

## BACKGROUND

With increasing age, patients with Duchenne muscular dystrophy (DMD) and spinal muscular atrophy type 2 (SMA II) suffer from progressive weakness in facial and oropharyngeal muscles and this affects their ability to speak, eat, swallow, cough and breathe. The underlying mechanism of dysphagia has been documented in several studies, so the aim of this study was to investigate how and to which extent patients experience orofacial problems in their daily life.

## METHODS

Based on knowledge from previous studies and patient experiences, we developed a questionnaire to identify problems related to respiratory treatment, speech, eating/drinking and teeth/tongue. The questionnaire was adjusted according to evaluations from Scandinavian specialists in NMDs and/or orofacial problems, and to interviews with seven adult persons diagnosed with DMD or SMA II. The questionnaire was sent to all patients  $\geq 10$  years of age registered with the Danish Rehabilitation Centre for Neuromuscular diseases (RCFM) with a generically verified diagnosis of DMD (n=139) and SMA II (n=62).

## CONCLUSION

Patients with DMD and SMA II report orofacial problems from an early age. In patients with DMD, problems increase over time. Problems are mainly related to feeding and speaking, but also to respiratory intervention. Only few patients in this study had received counselling from a dietician and/or speech therapist, and despite the fact that dysphagia was common, very few patients used a tube for supplementary feeding.

## RESULTS

101 patients with DMD and 48 patients with SMA II answered the questionnaire; age distribution of participants is illustrated in Table 1.

**Ventilation** Forty-four patients with DMD and 27 patients with SMA II used or had used non-invasive ventilation (NIV), 46 patients (DMD = 36, SMA II = 10) were tracheotomized and used full-time invasive ventilation (IV), Table 2. NIV was associated with side effects such as pressure sores and increased secretions. IV was associated with side effects such as air leaks from the tracheotomy and increased secretions (Fig 1).

**Speech** The majority of patients did not define speech as a problem, but 20 % of patients with DMD and 16 % of patients with SMA II could not speak distinctly for more than 30 minutes. This was related to age in DMD patients but not in SMA II patients, whose speech were

affected by other factors such as fatigue and posture; three patients with DMD used their ventilator to obtain a more distinct and vigorous speech. Only 15 patients with DMD and two patients with SMA II had received speech therapy. Factors that influence speech are illustrated in Table 3.

**Eating** Several patients refrained from eating certain types of food because of chewing problems or because of limited mouth opening; these problems were especially pronounced in SMA II patients. Dysphagia was present in both DMD and SMA II patients, but where problems with food transition and choking were almost equally common in the two groups, problems with food residue in the throat were far more common in patients with SMA II. Age had an influence on problems with eating and drinking in DMD; this was not the

case in SMA II. Twelve percent of patients with med DMD and 10 % of patients with SMA II had a feeding tube. Only one fourth of all patients – regardless of diagnosis – had received dietary counselling. Problems related to eating and drinking are illustrated in Figure 2.

**Tongue hypertrophy** was common in patients with DMD but also present in patients with SMA II. The ability to bite upper and lower incisors together was significantly affected in both groups of patients, whereas the ability to bite upper and lower molars together were more affected in patients with DMD. More than one third of the patients said that their tooth position had changed over time, Table 4.

### SIDEEFFECTS ASSOCIATED WITH VENTILATION

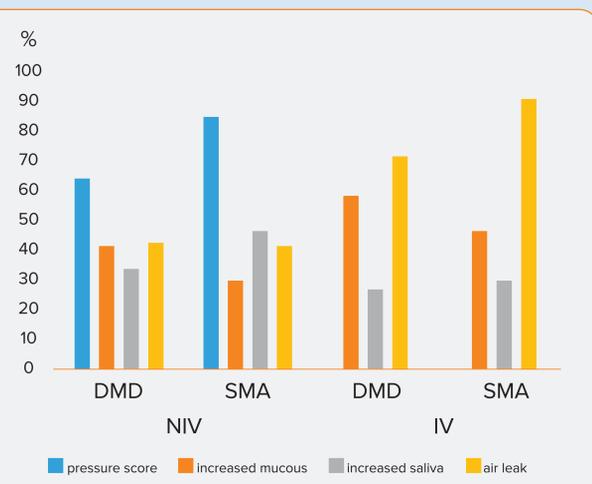


Figure 1 – Most common side effects according to number (percentage) of patients with DMD and SMA II



|          | 10-15 y | 16-20 y | 21-30 y | 31-40 y | 41-50 y | $\geq 50$ y | Total (n=) |
|----------|---------|---------|---------|---------|---------|-------------|------------|
| DMD (n=) | 41      | 15      | 26      | 15      | 4       | 0           | 101        |
| SMA (n=) | 8       | 7       | 13      | 8       | 8       | 4           | 48         |

Table 1 – Age distribution in 101 patients with DMD and 48 patients with SMA II

| %   | NIV  | IV   | Total |
|-----|------|------|-------|
| DMD | 18 % | 35 % | 53 %  |
| SMA | 35 % | 21 % | 56 %  |

Table 2 – Use of respiratory support in patients with DMD and SMA II; percentage of total number of patients. NIV = noninvasive ventilation, IV = invasive ventilation by tracheotomy.

### PROBLEMS RELATED TO EATING AND DRINKING

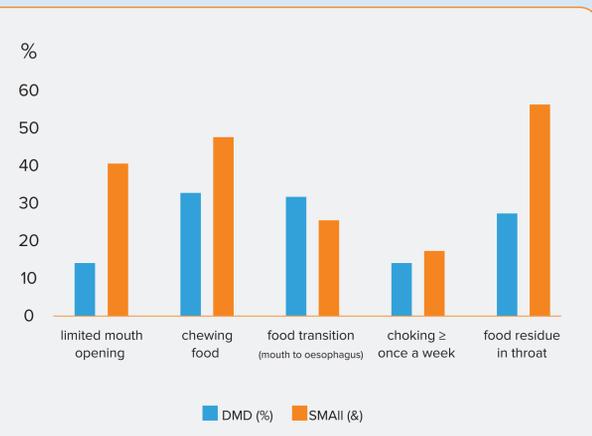


Figure 2 – Type of problems related to eating and drinking. Number of patients is illustrated as percentage of total number of patients with DMD or SMA II.

|        | Fatigue influences the ability to speak distinctly? (y/n) | Posture influences the ability to speak distinctly? (y/n) | Have you had speech therapy? (y/n) |
|--------|---|---|------------------------------------|
| DMD    | 13/87 (%)   | 9/91 (%)  | 15/85 (%)                          |
| SMA II | 23/77 (%)   | 25/75 (%)   | 8/92 (%)                           |

Table 3 – Factors that influence speech as reported by patients.

|     | Has tongue hypertrophy | Cannot bite front teeth together | Cannot bite molars together | Change of tooth position over time |
|-----|------------------------|----------------------------------|-----------------------------|------------------------------------|
| DMD | 37 %                   | 27 %                             | 14 %                        | 35 %                               |
| SMA | 13 %                   | 32 %                             | 4 %                         | 40 %                               |

Table 4 – Problems related to teeth and tongue. Numbers are a percentage of the total number of patients with DMD/SMA II.