

Orofacial dysfunctions in children and adolescents with myotonic dystrophy type - evaluation and intervention

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Abstract

Myotonic dystrophy type 1 (DM1) is a slowly progressive neuromuscular disease. The overall aim of this thesis was primarily to describe the characteristics, prevalence, and development of orofacial functions in a group of children and adolescents with DM1 and secondly to investigate the effect of lip strengthening exercises.

In total, the study population consisted of 66 individuals with DM1, five with Möbius syndrome, six with facioscapulohumeral muscular dystrophy and 106 healthy controls. Fifty-six of the patients (30 males, 26 females; median age 13 years [2–21 years]) with DM1 and 56 of the healthy age and gender matched controls were enrolled in Study I. Thirty-five patients and 31 controls were assessed twice with approximately 3–4-year intervals (Study II). Facial expression, intelligibility, oral motor performance, and lip force, were assessed by a speech-language pathologist and the families answered questions about eating and saliva control in a questionnaire. Eight individuals (7–17 years) from the same study group participated in an intervention study (Study IV). After baseline measurements, four participants began 16 weeks of treatment while the others acted as controls. Thereafter, those who had started as controls began treatment and the other had no training. Lip exercises were carried out five days a week. Follow-ups were conducted every fourth week. Assessments were performed with both quantitative and qualitative methods. These methods were developed and validated in a previous methodology study. Lip mobility was measured with 3D video analysis and lip strength with a lip force meter. Fifty healthy adults and 23 adults with diagnoses affecting the facial muscles participated in the methodology study (Study III).

All patients with DM1 had impaired facial expression. Intelligibility was considerably reduced in 30 patients (60 %), excluding 6 patients without speech. The majority had moderate or severe impairment of lip motility (76 %), tongue motility (52 %), and lip force (69 %). Deviant production of bilabial and dental consonants was common. Families reported problems with drooling (37 %) and eating (52 %). Oral motor dysfunction was most prominent in congenital DM1, and males were more affected than females. Intelligibility, eating and drinking ability, and saliva control improved during childhood in some patients. Facial expression deteriorated significantly, especially in patients with childhood DM1, but the progressive weakening of the orofacial muscles also manifested as reduced intelligibility and increased drooling. The measuring instruments were found to be reliable as well as clinically relevant and could therefore be used for evaluation of treatment as a complement to qualitative assessments. Seven of eight participants in the intervention study improved lip strength but not lip function.

Orofacial dysfunctions such as impaired facial expression, speech, eating and drooling are common in children and adolescents with DM1. Both improved and deteriorated orofacial functions could be seen in this group of patients at follow-up. The progression of muscle weakness in DM1 is clearly expressed in the deterioration of facial expression. Children and adolescents with DM1 can improve lip strength. However, improved lip strength will not automatically lead to improved lip function.

Key words: myotonic dystrophy type 1, children, facial expression, dysarthria, dysphagia, drooling, lip force, lip mobility, 3D motion analysis, oral screen.