compared the development of orofacial functions in children and adolescents with DM1 to healthy peers.

METHOD

Participants

Thirty-five patients with DM1 (congenital or childhood) and 31 age- and gender-matched controls were enrolled (Table 1). Inclusion criteria were that they had been examined twice by the same speech-language pathologist, had filled out a questionnaire on two occasions (assessments A and B), and were older than 2 years of age at the first assessment (A). The time between the two assessments varied from 3 years and 2 months (3:2) to 4:8 years (mean = 3:7 years). The patients were divided into three age groups according to age at assessment B. Age group I consisted of 11 children below age 11 years (prepuberty), age group II of 13 children 12–15 years (puberty), and age group III of 11 adolescents 16–21 years (postpuberty). Twen-

TABLE 1. Number, gender, and age of patients with myotonic dystrophy type 1 (DM1) and healthy controls at two assessments (A and B) made 3–4 years apart.

			Median Age (Range) Years:Month	
Participants	**No.**	**Gender, M/F**	**Assessment A**	**Assessment B**
Congenital DM1	22	13/9	10:6 (3:1–17:8)	14:1 (6:6–21:5)
Childhood DM1	13	5/8	9:5 (5:1–17:8)	12:7 (8:3–20:10)
DM1, whole group	35	18/17	10:6 (3:1–17:8)	13:9 (6:6–21:5)
Controls	31	17/14	10:0 (2:8–17:1)	13:3 (6:8–21:5)

ty-nine of the patients attended special schools for pupils with learning disabilities, and the remaining six attended ordinary schools.

Informed consent was obtained from each family, and the Ethics Committees of the Medical Faculties at Göteborg and Lund Universities approved the study.

Procedure

Videotaped recordings from assessment A were evaluated and compared to the results of recordings from assessment B. Assessment A recordings were made with an analogue video camera (Sony Handycam video Hi8, CCD-TR790E PAL, Sony Corporation, Tokyo, Japan) and assessment B with a digital video camera (Sony Handycam, 3CCD Mega Pixel, Sony Corporation, Tokyo, Japan). The setting for both assessments was a dental clinic near each patient's residence.

Assessment

An evaluation of facial expression and intelligibility of spontaneous speech was made from assessment A video recordings. The ratings were made on 4-point scales where 0 = normal function, 1 = mild deviation, 2 = moderate deviation, and 3 = severe deviation. Table 2 describes these definitions for intelligibility in detail. Three children with DM1 could not be evaluated for development of intelligibility: one child suffered from selective mutism, and two children had not developed speech or spoke less than 10 words at the first examination.

Questionnaire

All families filled out the same questionnaire (Andersson-Norinder, 1996) at assessments A and B. Questions about eating and drinking ability and drooling were selected for this study.

The families answered "yes" or "no" if the child had one or more of the following nine difficulties: "Has difficulty getting food off a spoon with lips," "Takes a long time to swallow bites of food," "Food and liquids leak out of the corners of the mouth," "Food gets stuck in the gums," "Swallows large pieces of food without chewing," "Chokes on food," "Coughs when drinking liquid," "Presses tongue forward when swallowing," "Food and liquid go up into the nose." Patients were assessed to have no (score = 0), mild (score = 1), moderate (score = 2), or severe (score = 3) difficulties eating and drinking if the families specified 0, 1, 2, or 3 or more such difficulties, respectively, on the questionnaire.

The questionnaire also asked whether the subject had any problems with drooling, and if so, the respondent was asked to estimate the amount of drooling on a scale. The definitions were saliva on the lips only (slight drooling), saliva on the chin (moderate drooling), saliva on the clothes (severe drooling), and saliva on hands and objects (profuse drooling) (Blasco, & Allaire, 1992).

Statistical analysis

Data were analyzed with the Statistical Package for the Social Sciences (SPSS for Windows, v. 14.0). Nonparametric tests were used to analyze categorical data and make comparisons between groups: Wilcoxon's signed rank test (two related samples), Mann-Whitney U Test (two unrelated samples), Kruskal-Wallis Test (more than two independent samples), and Kendall's tau_b (correlation analysis).

Reliability

To study inter- and intraobserver reliability of the assessment of facial expression and intelligibility, a speech-language pathologist not involved in the study evaluated 30 randomly chosen videotape recordings of patients with DM1 and their controls

TABLE 2. Definitions used to define intelligibility of individuals with myotonic dystrophy type 1 and their matched controls.

Assessment of Intelligibility
0 = Normal function: speech is fully understood
1 = Mild deviation: speech is largely understood; repetitions and verifications are occasionally needed
2 = Moderate deviation: there is an ongoing need for repetition and verification; listener effort is required
3 = Severe deviation: only a few words or phrases recognizable; alternative and complementary methods of communication are required

while the first examiner reevaluated 12 recordings. Table 3 presents exact percentage agreement ("point by point") for inter- and intraobserver reliability of assessment A and assessment B. Interobserver agreement was assessed using 70 videotape recordings and intraobserver with 52. When all results were combined, interobserver agreement was 80.4% (weighted kappa = 0.74) for facial expression and 77.5% (weighted kappa = 0.76) for intelligibility and intraobserver agreement was 87.5% (weighted kappa = 0.94) for facial expression and 87.5% (weighted kappa = 0.93) for intelligibility. Figure 1 presents agreement and disagreement for intra-observer reliability.

RESULTS

Facial Expression

All but two patients in assessment A and all patients in assessment B had impaired facial expression (Figure 2). Facial expression was significantly more impaired in assessment B than in assessment A ($z = –3.162$, $p = 0.002$). Patients with congenital DM1 had a significantly more impaired facial expression than patients with childhood DM1 (A: $z = –3.947$, $p = 0.001$; B: $z = –3.463$, $p = 0.001$). Males were significantly more affected than females (A: $z = –2.544$, $p = 0.011$; B: $z = –2.376$, $p = 0.018$).

Facial expression improved in none of the patients and deteriorated in 10 patients, 3 of whom had congenital DM1 (13%, group size 22) and 7 childhood DM1 (53%, group size 13). This difference was significant ($z = –2.508$, $p = 0.012$) for the group as whole. Deteriorated facial expression was not clearly associated with gender or age (Table 4).

Intelligibility

Of the 32 patients who were evaluated for speech, 27 at assessment A and 28 at assessment B had reduced intelligibility. Figure 2 illustrates frequency and degree of dysfunction. Both assessments revealed that males in general had significantly more reduced intelligibility than females (A: $z = –2.399$, $p = 0.016$; B: $z = –2.156$, $p = 0.031$).

Intelligibility was improved in four patients (three males and one female). Improvements were only found in children under 15 years (see Table 4). Of the eight patients who had deteriorated intelligibility, one belonged to the youngest age group and the others were 12 years or older at assessment B. Neither improved nor deteriorated intelligibility was found to be related to age, gender, or DM1 subgroup.

Eating and Drinking Ability

Less than half of the group had difficulties eating and drinking (see Figure 2). The most common problems were that it took a long time to swallow bites of food (A: n = 9, B: n = 6), the tongue was pressed forward when swallowing (A: n = 6, B: n = 2), food and liquids leaked out of corners of mouth (A: n = 6, B: n = 6), and it was difficult to get food off the spoon with the lips (A: n = 6, B: n = 6).

The development of eating and drinking skills fluctuated widely, especially in patients with congenital DM1 (see Figure 1). Nine patients had become better and five worse in the interval between assessments. Difficulties with eating and drinking ceased in four patients and started in four. Eating and drinking ability improved *and* deteriorated equally in all age groups. Deteriorated function was more frequent in males (4:1) than in females, but not significantly so ($z = –1.361$, $p = 0.174$).

Drooling

Drooling was reported for 12 patients in assessment A and 16 in assessment B (see Figure 2). Drooling was improved in four children under age

TABLE 3. Results of reliability testing of assessments A and B. Inter- and intrareliability are described as exact percentage agreement.

	Exact Percentage Agreement (%)					
	Interobserver			Intraobserver		
Orofacial Function	**A**	**B**	**A+B**	**A**	**B**	**A+B**
Facial expression	63.3	97.4	80.4	75.0	100	87.5
Intelligibility	80.0	75.0	77.5	80.0	95.0	87.5
Total	71.7	86.2	79.0	77.5	97.5	87.5

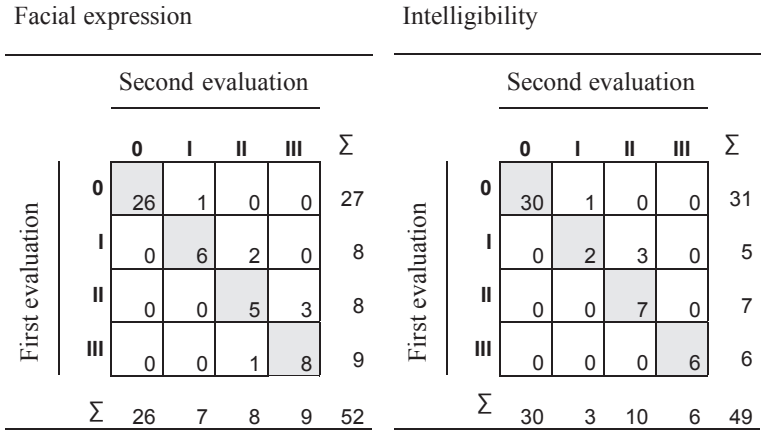

Figure 1. Intraobserver agreement concerning assessment of facial expression and intelligibility. The first observer reassessed 52 patients and controls (12 in connection to assessment A and 40 to assessment B). *Note:* 0 = no, I = mild, II = moderate, and III = severe deviation. Figures outside the shadowed boxes indicate disagreement.

TABLE 4. The frequency of improved and deteriorated orofacial functions observed in a retrospective follow-up of 35 children and adolescents with myotonic dystrophy type 1. Distributions according to gender, subgroup, and age group are listed.

		Gender		Subgroup		Age Group (Years)		
Improved functions	**No.**	**m**	**f**	**Congenital**	**Childhood**	**I (5–11)**	**II (12–15)**	**III (16–21)**
Intelligibility	4	3	1	2	2	2	2	0
Facial expression	0	0	0	0	0	0	0	0
Eating and drinking	9	5	4	7	2	4	3	2
Drooling	4	1	3	2	2	4	0	0
Deteriorated Functions								
Intelligibility	8	4	4	5	3	1	3	4
Facial expression	10	4	6	3	7	4	3	3
Eating and drinking	5	4	1	4	1	1	2	2
Drooling	11	7	4	9	2	3	3	5

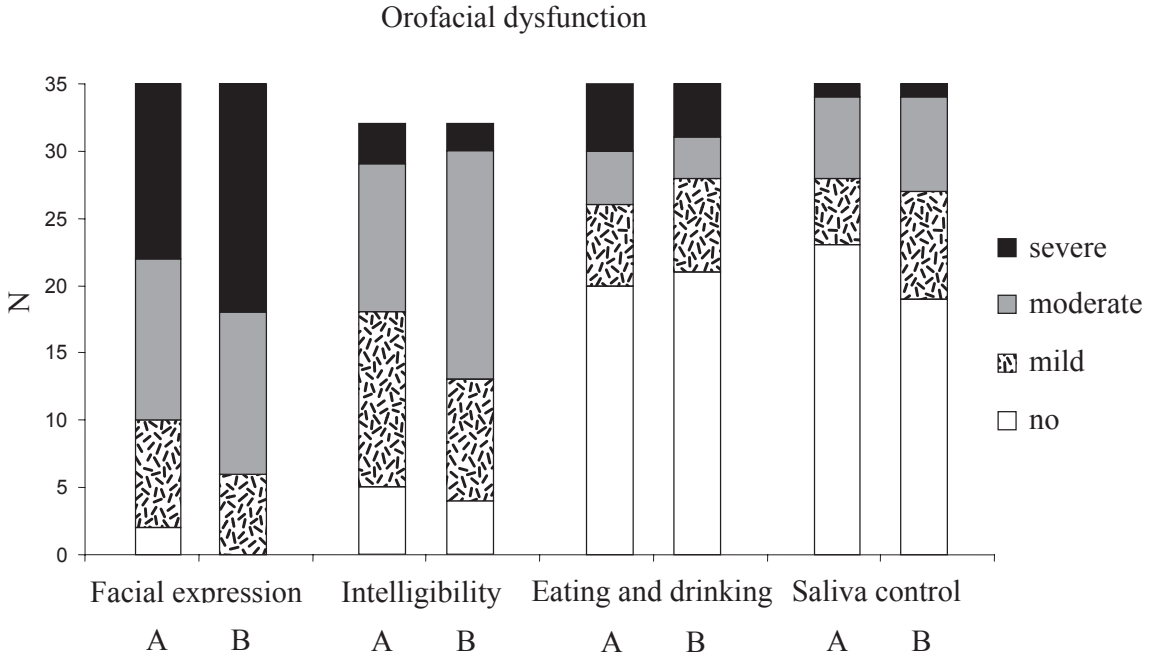

Figure 2. The frequency and degree of orofacial dysfunction in 35 patients with myotonic dystrophy type 1 assessed on two occasions with a 3- to 4-year interval between assessments (A and B).

10, one boy and three girls, at assessment B. Deteriorated saliva control was noted in 11 patients, of whom 9 had congenital and 2 childhood DM1 (see Table 4). Seven of these patients had no problems with saliva control at assessment A. No difference was found between males and females concerning changes in drooling.

General Aspects Concerning Changes in Orofacial Dysfunction

Figure 3 depicts the distribution of changes over time for the study participants. Thirteen individuals, eight males and five females, improved in one or more than one area of orofacial dysfunction in the interval between assessments. The median age for patients whose dysfunction had improved was lower (9:10 years) than the median age for the whole group (13:9 years) and for the patients with deteriorated functions (14:8 years). The correlation between age and improved orofacial functions was significant (Kendall's tau_b = –0.320, $p = 0.024$).

In 22 patients, 12 males and 10 females, one or more orofacial function had deteriorated between the two assessments. Deteriorations in saliva control and eating and drinking ability were combined in 4 patients, and the same number of patients had deteriorations in facial expression and reduced intelligibility.

A combination of improved and deteriorated functions was found in eight patients, of whom six had improved eating and drinking ability in combination with deterioration of facial expression, intelligibility, or saliva control. The orofacial functions of another eight patients were unchanged between the two assessments.

No one in the control group exhibited or reported any major deviation in orofacial functions at the first or the second assessment. Two young men reported that they swallowed large pieces of food without chewing and one that he sometimes choked on food.

DISCUSSION

Orofacial dysfunction manifests in most children and adolescents with DM1 as impaired facial expression, feeding problems in infancy, chewing and swallowing difficulties, and unintelligible speech (Sjögreen et al., 2007). In the present study we wanted to investigate differences in the development of orofacial dysfunction in young individuals with DM1 and healthy peers. We recorded whether an individual had improved, deteriorated, or unchanged facial expression, intelligibility, eating and drinking ability, and saliva control in the interval between two assessments. As we expected, the

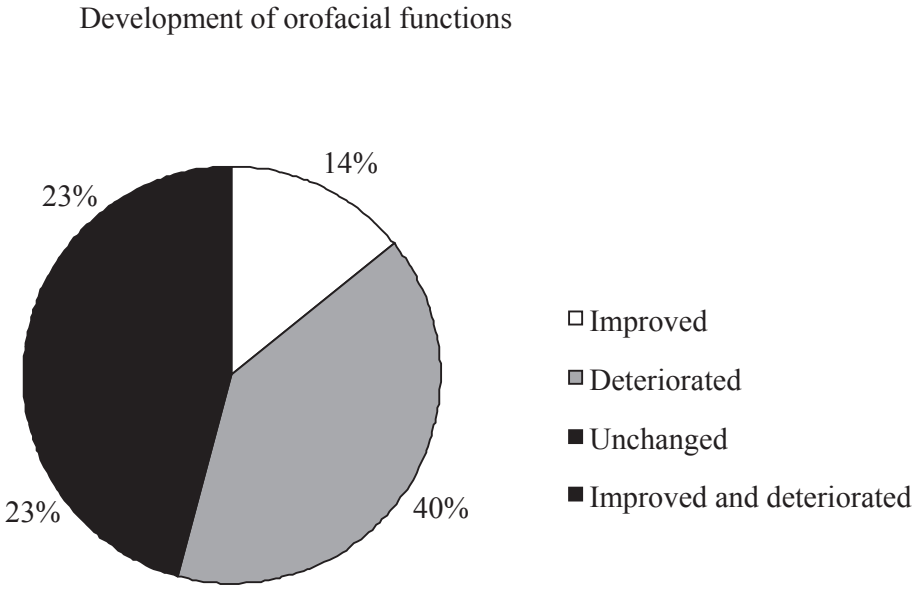

Figure 3. The proportion of patients with improved, deteriorated, unchanged, or improved *and* deteriorated orofacial dysfunction in a retrospective follow-up of 35 patients with myotonic dystrophy type 1.

healthy controls had no major orofacial dysfunction in either assessment. Some patients developed an orofacial dysfunction in one area and improved in another between the two assessments, which indicates that the changes have different explanations. General developmental delay is common in DM1 (de Die-Smulders, 2004), and therefore late maturation of oral motor skills could be one explanation for the improvement. Maturity also increases social awareness and could therefore be a reason for improved saliva control and perhaps improved eating skills. In Sweden, all children with disabilities are offered special care by a team of professionals. Most patients in this study had received treatment from speech-language pathologists and physiotherapists and most had excellent dental care. So improvement could also be an effect of therapy. According to van der Kooi, Lindeman, and Riphagen (2005) "normal" participation in sports and work does not appear to harm the muscles of patients with DM1, but there is insufficient evidence to establish that it is beneficial. Whether oral motor therapy in patients with DM1 has an effect is an urgent subject for further research.

It is well known among clinicians that muscle function and muscle strength of children with DM1 often improve during the first years of life, before muscle weakness sets in and motor functions are affected (de Die-Smolders, 2004; Roig et al., 1994). Most of the patients in this study worsened between assessments, and in some children, the progression of the disorder could be detected before puberty.

Deterioration of facial expression was more common in childhood DM1. This was expected since most patients with congenital DM1 already had severely impaired facial expression at the first assessment and could not get worse. We found no other pattern of improvement or deterioration that could be related to either subgroup of DM1. This might have been possible with a larger study group and comparable age groups.

The only significant gender difference found in this study was related to the degree of impaired facial expression and intelligibility. That males had more severe dysfunction could be explained by the high proportion of males in the subgroup of patients with congenital DM1. The cross-sectional study (Sjögreen et al., 2007) in which the patients of the present study participated found that males had more impaired oral motor skills than females.

Drooling is a symptom that often decreases when patients grow older, as we saw in 4 patients in this study group. A study of the prevalence of drooling in children with cerebral palsy found that the degree of drooling decreased as the child's dental age increased (Tahmassebi & Curzon, 2003). In this study, 11 patients had deteriorated saliva control, of which 7 began to drool in the time be-

tween the two assessments. We interpret this as a clear sign of disease progression. Worse drooling was related to worse eating and drinking ability in some patients.

There is a risk that some of the developmental changes registered in this study could be an artifact of uncertain or faulty ratings. Repeated assessment by the same observer and by a second observer, however, showed acceptable or good agreement, which supports the reliability of the results. Some parents with classical DM1 might have had cognitive deficits that affected their ability to answer the questions (Winblad, Lindberg, & Hansen, 2005).

CONCLUSIONS

The results of this study indicate that difficulties with intelligibility, eating and drinking, and drooling could be outgrown in some young individuals with DM1—at least temporarily. The study also shows that deterioration of orofacial functions is common and could start some years before puberty in congenital and childhood DM1. In general, improvements occurred earlier in life than deteriorations. Improvement and deterioration of different orofacial functions can be a parallel process in the same individual. The progression of muscle weakness in DM1 is clearly expressed in the deterioration of the facial expression. There is, however, a need for more reliable instruments for the objective evaluation of facial muscles in clinical practice and research. There is also a need for further and deeper investigations into the different aspects of motor speech, language, and communication disorders in congenital and childhood DM1.

Children with DM1 should get an early referral to a speech-language pathologist with special knowledge about neuromuscular diseases to get support for optimal development of feeding and communication and to get advice on compensatory strategies when intelligibility is reduced. The speech-language pathologist should follow the oral motor development of children and adolescents with DM1 and be aware that a deterioration of orofacial functions can begin early in life but also that improvements may occur throughout childhood and adolescence.

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