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CASE REPORT

Tracheostomy in children with nemaline core myopathy

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Summary Two children with nemaline core myopathy (NM) who required tracheostomy are reported. One had a severe neonatal form requiring tracheostomy in the neonatal period for continuous mechanical ventilation. The other had a milder form with obstructive sleep apnoea, who underwent tracheostomy at age 22 months after adenoidectomy and a trial of nocturnal nasal continuous positive airway pressure ventilation. These cases highlight the clinical heterogeneity of the condition and the important role of the otolaryngologist in its management.

1. Introduction

Nemaline core myopathy (NM) was first described in 1963 by Shy et al. [1], and is a rare disorder characterised by muscle weakness and hypotonia with a definitive histological appearance of rod shaped inclusion bodies in skeletal muscle. It is a clinically heterogeneous condition that can be classified according to severity and age of onset, although there is no single clinical parameter that separates the various classes. The most severe form typically occurs in the neonatal period and is characterised by total lack of spontaneous motor activity, muscle contractures and ventilator dependency [2]. Left untreated, these children die from respiratory failure. Milder forms generally appear later in life and are associated with delayed motor milestones, myopathic facies but intact anti-gravity muscle activity [3]. Death from respiratory failure in this group has been reported but these children are generally not ventilator dependent [4]. Adult onset disease is associated with a slowly progressive proximal motor weakness and gradual respiratory involvement that may also be progressive [2]. Mutations in several genes encoding protein components of muscle thin filaments have been implicated in all forms of the condition [5]. Both autosomal dominant and autosomal recessive inheritance of these various mutations has been documented [2,6].

Two patients with NM are described, one with a severe neonatal form requiring prolonged ventilation and early tracheostomy, and another who presented in early infancy with failure to thrive and

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recurrent aspiration who underwent tracheostomy for nocturnal ventilation and airway protection. These cases illustrate the variability of the condition and the important role of ventilatory support and tracheostomy in the management of children with congenital myopathies.

2. Case reports

2.1. Case 1

This term male infant was born to non-consanguineous Caucasian parents at 38 weeks gestation, at a regional hospital via elective caesarean section. Mild polyhydramnios was noted 1 week prior to delivery but there had been no other problems during pregnancy. At delivery APGAR scores were 1 at 1 min and 1 at 5 min. There were no spontaneous respirations and the child was immediately intubated and ventilated. The baby was noted to have arthrogryposis, fixed flexion deformities of limb and hand joints, very few spontaneous limb movements and no axial movements. Shortly after transfer to our institution the baby was noted to have pulmonary hypoplasia on echocardiogram but a normal heart and great vessels. A presumptive diagnosis of congenital myotonic dystrophy was made but later discounted when Southern blot analysis failed to show the characteristic cytosine—thymine—guanine (CTG) triplet repeat seen in that condition. Tests for congenital myasthenic syndrome and hypothyroidism were negative. Genetic screening for chromosomal abnormalities was also normal. MRI showed a focus of ischaemic change within the right corona radiata but there were no other abnormalities in the brain or spinal cord. Cranial and renal ultrasounds were normal. Masson-Trichrome staining of a muscle biopsy taken at day 13 revealed rounded, slightly elongated red bodies less than 1 μm diameter distributed throughout and concentrated toward the edges of the myofibres. The diagnosis of NM was then made. Both parents are phenotypically normal and there is no family history of genetic disorder.

During the period of investigation the child remained endotracheally intubated and ventilated. After extensive discussion with the parents that included information regarding the poor prognosis of NM in their child, tracheostomy was performed without complication on day 60. Feeds were provided via nasogastric tube prior to fundoplication and insertion of gastrostomy tube. At 100 days the baby had only minimal spontaneous limb movement and remained ventilated in hospital.

2.2. Case 2

This term baby was born via normal vaginal delivery to non-consanguineous Caucasian parents after an unremarkable pregnancy. At delivery his APGAR scores were 7 at 1 min and 6 at 5 min. Breastfeeding was commenced shortly after birth and the baby was discharged at day 6. He was first seen at our institution at 22 days of age with feeding difficulties and failure to thrive. He then represented at 6 weeks with weight loss and regurgitation of most feeds. Examination revealed significant bulbar disco-ordination with pooling of secretions and generalised hypotonia. The baby had retroverted ears, a small jaw and a tent-shaped mouth with paucity of movement of his lower face. He had no cleft palate. Initial investigations including metabolic screen, dystrophin gene assay and MRI brain were normal. Muscle biopsy at 6 months demonstrated NM.

Moderate conductive hearing loss secondary to otitis media with effusion was noted at 12 months and managed with insertion of Shepard ventilation tubes (Medtronic Xomed, Jacksonville, FL, USA). Persistent gastroesophageal reflux disease was treated with gastrostomy and fundoplication. At this stage the child was sitting upright and beginning to make some crawling movements. Polysomnography at 15 months demonstrated severe obstructive sleep apnoea with an oxygen saturation nadir of 55% during REM sleep. Flexible nasendoscopy revealed large adenoids obstructing 70—80% of his posterior choanae and small tonsils. A trial of nasal continuous positive airway pressure (nCPAP) was then commenced. The patient underwent uncomplicated adenoidectomy shortly thereafter, with the lower pole of the adenoid pad preserved to minimise the risk of velopharyngeal incompetence. Sheehy collar button ventilation tubes (Matrix Surgical, Vic., Australia) were inserted simultaneously for persistent middle ear effusions. At 22 months he was readmitted with worsening obstructive sleep apnoea despite CPAP and tracheostomy was performed. After a 90-day admission complicated by prolonged lobar collapse and sepsis, the child was discharged on nocturnal bilevel positive airway pressure ventilation (BiPAP). He was readmitted twice in the next 12 months with pneumonia and for insertion of Richards Modified T-tube ventilation tubes (Gyrus ENT LLC, Bartlett, TN, USA) and reinforcement of his fundoplication. At 28 months he was able to crawl around furniture and speak several words. He remained cognitively normal. A repeat sleep study showed adequate ventilation with nocturnal BiPAP.