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Nemaline Myopathy - A Case Report

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Background

Nemaline Myopathies (NM) are a group of inherited congenital myopathic conditions characterized clinically by weakness, hypotonia, and prominent hypoplasia of proximal muscles including the face. Muscle biopsy reveals large numbers of rod-shaped structures beneath the muscle fiber plasma membrane. This disorder is genetically heterogeneous and may occasionally present in adults[1].
Nemaline myopathy was originally identified and described in 1963 by Shy et al[2].
The disease affects all skeletal muscles, including the diaphragm, sparing the cardiac muscles. The weakest muscles are the facial muscles, the flexors of the neck and trunk, the dorsiflexors of the feet, and the extensors of the toes[3].

Objectives of Investigation

This case report describes a male child (M.S., born July 1998) with NM-Syndrome, who was 2 years 11 months at the time of the first examination in our department (June, 2001). He was referred from his speech therapist who had seen initial erosive and carious lesions on his anterior upper teeth.The aim of the case report is to present dental and orofacial status of this child with NM-Syndrome and exemplify dental and orofacial consequences in children with this disease.

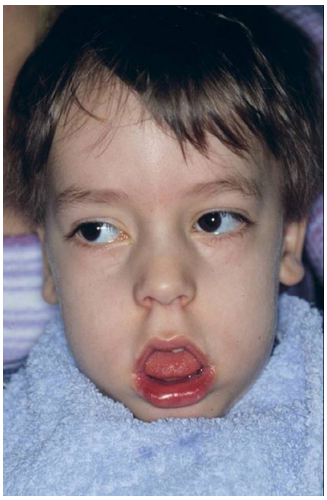
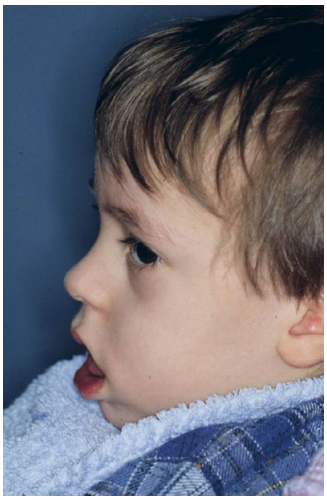

Fig. 1 and 2: facial photographs

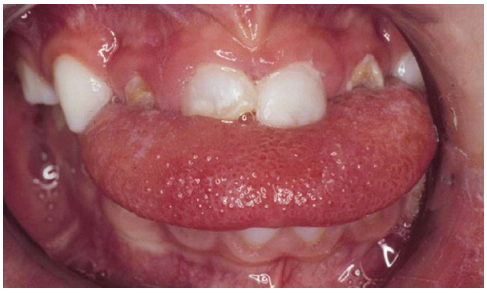

Fig. 3: Intraoral photograph

Findings

A mutation in the ACTA1 gene was identified in Patient M. It was a new, dominant mutation (not an inherited gene mutation) in which leucine takes the place of the amino acid glutamine 265.
Clinical examination of the patient showed a long, narrow face with weakness of facial muscles, tented upper lip and a fish-mouth appearance (Fig. 1 and 2). A poor masseteric and lip muscle function was recorded and was resulting in a permanent flow of saliva out of his mouth.
His tongue movement is restricted and he has difficulty swallowing. His ability to speak has been restricted to a small number of sounds or single-word statements, although his ability to understand speech is above average.
We found erosive lesions on 52, 51, 61 and 62 (palatinal and incisal) which were caused by a long-lasting reflux nearly one year ago (Fig. 3).
He is not able to stand, walk or run by himself.
Due to an occasional reflux, the upper part of his body should be placed higher than the rest during sleep.

Material and Methods

Dental prophylaxis: plaque control with polishing brush and paste (Zircate® Prophy Paste) and application of a fluoride varnish (Duraphat®) every 2-3 month.
He is undergoing logopedic therapy to train him to close his lips, improve his tongue movements and swallow better.
He also receives regular physiotherapy.

Results

After 9 month we observed a stagnation of the lesions. By undergoing a regular speech therapy, he now is able to control the perioral muscles for a short time and to close his lips.

Conclusions

Children with NM-Syndrome should be considered as patients with high risk for caries and dental erosions. Consequently they need special preventive dental care. Additionally, there is also a need for oral motoric training for these patients. Wallgren-Pettersson recommended for the management of NM active rehabilitation and vigorous treatment of respiratory infections. Physiotherapy should focus especially on the maintenance of cardiorespiratory capacity and the prevention and treatment of scoliosis. Long periods of immobilisation should be avoided[3].

References

1. Adams R, Victor M, Ropper AD: Principles of Neurology. McGraw Hill Text. 6th Edition.1997, p. 1453.
2. Shy GM, Engel WK, Somers JE, Wanko T: Nemaline myopathy: a new congenital myopathy. Brain 1963, 86, p. 793-810.
3. Wallgren-Pettersson, C: Congenital nemaline myopathy: a clinical follow-up study of twelve patients. J Neurol Sci 1989, 89, p. 1-14.

This Poster was submitted by Dr. Anahita Momeni.

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Poster Faksimile:

Nemaline Myopathy - A Case Report

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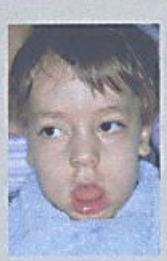

Fig. 1 and 2: Facial photographs

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We'd like to thank Mrs. Ch. Kulla, speech therapist, for participating in the treatment of this patient.