INTRODUCTION: Nemaline myopathy is a rare disease with an incidence of 1 in every 50,000 live births. It is the most prevalent of the congenital myopathies, a heterogeneous set of neuromuscular disorders present at birth or manifesting at a very early age, which affect the skeletal muscles and give rise to weakness, hypotonia and psychomotor retardation, although cognitive development remains normal.

AIM: To review the studies conducted to date on the communication difficulties and dysphagia of children with nemaline myopathy and their possible management based on speech therapy.

DEVELOPMENT: All the children presented dysphagia, with severe feeding problems during the first three years of life that nevertheless are somewhat mitigated as time goes by. In 50% of cases a gastrostomy will be used, although some oral ingestion is maintained in many of them. Nemaline myopathy gives rise to a clearly dysarthric pattern. The weakness of the muscles involved in ventilation and of the face, with a limited ability to close the mouth, leads to hypophonia, nasality and marked unintelligibility. Studies conducted on treatments based on speech therapy suggest that, in the most disabling cases of dysarthria, alternative systems of communication should be used in the first years of life so as to eliminate the frustration caused by the lack of meaningful expression. Later, communicators based on written language can be used. In the remaining cases, the aim must be to improve speech intelligibility.

CONCLUSIONS: Speech therapy can contribute to improve the quality of life of these children with two types of treatment: management of their dysphagia and improvement of their communication problems through speech or technical aids. These is insufficient scientific evidence of the effectiveness of these treatments.
Publication Types

PubMed Commons

0 comments

How to join PubMed Commons